Otulipenia

Description

Otulipenia is characterized by abnormal inflammation throughout the body. Inflammation is a normal immune system response to injury and foreign invaders (such as bacteria). However, the uncontrolled inflammation that occurs in otulipenia can damage many of the body's tissues and organs, including the gastrointestinal system, joints, and skin. Disorders such as otulipenia that result from abnormally increased inflammation are known as autoinflammatory diseases.

Signs and symptoms of otulipenia usually begin within the first few weeks of life, with recurring episodes of fever; diarrhea; painful, swollen joints; and skin rashes. The skin rashes are due to inflammation of the layer of fatty tissue under the skin (panniculitis), which causes painful red bumps. Some people with otulipenia have an abnormal distribution of fatty tissue in their bodies (lipodystrophy). Affected infants have difficulty growing and gaining weight at the expected rate (failure to thrive). Damage to the body's tissues and organs caused by inflammation is life-threatening if the condition is not treated.

Frequency

The prevalence of otulipenia is not known. At least four cases have been reported in the medical literature.

Causes

Otulipenia is caused by mutations in the *OTULIN* gene. The protein produced from this gene helps control inflammation. Inflammation can be turned on by a cellular process called ubiquitination, in which molecules called ubiquitin are attached to certain proteins. In particular, signaling pathways that lead to inflammation are stimulated by the attachment of chains of ubiquitin molecules linked end-to-end (linear ubiquitin chains). The OTULIN protein helps control inflammation by removing these linear ubiquitin chains.

OTULIN gene mutations that cause otulipenia lead to production of an OTULIN protein with reduced function. As a result, removal of linear ubiquitin chains is impaired, and signaling pathways that cause inflammation are abnormally active. The excessive inflammation that results causes fever, diarrhea, panniculitis, and the other signs and

symptoms of otulipenia.

Learn more about the gene associated with Otulipenia

- OTULIN

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

- AIPDS
- Autoinflammation, panniculitis, and dermatosis syndrome
- ORAS
- OTULIN-related autoinflammatory syndrome

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Infantile-onset periodic fever-panniculitis-dermatosis syndrome (https://www.ncbi.nlm.nih.gov/gtr/conditions/C4310614/)

Genetic and Rare Diseases Information Center

- Infantile-onset periodic fever-panniculitis-dermatosis syndrome (https://rarediseases.info.nih.gov/diseases/13198/index)

Patient Support and Advocacy Resources

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

Catalog of Genes and Diseases from OMIM

- AUTOINFLAMMATION, PANNICULITIS, AND DERMATOSIS SYNDROME, AUTOSOMAL RECESSIVE; AIPDSB (https://omim.org/entry/617099)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28otulin+deficiency%29+OR+%28otulipenia%29%29+AND+english%5Bla%5D+AND+%22last+3600+days%22%5Bdp%5D)

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