

APBDRF

Adult Polyglucosan Body Disease Research Foundation



Newsletter No. 1 March 2007

The Adult Polyglucosan Body Disease Research Foundation (APBDRF) is the only national non-profit health organization dedicated solely to finding the cause and cure for Adult Polyglucosan Body Disease (APBD). APBDRF strives to be the leading charitable funder and advocate of research worldwide.

Our mission is to improve the diagnosis and treatment of APBD, support individuals and families affected by the disease, increase awareness of APBD among health professionals and the public. Our ultimate goal, and the heart of our program is to find the underlying cause as well as a cure for APBD and its complications through the support of research, education, and patient services.

Letter from the President

Dear Friends,

As this is our first newsletter, I would like to take this opportunity to introduce myself and The Adult Polyglucosan Body Disease Research Foundation (APBDRF) and update you on our recent endeavors. I was diagnosed with Adult Polyglucosan Body Disease in 2002. Since my diagnosis, I was struck by the lack of awareness of APBD both among medical professionals and those that may be affected by the disease. In 2005, we organized the APBDRF to assist those who are affected by this disease and to support research and education.

In early 2006, the APBDRF launched its website. apbdrf.org is designed to reach those affected by APBD and doctors who may be treating patients with APBD. Through our website we have already been able to reach a number of people affected by APBD and have begun to build a community which is able to share their collective experiences. Based on our experience to date, we are reviewing our website and are planning a major redesign in the near future. I look forward to updating you on our progress.

In August 2006, the APBDRF sponsored the first conference of internationally recognized medical professionals to discuss the state of research on APBD. The conference laid the groundwork for the coordination of future research on APBD. We have included in this newsletter a story from *The Jewish Daily Forward* describing the conference.

Our next major initiative is organizing an exhibition at the 59th Annual American Academy of Neurology Expo in Boston, Monday, April 30 through Thursday, May 3, 2007. Our participation is our first major step in increasing awareness of APBD in the medical community. The APBDRF will be represented at the expo by many our members, supporters, volunteers, researchers and medical professionals who have made significant commitments to our cause.

It is our intention to continue to serve the interests of the APBD community and look forward to updating you on our progress.

Despite the progress we have made thus far, there is still enormous work that needs to be done. One day in the near future we hope to celebrate our victory over APBD. This is our first quarterly newsletter and we hope you find it informative.

Thank you for your continued support.

Best Regards,

Gregory Weiss
President

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Research Foundation**

APBD Foundation in the News!

**THE JEWISH DAILY
FORWARD**

Scientists Meet To Discuss Rare Disorder

In 1991, Alexander Lossos of the neurology department at the Hadassah University Hospital in Jerusalem discovered that a deficiency of the glycogen branching enzyme causes APBD. But it is still unclear how this occurs.

The best way to treat APBD is to "find a substance that increases the activity of the branching enzyme," said conference attendee Salvatore DiMauro, a professor of neurology at Columbia University and director of H. Houston Merritt Clinical Research Center at the Columbia Presbyterian Medical Center. Unfortunately that substance has yet to be discovered.

The research that Weiss's foundation promotes would not only encourage the development of prevention and treatment techniques for APBD, but also would potentially explain the causes of other disorders.

"Our knowledge about Alzheimer's disease, Parkinson's disease and a lot of other diseases almost exclusively is based on rare disorders — this very small subgroup of patients with rare single gene defects," said Raphael Schiffmann, head of clinical investigations in developmental and metabolic neurology at the National Institute of Health. "And really no research has been done on this disease at all."

In what was the first meeting of its kind, respected neurologists from around the globe held a conference

August 1 at which they committed themselves to increasing awareness of adult polyglucosan body disease, a rare genetic disorder occurring primarily among Ashkenazic Jews.

The nine-person scientific and medical advisory committee of the Adult Polyglucosan Body Disease Research Foundation made its decision after an all-day meeting at the New York University Medical Center in New York.

Committee members shared ideas about APBD, discussed the need to educate the medical and Jewish communities about the disorder and outlined further necessary research.

"We had all of the big minds in the field in the room," Gregory Weiss, who founded APBDRF and is an APBD patient himself, told the Forward. "We are trying to get involved patients and relatives and friends of people with this condition."

Spreading the word about the disease, which manifests itself in middle-aged patients, is difficult. With only 30 published cases, nobody knows exactly how many incidents of the chronic neurodegenerative disorder exist. Its symptoms — usually weakness and dementia — are easily confused with more common ailments, which often leads to misdiagnosis and mistreatment.

Mark I. Levenstein. Friday August 25, 2006

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We need your help! We need volunteers!!

Building up a foundation is not easy. Searching for the treatment and prevention of APBD is not easy. Increasing awareness of APBD is not easy. But we can do it with your help.

Ways You Can Help

Providing education and resources about testing as well as supporting research to help prevent APBD requires funding and collaboration. The APBDRF can only continue to make a difference in dealing with this deadly disease with your help. This help can come in the form of tax-deductible contributions, volunteering or as part of our local and national outreach efforts.

Help Spread the Word

Our efforts to build awareness can only be accomplished with your assistance.

- ✓ Please let us know about APBD families in your community.
- ✓ We need you to spread the word about APBD to individuals and families through your synagogues, local organizations and businesses, friends and healthcare providers.
- ✓ We need you to refer others to our Foundation and to other appropriate APBD sources.
- ✓ We will provide you with brochures and other literature for distribution and discuss setting up local chapters of the APBDRF in your area.

Conquering APBD requires collaboration, cooperation and commitment. Please help us to help you.

Your donation can start making the difference...

Your support makes it all possible!

To Make a Donation:

1. **Donate online on our webpage**
2. **Donate by Telephone**
Call the Foundation at the following phone number: 1-212-643-1221
3. **Donate by Mail**
Send your contribution to:
APBDRF
8 West 37th Street, Suite 901
New York, NY 10018
The Foundation is a 501(c)(3) non-profit, tax-exempt organization designated by the Internal Revenue Code. Our federal tax identification number is 20-3609790.

Join Our E-mail List

E-mail us at info@apbdrf.org so we may keep you posted about new developments and events. This list is for the APBDRF use only and will never be shared with anyone else.

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Medical Advisory Committee

The Foundation's Scientific/Medical Advisory Committee is comprised of respected researchers and practitioners knowledgeable about APBD and similar disorders.

From the outset in our quest for answers about APBD, our medical advisory board has provided professional advice and encouragement for all our efforts. They have always been thoughtful, caring and compassionate, even during discouraging times. Their guidance has been invaluable. The advisory panel is a vital part of our Foundation, and we are grateful for the commitment, time and expertise the members so graciously provide.

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