# Large autism mutation risk does not increase as parents age, study finds

Spontaneous, or de novo, mutations can occur in a sperm or egg cell, or very early on in an embryo's development. Many of these changes involve a single DNA letter, or base pair, and are known as 'point mutations.' They tend to accumulate over time in sperm and eggs and may contribute to higher rates of autism among children born to older parents, especially older fathers.

Larger mutations, such as deletions or duplications of DNA sequences, have also been linked to autism, but they are rarer and require sizeable cohorts to study. The new work examined structural variants involving 50 or more base pairs in two large groups of autistic people and their families.

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[T]he new findings from [researcher Aaron Quinlan's] lab make it clear that age plays a much smaller role for them than it does for point mutations.
The results are "fairly good evidence that if there is a parental age effect, it is weak," he says.



Point mutations and structural variants have different underlying mechanisms: The former tend to occur randomly during cell division and genome replication, whereas the latter are the result of cells repairing breaks in the chromosome.

"The rate at which those [chromosome breaks] occur don't really change as a function of age," Quinlan says.

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