Medical Xpress

Heart link discovered in sudden epilepsy deaths

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(PhysOrg.com) -- Sudden unexpected death in epilepsy (SUDEP) is the most common cause of epilepsy-related death and responsible for about 150 Australian deaths each year yet the underlying cause has remained a mystery.

New findings from the Centenary Institute and the University of Sydney have revealed faulty heart genes may be the missing link, according to research published today in *Brain Pathology*.

SUDEP is the term applied to sudden death occurring in a person with epilepsy for no apparent reason. When such a death occurs and all other possible causes of death are excluded, SUDEP is usually attributed as the cause of death.

"Sudden unexpected death in epilepsy occurs mainly in young people so these findings could have a huge impact in saving lives through early diagnosis. The ultimate goal will be to use genetic screening of patients with epilepsy to identify these gene mutations that could increase the risk of sudden unexpected death," said lead author Centenary Institute and University of Sydney Head of Molecular Cardiology, Professor Chris Semsarian.

The research of people with epilepsy who had died from a sudden unexpected death discovered the presence of certain genetic mutations found in a potentially fatal heart disorder known as long QT syndrome. Long QT syndrome is caused by mutations in more than 10 genes and

eight of these can interfere with the ion channel of cell membranes and disrupt their ability to regulate electrical activity in our body. This disruption of the ion channels can lead to abnormal, life-threatening heart rhythms.

The link was discovered by the researchers who checked the post-mortem blood samples of cases of sudden unexpected death in epilepsy from 1993 to 2009 for the three most common long QT syndrome genes (KCNQ1, KCNH2 (HERG), SCN5A). Of the 48 cases that could be analysed, the researchers found the faulty genes were present in six (13 percent) cases.

While the findings are a major first step in understanding the cause of sudden unexpected death in epilepsy, more research is needed to determine the exact role these genetic mutations play.

Professor Semsarian said: "These findings clearly demonstrate genetic mutations that disrupt the ion channels play a role in sudden death in people with epilepsy. However, we were unable to review medical histories to look at a family history of sudden death, epilepsy and/or long QT syndrome so it remains to be determined whether these changes are the genetic cause or an accompanying risk factor."

More information: www.wiley.com/bw/journal.asp?ref=1015-6305

Provided by University of Sydney

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