Adult polyglucosan body disease

Adult polyglucosan body disease is a condition that affects the nervous system. People with this condition have problems walking due to reduced sensation in their legs (peripheral neuropathy) and progressive muscle weakness and stiffness (spasticity). Damage to the nerves that control bladder function, a condition called neurogenic bladder, causes affected individuals to have progressive difficulty controlling the flow of urine. About half of people with adult polyglucosan body disease experience a decline in intellectual function (dementia).

People with adult polyglucosan body disease typically first experience signs and symptoms related to the condition between ages 30 and 60.

Frequency

Adult polyglucosan body disease is a rare condition; although its exact prevalence is unknown, at least 70 affected individuals have been described in the medical literature.

Causes

Mutations in the \textit{GBE1} gene cause adult polyglucosan body disease. The \textit{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 \textit{GBE1} gene mutations 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 \textit{GBE1} gene that cause adult polyglucosan body disease 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 adult polyglucosan body disease do not have identified mutations in the \textit{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

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
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+AND%22last+3600+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

• POLYGLUCOSAN BODY NEUROPATHY, ADULT FORM
  http://omim.org/entry/263570
Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/14755501

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20301758

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/9851430

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17994551

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11260627

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/23034915
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4329926/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16730990

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12874416

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