

NEWS

# Genetics: Spontaneous mutations lead to mental retardation

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Point mutant: Single DNA base changes that arise spontaneously in children can lead to genetic disorders, such as mental retardation.

Spontaneous mutations that change a single DNA base account for a large proportion of cases of unexplained mental retardation, according to a study published in the December *Nature Genetics*.

In the past few years, studies have linked copy number variations — large duplications and deletions of DNA segments — to mental disorders, including autism, schizophrenia and mental retardation. Spontaneous, or *de novo*, single DNA base changes — which are not present in the parents — appear at a rate of 50 to 100 new mutations in every child, and also account for some genetic disorders.

Seven of ten individuals in the new study who have mental retardation without a known genetic cause **have a single DNA base change that is likely to have caused the disorder**, the researchers found. To identify these mutations, the researchers sequenced the coding sequence of all ten individuals with mental retardation and their unaffected parents.

Of 21,755 changes they found in total, the researchers found 6 *de novo* mutations — in six different individuals — that affect the function of genes known to be linked to mental retardation.

One other male participant inherited a mutation in an X-linked mental retardation gene from his mother, who the researchers determined had acquired it *de novo*.

The fact that *de novo* mutations account for a large proportion of cases of mental retardation could explain why these cognitive disorders persist throughout evolution, even though they often lead to a lower rate of reproduction, the researchers say.