Chromosome 13

Humans normally have 46 chromosomes in each cell, divided into 23 pairs. Two copies of chromosome 13, one copy inherited from each parent, form one of the pairs. Chromosome 13 is made up of about 115 million DNA building blocks (base pairs) and represents between 3.5 and 4 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 13 likely contains 300 to 400 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 13.

8p11 myeloproliferative syndrome

A rearrangement (translocation) of genetic material involving chromosome 13 has been identified in most people with a rare blood cancer called 8p11 myeloproliferative syndrome. This condition is characterized by an increased number of white blood cells (myeloproliferative disorder) and the development of lymphoma, a blood-related cancer that causes tumor formation in the lymph nodes. The myeloproliferative disorder usually develops into another form of blood cancer called acute myeloid leukemia. 8p11 myeloproliferative syndrome most commonly results from a translocation between chromosome 13 and chromosome 8, written as t(8;13)(p11;q12). This genetic change fuses part of the *ZMYM2* gene on chromosome 13 with part of the *FGFR1* gene on chromosome 8. The translocation occurs only in cancer cells.

The protein produced from the normal *FGFR1* gene can turn on cellular signaling that helps the cell respond to its environment, for example by stimulating cell growth. The protein produced from the fused *ZMYM2-FGFR1* gene leads to constant FGFR1 signaling. The uncontrolled signaling promotes continuous cell growth and division, leading to cancer.

Feingold syndrome

Feingold syndrome type 2 is caused by genetic changes that remove (delete) small pieces of DNA from the long (q) arm of chromosome 13. These mutations are known as 13q31.3 microdeletions. Feingold syndrome type 2 is characterized by abnormalities of the fingers and toes, particularly shortening of the second and fifth fingers (brachymesophalangy). Other common features include an unusually small head size (microcephaly) and learning disabilities. 13q31.3 microdeletions involved in this condition delete the *MIR17HG* gene and sometimes part or all of other nearby genes. Loss of the *MIR17HG* gene is thought to underlie the characteristic features of the disorder, although loss of other genes may play a role in some cases.

The *MIR17HG* gene provides instructions for making the miR-17~92 microRNA (miRNA) cluster, which includes six different miRNAs. MiRNAs are short pieces of RNA, a chemical cousin of DNA. These molecules control gene activity (expression) by blocking protein production. MiRNAs in the miR-17~92 cluster help regulate signaling pathways that direct several cellular processes involved in growth and development before birth.

Deletion of one copy of the *MIR17HG* gene reduces the amount of miR-17~92 cluster miRNAs available to control the activity of specific genes during development. While it is likely that the resulting disruption of signaling pathways leads to the problems with growth and development characteristic of Feingold syndrome type 2, it is unclear exactly how a shortage of miR-17~92 cluster miRNAs causes the specific features of the condition.

Retinoblastoma

Retinoblastoma, a cancer of the light-sensing tissue at the back of the eye (the retina) that affects mostly children, is caused by abnormalities of a gene called *RB1*. This gene is located on a region of the q arm of chromosome 13 designated 13q14. Although most retinoblastomas are caused by mutations within the *RB1* gene, a small percentage of retinoblastomas result from a deletion of the 13q14 region.

In addition to retinoblastoma, deletions of the 13q14 region may cause intellectual disability, slow growth, and characteristic facial features such as prominent eyebrows, a broad nasal bridge, a short nose, and ear abnormalities. A loss of several genes is likely responsible for these developmental problems, although researchers have not determined which other genes in the deleted region are involved.

Trisomy 13

Trisomy 13 occurs when each cell in the body has three copies of chromosome 13 instead of the usual two copies. Trisomy 13 can also result from an extra copy of chromosome 13 in only some of the body's cells (mosaic trisomy 13).

In some cases, trisomy 13 occurs when part of chromosome 13 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 13, plus extra material from chromosome 13 attached to another chromosome. People with this genetic change are said to have translocation trisomy 13. The physical signs of translocation trisomy 13 may be different from those typically seen in trisomy 13 because only part of chromosome 13 is present in three copies.

Researchers believe that extra copies of some genes on chromosome 13 disrupt the course of normal development, causing the characteristic features of trisomy 13 and the increased risk of medical problems associated with this disorder.

Other cancers

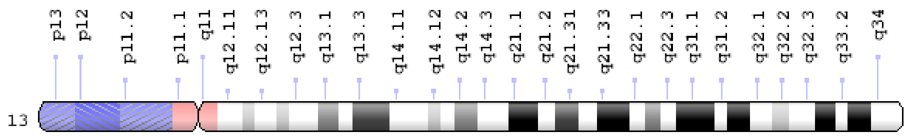
Changes in chromosome 13 have been associated with several types of cancer. These genetic changes are somatic, which means they are acquired during a person's lifetime and are present only in certain cells. The loss of genetic material from the middle of chromosome 13 is common in cancers of blood-forming cells (leukemias), cancers of immune system cells (lymphomas), and other related cancers.

Other chromosomal conditions

Partial monosomy and partial trisomy of chromosome 13 occur when a portion of the q arm of this chromosome is deleted or duplicated, respectively. The effect of missing or extra chromosome material varies with the size and location of the chromosome abnormality. Affected individuals may have developmental delay, intellectual disability, low birth weight, skeletal abnormalities, and other physical features.

Chromosome Diagram

Geneticists use diagrams called idiograms as a standard representation for chromosomes. Idiograms show a chromosome's relative size and its banding pattern, which is the characteristic pattern of dark and light bands that appears when a chromosome is stained with a chemical solution and then viewed under a microscope. These bands are used to describe the location of genes on each chromosome.



Credit: Genome Decoration Page/NCBI

Additional Information & Resources

Health Information from MedlinePlus

- Encyclopedia: Chromosome
  https://medlineplus.gov/ency/article/002327.htm

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://www.ncbi.nlm.nih.gov/pubmed?term=%28Chromosomes,+Human,+Pair+13%5BMAJR%5D%29+AND+%28Chromosome+13%5BTI%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Indexbychrom/idxa_13.html
- Cancer Genetics Web
  http://www.cancerindex.org/geneweb/clinkc13.htm
- Database of Genomic Variants: Chromosome 13
  http://projects.tcag.ca/variation/cgi-bin/tbrowse/tbrowse?source=hg17&table=Locus&show=table&keyword=&flop=AND&fcol=_C19&fcomp==&fkwd=chr13&cols=
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