Chromosome 3

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Humans normally have 46 chromosomes in each cell, divided into 23 pairs. Two copies of chromosome 3, one copy inherited from each parent, form one of the pairs.

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1. Introduction

Chromosome 3 spans about 198 million base pairs (the building blocks of DNA) and represents approximately 6.5 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 3 likely contains 1,000 to 1,100 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. 3p deletion syndrome

3p deletion syndrome is a condition that often results in intellectual disability, developmental delay, and abnormal physical features. 3p deletion syndrome is caused by the deletion of the end of the small (p) arm of chromosome 3. The size of the deletion varies among affected individuals, from approximately 150,000 DNA building blocks (base pairs) to 11 million base pairs and can include 4 to 71 known genes. In some individuals, the deletion involves material near the end of the chromosome but does not include the tip (the telomere).

The signs and symptoms related to 3p deletion syndrome result from the loss of genes in the 3p region; however, it is difficult to determine which genes influence specific features because not all affected individuals are missing the same genes.

2.2. 3q29 microdeletion syndrome

3q29 microdeletion syndrome is a condition that results from the deletion of a small piece of chromosome 3 in each cell. Features associated with the deletion vary widely but can include delayed development, intellectual disability, behavioral and psychiatric disorders, and physical abnormalities. Some individuals with this chromosomal change have very mild or no related signs and symptoms.

Most people with 3q29 microdeletion syndrome are missing about 1.6 million base pairs, also written as 1.6 megabases (Mb), on the long (q) arm of the chromosome at a position designated q29. It is the same region of chromosome 3 that is abnormally copied (duplicated) in people with 3q29 microduplication syndrome (described below). This chromosome segment is normally surrounded by short, repeated sequences of DNA that make it prone to rearrangement during cell division. The rearrangement can lead to missing or extra copies of DNA at 3q29.

The segment that is most often deleted in people with 3q29 microdeletion syndrome includes about 20 genes. Some of these genes are thought to be involved in brain development. However, it is unknown which specific genes, when abnormally deleted, are related to the signs and symptoms of 3q29 microdeletion syndrome. It is also unclear why some people with a deletion at 3q29 have no associated health problems. It is possible that genetic changes outside the 3q29 region can influence the features of this condition.

2.3. 3q29 microduplication syndrome

3q29 microduplication syndrome is a condition that results from the duplication of a small piece of chromosome 3 in each cell. Signs and symptoms related to this duplication vary widely. Some individuals with the duplication have no apparent signs or symptoms, or the features are very mild. Other individuals have delayed development and intellectual disability or learning difficulties. Eye abnormalities, heart defects, and an unusually small head (microcephaly) can also occur.

Most people with 3q29 microduplication syndrome have an extra copy of about 1.6 Mb of DNA at position q29 on chromosome 3. It is the same region of chromosome 3 that is deleted in people with 3q29 microdeletion syndrome (described above). This chromosome segment is prone to rearrangement during cell division, which can lead to extra or missing copies of DNA at 3q29.

The duplicated segment of 3q29 includes about 20 genes. Some of these genes are thought to be involved in brain and eye development. However, it is unknown which specific genes, when abnormally copied, are related to the varied signs and symptoms of 3q29 microduplication syndrome. It is also unclear why some people with a duplication at 3q29 have no associated health problems. It is possible that genetic changes outside the 3q29 region can influence the features of this condition.

2.4. Other chromosomal conditions

Other changes in the structure of chromosome 3 in each cell can have a variety of effects, including intellectual disability, developmental delay, distinctive facial features, birth defects, and other health problems. Changes to chromosome 3 include extra (duplicated) or deleted segments of the p arm or q arm of the chromosome in each cell. The size of the extra or deleted segments varies; the amount of genetic material involved contributes to the signs and symptoms that develop. Rarely, chromosome 3 can form a circular structure called a ring chromosome, which occurs when a chromosome breaks in two places and the ends of the chromosome arms fuse together. When the ring chromosome forms, genes near the ends of chromosome 3 are deleted, and because of the ring shape, the chromosome cannot copy (replicate) itself normally during cell division, likely contributing to health problems.

2.5. Cancers

Changes in chromosome 3 have been identified in a type of kidney cancer called clear cell renal carcinoma. This cancer can develop when one copy of chromosome 3 is missing or when part of the p arm of chromosome 3 is deleted. Additionally, clear cell renal carcinoma can be associated with abnormal exchanges of genetic material, called translocations, between chromosome 3p and another chromosome. Unlike the changes that cause the syndromes described above, the genetic changes associated with clear cell renal carcinoma are somatic, which means they are acquired during a person's lifetime and are present only in certain kidney cells. These genetic changes allow the cells to grow and divide in an uncontrolled way to form a tumor.

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