Behind the exoneration of Australian woman convicted of killing her children

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On December 14, Kathleen Folbigg's 2003 convictions for murder of three of her infant children and manslaughter of the fourth was quashed by the New South Wales Court of Criminal Appeal in light of the *Report of the 2022 Inquiry into the convictions of Kathleen Megan Folbigg* delivered in November by former chief justice Tom Bathurst KC.

Folbigg was convicted of murdering Laura (10 months), Patrick (8 months) and Sarah (19 months), and the manslaughter of Caleb (19 days), with their deaths occurring between 1989 and 1999. She was sentenced to 40 years in prison, reduced on appeal to 30 years with a 25-year non-parole period. Folbigg maintained her innocence throughout.

Folbigg's conviction in the 2003 court case constituted a miscarriage of justice for which she spent nearly two decades in jail. There were no direct eyewitnesses and no physical evidence of foul play on the babies. Her conviction rested heavily on the interpretation of her diaries, in which she agonised over the deaths of her children, as confessions of infanticide.

None of this amounted to proof beyond a reasonable doubt, yet the jury found her guilty. With her conviction, the media coverage denounced her in the blackest terms. The tabloid *Daily Telegraph*, for instance, headlined its article "Monstress: the diary of a child murderer."

Outside the court of appeal on December 14, a vindicated Folbigg told the assembled press conference "The system preferred to blame me rather than accept that, sometimes, children can and do die suddenly, unexpectedly, and heartbreakingly. I think the system and society needs to think before they blame a parent of hurting their children."

In his report, Bathurst found reasonable doubt about her guilt. The intervention of principled scientists played a central role in the decision to overturn the conviction.

The science of genetics was critical to Folbigg's exoneration. The case in 2003 was just three years after the Human Genome Project published the first draft of the human genome, which led to a revolution in genetics, with very rapid developments following. Central to this was the ability to sequence genomes relatively quickly and cheaply.

In 2015, a petition was lodged, raising doubts over the evidence presented in the trial. This led to a 2018 inquiry into Folbigg's case conducted by Reginald Blanch, former chief judge in the District Court of NSW, who reported in October 2019.

As part of the preparation for the inquiry, Folbigg's lawyers turned to a geneticist at the Australian National University (ANU), Carola Vinuesa, to see if genetic make-up had played a part in the death of the children. Vinuesa, along with her colleague geneticist Tudor Arsov, arranged the DNA sequencing of biological samples from Kathleen.

They found that she had the calmodulin 2 (CALM2) gene. This gene has undergone three mutations, producing three variants, CALM1, 2 and 3, that are known to be critical to the functioning of the heart. Calmodulin stands for calcium modulated protein, which senses and helps control calcium levels through cellular pathways known as ion channels. This process is particularly important in regulating the heartbeat. The CALM mutations cause too much calcium ion to be channelled, a result that has been found to be responsible for several sudden deaths, especially in infants.

This discovery occurred as the inquiry was starting, and an expert panel of geneticists and cardiologists was formed. They managed to obtain preserved samples from the Folbigg children. The scientists split into two teams, one based in Canberra and the other in Sydney. Both teams found that Folbigg's two female children had the CALM2 gene.

"Given that it was much more complicated to extract the genome of the children, there was a chance that Kathleen herself might be carrying one of these variants, because they tend to be inherited," Vinuesa told *Cosmos*.

Yet the 2019 inquiry upheld Folbigg's conviction. An important issue in upholding the conviction was the significance of the CALM2 gene—the two teams disagreed, with Canberra asserting its importance while Sydney pushed that it was not significant. A factor in the Sydney group's argument was that Folbigg herself had survived to adulthood, thus calling into doubt the lethality of the gene.

Vinuesa and Arsov, who were involved in the Canberra team's report, asserted that the presence of the gene in the female children was 'likely pathogenic."

The divergence between the two groups occurred because many genes were being identified by scientists, including some that were potentially dangerous, but their role in the functioning of the human organism was not necessarily known.

An important feature article by science journalist Nicky Phillips, entitled "Trials of the Heart," published in *Nature* in November 2022, explained the basis of the disagreement. "The medical literature is littered with papers that claimed to have identified

dangerous gene variants that later turned out to be harmless."

Before the inquiry findings were handed down, Vinuesa contacted several scientists who work on calmodulin to help resolve the disagreement. Peter Schwartz, a cardiologist specializing in arrhythmias of genetic origin at the Auxological Institute in Milan, responded. His research team had found a link between mutations in calmodulin and sudden death in childhood.

Schwartz had written a paper that identified a case involving the CALM3 gene in a family where a four-year-old boy had died suddenly while his five-year-old sister had survived a cardiac arrest. Their mother was a mosaic, meaning only some of her cells had the CALM3 variant, with the remainder normal.

Schwartz wrote to Vinuesa stating that "my conclusion is that the accusation of infanticide might have been premature and not correct."

Spurred by the adverse 2019 finding against Folbigg, Vinuesa contacted other scientists internationally to assist in gathering evidence to extend the scientific understanding.

Central to the campaign was the involvement of a protein scientist at Aalborg University in Denmark, Michael Toft Overgaard who had discovered the first mutation calmodulin gene in 2012.

His team of researchers included postdoctoral students Helene Halkjær Jensen and Malene Brohus, who constructed proteins with Folbigg's mutations of the CALM2 gene known as G114R. These were compared to two other calmodulin variants known to cause severe arrhythmias that occur due to an electric signal failing that cause the heart to beat.

The two scientists discovered the mechanism by which the G114R variant interferes with the channelling of calcium in the heart, causing these arrhythmias to occur.

They found that the G114R variant delayed the closure of the ion channels, letting extra calcium into the cell. "That's what we know to be one of the signatures of a pathogenic calmodulin mutation," Ivy Dick, an electrophysiologist at the University of Maryland School of Medicine in Baltimore, who was involved in the research, told *Nature*.

Dick continued, "If you asked me, 'Would this mutation be likely to cause sudden death? I would say somebody with this mutation is at very high risk of that."

The research was published in November 2020 in a paper entitled "Infanticide vs. inherited cardiac arrhythmias" in the journal of the European Society of Cardiology (ESC). It is known legally as the Brohus article, and presented the evidence proving the natural cause of the death of Kathleen Folbigg's two girls.

The researchers also identified that the two boys, Caleb and Patrick, carried a different and dangerous gene, BSN, that controls the Basson protein responsible for transmissions across the synapse between two nerve cells. The variant could cause a lethal form of epilepsy.

"In mice, BSN deficiency causes early onset severe epilepsy that is 50 percent lethal in the first six months of life," the study stated.

It was established that Kathleen Folbigg was a mosaic for the CALM2 variant, enabling her to survive to adulthood but unfortunately passing on the lethal gene to her two girls.

The Brohus article powerfully concluded: "The growing understanding of inherited cardiac arrhythmias forces a reassessment of the medical foundations, never challenged, on which several mothers were found guilty of infanticide on the basis of assumption instead of evidence.

"The genomic revolution heralds a new era for the assessment of recurring familial sudden deaths of infants and children, an era that reasserts the presumption of innocence for tragically unlucky families."

These words preceded an intensified fight against the miscarriage of justice and calumny perpetrated against Kathleen Folbigg in 2003.

Vinuesa contacted the chief executive of the Australian Academy of Science, Anna-Maria Arabia, who produced a petition of 90 eminent scientists, including Nobel prize laureates Elizabeth Blackburn and Peter Doherty, calling on the New South Wales government to override the 2019 finding and pardon Folbigg. This led to the establishment of what became the Bathurst inquiry.

The scientific evidence was critical in Folbigg's exoneration, with scientists from the Australian Academy of Science acting as consultants to the Bathurst inquiry.

The array of scientific evidence to the inquiry from world leading geneticists, protein scientists, cardiologists, neurologists, paediatricians, forensic pathologists and psychologists was itself virtually unprecedented.

Overgaard and his colleague Professor Mette Nyegaard made a crucial presentation, where they outlined results of the study of the gene variant CALM2 G114R, finding it inadequately performs its regulatory role in heart cells.

Bathurst heard from the remaining senior authors involved in the Brohus article, Arsov, Matthew Cook, Vinuesa and Schwartz. Other internationally-renowned cardiologists and geneticists appraised the work of the Brohus article, with most agreeing that the CALM2 G114R variant had the potential to be disease-causing.

Other experts in psychology, neuroscience, forensic pathology and paediatrics gave evidence relating to other aspects of the case. Two neurologists proposed epilepsy as a cause of death in Patrick Folbigg. Psychiatric and psychological experts determined Folbigg's diaries—centrally used in her conviction—should be interpreted through the lens of maternal grief, rather than as admissions of infanticide.

Science and scientists were critical to ensuring justice for Folbigg who had been egregiously dealt with after the tragic death of her four children. As her lawyer Rhanee Rego commented in June when Folbigg was pardoned before the final Bathurst report was delivered, she had endured "close to two decades locked away in maximum security prisons for crimes which science has proved never occurred."