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Patient's Name (Last, First, Middle):	DOB (MM/DD/YYYY):	Gender: ___ Female ___ Male
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**Your Clinical Features** (Please check all that apply)\*

Musculoskeletal	Hematologic	Neurologic	Psychiatric
<input type="checkbox"/> Osteoporosis	<input type="checkbox"/> Anemia	<input type="checkbox"/> Brain disease	<input type="checkbox"/> Aggression
<input type="checkbox"/> Spinal disease	<input type="checkbox"/> Blood coagulation disorder	<input type="checkbox"/> Intracranial hemorrhage	<input type="checkbox"/> Feeding disorder
<input type="checkbox"/> Pathological fracture(s)	<input type="checkbox"/> Petechiae	<input type="checkbox"/> Seizures	<input type="checkbox"/> Anxiety disorder
<input type="checkbox"/> Foot deformity	<input type="checkbox"/> Blood platelet disorder	<input type="checkbox"/> Headache	<input type="checkbox"/> Dementia
<input type="checkbox"/> Joint disease	<input type="checkbox"/> Blood protein disorder	<input type="checkbox"/> Migraine	<input type="checkbox"/> Autistic disorder
<input type="checkbox"/> Muscular disease	<input type="checkbox"/> Bone marrow disease	<input type="checkbox"/> Leukoencephalopathy	<input type="checkbox"/> Depressive disorder
<input type="checkbox"/> Microcephaly	<input type="checkbox"/> Leukocyte disorder	<input type="checkbox"/> Cerebellar atrophy	<input type="checkbox"/> Psychotic disorder
<input type="checkbox"/> Rhabdomyolysis	<input type="checkbox"/> Lymphatic disease	<input type="checkbox"/> Corpus callosum hypoplasia	<input type="checkbox"/> Schizophrenia
Dermatologic	Metabolic	<input type="checkbox"/> Choreatic disorder	Oncologic
<input type="checkbox"/> Dermatitis	<input type="checkbox"/> Acidosis	<input type="checkbox"/> Dystonic disorder	<input type="checkbox"/> Squamous-cell neoplasm
<input type="checkbox"/> Hair-disease	<input type="checkbox"/> Reye-like symptoms	<input type="checkbox"/> Parkinsonian disorder	<input type="checkbox"/> Neuroendocrine tumor
<input type="checkbox"/> Pigmentation disorder	<input type="checkbox"/> Dyslipidemias	<input type="checkbox"/> Tremor	<input type="checkbox"/> Paraganglioma
<input type="checkbox"/> Hyperhidrosis	<input type="checkbox"/> Diabetes mellitus	<input type="checkbox"/> Spinal cord disease	<input type="checkbox"/> Leukemia/lymphoma
<input type="checkbox"/> Paleness	<input type="checkbox"/> Hyperglycemia	<input type="checkbox"/> Neurogenic bladder	<input type="checkbox"/> Breast neoplasm
Endocrinologic	<input type="checkbox"/> Hyperinsulinism	<input type="checkbox"/> Ataxia	<input type="checkbox"/> Colorectal neoplasm
<input type="checkbox"/> Adrenal gland disease	<input type="checkbox"/> Hypoglycemia	<input type="checkbox"/> Speech disorder	<input type="checkbox"/> Hepatocellular carcinoma
<input type="checkbox"/> Adrenal insufficiency	<input type="checkbox"/> Hyperammonemia	<input type="checkbox"/> Consciousness disorder	<input type="checkbox"/> Leiomyoma
<input type="checkbox"/> Adrenocortical hyperfunction	<input type="checkbox"/> Hyperbilirubinemia	<input type="checkbox"/> Memory disorder	<input type="checkbox"/> Prostatic neoplasm
<input type="checkbox"/> Gonadal disorder	<input type="checkbox"/> Hemochromatosis	<input type="checkbox"/> Mental retardation	<input type="checkbox"/> Renal cell carcinoma
<input type="checkbox"/> Sex-differentiation disorder	<input type="checkbox"/> Abnormal amino acid levels	<input type="checkbox"/> Hallucinations	Ophthalmologic
<input type="checkbox"/> Parathyroid disease	<input type="checkbox"/> Water-electrolyte imbalance	<input type="checkbox"/> Psychomotor agitation	<input type="checkbox"/> Blepharoptosis
<input type="checkbox"/> Pituitary disease	<input type="checkbox"/> Obesity	<input type="checkbox"/> Irritability	<input type="checkbox"/> Cataract
<input type="checkbox"/> Thyroid disease	<input type="checkbox"/> Abnormal fatty acids	<input type="checkbox"/> Lethargy	<input type="checkbox"/> Pathologic nystagmus
Gastrointestinal	<input type="checkbox"/> Abnormal organic acids	<input type="checkbox"/> Neuromuscular manifestation	<input type="checkbox"/> Ophthalmoplegia
<input type="checkbox"/> Cholestasis	<input type="checkbox"/> Dicarboxylic aciduria	<input type="checkbox"/> Paralysis-Paresis	<input type="checkbox"/> Optic nerve disease
<input type="checkbox"/> Deglutition disorder	Genitourinary	<input type="checkbox"/> Abnormal reflexes	<input type="checkbox"/> Retinal disease
<input type="checkbox"/> Gastroenteritis	<input type="checkbox"/> Male infertility	<input type="checkbox"/> Hearing disorder	<input type="checkbox"/> Color vision defect
<input type="checkbox"/> Intestinal obstruction	<input type="checkbox"/> Hypospadias	<input type="checkbox"/> Voice disorder	Immunologic
<input type="checkbox"/> Gastrointestinal hemorrhage	<input type="checkbox"/> Cystic kidney disease	<input type="checkbox"/> Stroke-like episodes	<input type="checkbox"/> Autoimmune diseases
<input type="checkbox"/> Liver disease	<input type="checkbox"/> Nephrocalcinosis	<input type="checkbox"/> Developmental delay	<input type="checkbox"/> Immune deficiency
<input type="checkbox"/> Fatty liver	<input type="checkbox"/> Renal insufficiency	<input type="checkbox"/> Polyneuropathy	<input type="checkbox"/> Infections
<input type="checkbox"/> Pancreatitis	<input type="checkbox"/> Urination disorder	<input type="checkbox"/> Sleep disorder	Miscellaneous
<input type="checkbox"/> Abdominal pain	<input type="checkbox"/> Menstruation disorder	Cardiovascular	<input type="checkbox"/> Fever
<input type="checkbox"/> Feeding difficulties	<input type="checkbox"/> Pregnancy complications	<input type="checkbox"/> Arrhythmia	<input type="checkbox"/> Hypothermia
<input type="checkbox"/> Diarrhea	Respiratory	<input type="checkbox"/> Cardiomyopathy	<input type="checkbox"/> Exercise intolerance
<input type="checkbox"/> Vomiting	<input type="checkbox"/> Hyperventilation	<input type="checkbox"/> Cardiorespiratory-arrest	<input type="checkbox"/> Failure to thrive
	<input type="checkbox"/> Respiratory insufficiency	<input type="checkbox"/> Hypertension	<input type="checkbox"/> Growth deficiency
	<input type="checkbox"/> Asthma	<input type="checkbox"/> Hypotension	<input type="checkbox"/> Odor
	<input type="checkbox"/> Pneumonia	<input type="checkbox"/> Myocardial-ischemia	<input type="checkbox"/> RCC deficiency
	<input type="checkbox"/> Pulmonary edema		<input type="checkbox"/> Vitamin-responsive

\* Adapted from Scharfe C et al (2009) PLoS ONE 5(4):1-12

**Additional Clinical Features** (Please check all that apply)

- |   |  |
|---|--|
| <input type="checkbox"/> ADD (attention deficit disorder)   | <input type="checkbox"/> MERRF (Myoclonic Epilepsy associated with Ragged Red Fibers)                          |
| <input type="checkbox"/> ADHD (attention deficit / hyperactivity disorder)                                  | <input type="checkbox"/> OCD (obsessive-compulsive disorder)   |
| <input type="checkbox"/> Bipolar disorder   | <input type="checkbox"/> PANDAS (Pediatric Autoimmune Neuropsychiatric Disorder Associated with Streptococcus) |
| <input type="checkbox"/> Epilepsy   | <input type="checkbox"/> PEO (progressive external ophthalmoplegia)  |
| <input type="checkbox"/> Intellectual disorder  | <input type="checkbox"/> Tourette syndrome   |
| <input type="checkbox"/> LHON (Leber's Hereditary Optic Neuropathy)   | <input type="checkbox"/> Tics  |
| <input type="checkbox"/> MELAS (Mitochondrial Encephalomyopathy, Lactic acidosis, and Stroke-like episodes) |  |