#### 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	Relapsing-remitting	Reversible
Fecal incontinence	No Yes		Stable	Progressive	Relapsing-remitting	Reversible
Gait alteration	No Yes		Stable	Progressive	Relapsing-remitting	Reversible
Feet numbness	No Yes		Stable	Progressive	Relapsing-remitting	Reversible
Hand numbness	No Yes		Stable	Progressive	Relapsing-remitting	Reversible
Tremor	No Yes		Stable	Progressive	Relapsing-remitting	Reversible
Memory deficit	No Yes		Stable	Progressive	Relapsing-remitting	Reversible
Dementia	No Yes		Stable	Progressive	Relapsing-remitting	Reversible
Psychiatric disorder	No Yes		Stable	Progressive	Relapsing-remitting	Reversible
Behavioral problems	No Yes		Stable	Progressive	Relapsing-remitting	Reversible
Parkinsonism	No Yes		Stable	Progressive	Relapsing-remitting	Reversible
Autonomic symptoms (orthostatic hypotension, etc.)	No Yes		Stable	Progressive	Relapsing-remitting	Reversible
Swallowing problems	No Yes		Stable	Progressive	Relapsing-remitting	Reversible
Speech problems	No Yes		Stable	Progressive	Relapsing-remitting	Reversible
Other, describe:	No Yes		Stable	Progressive	Relapsing-remitting	Reversible

Physical/Neurological Exam

Age at exam		
	Eye Movements	
Pursuit	Normal	
	Saccadic	
Voluntary saccades	Normal	
	Slow	
	Muscle Tone	
LL muscle tone	Normal	
	Spastic	
	Flaccid	
UL muscle tone	Normal	
	Spastic	
	Flaccid	
	Strength Proximal No Yes	
LL weakness	Distal No Yes	
UL 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	Normal Increased Decreased Absent	
	Sensitivity	
Vibration	Decreased LL No Yes	
	Decreased UL No Yes	
Position	Decreased LL No Yes	
	Decreased UL No Yes	
Touch	Decreased LL No Yes	
	Decreased UL No Yes	
Pain	Decreased LL No Yes	
	Decreased UL No Yes	
	Gait/Coordination	
Ataxia	No	
Tenden	Yes	
Tandem	Normal	
	Abnormal	
Domhora	Unable	
Romberg	No	
Dysdiadochokinesia	Yes No	
Dysulduotilokiilesia	Yes	
Dysmetria	No	
Jonotha	Yes	
	Cognition	
Memory deficit	No	
	Yes	
Attention deficit	No	
	Yes	
Mini-Mental State Exam score		

### Other Clinical Tests

CK max	Not do	ne						
	Done							
	If Done	Age performed: Value (IU/I):						
Functional bladder study	Not do Done							
	If Done	Age performed:	Normal	 nal – Neu	rogenic			
				nal – Cent				
Brain MRI	Not do Done	ne						
	If Done	Age performed:						
		White matter lesions	5	Periventi Subcorti Internal External Cerebell Midbrain Pons Medulla Symmeti Asymmeti	cal capsule capsule um	No No No No No No No No	Yes Yes Yes Yes Yes Yes Yes Yes Yes	
		Atrophy		Cortex Ventricle Cerebell Corpus of Pons Cervical Dorsal sp	um callosum spine	No No No No No No	Yes Yes Yes Yes Yes Yes Yes	
Nerve biopsy	Not do	ne		Dersars		110	100	
	Done If Done	Age performed: Name of laboratory: Polyglucosan bodies		No	Yes			
		Axonal changes Demyelinated changes	nes	No No	Yes Yes			
Muscle biopsy	Not do Done If Done		900		103			
		Name of laboratory:						
		Polyglucosan bodies Myopathic changes Neurogenic changes		No No No	Yes Yes Yes			

Other Clinical Tests, continued

EN 0							
EMG	Not done						
	Done	Done					
	If Done	Age performed:					
		Spontaneous activ		No	Yes		
		Myopathic pattern		No	Yes		
	<b>N 1 1</b>	Neurogenic patter	'n	No	Yes		
Nerve conduction study	Not do	one					
	Done						
	If Done	Age performed:					
		0		NI	A I	Description	
		Sensory	LL	Normal	Axonal	Demyelinating	
			UL	Normal	Axonal	Demyelinating	
		Matan		N	A	Dennellingting	
		Motor	LL	Normal	Axonal	Demyelinating	
	Not do		UL	Normal	Axonal	Demyelinating	
GBE enzymatic activity		ne					
	Done						
	If Dono	Name of laborator	n./:				
	II DONE		у				
		Activity reduction, lymphocytes (%):					
		Activity reduction,					
Molecular study	Not do		1010010303 (	/0]			
	Done	no					
	Done						
	If Done Name of laboratory:						
	II Done		y				
		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

### Physician Information (optional)

Name	
Phone number	
Mailing Address	
Email address	
Institutional Affiliation	