***Summary of APBD, March 2016***

***What is adult polyglucosan body disease (APBD)?***

APBD is a condition that affects the nervous system. People with this condition have problems walking due to reduced sensation in their legs (peripheral neuropathy) and progressive muscle weakness and stiffness (spasticity). Also, they have progressive difficulty in controlling the flow of urine, a condition called neurogenic bladder. Finally, about half with APBD will experience a decline in intellectual function (dementia). Source: National Library of Medicine (NLM)

***How common is the disease?***

APBD is a rare condition. Although its exact prevalence is unknown, at least 50 cases have been described in the medical literature. The disease is concentrated in Ashkenazi Jews. But it is often misdiagnosed for MS, ALS, prostate disease (men) or other ailments---leading to much frustration, inappropriate medications, and even useless surgeries. Unknowingly, thousands may have APBD. Source: Adult Polyglucosan Body Disease Research Foundation (APBDRF)

***What genes are related to APBD?***

Mutations in the glycogen branching enzyme (*GBE)* gene cause adult polyglucosan body disease. The *GBE* gene provides instructions for making the glycogen branching enzyme. This enzyme is involved in the production of a complex sugar called glycogen, which is a major source of stored energy in the body. Most *GBE* gene mutations result in a shortage of the glycogen branching enzyme, leading to the overproduction of glycogen molecules.

These abnormal glycogen molecules, called polyglucosan bodies, accumulate within cells; especially nerve cells (neurons). Essentially with APBD, the polyglucosan bodies block the pathways for brain messaging, causing the functional problems. Source: NLM

***What research is going on?***

Recently, the Baylor Metabolic Institute completed a six-year clinical controlled study of triheptanoin oil for treating APBD. This oil is made from caster bean---containing seven carbon atoms whereas foods have an even number---that circumvents the GBE and provides 500 calories without producing more polyglucosan bodies. The results of the study have not yet been submitted to the FDA for approval. Baylor Research Institute

With the guidance of a scientific advisory board, the APBDRF has sponsored multifaceted research to reach a cure. This includes the following three approaches: (1) Increasing GBE activity (by stabilizing the mutated GBE with a peptide and other small molecules); (2) Lowering the accumulation of polyglucosan bodies in the cells (by down regulating glycogen synthase with compounds and antisense oligonucleotides); and (3) Correcting the non-functioning gene itself in compound heterozygotes (by using antisense oligonucleotides). We anticipate that the cure will be a combination of different approaches. Source: APBDRF