Sialuria

Description

Sialuria is a rare disorder that affects development. Infants with sialuria are often born with a yellow tint to the skin and the whites of the eyes (neonatal jaundice), an enlarged liver and spleen (hepatosplenomegaly), and unusually small red blood cells (microcytic anemia). They may develop a somewhat flat face and distinctive-looking facial features that are described as "coarse." Temporarily delayed development and weak muscle tone (hypotonia) have also been reported.

Young children with sialuria tend to have frequent upper respiratory infections and episodes of dehydration and stomach upset (gastroenteritis). Older children may have seizures and learning difficulties. In some affected children, intellectual development is nearly normal.

The features of sialuria vary widely among affected people. Many of the problems associated with this disorder appear to improve with age, although little is known about the long-term effects of the disease. It is likely that some adults with sialuria never come to medical attention because they have very mild signs and symptoms or no health problems related to the condition.

Frequency

Fewer than 20 cases of sialuria have been described worldwide. There are probably more people with the disorder who have not been diagnosed, as sialuria can be difficult to detect because of its variable features.

Causes

Variants (also called mutations) in the *GNE* gene cause sialuria. The *GNE* gene provides instructions for making an enzyme found in cells and tissues throughout the body. This enzyme is involved in a chemical pathway that produces sialic acid, which is a simple sugar that attaches to the ends of more complex molecules on the surface of cells. By modifying these molecules, sialic acid influences a wide variety of cellular functions, including cell movement (migration), the attachment of cells to one another (adhesion), signaling between cells, and inflammation.

A feedback system helps control the amount of sialic acid produced in cells. This system shuts off the enzyme produced from the *GNE* gene when no more sialic acid is

needed. The variants responsible for sialuria disrupt this feedback mechanism, resulting in an overproduction of sialic acid. This simple sugar builds up within cells and is excreted in urine. Researchers are working to determine how an accumulation of sialic acid in the body interferes with normal development in people with sialuria.

Learn more about the gene associated with Sialuria

- GNE

Inheritance

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. Most reported cases have occurred in people with no known history of the disorder in their family. These cases may represent new (de novo) variants in the gene that occur during the formation of reproductive cells (eggs or sperm) in an affected individual's parent or during early embryonic development.

Other Names for This Condition

- French type sialuria
- Sialuria, French type

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Sialuria (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0342853/)

Genetic and Rare Diseases Information Center

- Sialuria (https://rarediseases.info.nih.gov/diseases/4865/index)

Patient Support and Advocacy Resources

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

Clinical Trials

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

Catalog of Genes and Diseases from OMIM

- SIALURIA (https://omim.org/entry/269921)

Scientific Articles on PubMed

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

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