FOXP2 Syndrome

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Chromosome 7

Chromosome 7

Description

Humans normally have 46 chromosomes in each cell, divided into 23 pairs. Two copies of chromosome 7, one copy inherited from each parent, form one of the pairs. Chromosome 7 spans about 159 million DNA building blocks (base pairs) and represents more than 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 7 likely contains 900 to 1,000 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 7.

7q11.23 duplication syndrome

7q11.23 duplication syndrome, a condition that can cause a variety of neurological and behavioral problems as well as other abnormalities, results from an extra copy of a region on the long (q) arm of chromosome 7. This region is called the Williams-Beuren syndrome critical region (WBSCR) because its deletion causes a different disorder called Williams syndrome (described below), also known as Williams-Beuren syndrome. The region, which is 1.5 to 1.8 million DNA base pairs (Mb) in length, includes 25 to 27 genes.Extra copies of several of these genes likely contribute to the characteristic features of 7q11.23 duplication syndrome. Researchers are studying genes whose functions suggest that they might be related to particular features.

More About This Health Condition

FOXP2-related speech and language disorder

Several different changes affecting chromosome 7 can result in FOXP2-related speech and language disorder. These changes involve a region of the long (q) arm of chromosome 7 containing the FOXP2 gene. FOXP2-related speech and language disorder is an uncommon condition that affects the development of speech and language starting in early childhood. In some affected individuals, problems with speech and language are the only features of the condition. Other individuals also have delayed development of motor skills such as walking and tying shoelaces, and autism spectrum disorders, which are conditions characterized by impaired communication and social interaction.All of the genetic changes that underlie FOXP2-related speech and language disorder disrupt the activity of FOXP2, a critical gene for normal speech and language development. Some individuals with FOXP2-related speech and language disorder have a deletion that removes a

small segment of chromosome 7, including the FOXP2 gene and several neighboring genes.Â Other people with this condition have a variant (also known as a mutation) within the FOXP2 gene itself. Less commonly, FOXP2-related speech and language disorder results from a rearrangement of the structure of chromosome 7 (such as a translocation) or from inheriting two copies of chromosome 7 from the mother instead of one from each parent (a phenomenon called maternal uniparental disomy or maternal UPD, which is described in more detail with Russell-Silver syndrome, below). It remains unclear how having two maternal copies of chromosome 7 affects the activity of the FOXP2 gene.Additional features that are sometimes associated with FOXP2-related speech and language disorder, including delayed motor development and autism spectrum disorders, likely result from changes to other genes on chromosome 7. For example, in affected individuals with a deletion involving chromosome 7, a loss of FOXP2 is thought to disrupt speech and language development, while the loss of nearby genes accounts for other signs and symptoms. Â People with maternal UPD for chromosome 7 have FOXP2-related speech and language disorder as part of a larger condition called Russell-Silver syndrome (described below).

More About This Health Condition

Greig cephalopolysyndactyly syndrome

Abnormalities of chromosome 7 are responsible for some cases of Greig cephalopolysyndactyly syndrome, a disorder that affects development of the limbs, head, and face. These chromosomal changes involve a region of the short (p) arm of chromosome 7 that contains the GLI3 gene. This gene plays an important role in the development of many tissues and organs before birth.In some cases, Greig cephalopolysyndactyly syndrome results from a rearrangement (translocation) of genetic material between chromosome 7 and another chromosome. Other cases are caused by the

deletion of several genes, including GLI3, from the short arm of chromosome 7. The loss of multiple genes can cause a more severe form of this disorder called Greig cephalopolysyndactyly contiguous gene deletion syndrome. People with this form of the disorder have characteristic developmental problems involving the limbs, head, and face, along with seizures, developmental delay, and intellectual disability.

More About This Health Condition

Russell-Silver syndrome

Abnormalities involving the inheritance of chromosome 7 can cause Russell-Silver syndrome, a rare condition characterized by slow growth, distinctive facial features, delayed development, speech and language problems, and learning disabilities.People normally inherit one copy of each chromosome from their mother and one copy from their father. For most genes, both copies are expressed, or "turned on," in cells. For some genes, however, only the copy inherited from a person's father (the paternal copy) is expressed. For other genes, only the copy inherited from a person's mother (the maternal copy) is expressed. These parent-specific differences in gene expression are caused by a phenomenon called genomic imprinting. Chromosome 7 contains a group of genes that normally undergo genomic imprinting; some of these genes are active only on the maternal copy, while others are active only on the paternal copy.In 7 percent to 10 percent of cases of Russell-Silver syndrome, people inherit both copies of chromosome 7 from their mother (maternal UPD) instead of one copy from each parent. Maternal UPD causes people to have two active copies of some imprinted genes and no active copies of others. An imbalance in active maternal and paternal genes on chromosome 7 underlies the signs and symptoms of the disorder in these cases.

More About This Health Condition

Saethre-Chotzen syndrome

Abnormalities of chromosome 7 cause some cases of Saethre-Chotzen syndrome. This rare condition is characterized by the premature fusion of certain skull bones (craniosynostosis), which prevents the skull from growing normally and affects the shape of the head and face. The chromosomal changes involve a region of the short (p) arm of chromosome 7 that contains the TWIST1 gene. This gene plays an important role in early development of the head, face, and limbs.The chromosome abnormalities responsible for Saethre-Chotzen syndrome include translocations of genetic material between chromosome 7 and another chromosome, a rearrangement of genetic material within chromosome 7 (an inversion), or the deletion of a segment of chromosome 7. Each of these chromosomal changes alters or deletes the TWIST1 gene and may also affect nearby genes.When Saethre-Chotzen syndrome is caused by a chromosomal deletion instead of a variant within the TWIST1 gene, affected children are much more likely to have intellectual disability, developmental delay, and learning difficulties. These features are typically not seen in classic cases of Saethre-Chotzen syndrome. Researchers believe that a loss of other genes on the short arm of chromosome 7 may be responsible for these additional features.

More About This Health Condition

Williams syndrome

Williams syndrome is caused by the deletion of genetic material from the Williams-Beuren critical region at 7q11.23 (described above). Researchers believe that the characteristic features of Williams syndrome, which include mild to moderate intellectual disability or learning problems,

unique personality characteristics, distinctive facial features, and heart and blood vessel (cardiovascular) problems, are probably related to the loss of several of the genes in this region.While the genes related to Williams syndrome have been identified, the relationship between most of those genes and the signs and symptoms of Williams syndrome is under investigation or unknown.

More About This Health Condition

Other chromosomal conditions

Other changes in the number or structure of chromosome 7 can cause delayed growth and development, intellectual disability, distinctive facial features, skeletal abnormalities, delayed speech, and other medical problems. Changes in chromosome 7 include an extra copy of some genetic material from this chromosome in each cell (partial trisomy 7) or a missing segment of the chromosome in each cell (partial monosomy 7). In some cases, several DNA building blocks (nucleotides) are abnormally deleted or duplicated in part of chromosome 7. A circular structure called ring chromosome 7 is also possible. 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 the number or structure of chromosome 7 occur frequently in human cancers. These changes are typically somatic, which means they are acquired during a person's lifetime and are present only in tumor cells. Many forms of cancer are associated with damage to chromosome 7.

In particular, changes in this chromosome have been identified in cancers of blood-forming tissue (leukemias) and cancers of immune system cells (lymphomas). A loss of part or all of one copy of chromosome 7 is common in myelodysplastic syndrome, which is a disease of the blood and bone marrow. People with this disorder have an increased risk of developing leukemia.Studies suggest that some genes on chromosome 7 may play critical roles in controlling the growth and division of cells. Without these genes, cells could grow and divide too quickly or in an uncontrolled way, resulting in a cancerous tumor. Researchers are working to identify the genes on chromosome 7 that are involved in the development and progression of cancer.

Additional Information & Resources

Additional NIH Resources

National Human Genome Research Institute: Chromosome Abnormalities

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