

## In This Issue

Letter from the President

Videos

APBD Registry Update

APBD Registry Update

Prestigious Endorsement  
from Guidestar

Prestigious Endorsement  
from Guidestar

Prestigious Endorsement  
from Guidestar

APBDRF Thanks our Web  
Analytic Team

## Website Quick Links

[About Us](#)

[Donate](#)

[Do You See Yourself?](#)

[Letter to the Jewish Community](#)

[Living With APBD](#)

[The Patient Corner](#)

[Research](#)

[Resources](#)

[Videos](#)

[Sign up for the Registry](#)

[Instructions for Enrollment](#)

[Forward to a Friend](#)

[Join Our Mailing List!](#)

Stay Connected



## Recent Press and Blogger Coverage:

[The Argonaut News](#)

[Jewish Daily Forward](#)

[The Jewish Georgian](#)  
or see our ad on Page 32

[PLOS Blog](#)

[Center for Jewish Genetics](#)

[The Neuropathy Association](#)

From the  
Webmaster:

## 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 online. [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. Anjanette Levert.

Can you handle more good news? The APBD 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](mailto: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

[Click to see our new Landing Page](#)

and the therapeutic trials that are on the horizon are astronomical. Please do [click here](#) to donate! We need your support.

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

## 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.

## Contact Us

<http://apbdrf.org/about-us/contact-us>

## KINDSIGHT INITIATIVES

Follow the links to see our press for the following cities:

[NYC](#)  
[LA](#)

## VIDEOS



Akman short 1: Significance of 2nd APBD mutation discovery



Akman 2: Registry will facilitate clinical trials



Akman 3: Second mutation discovery's impact on other diseases



We Are APBD

## APBD TRIBE

While researchers chip away at finding the cause and cure for APBD, patients, partners, and care givers seek work-arounds for

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](mailto:Sharon@apbdrf.org). We want to hear from you!

[1] Other diseases that have similarities to aspects of APBD are discussed [here](#) 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](#) article:

<http://www.lupusresearchinstitute.org/lupus-news/2014/10/06/big-data-shows-big-promise-lupus>



## 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](http://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



Columbia University Discovery of 2nd Mutation -  
for scientist viewers

### [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 Adiv](#)

### **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](#)

### **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 braving 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**



## Rare Disease Day

[FAQ](#)

## 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

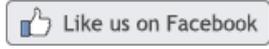
Gary Epstein  
Joshua Goldman  
David Epstein  
Jeff Levenson

### Staff

*From Argentina:*  
Pirsum Digital Marketing-Social media consultants  
Sofia Plager  
Ezequiel Singer  
Leonel Reckinger

### Our Web Master

Brand & Insight-Gabiel Baril



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

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