Vici syndrome

Description

Vici syndrome is a severe disorder that begins early in life and affects many body systems. It is characterized by abnormalities of the brain, immune system, heart, skin, and eyes. Other organs and tissues are less commonly affected.

A characteristic feature of Vici syndrome is a brain abnormality called agenesis of the corpus callosum, in which the tissue that connects the left and right halves of the brain (the corpus callosum) fails to form normally during the early stages of development before birth. A region of the brain known as the pons (pontine hypoplasia) may be underdeveloped in people with Vici syndrome. Affected individuals may also have lower levels of myelin, which is a fatty substance that covers and protects nerve cells. In addition to problems with brain development, breakdown (degeneration) of brain tissue may occur over time, resulting in an unusually small head size (microcephaly).

These brain problems contribute to profound developmental delays in individuals with Vici syndrome. Affected infants have weak muscle tone (hypotonia). Generally, children with Vici syndrome are not able to roll or sit, and those that can may lose this skill when they get older. In addition, affected children cannot walk or speak.

Another characteristic feature of Vici syndrome is impaired immune function (immune deficiency), which leads to recurrent infections that can be life-threatening. Respiratory infections are the most common type of infection, though gastrointestinal and urinary tract infections also frequently occur.

A potentially life-threatening heart condition called cardiomyopathy is common in children with Vici syndrome. This condition, which worsens over time, makes it difficult for the heart to pump blood efficiently. Some affected children also have heart defects that are present from birth (congenital).

.cf0{font-style:italic;font-family:Segoe UI;font-size:9pt;}People with Vici syndrome may have skin and hair that are lighter in color than that of family members and other people with the same ethnic background (hypopigmentation). They may also experience clouding of the lenses of the eyes (cataracts) or other eye abnormalities, which may reduce their ability to see.

Other, less common signs and symptoms of Vici syndrome include seizures; hearing loss caused by abnormalities of the inner ear (sensorineural hearing loss); an opening

in the upper lip (cleft lip) with or without an opening in the roof of the mouth (cleft palate) or other unusual facial features; and abnormal function of the thyroid, liver, or kidneys. Many affected infants grow and gain weight more slowly than expected.

Most people with Vici syndrome do not survive beyond childhood, though this can vary widely.

Frequency

Vici syndrome is a rare disorder, though its exact prevalence is unknown. Approximately 100 individuals have been diagnosed with this condition.

Causes

Variants (also called mutations) in the *EPG5* gene cause Vici syndrome. This gene provides instructions for making a protein that is involved in a cellular process called autophagy. Cells use this process to recycle or break down worn-out or unnecessary cell parts. Autophagy also helps cells use materials efficiently when energy demands are high. In addition to its role in autophagy, the EPG5 protein aids in the body's immune response to foreign invaders such as bacteria and viruses.

.cf0{font-style:italic;font-family:Segoe UI;font-size:9pt;}Some variants in the *EPG5* gene cause the gene to produce abnormal EPG5 proteins that do not respond properly to foreign invaders. This leads to recurrent infections. In addition, autophagy is impaired. . cf0{font-style:italic;font-family:Segoe UI;font-size:9pt;}Researchers believe that problems with autophagy can disrupt the normal development and survival of cells in the brain and other organs and tissues; however, they do not fully understand how this disruption leads to the signs and symptoms of Vici syndrome.

Learn more about the gene associated with Vici syndrome

- EPG5

Inheritance

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

Other Names for This Condition

- Absent corpus callosum cataract immunodeficiency
- Corpus callosum agenesis-cataract-immunodeficiency syndrome
- Dionisi Vici Sabetta Gambarara syndrome
- Dionisi-Vici-Sabetta-Gambarara syndrome

- Immunodeficiency with cleft lip/palate, cataract, hypopigmentation and absent corpus callosum

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Vici syndrome (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1855772/)

Genetic and Rare Diseases Information Center

- Vici syndrome (https://rarediseases.info.nih.gov/diseases/448/index)

Patient Support and Advocacy Resources

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

Catalog of Genes and Diseases from OMIM

- VICI SYNDROME; VICIS (https://omim.org/entry/242840)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28Vici+syndrome%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2160+days%22%5Bdp%5D)

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Last updated August 28, 2023