A world first: The discovery of a common genetic cause of autism and epilepsy

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Researchers from the CHUM Research Centre (CRCHUM) have identified a new gene that predisposes people to both autism and epilepsy.

Led by the neurologist Dr. Patrick Cossette, the research team found a severe mutation of the synapsin gene (SYN1) in all members of a large French-Canadian family suffering from epilepsy, including individuals also suffering from autism. This study also includes an analysis of two cohorts of individuals from Quebec, which made it possible to identify other mutations in the SYN1 gene among 1% and 3.5% of those suffering respectively from autism and epilepsy, while several carriers of the SYN1 mutation displayed symptoms of both disorders.

"The results show for the first time the role of the SYN1 gene in autism, in addition to epilepsy, and strengthen the hypothesis that a deregulation of the function of synapse because of this mutation is the cause of both diseases," notes Cossette, who is also a professor with the Faculty of Medicine at the Université de Montréal.

He adds that "until now, no other genetic study of humans has made this demonstration."

The different forms of autism are often genetic in origin and nearly a third of people with autism also suffer from epilepsy. The reason for this comorbidity is unknown. The synapsin gene plays are crucial role in the development of the membrane surrounding neurotransmitters, also referred to as synaptic vesicles. These neurotransmitters ensure communication between neurons. Although mutations in other genes involved in the development of synapses (the functional junction between two neurons) have previously been identified, this mechanism has never been proved in epilepsy in humans until the present study.

More information: The results of the present study were published in the latest online edition of *Human Molecular Genetics*
www.hmg.oxfordjournals.org/sea ... s&fulltext=SYN1+loss

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