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Types of Chromosomal Abnormalities Which are resulting in Mental Impairment

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Introduction

A single gene mutation should interfere with the biological process of MR. This gene codes for a defective protein that can disrupt functioning cellular pathways or processes, affecting cellular connections, synaptic structure, and function. The brain complex functions are compromised as a result of this common hereditary and physiopathological mechanism, with the clinical manifestation being a restricted ability to process information.

The genetic approach to MR, which incorporates the karyotyping technology's whole-genome analysis and the FISH test's tailored high-resolution Depending on the clinical selection of patients, genomic microarrays can detect uncommon, de novo, submicroscopic interstitial imbalances or CNVs in around 5-20% of instances with idiopathic MR and numerous congenital abnormalities, with a resolution 10-10000 times greater than traditional karyotyping [1]. The increased recognition of new microdeletion/microduplication syndromes is based on a precise genotype-phenotype correlation, which is defined by the connection of comparable chromosomal abnormalities and clinical manifestations among afflicted individuals.

1p36 microdeletion syndrome

Monosomy 1p36 is a well-known contiguous genes syndrome that is thought to be the most frequent terminal deletion in humans, accounting for 0.5-1.2% of all idiopathic MR cases. Microcephaly, a big and late closing anterior fontanel, a tower skull, a prominent forehead, straight eyebrows, deep-set eyes, a flat nasal bridge with midface hypoplasia, atypical ears, brachydactyly/camptodactyly, and small feet are all characteristics of this disease [2]. Seizures, oropharyngeal dysphagia, and cardiac abnormalities are all frequent symptoms.

2q23.1 microdeletion syndrome

A-CGH discovered this novel condition in individuals with severe MR and severe speech impairment, as well as microcephaly, coarse facial features, low height, and epilepsy. The clinical impression of Angelman, Rett, or Smith-Magenis syndromes is frequently characterised by stereotypic behaviours, abnormal sleep patterns, and a broad-based stride. The usual phenotype appears to be caused by haploinsufficiency of MBD5 or EPC2 genes found in the deleted genomic region [3].

2q37 deletion syndrome

Del(2q37) syndrome is now a well-known condition marked by facial dysmorphic traits, developmental delay, hypotonia, epilepsy, and severe abnormalities in around 30% of patients. Psychiatric disorders are commonly linked to delinquency (2q37). In 24-35% of del (2q37) cases, autism spectrum disorders are present, but severe speech delay, stereotypic movements, aggressive behaviour, attention-deficit/hyperactivity disorder (ADHD), and obsessive-compulsive disorder are also prevalent.

The appearance of facial dysmorphic features and congenital abnormalities in a kid with MR and various neuropsychiatric illnesses, which are commonly linked with short height, obesity, brachydactyly, dermatitis, and hypotonia, should be regarded strongly indicative of delusion. (2q37) [4].

7q11.3 microduplication syndrome

Williams-Beuren syndrome (WBS) is one of the most well-known microdeletion syndromes, caused by the deletion of a 1.4-1.5 Mb area at 7q11.23, although the reciprocal microduplication of this genomic region is less well understood. Patients' clinical phenotypes for 7q11.23 microduplication appear to range, ranging from moderate to severe MR. The neurobehavioral phenotype of dup 7q11.23 individuals is the polar opposite of

WBS: instead of fluent expressive language, they have significant speech delay and only moderately impaired visuospatial abilities.

Down syndrome(DS)

Abnormalities in chromosome 21 results in down syndrome. In the domains of sensoriomotor, adaptive, and social interaction abilities, children with Down syndrome may exhibit developmental phases that are similar to those of normal children, although at a slower rate. In the Personal, Social, and Adaptive Domains, developmental scores of young children with DS are similar to those of typical children. In the Battelle Developmental Assessment's Communication and Cognitive Domains, developmental scores are less comparable [5].

In DS children, cell density indicated a lower neuron number in late pregnancy (weeks 19–23) compared to early pregnancy. In the hippocampus, parahippocampalgyrus, cerebellum, and neocortex, the same cellular decrease is maintained from foetal to neonatal age. In children and adults with DS, reduced volumes of the hippocampus, entorhinal, frontal, prefrontal, and temporal cortices, amygdala, cerebellum, brain stem nuclei, and hypothalamic mammillary bodies have been discovered.

Wolf-Hirschhorn syndrome

The deletion of genetic material near the end of the short (p) arm of chromosome 4(4p-) the crucial region- 4p16.3. Wolf-Hirschhorn syndrome (WHS) is characterised by a loss of genetic material near the end of the short (p) arm of chromosome 4(4p-) the key region- 4p16.3. The amount of the deletion varies from person to person; bigger deletions are linked to more severe intellectual disabilities and physical deformities than smaller deletions.

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 are the most common phenotypes. The panel of intellectual disabilities is completed by central nervous system abnormalities, sensorial defects (hearing and sight), hypotonia, and seizures. Mild to moderate mental impairment affects one-third of people with WHS [6].

Cri du Chat syndrome

Cri du Chat syndrome (CdCS) is caused by a variable-sized deletion on chromosome 5's short arm (5p-). In the majority of instances, the deletion is terminal, 5p terminal, but we can also discover interstitial deletion, de novo translocation, and familial translocation.

Individuals with speech delay but no severe intellectual impairment had 5p15.3 deletion breakpoints, as did those with p15.2 deletion breakpoints. They also had a lower degree of cognitive impairment and fewer behavioural issues than those with p15.2 deletion breakpoints. Aberrant gene expression in this region has been linked to abnormal cerebral lateralisation, supporting the idea of a distinct area for speech delay distal to p15. Individuals' speech and language development is generally delayed, and others never develop spoken language. Although both are delayed, their receptive language is better than their expressive language. Subjects with 5p15.3 deletion breakpoints exhibited less cognitive impairment and fewer behavioural issues than those with p15.2 deletion breakpoints.

Severe types of MR are considered to be caused by bigger chromosomal abnormalities or single gene deficiencies, which may be detected with specialised genetic tests in the majority of instances. The existence of unexplained MR, numerous congenital abnormalities, neurological and psychiatric symptoms, and/or mild dysmorphisms should alert paediatricians. Many children with MR and dysmorphisms, on the other hand, may not have significant deformities and merely have a look that is different from that of their unaffected relatives.

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