alamya health

PATIENT INFORMATION

Patient Name	Date of Birth	Sex assigned at birth	Ancestry/Ethnicity

Postal Address	Email	Phone

Affected Status ■ Affected/Symptomatic ■ Unaffected/Asymptomatic ■ Carrier testing/At risk

Has the patient had a stem cell or bone marrow transplant? ■ No ■ Yes *If yes, date of transplant:*

Has the patient had a blood transfusion ■ No ■ Yes *If yes, date of last transfusion:*

FAMILY SAMPLE INFORMATION

Mother ■ Not available	Name	Date of Birth	■ Symptomatic ■ Asmyptomatic	MRN	Sample Type ■ Blood ■ Saliva
Father ■ Not available	Name	Date of Birth	■ Symptomatic ■ Asmyptomatic	MRN	Sample Type ■ Blood ■ Saliva
Other: ■ Not available	Name	Date of Birth	■ Symptomatic ■ Asmyptomatic	MRN	Sample Type ■ Blood ■ Saliva

PROVIDER INFORMATION

Provider Name	Institution	
Email	Phone	Fax

Copy results to:

TEST INFORMATION

Test Menu – Please select all that apply

- ■ **First in Class (Short-read genome w/ reflex testing)**
- ■ Next Era **(Short-read & long-read genome w/ reflex testing)**
- ■ Generation Beyond **(Long-read genome)**
- ■ Variant Clarification
  - ■ Short-read Transcriptome
  - ■ Infinium MethylationEPIC microarray
  - ■ Long-read genome
- ■ Long-read genome sequencing for researchers
- ■ Genomic **Health Screen**
- ■ Other

Secondary Findings

- ■ As per consent, I wish to receive information about secondary findings unrelated to the primary indication for testing as recommended by the ACMG.

PROVIDER STATEMENT

By signing this form, I acknowledge that written informed consent for genome-wide molecular testing has been obtained from the participant or the participant's parent or legal guardian and they understand the risks of genetic testing. De-identified clinical information and sequencing data will be stored for a minimum of three years and may be used by Alamya Health for the purpose of quality assurance. De-identified variant information will contribute to publicly accessible clinical and population variant databases; however, no personal identifying information will be disclosed without the patient's explicit consent.

Signature* Date

* Option 1: Click signature box to sign with Adobe Digital ID. Option 2: Select "Prepare Form", then "Sign Yourself" to click & drag your signature. Option 3: Print document & physically sign.

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CLINICAL INFORMATION

Clinical Diagnosis Age of onset:	MONDO Disease (Optional)	ICD-10 Codes (Optional)

HPO Terms (Optional – if HPO terms are not provided, please provide a clinical consult note and/or use the phenotype checklist provided on the following page)

Differential Diagnosis (Optional)	Genes or Locus of Interest (Optional)

Was the patient taking any medications at the time of sample collection? ■ No ■ Yes

If yes, please list past and current conditions and the medications prescribed

Condition	Medication	Dosage and duration (if known)
E.g. Seizures	Carbamazepine	200mg BID
1.		
2.		
3.		

Does the patient have a history of cancer? ■ No ■ Yes

If yes, please list the patient's cancer history and age(s) of diagnosis:

Primary diagnosis	Age of diagnosis	Histological or molecular subtype (if known)
E.g. Rhabdomyosarcoma	46 months	Embryonal anaplastic
1.		
2.		

If yes, has the patient received systemic chemotherapy? ■ No ■ Yes

If yes, please specify:

Please provide additional information about the patient's phenotype or medical history that may not be captured by the phenotype ontologies provided.

Molecular Findings

- ■ Conventional karyotyping/FISH
  - *Karyotype:*
- ■ Chromosomal microarray
  - *Result:*
- ■ Genome/exome sequencing
  - *Variants:*
- ■ Single gene or multigene panel
  - *Variants:*
  - *Gene, indication, or panel name and testing provider:*
- ■ MS-PCR/MS-MLPA
  - *Result:*
- ■ Other
  - *Specify:*

Metabolic Findings

- ■ Abnormal mitochondrial respiratory chain activity
- ■ Abnormal plasma/urine amino acids
- ■ Abnormal plasma/urine organic acids
- ■ Carnitine deficiency
- ■ Elevated creatine
- ■ Hyperammonemia
- ■ Hypoglycemia
- ■ Ketonuria
- ■ Metabolic acidosis
- ■ Other
  - *Specify:*

Other Tests and Imaging

- ■ CT
- ■ Echocardiogram
- ■ EEG
- ■ EMG
- ■ MRI
- ■ Ultrasound
- ■ X-ray
- ■ Other
  - *Specify:*

Are any clinical or molecular investigations pending for this patient? ■ No ■ Yes

If yes, please specify:

Please provide a family pedigree and/or information about family history of disease that may be relevant to the interpretation of the patient's test results.

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PHENOTYPE CHECKLIST (OPTIONAL)

Auditory/Ophthalmologic

- ☐ Anophthalmial
- ☐ Cataract
- ☐ Coloboma
- ☐ Ectopia Lentis
- ☐ External ophthalmoplegia
- ☐ Hearing impairment (specify):
  Sensorineural /Conductive
- ☐ Microphthalmia
- ☐ Myopia
- ☐ Nystagmus
- ☐ Strabismus
- ☐ Optic atrophy
- ☐ Optic neuropathy
- ☐ Ptosis
- ☐ Retinal detachment
- ☐ Other
  Specify

Cardiovascular

- ☐ Amyloidosis
- ☐ Aneurysm
- ☐ Aortic root dilation
- ☐ Arrhythmia
  Specify
- ☐ Arteriovenous malformation
- ☐ Cardiomyopathy
  Specify
- ☐ Congenital heart defect
  Specify
- ☐ Dissection
- ☐ Epistaxis
- ☐ Stroke
- ☐ Heterotaxy
- ☐ Hypertension
- ☐ Lymphedema
- ☐ Mitral valve prolapse
- ☐ Pulmonary hypertension
- ☐ Sudden death
- ☐ Other
  Specify

Craniofacial/Dysmorphism

- ☐ Brachycephaly
- ☐ Cleft lip
- ☐ Cleft palate
- ☐ Craniosynostosis
- ☐ Micrognathia
- ☐ Retrognathia
- ☐ Other
  Specify

Developmental/Psychiatric

- ☐ Autistic behavior
- ☐ Developmental regression
- ☐ Developmental delay (specify):
  Gross motor/ Fine motor/Speech
- ☐ Global developmental delay
- ☐ Intellectual disability (specify):
  Mild/ Moderate/Severe
- ☐ Psychiatric symptoms
- ☐ Other
  Specify

Endocrine

- ☐ Delayed puberty
- ☐ Diabetes Insipidus
- ☐ Diabetes Mellitus
- ☐ Hyperthyroidism
- ☐ Hypophosphatemia
- ☐ Hypothyroidism
- ☐ Maturity-onset diabetes of the young
- ☐ Other
  Specify

Gastrointestinal

- ☐ Constipation
- ☐ Diarrhea
- ☐ Duodenal stenosis/atresia
- ☐ Feeding difficulties
- ☐ Gastroesophageal reflux
- ☐ Gastroschisis
- ☐ Hepatomegaly
- ☐ Inflammatory bowel disease
- ☐ Intrahepatic biliary atresia
- ☐ Omphalocele
- ☐ Pancreatitis
- ☐ Pyloric stenosis
- ☐ Splenomegaly
- ☐ Other
  Specify

Genitourinary

- ☐ Ambiguous genitalia
- ☐ Cryptorchidism
- ☐ Cystic renal dysplasia
- ☐ Horseshoe kidney
- ☐ Hydronephrosis
- ☐ Hypospadia
- ☐ Micropenis
- ☐ Polycystic kidney
- ☐ Proximal renal tubulopathy
- ☐ Renal agenesis
- ☐ Other
  Specify

Growth Parameters

Please provide the following growth parameters where known:

	Length or height	Weight	Head circumference
At birth			
Current			

- ☐ Failure to thrive
- ☐ Growth restriction/short stature
- ☐ Hemihyperplasia
- ☐ Hemihypoplasia
- ☐ Macrocephaly
- ☐ Microcephaly
- ☐ Overgrowth
- ☐ Other
  Specify

Hematologic/Immunologic

- ☐ Allergic rhinitis
- ☐ Anemia
- ☐ Immunodeficiency
- ☐ Neutropenia
- ☐ Pancytopenia
- ☐ Recurrent infections
- ☐ Thrombocytopenia
- ☐ Other
  Specify

Musculoskeletal

- ☐ Arachnodactyly
- ☐ Arthrogryposis
- ☐ Bruising susceptibility
- ☐ Decreased muscle mass
- ☐ Ectrodactyly
- ☐ Fatigue
- ☐ Hypertonia
- ☐ Hypotonia
- ☐ Joint hypermobility
- ☐ Muscle weakness
- ☐ Myopathy
- ☐ Osteopenia
- ☐ Polydactyly
- ☐ Pectus deformity
- ☐ Recurrent fractures
- ☐ Scoliosis
- ☐ Skeletal dysplasia
  Specify
- ☐ Syndactyly
- ☐ Vertebral anomaly
- ☐ Other
  Specify

PHENOTYPE CHECKLIST (OPTIONAL)

Neurological

- ☐ Ataxia
- ☐ Cerebral palsy
- ☐ Chorea
- ☐ Cortical Visual Impairment
- ☐ Dementia
- ☐ Dysarthria
- ☐ Dysphasia
- ☐ Dystonia
- ☐ Encephalopathy
- ☐ Headaches
- ☐ Hemiplegia
- ☐ Infantile spasms
- ☐ Migraines
- ☐ Myoclonus
- ☐ Parkinsonism
- ☐ Peripheral neuropathy
- ☐ Seizure

Specify

- ☐ Spasticity
- ☐ Other

Specify

Prenatal/Perinatal

- ☐ Intrauterine growth restriction
- ☐ Oligohydramnios
- ☐ Polyhydramnios
- ☐ Premature birth
- ☐ Other

Specify

Respiratory

- ☐ Asthma
- ☐ Bronchiectasis
- ☐ Hyperventilation
- ☐ Hypoventilation
- ☐ Laryngomalacia
- ☐ Pneumothorax
- ☐ Pulmonary fibrosis
- ☐ Respiratory insufficiency
- ☐ Tracheomalacia
- ☐ Other

Specify

Skin/Hair

- ☐ Abnormal hair pattern/quantity
- ☐ Abnormality of nail
- ☐ Anhidrosis
- ☐ Blistering
- ☐ Café-Au-Lait Macules
- ☐ Cutis Laxa
- ☐ Eczema
- ☐ Hemangiomas
- ☐ Hyperextensible skin
- ☐ Hyperpigmentation
- ☐ Hypohidrosis
- ☐ Hypopigmentation
- ☐ Ichthyosis
- ☐ Telangiectasia
- ☐ Velvety skin
- ☐ Other

Specify

Structural Brain Abnormalities

- ☐ Abnormal myelination
- ☐ Abnormality of basal ganglia
- ☐ Agenesis of the corpus callosum
- ☐ Cerebellar atrophy
- ☐ Cortical dysplasia
- ☐ Hemimegalencephaly
- ☐ Heterotopia
- ☐ Holoprosencephaly
- ☐ Hydrocephalus
- ☐ Leukodystrophy
- ☐ Lissencephaly
- ☐ Polymicrogyria
- ☐ Other

Specify

PAYMENT	☐ INSTITUTIONAL		☐ PATIENT
Responsible party's name	Phone#		
Address	City	State	Zip
Email			
Signature			

KIT ORDERS		
■ I do not need any kits		
■ I need the following kit(s)/tubes:		
■ Saliva sponge kit (for individuals unable to spit)		How many? (0-10)
■ EDTA + PAXgene RNA kit ■ Adult (6.0ml EDTA + 2.5ml PAXgene) ■ Infant/child (2.0ml EDTA + 2.5ml PAXgene)		 How many? (0-10) How many? (0-10)
■ Saliva spit kit		How many? (0-50)
■ Freestanding tubes:	■ EDTA ■ PAXgene RNA	How many? (0-50)
■ Alamya branded shipping box		How many? (0-10)

Please provide an alternate address to ship the kits if not going to the ordering provider.

First name:

Last name

Address

Telephone number:

Email address