19p13.13 Deletion Syndrome

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19p13.13 deletion syndrome is a condition that results from a chromosomal change in which a small piece of chromosome 19 is deleted in each cell. The deletion occurs on the short (p) arm of the chromosome at a position designated p13.13

Keywords: genetic conditions

1. Introduction

Features commonly associated with this chromosomal change include an unusually large head size (macrocephaly), tall stature, and intellectual disability that is usually moderate in severity. Many affected individuals have significantly delayed development, including speech, and children may speak few or no words. Weak muscle tone (hypotonia) and problems with coordinating muscle movement (ataxia) contribute to delays in gross motor skills (such as sitting and walking) and fine motor skills (such as holding a pencil).

Other signs and symptoms that can occur with 19p13.13 deletion syndrome include seizures, abnormalities of brain structure, and mild differences in facial features (such as a prominent forehead). Many affected individuals have problems with feeding and digestion, including constipation, diarrhea, vomiting, and abdominal pain. Eye problems that can impair vision are also common. These include eyes that do not point in the same direction (strabismus) and underdevelopment of the optic nerves, which carry visual information from the eyes to the brain.

The signs and symptoms of 19p13.13 deletion syndrome vary among affected individuals. In part, this variation occurs because the size of the deletion, and the number of genes it affects, varies from person to person.

2. Frequency

This condition appears to be rare. About 10 affected individuals have been described in the medical literature.

3. Causes

People with 19p13.13 deletion syndrome are missing anywhere from about 300,000 DNA building blocks (300 kilobases or 300 kb) to more than 3 million DNA building blocks (3 megabases or 3 Mb) on the short arm of chromosome 19. The region of the deletion is usually referred to as p13.13, although some publications refer to it as p13.2. The region is the same; only the numbering differs. The exact size of the deletion varies among affected individuals, but it is thought to include at least 16 genes. This deletion affects one of the two copies of chromosome 19 in each cell.

The signs and symptoms of 19p13.13 deletion syndrome result from the loss of multiple genes in the deleted region. Some of these genes are suspected to have important roles in normal growth and development, and the loss of one copy of each of these genes likely underlies delayed development, intellectual disability, and the other features of this condition. For example, studies suggest that deletion of the *CACNA1A* gene may cause seizures in affected individuals. Researchers are working to determine which missing genes contribute to the other specific features of the disorder.

3.1. The genes and chromosome associated with 19p13.13 deletion syndrome

- CACNA1A
- CALR
- chromosome 19

3.2. Additional Information from NCBI Gene:

- BEST2
- MAST1
- NFIX

4. Inheritance

19p13.13 deletion syndrome is typically not inherited but results from the deletion of a chromosomal segment during the formation of reproductive cells (eggs and sperm) or in early fetal development. Most affected people have no history of the disorder in their family.

5. Other Names for This Condition

- 19p13.13 microdeletion
- 19p13.13 microdeletion syndrome
- chromosome 19p13.13 deletion syndrome

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