Autism, intellectual delay could be result of spontaneous mutations

Genetic variants across the genome contribute to about 8 percent of the risk for certain developmental conditions — much more than previously thought, according to a study published [September 16] in Nature .

The study looked at nearly 7,000 people who have a condition of brain development, such as intellectual disability, developmental delay or autism. The participants are all severely affected, suggesting that their conditions are the result of rare mutations, some of which are spontaneous or noninherited.

The new study found that common variants — those present in more than 5 percent of the population — are also important, however.

Scanning the sequences from the participants, the researchers found that certain combinations of a subset of common variants increase risk of the conditions. These combinations affect the severity of an individual's condition and yield a 'polygenic risk score' for that individual; this may explain why the same rare mutation can have diverse effects in different individuals.

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The participants all have the hallmarks of conditions that arise from a rare, harmful mutation in an important gene. For example, most of them have additional complications, such as an inherited heart problem or bone defect.

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[Jeffrey Barrett's] team is trying to trace the pathways that their set of common variants affect. He says researchers may eventually be able to use an individual's set of common variants to gauge her predisposition to specific conditions.

Read full, original post: Study links subset of genetic variants to autism, intellectual disability