Please shade the boc next to the condition

Pre/Perinatal History		
Cystic Hygroma	Growth delay	Nonimmune hydrops fetalis
Diaphragmatic Hernia	Increased nuchal translucency	Oligohydramnios
Encephalocele	Intrauterine Growth Retardation	Polyhydramnios
Prematurity GA:_____	Prolonged neonatal jaundice	

Structural Brain Abnormalities		
Abnormal myelination	Aplasia/hypoplasia of cerebellum	Leukodystrophy
Abnormality of basal ganglia	Arnold Chiari malformation	Lissencephaly
Abnormality of brainstem	Cerebellar atrophy	Pachygyria
Abnormality of periventricular white matter	Heterotopia (Periventricular nodular heterotopia)	Polymicrogyria
Abnormality of the corpus callosum	Holoprosencephaly	Ventriculomegaly
Aplasia/hypoplasia of cerebellar vermis	Hydrocephalus	

Developmental/Behavioral Findings		
Absent speech	Cognitive impairment	Gait disturbance
Aggressive behavior	Delayed speech & language development	Global developmental delay
Anxiety	Developmental regression	Hyperactivity
Autistic Behavior	Dysarthria	Incoordination

Neurological Findings		
Abnormality of nervous system	Dyskinesia	Migraines
Ataxia	Dysphasia	Myoclonus
Cerebral palsy	Paresthesia	Parkinsonism
Chorea	Dystonia	Peripheral neuropathy
Cortical Visual Impairment	Encephalopathy	Seizures
Dementia	Headaches	Sensory neuropathy
Dysarthria	Hemiplegia	Spasticity

	Hypoesthesia		Infantile Spasms		Syncope
	Tremors		Vertigo		
Craniofacial/Dysmorphism					
	Abnormal facial shape (Dysmorphic features)		Coarse facial features		Short neck
	Specify:_____		Craniosynostosis		Synophrys
	Brachycephaly		Macrocephaly		Widely patent fontanelles and sutures
	Cleft lip and/or palate		Microcephaly		
Eye Defects/ Vision					
	Abnormality of Vision		External ophthalmoplegia		Ptosis
	Anophthalmia		Microphthalmia		Retinal detachment
	Cataracts		Myopia		Retinitis pigmentosa
	Coloboma		Nystagmus		Strabismus
	Corneal opacity		Optic atrophy		
	Ectopia lentis		Optic neuropathy		
Hearing Impairment					
	Abnormal Newborn Screen:_____		Conductive hearing impairment		Sensorineural hearing impairment
Endocrine Findings					
	Delayed puberty		Diabetes Insipidus		Diabetes Mellitus
Hyperthyroidism					
	Hypophosphatemia		Maturity-onset diabetes of the young		Rickets
	Hypothyroidism				

Respiratory Findings		
Asthma	Hypoventilation	Respiratory insufficiency
Bronchiectasis	Pneumothorax	Dyspnea
Hyperventilation	Pulmonary fibrosis	

Hematologic or Immunologic Findings		
Allergic rhinitis	Autoimmunity	Thrombocytopenia
Anemia	Neutropenia	Hypoalbuminemia
Increased circulating antibody level	Pancytopenia	
Immunodeficiency	Recurrent infections	

Skin/Hair Findings		
Abnormal blistering of the skin	Anhidrosis	Coarse hair
Abnormality of nail	Angioedema	Cutis Laxa
Nail Bed Hemorrhage	Swollen Lip	Eczema
Alopecia	Café-Au-Lait Macules	Sparse hair
Hyperextensible skin	Hemangiomas	Telangiectasia
Hyperpigmentation of the skin	Ichthyosis	Vascular skin abnormality
Hypohidrosis	Skin rash	
Hypopigmentation of the skin	Velvety skin	

Cardiac Findings		
Abnormal heart morphology	Dilated cardiomyopathy	Prolonged QTc interval
Amyloidosis	Heterotaxy	Sudden death
Aortic root dilation	Hypertension	Tetralogy of Fallot
Arrhythmia	Hypertrophic cardiomyopathy	Ventricular septal defect
Atrial septal defect	Mitral valve prolapse	Ventricular tachycardia
Bicuspid aortic valve	Noncompaction cardiomyopathy	Hypotension
Bradycardia	Patent ductus arteriosis	Angina pectoris
Coarctation of aorta	Patent foramen ovale	

Gastrointestinal Findings					
	Constipation		Gastroesophageal reflux		Pancreatitis
	Dysphagia		Hepatomegaly		Pyloric stenosis
	Diarrhea		Inflammatory bowel disease		Splenomegaly
	Duodenal stenosis/atresia		Intrahepatic biliary atresia		Tracheoesophageal fistula
	Exocrine pancreatic insufficiency		Laryngomalacia		Vomiting
	Feeding difficulties		Nausea		Hyperlipidemia
	Abdominal Pain				

Genitourinary Findings					
	Ambiguous genitalia		Nephrotic Syndrome		Anuria
	Cryptorchidism		Horseshoe kidney		Renotubular dysgenesis
	Cystic renal dysplasia		Hydronephrosis		Nephrolithiasis
	Proteinuria		Hypospadias		Polycystic kidney disease
	Hematuria		Inguinal hernia		Renal agenesis
	Umbilical hernia				

Musculoskeletal Findings					
	Abnormal connective tissue		Hypertonia		Pectus excavatum
	Arachnodactyly		Hypotonia		Polydactyly
	Arthralgia		Joint hypermobility		Recurrent fractures
	Arthrogryposis		Muscle weakness		Rhabdomyolysis
	Bruising susceptibility		Myalgia		Abnormal uvula morphology
	Clinodactyly		Myopathic facies		Scoliosis
	Decreased muscle mass		Myopathy		Short stature
	Ectrodactyly		Osteoarthritis		Skeletal dysplasia
	Exercise intolerance		Osteopenia		Syndactyly
	Fatigue		Pain		Tall stature
	Hemihypertrophy		Pectus carinatum		

Metabolic Findings (Attached relevant lab reports/values)

	Abnormal activity of mitochondrial respiratory chain		Hyperammonemia		Hypoglycemia
	Abnormal Newborn Screen:		Abnormal Circulating Lipid Concentration		Increased serum pyruvate
	Abnormality of mitochondrial metabolism		Decreased HDL Cholesterol concentration		Lactic acidosis
	Elevated CPK		Hyperglycemia		Plasma AA:_____
	Elevated hepatic transaminase		Hypoammonemia		Urine OA:_____
Vascular System					
	Aneurysm		Arterial tortuosity		Lymphedema
	Arterial calcification		Arteriovenous malformation		Pulmonary hypertension
	Arterial dissection		Epistaxis		Stroke
	Pedal edema				
Other Testing/Imaging (Please provide copy or report if possible)					
	Echo:_____		Gene Sequencing*:_____		Microarray:_____
	EEG: _____		Results:		MRI:_____
	EMG:_____		Performed at:		Muscle Biopsy: _____
	Gene Panel:_____		If you would like us to comment on the presence/		Ultrasound:_____
	Results:_____		absence of previously identified variants, including		X-rays:_____
	Performed at:_____		parental status (if included), provide complete variant		
	Additional Clinical Findings:_____		information or a copy of the original report.		