Researchers crack rare gene variant and deliver hope to South Australian family

December 16 2019, by Michele Nardelli

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Working in a highly successful collaboration between the Centre for Cancer Biology (a University of South Australia and SA Pathology alliance), the University of Adelaide, and the Women's and Children's Hospital, UniSA Ph.D. student Alicia Byrne has helped to identify a rare genetic disease that causes severe neurodegeneration in infants.

The genetic disorder has now only been described in three patients

worldwide, two of those in one South Australian family.

The previously baffling condition, sees a normal healthy child start to lose muscle tone and motor skills, ultimately losing the capacity to walk and use language. The children go on to experience epileptic encephalopathy and cycles of serious gastric disruption, including severe vomiting.

The condition has an onset at between 12 and 14 months.

Using a genomics approach, where a patient's entire DNA sequence is examined, Byrne's Ph.D. research has a focus on tracking down the causes of rare diseases in the perinatal and infant period, particularly cases of stillbirth.

"When we started working with this local family, the disorder the children presented with had never been described but since our research began there has been one more case identified," Byrne says (pictured above).

"We discovered that the children carried genetic changes which meant they were unable to absorb vital B group vitamins, which are essential for normal development and function of the nervous system."

While the Adelaide family tragically lost one child to this disorder, with the cause now identified, the family's paediatric neurologist at the Women's and Children's Hospital, Dr. Nicholas Smith, and colleagues were able to devise a targeted therapy to overcome the problem.

Senior lecturer in paediatric medicine at the University of Adelaide, Dr. Smith says the treatment has made a huge difference.

"For the family's second child, weekly injections of the B group vitamins

in which he is deficient have been able to halt and even reverse some of the impacts of this devastating disease," Dr. Smith says.

Byrne's Ph.D. supervisor, UniSA Adjunct Professor at the Centre for Cancer Biology, Hamish Scott says, ironically, rare diseases are actually a broad and significant area of genomics research.

"While a rare genetic disease may only impact a handful of people, what we are quickly understanding in our work on the human genome is that there are myriad different rare diseases," he says.

"Genomic research opens an important path in identifying and, with strong partnerships such as we have here in South Australia between universities, government and our hospitals, in developing personalised precision medicine to treat rare diseases."

"In addition, the work we do in understanding genes and how they make the body work, constantly informs human biology and provides deeper understandings of human health that have population-wide relevance."

"Our goal is to develop genomic testing so that children can be diagnosed at or before birth and treatments can be delivered as early as possible."

More information: Alicia B. Byrne et al. Identification and targeted management of a neurodegenerative disorder caused by biallelic mutations in SLC5A6, *npj Genomic Medicine* (2019). DOI: 10.1038/s41525-019-0103-x

Provided by University of South Australia