

Amount of gene surplus determines severity of mental retardation in males

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Researchers have discovered a new explanation for differences in the severity of mental illness in males. The more excess copies of a certain gene, the more serious the handicap. The genetic defect is situated on the X-chromosome; and it is suspected that it is the amount of copies of the GDI1 gene that is responsible. The results are being published in the *American Journal of Human Genetics*, and are the result of work by the group of Guy Froyen connected to VIB, a life sciences research institute in Flanders, Belgium at the University of Leuven, in close collaboration with Hilde Van Esch of the Center for Human Genetics (University Hospital Leuven) and colleagues in Germany and Spain.

It is for first time that scientists have linked the degree of a [mental illness](#) to the number of copies of a gene on the [X-chromosome](#), normally present as a single copy in males. The mental handicap is much more severe in patients with 5 copies than in patients with 2 copies. An intermediate severity has been observed in case of 3 copies. In their publication, the scientists also present a new mechanism by which such defects can arise. This mechanism might also underlie other genetic disorders.

Differences in GDI1 production

Defects in the GDI1 gene have previously been found in a few XLMR (X-Linked [Mental Retardation](#)) patients. In these patients, the production of GDI1 in the brain is disturbed, which impedes the transfer of stimuli

in the brain. The new finding in this research is that over-production of GDI1 is also harmful. The higher the production, the greater the disruption of signals.

Only in male patients

The discovery was made through DNA research in several families in which only males are afflicted with a mental handicap. In such families, defects appear on the X-chromosome (thus the name X-Linked Mental Retardation). Males have only one version of the X-chromosome. Females have a reserve copy, through which defective information can be masked.

2 - 3% of the population has a mental handicap

Mental handicap occurs in 2 - 3% of the population. The handicap can be attributed to external factors, such as a deficiency in oxygen at birth, or to defects in the [DNA](#). When the cause is genetic (hereditary), exact identification of the defect is crucially important for providing the patient with the proper medical support or for assessing the risk involved for couples with child wish. Recent estimates are that a defect on the X-chromosome is the cause in about 12% of the patients. However, in over half of the XLMR patients, the responsible gene has not yet been identified. This research makes a contribution toward filling this gap in our knowledge.

Source: VIB (the Flanders Institute for Biotechnology) ([news](#) : [web](#))

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