New gene discovered for new form of intellectual disability

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The Centre for Addiction and Mental Health (CAMH) has discovered a new form of intellectual disability involving mental retardation (MR) along with the eye defect retinitis pigmentosa (RP). CAMH also discovered the previously unidentified gene that causes this disorder, CC2D2A.

This scientific advance will help understand the developmental and biological processes involved in brain development, and may help identify ways to diagnose and treat intellectual disabilities.

Under the direction of Dr. John Vincent, scientist at CAMH, the team identified a mutation in CC2D2A that causes the production of a shortened protein missing the C2, or calcium-binding, domain. This protein mutation results in faulty cell function, which leads to MR with RP.

Most genes for intellectual disabilities that have been found so far are on the X chromosome. As Dr. Vincent explains, this mutation was found on the autosome (The 22 pairs of non-sex chromosomes, that make up the 46 chromosomes in the human body). Autosomal-recessive inheritance (where both mother and father carry a gene mutation on one chromosome, but both maternal and paternal copies must be passed on to the offspring to cause the disorder) is believed to be relatively common in intellectual disability, though only four genes causing this type of disability have been identified to date.

”What’s really exciting is that the new gene, CC2D2A, encodes a protein with domains similar to those found in one of the previous four autosomal recessive MR genes. This link could suggest a common function that is essential for normal brain development,” says Dr. Vincent.

Dr. Vincent and his team will continue exploring these initial findings, to help identify more people with mutations affecting the CC2D2A gene. This additional research will provide scientists more clues to understand, diagnose and treat intellectual disabilities.

Intellectual disabilities, also known as developmental delay or mental retardation, are a group of disorders defined by deficits in cognitive and adaptive development. Impacting between one and three percent of the population, a higher proportion of men are affected by this type of disability.

Source: Centre for Addiction and Mental Health