Adult polyglucosan body disease

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.

Inheritance Pattern

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

Diagnosis & Management

Genetic Testing Information

- What is genetic testing?
  /primer/testing/genetictesting
- Genetic Testing Registry: Polyglucosan body disease, adult
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C1849722/

Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22adult+polyglucosan+body+disease%22

Other Diagnosis and Management Resources

- GeneReview: Adult Polyglucosan Body Disease
  https://www.ncbi.nlm.nih.gov/books/NBK5300
- MedlinePlus Encyclopedia: Neurogenic Bladder
  https://medlineplus.gov/ency/article/000754.htm
- MedlinePlus Encyclopedia: Spasticity
  https://medlineplus.gov/ency/article/003297.htm

Additional Information & Resources

Health Information from MedlinePlus

- Encyclopedia: Neurogenic Bladder
  https://medlineplus.gov/ency/article/000754.htm
- Encyclopedia: Spasticity
  https://medlineplus.gov/ency/article/003297.htm

- Health Topic: Dementia
  https://medlineplus.gov/dementia.html
- Health Topic: Peripheral Nerve Disorders
  https://medlineplus.gov/peripheralnervedisorders.html

Genetic and Rare Diseases Information Center

- Adult polyglucosan body disease
  https://rarediseases.info.nih.gov/diseases/108/adult-polyglucosan-body-disease

Additional NIH Resources

- National Institute of Neurological Disorders and Stroke: Spasticity Information Page
  https://www.ninds.nih.gov/Disorders/All-Disorders/Spasticity-Information-Page

Educational Resources

- Boston Children's Hospital: Neurogenic Bladder
  http://www.childrenshospital.org/conditions-and-treatments/conditions/n/neurogenic-bladder
- JAMA Patient Page: Peripheral Neuropathy
  https://jamanetwork.com/journals/jama/fullarticle/185714
- MalaCards: adult polyglucosan body disease
  https://www.malacards.org/card/adult_polyglucosan_body_disease
- Orphanet: Adult polyglucosan body disease
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=206583
- Washington University, St. Louis: Neuromuscular Disease Center
  https://neuromuscular.wustl.edu/synmot.html#pgb

Patient Support and Advocacy Resources

- Adult Polyglucosan Body Disease Research Foundation
  https://www.apbdrf.org/
- Family Caregiver Alliance
  https://www.caregiver.org/
- National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/adult-polyglucosan-body-disease/

Clinical Information from GeneReviews

- Adult Polyglucosan Body Disease
  https://www.ncbi.nlm.nih.gov/books/NBK5300

Scientific Articles on PubMed

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

Catalog of Genes and Diseases from OMIM

- POLYGLUCOSAN BODY NEUROPATHY, ADULT FORM
  http://omim.org/entry/263570

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