# Single gene mutations in chromosome leading to mental impairment.

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## Introduction

The biological mechanism of MR should be disrupted by a single gene mutation. This gene produces a faulty protein that affects cellular connections, synaptic structure, and function, as well as disrupting working cellular pathways or processes. As a result of this widespread genetic and physiopathological mechanism, brain complex functions are hampered, with the clinical manifestation being a limited ability to comprehend information.

The genetic approach to MR, which combines the whole-genome analysis of karyotyping technology with the customised high-resolution analysis of the FISH test In around 5-20 percent of cases with idiopathic MR and numerous congenital abnormalities, genomic microarrays can detect uncommon, de novo, submicroscopic interstitial imbalances or CNVs, with a resolution 10-10000 times greater than traditional karyotyping, depending on the clinical selection of patients. The increased detection of novel microdeletion/microduplication syndromes is based on a precise genotype-phenotype correlation, which is defined as the relationship between similar chromosomal defects and clinical symptoms in affected people [1].

### *1p36 microdeletion syndrome*

Monosomy 1p36 is a well-known contiguous genes syndrome that accounts for 0.5-1.2 percent of all idiopathic MR cases and is regarded to be the most common terminal deletion in humans. This illness is characterised by microcephaly, a large and late closing anterior fontanel, a tower skull, a prominent forehead, straight eyebrows, deep-set eyes, a flat nasal bridge with midface hypoplasia, unusual ears, brachydactyly/camptodactyly, and short feet. Symptoms such as seizures, oropharyngeal dysphagia, and heart abnormalities are common.

### *2q23.1 microdeletion syndrome*

A-CGH syndrome was identified in people who had severe MR and severe speech impairment, as well as microcephaly, coarse facial characteristics, short stature, and epilepsy. Angelman, Rett, and Smith-Magenis syndromes are usually associated with stereotypic behaviours, irregular sleep habits, and a broad-based stride. The typical phenotype appears to be caused by MBD5 or EPC2 gene haploinsufficiency in the deleted genomic region.

### *2q37 deletion syndrome*

Del (2q37) syndrome is now a well-known disorder that affects around 30% of individuals and is characterised by facial dysmorphic characteristics, developmental delay, hypotonia, seizures, and severe abnormalities. Delinquency is frequently connected to psychiatric problems (2q37). Autism spectrum disorders are present in 24-35 percent of del(2q37) cases, although severe speech delay, stereotypic movements, aggressive behaviour, Attention-Deficit/Hyperactivity Disorder (ADHD), and Obsessive-Compulsive Disorder (OCD) are also common [2].

The presence of facial dysmorphic traits and congenital anomalies in a child with MR and other neuropsychiatric diseases, which are usually associated with low stature, obesity, brachydactyly, eczema, and hypotonia, should be taken seriously as a significant indicator of hallucination. (2q37).

### *7q11.3 microduplication syndrome*

The deletion of a 1.4-1.5 Mb area at 7q11.23 causes Williams-Beuren syndrome (WBS), while the reciprocal microduplication of this genomic region is less well understood. The clinical characteristics of patients with 7q11.23 microduplication appear to be varied, ranging from mild to severe MR. Dup 7q11.23 people exhibit the polar opposite neurobehavioral phenotype to WBS: instead of fluent expressive language, they have substantial speech delay and only modestly impaired visuospatial ability.

### *Down Syndrome (DS)*

Down syndrome is caused by chromosome 21 abnormalities. Children with Down syndrome may go through developmental periods that are comparable to those of typical children, but at a slower rate, in the areas of sensoriomotor, adaptive, and social interaction capacities. The developmental scores of young children with DS are similar to those of normal children in the Personal, Social, and Adaptive Domains. The Battelle Developmental Assessment's Communication and Cognitive Domains have less comparable developmental scores [3].

In DS children, cell density in late pregnancy (weeks 19–23) revealed a decreased neuron number than in early pregnancy. From foetal to neonatal age, the same cellular reduction is maintained in the hippocampus, parahippocampalgyrus, cerebellum, and neocortex. Reduced volumes of the

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hippocampus, entorhinal, frontal, prefrontal, and temporal cortices, as well as the amygdala, cerebellum, brain stem nuclei, and hypothalamic mammillary bodies, have been identified in children and adults with DS.

### *Wolf-hirschhorn syndrome*

The loss of genetic material at the end of the short (p) arm of chromosome 4(4p-) the critical region- 4p16.3. Wolf-Hirschhorn Syndrome (WHS) is defined by the loss of genetic material near the end of the short (p) arm of chromosome 4(4p-) the crucial region- 4p16.3. The size of the deletion varies from person to person; larger deletions are associated with more severe intellectual impairments and physical abnormalities.

The most common phenotypes include prenatal and postnatal growth delays, a Greek warrior helmet facial appearance with microcephaly, high forehead, broad bridge of the nose continuing to the forehead, hypertelorism, epicanthus, highly arched eyebrows, short philtrum, downturned mouth, micrognathia, and poorly formed ears, congenital heart and urinary defect, and skeletal anomalies, congenital heart and urinary defect, and skeletal anomalies. Central nervous system anomalies, sensory deficits (hearing and sight), hypotonia, and seizures round out the list of intellectual disorders. One-third of patients with WHS have mild to moderate mental impairment [4].

### *Cri du chat syndrome*

A variable-sized deletion on chromosome 5's short arm causes Cri du Chat syndrome (CdCS) (5p-). The deletion is usually terminal, 5p terminal, but interstitial deletion, de novo translocation, and familial translocation can also be seen.

Individuals with 5p15.3 deletion breakpoints, as well as those with p15.2 deletion breakpoints, experienced speech delays but no serious intellectual impairment. Aberrant brain lateralization has been associated to abnormal gene expression in this region, supporting the hypothesis of a separate location for speech delay distal to p15. Speech and language development is often delayed, and some people never acquire spoken language at all. Despite the fact that they are both delayed, their receptive language is superior to their expressive language. 5p15.3 deletion breakpoints were associated with less cognitive impairment and fewer behavioural difficulties than p15.2 deletion breakpoints.

Severe forms of MR are thought to be caused by larger chromosomal abnormalities or single gene deficits, which can usually be diagnosed with specialised genetic tests. Paediatricians should be alerted if there is inexplicable MR, many congenital anomalies, neurological and mental problems, and/or minor dysmorphisms. On the other hand, many children with MR and dysmorphisms may not have major malformations and just have a different appearance than their unaffected siblings [5].

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