FACT SHEET: Multiple Sclerosis… or APBD?

WHAT IS ADULT POLYGLUCOSAN BODY DISEASE (APBD)?

- a rare glycogen storage disease type IV caused by mutations in the glycogen branching enzyme gene
- 40 known pathogenic variants in GBE1 gene
- inherited in an autosomal recessive pattern
- characterized by a deficiency of glycogen branching enzyme, resulting in the accumulation of polyglucosan bodies in muscles, nerves, and other tissues
- the gene involved in causing APBD is the same gene at the center of Andersen's Disease (glycogen storage disease type IV), a severe liver condition affecting infants

WHAT IS THE SCOPE?

- to date, 200 diagnosed patients worldwide
- affects men and women in equal proportions
- diagnosed most frequently in Ashkenazi Jewish populations, but it is pan-ethnic

HOW IS IT DIAGNOSED?

- GBE assay (blood test)
- sequencing genomic DNA/messenger RNA (saliva test)
- histologic examination of sural nerve biopsy

APBD SYMPTOMS

- Symptoms and severity can vary greatly from one person to another
- Develop around the fifth decade of life
- Diminish quality of life by interfering with the ability to work, mobility, and independence

- A – Adult onset, Ashkenazi Jewish ancestry

Although most patients are of Ashkenazi Jewish descent, it is pan-ethnic

- P – Peripheral neuropathy

Numbness, weakness, stiffness, and pain in the lower limbs, and sometimes the upper limbs

- B – Bladder dysfunction

An increased need to urinate and urgency to urinate, progressing to a complete loss of bladder control

- D – Decreased energy

Overwhelming need to rest and sleep throughout the day

WHY IS APBD CONFUSED WITH MS?

- MS has a similar adult onset
- MS has some of the same symptoms as APBD
- Physicians often are not aware of APBD clinical and imaging features and mistake the disease for MS and other neurodegenerative conditions

WHAT CAN YOU DO?

A study published in May 2019 in the journal *Multiple Sclerosis and Related Disorders* suggests that nearly 1 in 5 people with other neurologic conditions are mistakenly diagnosed with MS. Misdiagnoses result in patients receiving unnecessary investigations and potentially harmful therapeutic interventions.

If you find yourself questioning your MS diagnosis, and if you have questions about APBD, talk with your clinician about these concerns. Consider a saliva test to diagnose APBD. Please contact the APBD Research Foundation to learn more about APBD and speak with our community of patients, health professionals, and caregivers.

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