Chromosome 19

Description

Humans normally have 46 chromosomes in each cell, divided into 23 pairs. Two copies of chromosome 19, one copy inherited from each parent, form one of the pairs. Chromosome 19 spans about 59 million base pairs (the building blocks of DNA) and represents almost 2 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 19 likely contains about 1,500 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 19.

19p13.13 deletion syndrome

19p13.13 deletion syndrome results from the deletion of a small piece of the short (p) arm of chromosome 19 in each cell. Major features of 19p13.13 deletion syndrome include an unusually large head size (macrocephaly), tall stature, delayed development of speech and motor skills (such as sitting and walking), and intellectual disability that is usually moderate in severity. Seizures, feeding and digestive difficulties, and eye abnormalities are also common.

People with this condition 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 the features of this condition. Researchers are working to determine which missing genes contribute to which specific features of the disorder.

Other chromosomal conditions

Other changes in the number or structure of chromosome 19 can have a variety of effects on growth and development. These chromosomal changes can cause delayed development, intellectual disability, feeding difficulties, hearing and vision impairment, heart problems, or other birth defects. The signs and symptoms that occur in a particular individual depend on the specific chromosomal change and which genes are involved.

Among the changes in chromosome 19 that have been reported are microdeletions, which remove a relatively small number of genes. These include 19p13.13 deletions (described above) and small deletions in other regions of the chromosome. Other possible changes include the presence of an extra piece of the chromosome in each cell (partial trisomy 19) or the absence of a larger segment of the chromosome in each cell (partial monosomy 19). Translocations of genetic material between chromosome 19 and another chromosome can also lead to extra or missing material from chromosome 19. Rarely, chromosome 19 forms a structure called a ring chromosome. Ring chromosomes occur when a chromosome breaks in two places and the ends of the chromosome arms fuse together to form a circular structure.

Cancers

Changes in chromosome 19 have been identified in several types of cancer. These chromosome abnormalities are somatic, which means they are acquired during a person's lifetime and are present only in the cells that give rise to cancer. Rearrangements of genetic material between chromosome 19 and one of several other chromosomes have been found in some forms of blood cancer (leukemia). These rearrangements, called translocations, appear to be particularly common in a type of leukemia called acute lymphoblastic leukemia (ALL). These translocations likely disrupt genes that are critical for keeping cell growth and division under control. Unregulated cell division can lead to the development of cancer.

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+19%5BMAJR%5D%29+AND+%2819%5BTI%5D%29+AND+english%5Bla%5D+A

ND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D)

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- UCSC Genome Browser: Statistics (http://genome.cse.ucsc.edu/goldenPath/stats.html)

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