Essential Groundwork – Patient Registry, Genetic Testing, Natural History Studies, Biobanks

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The APBD Research Foundation is encouraging and facilitating many initiatives on many fronts in order to be prepared for clinical trials that are on the horizon.

One effort underway since 2014 is a patient registry called **CAP**, **C**olumbia University **A**PBD **P**atient Registry.

A natural history study, the objective of which is to gather prospective information on the manifestations of a condition over time, is just being launched by the National Organization for Rare Diseases (NORD). The study, called **FAN**, which stands for **F**DA and NORD-**A**PBD **N**atural History Study, will be discussed. Duke University is also planning to conduct a different natural history study. It is hoped that funding will come primarily from the FDA. Duke contemplates applying for a grant from the FDA in 2018.

Genetic testing for GBE1, the gene that contains mutations which lead to ABPD, was begun at Mount Sinai Medical in 2015. Screening through their prestigious Jewish genetic screening panel has already resulted in more refined information about the carrier rate for APBD mutations among people of Ashkenazi Jewish descent. In the near future, carrier screening for APBD will be offered by Counsyl, Inc., a genetic testing laboratory.

The Association for Glycogen Storage Disease (United States), or AGSDUS plans to write the Glycogen Storage Disease Type IV (GSD IV) health care guidelines in 2017.

Last but not least, a biobank for APBD patient skin fibroblasts has begun to collect samples from Israeli and American patients.

Presentations of all these initiatives will be provided.