





SYDNEY SOUTH WEST GENETICS SERVICE

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26th November 2018

SUMMARY OF PREVIOUS INVOLVEMENT BY DR ALISON COLLEY Information from - FOLBIGG GENETICS FILE 1564

In 1991 Mr Craig and Mrs Kathleen FOLBIGG were referred to the Newcastle and Northern NSW Genetics Service (now referred to as Hunter Genetics) by general practitioner Dr Chris Marley. Mr and Mrs Folbigg's two sons, Caleb and Patrick had died, aged 19 days and 8 months respectively, both with a diagnosis of SIDS. The concern was whether the boys had a genetic condition that caused or predisposed to their demise, the risk of recurrence in another child and whether there were options to mitigate such an event recurring.

I met Craig and Kathleen in the genetics clinic on 12th November 1991, for an initial discussion to obtain personal and family history information, draw a pedigree and get their consent to obtain doctors letters and documents, including post mortem reports, to assist me in providing them with information and counselling. I wrote to Professor Bridget Wilcken on 4th December 1991 to enlist her expert advice and received a reply dated 10th December 1991.

I met Craig and Kathleen a second time in the genetics clinic on 18th February 1992 for a consultation and wrote them a summary letter dated 27th February 1992 and also on that day wrote a follow up letter to Professor Wilcken with copies to the other doctors involved with the family. At this appointment Kathleen informed me she was pregnant.

On 30th October 1992 I documented in the genetics file that baby Sarah had been born at John Hunter Hospital on 14th October, was seen by Dr Gus Cooper, respiratory specialist, a sleep monitor was arranged and urine sample for metabolic screen was sent to Professor Bridget Wilcken at The Oliver Latham Laboratory in Sydney.

I wrote to Craig and Kathleen on 6th October 1993 after I returned to work after a period of leave to learn that Sarah had died on 30th August 1993 in similar circumstances to her two brothers. I had discussed the event and the subsequent investigations that had been arranged with my colleague Dr Matt Edwards. I offered my condolences and an appointment with me for a further discussion should they want to meet.

I met with Craig and Kathleen in the genetics clinic on 5th November 1993; Craig's sister Carol and her husband Robert were present also. I had discussed the situation with Professor John Christodoulou, metabolic specialist at Westmead Children's Hospital to ensure there wasn't any further metabolic investigations indicted. I wrote a letter to Craig and Kathleen dated 16th November 1993 summarising our discussion on 5th November.

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Courier Address: Department of Clinical Genetics Liverpool Hospital Clinic B, Reception 112, Health Services Building Elizabeth Street LIVERPOOL NSW 2170 I left the Newcastle and Northern NSW Genetics Service in early 1996 and had no further involvement or contact with the family. I learnt from the media that Craig and Kathleen had a fourth child, Laura, who died at 20 months also SIDS.

MEDICAL GENETIC ADVANCES

There has been a fundamental change in genetic testing since 2000, especially in the last five years with the development of robust genomic technology. Genomic interrogation, either by whole exome sequencing (WES) or whole genome sequencing (WGS) is the hypothesis-free study of the DNA; a known or presumed diagnosis as a starting point is not needed, but the DNA sequences are studied and variants interrogated against the known human healthy genome and the clinical features of the patient(s). Variants of unknown significance (VOUS) may need further clarification by in family (parental samples) segregation or functional studies. However, having DNA samples from four siblings with a similar phenotype - normal growth and development, no dysmorphic features, well general health and sudden death in infancy - will decrease the number of VOUS.

The whole exome refers to all the DNA content that is involved in coding for proteins (exons). The whole genome refers to the non-coding elements (introns) in the genome in addition to the exome, as well as the mitochondrial DNA. Increasingly deep intronic variants have been found to cause splice- site changes that lead to the disruption of the correct reading of the code and thus synthesis of a normal protein. There also may be regulatory elements that are not captured within the exome.

The laboratory technique of WES in fact only captures ~85% of the exome and is thus not a perfect technology. The whole genome platform ensures all exomes are fully captured and interrogated, and WGS captures all classes of genetic variation in one test. It has been consistently shown to be more efficient than WES in detecting mutations and is very useful when a specific clinical diagnosis is not achievable. Unexplained infant death can be studied by doing a molecular autopsy by WES or WGS. To capture the mitochondrial genome as well as the most complete nuclear genome, a WGS approach is needed. Variants would be considered if found in genes coding for proteins involved in cardiac arrhythmia, immunodeficiency, respiratory and central nervous system functions, including central hypoventilation, as well as inborn errors of metabolism/metabolic pathways.

RECOMMENDATIONS

1 A discussion of the utility and best strategy for performing genomic studies with a multidisciplinary team of specialists including clinical geneticists and genomicists, anatomical pathologist and molecular pathologists

1 Involve a NSW Health pathologist in molecular genetics, National Pathology Accreditation Advisory Council (NPAAC) accredited, to report the genomic study results and interpretation from a NATA accredited molecular genetics laboratory to issue diagnostic (not research) reports.

2 Consider the samples available from each child, the ability and method to extract DNA from these samples and whether it is possible to have DNA from the parents as in-family control to test variants that require segregation analysis.

3 Variant analysis by team of clinical geneticist and genetic pathologist. Only report pathogenic or likely pathogenic variants (class 4-5); VOUS will not allow diagnosis or exclusion of a genetic condition as a diagnosis.

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Dr Alison Colley Clinical Geneticist Director, Clinical Genetic Services SWSLHD

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Inquiry into the convictions of Kathleen Megan Folbigg

13 November 2018

Dr Alison Colley Senior Staff Specialist Geneticist, Director of Clinical Genetics Department of Clinical Genetics Liverpool Hospital Locked Bag 7279 LIVERPOOL BC NSW 1871

C/-

Deanne Tadros Senior Legal Officer NSW Ministry of Health 73 Miller Street NORTH SYDNEY NSW 2060

By email: Deanne.tadros@health.nsw.gov.au

Copied to: Ian.fraser@fjc.net.au; Blaise.lyons@health.nsw.gov.au

Dear Dr Colley

Letter of engagement

Summary

On 22 August 2018 the Governor of New South Wales directed that an inquiry be held into the convictions of Kathleen Megan Folbigg for three counts of murder, one count of manslaughter and one count of maliciously inflicting grievous bodily harm in respect of her four children on 21 May 2003 ("the Inquiry"). The Crown Solicitor is the Solicitor Assisting the Honourable Reginald Oliver Blanch AM QC ("the Judicial Officer") with the Inquiry.

As discussed, you are engaged to prepare a brief report for the Inquiry which summarises your previous involvement in the Folbigg matter. You are also engaged, together with Dr Bridget Wilcken, to prepare a joint report which:

1. identifies and explains any new research or medical advances in your area of expertise since 2002 relevant to the causes of death of any of the children and/or the cause of the apparent or acute life threatening event in respect of Patrick; and

Inquiry into the convictions of Kathleen Megan Folbigg Level 2 | Industrial Relations Commission | 47 Bridge Street | SYDNEY NSW 2000 T (02) 9258 0832 | E folbigg.inquiry@justice.nsw.gov.au W https://www.folbigginquiry.justice.nsw.gov.au

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2. provides your view as to whether, and if so what, further genetic testing can now be performed relevant to the causes of death of any of the children and/or the cause of the apparent or acute life threatening event in respect of Patrick.

It is requested that you provide your reports, through your solicitor, to Amber Richards, Senior Solicitor for the Crown Solicitor, by **Monday**, **26 November 2018**.

We anticipate scheduling a meeting with all of those geneticists engaged to assist the Inquiry in due course. You may also be required to give oral evidence at the public hearings of the Inquiry. It is expected the hearings will take place at the Industrial Relations Commission Building located on 47 Bridge Street, Sydney in **late February or early March 2019**. We will contact you once the hearing dates for the Inquiry have been listed.

Background

Trial

Between 1 April 2003 and 21 May 2003, Ms Folbigg stood trial in the Supreme Court before Barr J and a jury, upon an indictment containing five counts in respect of the deaths of her four infant children. Those counts were as follows:

- **Count 1**: the murder, on 20 February 1989, of Caleb Gibson Folbigg, 19 days old.
- **Count 2:** maliciously inflict, on 18 October 1990, grievous bodily harm upon Patrick Allen Folbigg with intent to do grievous bodily harm, 4 months old.
- Count 3: the murder, on 13 February 1991, of Patrick Allen Folbigg, 8 months old.
- Count 4: the murder, on 30 August 1993, of Sarah Kathleen Folbigg, 10 months old.

Count 5: the murder, on 1 March 1999, of Laura Elizabeth Folbigg, 19 months old.

The essential issue at trial was whether the Crown could establish that each child's death and Patrick's apparent life threatening event ("ALTE") was a result of a deliberate act of suffocation and not the result of natural causes. The Crown case was that the deaths and the ALTE; the circumstances in which they occurred (including by way of coincidence reasoning); evidence of Ms Folbigg's relationships with, and attitude towards, each child; entries made by Ms Folbigg in her diaries (some of which were said to amount to admissions); and the medical evidence, established that the only rational inference open was that Ms Folbigg unlawfully killed each child.

On 21 May 2003, the jury found Ms Folbigg guilty of three counts of murder in respect of Patrick, Sarah and Laura; one count of manslaughter in respect of Caleb; and one count of maliciously inflict grievous bodily harm in respect of Patrick in respect of the ALTE.

Ms Folbigg was sentenced on 24 October 2003 to an overall sentence of imprisonment of 40 years with a non-parole period of 30 years. This was later reduced on appeal to 30 years' imprisonment with a non-parole period of 25 years and she remains incarcerated at Silverwater Women's Correctional Centre. Ms Folbigg brought appeals against conviction on 17 February 2005, 2 September 2005 and 21 December 2007. These appeals were unsuccessful.

Petition for review of conviction

After having exhausted her right of appeal, on 10 June 2015, pursuant to s. 76 *Crimes* (Appeal and Review) Act 2001 ("the CAR Act") a petition on behalf of Ms Folbigg was

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presented to the Governor of New South Wales seeking an inquiry into her convictions. Accompanying Ms Folbigg's petition were several expert reports, including:

- an undated report by Professor Stephen Cordner (forensic pathologist); and
- a report dated 1 June 2015 by Dr Michael Pollanen (forensic pathologist).

Direction

On 22 August 2018, the Governor of New South Wales issued a Direction pursuant s. 77(1)(a) of the *CAR Act* that an inquiry be conducted into the convictions of Ms Folbigg. The preliminary focus of the Inquiry is on expert medical evidence, including:

- Any new research or advances in medical science relevant to the causes of death of each child and the cause of the apparent or acute life threatening event in respect of Patrick.
- Expert medical opinion as to the causes of death of each child and the cause of the apparent or acute life threatening event in respect of Patrick in light of any relevant new research or advances in medical science.
- Any new research or literature concerning the incidence of reported deaths of three or more infants in the same family attributed to unidentified natural causes.
- Any other related expert medical evidence.

The final scope of the Inquiry is yet to be determined.

Preparation of your reports

The Inquiry would be assisted if you could prepare a brief report for the Inquiry which summarises your previous involvement in the Folbigg matter. You are also engaged, together with Dr Bridget Wilcken, to prepare a joint report which:

- 1. identifies and explains any new research or medical advances in your area of expertise since 2002 relevant to the causes of death of any of the children and/or the cause of the apparent or acute life threatening event in respect of Patrick; and
- 2. provides your view as to whether, and if so what, further genetic testing can now be performed relevant to the causes of death of any of the children and/or the cause of the apparent or acute life threatening event in respect of Patrick.

Your reports should only offer opinions on these matters to the extent that they are based upon your knowledge, training and fields of specialist expertise.

In preparing your reports, please:

- i. identify (and reference as appropriate) any facts and assumptions from materials upon which you rely;
- ii. show how those facts and assumptions relate to your opinions;
- iii. provide an explanation of your reasons for each of your opinions; and
- iv. if necessary, set out any qualification or reservations you have about the opinions expressed in your reports (for instance, because of reservations you hold about a fact, or if further research or information is required, or for any other reason).

Documents with which you are briefed

For the purpose of your engagement, you are briefed with the following documents set out in **Annexure A** to this letter.

If you believe you would be assisted by any further documents, please advise us of the same.

Giving of evidence before the Inquiry

You may be required to give evidence orally before the Inquiry at the public hearings. It is expected the hearings will take place at the Industrial Relations Commission Building located on 47 Bridge Street, Sydney in **late February or early March 2019**. We will contact you once the hearing dates for the Inquiry have been listed. Please also keep us informed of any planned periods of leave.

Expert Code of Conduct and Curriculum Vitae

At **Annexure B** to this letter I set out the Expert Witness Code of Conduct and ask that you read it carefully. In each report you should acknowledge that you have read the Code and agree to be bound by it. I suggest the following form of words be included in the body of each report:

"I, Dr Alison Colley, acknowledge for the purpose of Rule 31.23 of the Uniform Civil Procedure Rules 2005 that I have read the Expert Witness Code of Conduct in Schedule 7 to the said rules and agree to be bound by it."

I also request that you please attach a copy of your curriculum vitae to each report.

Confidentiality

Please ensure you keep your engagement, the documents with which you are briefed, and your reports **confidential**.

Conclusion

Please do not hesitate to contact Amber Richards, by your solicitor, on (02) 9258 0832 or <u>amber.richards@cso.nsw.gov.au</u> if you have any queries or require anything further to assist in the preparation of your reports.

Kind regards

Bhill

Amber Richards Senior Solicitor for Crown Solicitor

Encl.

ANNEXURE A

Index to briefing documents for Dr Alison Colley

Tab	Document	Date
1.	Medical testing of Patrick Folbigg	13 February 1991
2.	Letter to Dr Bridget Wilcken re newborn blood sample	11 October 1999
3.	Genetics report re death of Caleb Folbigg	13 January 2000
4.	Genetics report re death of Patrick Folbigg	13 January 2000
5.	Genetics report re death of Sarah Folbigg	13 January 2000
6.	Genetics report re death of Laura Folbigg	13 January 2000
7.	Expert Certificate of Dr Bridget Wilcken and exhibits referred to therein: 7a – 7c:	14 January 2000
7.a	Letter from Dr Alison Colley to Dr Bridget Wilcken regarding Caleb and Patrick Folbigg	4 December 1991
7.b	Letter from Dr Bridget Wilcken to Dr Alison Colley regarding Caleb and Patrick Folbigg	10 December 1991
7.c	NSW Newborn Screening Programme Report re Caleb, Patrick, Sarah and Laura Folbigg	13 January 2000
8.	Report of Professor Peter Berry	November 2000
9.	Letter from Dr Alison Colley to Dr Bridget Wilcken	27 February 1992
10.	Letter from Professor David Isaacs re testing for serum levels of IgG of Folbigg children	3 March 2003
11.	Facsimile from Dr J. Vivian Wells re IgG levels	5 March 2003
12.	Medical Testing for IgG deficiency, prolonged QT and "Druckers" Gene	7 March 2003
13.	Letter from Dr Allan Cala re IL-10 Gene theory	19 March 2003
14.	Letter from Dr John Christodoulou to J Culver regarding genetic causes of some cases of SIDS	18 February 2003
15.	Summary of present situation re medical investigations prepared by Peter Krisenthal, Legal Aid	27 February 2003
16.	Letter from John Hilton to ODPP re genetic testing of Folbigg Children	27 February 2003
17.	Transcript of evidence of Dr Bridget Wilcken at trial (pages 817-823)	16 April 2003
18.	Supplementary Report of Professor Peter Berry regarding Dr Drucker's work on IL-10 gene polymorphism theory	26 March 2003
19.	Final Report of Professor Blackwell	8 May 2014
20.	Article, Exploring the risk factors for sudden infant deaths and their role in inflammatory responses to infection (frontiers in immunology, Volume 6, Article 44) by Caroline Blackwell, Sophia Moscovis, Sharron Hall, Christine Burns and Rodney J. Scott	March 2015
21.	Chapters from Book, SIDS - Sudden infant and early	

Tab	Document	Date
	childhood death: The past, the present and the future edited by Jhodie R. Duncan and Roger W. Byard (University Adelaide Press 2018):	
	Chapter 2 – Sudden Infant Death Syndrome: An Overview by Jhodie R Duncan and Roger W Byard	
21.b	Chapter 14 – Future Directions in Sudden Unexpected Death in Infancy Research by Heather E Jeffery	
21.c	Chapter 30 – Cytokines, Infection, and Immunity by Siri Hauge Opdal	
21.d	Chapter 31 – The Genetics of Sudden Infant Death Syndrome by Catherine A Brownstein, Annapurna Poduri, Richard D Goldstein and Ingrid A Holm	

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Uniform Civil Procedure Rules 2005, Sch 7: Expert Witness Code of Conduct

1 Application of code

This code of conduct applies to any expert witness engaged or appointed:

- (a) to provide an expert's report for use as evidence in proceedings or proposed proceedings, or
- (b) to give opinion evidence in proceedings or proposed proceedings.

2 General duties to the Court

An expert witness is not an advocate for a party and has a paramount duty, overriding any duty to the party to the proceedings or other person retaining the expert witness, to assist the court impartially on matters relevant to the area of expertise of the witness.

3 Content of report

Every report prepared by an expert witness for use in court must clearly state the opinion or opinions of the expert and must state, specify or provide:

- (a) the name and address of the expert, and
- (b) an acknowledgement that the expert has read this code and agrees to be bound by it, and
- (c) the qualifications of the expert to prepare the report, and
- (d) the assumptions and material facts on which each opinion expressed in the report is based (a letter of instructions may be annexed), and
- (e) the reasons for and any literature or other materials utilised in support of each such opinion, and
- (f) (if applicable) that a particular question, issue or matter falls outside the expert's field of expertise, and
- (g) any examinations, tests or other investigations on which the expert has relied, identifying the person who carried them out and that person's qualifications, and
- (h) the extent to which any opinion which the expert has expressed involves the acceptance of another person's opinion, the identification of that other person and the opinion expressed by that other person, and
- (i) a declaration that the expert has made all the inquiries which the expert believes are desirable and appropriate (save for any matters identified explicitly in the report), and that no matters of significance which the expert regards as relevant have, to the knowledge of the expert, been withheld from the court, and
- (j) any qualification of an opinion expressed in the report without which the report is or may be incomplete or inaccurate, and
- (k) whether any opinion expressed in the report is not a concluded opinion because of insufficient research or insufficient data or for any other reason, and

(I) where the report is lengthy or complex, a brief summary of the report at the beginning of the report.

4 Supplementary report following change of opinion

- (a) Where an expert witness has provided to a party (or that party's legal representative) a report for use in court, and the expert thereafter changes his or her opinion on a material matter, the expert must forthwith provide to the party (or that party's legal representative) a supplementary report which must state, specify or provide the information referred to in clause 3 (a), (d), (e), (g), (h), (i), (j), (k) and (l), and if applicable, clause 3 (f).
- (b) In any subsequent report (whether prepared in accordance with subclause (1) or not), the expert may refer to material contained in the earlier report without repeating it.

5 Duty to comply with the court's directions

If directed to do so by the court, an expert witness must:

- (a) confer with any other expert witness, and
- (b) provide the court with a joint report specifying (as the case requires) matters agreed and matters not agreed and the reasons for the experts not agreeing, and
- (c) abide in a timely way by any direction of the court.

6 Conferences of experts

Each expert witness must:

- (a) exercise his or her independent judgment in relation to every conference in which the expert participates pursuant to a direction of the court and in relation to each report thereafter provided, and must not act on any instruction or request to withhold or avoid agreement, and
- (b) endeavour to reach agreement with the other expert witness (or witnesses) on any issue in dispute between them, or failing agreement, endeavour to identify and clarify the basis of disagreement on the issues which are in dispute.