Effectiveness of posthumous molecular diagnosis from a kept baby tooth

Helen Leonard, Mark R Davis, Gavin R Turbett, Nigel G Laing, Carol Bower, David Ravine

In April, 2004, we met the parents of a young girl who had died almost 13 years earlier. In September, 1991, their 7-year-old daughter had died of pneumonia after a 5-year history of undiagnosed neurological decline. She was the second live-born child of a couple with a healthy son, now in his mid-twenties. The girl was born at term after a pregnancy complicated only by slight early bleeding. Two previous pregnancies had resulted in stillbirths due to abruptio placentae. The child thrived during her first year and there were no parental concerns. At 16 months, however, they became uneasy because she was not walking independently. Parental anxiety was further heightened by loss of previously acquired words. At 17 months, she fell down steps, hitting her head on a cement floor but with no loss of consciousness. By her second birthday functional hand use, which had developed to the stage of self-feeding and pushing toy cars, was replaced by repetitive hand movements, including relentless finger chewing and sucking. Seizures, which responded only partially to anticonvulsant therapy, started after her third birthday. Between the ages of 2 years and 4 years she underwent intensive neurological investigations, which did not reveal a cause for her condition. Her deterioration continued and, in the absence of a diagnosis, her parents began to wonder whether they had somehow been responsible. As well as fears about the role of a vaccination a month before the onset of symptoms, the mother harboured concerns about a possible inherited disorder, and the father feelings of guilt about the circumstances of the child’s fall.

During the child’s lifetime no definite medical diagnosis was ever made, although Rett syndrome had been suggested by an occupational therapist visiting her school. An article about Rett syndrome in a women’s magazine in 2002 rekindled the mother’s interest in neurological investigations, which did not reveal a cause for her condition. Her deterioration continued and, in the absence of a diagnosis, her parents began to wonder whether they had somehow been responsible. As well as fears about the role of a vaccination a month before the onset of symptoms, the mother harboured concerns about a possible inherited disorder, and the father feelings of guilt about the circumstances of the child’s fall.

The onset of symptoms, the mother harboured concerns about a possible inherited disorder, and the father feelings of guilt about the circumstances of the child’s fall. The onset of symptoms, the mother harboured concerns about a possible inherited disorder, and the father feelings of guilt about the circumstances of the child’s fall.

The result has also brought closure to many long-standing psychological stresses for a couple, one sign of which was the drive that eventually prompted the investigation 14 years after the death of their daughter.

Acknowledgments
We acknowledge the major contribution made by the family to this report, and thank Hayley Durling for the sequence analysis, and the National Institute of Child Health and Human Development for funding of the Australian Rett Syndrome project under NIH grant number 1 R01HD43100-01A1.

References