alamya health

BREAKTHROUGH GENOMICS

PATIENT INFORMATION

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

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	Collection Date
Father ■ Not available	Name	Date of Birth	■ Symptomatic ■ Asmyptomatic	MRN	Collection Date
Other: ■ Not available	Name	Date of Birth	■ Symptomatic ■ Asmyptomatic	MRN	Collection Date

PROVIDER INFORMATION

Provider Name	Institution	
Email	Phone	Fax
Copy results to:		

TEST INFORMATION

Test Menu – Please select all that apply

■ Short-read genome sequencing
* Reflex testing to short-read transcriptome and Infinium MethylationEPIC microarray for interpretative clarification when indicated.

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

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	Metabolic Findings	Other Tests and Imaging
■ Conventional karyotyping/FISH	■ Abnormal mitochondrial respiratory chain activity	■ CT
Karyotype:	■ Abnormal plasma/urine amino acids	■ Echocardiogram
■ Chromosomal microarray	■ Abnormal plasma/urine organic acids	■ EEG
Result:	■ Carnitine deficiency	■ EMG
■ Genome/exome sequencing	■ Elevated creatine	■ MRI
Variants:	■ Hyperammonemia	■ Ultrasound
■ Single gene or multigene panel	■ Hypoglycemia	■ X-ray
Variants: *Gene, indication, or panel name and testing provider:*	■ Ketonuria	■ Other
	■ Metabolic acidosis	*Specify:*
■ MS-PCR/MS-MLPA	■ Other	
Result:	*Specify:*	
■ 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.

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

alamya health

BREAKTHROUGH GENOMICS

INSTITUTIONAL BILLING

Billing Institution		PO Number	
Contact	Phone#	Email	
Address	City	State	Zip
Billing Account Number			
Email Invoice via secure email			

PATIENT PAY

Responsible party's name (must be 18 years or older)		Phone#	
Address	City	State	Zip
Email			
Signature*			
Credit Card Number	Expiration date	3-digit security number	

My signature authorizes Alamya Health to charge my credit card for services for which I am responsible.

Credit Card Holders Signature: Date:

INSURANCE INFORMATION

Indicate the type of insurance (attach copy of Insurance Card (both sides))

■ Private ■ Tricare ■ Medicare ■ Medicaid

Policy holder name	Date of birth	Relationship to patient

Primary insurance company name (required)	Phone#

Policy ID#	Group#	Authorization#

Secondary Insurance (attach copy of Insurance Card (both sides))

Testing will proceed unless:

- We (or you) are working in a required Pre-Authorization
- No insurance coverage is available. We will work with you or your patient to determine payment options