Chromosome 20

Subjects: Genetics & Heredity
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Humans normally have 46 chromosomes in each cell, divided into 23 pairs. Two copies of chromosome 20, one copy inherited from each parent, form one of the pairs.

Keywords: chromosomes & mtDNA

1. Indtoduction

Chromosome 20 spans about 63 million DNA building blocks (base pairs) and represents approximately 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 20 likely contains 500 to 600 genes that provide instructions for making proteins. These proteins perform a variety of different roles in the body.

2. Health Conditions Related to Chromosomal Changes

2.1. Alagille syndrome

Approximately 7 percent of individuals with Alagille syndrome have small deletions of genetic material on chromosome 20, in a region known as 20p12. This region includes the *JAG1* gene, which is involved in signaling between neighboring cells during embryonic development. This signaling influences how the cells are used to build body structures in the developing embryo. Loss of the *JAG1* gene probably disrupts the signaling pathway. As a result, errors may occur during development, especially affecting the heart, bile ducts in the liver, the spinal column, and certain facial features.

2.2. Ring chromosome 20 syndrome

Ring chromosome 20 syndrome is caused by a chromosomal abnormality known as a ring chromosome 20 or r(20). A ring chromosome is a circular structure that occurs when a chromosome breaks in two places and its broken ends fuse together. People with ring chromosome 20 syndrome have one copy of this abnormal chromosome in some or all of their cells.

It is not well understood how the ring chromosome causes the signs and symptoms of this syndrome. In some affected individuals, genes near the ends of chromosome 20 are deleted when the ring chromosome forms. Researchers suspect that the loss of these genes may be responsible for epilepsy and other health problems. However, other affected individuals do not have gene deletions associated with the ring chromosome. In these people, the ring chromosome may change the activity of certain genes on chromosome 20, or it may be unable to copy (replicate) itself normally during cell division. Researchers are still working to determine the precise relationship between the ring chromosome 20 and the characteristic features of the syndrome.

2.3. Cancers

Changes in chromosome 20 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 certain cells. Deletions involving the long (q) arm of chromosome 20 appear to be common in blood-related cancers such as leukemia and lymphoma. Deletions of this chromosomal region have also been identified in other disorders of the blood and bone marrow, including polycythemia vera (which causes an overproduction of red blood cells) and myelodysplastic syndrome (which leads to a shortage of healthy blood cells).

Researchers are working to determine which genes on chromosome 20 are disrupted in these conditions. Studies suggest that some genes on the long arm of the chromosome may play critical roles in controlling the growth and division of cells.

2.4. Other chromosomal conditions

Deletions or duplications of genetic material from chromosome 20 can have a variety of effects, including intellectual disability, delayed development, distinctive facial features, skeletal abnormalities, and heart defects. Several different changes in the structure of chromosome 20 have been reported. These include an extra segment of the short (p) or long (q) arm of the chromosome in each cell (partial trisomy 20p or 20q) or a missing segment of the short or long arm of the chromosome in each cell (partial monosomy 20p or 20q).

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