MED13L

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MED13L syndrome

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Description

MED13L syndrome is a developmental disorder characterized by developmental delay, intellectual disability, and minor differences in facial features. Additionally, some people with this condition have recurrent seizures (epilepsy) or heart abnormalities that are present from birth (congenital heart defects).Intellectual disability and developmental delay are usually moderate to severe in people with MED13L syndrome. Weak muscle tone (hypotonia) and delayed development of motor skills, such as sitting, standing, and walking, are early symptoms of the condition. After learning to walk, some affected individuals continue to have difficulty with coordination and balance (ataxia). Speech is also delayed, and most people with this condition develop only a few words or never learn to talk. People with MED13L syndrome may exhibit characteristics typical of autism spectrum disorder, including repetitive actions and difficulty with social interactions.Most people with MED13L syndrome have unusual facial features that consist of a depressed nasal bridge, a bulbous nasal tip, straight eyebrows, outside corners of the eyes that point upward (upslanting palpebral fissures), full cheeks, and an open mouth. Other facial features that sometimes occur are a pronounced double

curve of the upper lip (Cupid's bow), and a deep space between the nose and upper lip (philtrum).Different congenital heart defects can occur in MED13L syndrome. Affected individuals may have transposition of the great arteries, which is abnormal positioning of the large blood vessel that distributes blood from the heart to the rest of the body (aorta) and the artery that carries blood from the heart to the lungs (the pulmonary artery). Other congenital heart defects in MED13L syndrome include a hole between the two lower chambers of the heart (ventricular septal defect), a hole between the two upper chambers of the heart (patent foramen ovale), or a particular combination of heart defects known as tetralogy of Fallot.

Frequency

MED13L syndrome is a rare disorder that occurs in an estimated 1.6 per 100,000 newborns. More than 65 affected individuals have been reported in the scientific literature.

Causes

As its name suggests, MED13L syndrome is caused by mutations in a gene known as MED13L. This gene provides instructions for making a protein that helps regulate gene activity; it is thought to play an essential role in development both before and after birth. The MED13L gene mutations that cause this condition alter the function of the MED13L protein or reduce the amount of protein present, impairing normal control of gene activity. It is unclear how these changes lead to the particular developmental and physical features of MED13L syndrome.

Learn more about the gene associated with MED13L syndrome

MED13L

Inheritance

MED13L syndrome is inherited in an autosomal dominant pattern, which means one copy of the altered MED13L gene in each cell is sufficient to cause the disorder. Most cases of this condition result from new (de novo) mutations in the gene that occur 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. Very rarely, the condition is inherited from a parent with mosaicism. In these instances, the parent has a MED13L gene mutation in a small number of cells, including reproductive cells (eggs or sperm), and does not show any signs or symptoms of MED13L syndrome.

Other Names for This Condition

Asadollahi-Rauch syndrome ASRAS Cardiac anomalies-developmental delay-facial dysmorphism syndrome Developmental delay-facial dysmorphism syndrome due to MED13L deficiency Intellectual disability and distinctive facial features with or without cardiac defects MED13L haploinsufficiency syndrome MED13L-related intellectual disability MRFACD

Additional Information & Resources

Genetic Testing Information

Genetic Testing Registry: Cardiac anomalies - developmental delay - facial dysmorphism syndrome

Genetic and Rare Diseases Information Center

MED13L haploinsufficiency syndrome

Patient Support and Advocacy Resources

National Organization for Rare Disorders (NORD)

Catalog of Genes and Diseases from OMIM

IMPAIRED INTELLECTUAL DEVELOPMENT AND DISTINCTIVE FACIAL FEATURES WITH OR WITHOUT CARDIAC DEFECTS; MRFACD

Scientific Articles on PubMed

PubMed

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