Genetic epilepsy can originate in the embryo, study shows

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A study of identical twins shows that a rare genetic form of epilepsy can be caused by a genetic mutation that occurs in the embryo, and not necessarily passed down from parents.

The study was led by the University of Melbourne and Austin Health and published today in the New England Journal of Medicine.

Professor Berkovic, Director of the Comprehensive Epilepsy Program at Austin Health and Epilepsy Research Centre at the University of Melbourne and lead investigator on the study said this is an exciting finding revealing how a mutation in the embryo can cause genetic epilepsy to occur.

“While this study focused on an uncommon form of epilepsy, this finding may have implications for those with other forms of genetic epilepsy, and in fact, other types of genetic disease,” Professor Berkovic said.

While this discovery was made by studying a limited group of people - those who have an identical twin with a particular form of epilepsy known as Dravet’s syndrome - researchers believe it could have wider implications for siblings of people with other genetic diseases that could be caused by genetic mutation in an embryo rather than in the germ (sperm or egg) cells of the parents.

Researchers have previously believed that new mutations for epilepsy

and other diseases usually occur in the sperm or egg cells of the parents, but, by using identical twins, the study shows that mutation may occur shortly after fertilization.

If a test could be developed in the future to verify that there has been no mutation in the germ cells of the parents, siblings of a child with a disease caused by a genetic mutation could rule out the likelihood of passing it on to their own children.

“This really shows the value of studying identical twins to make genetic discoveries that are otherwise effectively invisible to scientists when studying other members of the population,” Professor Berkovic said.

The study was conducted in collaboration with the Epilepsy Research Program, SA, Pathology at Women’s and Children’s Hospital, North Adelaide, SA, and the National Center for Adult Stem Cell Research, Griffith University QLD.

Provided by University of Melbourne

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