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Study reveals gene mutation that increases risk of SUDEP in patients with mild forms of disease

Published on October 16, 2015 at 7:52 AM

Researchers in the Adult Genetic Epilepsy Program of the Krembil Neuroscience Centre have discovered a gene mutation that increases the risk of sudden unexpected death in epilepsy (SUDEP) in patients with mild forms of the disease.

The study, entitled "Two definite sudden unexpected deaths in epilepsy in a family with a DEPDC5 mutation" and published in the journal *Neurology: Genetic*s, studied a four generation family where nine members had epilepsy caused by a mutation of the DEPDC5 gene. Despite this form of epilepsy being categorized as a mild type, two of the family members passed away from SUDEP - a statistically significant finding for such a small sample size.

"This finding is very important because SUDEP is much more common in patients with very severe forms of epilepsy where they are having seizures almost daily," says Dr. Danielle Andrade, neurologist and Krembil Neuroscience Director of the Adult Genetic Epilepsy Program who was the principal investigator of the report.

"Now that we have evidence that a particular gene increases the risk of SUDEP in patients with mild epilepsy, we can not only screen them for this mutation but it also helps us better counsel our patients about their risk."

SUDEP occurs when someone with epilepsy who is otherwise healthy dies unexpectedly without an apparent cause. It is estimated that 1 in 1,000 patients with epilepsy die of SUDEP each year.

Unlike those with severe forms of the disease, patients with mild epilepsy are able to live relatively normal lives while managing their occasional seizures with medication. However, all seizure medications have strong side effects, and some patients who feel their seizures are well controlled are known to occasionally skip taking their medication to avoid side effects.

"This new evidence of the DEPDC5 gene being a risk factor means we may have more patients than we previously thought that are at risk of SUDEP," explains Dr. Andrade. "So this helps to strengthen a physician's argument as to why their patients need to stick to their medication regimen even if their seizures are mild and infrequent."

The DEPDC5 gene was first identified in 2013 and has been found to be involved in many different types of epilepsy thereby furthering researchers' understanding of the disease.

More research is needed to determine the exact percentage increase risk of SUDEP that can be attributed to the DEPDC5 gene as well as understand how the gene induces SUDEP.

Source:
Krembil Neuroscience Centre (KNC)
