

New gene that causes intellectual disability discovered

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A new study involving Canada's Centre for Addiction and Mental Health (CAMH) has found a gene connected with a type of intellectual disability called Joubert syndrome.

CAMH Senior Scientist Dr. John Vincent has identified this gene that, when defective, leads to Joubert syndrome. This research is published in the 13 May 2011 issue of *Cell*.

This international study combined Dr. Vincent's gene mapping of a family with Joubert syndrome, with the use of a [protein network](#) map established by researchers at [Genentech Inc.](#), Stanford University and the University of California at San Francisco (UCSF). Together this approach identified two genes associated with the group of disorders called ciliopathies.

Joubert syndrome, which is a ciliopathy, affects brain functioning, resulting in intellectual deficits, movement and coordination problems and other symptoms such as kidney and eye problems. This syndrome is reported to affect approximately 1 in 100,000 children, although this is likely to be a significant underestimate of the true prevalence.

Ciliopathies are caused by [genetic defects](#) to a part of the cell called the [cilium](#). The cilium is crucial as it is involved with [cell signaling pathways](#) during cell development in different parts of the body. The other ciliopathy gene identified in this study leads to a condition called nephronophthisis, which is also associated with kidney and eye problems.

"A defect in any aspect of this molecular pathway may have very similar effects at the clinical level," says Dr. Vincent, who is also head of the Centre for Addiction and Mental Health's Molecular Neuropsychiatry and Development Laboratory.

Dr. Vincent's team found defects in the TCTN2 gene occurring in a family in Pakistan, in which

four siblings had Joubert syndrome. The syndrome occurs when a child inherits a defective copy of the gene from each parent.

The researchers based in California led the mapping of a network of potential proteins related to ciliopathies that interact on common pathways, as an approach to identify the disease genes.

The study was a collaboration of researchers from six countries and more than a dozen centres. The lead authors were Peter Jackson and Liyun Sang at Genentech and Jeremy Reiter at UCSF. Other collaborators included Friedhelm Hildebrandt at the University of Michigan at Ann Arbor, Dan Doherty at the University of Washington in Seattle, Muhammad Ansar of Quaid-e-Azam University in Islamabad, and Julie Miller at Stanford University.

Dr. Vincent previously discovered the CC2D2A gene, which is also associated with Joubert syndrome. Defects on this gene account for about 9 percent of cases of Joubert syndrome. To date, 10 genes have been connected with Joubert syndrome.

Provided by Centre for Addiction and Mental Health

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