

Scientists find 'molecular trigger' for sudden death in epilepsy

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The most common gene for a syndrome associated with abnormal heart rhythms and sudden death triggers epileptic seizures and could explain sudden unexplained death in epilepsy, said researchers from Baylor College of Medicine in a report that appears today in the journal *Science Translational Medicine*.

The identification of this particular potassium channel KvLQT in neurons of the [central nervous system](#) gives scientists a clue about which epilepsy patients face the greatest risk of dying unexpectedly, said Dr. Jeffrey Noebels, the study's senior author and director of the Blue Bird Circle Developmental Neurogenetics Laboratory at Baylor College of Medicine. The channel has been identified in [heart](#) muscle cells and now for the first time in brain or [nerve cells](#).

"Idiopathic (unexplained) epilepsy is one of neurology's oldest mysteries. While most persons with epilepsy will have a normal lifespan, our finding now points the way to a simple and essential test to identify risk for [sudden death](#) in persons with seizures of unknown origin. In these patients, a routine cardiology evaluation consisting of an EKG, and if indicated, a genetic screening test for this family of genes can positively identify this new risk factor," said Noebels. "If the gene test is positive, there are effective treatments for the heart irregularity, including drugs known as beta blockers, as well as the use of a cardiac pacemaker to prevent lethal arrhythmias."

As many as 18 percent of deaths in epilepsy come suddenly without

warning, devastating families.

"Living with epilepsy is difficult enough, but unexpectedly dying from it, as happens in young adults with the disorder, is one of the greatest fears a family must face," said Dr. Alica Goldman, assistant professor in the BCM department of neurology. Noebels is a professor in the departments of neurology, neuroscience and molecular and human genetics at BCM.

No one knew why young people with epilepsy died suddenly, but Goldman built on previous work in Noebels' lab that found that an ion channel gene thought to work only in the heart was active in the brain as well. She examined five ion channel genes linked to long QT syndrome, a disorder associated with heart rhythm disorders and sudden death.

Long QT refers to an interval in electrocardiograms - the QT interval, which is prolonged in this disorder. An ion channel is a tiny pore in a membrane that controls the flow of ions such as calcium and potassium in and out of a cell.

Goldman found that mice with a mutation in the gene that encodes for the KvLQT1 [ion channel](#) had frequent epileptic seizures as well as life-threatening heart rhythm irregularities.

"This demonstrates the long-sought molecular link between heart and brain in epilepsy," said Noebels.

Goldman is now screening [epilepsy](#) patients to determine whether they have the same gene mutation.

Source: Baylor College of Medicine ([news](#) : [web](#))

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