MED13L syndrome

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 (https://www.ncbi.nlm.nih.gov/gtr/conditions/C4225208/)

Genetic and Rare Diseases Information Center

- MED13L haploinsufficiency syndrome (https://rarediseases.info.nih.gov/diseases/12999/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Catalog of Genes and Diseases from OMIM

- IMPAIRED INTELLECTUAL DEVELOPMENT AND DISTINCTIVE FACIAL FEATURES WITH OR WITHOUT CARDIAC DEFECTS; MRFACD (https://omim.org/entry/616789)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28MED13L+syndrome%5BTIAB%5D%29+OR+%28MED13L%5BTI%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

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Last updated May 1, 2019