Chromosome 18

Description

Humans normally have 46 chromosomes in each cell, divided into 23 pairs. Two copies of chromosome 18, one copy inherited from each parent, form one of the pairs. Chromosome 18 spans about 78 million DNA building blocks (base pairs) and represents approximately 2.5 percent of the total DNA in cells.

Identifying genes on each chromosome is an active area of genetic research. Because researchers use different approaches to predict the number of genes on each chromosome, the estimated number of genes varies. Chromosome 18 likely contains 200 to 300 genes that provide instructions for making proteins. These proteins perform a variety of different roles in the body.

Health Conditions Related to Chromosomal Changes

The following chromosomal conditions are associated with changes in the structure or number of copies of chromosome 18.

Distal 18q deletion syndrome

Distal 18q deletion syndrome occurs when a piece of the long (q) arm of chromosome 18 is missing. The term "distal" means that the missing piece (deletion) occurs near one end of the chromosome arm. The signs and symptoms of distal 18q deletion syndrome include delayed development and learning disabilities, short stature, weak muscle tone (hypotonia), foot abnormalities, and a wide variety of other features.

The deletion that causes distal 18q deletion syndrome can occur anywhere between a region called 18q21 and the end of the chromosome. The size of the deletion varies among affected individuals. The signs and symptoms of distal 18q deletion syndrome are thought to be related to the loss of multiple genes from this part of the long arm of chromosome 18. Researchers are working to determine how the loss of specific genes in this region contributes to the various features of the disorder.

Proximal 18q deletion syndrome

Like distal 18q deletion syndrome (described above), proximal 18q deletion syndrome is a chromosomal condition that occurs when a piece of the q arm of chromosome 18 is missing. The term "proximal" means that in this disorder the deletion occurs near the

center of the chromosome, in an area between regions called 18q11.2 and 18q21.2. The size of the deletion varies among affected individuals. Proximal 18q deletion syndrome can lead to a wide variety of signs and symptoms among affected individuals, including delayed development and intellectual disability, recurrent seizures (epilepsy), behavioral problems, and characteristic facial features. These signs and symptoms are likely caused by the loss of specific genes in the deleted region.

Tetrasomy 18p

Tetrasomy 18p results from the presence of an abnormal extra chromosome, called an isochromosome 18p, in each cell. An isochromosome is a chromosome with two identical arms. Normal chromosomes have one long (q) arm and one short (p) arm, but isochromosomes have either two q arms or two p arms. Isochromosome 18p is a version of chromosome 18 made up of two p arms.

Cells normally have two copies of each chromosome, one inherited from each parent. In people with tetrasomy 18p, cells have the usual two copies of chromosome 18 plus an isochromosome 18p. As a result, each cell has four copies of the short arm of chromosome 18. (The word "tetrasomy" is derived from "tetra," the Greek word for "four.") The extra genetic material from the isochromosome disrupts the normal course of development, causing intellectual disability, delayed development, and the other characteristic features of this disorder.

Trisomy 18

Trisomy 18 occurs when each cell in the body has three copies of chromosome 18 instead of the usual two copies, causing severe intellectual disability and multiple birth defects that are usually fatal by early childhood. (The word "trisomy" comes from "tri," the Greek word for "three.") In some cases, the extra copy of chromosome 18 is present in only some of the body's cells. This condition is known as mosaic trisomy 18.

Rarely, trisomy 18 is caused by an extra copy of only a piece of chromosome 18. This condition is known as partial trisomy 18. Partial trisomy 18 occurs when part of the q arm of chromosome 18 becomes attached (translocated) to another chromosome during the formation of reproductive cells (eggs and sperm) or very early in embryonic development. Affected individuals have two copies of chromosome 18, plus the extra material from chromosome 18 attached to another chromosome. If only part of the q arm is present in three copies, the physical signs of partial trisomy 18 may be less severe than those typically seen in trisomy 18. If the entire q arm is present in three copies, individuals may be as severely affected as if they had three full copies of chromosome 18.

Researchers believe that extra copies of some genes on chromosome 18 disrupt the course of normal development, causing the characteristic features of trisomy 18 and the health problems associated with this disorder.

Other chromosomal conditions

Other disorders associated with chromosome 18 occur when pieces of the p arm of this

chromosome are missing or when extra genetic material from chromosome 18 is present. Researchers are uncertain how missing or extra pieces of chromosome 18 lead to the specific features of these disorders.

Partial monosomy of chromosome 18p (18p-) occurs when a piece of the p arm of this chromosome is deleted. Individuals with this condition often have short stature, a round face, large ears, a shortened space between the nose and mouth (philtrum), droopy eyelids (ptosis), and mild to moderate intellectual disability. About 10 to 15 percent of people with this condition have serious abnormalities of the brain and spinal cord (central nervous system). The lifespan of individuals with partial monosomy of chromosome 18p is typically not reduced, except when severe brain abnormalities are present.

Some people have a chromosome 18 with a circular structure, which is called a ring chromosome 18. This type of chromosome is formed when breaks occur at both ends of the chromosome and the broken ends join together to form a ring. Individuals with this chromosome abnormality often have intellectual disability, an unusually small head (microcephaly), widely spaced eyes (hypertelorism), low-set ears, and speech problems. The signs and symptoms associated with ring chromosome 18 depend on how much genetic material is lost from each arm of the chromosome.

Additional Information & Resources

Additional NIH Resources

- National Human Genome Research Institute: Chromosome Abnormalities (https://www.genome.gov/about-genomics/fact-sheets/Chromosome-Abnormalities-Fact-Sheet)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28Chromosomes,+Human,+Pair+18%5BMAJR%5D%29+AND+%28Chromosome+18%5BTI%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D)

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