**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 YesDistal No Yes |
| Lower extremities weakness | Proximal No YesDistal 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 YesDecreased Lower extremities No Yes |
| Position | Decreased Upper extremities No YesDecreased Lower extremities No Yes |
| Touch | Decreased Upper extremities No YesDecreased Lower extremities No Yes |
| Temperature (cold) | Decreased Upper extremities No YesDecreased 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 DoneIf Done Age performed: \_\_\_\_\_\_\_\_\_\_\_\_ Value (IU/l): \_\_\_\_\_\_\_\_\_\_\_\_ |
| Functional bladder study |  Not done DoneIf Done Age performed: \_\_\_\_\_\_\_\_\_\_\_\_ Result: Normal Abnormal – Neurogenic Abnormal – Central |
| Brain MRI |  Not done DoneIf 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 DoneIf Done Age performed: \_\_\_\_\_\_\_\_\_\_\_\_ Name of laboratory: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ Polyglucosan bodies No Yes Axonal changes No Yes Demyelinated changes No Yes |
| Muscle biopsy |  Not done DoneIf 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 DoneIf Done Age performed: \_\_\_\_\_\_\_\_\_\_\_\_ Spontaneous activity No Yes Myopathic pattern No Yes Neurogenic pattern No Yes |
| Nerve conduction study |  Not done DoneIf 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 DoneIf Done Name of laboratory: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ Activity reduction, lymphocytes (%): \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ Activity reduction, fibroblasts (%): \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ |
| Molecular study of *GBE1* gene |  Not done DoneIf 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 |  |