



# APBDRF

ADULT POLYGLUCOSAN BODY DISEASE RESEARCH FOUNDATION

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## Letter from the President

Dear APBDRF Supporters,

The APBD Research Foundation will hold its 10th Scientific Advisory Board Meeting on Wednesday, December 4, 2013 in Midtown, Manhattan. As you know, there have been many new and exciting research developments that will be highlighted at the meeting. Below you can read about some of the new projects, resources, and talented people that have recently joined the fight to find a cure for APBD.

This year our veteran researchers:

Dr. Kakhlon, Dr. Minassian, Dr. Schiffmann (Mary Wallace) and Dr. Akman, will be joined by Dr. Yue of Oxford University and Richard Buchsbaum of Columbia University, both of whom will also be presenting at the meeting (see below). In addition, we will be receiving long anticipated results to a High Throughput Screening (HTS) study on the GBE from Dr. Michael Tropak. He has already moved into a new research lab and will no longer be working with the APBDRF after the presentation. We want to congratulate him on his new position and thank him for his hard work in obtaining and testing the GBE protein and bringing the research to where it is today.

What can you do?

Over this coming year, your involvement in our new registry both as registrants and workers is crucial. Getting the word out will make a difference in facilitating potential treatment in the foreseeable future. We cannot predict the future, but when looking back, we will ask ourselves did we do everything we could? Please get involved by sending in your personal narratives, (which can be anonymous, see below) share your ideas with Sharon ([sharon@apbdrf.org](mailto:sharon@apbdrf.org)) our new executive director and donate if you can. We need your help!

Sincerely,

Gregory Weiss  
APBDRF President



[Models](#)

[Human Trials for APBD](#)

Forward to a Friend

Join Our Mailing List!

Stay Connected



Adult Polyglucosan Body Disease: Natural History and Key Magnetic Resonance Imaging Findings

[Read here](#)

Triheptanoin Treatment Trial for Patients with Adult Polyglucosan Body Disease - *Currently the only human trial*

[Read here](#)

## Interesting Articles

Bits of Mystery DNA, Far From 'Junk', Play Crucial Role

[Read here](#)

Exon Skipping - Experimental drugs are being designed to repair genetic errors that cause muscular dystrophy-*Exon*

## What We are Working On

Our three research centers at Columbia University, Hadassah University Hospital, and Toronto University Sick Kids Hospital are working on several critical projects, which you can learn more about on our new [Donation Platform](#).



## Registry Announcement

The APBD Research Foundation is working with the Mailman School of Public Health at Columbia University, along with a team of international researchers and clinicians to establish and maintain an APBD registry. The team at Columbia is experienced maintaining patient registries and follows IRB approved protocols to ensure security and privacy. The registry will be hosted on a secure database which will be accessed via an encrypted website. We will be collecting patients' demographic data, family history, clinical observations, and the results of physical and neurological examinations.

## New Testing Information

Please know that a simple blood test for APBD is now available. [Click here](#) to see our new trifold brochure for patients and clinicians, as well as additional information and other resources available on our [website](#). At the same time, please contact us at [info@APBDRF.org](mailto:info@APBDRF.org) with any other questions.

## Ultragenyx Pharmaceuticals

We have some exciting news in regard to our human trials project led by [Raphael Schiffmann MD](#), with assistance from Mary Wallace, MSRD/LD. [Ultragenyx Pharmaceutical Inc](#), of Novato CA, has purchased licensing rights for triheptanoin, the oil used in the study ([A Treatment Trial of Triheptanoin in Patients with Adult Polyglucosan Body Disease](#)) conducted at [Baylor Research Institute](#). As part of the agreement, the company will provide travel funds for study subjects (those currently enrolled and any future participants) as well as a more purified form of triheptanoin for the study.

[Dr. Schiffmann](#) recently wrote, "regarding the Ultragenyx story, this emphasizes the importance of identifying clinical outcome measures and controlled clinical trials. As I noted on a number of occasions in the past, this study is not only important to see if triheptanoin is effective in APBD, but also to develop ways to assess efficacy of any potential future treatment for APBD."

Skipping may be important in 40% of APBD patients

[Read here](#)

Gene Therapy with a Difference

[Read here](#)

## Patient Corner

In this section you will find personal accounts of people affected by APBD. The APBDRF as a patient support group [does not](#) endorse any health practitioners, therapies, medicines, etc.

[Click here](#) to see our new trifold brochure for patients and clinicians

Thanks to the work of David Epstein, our organization has been included in the Resource Central Section of [Neurology Now](#) magazine. You can read it online here, and order a FREE subscription to the magazine as well

[Better Branches: Alma Hecht's personal blog about living with APBD](#)

## Fall 2013's Frequently Asked Question

*In every newsletter we will be addressing a different question related to APBD. If you have questions that you'd like answered in future newsletters please send them to [info@apbdrf.org](mailto:info@apbdrf.org)*

### Why can't you just inject the healthy GBE into APBD patients?

The enzyme, injected, will be immediately destroyed by enzymes in the blood, and will never even make it to the brain. Even if it made it to the brain, it will not be able to cross the blood-brain barrier and will be destroyed outside the barrier. (Thank you Dr. Minassian)

## New Scientist to Join APBDRF Meeting

We welcome Dr Wyatt W. Yue, who will attend the APBDRF Scientific Advisory Board Meeting for the first time this December. He leads a group at the [Structural Genomics Consortium, University of Oxford](#), where his primary interest is structural biology of inherited metabolic diseases and development of small molecule therapy. Further information about his work can be found [here](#).

His planned participation is in the following areas:

- Recombinant protein production of GBE1 (he has established a system that yields low milligram quantities for biochemical and structural work)
- Looking into the thermal stability of GBE1 protein comparing WT and mutants (e.g. Y329S)
- Characterizing small molecule hits (from various screening efforts) using in vitro and structural methods to evaluate binding and specificity.

## Part-time Executive Director

We welcome Sharon Steinberg, recently retired from Columbia University Pediatric Infectious Diseases where she worked for many years in the same laboratory doing actual antibody testing and pcr pre and post varicella and zoster vaccine reactions. She also managed a multi-center study for the licensure of the vaccine.

In addition, Sharon, who seems to have an extraordinary amount of energy, has always been an active volunteer for Jewish causes and organizations. She served as the president of the Metropolitan New York District of United Synagogue of Conservative Judaism for two years, and was the president of Women's League for Conservative Judaism Met branch (NYC) for two terms. She has worked with Camp Ramah in the Berkshires as their journal editor, and in many other volunteer capacities. Sharon has a certificate in Synagogue Management from Hebrew Union College and, until recently, served as administrator of her synagogue, Tifereth Israel, Town and Village Synagogue, managing caretakers and the synagogue calendar, among other duties. She also serves as ritual chair at the synagogue.

Sharon is excited to learn more about APBDRF and is ready to use her extensive experience and organizational skills to help our organization in its quest to raise awareness about APBD and develop treatments and a cure.

We are fortunate to have Sharon Steinberg working with us. We warmly welcome her to APBDRF.

[The Story of an APBD Patient by Phillip Adviv](#)

## Call for Personal Narratives

Each person's experience of living with APBD is unique. Here, real people living with APBD and their significant others, [share their stories](#) about adjusting to diagnosis, seeking treatment, and living with APBD. Your story, too, could inspire others whose lives have been affected by APBD. [Click here](#) to share your story today! (you can also submit anonymously)

## Allied Organizations

[Association for Glycogen Storage Disease](#)

[Association for Neuro-Metabolic Disorders \(ANMD\)](#)

[The Dana Foundation](#)

[The Doctor's Doctor](#)

[Genetic Alliance](#)

[Jewish Genetic Disease Consortium \(JGDC\)](#)

[Muscular Dystrophy Association](#)

[National Organization for Rare Disorders](#)

[National Tay-Sachs & Allied Diseases Association \(NTSAD\)](#)



[www.apbdrf.org](http://www.apbdrf.org)

APBDRF | [\(646\)-580-5610](tel:646-580-5610) | [info@apbdrf.org](mailto:info@apbdrf.org) |