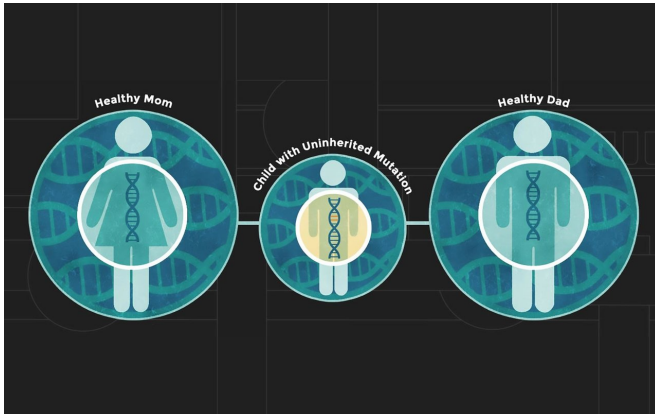


# Autism study finds de novo mutations associated with motor skill deficiencies

7 February 2018, by Bob Yirka



Mutations that appear in a child which are not present in either parent -- called de novo mutations -- can be important in autism. Severe, gene-disrupting de novo mutations are thought to be capable of causing the disorder in certain instances. New research shows that diminished motor skills, like low non-verbal IQ, correlate with the severity of de novo mutations. More broadly the study calls attention to role played by genetics in diminished cognitive functions in children across the autism spectrum. Credit: Dept. of Public Affairs, Cold Spring Harbor Laboratory

A team of researchers affiliated with several institutions in the U.S. has found an association between de novo mutations and motor skills in people with autism. In their paper published in *Proceedings of the National Academy of Sciences*, the group describes searching a database of information about people with autism and compared what they found with de novo mutations and the patterns that emerged.

Autism is a mental condition characterized by communication difficulties, but has also been associated with [behavioral problems](#) and IQ and motor skill deficiencies. It is believed to be biological in origin, though the cause is not known. Because of the variety and degree of symptoms

associated with [autism](#), medical scientists have given the condition the broad name [autism spectrum disorder](#) (ASD). Prior research has found an association between de novo [mutations](#) (mutations that are present for the first time in a given [family](#)) and low IQ levels in some people with ASD, though it is not known what causes such mutations. In this new effort, the researchers have found the same to be true of many people with ASD that have motor skill deficiencies.

Suspecting that de novo mutations are likely present in ASD people with motor skill deficiencies, the researchers analyzed data in the Simons Simplex Collection, a database of information on people with ASD and their families. It currently holds information on 2,760 such families, which includes genome information as well as behavioral information provided by family members. To find an association, the researchers compared known de novo mutations in ASD people with described behavioral patterns. They report that they were able to identify 57 recurrent mutations that could be associated with motor skill deficiencies. They note that they did not see any associations between de novo mutations and IQ versus motor skill deficiencies, which suggests the two have different causes. They also report that they found that de novo mutation associations in motor skill deficiencies were more distinctly associated than were mutation associations for people with IQ deficiencies.

**More information:** Andreas Buja et al. Damaging de novo mutations diminish motor skills in children on the autism spectrum, *Proceedings of the National Academy of Sciences* (2018). [DOI: 10.1073/pnas.1715427115](https://doi.org/10.1073/pnas.1715427115)

## Abstract

In individuals with autism spectrum disorder (ASD), de novo mutations have previously been shown to be significantly correlated with lower IQ but not with the core characteristics of ASD: deficits in social

communication and interaction and restricted interests and repetitive patterns of behavior. We extend these findings by demonstrating in the Simons Simplex Collection that damaging de novo mutations in ASD individuals are also significantly and convincingly correlated with measures of impaired motor skills. This correlation is not explained by a correlation between IQ and motor skills. We find that IQ and motor skills are distinctly associated with damaging mutations and, in particular, that motor skills are a more sensitive indicator of mutational severity than is IQ, as judged by mutational type and target gene. We use this finding to propose a combined classification of phenotypic severity: mild (little impairment of either), moderate (impairment mainly to motor skills), and severe (impairment of both IQ and motor skills).

[Press release](#)

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APA citation: Autism study finds de novo mutations associated with motor skill deficiencies (2018, February 7) retrieved 27 April 2021 from <https://medicalxpress.com/news/2018-02-autism-de-novo-mutations-motor.html>

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