

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

Gliososis

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

Pachygyria

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

Upslanted 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.)

Intralobular septal thickening
Neonatal respiratory distress
Pleural effusion
Pleuritis
Pneumothorax
Pulmonary fibrosis
Pulmonary hypoplasia
Recurrent bronchitis
Respiratory failure
Respiratory insufficiency

HEART

Abnormal cardiac septum morphology
Abnormal heart morphology
Aortic root aneurysm
Atrial septal defect
Bicuspid aortic valve
Conotruncal defect
Dextrocardia
Dilated cardiomyopathy
Double outlet right ventricle
Hypertrophic cardiomyopathy
Hypoplastic left heart
Mitral stenosis
Mitral valve prolapse
Noncompaction cardiomyopathy
Patent foramen ovale
Pericardial effusion
Pulmonic stenosis
Restrictive cardiomyopathy
Transposition of the great arteries
Tricuspid regurgitation
Ventricular septal defect

CARDIOVASCULAR SYSTEM

Abnormal cardiac test
Aortic aneurysm
Aortic dissection
Aortic regurgitation
Aortic valve stenosis
Arrhythmia
Arteriovenous malformation
Atrioventricular block
Bradycardia
Carotid artery calcification
Carotid artery dissection
Coarctation of aorta
Cutis marmorata
Hypertension
Mitral regurgitation
Patent ductus arteriosus
Prolonged QT interval
Pulmonary arterial hypertension
Pulmonary venous hypertension
Stroke
Supraventricular arrhythmia
Syncope
Tachycardia
Tetralogy of Fallot
Total anomalous pulmonary venous
Vascular dilatation
Ventricular arrhythmia
Ventricular tachycardia
Abdominal pain
Ascites
Asplenia
Biliary cirrhosis

ABDOMEN

Cholangitis
Cholelithiasis
Cholestasis
Chronic diarrhea
Cirrhosis
Diastasis recti
Exocrine pancreatic insufficiency
Hepatic cysts
Hepatic fibrosis
Malformation of the hepatic ductal plate
Pancreatic cysts
Pancreatic fibrosis
Peritonitis
Polysplenia
Protuberant abdomen
Umbilical hernia

GASTROINTESTINAL SYSTEM

Anal atresia
Anal stenosis
Constipation
Diarrhea
Duodenal stenosis
Dysphagia
Esophageal atresia
Feeding difficulties
Gastroesophageal reflux
Hepatomegaly
Inflammation of the large intestine
Intrahepatic biliary atresia
Laryngomalacia
Nausea
Pancreatitis
Pyloric stenosis
Rectal prolapse
Splenomegaly
Tracheoesophageal fistula
Vomiting

URINARY SYSTEM

Cystic renal dysplasia
Decreased numbers of nephrons
Ectopic kidney
Glomerular sclerosis
Glomerulonephritis
Hematuria
Horseshoe kidney
Hydronephrosis
Hypospadias
Multiple glomerular cysts
Nephrolithiasis
Polycystic kidney dysplasia
Polyuria
Proteinuria
Proximal tubulopathy
Renal agenesis
Renal amyloidosis
Renal cortical cysts
Renal dysplasia
Renal insufficiency
Ureteropelvic junction obstruction
Vesicoureteral reflux

REPRODUCTIVE SYSTEM

Ambiguous genitalia
Bifid scrotum
Clitoral hypoplasia
Cryptorchidism

REPRODUCTIVE SYSTEM (cont.)

Gonadal dysgenesis
Hyperplastic labia majora
Hypoplasia of the uterus
Hypoplastic labia majora
Inguinal hernia
Micropenis
Precocious puberty
Shawl scrotum
Uterine prolapse
SKIN
Abnormal blistering of the skin
Acanthosis nigricans
Acne inversa
Anhidrosis
Aplasia cutis congenita
Axillary freckling
Cafe-au-lait spot
Cutis laxa
Eczema
Erythema
Fragile skin
Hyperextensible skin
Hyperpigmentation of the skin
Hypohidrosis
Hypopigmentation of the skin
Ichthyosis
Intermittent generalized erythematous papular rash
Jaundice
Keloids
Lichenoid skin lesion
Oral mucosal blisters
Palmoplantar blistering
Palmoplantar hyperkeratosis
Palmoplantar keratoderma
Psoriasiform dermatitis
Pulmonary capillary hemangiomatosis
Single transverse palmar crease
Skin rash
Soft skin
Subepidermal blistering
Telangiectasia
Urticaria
Vascular skin abnormality
Vitiligo

HAIR AND NAIL
Abnormality of the nail
Absent eyelashes
Alopecia
Anonychia
Coarse hair
Hyperconvex fingernails
Hypertrichosis
Keratosis pilaris
Long eyelashes
Low anterior hairline
Lumbosacral hirsutism
Nail dystrophy
Sparse body hair
Sparse eyebrow
Sparse eyelashes
Sparse hair
Sparse scalp hair
Synophrys
Thick eyebrow
Trichorrhexis nodosa

HAIR AND NAIL (cont.)

Woolly hair
SOFT TISSUE
Abnormal muscle fiber dystrophin expression
Achilles tendon contracture
Antecubital pterygium
Axillary pterygium
Bulbar palsy
Centrally nucleated skeletal muscle fibers
Deposits immunoreactive to β -amyloid protein
Distal arthrogryposis
Distal lower limb amyotrophy
Distal muscle weakness
Elbow flexion contracture
Fatty replacement of skeletal muscle
Flexion contracture
Generalized muscle weakness
Gowers sign
Hypertonia
Hypotonia
Increased variability in muscle fiber diameter
Joint hypermobility
Minicore myopathy
Muscle fiber cytoplasmatic inclusion bodies
Muscle hypertrophy of the lower extremities
Muscle spasm
Myalgia
Myopathic facies
Myopathy
Nemaline bodies
Popliteal pterygium
Rhabdomyolysis
Shoulder girdle muscle atrophy
Skeletal muscle atrophy
Skeletal muscle hypertrophy
Type 1 muscle fiber predominance
Type 2 muscle fiber predominance
Upper limb muscle hypertrophy
Weakness of facial musculature
SKELETAL: THORAX
11 pairs of ribs
Abnormal clavicle morphology
Aplasia/Hypoplasia of the clavicles
Aplasia/Hypoplasia of the ribs
Aplasia/Hypoplasia of the sternum
Barrel-shaped chest
Bifid sternum
Broad clavicles
Cupped ribs
Enlarged thorax
Flaring of rib cage
Pectus carinatum
Pectus excavatum
Prominent sternum
Rib fusion
Short clavicles
Short ribs
Short sternum
Supernumerary ribs
Thin clavicles
SKELETAL: SPINE AND PELVIS
Cervical spina bifida
Coronal cleft vertebrae
Hip dislocation
Hyperlordosis
Hypoplastic pubic rami

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
Bowling 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 equinovaglus
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
Ketoneuria
Lactic acidosis
Metabolic acidosis
Metabolic ketoacidosis

ADDITIONAL NOTES