Inquiry into Convictions of Kathleen Folbigg

Submissions on behalf of Kathleen Folbigg

ADDENDUM

1. On 21 June 2019 Professor de Vinuesa forwarded to the Inquiry a submission. That submission attached a letter from Professor Peter Schwarz dated 20 June 2019.

2. Professor Schwarz is a world authority in the genetic causes of long QT syndrome (LQTS), Sudden Infant Death Syndrome (SIDS) and sudden cardiac death. His curriculum vitae was attached to the submission.

3. On 5 July 2019 Professors Skinner, Buckley and Kirk responded by way of supplementary report to the letter of Professor Schwarz. The report accepted that PM5 was another significant piece of genetic information[1].

4. The further report of Professors Skinner, Buckley and Kirk asserts:

> *If the clinical information described above is not taken into account, the addition of PM5 to the previously applied criteria, PM2, PP2 and PP3 would mean that the variant would be classified as Likely Pathogenic (class IV) with a 10% residual likelihood that this assessment of the level of pathogenicity is incorrect.*

5. If one applies the ACMG guidelines then the addition of the PM5 information provides a likely explanation for the sudden death of two of the children[2].

6. If the ACMG guidelines are applied, with the addition of the PM5 genetic information, then the Judicial Officer would have a reasonable doubt about the guilt of Ms Folbigg with respect to the deaths of Sarah and Laura, the female children.

---

[1] Paragraph 2.7 of the Kirk, Buckley, Skinner report dated 5 July 2019

[2] See also, Submission to Inquiry of Professor Robert Clancy dated 27 June 2019

7. Contrary to this position, in their report in reply Professors Skinner, Buckley and Kirk, review clinical information and give it undue weight outside of the ACMG criteria, to conclude the variant is not likely pathogenic.

8. This presents several problems for these experts, namely:

   A. They provide no scientific reason for their opinion justifying the departure from the ACMG guidelines.
   B. Counsel Assisting introduced the genetics experts by reference to the ACMG standards[3].
   C. Professors Colley, Kirk and Buckley agreed that the ACMG guidelines were to be used to assess pathogenicity[4].

9. At the Inquiry these experts professed the need to apply the ACMG guidelines strictly and yet, in their report in reply, they seek to depart from that position without any proper explanation. Indeed, they appear, to depart from the ACMG guidelines in order to undermine the pathogenicity of the calmodulin genetic variants. The inconsistent application of the ACMG guidelines indicate an exercise of advocacy.

10. Under the circumstances the Inquiry would treat the opinions of Professors Skinner, Buckley and Kirk with caution. The abandonment of application of the ACMG guidelines in their final submission is a mere *ipse dixit* without proper reasoning and should be rejected or, alternatively, given no weight.

11. The willingness of Professors Skinner, Buckley and Kirk to advance inconsistent application of the ACMG guidelines, without proper reasoning, to suit their ultimate conclusion and to resile from their previously held opinions regarding application of the ACMG guidelines should trouble the Inquiry.

12. Further concerns raised by Professor Skinner's unqualified views during the Inquiry[5] should also affect the assessment of his evidence. Such a view was

---

[3] T365.7 - T365.12
[4] T402.25 - T402.31
[5] T480.9 - T480.18: *To my knowledge this condition, Calmodulin, has never been linked to SIDS. I accept that I have never - not read every paper, but I believe I would have known if Calmodulin has presented in SIDS. I do*

reiterated in the 5 July 2019 report: "*To our knowledge, variants in calmodulin have still not been reported as a cause of sudden infant death syndrome, being death of an infant during sleep.*"

13. It is apparent from the further report of Professors de Vinuesa, Schwarz, Overgaard, Cook and Dr Arsov dated 12 July 2019 that Professor Skinner's view is wrong. There is reported death during sleep caused by calmodulin variants. If this is correct then it indicates that Professor Skinner's knowledge is deficient or alternatively he was prepared to advance a view without proper research.

14. If it be the case that the record of the infants' deaths during sleep referred to above was not available at the time of Professor Skinner's evidence at the Inquiry, that may excuse his unqualified opinion at that time. It does not, however, excuse the unqualified opinion expressed in his supplementary report.

15. It might have been expected of Professors Buckley, Kirk and Skinner to examine the Calmodulin register before making the relevant assertion.

16. It is perfectly clear that further scientific evidence has become available which demonstrates likely pathogenecity under the ACMG guidelines. As such, two of the deaths have a reasonable explanation, and this Inquiry should make a finding that there is a reasonable doubt regarding the guilt of Ms Folbigg with respect to those two deaths. That being the case there should be a reasonable doubt regarding the guilt of Ms Folbigg with respect to the other children and we rely on our earlier submissions.

Dated: 12 July 2019

Stuart Gray

Solicitor

---

accept that it 15 causes sudden cardiac arrest in infants, but to date there have been witnesses because the children or the infants were awake. So SIDS is a specific phenotype, you know, the children are asleep in bed, and so I think to say that that is supportive is possibly stretching the criterion.