**Instructions:**

You are being asked to provide medical information on someone diagnosed with Adult Polyglucosan Body Disease (APBD), who has chosen to participate in the APBD Registry, a research project sponsored by the APBD Research Foundation and hosted by Columbia University. If you have any questions regarding the information requested on this form or the APBD Registry generally, you can contact the Principal Investigator for the study, Dr. Salvatore DiMauro, at 212-305-1662 or sd12@columbia.edu.

Please complete the following pages as thoroughly as possible, using results from the most recent physical/neurological exam performed on the patient.

If you complete this form electronically, be sure to save your entries, and return the resulting file to your patient. The patient will then upload the file to the secure APBD web site.

If you complete this form on paper, return the completed pages to your patient. The patient will then enter the data you have supplied into the secure APBD web site.

Thank you for your time, and for your contribution to the understanding of Adult Polyglucosan Body Disease.

**Clinical Manifestations**

|  |  |  |  |
| --- | --- | --- | --- |
| Manifestation | Present | Age at Onset | Course |
| Bladder dysfunction | No Yes |  | Stable Progressive Medication use catheter use |
| Fecal incontinence | No Yes |  | Stable Progressive |
| Gait alteration | No Yes |  | Stable Progressive cane/walker wheelchair |
| Feet numbness | No Yes |  | Stable Progressive |
| Hand numbness | No Yes |  | Stable Progressive |
| Tremor | No Yes |  | Stable Progressive |
| Extremity pain | No Yes |  | Stable Progressive Pain Medication use |
| Memory deficit | No Yes |  | Stable Progressive Affects daily life yes no |
| Dementia | No Yes |  | Stable Progressive |
| Psychiatric disorder | No Yes |  | Stable Progressive Medication use |
| Behavioral problems | No Yes |  | Stable Progressive |
| Parkinsonism | No Yes |  | Stable Progressive Medication use |
| Autonomic symptoms (orthostatic hypotension, etc.) | No Yes |  | Stable Progressive |
| Swallowing problems | No Yes |  | Stable Progressive |
| Speech problems | No Yes |  | Stable Progressive |
| Other, describe: | No Yes |  | Stable Progressive |

**Physical/Neurological Exam**

|  |  |
| --- | --- |
| Age at exam |  |
| Eye Movements | |
| Pursuit | Normal  Saccadic |
| Voluntary saccades | Normal  Slow |
| Muscle Tone | |
| Upper extremities muscle tone | Normal  Spastic  Flaccid |
| Lower extremities muscle tone | Normal  Spastic  Flaccid |
| Strength | |
| Upper extremities weakness | Proximal No Yes  Distal No Yes |
| Lower extremities weakness | Proximal No Yes  Distal No Yes |
| Reflexes | |
| Snout | Normal Increased Decreased Absent |
| Bicipital | Normal Increased Decreased Absent |
| Tricipital | Normal Increased Decreased Absent |
| Patellar | Normal Increased Decreased Absent |
| Ankle | Normal Increased Decreased Absent |
| Plantar | Flexor Extensor (Babinski sign) Mute |
| Sensitivity | |
| Vibration | Decreased Upper extremities No Yes  Decreased Lower extremities No Yes |
| Position | Decreased Upper extremities No Yes  Decreased Lower extremities No Yes |
| Touch | Decreased Upper extremities No Yes  Decreased Lower extremities No Yes |
| Temperature (cold) | Decreased Upper extremities No Yes  Decreased Lower extremities No Yes |
| Gait/Coordination | |
| Ataxic gait | No  Yes |
| Tandem gait | Normal  Abnormal  Unable |
| Romberg test | Normal  Abnormal |
| Dysdiadochokinesia | No  Yes |
| Dysmetria | No  Yes |
| Cognition | |
| Memory deficit | No  Yes |
| Attention deficit | No  Yes |
| Mini-Mental State Exam score |  |

**Other Clinical Tests**

|  |  |
| --- | --- |
| CK max | Not done  Done  If Done Age performed: \_\_\_\_\_\_\_\_\_\_\_\_  Value (IU/l): \_\_\_\_\_\_\_\_\_\_\_\_ |
| Functional bladder study | Not done  Done  If Done Age performed: \_\_\_\_\_\_\_\_\_\_\_\_  Result: Normal  Abnormal – Neurogenic  Abnormal – Central |
| Brain MRI | Not done  Done  If Done Age performed: \_\_\_\_\_\_\_\_\_\_\_\_  White matter lesions Periventricular No Yes  Subcortical No Yes  Internal capsule No Yes  External capsule No Yes  Cerebellum No Yes  Midbrain No Yes  Pons No Yes  Medulla No Yes  Symmetrical No Yes  Asymmetrical No Yes  Atrophy Cortex No Yes  Ventricles No Yes  Cerebellum No Yes  Corpus callosum No Yes  Pons No Yes  Cervical spine No Yes  Dorsal spine No Yes |
| Nerve biopsy | Not done  Done  If Done Age performed: \_\_\_\_\_\_\_\_\_\_\_\_  Name of laboratory: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_  Polyglucosan bodies No Yes  Axonal changes No Yes  Demyelinated changes No Yes |
| Muscle biopsy | Not done  Done  If Done Age performed: \_\_\_\_\_\_\_\_\_\_\_\_  Name of laboratory: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_  Polyglucosan bodies No Yes  Myopathic changes No Yes  Neurogenic changes No Yes |

**Other Clinical Tests, continued**

|  |  |
| --- | --- |
| EMG | Not done  Done  If Done Age performed: \_\_\_\_\_\_\_\_\_\_\_\_  Spontaneous activity No Yes  Myopathic pattern No Yes  Neurogenic pattern No Yes |
| Nerve conduction study | Not done  Done  If Done Age performed: \_\_\_\_\_\_\_\_\_\_\_\_  Sensory Upper Extremities Normal Axonal Demyelinating  Lower Extremities Normal Axonal Demyelinating  Motor Upper Extremities Normal Axonal Demyelinating  Lower Extremities Normal Axonal Demyelinating |
| GBE enzymatic activity | Not done  Done  If Done Name of laboratory: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_  Activity reduction, lymphocytes (%): \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_  Activity reduction, fibroblasts (%): \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ |
| Molecular study of *GBE1* gene | Not done  Done  If Done Name of laboratory: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_  Allele 1: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_  Allele 2: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_  Homozygous No  Yes  Restriction Enzyme Analysis Done  Not done |

**Material Available for Study**

|  |  |
| --- | --- |
| DNA | No Yes Unknown |
| RNA | No Yes Unknown |
| Cell – specify: | No Yes Unknown |

**nknowny:\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_Physician Information (optional)**

|  |  |
| --- | --- |
| Name |  |
| Phone number |  |
| Mailing Address |  |
| Email address |  |
| Institutional Affiliation |  |