FOXP2 Syndrome

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FOXP2-related speech and language disorder

Description

FOXP2-related speech and language disorder is a condition that affects the development of speech and language starting in early childhood. Affected individuals have a speech problem known as apraxia, which makes it difficult to produce sequences of sounds, syllables, and words. This condition results from abnormalities involving parts of the brain that plan and coordinate movements of the lips, mouth, and tongue. Children with apraxia typically say their first words later than other children. Their speech is often difficult to understand, although the clarity of speech improves somewhat over time. Some affected individuals also cannot cough, sneeze, or clear their throats.In addition to having problems with producing speech (expressive language), people with FOXP2-related speech and language disorder may have difficulty with understanding speech (receptive language). Some also have trouble with other language-related skills, such as reading, writing, spelling, and grammar. In some affected individuals, problems with speech and language are the only features of the condition. Others also have delayed development in other areas,

including motor skills such as walking and tying shoelaces, and autism spectrum disorders, which are conditions characterized by impaired communication and social interaction.

Frequency

FOXP2-related speech and language disorder appears to be a relatively uncommon cause of problems with speech and language development. The total prevalence of apraxia is estimated to be 1 to 2 in 1,000 people, and it is likely that FOXP2-related speech and language disorder accounts for only a small portion of cases.

Causes

As its name suggests, FOXP2-related speech and language disorder is caused by changes involving the FOXP2 gene. This gene provides instructions for making a protein called forkhead box P2, which appears to be essential for the normal development of speech and language. The forkhead box P2 protein is active in many different tissues, including the brain, both before and after birth. It acts as a transcription factor, which means that it helps control the activity of other genes. Researchers suspect that many of the genes targeted by forkhead box P2 play important roles in brain development and the connections between nerve cells.Several changes involving the FOXP2 gene can result in FOXP2-related speech and language disorder. Some affected individuals have a deletion that removes a small segment of chromosome 7, including the FOXP2 gene and several neighboring genes. Other people with this condition have 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). It remains unclear how having two maternal copies of chromosome 7 affects the activity of the FOXP2 gene.The genetic changes that underlie FOXP2-related speech and language disorder disrupt the activity of the FOXP2 gene. Because forkhead box P2 is a transcription factor, these changes affect the activity of other genes in the developing brain. Researchers are working to determine which of these genes are involved and how changes in their activity lead to abnormal speech and language development.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. People with maternal UPD for chromosome 7 have FOXP2-related speech and language disorder as part of a larger condition called Russell-Silver syndrome. In addition to speech and language problems, these individuals have slow growth, distinctive facial features, delayed development, and learning disabilities.

Learn more about the gene and chromosome associated with FOXP2-related speech and language disorder

FOXP2

chromosome 7

Inheritance

The inheritance pattern of FOXP2-related speech and language disorder depends on its genetic cause. Mutations within the FOXP2 gene and deletions of genetic material from chromosome 7 that include FOXP2 have an autosomal dominant pattern of inheritance, which means one copy of the altered gene or chromosome in each cell is sufficient to cause the disorder. In most cases, the condition results from a new (de novo) mutation or deletion that occurs during the formation of reproductive cells (eggs or sperm) or in early embryonic development. These cases occur in people with no history of the disorder in their family. Less commonly, an affected individual inherits the genetic change from a parent with the condition; in at least one large family, a FOXP2 gene mutation has been passed through several generations.When FOXP2-related speech and language disorder results from maternal UPD of chromosome 7 as part of Russell-Silver syndrome, the condition is not inherited. UPD occurs as a random event during the formation of reproductive cells (eggs and sperm) or in early embryonic development. Affected people with maternal UPD of chromosome 7 typically have no history of the disorder in their family.When the condition is caused by rearrangements of the structure of chromosome 7, its pattern of inheritance can be complex and depends on the specific genetic change.

Other Names for This Condition

Speech and language disorder with orofacial dyspraxia Speech-language disorder 1

Additional Information & Resources

Genetic Testing Information

Genetic Testing Registry: Childhood apraxia of speech

Genetic and Rare Diseases Information Center

Isolated childhood apraxia of speech

Patient Support and Advocacy Resources

National Organization for Rare Disorders (NORD)

Catalog of Genes and Diseases from OMIM

SPEECH-LANGUAGE DISORDER 1; SPCH1

Scientific Articles on PubMed

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