Mucopolysaccharidosis type VI

Description

Mucopolysaccharidosis type VI (MPS VI), also known as Maroteaux-Lamy syndrome, is a progressive condition that causes many tissues and organs to enlarge, become inflamed or scarred, and eventually waste away (atrophy). Skeletal abnormalities are also common in this condition. The rate at which symptoms worsen varies among affected individuals.

People with MPS VI generally do not display any features of the condition at birth. They often begin to show signs and symptoms of MPS VI during early childhood. The features of MPS VI affect many bodily systems, including skeletal, cardiac, and respiratory.

MPS VI causes various skeletal abnormalities, including a large head (macrocephaly) with a buildup of fluid in the brain (hydrocephalus), distinctive-looking facial features that are described as "coarse," and a large tongue (macroglossia). Other skeletal features include short stature, joint deformities (contractures) that affect mobility, and dysostosis multiplex, which refers to multiple skeletal abnormalities seen on x-ray. Carpal tunnel syndrome develops in many children with MPS VI and is characterized by numbness, tingling, and weakness in the hands and fingers. People with MPS VI may develop a narrowing of the spinal canal (spinal stenosis) in the neck, which can compress and damage the spinal cord.

Cardiac problems in people with MPS VI typically includes heart valve abnormalities. Respiratory abnormalities in this condition may involve the airway becoming narrow, which leads to frequent upper respiratory infections and short pauses in breathing during sleep (sleep apnea).

Other features of MPS VI include an enlarged liver and spleen (hepatosplenomegaly), and a soft out-pouching around the belly-button (umbilical hernia) or lower abdomen (inguinal hernia). The clear covering of the eye (cornea) typically becomes cloudy, which can cause significant vision loss. People with MPS VI may also have recurrent ear infections and hearing loss. Unlike other types of mucopolysaccharidosis, MPS VI does not affect intelligence.

The life expectancy of individuals with MPS VI depends on the severity of symptoms. Without treatment, severely affected individuals may survive only until late childhood or adolescence. Those with milder forms of the disorder usually live into adulthood,

although their life expectancy may be reduced. Heart disease and airway obstruction are major causes of death in people with MPS VI.

Frequency

The incidence of MPS VI is unknown, although it is estimated to occur in 1 in 250,000 to 600,000 newborns.

Causes

Mutations in the *ARSB* gene cause MPS VI. The *ARSB* gene provides instructions for producing an enzyme called arylsulfatase B (also known as N-acetylgalactosamine-4-sulfatase), which is involved in the breakdown of large sugar molecules called glycosaminoglycans (GAGs). GAGs were originally called mucopolysaccharides, which is where this condition gets its name.

Mutations in the *ARSB* gene reduce or completely eliminate the function of arylsulfatase B. The lack of arylsulfatase B activity leads to the accumulation of GAGs within cells, specifically inside the lysosomes. Lysosomes are compartments in the cell that digest and recycle different types of molecules. Conditions such as MPS VI that cause molecules to build up inside the lysosomes are called lysosomal storage disorders. The accumulation of GAGs within lysosomes increases the size of the cells, which is why many tissues and organs are enlarged in this disorder. Researchers believe that the buildup of GAGs are toxic to cells and may also interfere with the functions of other proteins inside lysosomes, triggering inflammation and cell death. The loss of cells leads to atrophy of tissues and organs over time in MPS VI.

Learn more about the gene associated with Mucopolysaccharidosis type VI

- ARSB

Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- Arylsulfatase B deficiency
- Maroteaux-Lamy syndrome
- MPS VI
- MPS6
- Mucopolysaccharidosis 6

- Mucopolysaccharidosis VI
- Polydystrophic dwarfism

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Mucopolysaccharidosis type 6 (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0026709/)

Genetic and Rare Diseases Information Center

- Mucopolysaccharidosis type 6 (https://rarediseases.info.nih.gov/diseases/7095/index)

Patient Support and Advocacy Resources

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

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Mucopolysaccharidosis type VI%22)

Catalog of Genes and Diseases from OMIM

- MUCOPOLYSACCHARIDOSIS, TYPE VI; MPS6 (https://omim.org/entry/253200)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28Mucopolysaccharidosis+VI%5BMAJR%5D%29+AND+%28%28mucopolysaccharidosis+type+VI%5BTIAB%5D%29+OR+%28Maroteaux+Lamy+syndrome%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

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Last updated June 28, 2021