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Intellectual disability is frequently caused by non-hereditary genetic problems

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Mutations in a group of genes associated with brain activity frequently cause intellectual disability, according to a study led by scientists affiliated with the University of Montreal and the research centre at the Centre hospitalier universitaire Sainte-Justine. Intellectual disability is a severe handicap that affects between one and two percent of children worldwide. It can often be attributed to genetic causes, but the specific genes involved were mostly unknown. "The group of genes we identified all play important roles in nerve synapses, the structures that allow brain cells to rapidly transfer information," explained senior author Dr. Jacques Michaud. "These findings indicate that, in this case, intellectual disability occurs because there is a disruption in nerve cell communication."

Some intellectual disabilities are associated with physical abnormalities that indicate major genetic abnormalities. However, in other cases disabilities are not associated with obvious physical traits "We targeted non-syndromic patients," Michaud said. Together with his colleagues from the Synapse to Disease Project, Michaud analyzed DNA from the patients to identify mutations that were not inherited but were in fact newly formed. These are called de novo mutations. They studied 197 synaptic genes and identified de novo mutations in 10 of the 95 patients. Further studies indicated that all of these mutations affect nerve cell communication, which lead the team to conclude that at least two-thirds of the mutations are the definitive cause of the disorder.

"This finding aligns with our previous work that shows that de novo

mutations play important roles in disorders such as autism and schizophrenia," said co-author Guy Rouleau. "Our study indicates that a large fraction of cases with intellectual disability have a genetic origin but are not hereditary. These findings will lead to improved diagnostics," added co-author Fadi Hamdan.

More information: "Excess of De Novo Deleterious Mutations in Genes Associated with Glutamatergic Systems in Nonsyndromic Intellectual Disability" was published in the American Journal of Human Genetics

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