

Discovery of a gene responsible for familial scoliosis

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The discovery of the first gene causing familial scoliosis was announced by an international France-Canada research team today. Credit: Michael Perez

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"Mystery surrounds the cause of scoliosis, which is a three dimensional deformation of the vertebral column. Many researchers have been attempting to uncover the origins of this disease, particularly from a genetic point of view," explained leading co-author Dr Florina Moldovan of the University of Montreal and the CHU Sainte Justine research hospital. "To date, many genes have been suspected of causing scoliosis amongst different populations, but the gene that causes the familial form of the disease remained unknown. Our discovery of this first causative gene is due to the support of the Fondation Yves Cotrel and our international teamwork, in particular with leading co-author Dr. Patrick Edery of CHU de Lyon hospital and Dr. Pierre Drapeau of the CRCHUM."

A variation in the POC5 gene was initially identified by DNA sequencing (exome sequencing) in the samples Dr Patrick Edery collected from a large French family, of whom several members are affected by idiopathic scoliosis. Others variants of the POC5 gene were detected in scoliotic families and in people whose scoliosis had no precedence in their families. "The POC5 gene encodes for a centrosomal protein involved in microtubule-organising centres and cellular polarity," explained first author Dr. Shunmoogum (Kessen) Patten, who undertook his post-doctoral work at the CHU Sainte-Justine and CHUM research centres. "The pathogenicity of POC5 variants was documented by using the zebrafish, a well-established genetic animal model that has a spine." This model revealed that the over-expression of mutated human POC5 gene led to the rotational deformation of the anterior-posterior axis of the spine in half of the zebrafish embryos. The deformations are similar to the deformations observed in scoliosis patients.

The data suggest that the mutations are dominant, confirming the human genetic analysis. Interestingly, the protein is strongly expressed in the brain, within very precise structures in the midbrain. This leads the research team to believe that there is an association between the brain



and <u>idiopathic scoliosis</u>. "This is a very heterogenous disease and probably more than one gene is required for disease expression. This discovery has enabled the identification of the first causative gene and represents an important step towards decoding its genetic causes," Dr. Moldovan said. "This crucial first step will open the door to future studies that will identify the complementary genes and pathways that play a role in scoliosis in other populations. In particular, a full portrait of genetic events would enable the perfecting of effective preventative methods and strategies for understanding <u>scoliosis</u>," said Dr. Drapeau.

Provided by University of Montreal

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