CHAPTER 3: HEALTH OF THE CHILDREN AND KATHLEEN

The children

266. Dr Alison Colley is a clinical geneticist and the Director of Clinical Genetics Services for various local health districts in NSW. She has trained in paediatrics as well as clinical genetics. She is a conjoint Senior Lecturer at the University of New South Wales. Dr Colley is a renowned dysmorphologist.[347]

267. In 1991 Dr Colley worked at the Newcastle and Northern NSW Genetics Service (now Hunter Genetics) as a Staff Specialist Clinical Geneticist and in that capacity met Mr and Ms Folbigg following the death of their first two children.[348] She gave evidence that both parents agreed that Caleb and Patrick were normal healthy children prior to their sudden unexpected event, which was lethal for Caleb and resulted in severe subsequent neurological damage for Patrick.[349]

268. In 2019, Dr Colley together with Dr Buckley and Professor Kirk ("the Sydney team") prepared a joint report for the Inquiry interpreting the significance of genetic variants, identified through the Sydney pipeline, present in the children and in Ms Folbigg and potentially relevant to the children's causes of death ("the Sydney report").[350] These matters are dealt with in detail later in these submissions.

269. Dr Colley was the author of section three of the Sydney report. In preparing that section, she had regard to the medical evidence in relation to all children prior to the death of Caleb, Sarah and Laura and Patrick's ALTE. She stated as follows:

> *A characteristic of genomic testing in complex disease is that the laboratory data must be interpreted in the context of the clinical presentation. The clinical phenotype and the laboratory results together can be considered to form a single testing process…*[351]
>
> *The below pertains to each of the 4 children, including Patrick prior to his initial unexpected event at 4 months, so I will report collectively:*

[347] Exhibit Z, CV of Dr Alison Colley.
[348] Exhibit AA, Report of Dr Alison Colley (26 November 2019) p 1.
[349] Transcript of the Inquiry, 15 April 2019 T382.17-21.
[350] Exhibit Z, Joint report of Sydney genetics team (29 March 2019).
[351] Exhibit Z, Joint report of Sydney genetics team (29 March 2019) p 4.

1. *There was no evidence of pregnancy related complications; all children had good Apgar scores at birth and normal birth weights*

2. *There were no congenital malformations noted at birth*

3. *Each child had newborn screening reported as normal*

4. *There were no dysmorphic features noted at birth or subsequently by any medical officer, including paediatrician and paediatric specialists when examination occurred*

5. *Development was reported as normal for age for each child*

6. *Photographs [of] children were noted not to show dysmorphic features*

7. *All children were thriving at the time of their unexpected event (at 4 months for Patrick)*

8. *All of the children had normal growth parameters for their ages; there is no mention of small size or short stature*

9. *None of the children had a surgical operation or procedure*

10. *None of the children were admitted to hospital with a significant medical problem*

11. *None of the children were on continuous medication*

12. *None of the children were documented to have more than 8 respiratory infections a year*

13. *The retrieved blood spots on all the children were subject to tandem mass spectrometry with normal results, and negative for MCAD mutation testing*

14. *Paediatric metabolic specialists concluded that all children did not have evidence of an inborn error of metabolism*

15. *All children had autopsy examinations with no medical cause of death determined*.[352]

270. Dr Colley was asked about her statement at point 12 above. She gave the following evidence:

> *Well young children, particularly in the first year or so of life, have a lot of upper respiratory tract infections and, you know, some parents think oh my goodness my child's always got a cough or a cold, is that normal? And so studies have been done and I think I presented a couple of papers, looking at just what is normal for the number of respiratory tract infections per year for a child at different ages. I think eight is a very reasonable number. Some people would say six, some people might say up to ten, but eight is what's accepted.*[353]

271. Dr Colley continued:

> *That is about respiratory tract infections, and I think what's really important is the fact that these children were not admitted to hospital with more serious infections. We don't have any evidence of meningitis, encephalitis, peritonitis, widespread skin infections, whether it be pastoral infections from a bacterial infection, or candidiasis from fungal infections, because children who are born or who have genetic predisposition to infections, one would have expected to see more serious infections than just your usual running nose and cold.*[354]

272. Dr Colley agreed that myocarditis was found on Laura's autopsy, and further that her comments above related to the time before Patrick's ALTE.[355]

273. In her interview with Police, Ms Folbigg noted the following in relation to the health of her children:

    a. In relation to Caleb: "short of his feedin' problem that he had, he sort of didn't have a, didn't have sniffles or colds or any of that sort of thing but

[352] Exhibit Z, Joint report of Sydney genetics team (29 March 2019) pp 4-5.
[353] Transcript of the Inquiry, 15 April 2019 T383.7-14; Exhibit BF, Literature referred to in tranche 2 of substantive hearings, tabs 1-3.
[354] Transcript of the Inquiry, 15 April 2019 T383.26-33.
[355] Transcript of the Inquiry, 15 April 2019 T384.12-23.

he was very young so he sort of hadn't got to that point where you would notice"[356]

b. In relation to Patrick, prior to his ALTE: "he had no problems with breathing, no problems with health in general."[357]

c. In relation to Sarah: "we had a healthy baby."[358]

d. In relation to Laura: "there had been no other problems during that first 12 months other than when she had sort of had her first cold, during the first cold season she had come up with the sniffles a couple of times. Nothing ever serious and she always soldiered through it and it didn't last very long… [after 12 months] she did come down with a flu or bad cold once".[359]

274. Ms Folbigg also gave evidence before the Inquiry in relation to Laura's health:

> *Q. And you say, "nothing out of the ordinary anyway". What did you mean by that?*
>
> *A. Well, she didn't have any major health issues or concerns.*
>
> *Q. Well, nothing at all?*
>
> *A. Basically, yeah.*[360]
>
> *…*
>
> *Q. "Me, well I know there's nothing wrong with her."* [in relation to a diary entry]
>
> *A. Well I did know at the time there was nothing wrong with her, she was a very healthy baby.*[361]

[356] Exhibit E, ERISP of Kathleen Folbigg Q 38.
[357] Exhibit E, ERISP of Kathleen Folbigg Q 142.
[358] Exhibit E, ERISP of Kathleen Folbigg Q 261.
[359] Exhibit E, ERISP of Kathleen Folbigg Q 333.
[360] Transcript of the Inquiry, 29 April 2019 T660.37-42.
[361] Transcript of the Inquiry, 30 April 2019 T757.6-8.

275. Professor Jonathan Skinner is a paediatric cardiologist and cardiac electrophysiologist working as a consultant at Starship Children's Hospital in Auckland, New Zealand. He is an Honorary Professor in Paediatrics, Child and Youth Health at the University of Auckland and Chair of the Genetics Council of the Cardiac Society of Australia and New Zealand.[362]

276. Professor Skinner prepared a report for the Inquiry addressing, among other matters, the cardiac clinical presentation of each of them.[363]

277. From an ECG performed on Patrick on 18 October 1990, Professor Skinner concluded that it was "perfectly normal"[364] and in particular that there are no features to suggest Long QT syndrome,[365] or Brugada syndrome[366] and no evidence of heart muscle disease.[367]

278. In relation to Laura, Professor Skinner reviewed the "strips" from an overnight ECG in August 1997 and again in February 1998 and gave evidence that the rhythm was normal and there was no evidence of any conduction system disorder.[368]

279. In our submission, it is clear from the evidence of medical experts, in particular Dr Colley and Professor Skinner, and of Ms Folbigg, that prior to the catastrophic event leading to death in three of the children and severe neurological sequelae in Patrick which preceded his death, each of the children were healthy, well-grown, normally developing children who were normal in appearance.

[362] Exhibit Y, CV of Professor Jonathan Skinner; Transcript of the Inquiry, 15 April 2019 T369.8-21.
[363] Exhibit Y, Report of Professor Jonathan Skinner (31 March 2019).
[364] Transcript of the Inquiry, 15 April 2019 T385.17.
[365] A heart condition that may have a genetic component. In an individual with long QT syndrome it takes longer for the electrical signals to activate or inactivate the lower chambers of the heart, and this is described as a longer Q-T interval on an ECG, see NSW Health, 'Glossary', *Centre for Genetics Education* (Web Page, 26 February 2016) <https://www.genetics.edu.au/publications-and-resources/glossary#L>.
[366] A rhythm disorder of the heart that can cause the bottom chambers to beat abnormally fast causing the heart to become inefficient at pumping blood around the body, see 'Brugada Syndrome', *Brugada Syndrome - Australian Genetic Heart Disease Registry* (Web Page, 3 September 2014) <http://www.heartregistry.org.au/patients-families/genetic-heart-diseases/brugada-syndrome/>.
[367] Transcript of the Inquiry, 15 April 2019 T386.26-29.
[368] Transcript of the Inquiry, 15 April 2019 T387.8-10.

Craig Folbigg and his sister Carol Newitt

280. As part of her dealings with Kathleen and Craig Folbigg, Dr Colley met with Craig's sister, Carol Newitt.[369]

281. Dr Colley gave the following oral evidence of her observations of Craig Folbigg and his sister:

> *WITNESS COLLEY: … they were both of normal stature, normal facial appearance, they were not what I'd call dysmorphic. They were normal white Australian, Caucasian looking people. They also appeared to have normal intelligence and their behaviour was appropriate and normal for a consultation…*
>
> *Mr Folbigg and his sister and the information they gave me was that there was no known genetic disorder in the family, there was no known inherited disorder in the family. There was no known disorder that caused children to be unwell and spend time in hospital and, and not grow properly or anything like that.*[370]

282. Dr Colley gave evidence as to information provided to her by Craig Folbigg concerning the death of his brother's son. Craig Folbigg, who had been following the hearing of the evidence by the Inquiry, provided the following statement after that evidence was given:

> *This information comes from my brother Michael who I personally spoke with this morning and he quite freely gave this information to me to use as I wished.*
>
> *Michael and his wife Alexandria gave birth to a baby boy on the 12/5/1979 who was 5 and ½ weeks premature, he lived for 7 hours and passed away on the 13/5/79, they were told he suffered from respiratory distress due to his lungs not being properly formed.*
>
> *The cause of death listed on his death certificate is respiratory distress syndrome, a copy of which is provided in the attachment.*

[369] Exhibit AA, Report of Dr Alison Colley (26 November 2019) p 1.
[370] Transcript of the Inquiry, 15 April 2019 T379.48-380.1, T380.17-21.

Fortunately, Michael and Alexandria were blessed with two more children and are now Grandparents.

I only wish to bring this up to dispel any conjecture as to this baby's demise, given the theories that have been advanced during discussions in court.[371]

283. It is submitted that the Judicial Officer will be satisfied that each of the children were healthy, normally developing children prior to their death and in Patrick's case his ALTE. Craig Folbigg's statement as to the circumstances of his nephew's death should be accepted. There is no evidence that it was a SIDS or SUDI death.

Kathleen Folbigg

284. Professor Skinner considered the medical records which have been tendered in respect of Ms Folbigg dating from 1989 to 2019.[372] He concluded that there was no evidence of cardiac-related conditions or cardiomyopathy or heart muscle disease.[373] He also observed from a stress test carried out in May 2011 that there was unlikely to have been significant ventricular tachycardia during the test and there were no signs of ischaemia.[374]

285. Professor Skinner described his impression from the notes from 1989 to 2019 as:

really points to somebody who tends to nearly faint or faint in the presence of a vomiting illness or being dehydrated and during pregnancy so I think these don't sound like arrhythmic collapses. They don't sound like it's a primary cardiac problem. It's a circulatory problem, situational, common fainting, that's the general picture I'm getting.[375]

286. He drew the following conclusions:

Kathleen is now over 50 years of age and this is 50 years over which an inherited heart condition can present itself and signs on cardiac tests can present themselves. So conditions which cause sudden deaths such as

[371] Exhibit BG, Statement of Craig Folbigg (19 April 2019).
[372] Exhibit AC, Genetics tender bundle.
[373] Exhibit Y, Report of Professor Jonathan Skinner (31 March 2019); Transcript of the Inquiry, 15 April 2019 T392.15-20.
[374] Transcript of the Inquiry, 15 April 2019 T389.9-393.30; Exhibit Y, Report of Professor Jonathan Skinner (31 March 2019) p 7.
[375] Transcript of the Inquiry, 15 April 2019 T395.1-5.

hypertrophic or dilated cardiomyopathy, they tend to progress over time and if she was going to develop these conditions I think by 50 we could reasonably expect some clinical signs - an abnormal ECG or an echocardiogram by now. She has got no features of these cardiomyopathies. Regarding cardiac ion channelopathies you can't see these; even when the heart is taken out of the body there's nothing to see. It's a microscopic thing. But she has not had a cardiac arrest in her 50 years. The syncopal episodes that I've reviewed would be consistent with situational or vasovagal syncope rather than arrhythmic syncope. The ECGs show no features of Brugada syndrome or Long QT syndrome nor do they show any sign of conduction system disease.

So I think we can make some general conclusions. As a specialist in Long QT syndrome I don't think she has Long QT syndrome and I don't think she has got any ECG features to suggest she's a gene carrier for it either on her two 12-lead ECGs. So I don't think it's likely she has got Long QT syndrome.[376]

287. Professor Skinner gave evidence that catecholaminergic-induced polymorphic ventricular tachycardia ("CPVT")[377] is not excluded by the ECG results.[378] However, following the further cardiac testing of Ms Folbigg outlined below, Professor Skinner provided a supplementary report to the Inquiry in which he concluded "I am confident she does not have CPVT, and I would not recommend any further tests."[379]

288. Dr Arsov, a genetic counsellor from the Canberra team visited Ms Folbigg in prison in November 2019 for the purposes of taking buccal swabs and a saliva sample and compiling a Folbigg family tree.[380] That tree recorded information from Ms Folbigg including "11 to 12 year old fainted in swim race".[381]

[376] Transcript of the Inquiry, 15 April 2019 T397.15-33.
[377] Catecholaminergic polymorphic ventricular tachycardia is an inherited arrhythmia that causes the heart to beat abnormally fast, particularly during exercise, see 'Catecholaminergic Polymorphic VT (CPVT)', *Catecholaminergic Polymorphic VT (CPVT) - Australian Genetic Heart Disease Registry* (Web Page, 2011) <http://www.heartregistry.org.au/patients-families/genetic-heart-diseases/catecholaminergic-polymorphic-ventricular-tachycardia/>.
[378] Transcript of the Inquiry, 15 April 2019 T393.49-394.1.
[379] Exhibit BK, Letter from Professor Jonathan Skinner to the Inquiry (30 April 2019) p 1.
[380] Transcript of the Inquiry, 16 April 2019 T463.25-29.
[381] Exhibit AE, Pedigree.

289. In his evidence, Professor Skinner drew attention to that comment. He said it was important to know whether Ms Folbigg sank to the bottom of the pool, was pulled out and resuscitated or just won a race and felt a bit dizzy and was pulled out and recovered.[382]

290. Dr Arsov gave evidence that Ms Folbigg told him, "she was in the swimming competition, after swimming for a while she felt unwell and fainted."[383]

291. Professor Vinuesa gave evidence that Ms Folbigg told her "she fainted at the end of the race and I don't know if she fell to the bottom of the pool."[384]

292. As set out above, Dr Colley took a history from Ms Folbigg in 1991. She made the following observation in respect of that history:

> *It was interesting when I took the history from Mrs Folbigg when I… met her, she never mentioned any need for resuscitation, losing consciousness, going to hospital, and it's – I think that's quite interesting that if that had happened to someone I think you would have been told or you would remember, particularly when I'm asking about acute life-threatening events, that that would have come up, and it never did.*[385]

Further cardiac tests on 18 April 2019

293. On 18 April 2019 Dr Hariharan Raju, a cardiologist and electrophysiologist at Macquarie University, performed an exercise test, a standing ECG and a 24 ambulatory Holter test on Ms Folbigg.[386]

294. Dr Raju prepared a short report providing his opinion on the results of these tests.[387] He found:

295. Ms Folbigg's resting ECG was entirely within normal limits;

296. there was a normal Holter monitor in the absence of patient symptoms;

[382] Transcript of the Inquiry, 19 April 2019 T507.38-46.
[383] Transcript of the Inquiry, 16 April 2019 T506.40-41.
[384] Transcript of the Inquiry, 16 April 2019 T507.28-29.
[385] Transcript of the Inquiry, 17 April 2019 T572.47-573.2.
[386] Exhibit BH, Raw test results of Kathleen Folbigg's exercise testing.
[387] Exhibit BL, Letter from Associate Professor Hariharan Raju (18 April 2019).

297. the only abnormality detected was the presence of possible exertional ventricular ectopy, but this is likely of no clinical relevance; and

298. in respect of any further cardiac investigation of Ms Folbigg, the likely yield of cardiac pathology would be negligible.

299. He concluded:

> *In summary, following comprehensive non-invasive evaluation, Kathleen has no phenotypic evidence of either cardiomyopathy or primary arrhythmia syndrome… Her multiple syncopal episodes are likely to be of reflex aetiology, which is benign.*[388]

300. Professor Skinner was also provided with the raw data produced from the additional tests. He concluded:

> *The exercise test shows a good heart rate and blood pressure response and average exercise tolerance. The test is within normal for age…*
>
> *Therefore I find no evidence of Long QT syndrome, and no evidence of significant ventricular arrhythmia. There are a small number of single ventricular ectopic beats, common on exercise tests in people of this age and which are not likely to be of clinical significance. The ECG modified to detect Brugada syndrome was negative.*[389]

301. Ms Folbigg has now undergone extensive cardiac testing. Professor Skinner and Dr Raju agree that there is no evidence of either cardiomyopathy or primary arrhythmia syndrome. In addition, Professor Skinner concludes there is no evidence that Ms Folbigg has Long QT syndrome, Brugada syndrome or CPVT. There is no expert evidence to the contrary. In our submission, the Judicial Officer should be satisfied that all reasonable cardiac testing has been carried out and Professor Skinner's opinions should be accepted.

[388] Exhibit BL, Letter from Associate Professor Hariharan Raju (18 April 2019) p 3.
[389] Exhibit BJ, Further report of Professor Jonathan Skinner (24 April 2019) pp 3-4.