## Clinical Questionnaire for Whole Exome/Genome Sequencing

Patient name:					Da	te of birth:		
Ordering provider signature:			Provider NPI:		Phone:		Date:	
Ordering Provider: check any boxes	s that apply t	to the proba	and and his/her family (current or pre	evious history	of), includi	ng the proband's 1st and 2nd degree ing processes will not begin until thi	relatives. Aco	curate
			no the affected individual(s) is/are in I-890-0003; otherwise, include thi			linical information section below. Imple. For questions, call 866-248	-1265.	
Prenatal History	Proband	-	Dermatologic Findings	Proband		Metabolic	Proband	
IUGR	0	0	Alopecia	0	0	Elevated alanine	0	0
Prematurity/postmaturity	0	0	Angiokeratoma	0	0	Elevated pyruvate	0	0
Poly/oligohydramnios	0	0	Café au lait spots	0	0	CPK abnormalities	0	0
Cystic hygroma/increased NT	0	0	Hypo/hyperpigmentation	0	0	Ketosis	0	0
Other:	0	0	Ichthyosis	0	0	Lactic acidosis	0	0
General History	Proband	Family*	Rash/dermatitis/eczema	0	0	Organic aciduria	0	0
Failure to thrive	O		Nail dysplasia	0	0	Low plasma carnitine	0	0
Growth retardation/short stature	_	0	Abnormal connective tissue	0	0	Other:	_ 0	0
Overgrowth	0	0	Other:	_ 0	0	Neuromuscular	Proband	Family*
Fine motor delay	0	0	<b>Brain Malformations</b>	Proband	Family*	Ataxia	0	0
Gross motor delay	0	0	Agenesis-Corpus Callosum	0	0	Chorea	0	0
Speech delay	0	0	Holoprosencephaly	0	0	Dystonia	0	0
Intellectual disability/MR	0	0	Lissencephaly	0	0	Exercise intolerance/fatigue	0	0
		0	Cortical dysplasia	0	0	Headaches/migraines	0	0
IQ: Learning disability	0	0	Heterotopia	0	0	Hypotonia	0	0
9	0	0	Hydrocephalus	0	0	Hypertonia	Ō	0
Autism/Autism spectrum	0	0	Brain atrophy	0	0	Muscle weakness	0	0
Psychiatric disorder	0	0	Abnormalities of basal ganglia	Ö	Ö	Neuropathy	Ö	Ö
Behavioral disorder		_	Other:		Ö	Seizures/epilepsy	Ö	Ö
Cardiovascular Findings	Proband	-				Stroke/stroke-like episodes	Ō	0
ASD	0	0	Endocrine Findings	Proband	•	Spasticity	Ö	Ö
VSD	0	0	Adrenal abnormality	0	0	Torticollis	Ö	Ö
Cardiomyopathy	0	0	Diabetes mellitus	0	0	Other:	Ö	0
Arrhythmia/conduction defect	0	0	Hypothyroidism	0	0		-	_
Tetralogy of Fallot	0	0	Hypoparathyroidism	0	0	Skeletal/Limb Findings	Proband	-
Coarctation of the aorta	Ö	Ö	Pheochromocytoma	0	0	Contractures	0	0
Hypoplastic left heart	Ō	Ō	Sex hormone abnormality	0	0	Clubfoot	0	0
Aortic root dilation	Ö	Ö	<b>Gastrointestinal Findings</b>	Proband	Family*	Polydactyly	0	0
Other:	_	Ö	Abnormal liver function	0	0	Syndactyly	0	0
	-	-	Achalasia	0	0	Scoliosis	0	0
Craniofacial/Ophthalmologic/		- " +	Bile duct proliferation	0	0	Vertebral anomaly	0	0
Auditory	Proband	-	Biliary atresia	0	0	Other:	_ 0	0
Blindness	0	0	Cirrhosis	0	0			
Cataracts	0	0	Chronic constipation	0	0	Consanguinity ○ Yes ○ No		
Coloboma	0	0	Chronic diarrhea	0	Ō			
Glaucoma	0	0	Congenital anomaly:		Ö	Other Testing/Findings		
Retinal disorder	0	0	Hepatomegaly	_ 0	Ö	(Please summarize results or atta	ch them)	
Ptosis	0	0	Hirschsprung disease	Ö	Ö	Chromosomes		_
Optic atrophy	0	0	Jaundice	Ö	Ö			
Retinitis pigmentosa	0	0	Liver failure	0	0	FISH		
Cleft lip/palate	0	0	Other:	_	0	Microarray		
Craniosynostosis	0	0		_	-	Fragile X		
Microcephaly	0	0	Genitourinary Findings	Proband		Muscle biopsy		
Macrocephaly	0	0	Ambiguous genitalia	0	0			
Hearing loss	0	0	Abnormal renal function	0	0	Other (clinical or research)		
External ear malformation	0	0	Hypospadias	0	0			
Ototoxicity	0	0	Hydronephrosis	0	0			
Dysmorphic facial features	0	0	Kidney malformation	0	0	<b>Ethnicity</b> (check all that apply)	Maternal	
Other:	0	0	Obstructive renal disease	0	0	Caucasian/White	0	0
Cancer/Malignancy		Family*	Renal agenesis/hypoplasia	0	0	African American/Black	0	0
			Sex reversal	0	0	Hispanic	0	0
Tumor type:		0	Undescended testicles	0	0	Asian	0	0
Location:		0	Congenital genital anomalies:	0	0	Ashkenazi Jewish	0	0
Age of onset:			Hematologic/Immunologic	Proband	Family*	Sephardic Jewish	0	0
Affected family members:			• .	O		Sephardic Jewish Other:	0	0
			Anemia					
			Allergies:	_ 0	0	Additional relevant clinical info	rmation:	
			Autoimmune disorder					
			Enlarged lymph nodes	0	0			
(O) labco	1rn		Immunodeficiency	0	0			
	/I P		Neutropenia/pancytopenia	0	0			
	=		Thymic hypoplasia	0	0			
			Other:	0	0	I		,