Letter from the President

Hi All:

When the APBDRF was founded in 2005 on a small budget for orphan disease organizations, I never dreamed that we would have such a vibrant scientific collaboration. This fertile environment is nurturing some great researchers who are our heroes. Dr. H. Orhan Akman of Columbia University, in collaboration with Dr. Or Kakhlon of Hadassah Hospital and Dr. Berge Minassian of Sick Kids Hospital in Toronto, has made a discovery that will profoundly impact the understanding of APBD and also other diseases. [1]

As an autosomal recessive disease, one would expect that all APBD patients would present two mutated genes, one from mom and one from dad. But 40% of APBD patients don’t present that way. They’ve been called manifesting heterozygotes. That is, they express the disease, but only one mutated gene from one parent could be found; the other gene from the opposite parent appeared normal. Our researchers looked in a novel way at the GBE1 gene for a second mutation, and they found it! This sheds a light on a confounding phenomenon for a segment of the APBD population, as well as for manifesting heterozygotes who are in the ranks of patients having many other genetic diseases. The article about this milestone discovery appears now in JAMA Neurology. Click here to read.

And the news gets better. We are approaching several possible therapeutic treatments for APBD and will be reporting on them to you over the coming year.

I have even more to share with you. The Research Foundation has developed several videos that explain APBD at differing levels of complexity. To hear Dr. Akman explain his discovery in highly scientific terms, please click here. To hear the explanation at a mid-level of complexity, please click here. Our website, apbdrf.org, has a wealth of updated material, as well. If you’re just beginning to learn about APBD, I encourage you to start with this enlightening article "Rare Disease Gets the Spotlight of Discovery" by T. Anjannette Leven.

Can you handle more good news? The APBDRF Research Foundation has recently earned the Silver participation level from GuideStar.org. GuideStar collects, organizes, and presents information on non-profit organizations in an easy-to-understand format that holds great weight with donation providers, donor advised funds, foundations, grant providers, search engines, and more. Click here to see the GuideStar webpage with our Silver endorsement.

This is a very important moment in our fight to find a cure for APBD. Please get involved. We need:

- Help from the Jewish community, which we hope will understand their own self interest in getting to a cure for a disease that hides in its population. Here’s a letter that rabbis and leaders of Jewish communal organizations could share directly with their memberships.

- A strong outreach campaign to identify undiagnosed patients who are seeking an answer to a troubling combination of symptoms. Click here to see our video of APBD patients who describe the symptoms that sent them on a search for a diagnosis.

- A large contingency of supporters who are willing to work to keep the progress going. To help, please email me or our Executive Director Sharon Steinberg at Sharon@apbdrf.org.

- A robust, up-to-date patient registry. Click here if you’re a patient and haven’t yet registered.

- Assistance with funding. With our limited resources to date, see how much we have accomplished! But the costs associated with continued disease research
and the therapeutic trials that are on the horizon are astronomical. Please do [click here](http://apbdrf.org/about-us/contact-us) to donate! We need your support.

Thank you for taking interest. Thank you for taking action.

All the best,
Gregory Weiss
President, APBD Research Foundation

P.S.
If you have questions, comments, or insights of any kind, please contact me or Executive Director Sharon Steinberg at Sharon@apbdrf.org. We want to hear from you!

[1] Other diseases that have similarities to aspects of APBD are discussed [here](http://apbdrf.org/about-us/contact-us) on the APBDRF website.

### APBD registry update

PLEASE get your physician forms filled out and uploaded on a yearly basis onto the APBD registry. It is very important! Read how big data was used to help a Lupus patient in this [New York Times](http://www.lupusresearchinstitute.org/lupus-news/2014/10/06/big-data-shows-big-promise-lupus) article:

*Columbia University Discovery Videos*

*Columbia University Discovery of 2nd Mutation - for lay viewers*

*"New Discovery Informs Approach to Finding Cures"* 7:04

*Columbia University Discovery of 2nd Mutation - for lay viewers*

Columbia University Discovery of 2nd Mutation -for scientist viewers

Science Behind New Intronic Mutation Discovery 10:55
disease symptoms. “Tribe” members are constantly on
the lookout for tips and
ideas to help blunt their
challenges.
Visit apbdtribe.com, to see
what we have to share.

Searching for a
Diagnosis
click to open the brochure

Patients and Clinicians:
click to open our new trifold brochure

APBD in Neurology Now

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

The Story of an APBD
Patient by Phillip Adv

Article of
Interest:

The Search for a
Diagnosis
Having a rare disorder is
difficult. But having a rare
disorder and not knowing
what it is or how to treat it is
even worse. Find out how
long it takes between
symptoms and diagnosis.

Allied
Organizations

Association for Glycogen
Storage Disease
The Dana Foundation
The Doctor's Doctor
Foundation for Peripheral
Neuropathy
Genetic Alliance
Global Genes Project
Jewish Genetic Disease
Consortium (JGDC)
Muscular Dystrophy
Association
National Institutes of Health -
Office of Rare Diseases
Research
National Organization for
Rare Disorders

Columbia University Discovery of 2nd Mutation -
for scientist viewers

Dr Akman in APBD/Kindsight®

Dr. Hasan Orhan Akman
New York City
September, 2014

I’ve heard for some time about Dr. Akman and his pioneering research into the nature and possible cure for APBD, Adult Polyglucosan Body Disease, the untreatable, some call fatal, genetic disease with which I am afflicted and that currently has me paraplegic with other challenging symptoms. Based on what I’ve heard about him and his luminous navigation of uncharted genetic frontiers, I envision Dr. Akman as a white haired older sage in the vein of Gandolf or Yoda, with esoteric thoughts visibly circling his head like streaking comets. I am refreshingly surprised (and yet, not so surprised) when we meet to see a young man who could biologically be my son. We set out into the upper east side neighborhood and speak about many things, including his family, his native Turkey and APBD. I must admit that I get a little lost when he speaks of genetic mutations and such. We make some photos in the afternoon light and head back to the gallery where my KINDSIGHT® exhibit is on view and he expresses sincere appreciation for my stories about real people. I want to thank him in return for what he is doing, for bravery and forging a pathway of possibility and hope into the existence of uncertainty that those of us afflicted with APBD live with. I say to him a couple of the few Turkish words I know: “sag olun”. “Thank you for doing what you don’t have to do.”

SEE A PHOTO ALBUM OF APBD/KINDSIGHT® PIECES HERE

RARE DISEASE DAY: February 28, 2015
APBDRF Accepted into NORD

As of February 2, the APBD Research Foundation was accepted as a member organization in NORD, the National Organization for Rare Disorders. 

Through NORD, member organizations gain a stronger voice in rare disease visibility, advocacy, policy development, education, and research support.

Becoming a NORD member reflects well on the APBDRF. Acceptance was by no means a shoe-in. NORD reviewed and approved of our organizational infrastructure, as well as our actual work on behalf of APBD patients. They confirmed that the research foundation has appropriate by-laws, board membership, scientific advisors, financial budgeting, board meetings, and record-keeping. They reviewed our Annual Report and the educational materials that we distribute.

We’re quite proud of this achievement. Our thanks go to the many hands who developed our application package.

APBD is awarded Guidestar Level: Silver

Great News. The APBD Research Foundation has recently earned the Silver participation level from the GuideStar Exchange. This is a testament of the Foundation’s commitment to datatransparency. GuideStar collects, organizes, and presents the information on charitable organizations in an easy-to-understand format. By providing up-to-date information to GuideStar, we are ensuring that timely and accurate information about the APBDRF reaches the 10 million annual visitors to GuideStar’s website and millions of other viewers reached through GuideStar’s network of donation providers, search engines, donor advised funds, foundations, and more. This enables potential donors and grant providers with vital information about the Foundation.

The APBDRF would like to announce the formation of our web analytics team. We thank the following members for their ongoing work which include participating in monthly meetings with our Argentinian website and social media companies.

Volunteers
Gary Epstein
Joshua Goldman
David Epstein
Jeff Levenson

Staff
From Argentina:
Pirusm Digital Marketing-Social media consultants
Sofia Plager
Ezequiel Singer
Leonel Reckinger

Our Web Master
Brand & Insight-Gabriel Baril