APBDRF
ADULT POLYGLUCOSAN BODY DISEASE RESEARCH FOUNDATION

Dear Rabbis and Jewish Community Leaders:

Mount Sinai School of Medicine in New York recently completed a frequency study to estimate the number of Ashkenazi Jews who are carriers for two specific gene mutations. When a baby inherits either of two mutated genes—one from each parent, a metabolic disorder known as Adult Polyglucosan Body Disease, (**APBD),** results. The frequency discovery: 1 out of 68 Ashkenazi Jews is a carrier of at least one of the studied gene mutations. That surprising finding compelled Mount Sinai to add APBD to its Comprehensive Jewish Carrier Screening Panel.

Unlike many of its companions on the screening panel, APBD presents itself in adulthood rather than in infancy or early childhood. Symptoms become noticeable to some adults when they're in their 30's. But more commonly, the indicators demand attention when the affected person is between ages 40 and 60.

Most who suffer from APBD have no idea of what's causing their symptoms. Physicians, even specialists who might be consulted, are almost always unaware of APBD. For those affected, then, life typically becomes a medical disaster filled with misdiagnoses; frustration; unsuccessful surgeries and medications; and canes, walkers, and wheelchairs.

At its outset, there are four very common indicators among adult APBD patients. We created a memory tool to help keep them in mind:

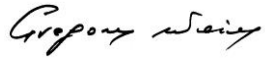
- **A -** Ashkenazi background
- **P -** Peripheral neuropathy (numbness, weakness, or tingling in the toes, fingers, hands, and/or feet)
- **B -** Bathroom frequency beyond the norm, or difficulty with bladder control
- **D -** Decreased energy -- We note that this symptom often develops a little bit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

Any Jewish adult experiencing these symptoms and searching for a diagnosis should talk to their physician about being tested for APBD. The disease is progressive and, the older one gets, the more intense and more extensive are the symptoms. The disease shortens the patient's life. The last few years follow a pattern similar to other debilitating degenerative diseases like ALS and MS.

Ashkenazi couples contemplating having children should be sure that any genetic testing they schedule screens them for APBD carrier status.

Would you use your connection with the Jewish community to put out the word about APBD? You could save many people many years of frustrating search. Additionally, with therapies and cures on the horizon, we need to get patients on our international registry.

A campaign to raise awareness about APBD was launched with an article in *The Jewish Daily Forward*. You can read it by clicking on this link. The APBD Research Foundation's website 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.



B'Shalom,
Gregory Weiss
President, APBD Research Foundation