



## LETTER TO THE JEWISH COMMUNITY

We would like to alert you to a new and emerging understanding of a serious genetic disease affecting Ashkenazi Jewish adults. It's called Adult Polyglucosan Body Disease (**APBD**). APBD strikes Ashkenazi Jews as early as age 35, but more commonly between ages 40 and 60. Most people now affected by APBD have no idea that the disease is the cause of their symptoms. Physicians, even specialists who might be consulted, are almost always unaware of the disease. For those affected, then, life typically becomes a medical merry-go-round filled with misdiagnoses; unsuccessful surgeries and medications; canes; walkers; and wheelchairs.

Thanks to the collaborative efforts of medical research teams around the world (including Canada, Israel, the UK, Spain, France and the US), there have been a number of important advances in our understanding of APBD. This improved understanding includes genetic causes, simple methods to diagnose, and a variety of possible approaches to treatment.

**At its outset, there are four very common indicators among adult APBD patients:**

- Ashkenazi background
- Peripheral neuropathy (numbness, weakness, or tingling in the toes or feet)
- Bathroom frequency beyond the norm, or difficulty with bladder control
- Decreased energy -- We note that this symptom often develops a little bit later in the disease process.

**Anyone experiencing these symptoms should be tested for APBD.** The disease is progressive, and subsequent symptoms are more intense and more extensive. The disease shortens the patient's life, and the last few years follow a similar pattern to other debilitating degenerative diseases like ALS and MS.

A recent study estimated that about 1 out of 35 Jews of Ashkenazi descent carries a "p.Y329S" genetic defect that can result in APBD. For every 1,000 Ashkenazi Jews, about 30 may carry the "p.Y329S" defect. And very new research has identified a related genetic defect that makes the carrier rate higher yet.

A campaign to raise awareness about APBD was launched with a recent article in "The Jewish Daily Forward." You can read it by [clicking here](#) or following the link at the bottom of the page. The APBD Research Foundation website - [www.apbdrf.org](http://www.apbdrf.org) - holds a wealth of information. And of course, please feel free to email questions or concerns to me or our foundation's Executive Director Sharon Steinberg directly at [Sharon@apbdrf.org](mailto:Sharon@apbdrf.org).

B'Shalom,  
Gregory Weiss  
President  
APBD Research Foundation

P.S. The newly identified, related defect I mentioned above is suspected of also having a prenatal impact, likely causing miscarriages. For that reason, we are currently working to get APBD added to existing genetic screening panels. At this moment adult carriers need your help with diagnoses and support for medical intervention.

<http://forward.com/articles/203717/genetic-nerve-disease-puts-photographer-on-other-s/?p=all>