

# FABRIC GENOMICS CLINICAL REQUISITION FORM

## Patient Information

Patient First Name:

Patient Last Name:

Patient Sex:

Patient DOB (mm/dd/yyyy):

Patient GA (in weeks):

Diagnosis:

## Case Information

Accession ID:

Project ID:

Proband Genome ID:

## Lab Tests

Blood Biochemistry Findings:

Urine Biochemistry Findings:

Cancer Screen Findings:

**PERINATAL HISTORY**

Abnormal placenta morphology  
Abnormality of the umbilical cord  
Breech presentation  
Congenital diaphragmatic hernia  
Failure to thrive  
Growth delay  
Hydrops fetalis  
Increased nuchal translucency  
Infantile spasms  
Intrauterine growth retardation  
Low APGAR score  
Nonimmune hydrops fetalis  
Oligohydramnios  
Omphalocele  
Polyhydramnios  
Postnatal growth retardation  
Premature birth  
Prolonged neonatal jaundice

**DEVELOPMENT AND BEHAVIOR**

Absent speech  
Aggressive behavior  
Anxiety  
Apathy  
Attention deficit hyperactivity disorder  
Autistic behavior  
Cognitive impairment  
Delayed speech and language development  
Depression  
Developmental regression  
Dyscalculia  
Dyslexia  
Global developmental delay  
Hyperactivity  
Impaired visuospatial constructive cognition  
Intellectual disability  
Lethargy  
Memory impairment  
Obsessive-compulsive behavior  
Polydipsia  
Restrictive behavior  
Sleep disturbance  
Speech articulation difficulties

**MOTOR FUNCTION**

Akinesia  
Areflexia  
Ataxia  
Athetosis  
Cerebral palsy  
Chorea  
Dyskinesia  
Dysmetria  
Dystonia  
Gait disturbance  
Hemiplegia  
Hyperreflexia  
Hypokinesia  
Hyporeflexia  
Inability to walk  
Incoordination  
Involuntary movements  
Loss of ambulation  
Motor stereotypy  
Myoclonus  
Paraplegia  
Parkinsonism

**MOTOR FUNCTION (cont.)**

Poor suck  
Postural instability  
Progressive muscle weakness  
Proximal muscle weakness  
Spasticity  
Tip-toe gait  
Torticollis  
Tremor  
Waddling gait  
**NEURAL MORPHOLOGY**  
Abnormal brainstem morphology  
Abnormal corpus callosum morphology  
Abnormal myelination  
Abnormal periventricular white matter morphology  
Abnormality of the basal ganglia  
Aplasia/Hypoplasia of the cerebellar vermis  
Aplasia/Hypoplasia of the cerebellum  
Brain stem compression  
Cerebellar atrophy  
Cerebral cortical atrophy  
CNS demyelination  
CNS hypomyelination  
Cortical dysplasia  
Cortical tubers  
Delayed CNS myelination  
Dysgenesis of the basal ganglia  
Encephalocele  
Gliosis  
Gray matter heterotopia  
Holoprosencephaly  
Hydrocephalus  
Hyperintensity of cerebral white matter on MRI  
Hypointensity of cerebral white matter on MRI  
Hypoplasia of the corpus callosum  
Leukodystrophy  
Lissencephaly  
Pachygryia  
Periventricular leukomalacia  
Periventricular nodular heterotopia  
Periventricular white matter hyperintensities  
Polymicrogyria  
Tethered cord  
Ventriculomegaly  
**NEURAL PHYSIOLOGY**  
Abnormal pineal melatonin secretion  
Agnosia  
Decreased intracranial pressure  
Decreased motor nerve conduction velocity  
Decreased nerve conduction velocity  
Decreased sensory nerve conduction velocity  
Demyelinating peripheral neuropathy  
Distal sensory impairment  
Dysarthria  
Encephalopathy  
Headache  
Increased intracranial pressure  
Lhermitte's sign  
Migraine  
Mononeuropathy  
Optic ataxia  
Peripheral neuropathy  
Polyneuropathy  
Psychosis  
Seizure  
Sensorimotor neuropathy

**NEURAL PHYSIOLOGY (cont.)**

Sensory neuropathy  
Vertigo  
**CRANIOFACIAL/DYSMORPHISM**  
Abnormal facial shape  
Abnormality of malar bones  
Anosmia  
Anteverted nares  
Bifid uvula  
Brachycephaly  
Choanal atresia  
Cleft lip  
Cleft palate  
Coarse facial features  
Craniosynostosis  
Dental enamel pits  
Dental malocclusion  
Depressed nasal bridge  
Depressed nasal tip  
Dolichocephaly  
Facial asymmetry  
Frontal bossing  
High palate  
Hypodontia  
Macrocephaly  
Malar flattening  
Mandibular prognathia  
Microcephaly  
Micrognathia  
Midface retrusion  
Narrow palate  
Retrognathia  
Short philtrum  
Sloping forehead  
Smooth philtrum  
Thick nasal alae  
Thin upper lip vermillion  
Trigonocephaly  
Wide anterior fontanel  
Wide nose  
Widely spaced teeth  
**EYE DEFECTS/VISION**  
Amblyopia  
Anophthalmia  
Blepharitis  
Blepharophimosis  
Cataract  
Cerebral visual impairment  
Coloboma  
Conjunctivitis  
Corneal dystrophy  
Corneal erosion  
Corneal opacity  
Developmental cataract  
Downslanted palpebral fissures  
Ectopia lentis  
Epicanthus  
Esotropia  
External ophthalmoplegia  
Foveal hyperpigmentation  
Hyperpigmentation of the fundus  
Hypertelorism  
Hypopigmentation of the fundus  
Lacrimal duct stenosis  
Macular atrophy  
Macular degeneration

**EYE DEFECTS/VISION (cont.)**

Macular purpura  
Microphthalmia  
Myopia  
Nystagmus  
Ophthalmoparesis  
Optic atrophy  
Optic neuropathy  
Posterior embryotoxon  
Proptosis  
Pterygium  
Ptosis  
Reduced visual acuity  
Retinal atrophy  
Retinal degeneration  
Retinal detachment  
Retinal dystrophy  
Retinoschisis  
Rod-cone dystrophy  
Strabismus  
Telecanthus  
Upturned palpebral fissure  
Vitreoretinopathy  
**EAR DEFECTS/HEARING**  
Conductive hearing impairment  
Cupped ear  
High-frequency hearing impairment  
Hypoplasia of the semicircular canal  
Low-frequency hearing loss  
Low-set ears  
Microtia  
Mild hearing impairment  
Posteriorly rotated ears  
Preauricular pit  
Protruding ear  
Sensorineural hearing impairment  
Unilateral deafness  
**NECK AND CHEST**  
Chest pain  
Cystic hygroma  
Gynecomastia  
Heterotaxy  
Inverted nipples  
Laryngeal stenosis  
Neck flexor weakness  
Neck muscle weakness  
Neck pterygia  
Shield chest  
Short neck  
Supernumerary nipple  
Webbed neck  
Wide intermamillary distance  
**RESPIRATORY SYSTEM**  
Apnea  
Asthma  
Bronchial wall thickening  
Bronchiectasis  
Central apnea  
Cyanosis  
Desquamative interstitial pneumonitis  
Dyspnea  
Hyperventilation  
Hypoventilation  
Hypoxemia  
Interstitial pneumonitis  
Intraalveolar phospholipid accumulation

RESPIRATORY SYSTEM (cont.)	ABDOMEN	REPRODUCTIVE SYSTEM (cont.)	HAIR AND NAIL (cont.)
Intralobular septal thickening	Cholangitis	Gonadal dysgenesis	Woolly hair
Neonatal respiratory distress	Cholelithiasis	Hyperplastic labia majora	SOFT TISSUE
Pleural effusion	Cholestasis	Hypoplasia of the uterus	Abnormal muscle fiber dystrophin expression
Pleuritis	Chronic diarrhea	Hypoplastic labia majora	Achilles tendon contracture
Pneumothorax	Cirrhosis	Inguinal hernia	Antecubital pterygium
Pulmonary fibrosis	Diastasis recti	Micropenis	Axillary pterygium
Pulmonary hypoplasia	Exocrine pancreatic insufficiency	Precocious puberty	Bulbar palsy
Recurrent bronchitis	Hepatic cysts	Shawl scrotum	Centrally nucleated skeletal muscle fibers
Respiratory failure	Hepatic fibrosis	Uterine prolapse	Deposits immunoreactive to $\beta$ -amyloid protein
Respiratory insufficiency	Malformation of the hepatic ductal plate	<b>SKIN</b>	Distal arthrogryposis
HEART	Pancreatic cysts	Abnormal blistering of the skin	Distal lower limb amyotrophy
Abnormal cardiac septum morphology	Pancreatic fibrosis	Acanthosis nigricans	Distal muscle weakness
Abnormal heart morphology	Peritonitis	Acne inversa	Elbow flexion contracture
Aortic root aneurysm	Polysplenia	Anhidrosis	Fatty replacement of skeletal muscle
Atrial septal defect	Protuberant abdomen	Aplasia cutis congenita	Flexion contracture
Bicuspid aortic valve	Umbilical hernia	Axillary freckling	Generalized muscle weakness
Conotruncal defect		Cafe-au-lait spot	Gowers sign
Dextrocardia	<b>GASTROINTESTINAL SYSTEM</b>	Cutis laxa	Hypertonia
Dilated cardiomyopathy	Anal atresia	Eczema	Hypotonia
Double outlet right ventricle	Anal stenosis	Erythema	Increased variability in muscle fiber diameter
Hypertrophic cardiomyopathy	Constipation	Fragile skin	Joint hypermobility
Hypoplastic left heart	Diarrhea	Hyperextensible skin	Minicore myopathy
Mitral stenosis	Duodenal stenosis	Hyperpigmentation of the skin	Muscle fiber cytoplasmatic inclusion bodies
Mitral valve prolapse	Dysphagia	Hypohidrosis	Muscle hypertrophy of the lower extremities
Noncompaction cardiomyopathy	Esophageal atresia	Hypopigmentation of the skin	Muscle spasm
Patent foramen ovale	Feeding difficulties	Ichthyosis	Myalgia
Pericardial effusion	Gastroesophageal reflux	Intermittent generalized erythematous papular rash	Myopathic facies
Pulmonic stenosis	Hepatomegaly	Jaundice	Myopathy
Restrictive cardiomyopathy	Inflammation of the large intestine	Keloids	Nemaline bodies
Transposition of the great arteries	Intrahepatic biliary atresia	Lichenoid skin lesion	Popliteal pterygium
Tricuspid regurgitation	Laryngomalacia	Oral mucosal blisters	Rhabdomyolysis
Ventricular septal defect	Nausea	Palmoplantar blistering	Shoulder girdle muscle atrophy
CARDIOVASCULAR SYSTEM	Pancreatitis	Palmoplantar hyperkeratosis	Skeletal muscle atrophy
Abnormal cardiac test	Pyloric stenosis	Palmoplantar keratoderma	Skeletal muscle hypertrophy
Aortic aneurysm	Rectal prolapse	Psoriasisiform dermatitis	Type 1 muscle fiber predominance
Aortic dissection	Splenomegaly	Pulmonary capillary hemangiomatosis	Type 2 muscle fiber predominance
Aortic regurgitation	Tracheoesophageal fistula	Single transverse palmar crease	Upper limb muscle hypertrophy
Aortic valve stenosis	Vomiting	Skin rash	Weakness of facial musculature
ARRHYTHMIA	<b>URINARY SYSTEM</b>	Soft skin	<b>SKELETAL: THORAX</b>
Arrhythmia	Cystic renal dysplasia	Subepidermal blistering	11 pairs of ribs
Arteriovenous malformation	Decreased numbers of nephrons	Telangiectasia	Abnormal clavicle morphology
Atrioventricular block	Ectopic kidney	Urticaria	Aplasia/Hypoplasia of the clavicles
Bradycardia	Glomerular sclerosis	Vascular skin abnormality	Aplasia/Hypoplasia of the ribs
Carotid artery calcification	Glomerulonephritis	Vitiligo	Aplasia/Hypoplasia of the sternum
Carotid artery dissection	Hematuria	<b>HAIR AND NAIL</b>	Barrel-shaped chest
Coarctation of aorta	Horseshoe kidney	Abnormality of the nail	Bifid sternum
Cutis marmorata	Hydronephrosis	Absent eyelashes	Broad clavicles
Hypertension	Hypospadias	Alopecia	Cuffed ribs
Mitral regurgitation	Multiple glomerular cysts	Anonychia	Enlarged thorax
Patent ductus arteriosus	Nephrolithiasis	Coarse hair	Flaring of rib cage
Prolonged QT interval	Polycystic kidney dysplasia	Hyperconvex fingernails	Pectus carinatum
Pulmonary arterial hypertension	Polyuria	Hypertrichosis	Pectus excavatum
Pulmonary venous hypertension	Proteinuria	Keratosis pilaris	Prominent sternum
Stroke	Proximal tubulopathy	Long eyelashes	Rib fusion
Supraventricular arrhythmia	Renal agenesis	Low anterior hairline	Short clavicles
Syncope	Renal amyloidosis	Lumbosacral hirsutism	Short ribs
Tachycardia	Renal cortical cysts	Nail dystrophy	Short sternum
Tetralogy of Fallot	Renal dysplasia	Sparse body hair	Supernumerary ribs
Total anomalous pulmonary venous	Renal insufficiency	Sparse eyebrow	Thin clavicles
Vascular dilatation	Ureteropelvic junction obstruction	Sparse eyelashes	<b>SKELETAL: SPINE AND PELVIS</b>
Ventricular arrhythmia	Vesicoureteral reflux	Sparse hair	Cervical spina bifida
Ventricular tachycardia		Sparse scalp hair	Coronal cleft vertebrae
Abdominal pain	<b>REPRODUCTIVE SYSTEM</b>	Synophrys	Hip dislocation
Ascites	Ambiguous genitalia	Thick eyebrow	Hyperlordosis
Asplenia	Bifid scrotum	Trichorrhexis nodosa	Hypoplastic pubic rami
Biliary cirrhosis	Clitoral hypoplasia		
	Cryptorchidism		

## SKELETAL: SPINE AND PELVIS (cont.)

Kyphoscoliosis  
Kyphosis  
Lumbar hyperlordosis  
Myelomeningocele  
Narrow iliac wing  
Platyspondyly  
Scoliosis  
Shallow acetabular fossae  
Spina bifida  
Spina bifida occulta  
Spinal canal stenosis  
Spinal rigidity  
Spondyloepiphyseal dysplasia  
Vertebral fusion  
Vertebral segmentation defect

## SKELETAL: LIMBS

Bifid femur  
Bowing of the legs  
Coxa valga  
Disproportionate short-limb short stature  
Fibular aplasia  
Fibular hypoplasia  
Fibular overgrowth  
Flat capital femoral epiphysis  
Genu valgum  
Hypoplasia of the ulna  
Mesomelia  
Mesomelic arm shortening  
Mesomelic leg shortening  
Mesomelic short stature  
Patellar hypoplasia  
Radioulnar synostosis  
Rhizomelia  
Rhizomelic arm shortening  
Rhizomelic leg shortening  
Short femoral neck  
Short humerus  
Short tibia  
Stippled calcification proximal humeral epiphyses  
Tibial bowing  
Widened proximal tibial metaphyses

## SKELETAL: HANDS AND FEET

Aplasia/Hypoplasia of metatarsal bones  
Aplasia/Hypoplasia of the hallux  
Aplasia/Hypoplasia of the radius  
Brachydactyly  
Broad hallux  
Broad thumb  
Carpal bone hypoplasia  
Clubbing of fingers  
Clubbing of toes  
Drumstick terminal phalanges  
Foot monodactyly  
Hammertoe  
Hand monodactyly  
Large carpal bones  
Large hands  
Metatarsus adductus  
Patellar dislocation  
Pes planus  
Polydactyly  
Postaxial hand polydactyly

## SKELETAL: HANDS AND FEET (cont.)

Prominent fingertip pads  
Proximal placement of thumb  
Short distal phalanx of finger  
Short metacarpal  
Short metatarsal  
Short phalanx of finger  
Split foot  
Split hand  
Syndactyly  
Synostosis of metacarpals/metatarsals  
Talipes equinovalgus  
Talipes equinovarus  
Tapered finger  
Triphalangeal thumb

## ENDOCRINE SYSTEM

Decreased response to growth hormone stimulation  
test Delayed puberty  
Diabetes insipidus  
Diabetes mellitus  
Hypercalcemia  
Hypergonadotropic hypogonadism  
Hyperparathyroidism  
Hyperphosphatemia  
Hyperthyroidism  
Hypocalcemia  
Hypoparathyroidism  
Hypophosphatemia  
Hypothyroidism  
Lymphedema  
Maturity-onset diabetes of the young  
Rickets  
Type I diabetes mellitus  
Type II diabetes mellitus

## HEMATOPOIETIC SYSTEM

Abnormal bleeding  
Anemia  
Anisocytosis  
Bone marrow hypercellularity  
Bruising susceptibility  
Epistaxis  
Gastrointestinal hemorrhage  
Hemolytic anemia  
Hypochromic anemia  
Hypoplastic anemia  
Macrocytic anemia  
Megaloblastic anemia  
Menorrhagia  
Microangiopathic hemolytic anemia  
Microcytic anemia  
Pancytopenia  
Poikilocytosis  
Prolonged bleeding time  
Sideroblastic anemia  
Stomatocytosis  
Thrombocytopenia

## IMMUNE SYSTEM

Abnormal T cell morphology  
Agammaglobulinemia  
Allergic rhinitis  
B lymphocytopenia  
Decreased circulating antibody level  
Decreased circulating IgA level

## IMMUNE SYSTEM (cont.)

Decreased circulating IgG level  
Decreased circulating total IgM  
Decreased proportion of CD4-positive helper T cells  
Eczematoid dermatitis  
Folliculitis  
Hashimoto thyroiditis  
Hemophagocytosis  
Immunodeficiency  
Leukopenia  
Lymphadenopathy  
Lymphopenia  
Myelokathexis  
Neutropenia  
Panhypogammaglobulinemia  
Predominantly dermal neutrophilic infiltrate  
Pyoderma gangrenosum  
Rectal abscess  
Recurrent bacterial infections  
Recurrent enteroviral infections  
Recurrent infections  
Recurrent otitis media  
Recurrent pneumonia  
Recurrent respiratory infections  
Recurrent sinusitis  
Reticulocytosis  
Schistocytosis  
Seborrheic dermatitis  
Verrucae

**METABOLISM**

Abnormal activity of mitochondrial respiratory chain  
Abnormality of mitochondrial metabolism  
Amyloidosis  
Conjugated hyperbilirubinemia  
Decreased adenosylcobalamin  
Decreased methylcobalamin  
Elevated circulating creatine kinase concentration  
Elevated circulating creatinine concentration  
Elevated circulating glutaric acid concentration  
Elevated hepatic transaminase  
Elevated plasma branched chain amino acids  
Ethylmalonic aciduria  
Generalized aminoaciduria  
Glutaric aciduria  
Glycosuria  
Homocystinuria  
Hyperammonemia  
Hyperbilirubinemia  
Hypercholesterolemia  
Hyperglycemia  
Hyperglycinemia  
Hyperinsulinemia  
Hyperketonemia  
Hypertriglyceridemia  
Hyperuricemia  
Hypoammonemia  
Hypoglycemia  
Increased blood urea nitrogen  
Increased serum pyruvate  
Ketonuria  
Lactic acidosis  
Metabolic acidosis  
Metabolic ketoacidosis

## ADDITIONAL NOTES