

Adult-onset Polyglucosan Body disease (APBD) is an adult-onset, neurological form of glycogen storage disease type IV. APBD is caused by recessive mutations in the glycogen branching enzyme (GBE1) gene. Deficiency of GBE1 results in the pathogenic accumulation of polyglucosan bodies in the nervous system. APBD symptoms typically develop in the fourth or fifth decade of life and include bladder dysfunction, gait disturbance, sensory and motor neuropathy, weakness, and fatigue. Cognitive decline is seen in approximately half of individuals with APBD. Progressive symptoms lead to wheelchair dependence and premature death. APBD is commonly misdiagnosed as multiple sclerosis, amyotrophic lateral sclerosis, and peripheral neuropathies. There are presently no treatments available for APBD.

The APBD Research Foundation is seeking research proposals that will advance the understanding of mechanisms of the disease or clinical phenotyping that will facilitate future treatment trials. Proposing studies with a clear therapeutic impact and a strong likelihood of future federal funding is a plus. A single grant of \$99,354 or two grants of \$49,677 will be awarded, depending upon the merits of the applications received.

The primary focus for this grant opportunity is the identification of a biomarker that could be used to demonstrate effectiveness of a therapeutic for the disorder.

Investigations related to the development of approaches that will prevent polyglucosan body accumulation or will facilitate its removal from the central and peripheral nervous systems will also be considered

Applicants are encouraged to collaborate with other scientists and clinicians and should include a statement on resource sharing in their proposal. Applicants are encouraged to use existing disease models (i.e., mouse models, cultured skin fibroblasts) and to contact the APBD Research Foundation (info@apbdrf.org) with any questions about these resources. All grant applications will be considered confidential. This grant is made possible by the APBD Research Foundation.

*Please submit a proposal for the total amount of \$99,354. The ODC may choose to fund two awards at \$49,677 each, at which point we will request a revised work plan and budget.