APBDRF
ADULT POLYGLUCOSAN BODY DISEASE RESEARCH FOUNDATION

As you know, Adult Polyglucosan Body Disease (APBD) is a highly variable disorder. Some patients have a neurogenic bladder and walking difficulties earlier than others. About half of our patients experience mild cognitive problems, but others see no change at all. Symptoms of fatigue are highly variable as well. On top of this, the known APBD community is very small, even by rare disease standards. All of this increases the difficulty of reaching our goal: To find effective treatments and a cure for APBD.

To help, the FDA and NIH urge rare disease communities such as ours to develop patient registries that follow the progression of their disorder over time. These so-called natural history studies are essential for several reasons:

- Understanding the causes of APBD;
- Developing hypotheses for basic and clinical research;
- Identifying patients who might have a superior/inferior response to various therapies;
- Designing appropriate clinical trials; and
- Contributing to drug development.

At this time, the APBD community has no such natural history data. We *did* establish a **C**olumbia University **APBD P**atient Registry (CAP) in 2014, and over 80 of you signed up for it. Although we intended enrollees to submit yearly updates, this has not happened. We've been told that it's daunting to provide the updates. We understand. But lacking updated information, the Columbia University registry (CAP) cannot produce data on the progression of the disease. Other rare disease communities have experienced a similar problem with their registries.

Fortunately, the FDA and the National Organization for Rare Diseases (NORD), an independent non-profit organization, have teamed up to overcome the daunting registry updating process. Twenty rare disease communities were chosen via an application process to be sponsored with a very patient-friendly natural history study registry. The APBD community was selected as one of the twenty.

The **F**dn & NORD-**APBD N**atural History Study (FAN) differs in significant ways from our existing **C**olumbia University **APBD** registry (CAP). Broadly speaking, one will be dynamic while the other remains relatively static.

**The APBD community needs information,**
**and the FAN study is a path that we hope with your help will address the need.**

1) Once 10 APBD patients enroll in FAN, a patient can get immediate feedback via charts and graphs about specific questions as they answer them which compare his/her situation to other patients who have also enrolled in the study.

2) Questions that the community has about the commonality of symptoms or events can be translated into questionnaires that are disseminated to enrolled APBD-FAN patients. In this way, important issues can be explored by the APBD community.

3) Eventually we expect that the FDA and NORD will make the registry available for queries from:
a. Physicians who are searching diagnostic criteria for their patients
b. Researchers who seek to compare one disease to another
c. Researchers who are seeking treatments for APBD and other allied diseases
d. Patient advocacy groups who are looking to join forces with other advocacy groups on activities, spending, technologies, research, etc. in order benefit from larger patient pools.

We need to be truthful. This initiative will take years to be fully realized. Indeed, five years of funding has been planned by the sponsors. But we believe that the front end work will begin to illuminate a clearer understanding of our orphan disease and potentially enhance the development of effective treatments and a cure.

**In the end, however, each of you will make decisions regarding your own participation. The accumulation of those decisions will dictate options that will or won't be available to the community as a whole.**

Since the FAN registry will take time to get up and running, the APBD Research Foundation must continue to maintain and develop the Columbia University APBD registry (CAP). When our scientific advisory board determines that the dual coverage is no longer necessary, we can take the appropriate action.

Yes, we are asking you to fill out both FAN and CAP questionnaires. We hope you will understand the wisdom of participating in both the interactive FAN natural history study and our established - although more static - CAP registry.