Adult polyglucosan body disease

Description

Adult polyglucosan body disease (APBD) is a condition that affects the nervous system. People with APBD typically first experience signs and symptoms related to the condition between ages 35 and 60. Initial symptoms of the disorder include numbness and tingling in the legs (peripheral neuropathy) and progressive muscle weakness and stiffness (spasticity). As a result, affected individuals can have an unsteady gait, poor balance, and an increased risk of falling.

Damage to the nerves that control bladder function, a condition called neurogenic bladder, is another feature that often occurs early in the course of APBD. Affected individuals have increasing difficulty starting or stopping the flow of urine.

Eventually, most people with APBD lose the ability to control their bladder and bowel functions and their limbs. Damage to the autonomic nervous system, which controls body functions that are mostly involuntary, leads to problems with blood pressure, heart rate, breathing rate, digestion, temperature regulation, and sexual response, and results in daily bouts of exhaustion. About half of people with APBD experience a decline in intellectual function (dementia).

Frequency

APBD is a rare condition, although its exact prevalence is unknown. Approximately 200 affected individuals have been diagnosed worldwide. Recently these have included younger individuals who have not yet experienced signs or symptoms but who are diagnosed in the course of genetic screening when considering parenthood. Researchers suspect that the disorder may be underdiagnosed.

Causes

Mutations in the *GBE1* gene cause APBD. The *GBE1* gene provides instructions for making the glycogen branching enzyme. This enzyme is involved in the production of a complex sugar called glycogen, which is a major source of stored energy in the body. Most *GBE1* gene mutations that cause APBD result in a shortage (deficiency) of the glycogen branching enzyme, which leads to the production of abnormal glycogen molecules. These abnormal glycogen molecules, called polyglucosan bodies, accumulate within cells and cause damage. Nerve cells (neurons) appear to be

particularly vulnerable to the accumulation of polyglucosan bodies in people with this disorder, leading to impaired neuronal function.

Some mutations in the *GBE1* gene that cause APBD do not result in a shortage of glycogen branching enzyme. In people with these mutations, the activity of this enzyme is normal. How mutations cause the disease in these individuals is unclear. Other people with APBD do not have identified mutations in the *GBE1* gene. In these individuals, the cause of the disease is unknown.

Learn more about the gene associated with Adult polyglucosan body disease

- GBE1

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

- APBD
- Polyglucosan body disease, adult form

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Adult polyglucosan body disease (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1849722/)

Genetic and Rare Diseases Information Center

- Adult polyglucosan body disease (https://rarediseases.info.nih.gov/diseases/108/index)

Patient Support and Advocacy Resources

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

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Adult polyglucosan bod

y disease%22)

Catalog of Genes and Diseases from OMIM

- POLYGLUCOSAN BODY NEUROPATHY, ADULT FORM; APBN (https://omim.org/entry/263570)

Scientific Articles on PubMed

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

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