EXHIBIT Q

Victorian Institute of Forensic Medicine
65 Kavanagh Street Southbank Victoria, Australia 3006
MONASH University

REPORT AND OPINION IN THE CASE OF KATHLEEN FOLBIGG

Stephen Cordner MB BS FRCPA FRCPath
Professor of Forensic Pathology (International), Monash University
Head, International Programmes (VIFM)

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Telephone: +61 3 9684 4444 Fax: -61 3 9682 7353 Web: www.vifm.org

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REFERENCES

REPORT AND OPINION IN THE CASE OF KATHLEEN FOLBIGG

INSTRUCTING SOLICITOR:

Shaun McCarthy
Director, University of Newcastle Legal Centre (UNLC)
Newcastle Law School
Faculty of Business and Law
The University of Newcastle

PRELIMINARIES

Materials:

Reports available to the trial

- Dr Cumming. Autopsy report of Caleb Folbigg
- Dr Bishop, Dr Singh-Khaira. Autopsy report of Patrick Folbigg (and the neurohistopathology report from Dr Khan in the form of a letter).
- Dr John Hilton, Autopsy report of examination of Sarah Folbigg.
- Dr Allan Cala. Autopsy report of Laura Folbigg
- Police report of death of Sarah Folbigg and the autopsy report
- Berry PJ (Professor) (2000) 28 page Report faxed to Singleton Police 1 Dec, 2000.
- Professor Peter Herdson. Report in the four deaths of the Folbigg children in the form of an Expert Certificate.
- Solicitor for PP letter to Legal Aid, 18/02/03 enclosing autopsy report for CMP Inst Case No: 00/13002. Autopsy date: 27 June, 2000

Transcripts of Evidence

- Dr Barry Springthorpe 7.4.03 p 264 – 272
- Dr Allan Cala 15/04/03 p 704-765.
- Prof Roger Byard 07/05/03 p1228-59
- Dr Khaira 14/04/03 p 554-9
- Dr John Cash 14/4/03 p656-60
- Dr Joseph Dezordi 9/4/03 p 445-505
- Dr Virginia Friedman 5/5/03 p 1152-6
- A/Prof John Hilton 14/4/03 p615-656; recalled 24/04/03 p 906-918; recalled 7/5/03 p1179-83
- Dr Paul Innes 15/4/03 p665-671
- Dr Owen Jones 8/5/03 p1260-1278
- Dr Alex Kane 24/04/03 p 924-932
- Dr Singh Khaira 11/4/03 p560-566
- Dr Christopher Marley 11/4/03 p 537-553
- Dr Christopher Walker 9/4/03 p471-477
- Dr Bridget Wilcken 16/4/03 p 817-823
- Dr David Cooper 14/4/03 p585-615

- Dr Dennis Seton 15/04/03 p689-699
- Prof Peter Berry 1/5/03 p 1053-1070
- Dr Susan Beale 5/5/03 p1131-1151
- Dr Barry Springthorpe 7/4/03 p 264-72

More recent reports/studies

- Pediatric Forensic Pathology: Limits and Controversies. Cordner S, Ehsani J, Bugeja L, Ibrahim J. Independent Research Studies. Roach K, Director. Queens Printer for Ontario. 2008. Commissioned by The Inquiry into Pediatric Forensic Pathology. Ontario, Canada. http://www.attorneygeneral.jus.gov.on.ca/inquiries/goudge/policy_research/pdf/Limits_and_Controversies-CORDNER.pdf
- Foundation of the Study of Infant Deaths. Care of Next Infant. Report on 10,000 babies. 2009
- Hill R (Professor) (Feb 2014) Review of the Kathleen Folbigg case. Report provided by Shaun McCarthy, Instructing Solicitor

Correspondence

- Letter from NSW State Coroner Barnes authorizing access to slides and blocks taken at the autopsies of the Folbigg children.
- Alan Cala letter to Det Ryan, 15/6/00 setting out histology material
- Assoc Prof Jo DuFlou to Shaun McCarthy, 6/01/14 confirming the availability of the histology to be examined
- Dr Allan Cala letter to Det SC Ryan, 19/6/01 (incomplete)

Standards

- National SIDS Autopsy Protocol April 1992
- Ethical Practice in Laboratory Medicine and Forensic Pathology. M El Nageh M, Linehan B, Cordner S, Wells D, McKelvie H. WHO Eastern Mediterranean Office. 1999.
- Sudden Unexpected Death in Infancy. A multi-agency protocol for care and investigation. The Royal College of Pathologists and The Royal College of Paediatrics and Child Health. 2004[1].
- Inquiry into Pediatric Forensic Pathology in Ontario. Report. Four Volumes. The Hon Stephen Goudge, Commissioner. Ontario Ministry of the Attorney General. 2008.
- Trans-Tasman Response AGAinst sudden Death in the Young (TRAGADY) 2008. Post Mortem in sudden unexpected death in the young: guidelines on autopsy practice.
- VIFM Minimum Standards: Investigation of Sudden Unexpected Death in Infancy (current)
- Gene testing for genetic heart conditions at Victorian Clinical Genetics Services. Information for referring practitioners. (As at August 2013; Appendix 2)

[1] This Protocol, produced by a working group chaired by Baroness Helena Kennedy QC, was initiated in the wake of three high profile criminal cases in the UK involving multiple infant and child deaths in the one family where convictions were subsequently overturned. The Protocol sets out in some detail the tasks of the multiple agencies involved in investigating and responding to sudden unexpected death in infancy, and how the agencies should work together.

Research undertaken for the purposes of this report

The National Coronial Information System was utilized for a number of searches as set out in further detail in the body of the report. These were undertaken on the author's behalf by Dr Colleen D'Arcy, VIFM Forensic Pathology Registrar in 2014. The detail of the audit of the forensic pathology investigations of the four deaths was also undertaken by Dr D'Arcy. The conclusions arising from the searches and the audit are the responsibility of the author of this report.

Note concerning some attributions in this report

Some material in this report has been taken from Paediatric Forensic Pathology: Limits and Controversies by Cordner S, Ehsani J, Bugeya L and Ibrahim J. This was a piece commissioned by the Inquiry into Pediatric Pathology in Ontario, also called the Goudge Inquiry, in 2008. As can be seen, I was its first author; thus I have not referenced the piece every time I have used material from it. This piece, and other material from the inquiry relevant to this case, can be seen at www.goudgeinquiry.ca/

CHAPTER 1

OVERALL APPROACH TO THIS REPORT AND ITS MAIN CONCLUSIONS:

The background to the deaths of the four Folbigg children, as provided by Professor Berry in his report available at the trial, is set out. There is then a brief discussion of child mortality, illustrating the difficulty for forensic pathologists, given the low numbers of infant deaths generally let alone intentional infant deaths, of obtaining deep experience in all aspects of the medical investigation of their deaths. This is followed by a general discussion of Sudden Unexpected Death in Infancy (SUDI) and Sudden Infant Death Syndrome (SIDS). This includes definitions and more recent categorisations of SIDS which would have been helpful had they been available prior to their publication in 2004. The forensic pathology investigations of the Folbigg deaths are compared with each other and with standards that existed in the mid 1990's and the mid 2000's.

The report then addresses the forensic pathology aspects of the trial as these are represented by the evidence of Dr Cala. (It is important to note that there were several other doctors, including pathologists, who were saying some of the same things. But it is simply too complicated, and risks losing sight of the wood for the trees, to refer to the evidence of all doctors. It is also important to note at this point that during the course of this report I try not refer to circumstantial information such as the diaries. The assessment of that information is for others).

In my view, much of the forensic pathology discussed at the trial is misconceived, based as it is on a flawed understanding of asphyxia. Asphyxia is not a helpful word in forensic pathology, is not understood in a uniform way, is not a diagnosis, and is not diagnosable. Yet the word is at the core of the main question asked repeatedly by the Prosecution: Did this child/these children suffer "an acute catastrophic asphyxiating event"? If this question was intended to be a technical question in forensic pathology, it has no content and is not capable

of an answer. Ultimately, and simply, there is no forensic pathology[2] support for the contention that any or all of these children have been killed, let alone smothered.

The report then addresses two aspects of the forensic pathology evidence which were respectively, outside forensic pathology and potentially confusing. The first aspect is the trial's apparent acceptance that the "circumstances" of these deaths are those of murder, and the forensic pathologists' acquiescence with this. This is without any established foundation, in the forensic pathology evidence, of what the circumstances were and how they were analysed, it seems to me. The second, and potentially confusing aspect is the reliance on the phrase "consistent with" in important parts of the forensic pathology evidence, and different understandings of this phrase in law and medicine.

The problem of the default diagnosis of murder is then discussed. The default diagnosis of murder came into play as an incorrect inference arising from the following question which was asked of some of the doctors: Have you ever heard of a family with three or four sudden unexplained infant or childhood deaths? The uniform answer was no, with the inference being that murder was therefore the only alternative. This default position was wrong, and was compounded by no doctor responding with: how many families are there with three, four or more surreptitious murders/smotherings of their infants and/or toddlers?[3] The fact that an infant can be smothered without leaving signs, the misunderstanding of asphyxia (in particular that it is a diagnosis and/or that it can be diagnosed), and there being no families in the literature with three or four SIDS, contributed significantly to a homicide hypothesis which in fact has little forensic pathology content.

The material above is then used to evaluate the evidence provided at the trial by Dr Wilkinson, being the main medical evidence in relation to the death of Patrick. In my view, his evidence fails to provide any medical basis for supposing that Patrick was, or might have been, killed let alone smothered; nor any medical basis for supposing that his ALTE was the consequence of a serious assault, let alone smothering.

The report's final part is an analysis leading to another view of how the Folbigg infant and toddler deaths might have occurred. When it is all pared back, the medical evidence at the trial consists of four unexplained deaths (and an acute life threatening event) in one family which in the view of the doctors were, with no actual medical evidence, likely to be four murders (and a very serious assault) by smothering. Actually, there are two unexplained deaths – not four - and an unexplained ALTE. It is the view of the author that taken in isolation, Laura's death is an uncontroversial case of death due to myocarditis. (Dr Cala was of the view that Laura died with myocarditis rather than from it. While this is possible, his reasons for concluding this do not hold water in my view). Patrick had brain pathology and seizures, which, taken in isolation, are sufficient to account for a sudden death; his acute life threatening event (ALTE) remains unexplained, although explicable. For Caleb, and Sarah, there have been considerable developments in the understanding of cardiac arrhythmias and genetics which, in the 20 or more years since, contribute to a fuller understanding of their

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[2] For the purposes of this report, I regard forensic pathology as the autopsy based medical specialty of the investigation of deaths reported to coroners.

[3] The answer to this question from my knowledge is perhaps one. The American case of Hoyt: http://en.m.wikipedia.org/wiki/Waneta_Hoyt Eventually the mother "confessed". I know of no forensic pathologist with experience of multiple homicides masquerading as SIDS. (I leave aside the case under discussion).

deaths, and possibly also to a fuller understanding of Patrick's ALTE. These medical developments are also important demonstrations of the inroads that continue to be made into further reducing the unknowns in infant deaths, inroads which will continue to be made in the future. At the very worst, one is actually left with an uncertain number of (in my view, two) infant and childhood deaths of undetermined cause, compatible with SIDS, also compatible with natural causes including cardiac arrhythmias, and an ALTE, in the one family.

In my view, it is wrong to rely on the forensic pathology evidence provided in this case to support the conclusion that one or more of the Folbigg children are the victims of a homicide. There is no merit in forcing certainty where uncertainty exists. The very existence of the enigma of SIDS demonstrates how little we know about why some babies die. It is not for a pathologist or physician to conclude that one or more of a number of infant or childhood deaths, with no significant injuries or related findings at all, are homicides on the basis of controversial circumstantial grounds. If the convictions in this case are to stand, I want to clearly state there is no pathological or medical basis for concluding homicide. The findings are perfectly compatible with natural causes. The findings cannot rule out smothering in one or more of the cases, but especially in the case of Laura, not only is there an acceptable natural cause of death easily visible microscopically, it is important that no signs of compression of the face are present.

CHAPTER II

BACKGROUND: NARRATIVE SUMMARIES OF THE BIRTH OF THE FOUR CHILDEN, THEIR MEDICAL HISTORIES, THEIR DEATHS AND THE SUBSEQUENT INVESTIGATIONS.

The following account of the deaths of Caleb, Patrick, Sarah and Laura Folbigg is taken verbatim, with minor edits, from Professor Berry's report.

Mr Folbigg was one of a family of nine children, one of whom had a child who died in infancy (but not of a genetic disorder or sudden infant death syndrome).

Craig Folbigg was working as a mobile crane operator in 1985 when he suffered a back injury and was off work receiving compensation. He met Kathy Marlborough at a nightclub and began a relationship. He learned that she was adopted and had a strained relationship with her adopted mother.

On 26th January 1986 they began to live together. In August 1986 they became engaged. At that time he was back working, and Kathy was working in a restaurant. They bought their own home and moved in in May 1987.

They were married on 5th September 1987. In 1988 Kathy became pregnant with Caleb Folbigg. In November 1988 Craig received a compensation payout from his employer with which he paid off the mortgage to their home and other debts. He then began working as a vehicle valuer.

1. Caleb Folbigg

Caleb Folbigg was born on 1st February 1989 following spontaneous onset of labour and artificial rupture of the membranes. There was meconium stained liquor and two variable decelerations with slow recovery. Delivery was by Kielland's forceps and episiotomy. His

birth weight was 3.28 kilograms. Apgar scores were 9 at one minute and 9 at five minutes. In the neonatal period he had transient tachypnoea requiring oxygen. His chest X-ray was normal. Caleb was slow to suck initially, but went home with his mother well on 5th February. Kathy used to get up at night to attend to Caleb. Craig noted that he would suck his bottle when feeding and then stop to take a breath. Kathy said that it took a little longer to feed Caleb than a normal baby as when feeding he would not breathe through his nose. He would have to stop and start feeding to catch his breath. By the time they left hospital this had improved a little bit so that it wasn't distressing him.

Caleb was seen on 17th February at two weeks of age with mild inspiratory stridor and slight costal recession, especially when upset or put on his back. His father had been concerned about his noisy breathing while feeding. His growth was on the 50th percentile. He was thought by their doctor to have congenital laryngeal stridor (floppy larynx), a common condition which resolves with time.

On 19th February 1989 he was given a feed and put down at in an adjoining room to his parents' bedroom. He was checked by Kathy Folbigg at about 10.00 – 10.30pm and was asleep in his cot. They went to bed leaving the door adjoining their bedroom and the sun room open with a lamp in the sun room on. His mother found him lifeless in his cot at 2:45am and noted him to be cold with a small amount of blood and froth around his mouth. Craig remembers waking up and hearing Kathy screaming that there was something wrong with the baby. She was standing over the cot in her pyjamas holding her hands on her forehead. When he picked Caleb up he noted that he was still warm. He commenced cardio-pulmonary resuscitation and she called an ambulance. The ambulance crew reported him as being warm and in asystole. He was pale around the nose and mouth (another ambulance report states that he was cold). After attempting further resuscitation the ambulance officers confirmed that he was dead.

(There is no material I have seen describing any scene examination in relation to this death)

Post-mortem examination

A coronial post-mortem examination was carried out the same day at 11:45am by Dr R Cummings.

External examination showed a normal baby, 3.97 kilograms, 55 centimetres long. There was posterior post-mortem staining and rigor mortis was well developed. There was no injury and the baby appeared well cared for.

Internal examination:
Normal heart weighing 25 grams.
Left lung 34 grams, right lung 53 grams with mottled pleural surfaces. The cut surfaces were moist.
The stomach contained a large quantity of curdled milk.
The liver weighed 178 grams.
The spleen weighed 15 grams and the thymus 18 grams.
Each kidney weighed 21 grams.
The brain weighed 465 grams.
There is no mention of froth or petechial haemorrhages in the post-mortem report. There is no record of any abnormality of the larynx or trachea.

Microscopic examination
The lungs showed incomplete aeration with extravasation of red blood cells and focal eosinophilic exudate.
No abnormality was described in other organs. The brain was apparently not examined histologically.
Toxicology of the liver and stomach contents was negative.

The cause of death was given as Sudden Infant Death Syndrome.

2. Patrick Folbigg

Patrick Folbigg was born on 3rd June 1990 after spontaneous onset of labour with artificial rupture of the membranes by vertex presentation at 39 weeks gestation.

The pregnancy was normal apart from an admission on 27th February 1990 at 25 weeks gestation with a three-day history of left groin pain. This was thought to be possibly due to a urinary tract infection, but the pain resolved spontaneously and she was discharged with a prescription for Amoxil (a urine culture subsequently showed a mixed growth only).

Apgar scores were 7 at one minute and 8 at five minutes. The birth weight was 3 410 grams. The placenta weighed 920 grams. His head circumference was 33.5 centimetres and crown heel length 48.3 centimetres. His mother suffered a minor perineal tear which was sutured. Her lactation was suppressed.

They went home on 8th June with Patrick being bottle-fed. They were both pleased with their new baby. He slept in a cot in the bedroom off the dining room. Craig left his job to spend all his time with his family. Again, Kathy would get up during the night to attend to Patrick because Craig was a heavy sleeper. (After about three months Craig began working again).

Sleep studies were arranged for 14th June. An electrocardiogram and serum electrolytes were normal. The sleep study was normal.

A barium swallow showed no gastro-oesophageal reflux. Contrast in the nose suggested incoordination of swallowing.

Patrick Folbigg attended his general practitioner on a number of occasions for vaccinations, mild viral infections and other childhood illnesses. However, his significant illnesses are detailed in his hospital records.

On 17th October Kathy put Patrick to bed in his cot at about 8:30pm. At about 10:30pm Craig saw Patrick lying on his back in the cot covered with a sheet and blanket. They went to bed leaving the lamp on in his room, His mother said that Patrick had been coughing at 3am when she attended to him. She was alerted at 4:30am because she heard him gasping, and noted that he was blue around the lips, lifeless, floppy, and making minimal respiratory effort. Craig woke up to the sound of Kathy screaming. He saw Kathy standing in front of the cot with the rail in the up position and Patrick lying on his back with the covers pushed down near his feet. He appeared lifeless. Craig began resuscitation and told Kathy to call an ambulance. He heard faint laboured breathing. Kathy stated that resuscitation was not performed, and that Patrick soon gave a high-pitched cry. The ambulance arrived at 04:41 about 20 minutes later and Patrick revived slightly when paramedics gave oxygen. A

paramedic recorded that the baby was having respiratory difficulties and was pale around the face and listless. The baby showed tracheal tug and intercostal recession.

On admission to hospital he was lethargic, cyanosed, and responsive only to painful stimuli. After about 15 minutes of oxygen treatment he became more alert and remained pink without high concentration oxygen. The admitting doctor noted that he was appropriately grown and arching his back. There were no signs to suggest any serious illness such as meningitis, and no evidence of trauma. Sugar in the urine in the absence of a high blood sugar with protein and blood was thought to be a response to an acute asphyxiating event. A chest X-ray was later reported to show signs which could have been due to bronchiolitis (lung fields of large volume with increased lung markings in the peri-hilar region). Virology was subsequently negative.

By the following day, he was back to his normal self. An electroencephalogram on 18th October 1990 was reported as normal. On the same day, an ECG was reported as follows: "Incomplete tracing and many leads are not labelled. Sinus rhythm at about 160/minute. Complexes appear normal on those leads that can be identified". At 9pm that day he developed a generalised seizure and was given diazepam. Following further fits he was started on phenobarbitone. He was seen by a paediatric neurologist and a lumbar puncture performed on 20th October which was normal. A metabolic screen was collected. A CT scan demonstrated hypodense areas in both temporal and occipital lobes. Phenytoin was added to his treatment because of further convulsions and acyclovir given to cover the possibility of herpes simplex encephalitis (investigations for herpes simplex were subsequently negative). He was discharged on 29th October with a diagnosis of a seizure disorder and a respiratory tract infection.

He was readmitted on 4th November with a prolonged seizure resembling an oculogyric crisis. This resolved spontaneously after 90 minutes. He was found to be febrile and to have bilateral conjunctivitis, a fine rash, and an upper respiratory tract infection. A lumbar puncture showed normal fluid, and cultures of blood and urine were negative. An eye swab yielded adenovirus. A repeat electroencephalogram showed multi-focal epileptogenic foci. Comparison with the two previous electroencephalograms showed a progressive deterioration. A further CT scan on 5th November showed generalised loss of brain substance with patchy enhancement in both occipital lobes. High-density in the pre-contrast scan was thought to be due to dystrophic calcification. These films were subsequently seen by Professor de Silva who suggested the possibility of child abuse such as shaking injury. Patrick was discharged on 10th November with the provisional diagnosis of a seizure of disorder perhaps due to an encephalopathy.

He was admitted again on a 14th November with a generalised seizure resulting in apnoea. He had an upper respiratory tract infection. It was noted that he had lost the ability to fix on a face or to follow, and he was found to have a degree of cortical blindness. A cardiac ultrasound scan on 16th November showed no evidence of intra-cardiac thrombus. On 18th November he developed gastroenteritis. Stools collected on 20th November were positive for rotavirus. He was discharged on 22nd November.

During this time Patrick was "a handful". Kathy had to give medication and physiotherapy.

On 22nd December he suffered an oculogyric crisis and was admitted to hospital again and discharged the following day.

In January 1991 he was assessed and treated in the physiotherapy department.

On 12th February 1991 he had a fever during the evening and his parents wondered whether he had a seizure at that time. He slept well and played with his father early in the morning of 13th February. Craig states he left for work at about 7.30am. Patrick appeared his normal self and was eating. At 10am that morning Kathy phoned him at work screaming "it's happened again". He drove straight home when he saw the ambulances arriving. Patrick was lying on his back in his cot with the inside rail in the up position. He again began resuscitation. Patrick was limp, blue around the lips and warm to the touch. An ambulance was called at 10.03am. On the arrival of the ambulance at 10.10am Patrick was pulseless and not breathing (the respiratory rate is recorded as zero, but the ambulance man noted shallow breathing. Another officer suggests it may be an error on the case sheet, and that no respiration was present. Another is categorical that the baby was not breathing). The baby was reported to be peripherally cyanosed with warm skin. On arrival in hospital an ECG monitor showed asystole. Despite full resuscitation no cardiac output was achieved at any stage and resuscitation was stopped after 20 minutes, death being pronounced at 10.40am on 13th February 1991.

The death was not reported to the Coroner. There was no scene examination. Craig gave authorisation for a hospital post-mortem examination.

Later he asked Kathy what had happened and she said "I put him to bed for a nap. When I went to check on him I found him how he was." She said she had put him down to sleep at about 7.30am and discovered him lifeless a couple of hours later. She then called her husband at work and also the paediatric neurologist.

Kathy Folbigg gives the following account (abstracted from statement of 23 July 1999); Patrick was an unplanned pregnancy. When he was born he had no problems with breathing and no general health problems. She thought he looked like Craig. On 18th October 1990 Patrick was around three months of age. He was sleeping in a different room which they had just done up. She had fed him as usual around 12 or 1 o'clock in the morning. She found herself awake and went to check on him on the way to the toilet. She noted his breathing was laboured to and so she put on the light finding him lethargic and unresponsive with closed eyes and trying to take a breath. She called for Craig and one of them called the ambulance. She remembers that as soon as the ambulance people put oxygen on him his eyes opened although he continued to have difficulty in breathing. One moment he was lying on the bed in the hospital unresponsive, and the next minute he was awake, screaming and panicking because of all people who were there. On the second or third day he began to have a fit in Craig's arms. From October through to Christmas they were in and out of hospital trying to control the fits. On his first birthday they were told that he was blind. He needed physiotherapy to help his development. Kathy describes herself as being on auto-pilot during this period but receiving a lot of family support.

On the day that Patrick died they followed their usual routine. Kathy put him to sleep in Caleb's old room for a morning nap. She looked into the room some time later and noticed that he was on his back which was unusual because she used to lay him on his side. He was pale and wasn't breathing. She did not remember what time this was and couldn't remember if Craig was home or not. The whole day was confused. However, she remembers that she was alone with Patrick in the house that day.

<u>Summary of metabolic and other investigations in life.</u>

Normal rectal biopsy with no neuronal inclusions.
Normal ammonia, calcium, magnesium, and glucose.
White cell enzymes were normal ruling out adrenoleukodystrophy, Refsum's disease, Zellweger's syndrome and other generalised peroxisomopathies.
Long chain fatty acid studies normal.
Urine mucopolysaccharide screen normal
Serum carnitine was normal.
Urine amino acids methylmalonic acid, organic acids and lactic acid values were normal.
Arterial blood lactate was slightly raised at 1.6 (normal 0.3-1.0) mmol per litre on one occasion.
A repeat blood lactate level was normal.
Anti-nuclear antibodies negative.
TORCH screen negative.
No leukocyte inclusions identified.

Post-mortem examination

A non-coronial post-mortem examination was carried out on 13th February 1991 at 12:30 hours.

External examination

The body was that of a normally formed a well-nourished male child weighing 8.57 kilograms, head circumference 44 centimetres, crown rump length 53 centimetres, crown heel length 77 centimetres, and foot length 10 centimetres. There was no external abnormality.

Internal examination

The skull was normal. The brain weighed 750 grams (versus 714 grams expected).

The larynx, trachea and bronchi contained frothy mucoid fluid. The right lung weighed 55 grams and the left 50 grams. Both lungs were congested posteriorly.

The heart weighed 49 grams and was structurally normal.

The thymus weighed 30 grams. It was described as enlarged. The spleen weighed 27 grams

The liver weighed 284 grams and was congested.

The pancreas weighed 15 grams and appeared normal.

The kidneys appeared normal, the right weighing 32 grams and the left 33 grams. 10 millilitres of urine were collected for metabolic studies.

The pituitary, thyroid gland, and adrenal glands were normal, the latter weighing together 6 grams.

Numerous samples were collected for microscopy, culture, toxicology, cytogenetics, metabolic studies and electron microscopy

Neuropathology

The brain weighed 750 grams after fixation. The gyri of both occipital lobes (visual cortex) were shrunken, thinner and more undulated than normal and the sulci were widened. On section, the cortical grey matter of the visual cortex in both hemispheres was thinner than normal and showed cystic degeneration, the cysts measuring 1 - 2 mm in diameter in a linear pattern at the junction of grey and white matter. Underlying white matter was firmer than normal and appeared expanded. Similar areas of firm white matter were present in the left frontal and both parietal lobes.

Microscopic examination showed no evidence of any neuronal storage disease or leukodystrophy. The major changes were old infarcts and gliosis of old laminar necrosis most severe in the parietal and occipital area. In the deeper parts of the cerebrum and in the cerebellar and brain stem nuclei there were neurones showing simple atrophy attributed to the baby's seizures. There was a slight lymphocytic infiltrate in the leptomeninges. There were no features suggestive of toxoplasmosis or cytomegalovirus, and the distribution of the lesions was unusual for herpes simplex. The appearance suggested the result of an episode of cardiorespiratory arrest that the baby suffered at about five months of age.

Microscopic examination of the lungs showed no significant abnormality apart from small foci of alveolar collapse in the periphery of the lung.

Microscope slides of the heart, skeletal muscle, liver, spleen, thymus, pancreas, kidneys, thyroid, adrenal gland, testes, and intestine showed no abnormality other than autolysis.

Results of additional post-mortem studies

Post-mortem blood cultures grew mixed organisms which were thought to reflect contamination. Cultures of lung tissue were negative for bacteria, viruses, and mycoplasma.

Phenobarbitone and carbamazapine levels were within the therapeutic range.

Investigation for primary lactic acidosis was not thought to be indicated.

Normal male karyotype.

Patrick was cremated.

The cause of death was given as:

I (a) Asphyxia due to airways obstruction (due to, or a result of)
I (b) Epileptic fits (due to, or a result of)
I (c) Encephalopathic disorder (underlying cause not determined on investigation)

3. Sarah Folbigg

Sarah Folbigg was born on 14th October 1992 following spontaneous onset of labour at 39 weeks' gestation by scan. The birth weight was 3.02 kilograms and the head circumference 34.5 centimetres. The pregnancy had been complicated by an admission for early bleeding on 21st February. This subsided spontaneously. The placenta was delivered by controlled cord traction. Apgar scores were 9 at one minute and 10 at five minutes. She was nursed on an apnoea mattress. Her mother chose to breast feed and the parents asked for early baptism.

The full neonatal examination was normal apart from mild plethora. Training in cardiopulmonary resuscitation was given and Sarah went home with an apnoea alarm on 19th October.

She was bottle fed and slept in a crib by her parents' bed. After two or three months she went into a cot in her own bedroom adjacent to the bedroom where Kathy and Craig slept. She used to snore when she was asleep. She slept with an apnoea blanket under the mattress. They found the apnoea mattress quite difficult to cope with.

When seen at 16 days of age she was 500 grams above her birth weight, and her general and neurological examination were normal.

A sleep study on 5th November showed very few sleep apnoeas and some periodic breathing. The results were judged to be normal. However, in a letter dated 16th November Dr David Cooper suggests that theophylline could be of help in view of some quite long episodes of hypoventilation.

A urine metabolic screen also on 5th November showed dicarboxylic aciduria without significant ketosis. "Further investigation is indicated if child is not on MCT containing feeds".

At four weeks of age she was developing normally.

At six weeks of age she was jittery at times but within the normal range. She appeared to be thriving and gaining weight. She was vigorous, alert, and interacted well.

When Sarah was about two and a half months old Kathy went back to work on Saturdays and Sundays for financial reasons.

When seen by her paediatrician on 21st January at the age of three months she appeared well with her weight along the 75th percentile, head circumference along the 75th percentile and length between the 50th and 75th percentiles. She was neurologically and developmentally normal. A test for MCAD deficiency was said to be normal. A further sleep study and urine metabolic screened was arranged. Her parents were described as understandably anxious, and Sarah as always wanting to be held.

At four-and-a-half months of age she was thriving and developmentally normal. When seen by her paediatrician she had a viral upper respiratory tract infection. Her mother requested a further sleep study.

She was seen five times by her general practitioner and given usual childhood vaccinations and treatment for a virus infection and a fungal skin rash. On 18th August she was prescribed flucloxacillin for a cold like illness (this was discontinued by her parents on about 26th August because of difficulty in administration) and on 26th August 1993 she was seen for a croupy cough.

Craig noted that Kathy became easily irritated and stressed with Sarah and he badgered her to "mellow out". Kathy left her job at the baby store in the middle of 1993.

On 29th August 1993 she ate normally and was put to bed in a single bed in her parents' bedroom at about 9pm. The apnoea monitor had been discontinued for about a week. At

about 9.30 or 10pm she was observed to be snoring. Her mother heard her tum over in her sleep at about 12 or 12:30am. She got up to go to the toilet at 1:30am and could not hear Sarah breathing. She turned on the bedroom light and saw that the child had a blue colour to her face and a discharge from the nose. She roused the father who commenced CPR and called an ambulance. A phone call was received at 01:25am. When an ambulance officer arrived at about 1:30am he found Craig Folbigg giving cardio-pulmonary resuscitation to Sarah on the floor. She was fully clothed in a Bond brand ski suit, blue around the mouth, and she had mucus and vomit in her mouth. She was not breathing. Full resuscitation was instituted. There was no electrical activity in her heart. The ambulance crew told her parents that she was dead. The ambulance report indicates her temperature as both normal and cold.

The child was conveyed to Maitland hospital where life was pronounced extinct at 4:30am. When a police officer attended the premises Sarah Folbigg was dressed in a yellow tracksuit, pink slippers, and wrapped in a crocheted blanket. The colour of the skin of the face and neck and hands was pale cream. He noted a small amount of semi-dried mucus material in the nostrils. An inverted U shaped mark on the bridge of the nose was thought to be consistent with that made by an oxygen mask. He observed that both parents were "genuinely very distressed".

According to the statement of Craig Folbigg, on Sunday 29th August 1993 Sarah was suffering from a cold like illness. They went out for the day and Kathy was agitated when they got home. Sarah had a bath and they had dinner at about 5:30pm when Sarah ate solids. Sarah and Craig played normally together and watched television. At 8pm Kathy took Sarah to their bedroom to put her to sleep in a single bed which they had placed in the room for her to sleep in. He had set this bed up in their bedroom because Kathy wanted Sarah out of her cot and he felt that she would be safer in the same room as them. This bed had been in that room for the past two nights and they had stopped using the apnoea blanket.
Sarah began to cry because she didn't want to go to bed which made Kathy angry and Craig heard her making growling noises. He went up to the room to inquire what the problem was and Kathy said "Nothing. Get out". He went back down to the lounge and heard Kathy return carrying Sarah in her arms. She stood about three paces in front of where he was sitting and dropped Sarah on to his lap in such a way that he had to catch her.

He settled Sarah down and put her to sleep which took about half-an-hour. Kathy got into bed while he was doing this. He put Sarah in her bed between 10:30 and 11.00pm. Kathy was either asleep or ignoring him. He awoke at about 1am and saw that Kathy was not in bed with him. He could sec by a street light that Sarah was in her bed. He thought that Kathy was in the bathroom and went back to sleep.

At about 1.30am he was woken by Kathy screaming and he sat up and saw that the light was on and Kathy was standing at the doorway of their bedroom with the door open. He saw that Sarah was lying on her back on her bed with the covers off. He began cardio-pulmonary resuscitation. Kathy was sitting on the floor in the hallway with her knees up to her face crying. He told her to call the ambulance which she did.

Kathy appeared to be devastated but managed better than Craig. A couple of days later he asked her what had happened before he woke up that morning. She said, "I had been to the toilet and just flicked on the light to check on the baby, and the rest you know." Kathy Folbigg gave the following account in her statement of 23 July 1999 (summarized); The birth of Sarah was straightforward, and they stayed in hospital for about five days. She was a good feeder and the result of a planned pregnancy. She was nicknamed the catnapper because she

would not sleep for longer than 15 or 20 minutes at a time. Kathy agrees that Sarah caused her more stress than Patrick.

The day before she died (29th August 1993) when she was about 10 months old they took her to the park and had a good day. That night they put her to sleep in a single bed in their own bedroom. The mattress was angled with pillows placed under it to face the wall. She slept for a couple of hours and then decided to get up and play in the lounge room. Kathy went to bed leaving Craig to play with her. He must then have put her to bed and gone to bed himself. (She denied having thrown the child down, but accepted the inconsistency between her account and Craig's concerning whether Sarah had gone to bed and then got up again).

Kathy got up to go to the toilet and on her return checked her and saw that she had moved. She was flat on her back with one of her arms hanging out. She was cool to the touch. She did not hear any breath sounds so she touched her arm and pulled back the covers. She woke Craig up straight away and he turned on the light. She called an ambulance while Craig attempted resuscitation. Two ambulances arrived, and events after that were pretty much a blur. She says that Sarah had a cold that day but was otherwise all right and was not on any medication. They were not using the apnoea mattress because they could not work out a way of using it in the single bed.

Following her death they were interviewed by the police.

Post-Mortem Examination

A post-mortem examination was ordered by the Coroner. This was carried out at 08.00 hours by Professor John Hilton at the New South Wales Institute of Forensic Medicine.

External examination.

The body was that of a well-nourished clean Caucasian female with minor abrading and drying of the lips. The body temperature was 25 degrees C by the rectal route at 11.00 am on 30th August. There was generalised rigor mortis. There was posterior hypostatic staining. Minor lividity was noted on the right side of the face with blanching of the left cheek and left side of the forehead. A 1.5 centimetres scratch was present on the anterolateral aspect of the right upper arm. The frenula of the lips were normal.

"There were two tiny punctate abrasions present, one immediately below the lower lip on the left side, the other slightly to the left side of the mid-line of the chin".[4]

Internal examination

The skull and membranes of the brain appeared normal. The brain was remained intact for formal neuropathology with a portion of the upper cervical cord. The middle ears were normal. Cerebrospinal fluid was clear.

The uvula was of normal size but appeared somewhat congested on its anterior surface. The epiglottis was normal. Stomach contents were present in the trachea and major bronchi. The larynx, trachea and major bronchi were otherwise normal.

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[4] Quoted from the autopsy report.

The lungs showed focal areas of collapse with a geographic pattern. Occasional petechial haemorrhages were present and there was minor congestion and minimal oedema.

The heart was normal.

The thymus showed occasional petechial haemorrhages on its surface and within the substance of the gland but was normal in size shape and location.

The stomach contained a moderate quantity of curdled material.

The liver appeared normal.

The pancreas, spleen, bone-marrow, adrenal glands, kidneys, bladder, and genital organs were normal.

No abnormality was detected in the bones, joints, or skeletal muscles.

The pathological findings were summarised as:

1. Focal pulmonary collapse
2. Modest pulmonary congestion and minimal oedema
3. Occasional petechiae on pleura, epicardium, and on and in the thymus
4. Congested haemorrhagic uvula lying anterior to the epiglottis
5. Aspiration of gastric content (?artefactual)

Subsequent microscopic examination showed marked vascular congestion of the pharyngeal aspect of the uvula.

The larynx showed a light lymphocytic inflammatory infiltrate deep to the respiratory epithelium.

Salivary gland showed two small acute inflammatory foci in the interstitium. There were no viral inclusions.

A section of diaphragm showed two foci of individual muscle fibrillary degeneration.

The spleen showed focal congestion.

The lungs showed congestion and oedema. In one section there was a light interstitial acute inflammatory infiltrate which could be seen around occasional bronchioles. A further section of lung showed multiple neutrophils within the lymphoid deposits and again some interstitial infiltration.

Examination of the brain showed no macroscopic or microscopic abnormality

Additional post-mortem studies

Lung: no virus isolated.

Lung: bacterial culture grew a mixed growth of doubtful significance.

Spleen: bacterial culture gave a moderate growth of coliforms of three types.

Large intestine contents: bacterial culture yielded no pathogen.

Small bowel contents: bacterial culture yielded no pathogen.

Biochemical analysis of vitreous humour showed a sodium of 145, chloride 133, urea 5.5 and glucose 0.4 mmol per litre.

Comprehensive screening tests for drugs and other common poisons were negative. The blood alcohol level was nil.

The cause of death was given as:

A. Sudden infant death syndrome.

Sarah was cremated.

4. Laura Folbigg

On 3rd May 1996 Craig and Kathleen Folbigg consulted Dr Christopher Seton, a staff specialist in the sleep disorders unit at the new Children's Hospital at Westmead. They were seeking advice about the risk of SIDS following the loss of their three previous children. The history they supplied was considered highly suggestive of familial clustering of obstructive apnoea. This included Mr Folbigg suffering heavy snoring as did his siblings and Patrick and Sarah Folbigg. Sarah in particular was a very loud snorer who suffered witnessed apnoea and choking episodes during sleep according to Dr Seton's letter. Dr Seton noted that Caleb was facially very similar to his mother, while Patrick and Sarah were facially similar to their father.

When Mr and Mrs Folbigg were expecting the birth of their 4th child arrangements were made for a sleep study when the child was born.

Laura Elizabeth Folbigg was born at term after an uncomplicated pregnancy on 7th August 1997 by spontaneous vaginal delivery. The placenta weighed 775 grams. Apgar scores were 9 at one minute and 10 at five minutes. The birth weight was 3.26 kilograms. She was initially breast fed for the first two weeks, but then was bottle fed.

A sleep study was carried out by Dr Seton on 19th August 1997 which demonstrated mild central apnoea and no obstructive apnoea. Laura also underwent a full biochemical, blood and metabolic investigations which were normal. In particular, urine amino acids, methylmalonic acid screen, lactate and organic acid profile were normal. She was discharged on 21st August 1997 with a home monitoring device designed to record and download breathing and heart information. They were instructed on cardio-pulmonary resuscitation and how to operate the monitor. (Laura was monitored on the machine for approximately 12 months without complication.)

Kathy started going to the gymnasium again and would give Laura to a neighbour to look after until Craig got home. He did not agree with this and she stopped. She used to leave Laura with others when she began to going to the gym regularly.

A sleep study on the 2nd October 1997 showed mild improvement. The mild central apnoea had improved and was later totally normal. Laura did not at any time show signs of suffering from obstructive apnoea which is "a potentially inherited breathing disorder associated with SIDS'.

When Laura was about three months old they moved her into her own bedroom adjacent to their bedroom. At first she slept in a cot. Kathy and Craig were sleeping in separate rooms and appeared to grow apart as they were both just living for Laura.

In March 1998 Craig Folbigg expressed concerns that his wife was not utilising the monitor as diligently as she should and wrote to Margaret Tanner a nurse consultant with the sleep study unit. "strangely though I feel that Kathy finds it all tedious and frustrating and would probably rather not use it at all, merely entrusting Laura's survival to fate! You would think that after all she had been through as a mother she of all people would be more diligent with the monitoring". The monitor was eventually returned to the new children's hospital around Laura's first birthday.

On 9th March 1998 she attended the accident and emergency department for vomiting and diarrhoea. A diagnosis of a viral gastroenteritis was made.

On 22nd June 1998 she was seen with croup following an upper respiratory tract infection over the previous two days. She was allowed home the same day after a period of observation and oxygen treatment.

Laura was seen by Dr Paul Innis her general practitioner on several occasions over seven months. On 14th August 1998 she had flu like symptoms. On 19th October 1998 she presented with a burn on her left forearm and palm which was treated over the next eight days with daily dressings.[5] On 19th January she presented with a rash which was initially thought to be allergic, but was subsequently accompanied by fever and a sore throat. On 5th February she was well and attended for her 18 month immunisation.

At about this time they purchased a single bed for Laura.

On Monday morning the 1st March 1999 Craig woke Laura up at about 6:30am. She was suffering from a runny nose and congestion of her chest. He fed her as usual and they watched
television.

Kathy got out of bed at about 7:00am and Craig began to get ready for work. Laura became upset and Kathy and Craig had an argument. Kathy became very agitated.

Craig drove to work, but at about 8.15am received a telephone call from Kathy when she apologised and Craig agreed to try harder.

At about 11am Laura and Kathy went to his place of work and she played in his office for about half an hour. Craig told them that he would be home for lunch as usual. At around lunchtime he received a phone call to go to hospital because there was something wrong with Laura. Kathy was sitting in a waiting room and Laura was lying on her back in an adjoining room being attended to by hospital staff. Kathy said "Laura fell asleep on the way home. I put

[5] I have not seen any material in relation to the circumstances of this event.

her into bed. I went to feed the dog. I had a play with the dog. I heard Laura coughing on the monitor but didn't check on her straight away. About 10 minutes later, I checked on her and found her."

An ambulance had attended at 12.14pm. Officers found Laura's mother giving cardiopulmonary resuscitation on a breakfast bar. She stated that child and had been heard coughing, and when checked approximately five minutes later was found not breathing. She said that Laura had been suffering from a runny nose and cough for a couple of days (she had been giving her Demazin (chlorpheniramine and phenylephrine) for the symptoms, last taken on 27th February). The baby was warm, blue and dressed in a pair of floral tights and a small top. There was no blood, vomit or foreign object in the child's mouth. An ECG monitor was attached and registered asystole or bradycardia (the accounts of the ambulance crews differ). The crew continued resuscitation and Laura was transferred to hospital at 12.35 pm where she was found to be cyanosed with no heartbeat, no respiration, and fixed dilated pupils. Despite full resuscitation death was declared at 12:45 pm. No bruise, mark, or other abnormality in physical appearance was found on examination. The parents were recorded as showing great anguish and anger.

Subsequent history from her mother was that Laura and had been in a bad mood, but did not appear to be seriously ill. She was last fed at 7 am. She took her to a gymnasium and to visit her father at his place of work. Laura fell asleep while travelling home in the car, and she put the sleeping child to bed in her own room at 11am, placing her on her side on top of the bed and then covering her with a woollen rug. At about 11.30am Kathleen heard the child coughing. At about 11.35am she checked on Laura and found her lying on her back and pale. She carried her to the breakfast bar in the kitchen where she rang for an ambulance and commenced cardio-pulmonary resuscitation.

Examination of the bedroom showed some small dark stains on the pillow of her bed. A screening test was positive for blood.

Kathy Folbigg gave the following account in her statement of 23 July 1999 (summarized); It was a difficult decision to have Laura. The birth itself was straightforward and she was born in good condition. They stayed in hospital for five days and she breast fed for about the first week. At about 10 days of age she spent a night in hospital undergoing sleep studies. They were given a more advanced monitor and she stayed in the bedroom with them for the first couple of months. There were many false alarms. After about three months they moved her into her own room, initially in a bassinet, and later in a cot. Kathy describes her as a good sleeper and a really good baby. At about six months of age they began to use the monitor less during the day. Kathy says this was probably more her decision then Craig's because it was she who responded to the alarms. When she had left Laura previously with friends, she did not give them the monitor.

They had a huge party for her first birthday. They were advised to discontinue using the monitor.

On the day of her death her mother took it to the gym as usual. She fell asleep in the car and her mother carried her into the house and put her to bed. She went out to check on her dogs, and around 11am about 15 to 20 minutes later she checked on Laura and found her flat on her back. She went to put her back on her side and there was no reaction. She may have been a little pale and was cool but not cold. She grabbed her and ran out into the kitchen to the breakfast bar and began resuscitation. She dialled the ambulance and continued until it

arrived. She phoned Craig at work but spoke to someone else, so that when Craig finally came home she was angry with him for having taken so long.

Post-mortem Examination

A coronial post-mortem examination was carried out at 9pm on 1st March 1999.

External examination

The body was that of a female infant consistent with the stated age of one year and eight months. The body weighed 11.52 kilograms crown heel length 80.5 centimetres, head circumference 47 centimetres, Chest circumference 49 centimetres, abdominal circumference 44.5 centimetres, and foot length 12 centimetres. The pupils were equal and measured approximately 3 mm. There was a small amount of clear fluid present at the nostrils. The lips were slightly dried and cyanosed. The frenulum was intact. The external genitalia were normal. Rigor mortis was fully developed. There was posterior lividity, and lividity was also present on the left side of the face where it was pronounced on the left cheek and left forehead.

An ovoid 5 by 3 millimetre brown bruise was present just medial to the left kneecap. An ovoid 12 by 1 0 millimetre brown bruise was noted on the right anterior lower leg. There were marks of medical intervention.

Internal examination

The scalp and skull were normal. The brain weighed 1154 grams and was saved for further examination. No retinal haemorrhages were present. The neck and cervical spine were normal.

The heart weighed 62 grams and was normal apart from an eight millimetre diameter area of haemorrhage on the posterior surface of the left atrium.

The airways were normal. The left lung weighed 122 grams and the right lung or 114 grams. Both showed focal haemorrhage and collapse on their cut surfaces.

The thymus weighed 28 grams and was normal apart from petechial haemorrhages on the anterior aspect of the suprasternal thymus.

The stomach contained a small quantity of milky-type fluid mixed with vegetable-type material. The intestines were otherwise normal.

The liver weighed 430 grams and was normal.

Each kidney weighed 36 grams. The bladder contained 10 ml of clear urine.

The spleen weighed 46 grams, Lymph nodes in the mesentery were mildly enlarged. The bone-marrow and ribs were normal.

The pituitary gland, the thyroid gland, and both adrenal glands were normal, the latter weighing two grams each.

No abnormality was found in the musculoskeletal system.

The body was re-examined on 2nd March and no additional significant findings were present. A facial dissection was carried out on 3rd March 1999 and no bruises or any other injuries were detected.

Microscopic examination

The heart showed a moderately dense infiltrate of lymphocytes with degenerate muscle cells interpreted as evidence of a viral myocarditis.

The spleen showed a markedly increased number of lymphocytes in the red pulp.

The lungs showed an increased number of lymphocytes within the interstitium and in some alveolar spaces. There were widespread areas of haemorrhage with numerous red blood cells within alveolar spaces which also contained oedema fluid, foamy macrophages, and fibrin.

Focal cortical haemorrhages were present in the thymus.

Microscope slides of the liver, kidney, stomach, oesophagus, adrenal, salivary gland, small and large intestine, thyroid, bone-marrow, pancreas, diaphragm, skeletal muscle, and ovary were all essentially normal.

Neuropathology report

The brain weighed 1307 grams after fixation. The brain stem and cerebellum weighed 11.8 per cent of the total brain weight. No macroscopic abnormality was seen in the brain or 20 cm segment of spinal cord examined.

No microscopic abnormality was seen in the multiple sections. Development was appropriate for age.

Additional post-mortem studies

Cerebrospinal fluid: no growth

Rectal swab: normal bowel flora

Lung: profuse post-mortem contaminants.

Spleen: mixed growth.

Toxicological analysis showed no alcohol in blood, and no chlorpheniramine in blood. Screening tests for various other common drugs were negative.

The cause of death was given as:

I(a) Undetermined.

CHAPTER III

CHILD MORTALITY[6] AND EXPERIENCE IN PAEDIATRIC FORENSIC PATHOLOGY

It is difficult for a forensic pathologist to obtain large experience in paediatric forensic pathology, including deaths in infancy. Nowhere in Australia is there sufficient case load to employ a full time paediatric forensic pathologist. An understanding of the paediatric forensic pathology caseload begins with an understanding of the major causes of neonatal, post-neonatal, infant, and child deaths. Death from injury is increasingly obvious as a cause of mortality with the improved survival rates from birth and reduction of early childhood diseases (e.g., infection).

To illustrate the issue, consider the situation in 2005 in Victoria with a population then of 5,022,346 people of whom 32, 606 died. Of these:

247 neonates died in the first month of their life;

82 post-neonatal/infants died between one and twelve months of their life;

43 children died aged one to four years;

34 children died aged five to nine years;

31 children died aged ten to fourteen years; and

57 children died fifteen to seventeen years.

In Victoria in 2005, of the 247 neonatal deaths, three (2.7%) resulted from causes other than natural. These were: Trauma (one) and Other (two). The natural cause category included causes such as congenital abnormality, extreme prematurity, and infection. (There was one case in this group regarded as SIDS). Of the 247, 182 had a gestational age of less than 32 weeks at birth, that is considerably premature. Very few of these deaths would be referred to the coroner and come into the field of forensic pathology.

Of (coincidentally the same number) 247 deaths of children aged 29 days to 18 years in Victoria in 2005, 47 (19%) died from unintentional injury and 7 (2.8%) from intentional trauma as set out in the table below. The remaining 193 died from other causes, overwhelmingly natural causes, including SIDS[7].

TABLE 1
Causes of post-neonatal infant and child deaths by age group
Victoria, Australia, 2005 [8]

	29-364 days	1-4 years	5-9 years	10-17 years	Total

[6] The (Victorian) Consultative Council on Obstetric and Pediatric Mortality and Morbidity, Annual Report 2005.

[7] However people use the term SIDS, smothering is not excluded on forensic pathology grounds when SIDS is the diagnosis. This overlap between natural and unnatural causes being caught in the rubric of SIDS is explicitly captured in the more recent categorization of SIDs by Krous et al (2004). See P 28ff

[8] The (Victorian) Consultative Council on Obstetric and Pediatric Mortality and Morbidity, Annual Report 2005.

Determined at birth	49	14	6	21	90
SIDS	15	-	-	-	15
Acquired Disease	12	15	17	22	66
Unintentional Injuries	4	11	8	24	47
MVA	-	6	4	16	24
Drowning	2	1	2	2	7
Asphyxiation[9]	2	-	-	-	2
Other	-	4	2	6	12
Intentional injuries	2	1	1	19	23
Intentional trauma	2	1	1	3	7
Suicide	-	-	-	16	16
Undetermined	-	2	2	2	6
Total	82	43	34	88	247

Very few of the deaths represented in the top row of the table above, deaths which were determined at birth (with a wide range of genetic and/or congenital disorders) would be referred to coroners; thus few of these deaths would be seen by forensic pathologists. Most of the acquired diseases (third row of the table) probably would also not be referred to the coroner. Clearly the number of pediatric cases, and especially deaths in infancy and the first years of childhood, that a forensic pathologist examines where the cause of death is due to intentional injury is quite small. To illustrate this issue, consider the situation at the Victorian Institute of Forensic Medicine set out in the Table below, which serves the population of Victoria. Of the 32, 606 people who died in 2005:

- approximately 5,000 were reported to the Victorian State Coroner's Office, 3,463 of whom were brought to the Victorian Institute of Forensic Medicine for medico-legal death investigation including possible post-mortem examination; (see Table below);
- 105 were under the age of 18 years, 75 of whom were under the age of 5 years;
- 3 under the age of 5 years, and a further 4 between the age of 5 and 18 years were determined to be a result of interpersonal violence; and
- 14 of the 55 deaths under the age of 1 year were either unclassified or regarded as unknown in terms of manner of death.

If the workload is shared evenly across the forensic pathologists[10], each pathologist would perform relatively few autopsies on a child and would be involved in many fewer cases of intentional injury (if any at all in a given year, especially of children under 5 years of age).

While these numbers will vary from jurisdiction to jurisdiction, we suspect the underlying message is fairly consistent: it is difficult for forensic pathologists to develop the same experience in paediatric forensic pathology as they do in adult forensic pathology.

[9] This category is used by the Consultative Committee on Obstetric and Paediatric Mortality and Morbidity. It probably represents accidental smothering.

[10] In 2014, VIFM had a complement of 10 consultant forensic pathologists and two registrars in forensic pathology.

TABLE 2
Number of medico-legal death investigations conducted
at the Victorian Institute of Forensic Medicine 2005–06 (July 1–June 30)

	<1	1-4 years	5-9 years	10-17 years	18+	Total
Natural	35	6	3	3	1890	1937
Intentional self harm	-	0	0	2	430	432
Unintentional	4	5	8	8	469	494
Interpersonal violence	2	1	2	1	65	71
Unknown	7	6	2	2	457	474
Unclassified	7	-	-	1	47	55
Total	55	18	15	17	3358	3463

CHAPTER IV

SUDDEN UNEXPECTED DEATH IN INFANCY (SUDI), SUDDEN INFANT DEATH SYNDROME (SIDS) AND ACUTE LIFE THREATENING EVENTS (ALTE'S):

SUDI refers to the unexpected death of a child between the ages of 1 month and 1 year ('post neonatal deaths')[11]. In the table above, this is represented by, at most, the 55 deaths in the first column. Based on the table preceding that, about 15 of these would have been SIDS. Two of the 55 SUDI's were regarded as being the result of homicide.

Byard[12] gives a brief historical account of SUDI, which is even more briefly summarized as follows.

There has been an awareness of SUDI since antiquity: *1 Kings 3:19 – "...and this woman's child died in the night because she overlaid it"*. Penalties were imposed for such deaths in some historical settings. This particular form of infant death, overlaying, is also found in the Bills of Mortality for the City of London in 1632.

In the 19th century, mors thymica, or thymic asthma - tracheal obstruction from an enlarged thymus[13] - was popularized. A related phenomenon named status thymo-lymphaticus became a popular explanation for sudden unexpected death in infancy. There were 820 papers on related issues in the 34 years following its publication. Boyd[14] in 1931 commented: "status thymo-lymphaticus is a good example of the growth of medical mythology, that a nucleus of truth is buried beneath a pile of intellectual rubbish, conjecture, bad observations, and rash generalization, and it is as accurate to attribute the cause of death to 'visitation by God' as to status lymphaticus".

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[11] Some authors define infancy, or the post neonatal period, as being the period from 7 days to one year.

[12] Byard R. Sudden Death in the Young. Cambridge University Press 2010. P558-9

[13] The thymus is an organ of the immune system in the upper chest just behind the upper sternum, or breast bone, of infants and young children. It involutes or shrinks as the child ages.

[14] Boyd W (1931) The pathology of internal diseases. Philadelphia, PA: Lea and Febiger, pp 675-6

The term "cot death" or "crib death" was popularized in the 1950's.

Even as recently as 2005, Krous with co-authors reported that 36 of 50 sudden unexpected deaths in infancy/childhood remained unexplained after thorough autopsies and other investigations (but not genetic testing).[15]

The current approach to SIDS owes its existence to the 1969 Stavenger definition[16]: "*The sudden death of an infant or young child which is unexpected by history and in which a thorough post-mortem examination fails to demonstrate an adequate cause of death.*"

As can be seen there are two main elements to this definition:

1. The infant seems healthy, or at least not particularly unwell, before death.
2. No cause of death could be found at a "thorough" autopsy.

It was not until 1971 that SIDS was accepted as a diagnosis on death certificates in the UK and not until 1979 that sudden infant death was given a separate coding in the WHO's International Classification of Diseases (coding number 798.0). The latter part of the 20th century saw a plethora of alternative explanations, hypotheses and theories on the possible pathogenesis of SIDS.

In 2004 Krous et al.[17] revised the general definition of Sudden Infant Death Syndrome to one which is probably now the most widely recognized definition in use today:

"*The sudden unexpected death of an infant <1 year of age, with onset of the fatal episode apparently occurring in sleep, that remains unexplained after a thorough investigation, including performance of a complete autopsy and review of the circumstances of death and the clinical history.*"

Despite its wide recognition, the categorization or classification of SIDS, which was also recommended by the authors, has not been so widely implemented. This classification is as follows:

- **Category IA SIDS (Classic features of SIDS Present and Completely Documented)**

An infant death that meets the requirements of the general definition with all of the following:
CLINICAL: Older than 21 days and under 9 months; a normal clinical history, including term pregnancy (≥37 weeks gestational age); normal growth and development; no similar deaths in

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[15] Krous, H.F., Chadwick, A.E., Crandall, L., & Nadeau-Manning, J.M. (2005). Sudden unexpected death in childhood: a report of 50 cases. Pediatric and Developmental Pathology, 8, 307-19

[16] Beckwith JB. Discussion of terminology and definition of the sudden infant death syndrome. In: Bergman AB, Beckwith JB, Ray CG, eds. Sudden infant death syndrome. Proceedings of the Second International Conference on the Causes of Sudden Death in Infants. Seattle: University of Washington Press, 1970: 14-22.

[17] Krous, H.F., Beckwith, J.B., Byard, R.W. et al. Sudden infant death syndrome and unclassified sudden infant deaths: a definitional and diagnostic approach. Pediatrics 2004;114:234–8.

siblings, close genetic relatives (uncles, aunts, and first degree cousins), or other infants in the custody of the same caregiver.
CIRCUMSTANCES: Investigation of the various scenes where incidents leading to death may have occurred and determination that they do not provide an explanation for death; found in a safe sleeping environment with no evidence of accidental death.
AUTOPSY: Absence of potentially lethal pathologic findings; minor respiratory system inflammatory infiltrates are acceptable; intrathoracic petechial haemorrhages are a supportive but not an obligatory or diagnostic finding; no evidence of unexplained trauma, abuse, neglect, or unintentional injury; no evidence of substantial thymic stress effect (ie. thymic weight less than 15g, and/or moderate to severe cortical lymphocyte depletion). Occasional "starry sky" macrophages or minor cortical depletion are acceptable; toxicology, microbiology, radiology studies, vitreous chemistry, and metabolic studies are negative.

- **Category IB SIDS (Classic features of SIDS Present but Incompletely Documented)**

An infant death that meets the requirements of the general definition and also meets all of the above criteria for Category IA except that: investigation of the various scenes were incidents leading to death may have occurred was not performed, and/or one or more of the following analyses was not performed: toxicology, microbiology, radiology, vitreous chemistry, and metabolic screening.

- **Category II SIDS**

An infant death that meets Category I criteria except for one of more of the following:
CLINICAL: Age range – outside Category IA or IB, ie. 0 to 21 days or 270-365 days; similar deaths of siblings, close relatives, or other infants in the custody of the same caregiver that are not considered suspicious for infanticide or for recognised genetic disorders; neonatal and perinatal conditions (e.g. those resulting from preterm birth) that have resolved by the time of death.
CIRCUMSTANCES: Mechanical asphyxia or suffocation by overlaying not determined with certainty.
AUTOPSY: Abnormal growth and development not thought to have contributed to death; marked inflammatory changes or abnormalities not sufficient to be unequivocal causes of death.

- **USID (Unclassified Sudden Infant Deaths)**

This includes deaths that did not meet the criteria for Category I or II SIDS, but where alternative diagnoses of natural or unnatural conditions were equivocal (including cases where autopsies have not been performed).

- **Post resuscitation cases**

Infants found in extremis who are resuscitated and later die ("temporarily interrupted SIDS) may be included in the aforementioned categories.

SUDC (Sudden unexpected death in children older than a year)

Krous et al (2004)[18] define this category of death as follows: "the sudden and unexpected death of a child older than 1 year of age which remains unexplained after a thorough

[18] Op cit 17, supra

investigation, including review of the clinical history and circumstances of death, and performance of a complete autopsy with appropriate ancillary testing."

Some features of Sudden Infant Death Syndrome (SIDS)

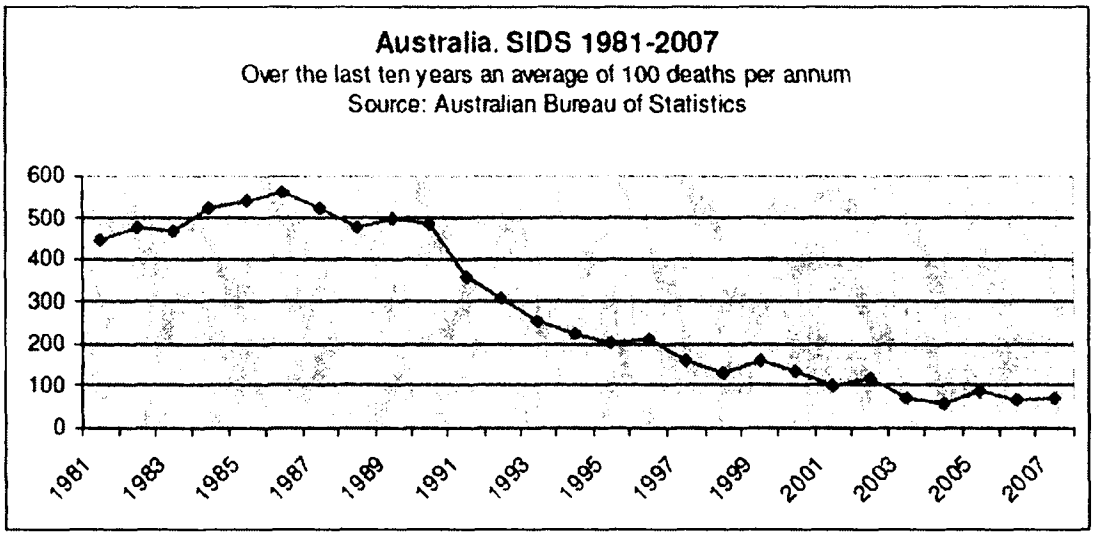
"Our understanding of the pathogenesis of SIDS is still incomplete, and this is reflected in the vast number of often contradictory papers that have been published in recent years. .... 9322 PubMed citations for "sudden infant death syndrome" listed in May 2010." (Byard, 2010)[19]

Incidence:

In Australia nationally, the incidence of SIDS was once greater than 2 deaths per 1000 live births. The rate has now dropped to well under 0.5 deaths per 1000 live births. This drop is largely attributed to risk campaigns having been undertaken resulting in many families no longer sleeping their babies on their tummies. While SIDS is an infrequent event, "it remains the most common cause of unexpected death in infants aged between one week and one year of life in Western countries" (Byard, 2010)[20]. See below for the situation in Australia.

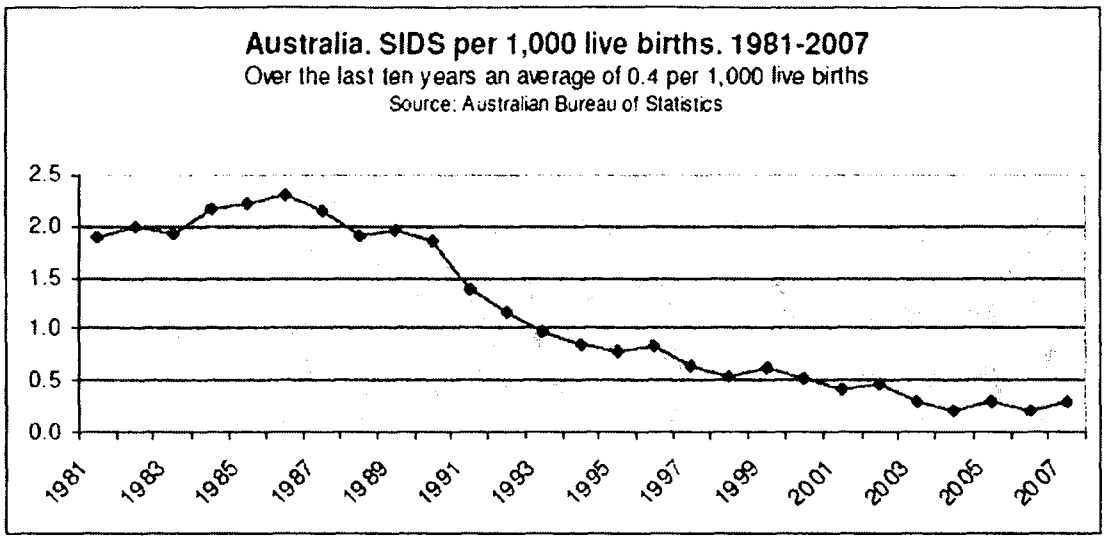
FIGURE 1

Number and rate of SIDS in Australia, 1981-2008



[19] Op cit 12, supra P560

[20] Op cit 12, supra P560



The rate may have substantially dropped, but there are still approaching 70-80 SIDS deaths a year in Australia.

Epidemiological factors associated or correlated with SIDS:

(These should be regarded as trends identified or associated with SIDS generally, and not regarded as being causative generally or in any particular case).

- Young age: 95% of deaths occur at an age of less than 6 months (peak 2-4 months)
- Males > females (Approx 55:45)
- Greater than average incidence of a history of prematurity – this is not demonstrated in all studies
- There may be a history of poor prenatal care
- Most often found in cot after sleeping; but can occur at any time of the day
- Victims often high in birth order
- History of minor respiratory of gastrointestinal illness in the days leading up to death – this is no longer a consistent finding
- Twins have a 2-4 fold increased risk of SIDS that has been attributed to low birth weight, prematurity and exposure to similar environmental factors
- Prone (ie face down) sleeping position
- Exposure to cigarette smoke before and after birth
- Poor post-natal weight gain
- Sharing a bed with a mother (as opposed to sleeping near mother, which reduces the risk)
- Mothers of SIDS infants tend to be young (<20y), have a lower socio-economic status, and to have had a number of children over a relatively short period.
- Generally more common in colder northern and southern climates

Possible mechanisms for SIDS

- Obstructive apnoea;

- anatomical abnormalities: narrowed nasal passages, shallow temporo-mandibular joints, short mandibular rami, close proximity of the soft palate to the pharyngeal walls, high position of the larynx in infants with relatively short necks predisposes to airways blockage when the tongue and mandible displace backwards; narrowness of infant aero-digestive tract is striking and could easily be aggravated by: pharyngeal hypotonia, increased secretions, submucosal oedema, tonsillar or adenoidal hyperplasia; (nb - protective effects of pacifiers)
- Neurological abnormalities – functional immaturity in autonomic control of the larynx; deficiency of neurons in the hypoglossal nucleus leading to impaired movement of the tongue;
- Physiological mechanisms – reduction in post palatal pressure causing suction of the tongue into the pharynx
- Central apnoea
- Influence of sleep state
- Possible sites for respiratory defects – brainstem nuclei, peripheral airways stretch receptors, chemoreceptors, suprapontine cerebral defects.
- Surfactant abnormality
- Evidence of chronic hypoxia
- Cardiovascular theories – defective brain stem cardiac control, autonomic imbalance, aberrant cardiac conduction pathways with a suggested terminal event being an arrhythmia rather than apnoea.

And so on, and so forth. The myriad of proposed possible mechanisms reflects the enigma of these infant deaths and how much remains unknown about the structure, detailed function and physiology of infants.

Byard (2010)[21], who has thought and written about SIDS more than most, summarizes this aspect of SIDS as follows: "Thirty years ago Luke J et al (1974)[22] commented that SIDS may well represent the final common pathway of various etiological factors, and the author would concur with this. It would appear unlikely that SIDS is a single disease entity with one cause. Rather, it seems to be the common end point for a variety of mechanisms that represent a complicated mix of predisposing factors, environmental stresses, and underlying vulnerabilities. It is also possible that different factors affect infants in particular ways, so that the population of SIDS infants may be quite heterogeneous, being composed of sub groups with unique predispositions and characteristics."

And some of these predispositions or sub groups may have genetic aspects to them. For example: "The demonstration of inherited substitutions in the first hypervariable region of the displacement loop of mitochondrial DNA in a small group of SIDS infants does raise the possibility of inherited predisposition in a certain number of victims (Arnestad M et al, 2002)[23]. This is clearly technical material. It is not a claim that there is anything as simple as a SIDS gene, however it is obvious that various forms of cardiac arrhythmias (and other diseases, including diseases yet to be discovered) could masquerade as SIDS.

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[21] Op cit 12, supra. P577

[22] Luke J, Blackbourne B and Donovan J (1974) Bed sharing deaths among victims of sudden infant death syndrome: a riddle within a conundrum. Forensic Science Gazette, 5: 3-4

[23] Arnestad M, Opdal S, Musse M, Vege A, Rognum T (2002) Are substitutions in the first hypervariable region of the mitochondrial DNA displacement-loop in SIDS due to maternal inheritance? Acta Paediatrica, 91, 1060-64

Since the Folbigg infant and childhood deaths were investigated, strong evidence has emerged of genetically determined cardiac arrhythmias capable of causing deaths indistinguishable from SIDS.

"After more than 30 years of research into the hypothesis that Long QT Syndrome (LQTS) might be a cause of arrhythmic sudden infant death, we are now at the point where we can state with certainty that some sudden unexplained deaths in infancy, about 10%, are indeed due to LQTS. The evidence for this lies in large population ECG screening programmes, post mortem molecular genetic testing of sudden infant death victims, and some informative case reports. The cardiac sodium channel gene SCN5A (LQTS Type 3) is the most common culprit, but LQTS types 1,2,6,9 and 12 have also been found. There is also new evidence that other arrhythmic syndromes sometimes cause SUDI, in particular Short QT Syndrome, and catecholaminergic polymorphic ventricular tachycardia. These conditions are also due to disordered cardiac ion channel function like LQTS, and are usually inherited in an autosomal dominant fashion. There remain however many unanswered questions, most particularly whether all populations are affected equally, and what should clinicians do with this knowledge? Should newborn ECG screening become mandatory? How should we best investigate SUDI at post-mortem in order to diagnose LQTS?"[24]

This is discussed in more detail later. This development is not only important in itself, but also as an indicator that medical science continues to make inroads into understanding infant deaths. We can expect further elucidation of new diseases and new causes of death to emerge as the years pass, and it is important, in my view, to take these currently unknown, yet to be discovered entities into account as we evaluate the Folbigg deaths. This is simply one form of acknowledgement that there is much that we do not know about why infants die.

Acute Life Threatening Events (ALTE's)

ALTE's can be regarded as falling into two categories: explained and unexplained. In this respect, they are analogous to SUDI.

".....apnoea may be a risk factor for SIDS if it takes the form of an apparent life threatening event (ALTE) in which an unexpected episode occurs that is frightening to an observer, and in which an infant is apneic for 20 seconds or longer, or where the cessation of respiration is shorter but is associated with cyanosis, pallor or bradycardia. .....infants who have had these episodes make up a very small percentage of SIDS victims (<7%) which compares with a general population frequency of 2-3 %. ALTE's may result from a wide variety of definable processes – see table – including inflicted suffocation". (Byard 2010)[25]

TABLE 3

Possible causes of an ALTE[26] (In Byard, 2010)

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[24] Skinner JR. (2010) Sudden unexplained death in infancy and Long QT Syndrome. Current Pediatric Reviews, 2010, 6, 48-55.

[25] Op cit 12, supra.

[26] Adapted from the following:
Brooks J (1998) SIDS and ALTE's. In Kendig's Disorders of the respiratory tract in children, 6th edition ed. Chernick V and Boat T. Philadelphia PA: WB Saunders, pp 1166-72
DeWolfe C (2005) Apparent life threatening event: a review. Pediatric Clinics of North America. 52: 1127-46

Cardiovascular	Arrhythmia; cardiomyopathy; congenital malformation; myocarditis; vascular rings
Respiratory	Infection; airway stenosis; obstructive sleep apnoea; breath holding episodes; laryngotracheomalacia; congenital alveolar hypoventilation; vocal cord paralysis; foreign body aspiration;
Neurological	Epilepsy; febrile convulsion; hyperekplexia; subdural haematoma; brain tumour; congenital malformation; ventriculo-peritoneal shunt malfunction; hydrocephalus; cerebral infection
Infectious	Septicemia;
Gastrointestinal	Gastro-oesophageal reflux/aspiration; pyloric stenosis; gastric volvulus; intussusception; gastroenteritis; colic; oesophageal dysfunction
Metabolic and endocrine	Hypoglycemia; hypocalcemia; hypothyroidism; hyponatremia; Reye Syndrome; carnitine deficiency; fructosemia; Leigh Syndrome;
Mechanical	Accidental mechanical asphyxia; Munchausen Syndrome by Proxy; shaken infant syndrome
Miscellaneous	Anaemia; hypothermia; anaphylaxis/allergy; medication/drug reaction
Idiopathic	Approx. 25-50% of cases

A quarter to a half of ALTE's are not explained. Those unexplained ALTE's belong in the same basket as SIDS/SUDI. Patrick's ALTE can really be thought of as SIDS/SUDI-like in character.

Multiple SIDS in one family

Although a study of multiple infant deaths within the same family by Carpenter (2005)[27] concluded that "repeat unexpected infant deaths are most probably natural", this has been criticized. Reanalysis of the data reduced the number of deaths attributed to natural causes from 87% to 43% (Bacon CJ et al, 2007)[28]. The authors of the latter paper concluded that the "likely cause of many of the second deaths could not be confidently ascertained" due to insufficient information. The Carpenter Report has also been called seriously misleading concluding that "the occurrence of two unexpected deaths in a family thus raises a definite suspicion of unnatural death which in my experience is confirmed.....in a third of cases"

Hall K and Zalman B (2005) Evaluation and Management of apparent life threatening events in children. American Family Physician, 71, 2301-8.
Kahn A, Rebuffat E, Sottiaux M, Blum D. (1988) Management of an infant with an apparent life threatening event. Pediatrician, 15: 204-11
KellyD, Shannon D. (1988). The medical management of cardiorespiratory monitoring in infantile apnea. In: Sudden Infant Death Syndrome: Medical Aspects and Psychological Management, ed. Culbertson J, Krous H, Bendell R. London. Edward Arnold, p 139-261.
Rahilly P (1991) The pneumographic and medical investigation of infants suffering apparent life threatening episodes. Journal of Paediatrics and Child Health, 27: 349-53
[27] Carpenter RG, Waite A, Coombs RC et al (2005) Repeat sudden unexpected and unexplained infant deaths: natural or unnatural? The Lancet, 365, 29-35
[28] Bacon CJ, Hey EN (2007). Uncertainty in classification of repeat sudden unexpected infant deaths in Care of Next Infant programme. British Medical Journal, 335, 129-31

(Gornall 2006)[29]. "The corollary is, of course, that there was no evidence of unnatural death in two thirds of cases" (Byard, 2010)[30]. The overall point is that as far as the research literature is concerned, more than half the subsequent deaths in families who have sustained a SIDS death are natural deaths and the remaining one third are largely unexplained, not necessarily homicides. Where is the evidence to "think dirty"[31] in families with multiple sudden unexpected and unexplained deaths in infancy?

Conclusion:

The introduction to the Kennedy Report (2004)[32] stresses the importance of remembering throughout investigations of SUDI/SIDS that:

An important starting point is the acknowledgement that in the vast majority of cases where babies suddenly die, nothing unlawful has taken place. Children are four times as likely to die in the first year of life from both natural and unnatural causes than at any other time. Parents suffering a terrible tragedy need sensitive support to help deal with their loss. It is every family's right to have their baby's death properly investigated. Families desperately want to know what happened, how the event could have occurred, what the cause of death was and whether it could have been prevented. This is important in terms of grieving, but is also relevant to a family's high anxiety about future pregnancies and may identify some hidden underlying cause, such as a genetic problem. And if there happens to be another sudden infant death in the family, carefully conducted investigations of an earlier death also help prevent miscarriages of justice.'

CHAPTER V

THE FORENSIC PATHOLOGY INVESTIGATION OF SUDI GENERALLY AND THE FOLBIGG DEATHS IN PARTICULAR.

1. Standards in the forensic pathology investigation of SUDI

Of interest, and perhaps importance, in this case is the quality of the four different forensic pathology investigations in this matter.

Rather than simply express a personal opinion, this report tries to quantify the quality of the forensic pathology investigation by reference to various standards, and by comparing the reports with each other. That is, with considerable assistance, the author has attempted to audit the forensic pathology investigations.

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[29] Gornall J. (2006) Was message of sudden infant death study misleading? British Medical Journal. 333, 1165-8

[30] Op cit 12, supra. P 569.

[31] "Think dirty" was a phrase introduced by Coroners in Toronto at the time of Dr Charles Smith, and which was roundly criticised by the Goudge Inquiry. Clearly, the objective for investigators must be to think truly.

[32] Kennedy H (Chair). Sudden Unexpected Death in Infancy. The report of a working group convened by the Royal College of Pathologists and the Royal College of Paediatrics and Child Health. The Royal College of Pathologists and The Royal College of Paediatrics and Child Health. 2004.

First, however, a brief introduction to standards may be useful. "Standards" is a general term describing a range of ways in which the quality of functions or performance are set or by which those functions or performance are measured.

Standards in medicine are generated in a number of ways including:

- By government bodies established for the purpose (eg National Pathology Accreditation Advisory Council)
- By learned medical colleges or societies
- By international organisations of repute
- By publications in recognised journals or books
- By individual organisations responsible for medical services

Forensic pathology is perhaps the smallest recognizable medical specialty in Australia with an organised basis in a specialist medical college. Because of its small size, the evolution of its standards has not been as substantial as in many larger and better resourced clinical specialties.

Standards can serve one or more of a number of purposes. Some are intended to act as guidelines in service provision, others represent to their audience a minimum standard below which the function should not fall, others are for audit purposes – a way to measure that the organisation or function is delivering, or doing what it should be doing.

There are a number of standards applicable to forensic pathology generally, and to the investigation of SUDI in particular. However, the time of their application does not overlap much with the cases here – most post-date them. Take for example a broad standard applicable to forensic pathology generally, published in 1999, in "Ethical Practice in Laboratory Medicine and Forensic Pathology".[33] It is set out in a section dealing with the duty of a forensic pathologist:

As with other health professionals, the more specific content of the forensic pathologist's duty is to exercise at least a reasonable degree of care and skill in his or her work, that is, in the production of valid and useful observations and conclusions. In assessing what is a reasonable degree of care and skill, reference can be made to the practice of colleagues of similar training and expertise. However such practice is sub-standard if it does not produce reliable and valid results. What this means in practical terms requires an understanding of the basic aims of the forensic autopsy. These are as follows:

- *To discover, describe and record all the pathological processes present in the deceased, and where necessary the identifying characteristics of the deceased.*
- *With knowledge of the medical history and circumstances of the death, to come to conclusions about the cause and time of death and factors contributing to death and, where necessary, the identity of the deceased.*
- *In situations where the circumstances of death are unknown or in question, to apply the autopsy findings and conclusions to the reconstruction of those circumstances.*

[33] El Nageh M, Linehan B, Cordner S, Wells D, McKelvie H. Ethical Practice in Laboratory Medicine and Forensic Pathology. WHO Eastern Mediterranean Office. 1999. P38

This will, on occasion, involve attendance at the scene of death, preferably with the body still in situ.

- *To record the positive and relevant negative observations and findings in such a way as to enable another forensic pathologist at another time to independently come to his or her own conclusions about the case. As forensic pathology is essentially a visual exercise, this involves a dependence on good quality and preferably colour photography.*

Other standards more applicable to SUDI particularly (set out also in the materials section at the beginning of this report) are:

- The National SIDS Autopsy Protocol from 1992
- Sudden Unexpected Death in Infancy. A multi-agency protocol for care and investigation. The Royal College of Pathologists and The Royal College of Paediatrics and Child Health. 2004.
- Trans-Tasman Response AGAinst Sudden Death in the Young (TRAGADY) 2008. Post Mortem in sudden unexpected death in the young: guidelines on autopsy practice.
- VIFM Minimum Standards: Investigation of Sudden Unexpected Death in Infancy (current)

The "Proposed minimal acceptable investigation necessary for a diagnosis of SIDS", contained in Bergman et al[34] (1970) is as follows:

"1. Adequate history
2. Gross examination, including thorax, abdomen, brain, entire larynx, and spinal cord.
3. Blood culture
4. Histological examination including: brain, heart, lungs, liver, kidneys, other organs indicated by 1 and 2 above.
5. Ancillary studies (toxicological, chemical, special cultures, virological studies, and so forth) as indicated by the results of the above.
6. Counselling of family."

This standard existed and could have been regarded as applicable to the first two Folbigg autopsies. It is likely that Dr Cumming would have been aware of this publication. One standard applicable to some of the deaths being considered here is the (Australian) National SIDS Autopsy Protocol produced in 1992. It was sponsored by the National SIDS Council and was tied to the establishment of a database of autopsies undertaken in accordance with the protocol.

2. Audit of the forensic pathology investigations of the Folbigg deaths

To try to develop a measure of the standard of the four autopsies in these deaths, with assistance from Dr Colleen D'Arcy (VIFM Forensic Pathology Registrar, 2014), a table was developed, which is attached as Appendix 1. It uses as a reference point the internal minimum standard currently in use at the Victorian Institute of Forensic Medicine for the medico-legal death investigation in circumstances of SUDI. This standard itemises the elements of the contemporary investigation that will occur, or be referenced or used in

[34] See Ref 16 supra. P18

coming to a conclusion about the cause of the death in a sudden unexpected death in infancy. We compared the 1992 National SIDS Autopsy Protocol with this, which, as expected, confirmed that even formal standards are not static. Standards in medicine generally improve as time passes. The information elements in each of the four autopsies of the Folbigg children are set out to enable comparison with these standards.

This is not presented as high science – but it does, I believe, provide one representation of the differences between the extent of the examinations and investigations in the four autopsies; and between those four examinations/investigations and the two standards, one of which was available for and applicable to the investigation of Sarah's and Laura's death. The differences are explicable in terms of expectations, available resources and knowledge changing over the time period we are considering. There is a gap in these comparisons – in the case of Patrick, for example, the number of post mortem examinations (beyond the requirements of the two standards, driven by what was regarded as the particular differential diagnosis in that case – herpes simplex encephalitis) was extensive, and is not reflected adequately in this comparison.

See Appendix 1 (**An audit of the four Folbigg autopsies against each other, the National SIDS Autopsy Protocol (1992) and the VIFM SUDI Minimum Standards (2012))** for details.

TABLE 4

OVERALL COMPARISON OF THE FOUR POST-MORTEM EXAMINATIONS, NATIONAL SIDS AUTOPSY PROTOCOL (1992) AND THE VIFM SUDI MINIMUM STANDARDS (2012).

	Number of pages in the autopsy report	Number of content elements[35] (cf those in VIFM Minimum Standard)
Caleb	**3**	**77**
Patrick	**6**	**81**
Sarah	**9**	**109**
Laura	**13**	**96**
Nat SIDS Autopsy Protocol (1992)		**122**
VIFM Minimum Standard (Current)		**194**

The table above speaks for itself. Obviously, one would expect the most recent standard to be the most demanding. An unanswerable question is what difference to the outcome would it have made if today's standard had been applied to all four of the Folbigg children's medico-legal death investigations.

A further comment should be made here. The autopsies of Caleb, Sarah and Laura were undertaken by qualified, specialist (forensic) pathologists. Patrick's autopsy was undertaken

[35] Content elements of the VIFM Minimum Standard mentioned specifically or encompassed within a broader general statement have been included in these numbers. (In the notation of the table, this means those items which have a tick or are designated with a '0').

by Dr Singh-Khaira, according to his evidence, under the supervision of Dr Bishop, the Head of his department. Also from his evidence, it seems at the time he was a qualified specialist pathologist (ie FRCPA). Even so, Chapter III applies.

3. Elements of all four death investigations missing from compliance with a contemporary standard

Below are listed some of the procedural and data elements which are required today at VIFM (as a surrogate for "today's" standards) which are apparently missing from all four death investigations[36]:

- Radiological skeletal survey (a 'babygram' showing the whole skeleton looking for fractures or underlying metabolic or other bone disease)
- CT scan (which can reveal a range of pathologies which will generally be discovered at autopsy, but can be better anticipated) but which also represents a powerful record available for later review if required.
- Whole body photography
- Intercanthal distance (the distance between the inner canthus of each eye); depth of subcutaneous fat at the chest and abdomen; signs of decomposition; state of the fontanelles; eyes: colour, icterus, petechiae, other abnormalities; specific reference to the absence of bruising, abrasion or laceration on the neck, chest, abdomen and back (rather than a general requirement in 1992 to check skin in general); anus; hyoid bone;
- Heart valve circumference; left ventricular thickness; right ventricular thickness;
- Contents of the oesophagus
- Additional investigations: mandatory histology – epiglottis, true cords, ventricular septum, bladder; bacteriology – middle ear swabs; C-reactive protein; skin for fibroblast culture; blood spot – Guthrie Card.
- EDTA tube of blood for molecular genetics screen

The Minimum Standard at VIFM for the Medico-Legal Death Investigation of SUDI also refers as follows to the issue of genetic screening:

"In the absence of a satisfactory cause of death, consideration should be given to referring the family to a paediatric cardiology service for assessment of the likelihood of an inherited abnormality of cardiac rhythm as these disorders may be fatal with no abnormality identifiable at autopsy."

In practice, pathologists refer all cases to the Pathology Liaison Nurse (PLN). (Personal Communication). The PLN:

1. In all SUDI/SIDS cases referred to her aged less than 1 year, obtains a family history; if there is something suspicious for an arrhythmia elsewhere in the family she refers for cardio-genetic follow up.

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[36] This is not to say these things should have been done (some simply were not available) but it is to say these things would be done today, and were not done in any of the four Folbigg cases.

2. In all cases >1 year with undetermined cause of death, obtains a family history, and refers all for cardio-genetic follow up.

This difference reflects the different yield on either side of this age divide – after 12 months of age, the yield is very high. [37]

This aspect of the investigation of SUDI/SIDS – a cardio-genetic follow up - is a particularly significant difference between the approach now, and the approach in the 1980's, 1990's and the early years of the last decade.

The TRAGADY (Trans-Tasman Response AGAinst sudden Death in the Young) group in 2008 developed its guidelines on autopsy practice in sudden unexpected death in the young.

"*Background.*
Inherited cardiac diseases that predispose to sudden and unexpected death in young people are being increasingly recognized and managed with life saving interventions. The impetus for this document arises from ongoing evidence of inadequate or inconsistent investigation of young sudden deaths, which results in failure to identify potentially fatal, yet treatable familial disease. The document had also been prompted by the collective experiences of family support groups in many regions, which reveal that surviving relatives find the post mortem process hard to understand and that the communications between family members and medical and legal professionals are frequently inadequate from their perspective..... ....An adequately detailed investigation of sudden death in children and young adults can identify inherited cardiac disease in more than 40% of cases.[38] *For each of these diagnosed cases, an average of 9-10 high risk relatives are identified. Increasingly, effective screening and therapy are available, which has the potential to reduce greatly the risk of future sudden deaths in this high risk group. However, the recognition of these disorders in the sudden death victim depends primarily on a detailed and thorough post mortem examination, followed by expert evaluation of first degree relatives, which may include analysis of DNA.*[39]

The overlap of SUDI/SIDS with cardio-genetics is discussed in more detail below in Chapter VIII 'How might the Folbigg children have died?' See Appendix 2: Genetic testing for genetic heart conditions at the Victorian Clinical Genetics Service. Information for referring practitioners.

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[37] The extent of follow up of relatives is resource dependent. In the TRAGADY Guidelines (see below), evaluation of first degree relatives of the deceased may extend beyond simple history taking to genetic analysis. This is not part of VIFM's follow up, but individual families may pursue this privately.

[38] Behr E, Wood DA, Wright M, Syrris P, Sheppard MN, Casey A, Davies MJ, McKenna W. Cardiological assessment of first degree relatives in sudden arrhythmic death syndrome. Lancet. 2003; 362:1457-9.
Tan HL, Hofman N, van Langen IM, van der Wal AC, Wilde AA. Sudden unexplained death: heritability and diagnostic yield of cardiological and genetic examination in surviving relatives. Circulation. 2005;112:207-13

[39] TRAGADY. Post-mortem in Sudden Unexpected Death in the Young: Guidelines on Autopsy Practice, 2008.

CHAPTER VI

THE FORENSIC PATHOLOGY EVIDENCE PROVIDED AT TRIAL ABOUT THE DEATHS

1. Issues related to the idea of asphyxia generally.

i) Introduction

Consider the following examination in chief of Dr Cala:

15.4.03 P706

Q. And what does asphyxia entail? What does it mean?
A. Asphyxia is a broad term which means any condition resulting in death whereby there is failure terminally to either adequately oxygenate tissue or failure to take carbon dioxide away from tissue.
Q. Is it in fact death from a failure to breathe?
A. Yes

Dr Cala captures some of the complexity of the word asphyxia in his answer to the first question above, but then is led into error by agreeing that asphyxia equates with a failure to breathe. Dr Cala may simply have wanted to be obliging, trying to keep things simple. But in my view this simplification underlies a misconception which is implicit throughout the trial subsequently. The misconception is that expert forensic pathology can tell whether a person has died because they stopped breathing, as opposed to having died because their heart stopped, or died from more complicated mechanisms. Forensic pathology cannot distinguish between, or identify or diagnose any of them. Forensic pathology cannot, by its work in the mortuary, point to the pathophysiological mechanisms leading to death, except perhaps in those cases where an obvious lesion is present which is incompatible with life. Even in these cases (massive haemorrhage, decapitation, ruptured heart, cerebral haemorrhage) the subsequent pathophysiological events are inferred from the incompatibility with life of the observed pathology, not concluded directly because of post-mortem findings in or on the body.

Yet, as we see repeatedly below, the prosecutor asks (and not only of Dr Cala but the other pathologists who gave evidence) whether the Folbigg children died of "an acute catastrophic asphyxiating event", meaning an acute catastrophic failure to breathe (albeit, of unknown causes). Forensic pathology as a matter of expertise cannot say; it cannot say in general, and it cannot say in these cases.[40] The answer to the question purported to be an expert forensic

[40] If a person was found dead with a piece of food impacted in and completely blocking their trachea (or windpipe), and we could be sure that it had not somehow arrived there during the process of dying, during resuscitation or after death, then we would be on good ground in concluding that their trachea had been blocked in life, and that this had therefore resulted in their death. Even in such a case, there would not be general or specific signs present (other than the piece of food) from which we could conclude that the person had died because their airway had been blocked. So, if the food bolus was completely removed prior to examination by the pathologist, there would be nothing (no general or specific signs of 'asphyxia' for example) to indicate to the pathologist how that person had died.

pathology answer, but was not an expert forensic pathology answer founded in autopsy findings; it was founded in an assessment of circumstances (which in my view was superficial), that four unexplained deaths occurred in the one family.

16.4.03 P746
Q If you yourself had conducted the post-mortem examination for Caleb, without any knowledge of what happened to the other children subsequently, if you had conducted his post-mortem what would your diagnosis have been as the cause of death?
A. Undetermined.
Q And in your view were the findings on Caleb's post-mortem examination consistent with him having been deliberately suffocated
A. Yes
Q Are you able to say whether or not Caleb died from a catastrophic asphyxiating event of unknown causes?
A. I believe that is likely.....

16.4.03 P747
Q Are you able to say whether or not (Patrick's) ALTE was consistent with him being deliberately smothered?
A. Yes
Q Was it?
A That is a possibility.
Q And are you able to say whether or not Patrick's ALTE was a result of an acute catastrophic asphyxiating event?
A. That is a possibility
Q (Looking at Patrick's death in isolation) if you had conducted the post-mortem examination on Patrick what would your diagnosis have been?
A. Undetermined
Q Are you able to say whether Patrick's death was consistent with having been caused by deliberate smothering?
A. Yes, that is a possibility.
Q. What are you able to say about whether Patrick's death was the result of an acute asphyxiating event of unknown causes?
A. I believe that is a possibility.

16.4.03 P747
Q Moving now to Sarah (looking at her death in isolation) what in your view would have been the diagnosis of her cause of death?
A. Undetermined
Q In your view, are the findings.....consistent with her having been deliberately smothered?
A Yes.....
(P749)
Q What do you say about whether or not Sarah, the third child, died of an acute catastrophic asphyxiating event?
A I believe that's a possibility.....

16.4.03 P749
Q In relation to Laura you have already told us that your diagnosis was that her cause of death was undetermined?
A Yes.

Q That it was consistent with smothering?
A Yes
Q Including deliberate smothering?
A Yes
Q And that she probably died from an acute catastrophic asphyxiating event of unknown causes?
A Yes.....

16.4.03 P749
Q Now putting those four individual children together, is this correct, that they have all died from what in your view should have been diagnosed as undetermined causes.
A Yes
Q That they all died in circumstances consistent with deliberate smothering.
A Yes
Q And that they all possibly died from an acute and catastrophic asphyxiating event of unknown causes?
A Yes.
Q Is there any natural cause of death that could account for all those four deaths and the ALTE?
A No

Some, but not all, of Dr Cala's phraseology employs words such as probable and possible. But, the repeated phrase in the prosecutor's questions - "acute catastrophic asphyxiating event" - was in my view mistakenly given oxygen. It should have been countered by: "what do you mean by an acute catastrophic asphyxiating event?; we cannot diagnose that; the pathology findings are also consistent with natural causes; there is nothing to say from a forensic pathology point of view that these children were killed". And the answer appears to be a default answer, because he did not believe there "was a natural cause of death to account for all four deaths and the ALTE". That is, because he could not explain all deaths from natural causes, the likelihood is that they are murders. As discussed later, approaching this case on that basis is a flawed approach, and the Prosecutor's question – asking for a natural cause of death to encompass all four – is based on a mistaken analysis of the four deaths.

To begin unwrapping all of this and to understand the Prosecutor, Dr Cala and others' use of the phrase "acute catastrophic asphyxiating event", we need to untangle the difficult use and meaning of the word "asphyxia". Furthermore, not only has the word been misused here, it is loosely used throughout the trial. In my view, this is a serious problem in a case such as this. And if the word is equated with "failure to breathe", and the word was intended to convey the primary or precipitating event which led to death, then it needs to be understood that this is an undiagnosable entity in forensic pathology. Forensic pathology cannot tell if the heart stops first, or breathing stops first, or they both fade away together, or some other pathophysiological cascade leads to death in any particular case.

ii) What does "asphyxia" mean ordinarily?

It is difficult to discuss "asphyxia" coherently because it encompasses a number of concepts over time in forensic pathology, it is used differently by pathologists and the common lay usage is not aligned with its technical usage. It is important in this case that this is untangled because of the emphasis placed on the word in evidence, as above for example.

The confusion starts at the beginning, with its etymology and dictionary definition. Etymologically, the word has Greek roots: the prefix "a," meaning without; and the stem of the word "sphyx," meaning pulse. One definition of "asphyxia" is given as follows[41]:

1. stoppage of the pulse[42];
2. the condition of suspended animation produced by a deficiency of oxygen in the blood; suffocation.

Asphyxiate and asphyxiation are correspondingly defined.

Suffocation is defined in the same dictionary, by reference to the verb, as

1. deprivation of air;
2. smothering;
3. killing by stopping the supply of air through the lungs;
4. the interruption of respiration in a person;
5. stifling, choking;
6. throttling (the windpipe);
7. stifling (the breath).

It is important to appreciate some other technical words that overlap with asphyxia. These include hypoxia, suboxia, and anoxia. These simply mean a low level of oxygen (hypoxia, suboxia) in the blood or person, or an absence of oxygen (anoxia) in the blood or person. Thus these technical terms correspond with the first part of the second limb of the dictionary definition above. I doubt that anyone in the street thinks of this when they use or hear the word asphyxia. Dr Cala included a form of this in his first try at telling the court what asphyxia was. (But then went on to agree with the prosecutor that it was the same as a failure to breathe).

In my view, it is the second part of the second limb of the dictionary definition of suffocation which resembles what most people think when they hear the word "asphyxia": mechanical forms of interference with respiration or breathing[43]. (This is an assertion on my part and not based on evidence, but based on excluding the other parts of the definition as really being part of the common sense of asphyxia). Numbers 1, 3, and 4 above can be, but are not necessarily, mechanical in nature and encapsulate a general mode of dying by respiratory failure (or the inhalation of fumes replacing air, or of fumes which are noxious themselves). The dictionary definition also includes some

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[41] The Shorter Oxford English Dictionary on Historical Principles. 3rd Ed. 1973, Oxford: Clarendon Press

[42] In 33 years of forensic pathology practice, I have never heard the word used in this sense. I do not believe in the exchange above the prosecutor was saying: acute catastrophic heart stoppage of unknown cause. If the prosecutor was saying this, s/he could equally have said unemotively: we do not really know why these children died.

[43] 'Mechanical' interference may sound a little strange to the layman; it means physical interference (including human interference) as distinct from, for example, the inhalation of fumes replacing air, or of toxic fumes.

mechanisms that bring to mind homicide (smothering, throttling— the latter being synonymous with manual strangulation). Smothering of course can be accidental, for example, when an infant sleeps with one or both parents who accidentally obstruct the infant's mouth and nose in one of many ways possible in such circumstances. (This is not the same as saying that all babies found dead after sleeping with their parents have been accidentally smothered. Such a death might well be due to natural causes which may or may not be diagnosable at autopsy. As I hope the reader will understand, forensic pathology usually cannot say). Fatal choking can be accidental, for example, choking on a food bolus. Deprivation of air or stifling the breath can be natural, for example, asthma by constriction of the airways and/or their occlusion by tenacious mucus.

On the dictionary test, "asphyxia", when it is regarded as a mechanical form of interference with breathing, is a non-specific term. It says nothing about a particular means of interfering with breathing and, with the exception of throttling, says nothing about the manner of its cause (i.e., natural, accidental, or homicidal). Already one can sense that for the word to be useful in a technical sense, it has to be explained and specified. This is particularly so because "asphyxia" is not a medical condition, cannot be diagnosed, and is a word which encompasses a number of disparate circumstances, medical conditions or externally imposed factors with pathological consequences to the affected individual.

iii) What does the pathologist mean when s/he uses the word "asphyxia".

The table below shows something of how the word has been used in the technical forensic pathology literature over time. A further source of confusion among lay readers/consumers of forensic pathology – as mentioned three paragraphs above, and set out in Table 5 below - is that "asphyxia" has been used by pathologists to describe the deprivation of oxygen at the level of cells and tissues, not just at the level of air entry into the body. On this basis, "asphyxia" has been regarded as a synonym of hypoxia, suboxia or anoxia—a lowered level, or absence, of oxygen in the blood. There are innumerable causes of this, most of which are natural consequences of disease states, including diseases affecting the heart, as reduced blood flow exerts its effect (at least in part) by causing hypoxia/suboxia/anoxia in the tissues.

TABLE 5

"Asphyxia": What does it mean in the forensic pathology literature?

Reference	Meaning
Black and Black, 1946. Black A, Black C. Forensic Medicine. 4th ed. 1946, London: Adam and Charles Black.	Asphyxia is a mode or mechanism of death and is the equivalent of hypoxia or anoxia
Bowden, 1949 Bowden KM, Mollison's Forensic Medicine Lectures. 5th ed. 1949, London: W. Ramsay (Surgical).	Asphyxia is the same as hypoxia or anoxia and, from a forensic point of view, the interest is in the various forms of asphyxia: "drowning, hanging, strangling, suffocation, and also asphyxia due to various poisonous gases, eg carbon monoxide"
Gonzales, Vance, Helpern and Umberger, 1954. Gonzales T et al, Legal Medicine, Pathology and Toxicology. 2nd ed.	Asphyxia occurs when oxygen transfer from air into the blood in the lungs is interfered with. This interference includes not only mechanical forms of asphyxia but also many natural

1954, New York: Appleton Century Crofts.	conditions. Asphyxia also refers to anoxia.
Smith and Fiddes, 1955 Smith S, Fiddes F, Forensic Medicine: A textbook for students and practitioners. 10th ed. 1955, London: Churchill.	Asphyxia is the same as anoxia or hypoxia. We should not attribute asphyxia to a violent cause unless the evidence of that violent cause is present.
Bowden, 1965 Bowden KM, Forensic Medicine. 2nd ed. 1965, Brisbane, Australia: The Jacaranda Press.	Asphyxia is the same as hypoxia or anoxia, and in forensic medicine we are mainly concerned with mechanical interference to the entry of air into the lungs.
Simpson, 1979 Simpson K, Forensic Medicine. 8th ed. 1979, London: Arnold.	Asphyxia is the same as mechanical asphyxia
Gordon and Shapiro, 1982 Gordon I, Shapiro HA, Forensic Medicine: A guide to principles. 2nd ed. 1982, Edinburgh: Churchill Livingstone.	"The concept that asphyxia is a pathological entity which can be recognized by certain pathological changes has led to considerable confusion in the literature on forensic medicine". Asphyxia is not a distinct pathological entity. The word is used in a variety of ways. In forensic practice it is usually intended to convey mechanical interference with respiration.
Jaffe, 1999 Jaffe FA, A Guide to Pathological Evidence for Lawyers and Policy Officers. 4th ed. 1999, Toronto: Thomson Professional Publishing.	Asphyxia signifies a terminal state of oxygen lack and not the manner in which such a state was brought about.
Dolinak, Matshes and Lew, 2005 Dolinak D, Matshes EW, and Lew EO. Forensic Pathology: Principles and Practice. 2005, New York: Elsevier Publishing.	"Technically speaking, everyone dies of asphyxia. There comes a point, arising from natural disease, injury, drug toxicity, or some combination thereof, at which blood flow to and from the brain, heart and other organs is insufficient, and terminal asphyxia is the end point of life. However in the majority of these cases, the death is not attributed to asphyxia, but rather to the underlying condition leading to a cessation of respirations."

The table shows the variations in the meaning of asphyxia. We have seen above that "asphyxia" used alone is not a helpful term in forensic medicine unless it is specified: what particular type of asphyxia are we talking about. When this happens (eg asphyxia due to compression of the neck) there is no particular contribution from the word asphyxia itself. Asphyxia is not a helpful term in technical forensic medicine. It is a non specific term for a number of entities of different kinds historically collected together.

iv) 'Asphyxia' as a diagnosis, including as a cause of death

Historically, before modern medicine, doctors would often refer only to the supposed mode of dying when completing a death certificate. The common modes were: heart failure,

respiratory (lung) failure or brain failure.[44] In old medical parlance these modes of dying were referred to, respectively, as syncope, asphyxia, and coma. Definitions 1, 3, and 4 above represent the meaning of asphyxia when it is used in this way.

Saying that someone died of respiratory failure does not advance very far the understanding of why that person died. There are very many causes of respiratory failure, or diseases that might lead to it. (Was it pneumonia: bronchial or lobar; bacterial or viral or even protozoal or fungal? Was it due to aspiration of food, or of gastric contents/vomit; or emphysema or bronchiectasis; fibrosing alveolitis; pneumothorax or a tumour compressing or obstructing the trachea or major bronchi? etc, etc). In addition, these conditions may be directly the result of a further underlying condition. It is the underlying cause of the respiratory failure that needs to be established in order to understand why the respiratory failure occurred and therefore why the person died. Clearly, before the advent of more modern understanding of pathology and medicine, the mode may have been all that was apparently understood by those caring for the dying person. And even that very superficial understanding (as it now appears in retrospect) was more apparent than real, as in fact, dying from heart failure or respiratory failure might be virtually indistinguishable and the two modes often overlap.

When "asphyxia" appears alone on the death certificate or in the cause of death on an autopsy report, unless it is accompanied by a definition, the reader cannot know what is meant by it. Is it meant as a very general mode of death, perhaps equating with hypoxia or anoxia, which by itself is completely non-specific and meaningless as a cause of death? Or is it meant to equate with mechanical asphyxia? If so, there would then have to be reference to the specific form of mechanical asphyxia for the word to be in any way useful. The specific form having been specified, there is nothing to be gained by using the word "asphyxia."

Asphyxia is not a medical condition. There is no such thing as a medical diagnosis of asphyxia. There is no constellation of findings that leads to a meaningful conclusion that a person has died of asphyxia, or even mechanical asphyxia. There are diagnoses of compression of the neck (ligature strangulation, manual strangulation, hanging), smothering, and traumatic asphyxia.

Again, what have the learned authors had to say about this?

TABLE 6

Asphyxia as a diagnosis and/or a proper term in the Cause of Death

Author	Author's interpretation
Black and Black, 1946	The general signs of asphyxia can occur in deaths from natural causes. Asphyxia is not really a cause of death unless the cause of the asphyxia is specified.
Rentoul and Smith, 1973 Rentoul E, Smith H. Eds. Glaisters Medical Jurisprudence and Toxicology. 13th ed 1973, Edinburgh. Churchill Livingstone.	Asphyxia, along with coma and syncope, alone is not acceptable as a cause of death
Gordon and Shapiro, 1982	The general signs of asphyxia, coma or syncope

[44] In contemporary practice, other modes of death include acute or chronic renal failure, liver failure, multiple organ failure, coagulopathy and septic shock. These terms, not otherwise explained are insufficient for the proper understanding of the death.

	cannot be distinguished post mortem. Asphyxia, as a general phenomenon, is not a pathological entity or recognizable disease state. It is therefore not sensible to think that it could ever be a cause of death. Since the beginning of forensic pathology, there has been a tendency to over interpret post mortem findings, both of a general and specific kind.
Jaffe, 1999	"Asphyxia without qualification is not an acceptable diagnosis".

The significance of the above is as follows.

- Asphyxia is not a useful term in forensic medicine
- It is only useful when it is further particularized – and when this is done, there is no contribution from the word asphyxia.
- If it is thought that it might loosely correspond with mechanical means of interfering with respiration, it is still of little or no technical use as there are many ways this can occur, and these different ways cause death by different mechanisms.
- There is no contribution from the term 'mechanical asphyxia' except as a time honoured term beneath which a variety of circumstances and/or conditions are collected.
- Asphyxia is not a diagnosis, mechanical asphyxia is not a diagnosis and neither is diagnosable.
- Asphyxia is a non specific term for a number of entities of different kinds historically collected together (and likewise for mechanical asphyxia).

In Vol 3 of the Report of the Inquiry into Pediatric Forensic Pathology in Ontario, Justice Goudge writes in much the same vein (P409):

"*However the evidence at this Inquiry demonstrated that the term has commonly been used to mean simply that the deceased stopped breathing or was deprived of oxygen. It has also been used frequently to denote mechanical asphyxia through the intervention of a third party. The latter meaning is radically different from the former in that it generally implies non accidental injury..... The varied meanings that can be given to the term not only invite caution in its use but present a compelling argument to avoid its use altogether, if confusion and misunderstanding are to be avoided.*"

v) Catastrophic asphyxia, acute asphyxial event or similar terms, as a cause of death or a term in autopsy reports in Australian forensic pathology.

The NCIS was searched for the following terms in the cause of death and the autopsy report itself in all deaths of children 2 years of age or less in NSW and Vic since 2000. The terms used were as follows:

- "catastrophic asphyxial event"
- "catastrophic asphyxiating event"
- "acute asphyxial event"

- "acute asphyxiating event"
- "asphyxial episode"
- "asphyxiating episode".

This search produced zero (0) cases. These terms are not used by pathologists in formulating either their autopsy reports or the cause of death.

The Prosecutor's formulation, the one used in his questions, and accepted by Dr Cala, that each of the Folbigg children died of an "acute catastrophic asphyxiating event", is a nullity. The prosecutor was using a technically meaningless phrase, and one used in questions to the other doctors as well. Whether the phrase was intended as a rhetorical flourish or ran risks of creating unjustified alarming prospects in the jury's mind is none of my business. But pointing out that it is meaningless in technical forensic pathology usage is, and the forensic pathologist(s) could, and should have avoided it.

2. "Mechanical asphyxia" and the absence of signs of smothering in the Folbigg deaths

(i) "Mechanical asphyxia" – a group of conditions historically considered together.

Mechanical asphyxia is the name given to a group of conditions historically considered together. Like asphyxia, it is not a diagnosis.

A classification of the conditions grouped under "mechanical asphyxia"

A External Forms

1. Compression of the neck
    - Hanging (where the force on the ligature is the weight of the deceased's own body, whether partially or wholly)
    - Ligature strangulation (where the force on the ligature is applied by another person, or accidentally as in a curtain cord, or very rarely, suicidally)
    - Manual strangulation (throttling)
    - Law enforcement holds (forearms across the front of the neck)
    - Other (eg by arms, knees, feet)
2. Compression/obstruction of the mouth and nose
    - Smothering
    - Plastic bag asphyxia
3. Compression of the chest (and abdomen)
    - Traumatic asphyxia (such as collapse of a sand pile or ditch, compressing the trunk)
4. Combination of two or more of the above such as where an infant becomes caught in a dangerous position between a mattress and a cot side, perhaps being a combination of 2 and 3 above.

B Internal

1. Obstruction of the larynx, laryngo-pharynx (back of the throat)
    - Choking (eg by a food bolus (when it is called a café coronary) or material inserted

into the mouth obstructing breathing
    - Postural/positional asphyxia
2. Obstruction of the trachea (windpipe)
    - Compression of the trachea (windpipe) by tumour
3. Obstruction of small airways
    - Asthma: obstruction by constriction of the airways and/or mucus
4. Obstruction of the alveoli
    - Drowning: inhalation of fluid, usually water; blood from a tumour, tuberculosis, facial fractures or mechanical intra-oral trauma.

(ii) General signs of some conditions included under "mechanical asphyxia"

"Mechanical asphyxia" is a term which gathers together various forms of mechanical obstruction to breathing, although death in the particular forms of it (eg hanging) may result from a variety of mechanisms, not simply the inability to breathe, or the deprivation of air or oxygen.

Vanezis (1989)[45] formally reviewed the general signs of many of the forms of mechanical asphyxia in his well regarded text, bearing the fruits of his MD research. The signs he regarded as relevant are: facial and conjunctival petechiae; skin petechiae above the level of a neck compression; cyanosis above the level of a neck compression. In relation to smothering, the relevant ones of these are facial and conjunctival petechiae. (The others mentioned being restricted to cases of compression of the neck. I would also add, as I think most forensic pathologists would, petechiae in the inner aspects of the lips, the mouth and the epiglottis).

In none of the four deaths is there any suggestion of the presence of any of these signs. In particular there is no suggestion of the presence of petechial haemorrhages: facial, post auricular (behind the ears), gingival (inner aspect of the lips), periorbital (around the eyes), conjunctival or epiglottic petechial haemorrhages. Neither am I aware of any material suggesting that any petechial haemorrhages were present in Patrick following the ALTE.

Traumatic asphyxia, plastic bag asphyxia and postural asphyxia are terms applied to particular forms of respiratory embarrassment. Traumatic asphyxia occurs when the chest is pinned or otherwise prevented from acting as a bellows because of the external imposition of force. For example when a car jack fails, one of the ways a person under the car might be killed is through the weight of the car pinning the chest and abdomen; or the walls of a ditch collapsing on a worker leaving the head exposed but heavily burying the trunk. Plastic bag asphyxia is the placement of a plastic bag over the head, often with the tying of the bag to prevent ingress of air. Postural asphyxia is when the person is in such a position, often aggravated by alcohol and/or drugs and/or obesity, such that there is respiratory obstruction at the level of the larynx. The mechanism of death for these three entities is different and there is therefore no reason to suppose that they would leave "general signs of asphyxia". Arriving at a diagnosis in each of the three instances is possible and in the last two it is particularly reliant on an interpretation of the circumstances. In the first, the circumstances are usually quite apparent. In none of these three entities does the word asphyxia do any real work.

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[45] Vanezis P. Pathology of Neck Injury. Butterworths. 1989. P44ff

(iii) Artefacts or mimics of inflicted injury – the abrasions to Sarah's chin.

In the four deaths there are no injuries present on the head or neck with the exception of Sarah's case.

In her case there are two small abrasions on the lower face. "There were two tiny punctate abrasions present, one immediately below the lower lip on the left side, the other slightly to the left side of the mid-line of the chin".[46] In view of the absence of any photographs, histology or other information about these abrasions, in my opinion no significance can be ascribed to them. What do they look like? Were they present before Sarah was found dead? Were they seen by the first attenders? Who saw them first? Could they be the result of resuscitation attempts? There is no objective evidence to say whether they occurred before death, around the time of death or after death.

In relation to the possibility that these abrasions were the result of resuscitation attempts, see the following discussion with Dr Cala from the transcript.

16.4.03 P753
Q. And similarly in relation to Sarah, we need to understand that in the area where the punctate marks appear the injuries to the face appear to be consistent with there being attempts to resuscitate that child?
A. Well to answer that I think I would like to know exactly as far as would be possible what resuscitation occurred, how it occurred, who gave it, what they did, before I accepted that the abrasions on her face were due to resuscitation. Because I have to say I have seen a lot of children who have been resuscitated, successfully and unsuccessfully, and most don't have marks on their face.
Q But it is common to have marks if there has been an attempted resuscitation?
A. No, it is not common to have marks.

Dr Cala is technically correct in these remarks, but the impression that is left is that the marks could not have been associated with resuscitation. If others were left with this impression, then this would have been wrong.

In the conclusion to their article, "Do resuscitation-related injuries kill infants and children?", Matshes and Lew write[47]: "*Did resuscitative efforts cause orofacial and upper airways injuries? Yes, superficial injuries of the face, mouth and upper airways could be attributed to resuscitative efforts such as attempted endotracheal intubation, facial manipulation, etc. These findings have a population prevalence of 3.4%*".

Kaplan and Fossum, in their paper "Patterns of facial resuscitation injury in infancy"[48] found that 9/25 sequentially examined infant deaths from SIDS, or histologically proven natural causes, had abrasions to the nose, face, cheeks and/or chin associated with resuscitation attempts. To the author of this opinion, this is unexceptional as desperate attempts – not always by trained emergency responders - are made to secure an airway in extreme circumstances. Resuscitation is a reasonable explanation for the abrasions on Sarah's face (if indeed they occurred around the time of death, and not on some other occasion).

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[46] Quoted from the autopsy report.
[47] Am J For Med Path. 31(2) June 2010, 178ff,
[48] Am J For Med Path. 15(3): 187-91, 1994,

(iv) Smothering <u>can</u> result in general and specific signs.

There is repeated mention throughout the trial transcripts that fatal smothering may leave no signs. While this is true, the converse is also true, which is that smothering can result in general and specific signs in infants and adults. Neither general nor specific signs were seen in any of four cases where death, on the homicide hypothesis, was due to smothering; and nor on a fifth occasion, where it is alleged an attempted homicidal smothering occurred, Patrick's ALTE. An absence of any such signs on this number of occasions might be thought to be worthy of remark.

Let us consider some of the literature.

The starting point is set out in Saukko and Knight (2004)[49]:

"*In relation to infants...it is essential to appreciate that smothering, whether intentional or accidental, is both rare and difficult to prove. The so-called classic signs of asphyxia (ie facial congestion, facial and conjunctival petechiae), for what they are worth, are rarely present in proven suffocation – and as intrathoracic petechiae are common in undoubted 'cot deaths', these signs cannot therefore be accepted in isolation as evidence of suffocation.*"

So, while facial petechiae, relatively common in adult cases of smothering, are rarely present in infants, had they been present they would almost certainly have been pointed to by Dr Cala and others as evidence in favour of smothering. Their absence therefore weighs on the other side of the balance, especially as these allegedly represent four cases of smothering (or five events of smothering if you include the ALTE suffered by Patrick). In addition, Laura is no longer an infant and the chance of facial petechiae occurring in her age group with smothering is, probably, somewhat greater. Their absence is a relevant negative tending particularly to weigh against the diagnosis in her case.

As mentioned, in none of the cases are there any conjunctival petechiae. Conjunctival haemorrhages do occur in infants in at least one form of mechanical asphyxia. Betz et al (1998) showed this in relation to manual neck compression in infants and toddlers under 2 years of age.[50] (7/7 of those who were manually strangled, 5/10 of those who were severely head injured, and 5/115 SIDS infants, all 5 of whom had undergone resuscitation, had conjunctival/eyelid petechial haemorrhages. 81/115 other SIDS deaths without petechiae had also had resuscitation.). In a study of 250 cases of SIDS, 37 natural deaths and 32 cases of lethal trauma (including 6 cases of manual strangulation), Kleeman et al (1995) found that "subconjunctival petechiae were low in density and found in only 2.4% of the SIDS group, 8.1% of the natural deaths group and 21.9% of the lethal trauma group. Subconjunctival petechiae were found at the highest density in strangulation", and in 5/6 of the strangled group[51].

The two papers appear to show that petechiae can and do occur in at least one form of paediatric mechanical asphyxia. The authors also discuss the appearance of over-inflation of

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[49] Saukko P, Knight B. Forensic Pathology. Edward Arnold. 3rd Edition. 2004 P 358

[50] Betz et al. A contribution to a possible differentiation between SIDS and asphyxiation. Forensic Science International 91 (1998) 147 – 152

[51] Kleeman WJ, Wiechern V, Schuck M, Troger HG. Intrathoracic and subconjunctival petechiae in sudden infant death syndrome (SIDS). For Sci Int 72 (1995) 49-54

the lungs as being a feature of their manual strangulation cases. Over-inflation of the lungs is not an observation I would make in such a case, although perhaps I should, and in any event it is likely to involve a degree of subjectivity. I have therefore not mentioned the absence of that finding by Dr Cala (or any of the other pathologists) as having any significance. But clearly if conjunctival or eyelid petechiae had been present, it is highly likely that Dr Cala and others would have mentioned them in support of the homicide hypothesis, especially since they are not a feature of SIDS (or, at least, not a feature of SIDS infants who have not had any resuscitation). The absence of petechiae, therefore, should be acknowledged and should be regarded as having some weight on the other side of the ledger.

(v) Deaths from smothering or compression of mouth and nose on the National Coronial Information System (NCIS) for NSW and Victoria since 2000.

One of the searches undertaken of the NCIS was for cases of smothering in NSW and Vic since 2000. A total of 17 cases were identified with "smothering" or "compression of mouth and nose" as the cause of death concluded by the forensic pathologist. Of these, six cases (35%) had no available post-mortem report accessible to analyse. The 11 cases with autopsy reports were analysed. Six (55%) cases had general and/or specific signs of smothering.

Five of the 11 cases were of less than or equal to 2 years of age. Of these five infant or childhood cases, three were female and two male, with the age ranging from 24 days to 2 years. Two of the five cases had general signs associated with smothering (such as conjunctival, facial or oral petechiae) and these two cases also had injuries suggestive of smothering or compression of the mouth such as facial or mucosal bruising or abrasions. Four of the five cases were concluded to be unintentional or accidental smothering and one was concluded to be intentional smothering[52].

These results are presented in Table 7 and demonstrate that since 2000 "smothering" or "compression of the mouth and nose" have been rarely concluded as the cause of death in children of less than or equal to two years of age in Victoria and NSW. They also demonstrate that smothering does leave signs in some infant and childhood cases, with 2/5 cases having both general and some specific signs of smothering. Reading the material in the trial in this case, you would be forgiven for thinking that such signs are more exceptional than they may be in practice. (The material from the NCIS search, although yielding very small numbers, also appears to be at odds with the quote from Saukko and Knight mentioned above). This is another reminder of the care that needs to be taken when speaking from experience, as opposed to relying on evidence, especially when the experience is not likely to be wide or deep.

(vi) What does this mean when evaluating the Folbigg children's deaths?

a) There is no forensic pathology evidence to suggest that the Folbigg children were deliberately smothered or killed.

It seems not to have been explicitly stated in the trial, but there is no forensic pathology evidence, no signs in or on the bodies, to positively suggest that the Folbigg children were smothered, or killed by any means.

[52] This is the case photographs from which are included in Appendix 3.

b) The lack of facial injuries is evidence against the conclusion of smothering.

The lack of external or deeper facial injuries is not a neutral factor in evaluating these deaths. In my view the lack of such injuries in all four cases is evidence against the conclusion of smothering as being the explanation for the four deaths. In none of the four Folbigg children, who all underwent autopsy, is there any bruising of the inside of the lips or frenulum or externally around the mouth or nose. Apparently, no reference to any such finding was made in relation to Patrick's ALTE. Quoting again from Saukko and Knight[53]:

> *"Where smothering is suspected, local signs must be sought to try to substantiate pressure on the face. Such signs include bruising around the mouth, chin and nose, though these are rarely seen except in the more violent incidents. Pressure of the lips on the teeth or dentures may cause the buccal surfaces to be bruised or abraded....It must be remembered that as small infants...have no teeth, these injuries are less likely."*

Again, in a series of four smothering deaths and one ALTE, especially where one of the deaths involved a child of 19 months (ie with teeth), the absence of any positive signs for compression of the face in all cases[54] represents a significant negative finding, which must be given due weight in any proper evaluation.

c) The lack of injuries inside the mouth is a particularly significant factor against the conclusion of smothering in Laura's case.

Appendix 3 shows photographs[55] from case 812/02, autopsied at the VIFM.[56] This two year old boy was smothered by his psychiatrically ill mother and was subject to the forced administration of drugs. Thus, the case is not completely analogous to the cases under consideration here. The autopsy findings included conjunctival petechial haemorrhages, linear facial abrasions and bruises, and mucosal abrasions and bruises

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[53] Saukko P, Knight B. Forensic Pathology. Edward Arnold. 3rd Edition, 2004. (P360)

[54] The two chin abrasions in Sarah's case I regard as neutral. They cannot be taken to represent evidence of compression of the face. See VI. 2. (iii) above.

[55] The photographs will upset some people and therefore Appendix 3 should be accessed judiciously.

[56] Burke M, Alamad S, Opeskin K. Death by smothering following forced quetiapine administration in an infant. Am J For Med Path. (2004). 25; 3:243-5

Victorian Institute of Forensic Medicine
65 Kavanagh Street Southbank Victoria, Australia 3006
MONASH University

TABLE 7

Deaths of two years of age or less with "smothering" or "compression of the mouth and nose" as the cause of death in Victoria and NSW since 2000.

Age at Death	Age Unit	Sex	Intent - Completion	Cause of Death 1a	Circumstances	General signs of smothering	Specific signs/Injuries related to smothering
24	Day(s)	F	Unintentional		Accidental smothering whilst breast feeding	Nil, circumstantial evidence only	Nil
10	Month(s)	F	Unintentional	Smothering (Accidental)	Father found child caught between cot mattress and adjustable railing. Died at Hospital.	Nil	Contusion 2cm right forehead (2cm above mid eyebrow)
5	Month(s)	M	Unintentional	Smothering	Baby found face down on pillow beside bed, underneath a wardrobe door. Had recent cold.	Nil	Nil
4	Month(s)	F	Unintentional	Smothering (Accidental)	Found face down in cot, prone and with no signs of life. Head slipped off pillow.	Petechia of forehead, malar areas and upper eyelids	Nose faint surface contusion nasal septum, nose tip and alar of left nose, red indentation left frontal head
2	Year(s)	M	Assault	Smothering	Deceased found naked, lying on cement flooring in stairwell outside residence. Mother history of schizophrenia and was found psychotic.	Petechial haemorrhages conjunctiva	Abrasions and bruises to eyelids, nose, lower/upper lip, face, buccal mucosa, right nasal bridge, cheeks, right ear, upper neck and chest, refractile material within airway

F = Female, M = Male

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inside the mouth. These last injuries were due to the pressure against the teeth. (This case is the same as the last case in Table 7).

The photographs show the injuries which can be present on the inner aspect of the mouth when pressure forces the lips against the teeth. Dr Cala was insistent that no such injuries of any type were present in Laura (who had teeth).

In relation to this, consider the following exchange with Dr Cala at the trial:

16.4.03 P752
Q. (in relation to Laura) In other words you were looking for even the most minor of bruising or injury that in fact is obviously below the skin surface?
A. Yes
Q. And again this is a very detailed process?
A. Yes
Q. And you found nothing?
A. I didn't find anything.
Q. And when you returned the following day to again examine the face on the outside again you saw nothing?
A. That's correct.
Q. You can point to nothing so far as your findings overall of Laura are concerned that can specifically be attributed to suffocation?
A. Because there are no positive findings for suffocation and my finding of no positive findings doesn't exclude suffocation. .....

The absence of such signs may very well not exclude smothering, but the first and most important conclusion in Laura's case, where one would reasonably expect such signs if she had been smothered, is that such absence is a very significant factor weighing against the diagnosis of smothering in her case.

CHAPTER VII

OTHER ASPECTS OF THE FORENSIC PATHOLOGY EVALUATION OF THE FOUR DEATHS AT THE TRIAL

15.4.03 P726
Q. You have considered all four deaths of the Folbigg children
A. Yes....
Q. Can you tell the court what the documents were that you reviewed?
A. I saw the post-mortem reports on Caleb, Patrick and Sarah and I was able to look at the medical records, that is, hospital records and GP visits and so on, and the two previous deaths that had been referred to the coroner. I examined the police statement to the coroner. [57]
Q. And doctor, what is your view about the possible cause of death for the other Folbigg children, that is, other than Laura.
A. It is my view that I suspect they died in the same way Laura Folbigg did.

[57] I imagine, if Dr Cala was examined in chief about all four cases, that he wrote a report evaluating all four deaths. I have not seen a copy of this report. It would be important to see this.

Q. And what in your view, in what way did they die
A. Well I suspect, I can't prove it medically, but I suspect that they were deliberately smothered.
Q. Are all the findings that you have seen on those four children consistent with deliberate smothering
A. Yes.
Q. Is there any explanation that you could think of that would apply to all four children, other than deliberate smothering?
A. To account for all four deaths, I don't believe there's one other entity that could account for all four deaths apart from that.....

15.4.03 P727
Q. And to what degree of suspicion would you attach your conclusion that they may have been deliberately smothered?
A. I have a high degree of suspicion that that's what happened, based on the circumstances surrounding each child's death, the essentially negative autopsy that followed each death, and therefore in combination with the circumstances and the largely negative autopsy, albeit with some caveats about Patrick's underlying condition and Caleb, who may have had an underlying laryngeal problem, that it is my suspicion that that's what happened to the four children.....
Q. Would you then suspect smothering in any unexplained death of a little baby?
A. I would suspect it until it had been excluded by a police investigation and/or the results of my autopsy.....

15.4.03 P730
Q I am asking you to identify with some precision if there is anything else (other than negative autopsies and the number of deaths) that you rely on to connect these these together to use to support that suspicion.
A. No. Beyond what I have – how I have answered and what I have said I don't believe there is any other connection.....

15.4.03 P731
Crown Pros: .....I would wish to put to him that, in effect, he is of the view that each of these children died from an unexpected catastrophic asphyxiating event of unknown origin.

1. The pathologist as an expert in the circumstances of death.

Consider these two questions from the Prosecutor:

Q Now putting those four individual children together, is this correct, that they have all died from what in your view should have been diagnosed as undetermined causes?
A Yes
Q That they all died in circumstances consistent with deliberate smothering?
A Yes

Dr Cala confirms it is his view, without obvious qualification, that all the Folbigg children "died in circumstances consistent with deliberate smothering". (See below for a discussion of the trouble that has been caused by the use of the phrase "consistent with". Its use has been effectively banned in forensic usage in Ontario, Canada.)

It is interesting to see Dr Cala being asked a question based, it seems, entirely on the circumstances. What investigation or enquiry did Dr Cala conduct into the circumstances of the four deaths? Where is there an expert report about that investigation by him? Or by anyone? What expertise did/does Dr Cala rely upon to render him an expert in the evaluation of the circumstances of infant and childhood deaths? Where is the evidence based literature to underpin such expertise? In particular, what expertise is there that enables one to conclude that specific circumstances are circumstances of multiple deliberate smotherings as opposed to multiple natural death? What evidence is there that these circumstances can be distinguished? In answering the question as he did, what were the specific circumstances relied upon by Dr Cala to render these deaths 'consistent with deliberate smothering'?

In an ordinary every day sense, a forensic pathologist acquires some general, overall familiarity with circumstances surrounding sudden unexpected death and the occurrence of injuries and related medical matters. However, when those circumstances become controversial, it soon becomes clear that pathologists may have no stronger, scientific evidence-based, foundations for evaluating these circumstances than others.

The questions and answers above, and similar questions to the other doctors, are couched in quasi-scientific terms; but they are a nullity. The pathologists should have resisted being drawn into answering them.

2. The use of the phrase 'consistent with' in forensic pathology is dangerous.

I acknowledge that I have used this phrase in giving evidence from time to time; consequently, this report may have wider implications. As a result of the consideration involved in producing this report, I am going to have to change my practice.

A simple way of thinking of the issue is to consider the following:

- Consistent with A, but inconsistent with B
- Consistent with A, but also consistent with B

In giving evidence on a particular point, one of these versions is usually meant, but most often the second half of the couplet is left unsaid. For example, 'all the findings are consistent with deliberate smothering' is not, as far as I can see, accompanied by what the findings might also be consistent with, or what they are inconsistent with. This omission may be a significant contributor to the problem with this phrase generally, and in this case in particular.

In Volume 3, P433 of the Report of the Inquiry into Pediatric Forensic Pathology in Ontario, Justice Goudge deals with the use of the phrase "consistent with" much more comprehensively.

"However, "asphyxia" is only one of a number of words and phrases that may be seriously misinterpreted or misunderstood. The phrase "consistent with" is particularly problematic. Where forensic pathologists are unable to narrow their opinions to a single cause or mechanism of death, they may indicate that the pathology is "consistent with" a particular cause or mechanism of death or a scenario presented by a questioner. Indeed, I saw instances in which Dr Smith was asked whether his findings were "consistent with" suffocation or smothering or asphyxia.

This phrase is fraught with danger. That observation, supported by the testimony of a number of forensic pathologists at this Inquiry, is hardly a new one. The danger was identified by Commissioner Fred Kaufman at the Morin Inquiry, specifically in connection with hair and fibre comparisons and generally for the forensic sciences. The following quotation he offered also resonates with the work of this Inquiry:

Bernard Robertson and G.A. Vignaux, in their book "Interpreting Evidence: Evaluating Forensic Science in the Courtroom", offer the following explanation of the difficulty with the term "consistent with": Worst of all is the word "consistent", a word in (unfortunately) common use by forensic scientists, pathologists and lawyers. To a scientist, and to a dictionary, "consistent with" is simply the opposite of "inconsistent with". The definition of "inconsistent with" is precise and narrow. Two events are inconsistent with one another if they cannot possible occur together. Thus a person cannot be in two different places at the same instant and so evidence that he was in New York at a particular instant is inconsistent with the proposition that he was in London at the same instant. Anything which is not inconsistent is consistent. Thus the proposition "several murders were committed in New York today" is quite consistent with the proposition "it rained in London today," although it may be irrelevant.

Unfortunately for clear communication, Craddock, Lamb and Moffat found that lawyers usually interpret "consistent with" as meaning "reasonably strongly supporting", while scientists use it in its strict logical and neutral meaning. When a pathologist says that certain injuries are consistent with a road accident there is no implication about whether or not there has been a road accident. It is possible the injuries could occur given the circumstances which have been described. It is therefore perfectly sensible to say that something is "consistent but unlikely". If there is some genuine dispute about the cause of the injuries what would the pathologist be able to say? He might say the injuries were consistent with either an assault or a road accident but are more likely to have occurred if there had been an assault than if there had been a road accident. If they are equally consistent with both, then they do not help us decide which of them occurred.

This example reinforces the desirability of using plain, common language that is not potentially misleading and that enhances understanding. It also supports the need to avoid specific language such as "consistent with", that is demonstrably misleading. If "consistent with" a particular cause of death means no more than "may or may not be the case", it is surely of little help. If reference must be made to this point then the pathologist must use neutral language rather than mask the opinion in language that may leave the impression that the pathology provides some support, or even strong support, for that cause of death..... .....

Recommendation 99

a) *Forensic Pathologists should avoid potentially misleading language, such as the phrase "consistent with", and adopt neutral language that clearly reflects the limitations of the opinion expressed.*
b) *Work should be done in a multi-disciplinary setting to build consensus on words and phrases that forensic pathologists should utilize or avoid as potentially misleading. The results of this work should be reflected in the Code of Practice and Performance Standards for forensic pathologists"*

.....One of the principal lessons learned at the Inquiry is that, although it is vital that forensic pathologists be highly skilled scientists, it is equally vital that they be able to communicate their opinions effectively to the criminal justice system. Improvements in the quality of forensic pathology must be paralleled by improvements in the effectiveness with which forensic pathologists are able to communicate to the criminal justice system.

The above speaks for itself and is directly applicable to the forensic pathology evidence provided in the Folbigg trial.

CHAPTER VIII

THE DEFAULT DIAGNOSIS OF MURDER IS A MISTAKE

Another important aspect of this case is implicit, not explicit. This aspect is related to the mistaken thought that all four deaths are either murder on the one hand, or a known natural cause on the other. Because the deaths were not explained, non-accidental injury became the default explanation. (Clearly this was assisted by other circumstantial information which it is not for me to assess). As mentioned early in this report, the point was made in evidence that no one has heard of a family with (three or) four such deaths; therefore, so the default corollary appears to be, these must be homicides. Nowhere in the transcripts do I see the rejoinder: how often have you seen four homicides of infants/toddlers in one family, let alone four homicides masquerading as SIDS, or SIDS like deaths.

In the Goudge Inquiry Report, Justice Goudge is critical of the approach which can be summarized as follows, (and which could be at the heart of the pathology evidence in R v Folbigg): In the absence of a credible explanation to the contrary, these children died of non-accidental injury[58]. In remarks to the Inquiry, Dr Pollanen gave the following clinical example to refute this approach:

"For example, in pathology in general, when someone goes to a surgeon with a lump.....a tumour, and the pathologist is given a biopsy of the tumour, and when we look at the section under the microscope and we're uncertain if it's cancer or not, we don't say "in the absence of evidence to the contrary this is cancer"; what we say is that the findings of the histology are not sufficient to come to a diagnosis; re-biopsy, do more investigations to find out".

Justice Goudge commented on this issue in his report as follows (Vol 3 P 415):

"Pathologists should be entitled to express their opinions if the science permits them to do so as to whether explanations given for the deceased's injuries or condition can be excluded, or conversely are supported by the pathology evidence..... But that is very different from allowing the absence of a credible explanation to serve as a substitute for pathology evidence sufficient to support a cause of death. If the evidence is insufficient to support a cause of death, the death should be characterized as undetermined. The same reasoning applies to opinions about issues other than the cause of death which may be within the forensic pathologist's expertize."

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[58] Inquiry into Pediatric Forensic Pathology in Ontario. Report. Four Volumes. The Hon Stephen Goudge, Commissioner. Ontario Ministry of the Attorney General. 2008. Vol 3. P415

It seems possible that the weight of a case such as the Folbiggs ("it is easy to smother babies and it will probably leave no signs") somehow outweighs the fact that the same pattern could occur in older children or young adults and the conclusion we would all come to would be an inherited arrhythmic disorder.

CHAPTER IX

APPLYING THE LESSONS LEARNED TO THE EVIDENCE AT TRIAL ABOUT PATRICK'S DEATH

The main evidence about Patrick's death was provided by Dr Wilkinson, the paediatric neurologist who led the management of Patrick following his ALTE. Along with Dr Cala's evidence about the death of Laura, this was the death (along with its preceding ALTE) most thoroughly examined at the trial. I have not seen any statements from any of the three hospital based pathologists involved with Patrick's autopsy.[59]

I have dealt with Dr Wilkinson's evidence by excerpting parts of it, and commenting via footnotes, in an attempt to keep the discussion flowing.

At the time of Patrick's death, Dr Wilkinson was of the view that Patrick's death was due to his epilepsy which in turn was a consequence of the brain damage from the ALTE.

In his statement of 8 October 1999, Dr Wilkinson states:

"At the time of Patrick's death (13.2.91) I saw no evidence of foul play and it did not appear necessary for the police to be notified[60]. I was satisfied with my diagnosis, however after becoming aware that a further two of Patrick's siblings have died since, I have doubt in my mind. I still believe that Patrick could have been asphyxiated[61] but I have doubts that it was the result of an epileptic fit. I must stress that I cannot positively rule out that an epileptic fit did cause the asphyxiation. Other causes of asphyxiation must now be considered in light of the other deaths in the family[62]. I would not have issued a DC if Patrick's death had been preceded by the death of his three siblings."

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[59] Of these three, I only have a transcript of the evidence given at trial of Dr Singh-Khaira.

[60] Dr Wilkinson was aware of Caleb's death from the time of his first involvement with Patrick. Not referring Patrick's death to the police (or coroner) is probably the strongest indicator of all of his state of mind about this death at the time it occurred. This experienced specialist clearly thought it was due to natural causes. To cut a long story short, considered alone, this death is reasonably thought of as being due to the epileptic consequences of the brain damage following the ALTE . This approach still leaves the ALTE unexplained.

[61] As explained above (Chapter VI), this sentence combines all of the problems with the use of the word asphyxia, with particular prominence being given to the thought that it is a diagnosable entity. The sentence has no content. If Dr Wilkinson is saying "I think Patrick was smothered" there is no evidence for this, and it is based on his interpretation of what is likely in circumstances of four deaths in one family. If Dr Wilkinson is not saying this, what is he saying?

[62] This is the stand out, single, reason why Dr Wilkinson changed his mind about the epileptic consequences of the brain damage (following the unexplained ALTE) being the reason for Patrick's death. He would have thought that whatever caused the death of the other

Dr Wilkinson seems to have initiated the autopsy which was undertaken with parental consent. The particularly important part of the autopsy discussed in evidence was the neurohistopathology report by Dr Khan, which was provided in the form of a letter, dated 24/6/91, to Dr Bishop and Dr Singh-Khaira, the joint issuers of the autopsy report.

"The major changes in this extensively sectioned brain are old infarcts and gliosis mostly in the form of old laminar necrosis which, in keeping with the macroscopic findings, is most severe in the parieto-occipital area. The only spongy change is seen in the gliotic cortical scars and the subjacent white matter in the old infarcts. The cerebellar cortex is unaffected..... In the deeper parts of the cerebrum and in the cerebellum and brain stem nuclei there are neurons showing simple atrophy. They could have resulted from this baby's epileptic seizures. In the leptomeninges there appears to be a light lymphoid infiltrate which is in addition to the small amount of residual haemopoiesis normal in this age group. This could be either non-specific and related to the cortical infarcts or related to the treated encephalitis (?assumed or proven).

I believe that the small amount of linear cortical calcification in the occipital region is just part of the laminar cortical necrosis. I can see no suggestive changes of toxoplasmosis or cytomegalovirus infection, and the distribution of lesions is unusual for herpes simplex encephalitis and they certainly appear far more likely to be the result of the episode of cardio-respiratory arrest this baby suffered at about 5 months of age."

The final diagnosis reached by Dr Bishop and Dr Singh-Khaira following Patrick's autopsy, which includes consideration of the above histology of the brain by Dr Khan, reads as follows:

"Old infarcts and gliosis in the parieto-occipital area (both cerebral hemispheres) which are probably secondary to the cardio-respiratory (sic) suffered at about five months of age".

The clinical diagnosis listed at the beginning of the autopsy report is:
"Encephalopathic disorder leading to intractable seizures. The underlying cause of the encephalopathy not determined on investigation. Asystolic cardiac arrest at home leading to death"

At the trial, Dr Wilkinson gave the following evidence:

10.4.03 P509
Q. Was that damage to Patrick's brain consistent with him having suffered from a catastrophic asphyxiating event from unknown causes?
A. Absolutely[63]
Q. If there is such damage to the brain, can that damage in turn cause seizures to develop within a few days?
A. Yes. It's a very typical sort of story that a child who's suffered some asphyxial[64] *damage to the brain may then over the next few days and weeks develop progressive change within the*

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three children probably caused the death of Patrick, and/or his ALTE. This is an understandable reaction; what is not so readily acceptable is the jump he makes to homicide. This jump falls at the 'default diagnosis is murder' hurdle. See Chapter VIII above.

[63] See the discussion of this same liturgy with Dr Cala. Chapter VI.

brain that produces seizures. So it is quite common that although the child having suffered such an event and survived it may not have seizures initially. It's quite common to find that further down the road they may have seizures..... .....That is again something I have seen in a number of situations where children have suffered various asphyxial events[65] *and subsequently developed visual problems. I believe that is because the visual part of the brain is extraordinarily sensitive to lack of oxygen..... ..... Subsequently, development of his seizures and the progressive changes on the EEG, electroencephalogram, and the changes on the CAT scan which became progressive over time too – I think that was all quite in keeping with his having suffered an asphyxial event*[66] *at the beginning of that, and then evolved over time.*
Q. Can you explain to us why it is that the seizures would normally happen a few days after the catastrophic asphyxiating event?
A. Yeah, well it's not clearly understood. I mean there is a lot of swelling that goes on in the brain as a consequence of asphyxia[67]*. That may not reach its maximum until the second, third or fourth day..... .....*

10.4.03 P511
(After having seen Patrick in the Emergency Department and participated in his attempted resuscitation)
Q. Was his appearance consistent with his having suffered a recent catastrophic asphyxiating event from an unknown cause?
A. Yes, it certainly could have been.
Q. Did you accordingly list asphyxia due to airway obstruction on Patrick's death certificate as a condition leading directly to his death[68]*?*
A. Yes I did.
Q. Did you know specifically what had caused that asphyxiation[69]*?*
A. No. I had no specific knowledge at that stage.
Q. Did you ever conclusively find what caused that asphyxiation[70]*?*
A. No we didn't. The post mortem certainly did not help us. There was no evidence of the things that might be associated with asphyxiation such as vomit..... .....
Q. In the absence of knowing what caused Patrick's asphyxiation did you form theories as to what could have caused asphyxiation?
A. Yes. I felt it was quite possible that he had had an epileptic seizure..... .....
Q. .....did you think back then that Patrick could have experienced an epileptic fit which resulted in obstruction of his airway?
A Yes, I did.
Q. Also, with that knowledge did you think that an epileptic fit could have resulted in cardiac arrest and cerebral anoxia; in other words, lack of oxygen to the brain?

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[64] The correct term to use here is "hypoxic-ischaemic damage". This is neutral as to aetiology and describes what is known about the damage, that it was caused by either or both of reduced oxygen in the blood and reduced blood supply to the region.
[65] For 'various asphyxial events' insert 'hypoxic-ischaemic damage to the brain'
[66] for 'an asphyxial event' insert 'hypoxic-ischaemic damage to the brain'
[67] For 'asphyxia' insert 'whatever caused the hypoxic-ischaemic damage to the brain'.
[68] The word asphyxia adds nothing to this sentence. Essentially Dr Wilkinson, believing Patrick had had an epileptic seizure which resulted in death, believed the epileptic fit had exerted its fatal effect by leading to obstruction of his airway.
[69] In this context it appears that asphyxiation is being used as a synonym for obstruction of the airway.
[70] Likewise, obstruction of the airway.

Yes, I did.
Q. In the absence of any other findings medically of what caused the asphyxia, did you list on the death certificate of Patrick that epileptic fits gave rise to Patrick's asphyxia?
A. Yes, I did.
Q. That was a theory at the time?
A. That was a theory at that time of his death, in advance of the knowledge of the post mortem and in advance of other knowledge as well.....

10.4.03 P514
(after considering the hospital records, the post mortem report and the neurohistopathology report)
Q After considering all those documents and with your knowledge at the time of the tests conducted in respect of Patrick, are you still of the view that the direct cause of Patrick's first emergency trip to the hospital on 18 October 1990 was a catastrophic asphyxiating event of unknown cause?
A. Yes, I am.
Q. From your experience, are you able to say whether or not smothering could have been the cause of that asphyxiation?
A. I believe it could.
Q. After considering the (the above documents) are you still of the view that the direct cause of Patrick's death later, on 13 February 1991, was a catastrophic asphyxiating event of unknown cause?
A. I believe it could have been.
Q. Why is it that you believe that?
A. The presentation at the time, at the time of his demise, I think, was in keeping with that, in that he had no breathing; no heart beat. I think that's in keeping with an asphyxia event. The fact that we found absolutely no other cause at post mortem to explain his death I think is in keeping with, consistent with, there having been an asphyxia event. There is certainly no other cause that was found that I know of at post mortem that explained his actual death.
Q. From your experience, are you able to say whether or not smothering could have caused the asphyxiation at Patrick's death?
A. Yes, I believe it could have.
Q. Was there anything in the post mortem report which points more to asphyxiation than for any other cause of Patrick's death?
A. Yes. The changes on the histopathology within – carried out by Dr Khan, had some fairly clear cut changes that I think are consistent with an asphyxial episode[71]*. He believed that at the time. In particular there are things called laminar cortical necrosis, and also the particular....*
Q. Can I just ask you to explain laminar cortical necrosis before we move on?
A. I am not a pathologist, but basically, the nerve cells in the brain are lined up in different layers, nine or so layers in the cerebral cortex. They form laminae. A lamina is a layer. By

[71] This is where the use of the word asphyxia is particularly confusing and even dangerous. In its previous two usages, the word has been used in the context of obstruction of the airway at the time of Patrick's death. The question has jumped, without any signaling, to the brain changes from the ALTE. If the former use of 'asphyxia' is intended here, then the clear implication is that the changes in the brain distinguish between obstruction of the airway at the time of Patrick's ALTE and any other cause of the brain changes; which they do not. As mentioned above the changes in the brain are best described as 'hypoxic ischaemic brain damage'.

examining the brain under the microscope you can look at the positioning of the different nerve cells and say whether that was in the lower layers or the superficial layers. Laminar necrosis is where the death appears to carry through in certain layers more than others.
Q. Is that typical of an asphyxiating event causing that?
A. I believe so. Totally so.....[72]
Q. What else in the post mortem were you going to refer to as pointing more to asphyxiation that any other cause of his death?....
A. It was also the areas of the brain that were affected at post mortem, and the occipital areas, which are the parts at the back of the brain. That, it was felt, was consistent with asphyxial episodes[73]*. That part of the brain is exquisitely sensitive to lack of oxygen.*
Q. In your experience in what other sorts of asphyxiating events would you expect to see this laminar cortical necrosis?
A. It could be any range of causes of asphyxia[74]*. I have certainly seen it amongst patients who have had drowninig episodes and things like that. Obstruction to airways for a range of causes. Suffocation from a range of causes could do it.....He (Dr Khan) commented that the distribution of the changes he found in the microscope slides was far more likely to be the result of an episode of cardiorespiratory arrest. He felt this was quite in keeping with the changes that one would see following cardiorespiratory arrest or asphyxia*[75]*.*
Q. So, does that strengthen your view that Patrick died from a catastrophic asphyxiating event?
A. Yes, but also more so that the original event was a catastrophic asphyxiating event[76]*.*
Q. In terms of the asphyxiation that you say in your view caused Patrick's death, are you able to say whether or not smothering could have caused that asphyxiation to Patrick?
A. Yes, it could have.
Q. Now, again from your experience, are you able to say whether or not you would necessarily see signs that Patrick had been smothered as opposed to experiencing some other asphyxiating event?
A. No. In a child of that age, I think it is quite possible that smothering could occur without any external visible signs.
Q. Having considered the (documents above) are you still of the view that epileptic fits led to the asphyxiation which caused Patrick's death?
A. No, I am not of that conviction any more[77]*.*

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[72] This is the same liturgy as described above with Dr Cala in Chapter VI.
[73] For 'asphyxial episodes' insert ' events causing hypoxic ischaemic damage to the brain'.
[74] For 'causes of asphyxia' insert 'events resulting in hypoxic ischaemic damage to the brain'.
[75] Dr Khan did not use the word asphyxia in his report, as submitted by letter to Dr Bishop and Dr Singh-Khaira. It may be that this answer from Dr Wilkinson is good evidence that in his mind he believes the consequences of 'asphyxia' and of 'cardio-respiratory arrest' are the same. If so, Dr Wilkinson would have no difficulty with all the substitutions I have made above, which allow a much wider range of possible causes of Patrick's brain damage than obstruction of the airway. In particular it allows for all of the possible events leading to SIDS including the whole range of genetically based cardiac arrhythmias.
[76] See the liturgy with Dr Cala in Chapter VI. Also, Dr Wilkinson is picking up on the switching, without much in the way of signaling from the barrister, between the ALTE and the death.
[77] I cannot see any basis in the exchange between the Prosecutor and Dr Wilkinson which could lead him to this conclusion. He has not discussed that part of his statement which clearly provides the basis for his change of view: the fact of four deaths. As mentioned, I

23.4.03 P860 (Cross examination; after a delay)
Q. And is it your opinion that (Patrick) did not die of an epileptic seizure?
A. I can't say that is impossible. It is a possibility.
Q. Is this possible, that he suffered from epileptic seizures which caused asphyxia which caused damage to the brain. Is that a possible sequence?.....
A. Yes, that is a possibility..... ...
(P865)
Q. Did (the post mortem report) have some effect on your thinking about the cause of this child's death?
A. Yes, it did.
Q. What effect was that?
A In two senses. There was nothing that indicated obstruction of the airways that one might perhaps have seen if he had had an epileptic seizure, vomited, inhaled vomitus, and that is one of the mechanisms of death associated with epileptic fits. There was no evidence of that. There was no evidence at post mortem that his tongue had obstructed his airway[78]*, so those were things which I might have thought, you know, possible at the post mortem which weren't there. But in a different sense there were changes in the brain, and Dr Khan, and this was given in evidence when I was first there, there were changes in the brain which certainly made my thinking very different. Those changes were consistent with an initial asphyxial episode and certainly in no way consistent with herpes simplex or an encephalitis*[79]*..... .....*

(P866-70 Dr Wilkinson agrees, in a letter written some months after Patrick's death to his parents, that he accepted that encephalitis may have been the cause of Patrick's ALTE, and therefore, ultimately, of his death).

23.4.03 P871
Q. ....It is possible isn't it, that epilepsy can cause sudden death?
A. It can do.
Q. Is there a phenomenon called SUDEP?
A. Yes there is.
Q. What does it mean?
A. Sudden unexpected death in an epileptic patient
Q. Could you describe what the biological process of that is, if you can?
A. Well I don't think anyone knows and there may be many causes for such unexpected deaths ranging from, as I said earlier, episodes of seizure leading to vomiting, leading to inhaling vomit and suffocating, seizure resulting in the tongue, I think the colloquial phrase

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think this is reasonable, but what is not correct is the immediate jump to a homicide hypothesis.

[78] This is an interesting example of a clinician interpreting an autopsy. Upper airway obstruction by the tongue is a real phenomenon in a number of circumstances (seizures, unconsciousness from any cause) causing respiratory embarrassment. But it is never visualized at autopsy because post mortem relaxation of muscles and movement of the body mean that such structures do not remain in place for the pathologist to see. This conclusion is always an inference in forensic pathology. So, the fact this was not seen cannot be used as evidence that it was not there. (This can surely be understood in a case where so much hinges on smothering leaving no signs!!). Upper airways obstruction as a result of a seizure cannot be ruled out, and remains as an entirely plausible mechanism for Patrick's death.

[79] For 'asphyxial episode' insert 'episode causing hypoxic-ischaemic brain damage'.

is "being swallowed", but the tongue can certainly obstruct the airway in an epileptic event. A patient may roll over and put their head into a pillow. There are other possible causes such as cardiac rhythm disturbances during epileptic seizures. I think it is fair to say that a lot of it is speculation as to what the cause of death is. Unless there is particular evidence found it has been my experience that often one just knows that the child was an epileptic and the child was found dead without any more clear evidence than that.

23.4.03 P874 (Re-examination)
Q. Was it (the damage to the brain) consistent with asphyxiation?
A. Yes, it was. Certainly[80]*.....*
Q (The pattern of seizures, their onset) is that consistent with asphyxiation?
A. Yes, I believe it is.....
Q (Progressive changes in the CAT scan) Is it consistent with asphyxiation?
A. Yes, I believe it is.....
(P876) Q. Putting all of those together (ten reasons why Patrick did not have herpes simplex encephalitis as the cause of his ALTE) are you now able to exclude encephalitis as a possible cause of Patrick's admission when he first came to hospital?
A. Yes, I can.
Q. And what do you now say is the most likely cause of the first admission to hospital?
A. I think the most likely cause was asphyxia.
Q. And what does asphyxia mean?
A. Asphyxia is a situation where the end result is that the blood cannot deliver oxygen to the tissues and that may be as a result of a number of issues. It would be as a result of just obstructing the passage of air and oxygen into the lungs, it can be other situations, carbon monoxide poisoning where the oxygen can't be carried, but I think asphyxia is most commonly the result of oxygen not getting into the body[81]*.....*
(P883)
Q. One further matter doctor. You have used the term hypoxia. Would you tell the court what that means?
A. Hypoxia means lack of oxygen, effectively. The hypoxia is a process whereby the blood is not able to carry enough oxygen to look after tissues and hypoxia results in changes of a damaging type in the tissues.
Q. Can hypoxia be caused by deliberate smothering?
A. It can[82]*.*

The evidence above is the main evidence provided in relation to Patrick's death. In so far as it relates to understanding the cause of Patrick's ALTE and death, it is confusing and potentially misleading. This has no doubt occurred through no conscious intent on Dr Wilkinson's part. The problems are as follows:

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[80] Liturgy discussed above, and repeated in next few questions and answers.
[81] This exchange is a manifestation of the confusion about the word asphyxia. Specifically, that asphyxia is diagnosable and that the term can be taken to mean there is a very good chance that there has been obstruction to breathing. As set out in Chapter VI all this is misleading, and this sort of exchange is devoid of technically reliable medical content.
[82] I suppose this could just be a rhetorical prosecutorial flourish – but ultimately everyone dies of hypoxia. This exchange really was "Can death be caused by smothering?". "Yes, it can".

1. All of the issues set out in Chapter VI in relation to "asphyxia" are played out in Dr Wilkinson's evidence. Once it is understood that asphyxia is not a meaningful entity and cannot be diagnosed, much of the examination in chief and parts of the cross examination become clearer, and seen to be non-contributory to understanding these deaths.
2. Dr Wilkinson at one point says the post mortem was of no help in understanding Patrick's death, and then repeatedly uses the histopathology report to support a view that the death was asphyxial in nature. It is possible that his whole emphasis was to ensure that everyone understood that the ALTE was not due to herpes simplex encephalitis.
3. At one point Dr Wilkinson conflates the word asphyxia with cardio-respiratory arrest. This would seem to be acceptance that he cannot tell, from having seen Patrick or from any of the subsequent tests including the post-mortem examination, whether Patrick had suffered from an inability to breath because of obstruction to the mouth and nose, or from lowered oxygen levels and eventually cessation of breathing following a disturbance to the rhythm or stoppage of the heart. If so, then much of Dr Wilkinson's evidence in relation to the ALTE and Patrick's death takes everyone nowhere. I think this is as much because of failure by the barristers to understand asphyxia, and acquiescence in this by Dr Wilkinson, who after all is not a forensic pathologist.
4. I am not in any position to take issue with Dr Wilkinson's evidence in relation to herpes simplex encephalitis. I accept that it is most unlikely that this was ever present.
5. Dr Wilkinson sets much store on Dr Khan's report as establishing that Patrick suffered an asphyxial episode. Leaving to one side all the issues with asphyxia mentioned already, Dr Wilkinson refers to laminar necrosis of the occipital cortex as particularly supporting this. Laminar necrosis of the occipital cortex in both cerebral hemispheres is most likely occurring in vascular zones of the brain; ie parts of the brain served by the posterior cerebral artery. It is just as likely, if not more so, that the laminar necrosis has occurred because of hypotension (low blood pressure) resulting from a primary cardiac event as it is from a primary respiratory obstruction event.
6. The pressing by the Prosecutor that many of the reasons why Patrick did not have herpes simplex encephalitis were factors "consistent with" asphyxia raises the issues of the use of this phrase which have been canvassed above. They leave a distinct impression with this reader that the prosecutor believed he was making a point of substance, not merely a rhetorical flourish. If this is so, it is important to understand that such questions and their answers in this case have no medical content.
7. No reference is made during the evidence of any significance attaching to the serious seizures resulting in admissions to hospital on November 4 and November 14. The latter was noted as being associated with apnoea (ie cessation of breathing). This shows the serious nature of his illness. Even in the context of the murder charges, it was not alleged that these events were other than the natural consequences of Patrick's underlying encephalopathy. This would appear to be strong internal evidence of the fatal potential of the seizure disorder assoxciated with his encephalopathy.

CHAPTER X

HOW MIGHT THE FOLBIGG CHILDREN HAVE DIED?

1. Introduction

The continuing enigma, even at a reduced rate, of Sudden Infant Death Syndrome is a continuing reminder of how far we still have to go to understand why infants die.

Essentially, when stripped of its trappings, the forensic pathologists' – and other doctors' - conclusion at the trial that supported the homicide hypothesis is based on the coincidence of four deaths, an apparent lack of identifiable causes of death, and an ALTE in one family. If police, prosecutors and the courts do not like coincidences, or believe there are other grounds for concluding that crimes have been committed in relation to the deaths of the Folbigg children, that is entirely a matter for them. I would like to make as clear as I can that there is no forensic pathology basis for concluding that these children have been killed. In my view, any conclusion that the Folbigg children have been killed should be reached in the knowledge that such conclusion has no forensic pathology basis or support, and indeed there is, in my view, forensic pathology evidence against such a conclusion. I do not say that reaching such a conclusion is impossible; I am simply emphasizing that any such conclusion must be reached without relying at all on forensic pathology, and in my view, in the face of forensic pathology evidence to the contrary.

The question was repeatedly posed (eg 16.4.03 P749): Is there any natural cause of death that could account for all these four deaths and the ALTE? That is a fair question, but there are a couple of related questions that do not appear to have been asked, and should have been to place the question as posed in its proper place. For example, could one or two of the deaths be explained, leaving only two (or three) deaths unexplained? Could there be a mix of explained and unexplained deaths? The answer is clearly yes, but this does really seem to have been explored. Dr Cala's answer to the question as posed – is there any natural cause for all four deaths and the ALTE – was 'No'. This answer only makes sense if both the Prosecutor and Dr Cala were assuming that the question was: Is there any known natural cause.....? In recent years more known natural causes for these events have been clarified (and that is discussed in the coming pages) and there are also obviously yet to be discovered natural causes for these events. And we need to recall that a significant unknown possibility is an inherited vulnerability to physiological stresses precipitated in different ways in different cases.

2. The death of Laura:

Having said that, let us move on to consider how, and from what, might these children have died. Let us start with Laura where, as the reader will recall, a diagnosis of myocarditis – inflammation of the heart muscle – was made but was discarded as a reasonable cause of death even if the death was considered in isolation.

Using the National Coronial Information System (NCIS), Dr Colleen D'Arcy, investigated deaths from myocarditis in children under 2 years of age in NSW and Victoria since 2000. A total of thirty-nine cases of children from NSW and Victoria aged 2 years or less, with myocarditis as the cause of death, were identified on the NCIS. Nineteen cases (49%)[83] were from NSW and twenty cases (51%) from Victoria. Nineteen cases were male (49%) and the remaining twenty cases female (51%). Of the 39 cases, seven had either no post-mortem report, circumstances available for review or had confirmed sepsis or multiple

[83] All percentages that follow in relation to this study are rounded to the nearest whole number; therefore in some situations they do not add to 100%

infective/inflammatory foci elsewhere, including lung, heart and brain, hence did not die of isolated myocarditis. Five cases were also still open and under investigation and consequently were not accessible, thus 12 cases could not be used.

The remaining 27 cases were therefore included in the analysis. Of those, fourteen were from NSW and thirteen cases were from Victoria. Fifteen were male (56%) and twelve were female (44%). The age at death ranged from 7 days to 2 years.

The main circumstances of these are presented in Table 8.

Of the 27 cases, two (cases 4 and 5) had no accessible police form or details of circumstances, however post mortem reports were available; these cases were therefore included in the analysis. Therefore two cases (7%) had no known circumstances. Thirteen cases (48%) died in hospital either in the Emergency Department of Intensive Care Unit. The remaining twelve (44%) cases were dead on arrival at the emergency department and were either found unresponsive in bed (30%), died during transfer to hospital (4%) or whilst at a grandparent's house (7%). Of the nine infants found dead in bed, three cases were co-sleeping with parents in a double bed at the time of death.

One myocarditis case (4%) had an incomplete history available. Of the remaining 24 cases with histories to analyse, thirteen cases (51%) had evidence of a preceding illness including symptoms of an upper respiratory tract infection, lethargy and/or poor oral intake. This is likely an underestimate given two (7%) cases had features suggestive of a preceding viral illness (such as poor oral intake) or did not have complete circumstances and history available at the time of review.

Two cases (7%) had a second registered cause of death (atrial septal defect and focal brainstem encephalitis), however both had myocarditis registered as the primary cause of death. In two cases (7%) the cause of death included cardiomyopathy/myocarditis and after reviewing the autopsy reports this term "cardiomyopathy" was interpreted to be a macroscopic description of the heart (eg dilated, enlarged, heavy), rather than implying an inherent pathological disease of the myocardium independent of the myocarditis, as the term is applied more commonly. The appearance may well have represented the myocarditis having been present for some time.

One myocarditis case (4%) had an incomplete history available. Of the remaining 24 cases with histories to analyse, thirteen cases (51%) had evidence of a preceding illness including symptoms of an upper respiratory tract infection, lethargy and/or poor oral intake. This is likely an underestimate given two (8%) cases had features suggestive of a preceding viral illness (such as poor oral intake) or did not have complete circumstances and history available at the time of review.

Two cases (7%) had a second registered cause of death (atrial septal defect and focal brainstem encephalitis), however both had myocarditis registered as cause of death number 1. In two cases (7%) the cause of death included cardiomyopathy/myocarditis and after reviewing the autopsy reports this term "cardiomyopathy" was interpreted as representing a descriptive term to describe the heart macroscopically (eg. dilated, enlarged, heavy), rather than implying an inherent pathological disease of the myocardium independent of the myocarditis, as the term is applied more commonly. The appearance may well have represented the myocarditis having been present for some time.

The internal examination included the measured and expected heart weight (g). These, and the macroscopic and microscopic features of the heart and results from ancillary testing are presented in Table 9.

i) Myocarditis – an introduction

"*Myocarditis is an important cause of mortality in infants and older children, and sudden death is a well recognized presentation. This may be related to ventricular asystole, ventricular fibrillation or conduction defects. Infants are more commonly affected than older children, and more of the deaths occur in winter months. Of 207 cases of sudden death among individuals aged between 1 and 21 years, Neuspiel and Kuller*[84] *found myocarditis to be the predominant cause of cardiac death. Human myocarditis has been associated with a number of viruses, most commonly of the coxsackie group.*"[85]

The authors describe the macroscopic findings which can be associated: cardiac dilatation, mottling of the myocardium; opacification of the endocardium. And also the

[84] Neuspiel DR, Kuller LH. Sudden and unexpected natural death in childhood and adolescence. J Am Med Assoc 1985; 254: 1321-5

[85] Busuttil A and Keeling J. Paediatric Forensic Medicine and Pathology Edward Arnold. 2009(P227)

Table 8: Cause and main circumstances of myocarditis deaths included on NCIS in children less than or equal to 2 years of age in Victoria and NSW since 2000.

Case No.	Age at death	Age unit	Sex	State	Cause of death 1a	Place of death	Arrived dead or died in ED	Preceeding viral illness
1	7	Month(s)	F	NSW	Acute (probable viral) Myocarditis	Hospital	Died in hospital	Yes, decreased oral intake
2	18	Month(s)	M	NSW	*Dilated Cardiomyopathy and Lymphocytic Myocarditis	Hospital	Died in hospital	Yes, 3/52 dyspnoea
3	10	Day(s)	F	NSW	Bacterial Myocarditis	Hospital	Died in hospital	No
4	1	Year(s)	F	NSW	Myocarditis	N/A	N/A	N/A
5	2	Year(s)	M	NSW	Myocarditis	N/A	N/A	N/A
6	1	Year(s)	F	NSW	Myocarditis	Hospital	Died in hospital	Yes, pharyngitis
7	10	Month(s)	M	NSW	Idiopathic Focal Myocarditis	Hospital	Died in hospital	Yes, periorbital cellulitis
8	19	Day(s)	F	NSW	Myocarditis	Hospital	Died in hospital	Yes,poor oral intake
9	6	Month(s)	F	NSW	Viral Myocarditis with Pyelonephritis	Hospital	Died in hospital	Yes, rhinorrhoea
10	1	Month(s)	F	NSW	Lymphocytic Myocarditis	Basinet/bed	Arrived dead	Yes, vomiting, immunisations 2/7 prior
11	16	Day(s)	M	NSW	Acute (presumed viral) Myocarditis	Hospital	Died in hospital	No
12	14	Month(s)	F	VIC	Acute Myocarditis – presumed viral	Hospital	Died in hospital	Yes, unwell 2/52
13	2	Week(s)	M	VIC	Acute Myocarditis	In car during transfer	Arrived dead	Yes, poor oral intake
14	1	Week(s)	M	VIC	Myocarditis and Meningoencephalitis	Bed/home	Arrived dead. Resuscitation unsuccessful.	No
15	5	Week(s)	F	VIC	Myocarditis	Mother's bed	Arrived dead	Yes, cold 2/52
16	9	Week(s)	M	VIC	Interstitial pneumonitis and myocarditis (of probable viral aetiology)	Crib/bed	Arrived dead	No
17	8	Week(s)	M	VIC	Myocarditis and Interstitial pneumonitis	Parent's bed.	Arrived dead	Yes, off food, immunisations 1/7 prior
18	2	Year(s)	M	VIC	Myocarditis in the setting of immersion	In grandparent's pool	Arrived dead	No
19	10	Week(s)	M	VIC	Myocarditis	Parent's double bed	Arrived dead	Yes
20	1	Week(s)	F	VIC	Viral myocarditis and meningoencephalitis	Grandmother's house	Arrived dead. Resuscitation unsuccessful.	No
21	1	Week(s)	M	VIC	Myocarditis	Hospital	Died in hospital	No
22	10	Week(s)	M	VIC	Myocarditis and Encephalitis	Bed	Arrived dead	N/A
23	4	Month(s)	M	NSW	*Cardiomyopathy/Myocarditis, possibly of Metabolic Origin	Hospital	Died in hospital	Likely, unsettled, vomiting
24	5	Month(s)	M	VIC	Myocarditis and Pneumonitis (Picornavirus identified)	Bed/home	Arrived dead	No
25	27	Day(s)	F	VIC	Myocarditis, interstitial presumed lymphocytic meningitis	Hospital	Died in hospital	Likely, off food, sibling had URTI
26	2	Month(s)	M	NSW	Myocarditis	Cot/bed	Arrived dead.	Yes, cold 2/7
27	47	Week(s)	F	NSW	Myocarditis	Hospital	Died in hospital	N/A

Legend: M = Male, F = Female, VIC = Victoria, NSW = New South Wales, N/A = not available, URTI = upper respiratory tract infection
*This term, cardiomyopathy, is being used in a descriptive sense.

Table 9: Macroscopic and microscopic features of hearts in 27 deaths from myocarditis included on NCIS in children less than or equal to 2 years of age in Victoria and NSW since 2000.

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Case No	Heart Weight (g)**	Myocardium Macroscopic Features	Myocardium Microscopic Features	Myocyte necrosis	AV node	NPA
1	60 (42/36)	Pale	Florid myocarditis, dense lymphocytic infiltrate, myocytolysis	Yes	Not examined	Negative
2	132 (44/51)	Pale light brown, LVH (10mm), dilated LV>RV	Lymphocytic inflammation, oedema, myocyte necrosis	Yes	Not examined	n/r
3	24 (19/24)	Normal	Myocyte necrosis, focally diffuse neutrophiic infiltrate	Yes	Not examined	Negative
4	n/r (66/41)	Enlarged, dilated LV	Dense lymphocytic infiltrate, myocytolysis, myofibre degeneration, collagen scar	Yes	Dense lymphocytic infiltrate	n/r
5	124 (92/51)	Normal	Lymphocytic and macrophage infiltrate, occsional neutrophils/eosinophils, necrotic myofibres	Yes	Inflamed	n/r
6	50 (50/46)	Reddish/brown, subtle pale mottling	Transmural diffuse myocarditis, lymphocytes, atypical lymphocytoid cells, myocyte necrosis	Yes	Not examined	n/r
7	42 (48/46)	Focal haemorrhagic discolouration in RV	Extensive myocardial necrosis, mixed inflammatory infiltrate (neutrophils, lymphocytes, eosinophils)	Yes	Normal	n/r
8	36 (19/19)	Multifocal scarring and haemorrhage LV	Lymphocytic (CD3, some CD20) myocarditis, myocyte loss, organising connective tissue	Yes	Not examined	Staph aureus++, mixed oral flora
9	92 (42/31)	Pallor, oedema, softening of LV and focally RV	Florid inflammation, lymphocytes (CD3/CD6a/CD7+), plasma cells, myocyte necrosis, oedema	Yes	Not examined	n/r
10	18 (19/19)	Uniformly dark red/brown, firm and unremarkable	Lymphocytic myocarditis, focal destruction of myocytes	Yes	Not examined	Negative
11	39 (21/22)	Severely mottled, haemorrhagic and pale LV and IVS	Florid, acute myocarditis, dense lymphocytic, occasional neutrophils, eosinophils and plasma cells, myocyte death/necrosis	Yes	Not examined	n/r
12	65 (74/46)	Tan, slightly glistening, otherwise unremarkable	Diffuse lymphocytic and neutrophilic infiltrate associated with myocyte necrosis	Yes	Not examined	Negative
13	22.7 (16/18)	Firm red and unremarkable	Widespread acute inflammatory infiltrate, extensive myofibre necrosis, basophilic smudged inclusions	Yes	Not examined	Respiratory syncytial virus
14	32.8 (16/22)	Congested LV	Focal lymphoplasmacytic inflammation, occasional neutrophils, eosinophils in LV, IVS and epicardium, myocyte necrosis	Yes	Not examined	Picornavirus, enterovirus
15	23 (24/24)	Uniformly red/brown	Two foci of lymphocytic infiltrate a/w myofibre necrosis, oedema	Yes	Not examined	n/r
16	20.9 (16/22)	Uniformly red/brown	Essentially normal. Occasional foci of interstitial inflammation with only one foci with myofibre necrosis, elsewhere no myofibre necrosis.	Yes	Not examined	Negative
17	18.8 (16/18)	Pale red and normal	Lymphocytic infiltrate, myofibre degeneration, few neutrophils	Yes	Not examined	Picornavirus
18	79 (57/71)	Unremarkable	Numerous foci of interstitial lymphocytes, myocyte necrosis	Yes	Not examined	n/r
19	47.8 (30/35)	Uniform red/brown, unremarkable	Focal interstitial lymphocytes and focal myocyte destruction	Yes	Not examined	Negative
20	19.3 (19/12)	Reddish/brown, no haemorrhages	Acute myocarditis, diffuse, myocyte necrosis, mixed inflammatory infiltrate	Yes	Not examined	Picornavirus
21	26 (16/26)	Mottling, intense congestion anteroseptally	Conspicuous infiltrate, plasma cells, eosinophils, macrophages, lymphocytes, myocyte degeneration/necrosis	Yes	Not examined	Negative

22	16 (16/22)	Unremarkable	Focal LV lymphocytic infiltrate, myocyte necrosis	Yes	Normal	CMV DNA detected
23	47 (35/30)	Pale LV wall	Moderate diffuse mononuclear infiltrate (neutrophils, plasma cells, eosinophils)	No	Normal, inflammation adjacent	n/r
24	30.6 (30/35)	Unremarkable	Patchy interstitial lymphocytes, no myocyte necrosis	No	Not examined	Picornavirus
25	21.4 (19/19)	Red, firm.	Sparse, patchy lymphocytic infiltrate, convincing myocyte necrosis not seen	No	Not examined	Picornavirus
26	N/A	N/A	N/A	N/A	N/A	N/A
27	N/A	N/A	N/A	N/A	N/A	N/A

Legend: n/r = not recorded, NPA = nasopharyngeal aspirate, AV = atrioventricular, LV = left ventricle, RV = right ventricle

**** Expected normal heart weight is provided in brackets, respectively against the baby's weight/height. Based on Scholtz DG et al. Age related changes in normal human hearts during the first 10 decades of life. Part 1 (Growth): A qualitative anatomic study of 200 specimens from subjects from birth to 19 years old. Mayo Clin Proc 63: 126-136, 1988.**

microscopic findings. The heart may be specifically affected, or part of a generalized involvement. "A problem often faced by pathologists and for which there is no easy answer is how many foci of inflammatory cells in the myocardium are sufficient to cause death". Myocarditis can coincide with other obvious causes of death such as trauma and therefore can be incidental.

ii) Could myocarditis be the cause of Laura's death?

The following excerpts from the transcript of Dr Cala's evidence sets out his view of myocarditis as a possible cause of Laura's death:

P714
Q. Now, is that (ie the myocarditis) sort of finding the finding that you found on Laura's heart of inflammatory infiltrate, consistent with the after effects of a cold or flu?
A. I believe so.
Q. In your opinion did it play any role in causing her death?
A. I don't believe so
Q. Would you explain to the court why you have that opinion?
A. As I have said, the heart was normal to the naked eye, but my microscopic examination did reveal inflammation of the heart. Having said that, the inflammation was quite patchy and rather mild in the sense that although the inflammation existed it was of a rather low amount as opposed to other cases that I've seen where the inflammation was much heavier in the heart and in other organs.
Q. Where the inflammation is much heavier it can cause death?
A. Yes.....
Q. And if someone had died from myocarditis of the kind that you have described, what would you expect to see in and around the heart?
A. I'd expect to see a number of things. The heart may, but not always, I have to say, it may be flabby and have a – when you cut through the pump of the heart, the left ventricle in particular, it may have a stripey appearance. In other words areas of paleness against areas of more normal looking heart, and that is just the way the inflammatory process is.
Q. Did you find any of those in Laura's case?
A. No. This is with the naked eye, looking at the heart with the naked eye. The left ventricle, that is the main chamber of the heart, may be a bit flabby and the chamber itself may be a bit dilated. I didn't find those changes in this case. Then there might be evidence of heart failure because a number of these people both children and adults may have myocarditis and it presents clinically to doctors as heart failure, so they may have fluid around the lungs and they may have fluid in the abdomen and I didn't see either of those things in this case.
Q. And in your experience as a pathologist have you sometimes come across persons who have died from totally unrelated causes like car accidents who have been found to have these, this mild inflammatory infiltrate of the heart.
A. Yes, I've seen them personally and they have been written in the literature.
.....

P719
(Videotape of Laura taken the day before she died showing her in normal good health)

Q. Doctor, if you accept that that tape was made of Laura the day before she died, does that assist you in any way in your opinion that inflammatory infiltrate of the heart or myocarditis did not cause her to die?
A. I think that Laura Folbigg appeared to me in quite normal health on that video, and I think that was about roughly 23 hours before she died. I think from that, given that she appears in quite good health, I think it is quite unlikely that she died as a result of the effects of myocarditis.
Q. What do you say to the possibility that she died of myocarditis?
A. I think it is known that myocarditis can cause sudden death, usually by a cardiac rhythm disturbance and I can't say that didn't happen with Laura Folbigg but I think it's, in all likelihood, very unlikely.
Q. Is it a reasonable possibility in your opinion that she died from myocarditis.
A. I don't believe it is....

16.4.03 P754
Q Would you agree that as a general principle it is recognized in paediatrics.... That children with myocarditis may die suddenly and unexpectedly with no symptoms or signs?
A The answer is yes, but in a minority of cases..... ....a small percentage may die suddenly and unexpectedly from myocarditis.
Q Without there being any signs externally?
A Yes.....
P756
Q .....and the range is from no manifestations to overt manifestations
A .....But I think I would have to say on careful review of certainly of a death from myocarditis I think it would be very unusual to have absolutely no symptoms or signs of some abnormality prior to the death by way of a fever or constitutional symptoms of being unwell, aches and pains in the joints, maybe a bit of shortness of breath, a bit of chest pain and so on
.....

P757
Q Do you keep a record of how many slides you prepared from the heart?
A. I don't keep a record but a record is kept in the histology section of the Institute
Q Do you recall how many you have taken in this case?
A Well, I recall it would have been at least 8. Four blocks taken routinely anyway and then once I had looked at the heart and found an abnormality I took more sections to see in fact how florid or otherwise her condition was.....Collectively I made some notes.....(Did not look at the conduction system)
P759
Q Was it the case that within the myocardium that there was a moderately dense infiltration of lymphocytes?
A Yes it was....not heavy, not light, somewhere in between.....
P760 Q Was it the case that these lymphocytes had however aggregated in certain areas of the heart?
A Yes
Q Particularly subendocardial?
A Yes.....

Q Were there also aggregates on the surface of the myocardium or the heart
A Yes
Q Were there large aggregates in the central area of the left ventricle?
A. Yes....

P761
Q Is it the case that all the slides, all the samples, of heart showed the presence of myocarditis? [86]
A. I don't think all but I think most.....but that is not to say that the amount of inflammation was florid and heavy in those sections. It was actually.....fairly patchy in the area that I have described[87].
Q Looking at this case in isolation.....can you exclude myocarditis as the cause of death?
A I can't exclude it as the cause of death.
Q Might you have given the cause of death as myocarditis looked at individually
A I don't think I would because, although it was present the amount of inflammation was not particularly heavy. There wasn't any evidence of heart failure, the heart to the naked eye looked pretty normal so – and not only that, there was evidence in other organs, the lungs and the spleen in particular of lymphocytes being in there as well. In other words indicative of some viral infection that Laura Folbigg was suffering from around the time of her death.

What now follows is a distillation of Dr Cala's evidence at the trial insofar as it relates to existence of myocarditis in Laura's heart, and the causal relationship between that and the cause of Laura's death. In summary, Dr Cala accepted the existence of the myocarditis, but there were a number of elements which all led to his view that Laura did not die of myocarditis. These elements and my view about them are set out below.

- In Laura's case the myocarditis was patchy and mild compared to other cases where the inflammation was more marked.

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[86] All seven of the heart slides evaluated microscopically in fact show myocarditis.

[87] Note Dr Cala states in his letter to Det Ryan on 19 June 2001: "The inflammatory infiltrate in the sections of heart which I examined in the case of Laura Folbigg was light in amount and patchy in distribution....My opinion that the inflammatory infiltrate in the heart represents an incidental finding is not based on the family history but rather, after consideration of the history provided of Laura's very sudden and most unexpected death, the post mortem findings of Laura and the histological assessment of the heart together with my own knowledge and experience of the condition of myocarditis. In other cases I have seen where the death of a child or an adult has been due to myocarditis the inflammatory infiltrate has been much heavier in number and more diffuse in distribution throughout the heart, although the amount of inflammation is variable from case to case. There are often observable naked eye changes when examining the heart. These changes may consist of dilation, flabbiness and pallor of the heart and a striped appearance of the heart on cut section. There may be features at post-mortem examination suggestive of heart failure. This may take the form of pleural effusions, straw-coloured fluid in each pleural cavity and ascites – fluid in the abdominal cavity. I should point out that these findings are not seen in every case and there are other causes for these findings. These findings were not present with Laura Folbigg whose heart looked normal on naked eye inspection."

I do not think the myocarditis is patchy and mild; I think it is better described as widespread and at least moderate in degree. How does one test this difference in view? In circumstances where colleagues were not aware of my involvement in this case, I circulated the following email to my ten consultant forensic pathology colleagues at the VIFM together with a number of photomicrographs which are attached as an appendix.

"This girl was 19 months old when she died. She had a runny nose for a couple of days. She was fed at 7 am, playing normally at about 11 am. She then had a sleep and when her mother went to check on her around midday, she was not breathing. Pathologist gave the cause of death as unascertained. Apart from myocarditis, which the pathologist reported as being present, the autopsy was negative. I would be happy with myocarditis as the cause of death. Any comments on this, or on the myocarditis itself? Would appreciate feedback."

Attached to the email were photomicrographs of the myocardium. These photomicrographs are attached as Appendix 3. I received the following replies:

Path 1	Certainly this is a very impressive myocarditis, I have 2 strikingly similar cases on my desk at the moment where there was enterovirus in bowel and picornavirus in myocardium (likely entero). Some of the inflammatory cells look quite immature but I assume that is just because this has been going on for a few days. Is the bone marrow ok?
Path 2	If a pathologist is not willing to call this myocarditis, I am wondering what it would take!
Path 3	Apparently wide-spread, predominantly lymphocytic infiltrates with areas of myocyte necrosis and interstitial oedema. Certainly looks like myocarditis as COD (cause of death).
Path 4	I agree myocarditis
Path 5	I think myocarditis and would have put it as cause of death as it appears so florid
Path 6	I would go with myocarditis, no doubt.
Path 7	Clear case of myocarditis. I would be happy with this as the cause of death in the circumstances indicated
Path 8	Looks like myocarditis to me-probably viral. Eosinophils look conspicuous which makes me think drugs but distribution of infiltrate not right. In the described circumstances I am happy with it as the cause of death. Results of virology? Does report make it apparent why pathologist uncomfortable with that as COD?
Path 9	I have to agree with you - I would give the COD as myocarditis.
Path	It cannot be anything else other than myocarditis.

I have included all the replies here. Truth is not a matter of democracy, even if it is unanimous; and in forensic pathology there is room for disagreement. But I think the tenor of these replies goes to the following point: for a number of pathologists, in the circumstances as described (which did not include the fact of three previous infant deaths in the same family) the myocarditis was regarded as sufficient to be a reasonable cause of death. (The comments cannot be used to support my view that the myocarditis is more than patchy and mild, as obviously the pathologists only saw what I sent them and did not view the entirety of the 7 slides.)

In this regard it is interesting to see in the report of Professor Berry, Professor of Paediatric Pathology at the University of Bristol, who concluded in regard to Laura's death: "Nevertheless, taken in isolation, I would have ascribed this death to myocarditis, recognizing that although the infiltrate was quite extensive, I could not see actual damage to heart muscle."[88] The conclusion of Professor Berry's report overall is a conclusion about the sudden unexpected deaths of **three** infants in the one family, not four.

- In Laura's case the heart seemed (to the naked eye) normal compared to other cases where death is due to myocarditis; in the latter the heart is possibly flabby, the left ventricle a bit dilated and has a striped appearance. In other words, Dr Cala would expect to see these signs to support the view that death was due to myocarditis.

In assessing this aspect in the 27 cases in our study, we have interpreted any naked eye description which we could reasonably infer as abnormal, as in fact abnormal. 12/27 cases were regarded as having an abnormal naked eye appearance; 13/27 were normal and for two there was no information.

Weber et al (2008) reported a series of 1516 paediatric autopsies over a 10 year period from Great Ormond Street. Histologically proven myocarditis was diagnosed in 28 cases (1.8%; age range 10 days -16 years; median age 10 months). In 11 (40%) of the cases, there was no macroscopic evidence of abnormality in the heart[89].

- In Laura's case there were no preceding symptoms or signs referable to myocarditis or viral illness. Dr Cala believes this absence is very unusual in deaths from myocarditis

In fact it seems that Laura did indeed have a runny nose in the couple of days prior to her death; this would normally be regarded as a sign of likely, albeit mild, viral illness. But

[88] I respectfully disagree with this observation of Prof Berry's; I think this damage is present.
[89] Weber MA, Ashworth MT, Risdon RA, Malone M, Burch M, Sebire NJ. Clinicopathological features of paediatric deaths due to myocarditis: an autopsy series. Arch Dis Child 2008;93;594-598.

leaving that to one side (because actually it is not a material criterion in assessing whether the cause of death is or is not due to myocarditis as Dr Cala seems to believe), consider the material from the NCIS search.

In Table 9, there are 27 cases where myocarditis was regarded by the pathologist as the precipitating cause of death. Of these, 15/27 had symptoms referable to a viral illness. For the purposes of this survey, a single symptom, such as being 'off food' was regarded as sufficient to establish such an illness. In 8/27 cases there were no symptoms reported of a preceding illness. In 4/27 cases no information enabling such an assessment was available.

In the series of myocarditis deaths reported by Weber et al (2008), 16 (57%) presented as a sudden death[90].

- In only a small percentage of cases when myocarditis causes death does it cause a sudden and unexpected death.

(It is not clear what definition of sudden and unexpected death Dr Cala had in mind. Some authors regard a sudden death as one which occurs within 24 hours of the onset of symptoms.).

For the purposes of this review we have taken death at home as the measure of its sudden and unexpected nature. At the very least, if the child was ill, caregivers did not believe that the infant/child needed to be at hospital. Death in hospital we have regarded as not sudden and unexpected being at least slightly delayed. (We are not able to say from the information on the NCIS how long the deceased was in hospital).

- 13/27 died in hospital
- 12/27 arrived dead at hospital (of which 10 were found in a cot or bed; a 2 year old was found in a swimming pool; and a one week old was found at the grandmother's house).
- 2/27 no information available

On this basis it would appear that sudden and unexpected death is not all that unusual in this population of infants and toddlers dying from myocarditis, happening in about half the cases. This conclusion is supported by the experience of Weber et al (2008) from Great Ormond Street, as set out above.

(It is possible that our own conclusion understates the proportion who collapsed suddenly at home, as it seems three of the deceased (cases 3, 11 and 21) had no preceding symptoms of viral illness yet died in hospital. The likelihood is that they were rushed to hospital and death occurred and/or the fact of death was certified in hospital.)

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[90] Weber MA, Ashworth MT, Risdon RA, Malone M, Burch M, Sebire NJ. Clinicopathological features of paediatric deaths due to myocarditis: an autopsy series. Arch Dis Child 2008;93;594-598.

- Laura appears normal in a video taken 23 hours before her death. "....given that she appears in such good health I think it is quite unlikely that she died of the effects of myocarditis".

There is no specific data on the NCIS which allows us to explicitly address the health of the 27 deaths in our study 23 hours before they died. But the two preceding elements amount to much the same thing by reasonable inference. 12/27 died at home and 8/27 had no record of any symptoms in the period leading up to being found dead. It is reasonable to say that between 8 and 12 of these 27 infants could have had a normal video taken of them 23 hours before their deaths.

- If this had been an isolated death, Dr Cala would not have given the cause of death as myocarditis because the amount of inflammation was not particularly heavy; there was no evidence of heart failure; the heart to the naked eye looked normal.

I imagine the evidence of heart failure Dr Cala is referring to is evidence of congestive cardiac failure, which would really be a marker of the myocarditis interfering with the efficient functioning of the heart over time, resulting in pulmonary oedema and perhaps pleural and pericardial effusions (fluid around the lungs and heart), congestion of the liver ("nutmeg change"), perhaps even peripheral oedema, and dilatation of the left and right ventricle with some thinning of the wall of the ventricles. In one sense all of the deaths in our review are deaths from acute heart failure, but apart from some terminally developing pulmonary oedema, there will usually not be sufficient time for the other signs mentioned to develop. We did not assess congestive cardiac failure in these cases.

The actual degree of myocarditis present was, in my opinion, substantially more than mild, and at least of moderate severity. And I have mentioned above that 13/27 cases were regarded as having a normal looking myocardium when assessed with the naked eye.

iii) Conclusion about myocarditis as the cause of Laura's death

Q: "Is it a reasonable possibility that she died from myocarditis?
A (Dr Cala). I don't believe it is."

I believe the middle of the road conclusion in relation to Laura's death is that considered alone, most forensic pathologists would be comfortable ascribing the death in similar circumstances to Laura's as being due to myocarditis. This is indeed my own view. It would have been acceptable, and I would support a pathologist who gave the cause of death as "I(a): Undetermined", but in the comments section of the report, fully canvassed the possibilities that death could be due to myocarditis, but because it was the 4th death in the particular family there could be other factors, including but not limited to homicide, at work.

Thus, I do think Dr Cala could have justified the cause of death as he gave it: Undetermined. He was incorrect to argue that there were medical and pathology reasons for excluding myocarditis. He could simply have said that, as this was the fourth infant/childhood death in the family, he was worried about smothering (but see below) or other natural causes having played a part. He

could have accepted that death might have been due to myocarditis – and he does accept this as a minor theoretical possibility - but the circumstantial information – four deaths in one family - concerned him. And simply left it at that. To the extent that he says that death could not be due to myocarditis, I disagree, and the evidence from the NCIS is against Dr Cala's view. Laura could very well have died from myocarditis, and it is my view that a clear preponderance of forensic pathologists would so conclude if Laura's was an isolated case.

Consideration of Laura's cause of death could easily extend into a discussion about how pathologists go about, or should go about, concluding the cause of death generally. See Appendix 5 for a discussion of this.

iv) Myocarditis as incidental to other causes of death

An NCIS search was undertaken to try and clarify this aspect of myocarditis. Autopsy reports for all NSW and Victorian cases on the NCIS since 2000 were all searched for myocarditis to find those cases where myocarditis appears in the autopsy report, but neither in Part I nor II of the cause of death.

This search produced a total of 7091 reports with the word "myocarditis" mentioned in the autopsy report but not in the cause of death (in neither Part I nor Part II of the cause of death statement). The vast preponderance of these cases were not cases of myocarditis but cases where myocarditis was mentioned as a negative finding (ie there is "no evidence of myocarditis").

Of these 7091 cases, a random 200 cases of patients aged 2 years or less were screened to determine the context for the word "myocarditis". Of the screened set, 7 cases (3.5%) had reported features of myocarditis either in the cardiovascular histology section or in the positive anatomical findings section of the autopsy report.

Of these cases (presented in table 10), the age range was two months to two years. Five cases (71%) were male and two cases female (29%). The registered cause of death included unascertained (2 cases), drowning (2 cases), multiple injuries sustained in motor vehicle accident (1 case), Neisseria Meningitis septicaemia (1 case) and Consistent with Sudden Infant Death Syndrome (1 case).

Table 10: Cases with myocarditis incidental to the cause of death.

Case No	Age	Sex	Cause of death	Heart weight (g)	Macroscopic	Heart Histology
1	2 years	Male	Multiple injuries sustained in a motor vehicle collision (passenger)	64	N/A	N/A
2	2 months	Female	Consistent with Sudden Infant Death	22	Unremarkable	One small focus lymphohistiocytic infiltrate, possible

			Syndrome			myocyte damage
3	2 years	Male	Drowning	82	Uniform red/brown	Multifocal lymphohistiocytic infiltrate, myocyte necrosis
4	1 year	Female	Unascertained	52	Unremarkable	Single focus myocarditis right ventricle
5	2 years	Male	Undetermined	88.5	Unremarkable	Single focus myocarditis left ventricle
6	1 year	Male	Neisseria meningitidis septicaemia	64	Uniform red/brown	Multiple micro-foci neutrophils and myocyte necrosis
7	15 months	Male	Drowning	52	Homogeneous brown, unremarkable	Mild, multifocal inflammation, chronic, few neutrophils, mild early myocyte damage

Legend: N/A = not accessible.

3. The death of Patrick

It will be recalled that Patrick suffered from difficult to control seizures as a consequence of hypoxic ischaemic encephalopathy following his ALTE. Indeed the cause of death concluded by the pathologists responsible for his autopsy was finally worded as follows:

I (a) Asphyxia due to airways obstruction[91]
I (b) Epileptic fits
I (c) Encephalopathic disorder (underlying cause not determined on investigation)

At the trial this cause of death was disowned and his death was subsequently dealt with as though it was much the same as the other deaths.

The point to be made here is that Patrick's death has been considered by all participants in the trial, it seems to me, as an event quite independent of the ALTE. However, once he had suffered the ALTE, is perfectly understandable as being from the delayed effects of the ALTE. This, it seems to me, is a non-controversial, ordinary thought.

4 The death of Sarah

[91] The issue of asphyxia has been canvassed above.

It will be recalled that Professor Hilton conducted this autopsy knowing it was the third death in the one family. According to the definition of SIDS and categories proposed by Krous et al (2004)[92] (see Ch IV), Sarah's death falls squarely within the category of SIDS II. These categorizations were published in 2004, so clearly Dr Cala and the other pathologists cannot be remotely criticized for forming views contrary to this. As mentioned in Ch IV, Krous et al's view about the definition of SIDS is the dominant view in the western world currently. Prof Hilton correctly anticipated this sort of approach when he gave the cause of death as he did: Sudden Infant Death Syndrome. Dr Cala can also be credited with anticipatory antennae because his major criterion for disagreeing with Prof Hilton's diagnosis of SIDS was Sarah's age.[93] (His other disagreement related to abrasions on Sarah's chin, which I have discussed above).

Considered alone, Sarah's death is properly diagnosed as : Sudden Infant Death Syndrome (Category II).

It may be thought this does not advance things much. Since the time of Sarah's death, if the same situation was to present itself today, there would be thorough cardio-genetic investigations of possible inherited arrhythmias.

5 The death of Caleb

Caleb's death was diagnosed at the time as SIDS. Dr Cala gave evidence that he would diagnose his death as undetermined. According to approach of Krous et al (2004), the correct diagnosis for Caleb's death would be: Sudden Infant Death Syndrome (Category II). This is because of the age of death being 19 days. Had Caleb died 2 days later, his death by contemporary standards would have been properly diagnosed as SIDS (Category IB) because of the incomplete level of investigation/documentation according to today's standards.

6 The death of Caleb, Sarah and Patrick's ALTE considered together

This section considers Caleb and Sarah's deaths and Patrick's ALTE as a collective. These events considered individually and alone are effectively not explained. This section does not include the deaths of Laura and Patrick because the author believes that, considered alone, those deaths are explicable, if not explained. (The reader should be aware that relatively few deaths are from causes incompatible with life; ie explained. Most are from causes which are compatible with life; ie explicable). This section also needs to be read in the light of the fact that no witness at trial could think of any explanation to tie the four deaths and the ALTE together. This was the wrong challenge. In my view the core challenge is to tie together medically, forensic pathologically, Caleb and Sarah's deaths and Patrick's ALTE.

It is at this point in this report that the issue of multiple SIDS in one family needs to be considered. Most readers of this report would be aware of the intense controversy that has

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[92] Krous HF, Beckwith JB, Byard RW et al. Sudden infant death syndrome and unclassified sudden infant deaths: a definitional and diagnostic approach. Pediatrics 2004;114:234–8.

[93] Trial transcript: 16.4.03 P748 Line 10ff

attached to this subject, particularly in the UK. I am aware that separate advice has been sought on this issue by the Instructing Solicitors in this case. This has primarily been a statistical debate which is outside my field, although earlier in this report I included some material informing this debate. (Carpenter et al, Bacon et al, Gornall et all).

As a general proposition, if SIDS is regarded as a collection of (currently) unidentifiable natural diseases or natural disorders of physiological or biochemical processes, then it follows that some of these will have a genetic basis and be capable of being inherited. Some may not be diseases as commonly understood, but potential diseases or states which are uncovered when the infant physiology and biochemistry is stressed in some way, perhaps by a mild respiratory illness, or by being placed face down to sleep.

Take for example a paper co-authored by Opdal et al (2004), well respected experts in Sudden Infant Death Syndrome.[94] It concludes as follows:

> *"It is unlikely that one mutation or polymorphism is the predisposing factor in all SIDS cases. However, it is likely that there are "SIDS genes" operating as a polygenic inheritance predisposing infants to sudden infant death, in combination with environmental risk factors...In the future, some of the cases now diagnosed as SIDS will probably be diagnosed as, for instance, metabolic or cardiac disease based on a better knowledge of the genetic basis of these and other diseases"*

Opdal and Rognum's paper analyses a number of gene polymorphisms that may predispose infants to sudden infant death under certain circumstances. These included:

"IL-10, an important immuno-regulatory cytokine that plays an important role in the development of infectious disease;....serotonin transporter gene influenc(ing) a broad range of physiologic systems, including the regulation of breathing, the cardiovascular system, temperature, and the sleep wake cycle."

The truth of Opdal's thesis is now very much clearer. Genetic factors have been found to play a role in some patients with SIDS. Since the 1970's, the possible association between congenital LQTS and SIDS has been pointed out.[95] It is currently estimated that 5-20% of SIDS are related to ion channelopathies.[96] So far, no fewer than 13 cardiac channelopathy susceptibility genes have been implicated in the pathogenesis of SIDS. These include potassium channel genes (KCNQ1, KCNH2, KCNE1, KCNE2, KCNJ8), sodium channel genes and related regulatory

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[94] Opdal SH, Rognum TO. The Sudden Infant Death Syndrome Gene: Does it exist? Paediatrics 2004;114;506-512.

[95] Kelly DH, Shannon DC, Liberthson RR. The role of the QT interval in the sudden infant death syndrome. Circulation 1977; 55:633-5.
Schwartz PJ. Cardiac sympathetic innervation and the sudden infant death syndrome. A possible pathogenetic link. Am J Med 1976; 60:167-72

[96] Klaver EC, Versluijs GM,Wilders R. Cardiac ion channel mutations in the sudden infant death syndrome. Int J Cardiol. 2011; 152: 162-70. Hunt CE, Hauck FR. Sudden Infant Death Syndrome. Can Med Assoc J. 2006; 174: 1861-9

proteins (SCN5A, SCN3B, SCN4B, GPDL1, CAV3, SNTA1), the calcium release channel of the sarcoplasmic reticulum (RYR2) and other genes (GJA1).[97] The clinical syndromes associated with these mutations are LQTS, short QT syndrome, Brugada Syndrome, Catecholaminergic Polymorphic Ventricular Tachycardia or idiopathic ventricular fibrillation, Early Repolarisation Syndrome and Progressive Cardiac Conduction Disease. Of these clinical phenotypes long QT syndrome is the most extensively studied in the new born. In a prospective study of a large cohort of 34,000 neonates with an ECG performed on the third or fourth day of life, the authors demonstrated that 50% of infants who subsequently died with a diagnosis of SIDS showed a prolonged QT interval.[98] The same research group subsequently performed genetic testing which resulted in finding genes related to LQTS.[99] () More recently Arnestad et al reported that the

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[97] Tan BH, Pundi KN, Van Norstrand DW, Valdivia CR, Tester DJ, Medeiros-Domingo A et al. Sudden infant death syndrome-associated mutations in the sodium channel beta sub-units. Heart Rhythm. 2010; 7:771-8.
Ackerman MJ et al. Post mortem molecular analysis of SCN5A defects in sudden infant death syndrome. JAMA. 2001; 286:2264-9.
Tester DJ, Ackerman MJ. Sudden infant death syndrome: how significant are the cardiac channelopathies? Cardiovasc Res. 2005; 67: 388-96.
Plant LD et al. A common cardiac sodium channel variant associated with sudden death in African Americans, SCN5A S1103Y. J Clin Invest. 2006; 116:430-5.
Wedekind H et al. Sudden infant death syndrome and long QT syndrome: an epidemiological and genetic study. Int J Leg Med. 2006; 120:129-37.
Arnestad M et al. A mitochondrial DNA polymorphism associated with cardiac arrhythmia investigated in sudden infant death syndrome. Acta Paediatr. 2007; 96:206-10.
Otagiri T et al. Cardiac ion channel gene mutations in sudden infant death syndrome. Paed Res 2008; 64: 482-7.
Millat G et al. Contribution of long QT syndrome genetic variants in sudden infant death syndrome. Paediatr Cardiol. 2009; 30:502-9.
Tester DJ et al (incl Ackerman MJ). Loss-of-function mutations in the KCNJ8-encoded Kir6.1 KATP channel and sudden infant death syndrome. Circ Cardiovasc Genet 2011; 4:510-5.
Tester et al. A mechanism for sudden infant death syndrome(SIDS): stress-induced leak via ryanodine receptors. Heart Rhythm. 2007; 4:733-9.
Cronk LB et al. Novel mechanism for sudden infant death syndrome: Persistent late sodium current secondary to mutations in caveolin-3. Heart Rhythm. 2007; 4:161-6.
Van Norstrand DW et al. Molecular and functional characterization of novel glycerol-3-phosphate dehydrogenase like gene (GPD1-L) mutations in sudden infant death syndrome. Circulation. 2007; 116:2253-9.
Cheng J et al. Alpha -1 syntrophin mutations identified in sudden infant death syndrome cause an increase in late cardiac sodium current. Circ Arrhyth Electrophysiol. 2009; 2:667-76.
Van Norstrand DW et al. Connexin 43 mutation causes heterogeneous gap junction loss and sudden infant death. Circulation. 2012; 125:474-81.
[98] Schwartz PJ et al. Prolongation of the QT interval and the sudden infant death syndrome. NEJM. 1998; 338:1709-14
[99] Schwartz PJ et al. A molecular link between the sudden infant death syndrome and LQTS. NEJM. 2000; 343:262-7.

prevalence of LQTS disease causing mutations in SIDS was 9.5%.[100] It follows that in families with multiple SIDS this prevalence would be expected to be higher.

Currently SIDS related mutations in cardiac sodium channel genes (SCN5A, SCN3B, SCN4B) constitutes 43% of all mutations found and the majority are missense mutations[101]. Functional analysis of several mutations has revealed that some mutations cause gain-of-function, while others cause loss-of-function. Clinically, this leads to the LQT3 and Brugada Syndrome phenotypes respectively. Another mutation found in SNC3B (V36M) showed both gain-of-function and loss of function phenotypes secondary to decreased peak Ina and increased late Ina. The SCN5A polymorphism S1103Y, common in African Americans, has also been implicated as a possible modifier to increase susceptibility to SIDS by increasing late Ina.[102] This is relevant as a pointer to the complexity of interactions that might be involved in any particular case of SUDI/SIDS – that is, a pointer to the complexity of the enigma that is SUDI/SIDS.

Although the mechanism of SIDS is not fully elucidated, some potential SIDS victims will certainly benefit from genetic analysis, considering that the prevalence of channelopathies in SIDS is up to 20%. Genetic testing in SIDS victims may also help to evaluate the variable expressivity in the ion channelopathies.

Ackerman is a pre-eminent researcher into inherited cardiac arrhythmias. In 2004, he and co-authors reckoned that "pathogenic mutations in the cardiac sodium channel gene, SCN5A cause approximately 15-20% of Brugada Syndrome, 5-10% of Long QT Syndrome (LQT3) and 2-5% of SIDS".[103] This is but one of the genes related to cardiac arrhythmias.

Skinner is the leading New Zealand researcher in the field of the role of cardiac arrhythmias in sudden unexpected death. In this 2011 paper he and co-authors recommend that, on the basis of their genetic testing work resulting in the conclusion that 15% of sudden unexpected deaths in the young (ie those aged 1-40 years) are caused by Long QT syndrome, genetic autopsy should become routine practice. This reinforces the developing knowledge in the field and emphasizes that understanding of the role of inherited arrhythmias, very limited 10-15 years ago, is improving.[104]

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[100] Arnestad M et al. Prevalence of long QT syndrome gene variants in sudden infant death syndrome. Circulation. 2007; 115:361-7.

[101] Klaver et al. Cardiac ion channel mutations in the sudden infant death syndrome. Int J Cardiol. 2011; 152:162-70

[102] Cheng J et al. The common African American polymorphism SCN5A-S1103Y interacts with mutation SCN5A-R680H to increase late Na current. Physiol Genomics. 2011; 43:461-6

[103] Ackerman MJ, Splawski I, Makielski JC, et al. Spectrum and prevalence of cardiac sodium channel variants among black, white, Asian, and Hispanic individuals: implications for arrhythmogenic susceptibility and Brugada/long QT syndrome genetic testing. Heart Rhythm 2004;1:600–7.

[104] Skinner JR, Crawford J, Smith W, et al. Prospective, population-based long QT molecular autopsy study of postmortem negative sudden death in 1 to 40-year-olds. Heart Rhythm 2011;8:412–19

This experience is similar to that in the Netherlands.[105]

Eddy and co-authors from Skinners group in New Zealand showed in a 2008 paper that the contemporary genetic tests failed to identify the genetic defects in about 10% of patients with LQTS.[106] That is, a patient could clinically be determined to have LQTS on ECG testing, but that genetic testing could not identify the genetic defect(s). This means:

i) there are probably other genes involved which are yet to be identified; and
ii) people could die of LQTS (for example, and be diagnosed with SIDS, or with an undetermined cause of death) and existing genetic testing would not pick up the genetic abnormality.

Complex interactions litter the field. Sun et al showed a significantly higher incidence of a particular genetic variation amongst patients who developed Long QT Syndrome when on a particular drug for another form of arrhythmia. It is open, but genetic variations may expose particular individuals in particular situations to the development of significant arrhythmias.[107] This has obvious implications in understanding Sudden Unexpected Death in Infancy (SUDI)/SIDS. Subtle, or even not so subtle stressors, which would be handled by most individuals may trigger a genetically determined potential to develop an arrhythmia.

An international consensus statement published late in 2013[108] on the diagnosis and management of patients with inherited primary arrhythmia syndromes contains valuable background information on these conditions:

- Long QT Syndrome. The first three genes responsible for LQTS were identified in 1995. Since then, a total of 13 genetic forms of congenital LQTS caused by mutations in genes encoding potassium channel proteins, sodium channel proteins, calcium channel related factors and membrane adaptor proteins have been revealed. Up to 15-20% of patients with LQTS remain genetically elusive. The fact that sudden death may be the first manifestation represents the main rationale for the treatment of asymptomatic patients. The conditions associated with arrhythmic events are to a large extent gene specific with most arrhythmic events occurring during physical or emotional stress in LQT1, at rest or in association with sudden noises in LQT2 patients and at rest or during sleep in LQT3 patients. Some patients with the gene defects do not have prolonged QT intervals but are

[105] van der Werf C, Hofman N, Tan HL, et al. Diagnostic yield in sudden unexplained death and aborted cardiac arrest in the young: the experience of a tertiary referral center in The Netherlands. Heart Rhythm 2010;7:1383–9.
[106] Eddy CA, MacCormick JM, Chung SK, et al. Identification of large gene deletions and duplications in KCNQ1 and KCNH2 in patients with long QT syndrome. Heart Rhythm 2008;5:1275–81
[107] Sun Z, Milos PM, Thompson JF, et al. Role of a KCNH2 polymorphism (R1047 L) in dofetilide-induced Torsades de Pointes. J Mol Cell Cardiol 2004;37:1031–9
[108] Priori SG, Wilde AA, Horie M, et al. HRS/EHRA/APHRS expert consensus statement on the diagnosis and management of patients with inherited primary arrhythmia syndromes: document endorsed by HRS, EHRA, and APHRS in May 2013 and by ACCF, AHA, PACES, and AEPC in June 2013. Heart Rhythm 2013;10:1932–63

regarded as having the condition. Patients are stratified according to risk and treatments include beta blockers, implantable cardioverter-defibrillators, left cardiac-sympathetic denervation.

- Brugada Syndrome. Therapies include lifestyle changes (avoid certain drugs, avoid excess alcohol), avoid anti-pyretics; cardioverter-defibrillator implantation; quinidine; isoproterenol;). Brugada is 8-10 times more prevalent in males than females. (This may involve aspects other than genetics eg higher testosterone levels). Inheritance is autosomal dominant. There are 12 responsible genes reported so far. All 12 genotypes involve either decreased inward sodium or calcium currents, or increased outwards potassium currents. Clinical manifestations: VF or aborted SCD (more often during the night than the day); syncope; nocturnal agonal respiration; palpitations; chest discomfort. These occur during rest or sleep; during a fever or with vagotonic conditions; rarely during exercise. Mean age of sudden death is 41 +/- 15 years.
- Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT). This is a rare arrhythmogenic disorder characterized by adrenergic-induced (ie adrenaline) bidirectional and polymorphic ventricular tachycardia. Resting ECG is normal and the otherwise unexplained arrhythmia is induced by exercise or other adrenergic stimulus. Treatment: Lifestyle changes are recommended: avoid competitive sports; avoid strenuous exercise; limit exposure to stressful environments; beta blockers; ICD implantation; flecainide; left cardiac sympathetic denervation. Inheritance is mainly autosomal dominant; there is a rarer autosomal recessive form. Family screening is recommended to be mandatory.
- Short QT Syndrome (SQTS). One of the rarer cardiac channelopathies. DNA variants in three potassium channel genes have been described. Mutations in the same three genes are also linked with three variants of LQTS. Treatment: cardioverter-defibrillator implantation; quinidiene; sotalol.
- Early Repolarization Syndrome. A primary cause of sudden death or a cause in conjunction with concurrent cardiac disease; may turn out to be not so uncommon. The relevant ECG pattern occurs in 1-13% of the general population and in 15-70% of idiopathic ventricular fibrillation cases. In the paediatric age group it is even more prevalent. 70% are male. There is some overlap with Brugada: 11-15% of Brugada patients have the ECG changes of ER Syndrome also. ER also seen in SQTS. There is obviously some difficulty in untangling the high prevalence of the ECG changes from the disease causing syndrome. So ECG pattern with demonstrated genetic change, family history or clinical history (eg syncope) generally required. Life threatening arrhythmias often the first and unexpected manifestation of ER Syndrome. Treatment: ICD implantation; isoproterenol; quinidine;
- Progressive Cardiac Conduction Disease (PCCD). Heterogeneous disorder of uncertain aetiology. Can be serious and potentially life threatening. Underlying mechanism can be functional or structural. Discovery of gene mutations causally involved in PCCD is relatively recent (2005). Can be associated with structurally abnormal heart. Treatment: cardiac pacemaker implantation; ICD implantation.
- Unexplained cardiac arrest: Idiopathic Ventricular Fibrillation (IVF; cardiac, respiratory, metabolic and toxicological aetiologies excluded by clinical evaluation). Genetic screening triggered by suspicion. Treatment: ICD implantation; quinidine; ablation of Purkinje potentials; all first degree relatives should be evaluated – resting ECG, exercise

stress testing and echocardiograph. In the CASPER registry of cardiac arrest survivors, in whom overt coronary and structural disease had already been excluded, 44% remained without a diagnosis after further comprehensive evaluation. In IVF there is by definition no evidence for pathogenesis.

- Unexplained Sudden Cardiac Death: Sudden Unexplained Death Syndrome (SUDS)is the term recommended for an unexplained sudden death occurring in an individual older than one year of age. Such a death with a negative pathological and toxicological assessment is recommended to be called Sudden Arrhythmic Death Syndrome (SADS). In relation to infants less than one year of age who have an unexplained sudden death with a negative pathological and toxicological assessment, the statement recommends the use of the term Sudden Unexplained Death in Infancy (SUDI). "The investigation of family members of cases of SUDI deaths often occurs on an ad hoc basis yet there are little data on its yield. Molecular autopsy identifies a lower burden of ion channel disease in SIDS compared to SUDS and there is a greater likelihood of sporadic genetic disease as a cause of sudden death in infancy. It is therefore likely that the yield of clinical evaluation of first degree relatives will be significantly lower than in SUDS. Nonetheless, if there is a positive molecular autopsy result, a family history of other cases of SUDI, SUDS or premature unexplained sudden death or of inherited heart disease then the yield is likely to be greater and familial evaluation more worthwhile."[109]

Earle and co-authors in their 2013 paper represent the increasing momentum for developing clinical registries of LQTS, as an alternative to community ECG screening, using the argument that for each identified person with LQTS there are 2-3 pre-symptomatic individuals identified in the family.[110]

The preceding information about inherited cardiac arrhythmias is not only important in itself, but also evidence of the continual discovery of new knowledge in medicine. In this instance the new knowledge is about deaths which would otherwise be regarded as sudden and unexplained. In relation to SIDS in particular, these would be deaths where smothering would lurk as a dark possibility. We did not really know of these conditions10 or so years ago. This progress will not stop, and more will be discovered about these and other conditions causing infant deaths. This needs to be taken into account when assessing the cause of the Folbigg's deaths.

Consider the consequences of regarding the state of knowledge as frozen, meaning that we should not take into account future developments in medicine when we consider the cause of the Folbigg deaths. Such an approach would have been a justification, prior to the identification and definition of SIDS as a syndrome, for concluding homicidal suffocation in many of those cases. This would have been on the basis of a diagnosis of exclusion (nothing found at autopsy and it

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[109] Priori SG, Wilde AA, Horie M, et al. HRS/EHRA/APHRS expert consensus statement on the diagnosis and management of patients with inherited primary arrhythmia syndromes: document endorsed by HRS, EHRA, and APHRS in May 2013 and by ACCF, AHA, PACES, and AEPC in June 2013. Heart Rhythm 2013;10:1951

[110] Earle N, Crawford J, Smith W, et al. Community detection of long QT syndrome with a clinical registry: an alternative to ECG screening programs? Heart rhythm 2013;10:233–8

being known that suffocation may leave no signs) where the mother was alone with the baby at around the time of the death and for some reason not believed. This would clearly have been wrong.

Some of the above may open up questions about how forensic pathology goes about concluding cause of death in general. I have included in Appendix 4 some material bearing on how a pathologist goes about this, and some related material showing that causes of death, especially in more difficult cases, are not highly reproducible between pathologists.

CHAPTER XI

THE FORENSIC PATHOLOGY CONCLUSION ABOUT THE FOLBIGG DEATHS

In my view, it is wrong on the forensic pathology evidence available in this case to conclude that one or more of the Folbigg children are the victims of a homicide. There is no merit in forcing certainty where uncertainty exists. The very existence of the enigma of SIDS demonstrates how little we know about why some babies die. It is not for a pathologist or physician to conclude that a number of infant or childhood deaths, with no significant injuries or related findings, are homicides on the basis of controversial circumstantial grounds.

If the convictions in this case are to stand, I want to clearly state there is no pathological or medical basis for concluding homicide. The findings are perfectly compatible with natural causes. The findings cannot rule out smothering in one or more of the cases, but especially in the case of Laura, not only is there an acceptable natural cause of death easily visible, it is important that absolutely no signs of asphyxia or compression of the face are present.

In my view, the correct way to view this case – as mentioned above - is not as four unexplained deaths and an unexplained ALTE.

Considered separately:

- Caleb's death is best regarded as SIDS (Category II)
- Patrick's ALTE is unexplained
- Patrick's death is an unsurprising consequence of the state he was left in following the ALTE
- Sarah's death is best regarded as SIDS (Category II)
- Laura's death has been caused, unexceptionally, by myocarditis.

In other words, as a collective, it is a case of two deaths and an ALTE which are unexplained. In considering the deaths of Caleb and Sarah, and Patrick's ALTE, one needs to consider the recent developments in cardiac arrhythmias (developments still to be fully realized), the likelihood that multiple SIDS in one family are due to natural causes, and the role of conditions yet to be discovered which will further unravel the enigma of Sudden Infant Death Syndrome. One should avoid the trap of concluding that, because the deaths and ALTE are unexplained, the default explanation is homicide. While both situations are rare, two or three unexplained natural deaths in the one family probably occur more frequently than the same number of hidden homicides.

Whenever a forensic pathologist in this case says it is possible that one or more of the Folbigg children was smothered[111], s/he must also say in the very same breath that

- there is nothing from a forensic pathology viewpoint to suggest that any of the children have been killed,
- it is surprising that in five alleged smothering events there are no signs of smothering,
- in Laura's case there are good grounds for concluding that she was not smothered, and that
- from a medical viewpoint, there are identifiable natural causes of death for two of the children and natural causes - whether inherited or not, whether known or yet to be discovered - is a plausible explanation for the other two deaths and the ALTE.

If the convictions are to stand, they must do so without the support of forensic pathology, and in Laura's case at least, against the forensic pathology view.

[111] It is worth pointing out that at no point have any of the pathologists or physicians put the homicide hypothesis as 'one or more of the Folbigg children were smothered'. It was always a binary proposition: all four or none. However, it seems logical, leaving aside the legal imperatives, that with the combination of the existence of SIDS, the newly characterized arrhythmic diseases, the pathological explanations for Laura's and Patrick's deaths, and the knowledge that smothering can occur without leaving signs, one must necessarily agree that it is possible, for example, that one child was smothered and the others died naturally. Of course, as soon as that is agreed, it is not possible to say which one was smothered, and at that point it has to be accepted that they all could well have died from natural causes.

APPENDIX 1

An audit of the four Folbigg autopsies against each other, the National SIDS Autopsy Protocol (1992) and the VIFM SUDI Minimum Standards (2012)

PM Procedure and/or data item				Folbigg PMs 1989-1999				National SIDS Autopsy Protocol (1992)	Current VIFM SUDI investigation standard (2012-14)
				Caleb Gibson Folbigg, 20/02/1989	Patrick Folbigg, 13/02/1991	Sarah Kathleen Folbigg, 31/08/1993	Laura Folbigg, 01/03/1999		
Details of patient	Identification			X	X	Identified by Const Saunders of Maitland police station Wristband ID E.45824	X	X	✓
	Name			✓	✓	✓	✓	X	✓
	Age			✓	✓	✓	✓	X	✓
	Sex			M	M	F	F	X	✓
	Date of birth			X	✓	X	X	X	✓
	Date and time of death			X	✓	✓		X	✓
	Date and time of PM			✓	✓	✓	✓	X	✓
Circumstances of the death	Form 83			X	X	X	X	X	✓
	Police Investigative Checklist for Sudden Infant or Child Deaths			X	X	X	X	X	✓
	Other sources			Clinical history	Clinical history	X	Clinical history		✓
	Scene investigation			X	X	X	X	X	+/- as required
Radiology	Skeletal survey			X	X	X	X	Recommended	+/-✓
	CT scan			X	X	X	X	X	✓
Photography	Whole body on admission			X	X	X	X	X	✓
	Scene photography should be reviewed if available.			X	X	X	X	X	✓
Weights and measures	Body weight			✓	✓	✓	✓	✓	✓
	Crown – heel length			✓	✓	✓	✓	✓	✓
	Crown – rump length			X	✓	✓	X	✓	✓
	Head circumference			X	✓	✓	✓	✓	✓
	Chest circumference			X	X	✓	✓	✓	✓
	Abdominal circumference			X	X	X	✓	✓	✓
	Intercanthal distance			X	X	X	X	✓	✓
	Foot length			X	✓	X	✓	X	✓
	Subcutaneous fat chest			X	X	X	X	✓	✓
	Subcutaneous fat abdomen			X	X	X	X	✓	✓
External examination	General	Identifying marks		✓ Hair, eye colour	X	X	X	Jaundice, pallor, cyanosis	✓
		Signs of medical intervention		X	X	✓	0	✓	✓
		Signs of decomposition		X	X	X	X	✓	✓
		Lividity		✓	X	✓	✓	✓	✓
		Rigor mortis		✓	X	✓	✓	X	✓
	Head	Configuration		X	X	X	X	✓	✓
		Scalp and Hair		✓	X	X	X	X	✓
		Fontanelles		X	X	X	X	✓	✓
		Eyes	Colour; cataracts; icterus; Petechiae; other abnormalities	✓ colour	X	X	X	✓ Conjunctival petechiae, sclera	✓
		Ears	Setting; rotation; discharge; other abnormalities	X	X	X	X	✓ Shape, meati	✓
External examination (continued)		Nose	Discharge; Septum; choanal atresia; other abnormalities	X	X	X	✓	✓ Shape, nares, chloanae	✓
	Head (continued)	Mouth	Discharge; labial frenulum; teeth (number upper and lower; abnormalities); mandible	X	X	✓ Lips, frenulum, abrasions, uvula mentioned	✓ Lips, frenulum	✓ Contents, buccal mucosa, gums, teeth	✓
		Tongue	Size; frenulum	X	X	✓ presence and frenulum noted	✓ frenulum noted	✓	✓

		Palate	Cleft; high arched; other abnormalities	X	X	X	X	✔	✔
	Neck	Bruising; abrasion or laceration; other abnormalities		X	X	X	✔	X Check skin in general	✔
	Chest	Bruising; abrasion or laceration; other abnormalities		X	X	X	X	✔ shape, breast development	✔
	Abdomen	Distension; umbilicus; hernias		X	X	X	X	✔ Shape, umbilicus, herniae	✔
		Bruising; abrasion or laceration; other abnormalities		X	X	X	X	✔	✔
		Spine/midline skin of back		X	X	X	X	✔ Overlying skin	✔
	External genitalia			X	X	X	✔	✔ Normality, testes in scrotum	✔
	Anus			X	X	X	X	X	✔
	Limbs	Bruising; abrasion or laceration; other abnormalities		X	X	✔	✔	✔ development, creases, nails, digits	✔
		Hands and Feet	Digits; nail beds; fluff in palmar creases	X	X	✔ Palmar/plantar creases	X	✔	✔
Internal examination	Soft tissues and bone of face and neck	Haemorrhage		X	X	X	0 Facial dissection	✔ Abrasions, bruises, congenital abnormalities	✔
		Hyoid bone		X	X	X	X	✔	✔
	Upper aerodigestive tract	Epiglottis		X	X	✔	X	✔	✔
		Larynx		X	X	✔	X	✔	✔
		Trachea	Stenosis; exudates/haemorrhage	X	X	✔	X	✔	✔
	Cardiovascular system	Pericardium	Surface; petechiae (visceral/parietal); fluid (amount)	✔	✔	✔	X	X	✔
		Heart	Weight	✔	✔	✔	✔	✔	✔
			Valve circumferences	X	X	X	X	X	✔
			LV thickness (10 mm inferior to AV groove)	X	X	X	X	X	✔
			RV thickness (10 mm inferior to AV groove)	X	X	X	X	X	✔
			Cardiac situs	X	X	✔	X	✔	✔
			Chamber blood (fluid/clotted)	✔	X	X	X	✔ chamber size	✔
			Ventricular inflow/outflow tracts	X	0 ventricles examined	X	X	✔ not specified	✔
			Valves (Structure; vegetations; cordae)	✔	✔	✔	X	✔	✔
			Myocardium	✔	✔	✔	✔	✔	✔
			Ductus arteriosus	0	0	0	X	✔	✔
			Coarctation	0	0	0	X	✔	✔
			Coronary arteries (ostia; course)	0	✔	0	X	✔	✔
			Interatrial/interventricular septa	X	0	✔	X	✔	✔
			Pulmonary veins	X	✔	✔	X	✔	✔
			Other abnormalities						✔
Internal examination (continued)	Respiratory system	Pleural cavities	Surfaces; parietal pleura should be stripped	✔	X	✔	X	X	✔
			Fluid/blood (amount)	X	X	✔	X	✔	✔
		Bronchi	Structure; contents; mucosa	✔	0 Frothy mucoid secretion	0 Gastric contents	✔	✔	✔
		Lungs	Weights	✔	✔	✔	✔	✔	✔
			Lobation/situs	X	X	X	X	✔	✔
			Visceral pleural surfaces	✔	✔	0	X	✔	✔
			Parenchyma	✔	✔	✔	✔	✔	✔
			Congestion/haemorrhage	0	✔	✔	✔	✔ cut surface	✔
			Oedema	0	0	✔	X	✔ cut surface	✔

			Consolidation	0	0	0	X	✓ cut surface	✓
			Other abnormalities				Soft palate, strap muscles for bruising		✓
	Alimentary system	Peritoneal cavity	Surfaces	0	✓	X	X	X	✓
			Fluid/blood	0	X	✓	X	✓	✓
			Diaphragm (intact; arched bilaterally)	0	✓	X	✓	X	✓
		Oesophagus	Mucosa	✓	X	X	✓	✓	✓
			Tracheo-oesophageal fistula	X	X	0	X	X	✓
			Contents	X	X	X	X	✓	✓
		Stomach	Contents	✓	X	✓	✓	✓	✓
			Mucosa	✓	X	✓	✓	✓	✓
		Small and large bowel	Rotation	X	X	✓	0	X	✓
			Serosal surfaces	0	X	0	0	X	✓
			Appendix	✓	X	✓	X	X	✓
			Contents	0	X	0	✓	✓	✓
			Mucosa	✓	X	0	✓	✓	✓
		Mesentery		0	X	X	X	X, LN mentioned	✓
		Liver and Gallbladder	Weight	✓	✓	✓	✓	✓	✓
			Gross lobation	0	0	✓	0	✓	✓
			Capsule	0	0	✓	0	✓	✓
			Parenchyma	0	✓	✓	✓	✓	✓
		Pancreas		✓	✓	✓	X	✓	✓
	Reticuloendothelial system	Thymus	Weight	✓	✓	✓	✓	✓	✓
			Outline/surface anatomy	0	0	0	✓	X	✓
			Capsule (Petechiae)	0	0	✓	✓	✓	✓
			Parenchyma	✓	✓	✓	✓	X	✓
		Spleen	Weight	✓	✓	✓	✓	✓	✓
			Surface anatomy	0	0	0	X	✓	✓
			Capsule	0	0	0	X	✓	✓
			Parenchyma	0	0	✓	✓	✓	✓
		Mesenteric lymph nodes		X	X	✓	✓	✓	✓
		Other lymph nodes		✓	X	✓	X	✓	✓
		Bone marrow	Colour	X	✓	✓	✓	✓	✓
Internal examination (continued)	Genitro-urinary system	Kidneys	Weights	✓	✓	✓	✓	✓	✓
			Surface anatomy	0	0	✓	0	✓	✓
			Capsules	0	0	✓	0	✓	✓
			Parenchyma	✓	0	✓	0	✓	✓
			Pelvicalyceal system	✓	✓	✓	0	✓	✓
			Ureters	✓	✓	✓	0	✓	✓
			Retroperitoneal/perirenal fatty tissue	X	X	X	X	X	✓
		Bladder	Contents	0	✓	✓	✓	✓	✓
			Mucosa	✓	✓	✓	0	✓	✓
		Genitalia	Ovaries; tubes; uterus	N/A	N/A	✓	✓	✓	✓
			Testes and prostate	X	X	N/A	N/A	✓	✓
	Endocrine system	Thyroid		✓	✓	✓	✓	✓	✓
		Adrenal glands	Weight (combined)	X	✓	X	✓	✓	✓
			External appearance	✓	0	X	✓	X	✓
			Parenchyma (cortex and medulla)	✓	✓	X	✓	X	✓
		Pituitary		✓	✓	X	✓	✓	✓
	Central nervous system	Whole brain weight (unfixed)		✓	✓	✓	✓	✓	✓
		Skull		✓	✓ Formal	✓	✓	✓	✓

		Meninges		✔	neuropathological examination performed post-fixation	✔	✔	✔	✔
		Dura, falx and tentorium		X		0 Dura mentioned only	0	✔	✔
		Sinuses: patency		X		X	X	✔	✔
		Cerebrum		X		✔	✔	X	✔
		Midbrain/brainstem		X		✔	✔	X	✔
		Cerebellum		X		✔	✔	X	✔
		Spinal cord		X		0 Cervical cord	0 Cervical cord	Op	✔
		Middle ears		X	X	✔	X	X	✔
	Musculoskeletal system	Muscle development		X	✔	X	✔	✔	✔
		Ribs, skull & vertebrae	Fractures	X	X	X	✔	✔	✔
		Long bones	Fractures	X	X	X	✔	✔	✔
ADDITIONAL INVESTIGATIONS	Histopathological examination (Mandatory sections)	Thymus		✔	✔	✔	✔	SR	✔
		Lymph node		✔	X	✔	X	X	✔
		Epiglottis (vertical)		X	X	X	X	SR	✔
		Larynx		X	X	✔	X	X	✔
		Supraglottic transverse		X	X	0 Uvula	X	C to soft palate	✔
		True cords transverse		X	X	X	X	SR	✔
ADDITIONAL INVESTIGATIONS (continued)	Histopathological examination (Mandatory sections continued)	Trachea/thyroid transverse		X	✔ thyroid only	✔	✔	✔ Must sample trachea, thyroid strongly recommended	✔
		Lungs, all lobes; Perls Prussian Blue on all lung sections.		✔ lobes analysed not specified, Perls performed	✔ Left lungx2	✔ lobes analysed not specified, Perls performed	✔ lobes analysed not specified, Perls performed	✔ except RML, pleura and bronchi, Perl's not specified	✔
		Diaphragm		X	✔	✔	✔	X	✔
		Heart	Septum	X	X	X	X	C to A-V sulcus	✔
			R and L ventricles	0 " myocardium" analysed not specified	0 myocardium analysed not specified	0 myocardium analysed not specified	✔ Myocarditis	✔ Must sample ventricular only, not specified to take R & L	✔
			AV node	X	✔	X	X	X	✔
		Oesophagus (distal 30 mm)		X	X	X	✔	X	✔
		Terminal ileum		X	✔	0 "Intestinal"	0 Small intestine	C to small bowel	✔
		Rectum		X	X	0 "Intestinal"	X	X	✔
		Liver		✔	✔	✔	✔	SR	✔
		Pancreas with duodenum		X	✔	✔ no duodenum	✔ pancreas	X	✔
		Spleen		✔	✔	✔	✔	SR	✔
		Kidney with capsule		✔ Didn't specify capsule	✔ didn't specify capsule	✔ Didn't specify capsule	✔ Didn't specify capsule	SR (capsule not specified)	✔
		Adrenal glands		✔	0	✔	✔	SR	✔
		Rib with costo-chondral junction		X	X	X	✔ Bone marrow, rib	X	✔
		Submandibular gland		X	X	✔ and Parotid	✔	SR	✔
		CNS Neuropathology referral of the brain for examination with sampling of all levels of the brainstem, both cerebellar hemispheres and vermis; bilateral cortical samples to include all three vascular territories and watershed zones, periventricular white matter, thalami, basal ganglia and hippocampi + any macroscopic 96bnormalities; dura; samples of thoracic cord (sectioned transversely)		X	Brain and spinal cord fixed for neuropathological examination: Encephalopathy leading to seizures	Brain and higher cervical cord fixed for neuropathological examination: Normal histology, 4 blocks sampled, not specified	Brain and spinal cord fixed for neuropathological examination: Multiple sections examined, 16 blocks, site sampled not specified.	✔ Must sample Cortes, meninges, medulla, cerebellum, thalamus.	✔
	Histopathological examination (Discretionary sections)	Supraglottic soft tissue		X	X	✔ tongue	X	X	✔
		Lung hilum		X	X	X	X	SR	✔
		Pancreatic tail		X	X	X	✔ pancreas	SR	✔
		Mesentery		X	✔ LN	X	X	X	✔
		Stomach		X	✔	X	✔	X	✔
		Colon		X	✔	0 "Intestinal" section only, site not specified	✔	C	✔
		Appendix		X	X	✔	X	X	✔

		Ovaries / testes	X	✔ testis	✔ ovary	✔ Ovary	C	✔
		Bladder	X	X	X	X	X	✔
		Psoas muscle; peripheral nerve	X	✔ "Muscle" only	X	✔ Skeletal muscle	C	✔
		Tonsil	X	X	✔	X	X	✔
	Toxicology	Blood	X	✔ Tegretol and phenobarbitone levels	✔	✔	Not specified, "where this is performed"	✔
		Stomach contents	✔	X	✔	✔		✔
		Liver	✔	X	✔ and bile	✔		✔
ADDITIONAL INVESTIGATIONS (continued)	Bacteriology	Blood	X	✔	"Representative samples for microbiology, not specified	✔	✔	✔
		CSF	X	X		✔	✔ only if macroscopic evidence of inflammation	✔
		Lung swabs (left and right)	X	✔ Lung tissue	✔ Lung grew Streptococcus profuse + staphylococcus scanty +	✔ Lung tissue	✔ Right and left not specified	✔
		Liver swab	X	X	"Representative samples for microbiology", not specified	X	✔ gut swab only if macroscopic evidence of inflammation	✔
		Spleen swab	X	X		✔ Spleen tissue	X	✔
		Middle ear swabs (left and right)	X	X		X	✔ only if macroscopic evidence of inflammation	✔
	Virology	Nasopharyngeal aspirate	X	✔ Blood and lung sampled for virology, R/L lung not specified	X		✔ take in accordance with recommendation from local lab	✔
		Bowel contents (small/large)	X		X	✔ (Culture for bacteriology)	✔ take in accordance with recommendation from local lab	✔
		CSF	X		✔ for biochemistry not virology	X	✔ take in accordance with recommendation from local lab	✔
		Lung tissue (left and right)	X		0	X	✔ take in accordance with recommendation from local lab	✔
		Meningeal swab/Cerebral tissue for viral pcr (if encephalitis considered)	X		X	X	✔ take in accordance with recommendation from local lab	✔
	Biochemistry/ electrolytes	Vitreous fluid	X		✔ Liver and CSF for biochemistry	X	MB	✔
		Blood (C-reactive protein if considered useful)	X	✔ FBC	X	X		✔
	Metabolic screen	Bile	X	Heart tissue, Liver tissue, Kidney tissue and Muscle tissue for metabolic studies.	X	X	Enzyme assay May be performed	✔
		Skin	X		X	X		✔
		Blood spot (Guthrie card)	X		X	X		✔
	Additional samples	Electron microscopy. This should be considered in cases where metabolic diseases are possible or likely. Glutaraldehyde has to be ordered prior to autopsy from Anatomical Pathology, Royal Children's Hospital. Fragments for glutaraldehyde fixation should be 1 mm^3. Material should be	X	Heart tissue Liver tissue Kidney tissue Muscle tissue	X	X	X	✔

		submitted only if there is a specific question of metabolic or mitochondrial disease.						
		EDTA tube of blood for karyotyping	X	✓ Blood and Skin sampled for chromosome analysis	X	X	Cytogenetics MB	✓
		EDTA tube of blood for molecular genetics screen (see table below for genes screened	X	X	X	X	X	✓
		Frozen tissue. Consideration should be given to freezing and retaining fragments of liver, heart and voluntary muscle to allow the possibility of subsequent investigation of metabolic diseases.	X	Urine, tissue snap frozen	X		MB	✓
		Other	Acyl carnitine Amino acid profile Phenylketonuria Medium chain Acyl CoA dehydrogenase	Skin sampled for fibroblast culture Rectal biopsy for neuronal inclusions Serum ammonia, calcium, magnesium, glucose White cell enzymes analysis for adrenoleukodystrophy, Refsum's disease, Zellweger's syndrome, peroxisomopathies Long chain fatty acid studies Urine mucopolysaccharide screen Serum carnitine Urine amino acids, methylmalonic acid, organic acids, lactic acid Arterial blood lactate Anti-nuclear antibodies TORCH screen Leukocyte inclusions. ECG	Spleen for DNA Acyl carnitine Amino acid profile Phenylketonuria Medium chain Acyl CoA dehydrogenase	Urine for tox Rectal swabs for culture Acyl carnitine Amino acid profile Phenylketonuria Medium chain Acyl CoA dehydrogenase	Fat stain on frozen section of Heart, liver, kidney, adrenal if sampled	Referral for cardiology genetics screening and counselling
Cause of death (as given at the time)			Sudden infant death syndrome	Ia Asphyxia due to airways obstruction (due to) Ib Epileptic fits (due to) Ic Encephalopathic disorder (the underlying cause of encephalopathy not determined on investigation)	Sudden infant death syndrome	Unascertained		
Applicable standard specified							*"The autopsy in this case incorporated all the observations recommended in the National SIDS Autopsy Protocol"*	

LEGEND: Post mortem procedure list = VIFM 2014 SIDS Investigation Standard, **X** = Not mentioned, ✓ = Mentioned specifically, 0 = subset of general statement only, M = Male, F = Female, SR = Strongly recommended, Op = Optional , C = Consideration given, MB = May be performed

APPENDIX 2

Gene testing for genetic heart conditions at the Victorian Clinical Genetics Service. Information for referring practitioners.
(Attached as a separate PDF file).

APPENDIX 3

VIFM Case 812/02 Smothering of a two year old boy following forced administration of a drug.
(Attached as a separate PDF file).

APPENDIX 4

Photomicrographs of sections of Laura's heart showing myocarditis.
(Attached as a separate pdf file)

APPENDIX 5

Chapter 4—Establishing the Cause and Manner of Death: The Hidden Controversy[112]

Introduction

The attribution of "cause" in forensic pathology is a fascinating subject about which too little has been written. Pollanen has made a significant recent contribution. It is one particular area, amongst many, of confusion at the dynamic interface of law and medicine. One reason for this confusion is that those involved have to grapple with (at least) two different conceptions of cause: that used in medicine and that used in law. This chapter has been written from a medical perspective, which is clearly all the authors can try to do.

The chapter begins by outlining the terrain covered by forensic pathology and the main features in the landscape. It then briefly looks at the form in which the cause of death is certified. The main aim is to arrive at something like a coherent schema for the attribution of the cause of death following autopsy. While in most cases this is straightforward, in many cases in pediatric forensic pathology it is not, and in those cases there is not a completely uniform approach. This is not surprising as the attribution of cause, particularly in retrospect and in as complex and unpredictable field as biology, is essentially a philosophical question. During the course of this chapter, examples of the issues arising in a number of cases will illustrate the breadth of the

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[112] Pediatric Forensic Pathology: Limits and Controversies. Cordner S, Ehsani J, Bugeja L, Ibrahim J. Independent Research Studies. Roach K, Director. Queens Printer for Ontario. 2008. Commissioned by The Inquiry into Pediatric Forensic Pathology. Ontario, Canada. http://www.attorneygeneral.jus.gov.on.ca/inquiries/goudge/policy_research/pdf/Limits_and_Controversies-CORDNER.pdf (References from this chapter not included in the Appendix)

forensic pathologist's contribution and the complexity of the associated causal issues, which, it must be said, are sometimes not appreciated by either pathologists or lawyers.

Forensic Pathology—The Terrain

Forensic pathology is the application of the principles and practice of pathology to the needs of the courts, or more generally, the law. Pathology is the study of disease. Anatomical pathology, its largest subdiscipline, has left its morbid anatomical roots and concentrates in the main on surgical and biopsy pathology. Forensic pathology, closely allied to or even derived from anatomical pathology, is a mortuary-based specialty that provides the knowledge basis for the performance of autopsies in deaths reported to Coroners, or their equivalents.

It is important to appreciate, at the outset, the scope of the forensic autopsy. The aims of this part of a death investigation are:

1. To discover, describe, and record all the pathological processes present in the deceased and, where necessary, the identifying characteristics of the deceased;
2. With knowledge of the medical history and circumstances of the death, to come to conclusions about the cause and time of death and factors contributing to death and, where necessary, the identity of the deceased;
3. In situations where the circumstances of death are unknown or in question, to apply the autopsy findings and conclusions to the reconstruction of those circumstances. This will, on occasions, involve attendance at the scene of death, preferably with the body in situ; and
4. To record the positive, and relevant negative, observations and findings in such a way as to enable another forensic pathologist at another time to independently come to his or her own conclusions about the case. As forensic pathology is essentially a visual exercise, this involves a dependence on good quality, and preferably colour, photographs.

Encapsulated in this approach to the forensic autopsy are two consequences at odds with a common perception of the specialty. Firstly, forensic pathology could be regarded as the "what happened" specialty (and not the "whodunnit" specialty). It is as part of this that the pathologist is concerned with coming to the best conclusion about the cause of death. However, and this is the second consequence, in pursuit of answers to "what happened," conclusions about the cause of other findings on or in the body, or at the scene, or of events described in witness statements, may require the pathologist to attribute "cause" in areas other than the cause of death. Provided the pathologist keeps to his or her expertise, this is a quite proper exercise.

Certification of the Cause of Death

There is a preliminary area that needs a little exploration, which is the formality of framing the cause of death.

Table 21—Cause of Death

1	
Disease or condition directly leading to death.**	(a)
Antecedent causes (morbid conditions, if any, giving rise to the above cause, stating the underlying condition last).	(b)
2	
Other significant conditions (contributing to the death but not related to the disease or condition causing it).	

** This means the disease, injury, or complication that caused death, NOT ONLY, for example, the mode of dying such as "heart failure, respiratory failure," etc.

One needs to understand the form of the cause of death to appreciate what is being conveyed. The cause is divided into 1 and 2. "1" is the direct cause of death or the disease or condition directly leading to death. "1" is subdivided, if necessary, into a, b, and c (and theoretically d and e, etc., if necessary; it very rarely is). "1a" is due to or a result of "1b" and so on. The last listed condition under "1" is the main, central or underlying cause of the patient's death. "2" represents those other significant conditions contributing to the death but not directly related to the disease or condition causing it.

This is the internationally accepted form that favours singular particular causes of death (which makes coding, classification, and statistics easier) and does not invite multiple interacting causes. There is an ill-defined distinction drawn between cause ("1a, b" above) and contribution ("2" above) that is essentially arbitrary where the latter is some form of lesser cause of death acting independently of the main cause of death.

The cause of death is not often a contentious issue in litigation, and when it is, there will almost always have been an autopsy. It is to these cases that we now turn.

Deciding the Cause of Death following Autopsy

Deciding the cause of death is a fundamental responsibility for all anatomical pathologists after autopsy. The responsibility is greatest for forensic pathologists, yet very little has been written about the criteria that need to be satisfied to make a decision. This issue causes confusion in some court cases because both pathologists and lawyers fail to appreciate something of the philosophy of causation. Leaving aside the minority of cases where the lesion observed at autopsy is incompatible with life (e.g., decapitation), what in fact usually happens in coming to a conclusion is that *a* cause of death discovered at autopsy, which accords with the medical history and circumstances, is elevated to *the* cause of death. In general terms, the pathologist makes a decision that a certain autopsy finding is capable of leading to death, and that as this is consistent with the deceased's medical history and the supposed circumstances of death, and there is no other competing cause, it is the cause of death. Such a conclusion about the cause of death is retrospective and therefore cannot generally be tested. This approach emphasizes the need to discover all the pathological processes present in the deceased before considering them in relation to the medical history and the circumstances of death. A corollary of this is that if there is no autopsy finding discerned that is capable of leading to death, the pathologist will be reliant completely upon the circumstances and medical history. To the extent these are likely to be disputed, or are inherently difficult to corroborate, the pathologist will need to be careful.

Causation: Philosophy and Problems

The authors are not philosophers, but have found the following to be a useful framework for discussion. David Hume (1711–1776), believed that for X to be the cause of Y, X must be both sufficient and necessary for the effect, Y: thus, X is always followed by Y, and Y never occurs unless X occurred. Somewhat differently, John Stuart Mill (1806–1873) thought that the cause was the sum total of the conditions in which an event occurred: it was not correct to isolate *one* of the conditions in which an event occurred as the exclusive cause. To Hume, the statement "*the rising of the sun causes daylight*" would have been reasonable, since the rising of the sun is always followed by daylight, and daylight never comes about unless the rising of the sun has occurred. The statement is, in fact, incomplete because daylight could not occur unless there was an atmosphere. Mill's approach would include an atmosphere in any statement about what caused daylight because it is one of the conditions in which the event occurs.

The restrictiveness of Hume can be seen in the commonest cause of death in the Western world: coronary atherosclerosis ("hardening of the arteries of the heart," "heart disease," "heart attack," "myocardial infarction"). The development of coronary atherosclerosis is not always followed by death, and death does not occur only when coronary atherosclerosis has developed. Yet clearly it is a reasonable proposition that coronary atherosclerosis has been the pathological basis for an enormous number of deaths. It seems that Hume's approach is suited more to those cases where the cause of death is incompatible with life, for example, decapitation. This is not to say that Mill's approach is necessarily the complete answer. Take the example of the heavy smoker who dies of carcinoma of the lung. One of the conditions in which the death occurred is smoking, but there are more: a person may smoke because of the effect of advertising, because of parents' smoking, because of particular personality traits, and so on *ad absurdum.*

Pathologists (and in some cases, courts) have to make a practical decision that cause stops somewhere.

In general, the line is drawn at the "medical" cause of death, but, as the example shows, this may be unsatisfactory: smoking is increasingly noted on death certificates.

Establishing the cause of death is heavily dependent upon an interpretation of the circumstances of death

This is a particularly important issue in pediatric forensic pathology, all the more so because it may be difficult to clearly establish what the circumstances are. Let us start with an adult case.

Mr. A. was a 29-year-old man with no known previous illnesses, was working with electrical machinery when he suddenly collapsed and died. His workmates thought he had been electrocuted, although others in contact with the same machine had felt no shock. The results of the examination of the machinery were controversial; government inspectors were saying that it was conceivable that the machine had been electrically alive, electrical engineers retained by the factory were saying it was not. At autopsy, there were no marks of electrocution. (It is quite possible to be electrocuted and for there to be no marks.) The only positive finding was appreciable hydrocephalus ("water on the brain," or dilatation of the ventricles of the brain) but no acute cerebral edema (or brain swelling due to intercellular fluid accumulation). Some basal meningeal thickening suggested the hydrocephalus may have been secondary to meningitis in infancy or childhood. There were no abnormal histological or toxicological findings. If it is assumed that uncomplicated hydrocephalus (as in this case) can cause sudden unexpected death (a matter of some dispute at the inquest), it is easy to see that the cause of death is completely dependent on the assessment of the machinery by electrical experts. Even the assessment of the circumstances contained causal issues because the inspectors, who said it was conceivable that the machine could become electrically live, could not say that it actually had been. For the purposes of discussion, let us consider this real case.

Baby A was 3 months old and left in the care of a local authority nursery. It was windy and snow was on the ground. She was left outside in a pram unattended for three hours. When she was fetched in, it was found that she was dead. There were no significant pathological findings at autopsy.

The definition of the Sudden Infant Death Syndrome (SIDS) is the sudden unexpected death of an infant during a period of supposed sleep in whom a thorough autopsy, and review of the medical history, scene, and circumstances of the death, fails to find any adequate cause for the death. The absence of any pathological findings in the case above puts this death in the category where SIDS would be considered, subject to a review of the medical history, scene, and the circumstances. It also needs to be understood that death from exposure (or, more technically, hypothermia—low body temperature) may also have no pathological findings.

Clearly, the circumstances of the death in this case, having been left outside in the wind on a freezing cold day, mean that hypothermia will have to be considered as a realistic cause of death. One approach would be for the pathologist to conclude that the cause of death was "unascertained," and then to have discussed in his/her report the extent to which the exposure may have been involved in this death by a consideration of the circumstances.

Another pathologist may have concluded that the cause of death was indeed "hypothermia" or was "consistent with hypothermia." There may have been information that only surfaced months after the death that in fact the nurse at the nursery took the baby's temperature soon after she was brought inside, and it was 28 degrees Celsius. This observation might be regarded as supporting death from hypothermia, but as a matter of fact cannot exclude death from some other (possibly natural cause) soon after being left outside, and the drop in temperature was simply cooling after death in a cold, windy environment.

Table 22—Possible approaches to the cause of death in the case of an infant left outside on a very cold day

Cause of Death	Comments
1 (a) Sudden Infant Death Syndrome	No. Hypothermia as a realistic possibility, based solely on the circumstances of the death, excludes a diagnosis of SIDS
1 (a) Hypothermia	The pathologist's opinion is that the cause of death is hypothermia. In his/her experience, the circumstances are such that hypothermia could explain the death, and in the absence of any competing cause of death, s/he believes it is reasonable to conclude that hypothermia indeed caused the death.
1 (a) Consistent with hypothermia*	As above, except that the pathologist believes that other, perhaps slight, possibilities cannot be excluded. For example, the baby might have had an underlying cardiac arrhythmia (e.g., long QT syndrome, or similar) that manifested itself when the baby was put in a stressful situation, or manifested itself spontaneously within minutes of the baby being left outside.
1 (a) Unascertained (Undetermined, or similar word)	The pathologist is not sufficiently confident that the circumstances can be held responsible for the death, because s/he is slightly surprised that, even though the pram was uncovered, the baby was well wrapped, and was several inches below the upper level of the pram such that the worst effect of the wind would not have been felt. The other slight possibilities assume greater significance in this pathologist's mind, and while hypothermia is discussed in the report as a possible cause of death, in the end the pathologist's opinion is that s/he cannot say.

* The use of the phrase "consistent with" generates its own controversy. The controversy sits between its technical usage by the expert, and the thought that a jury member might interpret the phrase as meaning "highly likely", "highly probably" or, even "is" hypothermia. Logically, "consistent with" carries with it the inference that the thing could be consistent with something else. It is in this latter sense that it is used here. Its usage in reports and in oral evidence should probably always be qualified by this explanation. One only has to consider that if this baby had been found dead at home, at 6 a.m. in her cot, with precisely the same autopsy findings, the cause of death would have been correctly given as SIDS. This is an example of dependence upon information about the circumstances. The pathologist in the mortuary, blind to the circumstances, would not be able to tell the difference between the two deaths.

1 (a) Unascertained in an infant left outside in a pram on a very cold day	Very similar to 1 (a) Unascertained, except that the pathologist wants to indicate formally that s/he believes the circumstances of being left out in the cold are implicated in the death.

Let us consider another actual case. D.L., a two-year-old boy who died following surgical evacuation of a subdural hemorrhage resulting from a head injury. The defence wanted to establish that after whatever injury caused the subdural hemorrhage there was a 6 hour delay (lucid interval) before the child deteriorated. The neurosurgeon involved did not believe that such an interval was a realistic possibility. (The existence of lucid intervals is a controversy in pediatric forensic pathology that has not been discussed in this paper). Notwithstanding that the accused admitted hitting the child while he was sitting in a car seat, he was acquitted because the pathologist discovered a previously existing old subdural hemorrhage that complicated the assessment of when the acute subdural may have started and the force required to cause the acute bleeding. The formal cause of death given by the pathologist was

1(a): Head injury.

The cause of death does not really address the issues of concern to the court:

1. Was the injury admitted as having been inflicted by the father the injury that caused the death?

2. If so, what sort of force must have been involved?

In another case, a two-and-a-half-year-old boy was left alive and well by his mother in the care of her de facto husband. When she returned one hour later, the child could not be wakened. An ambulance was called and it arrived some 15 minutes later. The child could not be resuscitated. The account of events leading to the child's death came from the de facto husband, who described himself losing control and striking the boy to the back of the head with an open hand some nine times with the child perched prone over his lap. The blows occurred in the one- hour period when the mother was absent and the de facto husband described putting the child to bed after the final blow. No lucid interval was described. At autopsy, there was evidence of several discrete areas of bruising subcutaneously on the back of the head. There was no significant subgaleal hemorrhage or skull fracture. There was no extradural, subdural, or subarachnoid hemorrhage. The brain was, however, swollen, showing mild tentorial grooving of both hippocampi. Formal neuropathological examination was undertaken including ophthalmological examination. No cerebral parenchymal injury was demonstrated. There was no evidence of axonal spheroids to suggest diffuse axonal injury as a mechanism. There were no retinal hemorrhages. The cause of death was cerebral edema, most likely secondary to trauma. The autopsy findings were relatively subtle and arguably non-specific. The description of malignant cerebral edema or rapid brain swelling following head injury (often trivial) is, however, well described in the literature [25, 153, 154]. Nonetheless the admissions of the accused were essential to the marrying of the relative non-specific evidence of trauma to the back of the head (discoid bruises) to the ultimately determined cause of death. He was found guilty of manslaughter and sentenced to seven years imprisonment. Had the accused asserted his right to silence, the outcome

Concluding the Cause of Death: A General Formulation

What general rule, if any, can we formulate following this? If the autopsy discovers a finding, a disease or condition that is operating at the time of death and that is capable itself of causing or accelerating death, and in the circumstances of the particular death, its effects apparently exerted themselves, then the disease or condition should be included as part of the cause of that person's death. (This formulation deals, inter alia, with the situation where it would be highly pedantic to include coronary atherosclerosis in the cause of death of a man decapitated by a train having been seen by the driver to place himself on the line in the path of an oncoming train.) In addition, if a disease or condition (whether or not a potentially fatal condition) aggravates or complicates another disease or condition (whether or not a potentially fatal condition) such that death occurs, then both diseases or conditions should generally be included as part of the cause of death.
This formulation has its weaknesses; for example, it does nothing to help draw the line in relation to remoteness of cause.

Conclusions as to Manner of Death

It will have been apparent in some of the cases above that conclusions as to the cause of death often carry with them an implication as to the manner of death; but not always. Making conclusions as to the manner of death is a fundamental responsibility of medical examiners in the United States and of many Coroners' jurisdictions around the world, but not all.
Making these conclusions can be a bit like pushing square pegs into round holes. So much so that some jurisdictions have moved away from requiring such conclusions. In Victoria, the Coroner must find who the deceased was, the medical cause of death, and how the deceased died [155]. The last requirement is met in descriptive terms. For example:

> *"The deceased, who was an intravenous drug addict, unemployed and depressed, was found on the floor in the kitchen of the house he was sharing with friends. Intravenous drug paraphernalia was found on the kitchen table, recent injection marks were identified on his forearms at autopsy, and metabolites of heroin together with alcohol and a benzodiazepine were found on toxicology. The cause of death was 'Mixed Drug Toxicity.'*

"Accidental" overdose is the commonest situation in circumstances of heroin abuse leading to death. In this case the possibility of suicide also exists. The Coroner has chosen to leave the matter open. The word accident or suicide is not used.

The reverse is true in England where Coroners are required to classify the death into one of a number of categories. For example, natural cause, industrial disease, want of attention at birth, dependence on drugs, killed himself (while the balance of his mind was disturbed), accident, misadventure, murder, manslaughter, infanticide, accident, open verdict. Doing so may not be straightforward as some work from the United States has verified.

A study by Hazlick and Goodin (1997) is interesting in demonstrating the high level of disagreement in this area.[113] Twenty-three succinct, well-described classical forensic pathology situations were presented to more than 700 medical examiners/Coroners who were members of the National Association of Medical Examiners, eliciting responses from 198 of them. The manner of

[113] Hanzlick R and Goodin J. Mind your manners, Part III: Individual scenario results and discussion. The National Association of Medical Examiners Manner of Death Questionnaire, 1995. American Journal of Forensic Medicine and Pathology. 1997. 18(3): 228-45

death inferred from the ICD Code that was assigned by the (U.S.) National Center for Health Statistics matched the most common response of participants in 18 (78%) of the 23 scenarios. Table 23 shows the percentage of agreement for the most popular conclusion (homicide, suicide, accident, natural, undetermined, other/blank).

Table 23—Percentage of 198 forensic pathologists agreeing on the most popular manner of deaths in 23 scenarios

%	No. of scenarios
41-50	2
51-60	4
61-70	5
71-80	1
81-90	7
91-100	4

In fewer than half the cases, admittedly chosen because they were at the boundary of different manners of death, was there greater than 80% agreement. In other words, in only 11 out of 23 cases did more than 80% of 198 U.S. forensic pathologists agree on the manner of death. There was considerable diversity of opinion amongst the relevant experts as to the manner of death in these examples. This points to different understandings, even on the same facts, of the criteria to establish particular manners of death. Such differences commonly include: how "certain" one should be to make a conclusion (e.g., the special consequences of concluding suicide may be such as to mean some will require a higher standard of proof than others); and different understandings of intention (e.g., the reckless killing of another may be a homicide whereas the reckless killing of oneself will generally be considered an accident and not a suicide).

The differences amongst pathologists and between them and the health statisticians about the manner of death shows how contentious assigning this form of cause can be, albeit in cases designed to be contentious.

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VCGS

Gene testing for genetic heart conditions at VCGS
Information for referring practitioners

The Molecular Genetics Laboratory at the Victorian Clinical Genetics Services (VCGS) is offering gene testing for a range of genetic heart conditions using massively parallel or "next generation" sequencing. Four cardiac gene panels are available:

Arrhythmia (ARR) syndrome panel - 28 genes

AKAP9	CASQ2	KCNA5	KCNE3	KCNQ1	SCN1B
ANK2	CAV3	KCND3	KCNH2	NPPA	SCN3B
CACNA1C	GJA5	KCNE1	KCNJ2	RYR2	SNTA1
CACNA2D1	GPD1L	KCNE1L	KCNJ5	SCN4B	
CACNB2	HCN4	KCNE2	KCNJ8	SCN5A	

Cardiomyopathy (CM) panel - 64 genes **[65 genes from end-2013]**

ABCC9	DES	[GAA]	MYH7	PLN	TMEM43
ACTC1	DMD	GLA	MYL2	PRKAG2	TMPO
ACTN2	DSC2	HCN4	MYL3	PTPN11	TNNC1
ALPK3	DSG2	ILK	MYLK2	RAF1	TNNI3
ANKRD1	DSP	JPH2	MYOM1	RBM20	TNNT2
ANO5	DTNA	JUP	MYOZ2	SCN5A	TPM1
BAG3	EMD	LAMP2	MYPN	SGCD	TTN
CALR3	EYA4	LDB3(ZASP)	NEBL	SLC25A4	TTR
CAV3	FKTN	LMNA	NEXN	TAZ	TXNRD2
CRYAB	FRYL	MYBPC3	PDLIM3	TCAP	VCL
CSRP3	FXN	MYH6	PKP2	TGFB3	

Aortopathy (AOR) panel - 3 genes **[11 genes from end-2013]**

[ACTA2]	[COL3A1]	[FBN2]	[SLC2A10]	[TGFB2]	TGFBR2
[CBS]	FBN1	[MHY11]	[SMAD3]	TGFBR1	

Sudden death panel (SD) - 92 genes **[101 genes from end-2013]**

All of the genes in the above panels

Why panel testing?

Mutations in a single gene can cause multiple cardiac phenotypes. For example, mutations in *MYBPC3* can cause hypertrophic and dilated cardiomyopathies as well as left ventricular non compaction. Such overlap is common in genetic heart diseases and forms the rationale for panel based gene testing.

Reporting and the possibility of additional findings

- A comprehensive gene report will be produced for the panel of cardiac genes requested by the referrer (as indicated on the referral form) and will include information about mutations and variants of unknown significance as per the laboratory variant classification system.
- The laboratory will also provide information about additional findings of **ACTIONABLE pathogenic mutations** identified in genes from the other cardiac gene panels, as DNA sequence is produced and analysed for <u>all genes from all cardiac panels</u> as part of the test design.

Variant classification

There are several types of variants that may be identified in a mutation detection test. The table overleaf provides a summary of the clinical implications of variant types. VCGS has multi-disciplinary team comprising clinical and laboratory staff that meets regularly to review variants as necessary.

Clinical implications of variant types - summary

Pathogenic mutation(s)

One or more pathogenic mutations may be identified.

Clinical interpretation:

- at-risk unaffected relatives can be offered *predictive* gene testing
- other affected relatives can be offered *confirmatory* testing
- *prenatal diagnosis* is possible

Likely pathogenic mutation(s)

The level of evidence that likely pathogenic mutations are disease-causing is very high, but they lack some of the evidence to be classified as pathogenic.

Clinical interpretation:

- at-risk unaffected relatives can be offered gene testing together with clinical screening to correlate gene and clinical status (taking into consideration the possibility of reduced penetrance)
- other affected relatives can be offered *confirmatory* testing

Variant(s) of unknown significance

VUS cannot be used for predictive testing or prenatal diagnosis.

Clinical interpretation:

- co-segregation studies in affected relatives may help to clarify pathogenicity of VUS

Polymorphism(s)

These are known common variants in the population with no clinical significance. Polymorphisms will not routinely be listed on reports and testing of relatives for polymorphisms is not recommended.

No mutation(s)/variants(s)

For nearly all cardiac genetic conditions the mutation detection rate is less than 100%, as not all the genes for these conditions have been discovered or because mutations in known genes may not be detectable with the gene testing technique employed or because a mutation is not present in the patient (phenocopy).

Clinical interpretation:

- relatives of index cases in whom no mutation is found should continue to have cardiac screening.

Additional findings

The laboratory will report *ACTIONABLE pathogenic mutations* in genes on cardiac gene panels other than the panel requested.

Clinical interpretation:

- proband should have cardiac investigations for the condition associated with the gene in which the mutation was identified.
- at-risk unaffected relatives can be offered *predictive* gene testing.
- other affected relatives can be offered *confirmatory* testing.

Mutation identification rates in INDEX cases

Condition	Panel	No. of genes*	Mutation detection rate
Anderson Tawil syndrome	**ARR**	**1**	**unknown**
Arryhthmogenic cardiomyopathy (ARVC/D)	CM	8	~50%
Barth syndrome	**CM**	**1**	**Up to 100%**
Brugada syndrome	ARR	10	~40%
Carvajal syndrome	**CM**	**1**	**unknown**
Catecholaminergic polymorphic ventricular tachycardia	ARR	4	50%-70%
Danon disease	**CM**	**1**	**up to 100%**
Dilated cardiomyopathy	CM	39	20%-30%
Fabry disease	**CM**	**1**	**up to100%**
Familial atrial fibrillation	ARR	8	unknown
Familial conduction system disease (eg Lenegre's disease)	**CM**	**2**	**unknown**
Familial thoracic aortic aneurysm and dissection	AOR	3 [8]	~20%
Familial sick sinus syndrome	**ARR**	**2**	**unknown**
Hypertrophic cardiomyopathy	CM	35 [1]	50%-70%
Left ventricular non compaction	**CM**	**8**	**unknown**
Loeys Dietz syndrome	AOR	2	Up to 100%
Long QT syndrome	**ARR**	**13**	**~80%**
Marfan syndrome	AOR	3	>90%
Naxos disease	**CM**	**1**	**unknown**
Short QT syndrome	ARR	3	unknown
Sudden arrhythmic death syndrome	**SCD**	**92 [9]**	**10-20%**
Timothy syndrome	ARR	1	unknown

** [ ] number of genes to be added at the end of 2013*

Death by Smothering Following Forced Quetiapine Administration in an Infant

*Michael Philip Burke, MB, BS, BSc, FRCPA, Dip Forens Path,**
Suhail Alamad, BDS (Dentistry), Grad Dip (Forensic Odontology), and*
Kenneth Opeskin, MB, BS, FRCPA†

Abstract: We present a case of smothering of a 2-year-old male infant by his schizophrenic mother who was having a psychotic episode. In addition to the initial autopsy findings of conjunctival petechial hemorrhages, facial linear abrasions and bruises, and mucosal abrasions and bruises, expert odontologic examination revealed indentations of the cusps of central incisors and molars, providing additional evidence of smothering. The postmortem and toxicological examination revealed features of forced quetiapine administration. The case presented is the first case of forced administration of quetiapine described. Our case also highlights the value of expert forensic odontological examination.

Key Words: smothering, quetiapine, infant

(*Am J Forensic Med Pathol* 2004;25: 243–245)

A 2-year-old naked male infant was found deceased in the stairwell of a block of units outside the front door of his mother's apartment. The deceased's mother was schizophrenic and having a psychotic episode for which she was subsequently hospitalized. Examination of the scene revealed the deceased's clothing in garbage bags within large industrial garbage bins. Also discovered were a number of photographs of the mother and deceased, some of which had the deceased's image cut out with scissors. Empty packets of quetiapine in the deceased's mother's name were located in a rubbish bin within the apartment.

Postmortem examination revealed a well-nourished Caucasian male infant. Scattered petechial hemorrhages were present within the conjunctivae. Punctate and linear abrasions were noted over the nose and left and right cheek (Fig. 1).

A formal neck dissection showed no bruises within the strap muscles of the neck. A large amount of whitish pastelike material was noted within the oropharynx and oral cavity, with similar material seen within the major airways and stomach. Residual grape flesh was seen in association with the pastelike material in the oropharynx.

Subcutaneous dissection of the face showed a bruise to the left cheek measuring 1 cm and 2 transverse superficial lacerated injuries to the lateral aspect of the left upper lip, 2 cm to the left of the midline and measuring 0.4 cm in maximum extent. Forensic odontologists were consulted in relation to a forensic intraoral examination. The examination confirmed whitish material on the palate. Lacerations and indentations of the buccal mucosa were seen adjacent to central incisors and molars (Figs. 2 and 3). The features indicated external pressure applied to the right cheek directed upward and inward, consistent with the act of manual smothering.

The material within the oropharynx and stomach was sent for toxicological examination. Toxicological examination showed a femoral vein blood quetiapine concentration of 106 mg/L. The reported therapeutic range for a single 75-mg oral dose in adults is 0.1 to 0.4 mg/L. Peak serum levels after an oral 450-mg dose range up to 0.6 mg/L. There is no indication for the use of the drug in infants. One hundred sixty-five milligrams of quetiapine were recovered from stomach contents and 760 mg from within the oropharynx.

The cause of death was issued as manual smothering, with comments in regard to evidence of prior administration of quetiapine.

DISCUSSION

The cause of death in pediatric homicide varies with the age of the child. In babies and infants, head injury from shaking and blunt trauma is the most common cause of death.[1] Death from deliberate poisoning is unusual. A retrospective study in America analyzing 10 years of pediatric

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From the *Victorian Institute of Forensic Medicine, the Department of Forensic Medicine, Monash University; and the †Department of Anatomical Pathology, St Vincent's Hospital, Victoria, Australia.
Reprints: Michael Philip Burke, MB, BS, BSc, FRCPA, Dip Forens Path, Victorian Institute of Forensic Medicine, 57-83 Kavanagh Street, Southbank, Victoria 3006, Australia. E-mail: michaelb@vifp.monash.edu.au.

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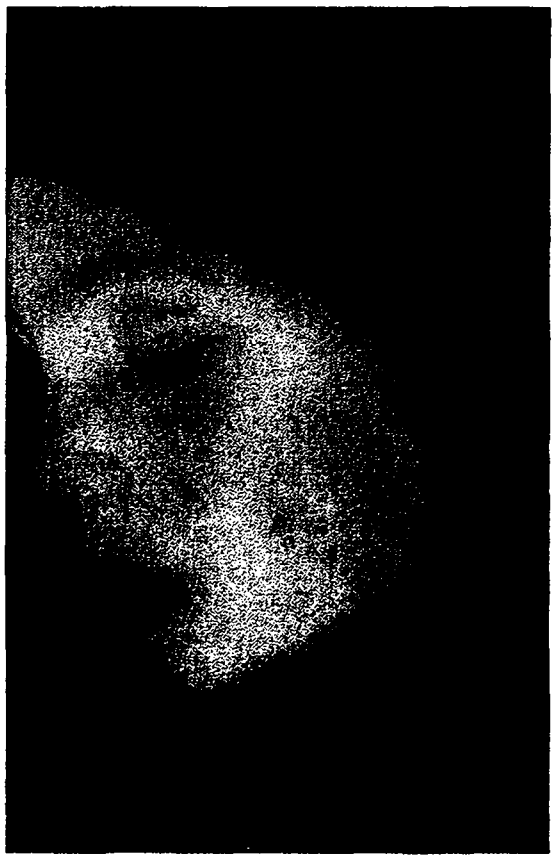

FIGURE 1. Linear skin abrasions to the nose and cheek.

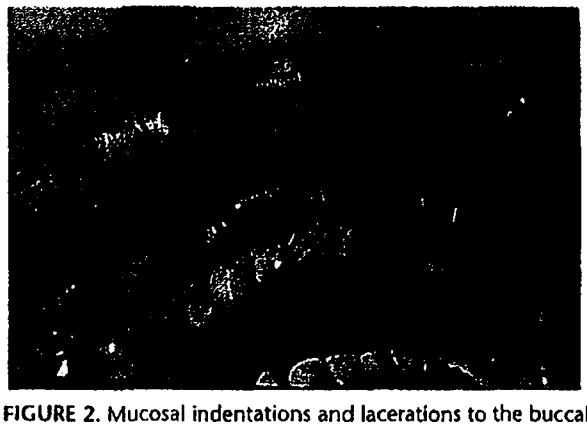

FIGURE 2. Mucosal indentations and lacerations to the buccal mucosa.

toxicologic deaths found 5 victims <5 years of age in a population of 709 pediatric autopsies.[2] The manner of death was ruled as an accident in 3 cases and homicide in 2 cases. The substances involved in the homicide cases were alcohol and cocaine. Nonaccidental injury and occasional deaths from poisoning and asphyxia are seen in Munchausen syndrome by proxy. A paper by McClure et al[3] investigating the epidemiology of Munchausen syndrome by proxy found that anticonvulsants and opiates were the most common drugs involved in their 44 cases.

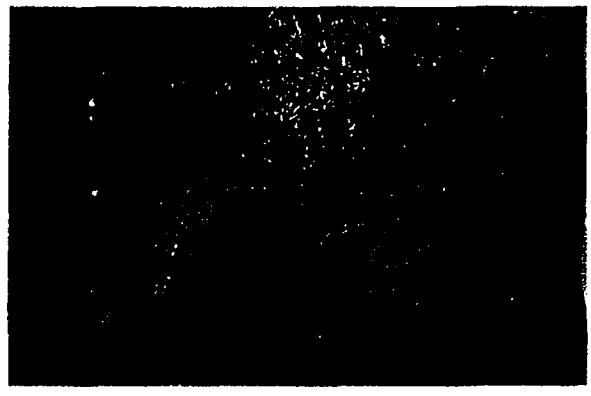

FIGURE 3. Close-up of indentations and lacerations to the buccal mucosa.

Homicidal smothering may leave no objective findings at postmortem examination. This is especially the case when there is a substantial difference in stature between the victim and the assailant, as occurs in homicidal smothering of infants. The presence of abrasions and bruises to the nose, mouth, cheeks, and neck are important findings, and mucosal injuries to the nose and mouth are of considerable diagnostic significance.[4,5] Forensic odontological examination may reveal subtle injuries that are not recognized at the initial autopsy. In the case in question, forensic odontological examination revealed additional regions of indentation and bruising to the buccal mucosa associated with the deceased's teeth, indicating forceful and upward pressure of the buccal mucosa against the teeth. The forensic odontological examination was significant for the finding of additional evidence for smothering.

The pastelike material noted within the oropharynx and airways was shown to be composed of quetiapine, an antipsychotic medication the deceased's mother was known to have been prescribed. The presence of material within the nasopharynx and airway in the setting of evidence of smothering supports forced ingestion of the medication. In this setting, it is considered inconsistent that possible voluntary access of the child to quetiapine could have accounted for the drug being found in the airways. The presence of this material in the nasopharynx/larynx may have contributed to the obstruction of the airways.

Quetiapine is a novel (dibenzothiazepine) antipsychotic used in the treatment of schizophrenia.[6] The drug is generally

considered safe following overdose, although QT prolongation on electrocardiogram, sinus tachycardia, hypotension, and loss of consciousness have been described.[7] In our case, the quetiapine level was markedly raised, at 106 mg/L. Although the deceased's level of consciousness may well have been impaired at this blood concentration, the presence of petechial hemorrhages, facial abrasions and bruises, and mucosal indentations indicate a functioning circulation at the time of the assault.

A single case of death due to quetiapine has been reported.[8] This was the suicidal death of a 52-year-old Caucasian schizophrenic man. The blood quetiapine level was some 18 times the upper limit of normal. The deceased's medical history was significant for cardiac arrhythmias and hypertension prior to the episode of overdose. Forced/homicidal administration of quetiapine has not previously been described.

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APPENDIX 4: PHOTOMICROGRAPHS OF LAURA'S HEART