



Yossi is slowly losing his life's greatest joy—the ability to run and play with his daughter.

Help us fight for all men and women APBD sufferers like Yossi

GREGORY WEISS, PRESIDENT  
APBD RESEARCH FOUNDATION  
2710 AVENUE S,  
BROOKLYN, NY 11229  
646 580 5610  
INFO@APBDRF.ORG  
WWW.APBDRF.ORG



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Contributions are tax deductible  
to the extent permitted by law.*

APBD  
ADULT  
POLYGLUCOSAN  
BODY DISEASE  
SHARES  
SYMPTOMS WITH  
ALS,  
MULTIPLE  
SCLEROSIS  
AND  
PROSTATE CANCER.  
IF YOU'RE A JEWISH  
MAN OR WOMAN  
OVER 35  
YOU NEED TO  
LEARN MORE.

## SYMPTOMS

- Adult onset (men and women)
- Problems walking
- Problems with urination
- Unexplained fatigue
- Some patients experience a mild cognitive decline

## PREVALENCE

- A recent study found the mutation that causes APBD has a carrier frequency of 1 in 34.5 Ashkenazi Jews.

<http://www.ncbi.nlm.nih.gov/pubmed/?term=22943850>

(This study still has to be validated.)

## LABORATORY FINDINGS

- Y329S mutation
- Low GBE1 enzyme activity
- Medulla and spine atrophy
- Neurogenic bladder

## TESTING

Simple blood test available for APBD

For testing information go to:

[www.APBDRF.org](http://www.APBDRF.org) "Living with APBD-testing info"

Contact us at [info@APBDRF.org](mailto:info@APBDRF.org)

**A**dult Polyglucosan Body Disease (APBD) is caused by a mutation in the glycogen branching enzyme (GBE1) gene. It generally begins to affect the patient's nervous system as early as age 35 and progresses slowly. The symptoms shared by almost all male and female APBD patients are gait disturbances, neurogenic bladder and GBE1 mutation. Mild cognitive decline may affect up to 50% of patients. Most APBD patients are of Ashkenazi Jewish heritage. Due to symptoms shared with other illnesses such as prostate cancer, dementia, ALS, Alzheimer's, Multiple Sclerosis, Peripheral Neuropathy and Spinal Stenosis, APBD is often misdiagnosed.

Gregory Weiss, a patient himself, founded the Adult Polyglucosan Body Disease Research Foundation (APBDRF) in 2005.

The Foundation is dedicated to:

- Improving the diagnosis and treatment of APBD
- Supporting individuals and families affected by the disease
- Increasing awareness of APBD among health professionals and the public

See a video of [Gregory's story](#) under personal narratives on the APBDRF website.

## PATIENT REGISTRY

The APBD Research Foundation, with the help of the Mailman School of Public Health at Columbia University and a team of international researchers and clinicians, is working to establish and maintain the first APBD registry. The team at Columbia is experienced in maintaining patient registries and follows IRB (Institutional Review Board) approved protocols to ensure security and privacy. The registry will be collecting patients' demographic data, family history, clinical observations, and the results of physical and neurological examinations.

Patients and their caregivers are encouraged to participate in the registry to aid the researchers studying the disease and the effects of various treatment programs. There are a number of research initiatives underway and the success of potential treatments will need to be tested over a period of time. The registry will enable the researchers to more effectively assess the results of upcoming treatment programs and will facilitate human trials in the foreseeable future.

For Registry Information, go to:

[www.APBDRF.ORG](http://www.APBDRF.ORG)