Australian Academy of Science
Australian Academy of Law
JOINT SYMPOSIUM – ONLINE

Speech – Professor Carola Vinuesa MD (MBBS) PhD FAA

Event: The reception, quality and evaluation of scientific evidence in Australian courts
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Introduction

In April last year I participated in the Inquiry into the Convictions of Kathleen Folbigg. My colleague, Professor Matt Cook, and I were asked to act as unpaid expert witnesses. I was approached by Ms Folbigg's solicitors, while Matt was approached by the Crown Solicitor for New South Wales. Together we wrote a joint report for the Inquiry.

I have not been involved in any other legal proceedings before, but I want to talk to you about my experience as a scientist expert witness. You may wonder what insights a first-time expert witness may have about legal proceedings, but I think it is exactly my naivety that allows for such insights.

While the legal system has an unparalleled ability to draw and distil information from varying sources, my experience left me thinking it has several blind spots when it comes to evaluating scientific evidence. I readily acknowledge that my observations come from a sample of an N of one, so where my experiences are not indicative of usual practice, please feel free to discard them.

My hope is that by engaging in discussions like this, we can build a more scientifically sensitive and informed legal system. One that is not only skilled in applying the scientific method but also welcoming of scientists.

Before delving into my observations, I think it is necessary to provide some context about the case.

Background

Kathleen Folbigg is a woman from the Hunter region of New South Wales. She and her then husband, Craig Folbigg, had four children that died over a period of 10 years in the 1990s. The children, Caleb, Patrick, Sarah, and Laura, all died while asleep at varying ages, ranging from 19 days to 18 months old.

In 2003 Kathleen was convicted of the manslaughter of Caleb and the murders of Patrick, Sarah, and Laura. The jury was persuaded by the prosecution's case that Kathleen had smothered her four children.

There had been no history of abuse and no physical signs of smothering. The circumstantial evidence against Kathleen consisted of coincidence evidence and diary entries. The coincidence evidence was that it was extremely rare for multiple SIDS-like deaths to occur in a family, while the diary entries were considered to contain inculpatory statements as they

expressed sentiments of remorse and guilt. Kathleen has always denied smothering her children and claims the sentiments reflected in her diaries were interpreted wrongly.

At the time of the trial 17 years ago, genomics was in its infancy and the geneticists involved in Kathleen's case could not find a genetic cause for the deaths of any of the children. Instead the sentiments of infamous British paediatrician, Sir Roy Meadow, influenced the discourse about multiple familial SIDS deaths. Meadow's now notorious maxim stated that "one SIDS death is a tragedy, two is suspicious and three is murder until proved otherwise". Meadow has since been discredited by statisticians and scientists, yet his suspicion of multiple familial SIDS deaths appears to have lingered in the wider medical and legal community.

In all four Folbigg children there was medical and pathological evidence that pointed towards natural causes of death. Caleb had difficulty in breathing and swallowing, Patrick suffered from epilepsy and blindness, and, prior to their deaths, Sarah had a bacterial lung infection and Laura had myocarditis. Kathleen herself suffered several episodes of fainting suddenly that were witnessed, including an episode during a school swimming race where she was dragged out of the pool. Fainting in this manner is typical of cardiac arrhythmias that can cause sudden death in infancy and adulthood.

In early 2019, I was asked to form part of a team of geneticists to analyse the genomes of Kathleen and her four children.

From a technical perspective it was an incredible achievement. The Victorian Clinical Genomics Service sequenced entire genomes from neonatal heel-prick blood cards that were over 20 years old. We performed bioinformatic analysis of the approximately 3 billion base pairs of DNA for each subject.

Unfortunately, the analysis was hindered by the fact that Craig Folbigg, the children's father, declined to provide his DNA. I assumed the Inquiry would compel Craig to provide a saliva sample, as we requested, but it declined to make such an order.

Despite this disadvantage, we found a novel, never-before reported, mutation in Sarah and Laura that had been inherited from Kathleen. The variant was in a gene called *CALM2* (that encodes for calmodulin). Calmodulin variants can cause sudden cardiac death.

The *CALM2* Variant

After the end of the hearings but before the Inquiry made its findings, an internationally peer-reviewed article was published containing a registry of 74 people with variants in calmodulin genes. Of those, 20 had died suddenly, 5 of them during periods of sleep, of which 3 were less than 2 years old, just like Sara and Laura.

One boy who died at age four, and his brother, who suffered a cardiac arrest at age five, had a calmodulin mutation at the exact same amino acid location as Sarah and Laura. The boys had also inherited the mutation from their seemingly healthy mother.

Our team reached out to the senior author of the paper, Professor Peter Schwartz; a cardiologist and the world leading authority in genetic causes of cardiac arrhythmias and sudden unexpected death. Professor Schwartz reviewed the clinical and genetic information of the Folbigg family and wrote to the Inquiry, indicating the *CALM2* variant was the likely cause of Sarah and Laura's deaths.

The Inquiry rejected Professor Schwartz's assessment, finding the inculpatory interpretation of the diary entries to be more persuasive. In reaching this conclusion, the Inquiry relied on

evidence from another group of clinicians, including the sole cardiologist that attended the Inquiry.

The other group of clinicians emphasised the fact that the genetic evidence did not explain the deaths of Caleb and Patrick. The sole cardiologist also made several incorrect statements about the effects of calmodulin variants that could have easily been checked by contacting the authors of the CALM registry paper.

Since the conclusion of the Inquiry, in which the judge ruled against reopening the case, Prof Michael Toft Overgaard from Denmark, in collaboration with calmodulin experts from US and Canada performed a series of experiments on the *CALM2* variant. Their findings have been published **[today]** in the Oxford journal, Europace. The 27 authors conclude that the calmodulin variant likely precipitated the deaths of Sarah and Laura, if their deaths were natural.

Natural causes

As a scientist and medical doctor, I found the procedure of the Inquiry bewildering. Even before Professor Toft Overgaard's team and collaborators completed their experiments, there was credible medical and scientific evidence to indicate the Folbigg children had died of natural causes.

For instance, all the forensic pathologists involved in the Inquiry considered Laura's myocarditis to be sufficiently severe to have caused her death. Myocarditis is known to trigger cardiac arrhythmias, and our genetic findings showed that Laura had a mutation in a gene associated with cardiac arrhythmias. Three of the four pathologists went further and said they would have recorded myocarditis as Laura's cause of death. The only hold out was the pathologist that had originally performed Laura's autopsy.

Although the lethal cardiac mutation was only present in the two female Folbigg children, the remaining two male children had medical conditions that further hinted at natural causes of death. Caleb died at 19 days and had difficulties breathing and swallowing since birth due to laryngomalacia (or a "floppy larynx"). Patrick died at 8 months from airway obstruction due to epileptic fits that he had had for the previous 4 months.

Recently we found that both male Folbigg children had biallelic rare missense variants in a gene, that when defective, causes early-onset lethal epilepsy in mice. These variants require further investigation, but their existence highlights the fact that a single unifying cause is not required to explain the deaths of the 4 children. There are countless papers with cases of similar or overlapping phenotypes occurring in a single family but arising from different genetic aetiologies.

Rarity of events

I understand that the law is in a difficult position as it is required to weigh disparate forms of evidence against one another. In this case, medical and scientific evidence was weighed against diary entries. But this is not unique to the law. In science, we regularly weigh evidence from varying sources against one another, and we have developed reproducible methods to categorise and evaluate the strength of evidence to inform clinical decision-making. These methods are extensively used when conducting meta-analyses, which are studies that synthesise the findings of multiple separate papers.

However, it appears the weighting of evidence in law occurs retrospectively and by intuition, rather than by following a process that is set out prospectively. In this case, as far as I can tell, Kathleen was selected for investigation because she had four children die. The rarity of

the events was the sole basis for a police-led investigation, which later discovered and relied on the diaries as evidence of her guilt. The rarity of such occurrences appears to have created a prosecutor's fallacy, through which the scientific and medical evidence was then parsed retrospectively.

Such an approach forgets that rare events do occur, but once the mystery is unravelled the rarity of the event becomes less the focus and more a curious footnote. For instance, we recently found a genetic cause for the deaths of four children in another family. All four children were male and inherited 1 variant from their father and 2 variants from their mother. The specific combination of variants in male children was lethal. The probability of this pattern of inheritance occurring was 1 in 64,000. The probability of another family carrying the same variants and experiencing the same pattern of inheritance is infinitesimally small. So small it is unlikely to ever happen again in human history.

These one-off events in genetics are commonplace, but a focus on the statistics of their rarity in isolation, would mislead you into thinking that such events do not occur. Scientists are not immune to these mistakes. We regularly seek guidance from statisticians to help us design our experiments and interpret our results. However, the law seems apprehensive about letting statisticians into the courtroom. The Inquiry spent much of the first week of the hearings discussing the statistics of multiple SIDS deaths but called no statisticians to give evidence.

Domain experts

In the lead up to the hearing, I was expecting the points of disagreement between the two genetics teams to have been referred to domain experts. But there were no domain experts invited to the hearings. There was not a single expert in the genetics of cardiac arrhythmias. There was a single cardiologist, who had publicly declared not to be a cardiac geneticist. And there were no calmodulin experts in the room.

I understand the difficulty in organising experts to give evidence when there may only be a handful of people in the world qualified to talk on a subject. However, in this Inquiry, the international cardiac genetics and calmodulin experts wrote letters and were willing to be involved. But their advice was not taken as seriously as that of the non-domain experts that were in the courtroom and who unfortunately, got several key facts, in my view, quite wrong.

In science, we have rigorous safeguards against getting things wrong. That is not to say that we always get things right or errors do not occur, but we have built a system to try and minimise such occurrences. For instance, clinical trials are performed in a double-blinded manner and there is anonymous peer-review of the scientific discoveries by the world experts in that domain before a paper is accepted for publication. And, if despite this scrutiny, it emerges after publication that there were major flaws, the paper is retracted from the journal.

This absence of scientific evidentiary safeguards and lack of rigour, made for a negative experience for me. It is possible that what I experienced might be the one-off, but it has discouraged me from engaging in similar court cases in the future. If my experience is not unique and this is common, the law runs the real risk that career scientists will not want to engage in legal matters.

If we want scientists to participate in cases involving complex or technical scientific issues, I think we need to improve the process of recruiting expert witnesses. Priority should be given to selecting scientists who support their reasoning based on peer-reviewed scientific evidence, rather than anecdotal evidence taken from their personal experiences.

There appears to be an obvious and inherent conflict of interest, in parties to a hearing being entirely responsible for selecting and remunerating their own expert witnesses. For less scientifically-technical matters, an experienced judge is presumably capable to deal with these conflicts, but for cases involving novel technologies, it may be prudent to have scientific advisors assist the court or Inquiry.

I am told that civil cases can involve the Court itself appointing a single expert or experts to give an opinion, as opposed to the parties each getting their own experts. While processes such as this may be well intentioned more questions arise such as, *how does a judge know who to appoint?* Or: *Is there a debate that should be had if the area of science is not settled?*

Potentially, an independent scientist with experience in the field of enquiry, could provide guidance to the judge or the parties about the selection of suitably qualified expert witnesses. The Academy of Science would be an obvious choice for fielding such helper scientists.

As a last refrain, I hope the experience of giving evidence could be made less combative. I felt intimidated throughout the hearing being forced to answer yes or no to many questions and being cut off repeatedly. The natural world rarely exists in binary and the use of legal sophistries does nothing to elucidate its complexity. If scientists do not feel they are treated as equals by their legal peers, they are unlikely to volunteer their time to assist the court. Instead the law will be left with only a small handful of professional expert witnesses that are unlikely to be representative of their respective fields.

In summary, I hope that in the coming years we will see an increased appreciation for the scientific method in a legal setting. Complex cases like this one are likely to become more frequent as our scientific tools improve and increasingly find their way out of the lab and into the courtroom.

In recognising this trend, I think it is our responsibility as scientists to assist our legal peers. They should feel confident they have the right scientists at their disposal to resolve the issues of a difficult case. Similarly, I hope our legal peers will continue to value scientists and make them welcome in their courtrooms.

Acknowledgement
I would like to end by thanking Dave Wallace, a talented young lawyer who instigated the sequencing of the Folbigg genomes. His interest in the case stems back to his time in law school and comes from his deep understanding of statistics and genetics.