Questionnaire for WES solo and WGS solo

Bumrungrad International HOSPITAL

Test ordered

☐ Whole Exome Sequencing (WES)-Solo
☐ Whole Genome Sequencing (WGS)-Solo

Patient Information

1. **Patient ancestry**

☐ Arabs	☐ Ashkenazi Jewish
☐ Asian	☐ Black/African-American
☐ French Canadian	☐ Hispanic
☐ Mediterranean	☐ Native American
☐ Pacific Islander	☐ Sephardic Jewish
☐ White/Caucasian	☐ Others (please specify) .....

Patient sticker

Clinical Information

2. **Patient's primary indication(s)**

   Please specify:.....

3. **Age of onset**:.....

4. **Are there any suspected genes related to the patient's condition?**

   ☐ No ☐ Yes (please specify).....

5. **Patient has a current or history of a hematological malignancy**

   ☐ No

   ☐ Yes, please specify: ○ Active/current ○ History

   (*If active, we are unable to accept the specimen, please do not order)**

6. **Patient had a blood transfusion less than four weeks prior to specimen collection (blood and saliva)**

   ☐ No ☐ Yes (please specify date of last transfusion) .....

   (*If yes, we are unable to accept the specimen, please do not order)**

7. **Patient who have had an allogenic bone marrow or stem cell transplant**

   ☐ No ☐ Yes **(*** If yes, we are unable to accept the specimen, please do not order)**

8. Patient's sign(s) or clinical symptom(s)

Please check all that apply to the patient (current or previous history of) and specify in the space provided. Accurate clinical information is critical for exome/genome analysis and interpretation. Clinical information indicated below follow HPO nomenclature.

Pre/Perinatal History

- ☐ Cystic hygroma
- ☐ Congenital diaphragmatic hernia
- ☐ Encephalocele
- ☐ Growth delay
- ☐ Increased nuchal translucency
- ☐ Intrauterine Growth Retardation
- ☐ Nonimmune hydrops fetalis
- ☐ Oligohydramnios
- ☐ Omphalocele
- ☐ Polyhydramnios
- ☐ Prolonged neonatal jaundice

Cardiac Findings

- ☐ Abnormal heart morphology
- ☐ Amyloidosis
- ☐ aortic root aneurysm
- ☐ Arrhythmia
- ☐ Atrial septal defect
- ☐ Bicuspid aortic valve
- ☐ Bradycardia
- ☐ Coarctation of aorta
- ☐ Dilated cardiomyopathy
- ☐ Heterotaxy
- ☐ Hypertension
- ☐ Hypertrophic cardiomyopathy
- ☐ Mitral valve prolapse
- ☐ Noncompaction cardiomyopathy
- ☐ Patent ductus arteriosis
- ☐ Patent foramen ovale
- ☐ Prolonged QTc interval
- ☐ Sudden death
- ☐ Tetralogy of Fallot
- ☐ Ventricular septal defect
- ☐ Ventricular tachycardia

Craniofacial/Dysmorphism

- ☐ Abnormal facial shape (Dysmorphic features) (Specify):
  _____
- ☐ Brachycephaly
- ☐ Cleft lip
- ☐ Coarse facial features
- ☐ Craniosynostosis
- ☐ Macrocephaly
- ☐ Microcephaly
- ☐ Short neck
- ☐ Synophrys

Developmental/Behavioral Findings

- ☐ Absent speech
- ☐ Aggressive behavior
- ☐ Anxiety
- ☐ Autistic Behavior
- ☐ Cognitive impairment
- ☐ Delayed speech & language development
- ☐ Developmental regression
- ☐ Dysarthria
- ☐ Gait disturbance
- ☐ Global developmental delay
- ☐ Hyperactivity
- ☐ Incoordination
- ☐ Intellectual disability
- ☐ Specific learning disability
- ☐ Memory impairment
- ☐ Sleep disturbance
- ☐ Stereotypy

Endocrine Findings

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

Eye Defects/ Vision

- ☐ Abnormality of Vision
- ☐ Anophthalmia
- ☐ Cataracts
- ☐ Coloboma
- ☐ Corneal opacity
- ☐ Ectopia lentis
- ☐ External ophthalmoplegia
- ☐ Microphthalmia
- ☐ Myopia
- ☐ Nystagmus
- ☐ Optic atrophy
- ☐ Optic neuropathy
- ☐ Ptosis
- ☐ Retinal detachment
- ☐ Retinitis pigmentosa inversa
- ☐ Strabismus

Gastrointestinal Findings

- ☐ Constipation
- ☐ Diarrhea
- ☐ Duodenal stenosis/atresia
- ☐ Exocrine pancreatic insufficiency
- ☐ Failure to thrive
- ☐ Feeding difficulties
- ☐ Gastroesophageal reflux
- ☐ Hepatomegaly
- ☐ Inflammatory bowel disease
- ☐ Intrahepatic biliary atresia
- ☐ Laryngomalacia
- ☐ Nausea
- ☐ Pancreatitis
- ☐ Pyloric stenosis
- ☐ Splenomegaly
- ☐ Tracheoesophageal fistula
- ☐ Vomiting

Genitourinary Findings

- ☐ Ambiguous genitalia
- ☐ Cryptorchidism
- ☐ Cystic renal dysplasia
- ☐ Horseshoe kidney
- ☐ Hydronephrosis
- ☐ Hypospadias
- ☐ Inguinal hernia
- ☐ Micropenis

Hearing Impairment

- ☐ Conductive hearing impairment
- ☐ Sensorineural hearing impairment

Hematologic or Immunologic Findings

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

Metabolic Findings

- ☐ Abnormal activity of mitochondrial respiratory
- ☐ Abnormality of mitochondrial metabolism
- ☐ Elevated CPK
- ☐ Elevated hepatic transaminase
- ☐ Hyperammonemia
- ☐ Hyperglycemia
- ☐ Hypoammonemia
- ☐ Hypoglycemia
- ☐ Increased serum pyruvate
- ☐ Lactic acidosis

Musculoskeletal Findings

- ☐ Abnormal form of the vertebral bodies
- ☐ Abnormality of the ribs
- ☐ Arachnodactyly
- ☐ Arthralgia
- ☐ Arthrogryposis syndrome
- ☐ Bruising susceptibility
- ☐ Clinodactyly
- ☐ Decreased muscle mass
- ☐ Ectrodactyly
- ☐ Exercise intolerance
- ☐ Fatigue

Musculoskeletal Findings (cont.)

- ☐ Hemihypertrophy
- ☐ Hypertonia
- ☐ Hypotonia
- ☐ Joint hypermobility
- ☐ Muscle weakness
- ☐ Myalgia
- ☐ Myopathic facies
- ☐ Myopathy
- ☐ Osteoarthritis
- ☐ Osteopenia
- ☐ Pain
- ☐ Pectus carinatum
- ☐ Pectus excavatum
- ☐ Polydactyly
- ☐ Recurrent fractures
- ☐ Rhabdomyolysis
- ☐ Scoliosis
- ☐ Short stature
- ☐ Skeletal dysplasia
- ☐ Syndactyly
- ☐ Tall stature

Neurological Findings

- ☐ Abnormality of central nervous system electrophysiology
- ☐ Ataxia
- ☐ Cerebral palsy
- ☐ Chorea
- ☐ Dementia
- ☐ Dysarthria
- ☐ Dyskinesia
- ☐ Dysphasia
- ☐ Dystonia
- ☐ Encephalopathy
- ☐ Headaches
- ☐ Hemiplegia
- ☐ Infantile Spasms
- ☐ Migraines
- ☐ Myoclonus
- ☐ Parkinsonism
- ☐ Peripheral neuropathy
- ☐ Seizure
- ☐ Sensory neuropathy
- ☐ Spasticity
- ☐ Syncope
- ☐ Tremors (Specify): _____
- ☐ Vertigo

Respiratory Findings

- ☐ Asthma
- ☐ Bronchiectasis
- ☐ Hyperventilation
- ☐ Hypoventilation
- ☐ Pneumothorax
- ☐ Pulmonary fibrosis
- ☐ Respiratory insufficiency

Structural Brain Abnormalities

- ☐ Abnormal myelination
- ☐ Abnormality of brain morphology
- ☐ Aplasia/hypoplasia of cerebellum
- ☐ Arnold Chiari malformation
- ☐ Cerebellar atrophy
- ☐ Heterotopia (Periventricular nodular heterotopia)
- ☐ Holoprosencephaly
- ☐ Hydrocephalus
- ☐ Leukodystrophy
- ☐ Lissencephaly
- ☐ Pachygyria
- ☐ Polymicrogyria
- ☐ Ventriculomegaly

Skin/Hair Findings

- ☐ Abnormal blistering of the skin
- ☐ Abnormality of nail color
- ☐ Alopecia
- ☐ Anhidrosis
- ☐ Café-Au-Lait Macules
- ☐ Coarse hair
- ☐ Cutis Laxa
- ☐ Eczema
- ☐ Hemangioma
- ☐ Hyperextensible skin
- ☐ Hyperpigmentation of the skin
- ☐ Hypohidrosis
- ☐ Hypopigmentation of the skin
- ☐ Ichthyosis
- ☐ Skin rash
- ☐ Sparse hair
- ☐ Telangiectasia
- ☐ Vascular skin abnormality
- ☐ Acanthosis nigricans

9. Patient is adopted

☐ No ☐ Yes

10. Patient has previous genetic result related to the disease(s)/symptom(s)?

☐ No ☐Yes (please specify mutation result) ……

11. Patient has previous WES or WGS result from Bumrungrad hospital

☐ No ☐ Yes (please specify mutation result) ……..

12. Patient's family has previous WES or WGS result from Bumrungrad hospital

☐ No ☐ Yes (please specify mutation result) ……

13. Patient has family history (including 1st and 2nd degree relatives) related to the disease(s)/symptom(s)?

☐ Unknown
☐ No
☐ Yes (please complete section 10.1)

10.1 **Father** ☐ Affected …… ☐ Unaffected ☐ Unknown
Mother ☐ Affected …… ☐ Unaffected ☐ Unknown

Additional family member 1 relationship……

☐ Affected ……
☐ Unaffected
☐ Unknown

Additional family member 2 relationship……

☐ Affected ……
☐ Unaffected
☐ Unknown

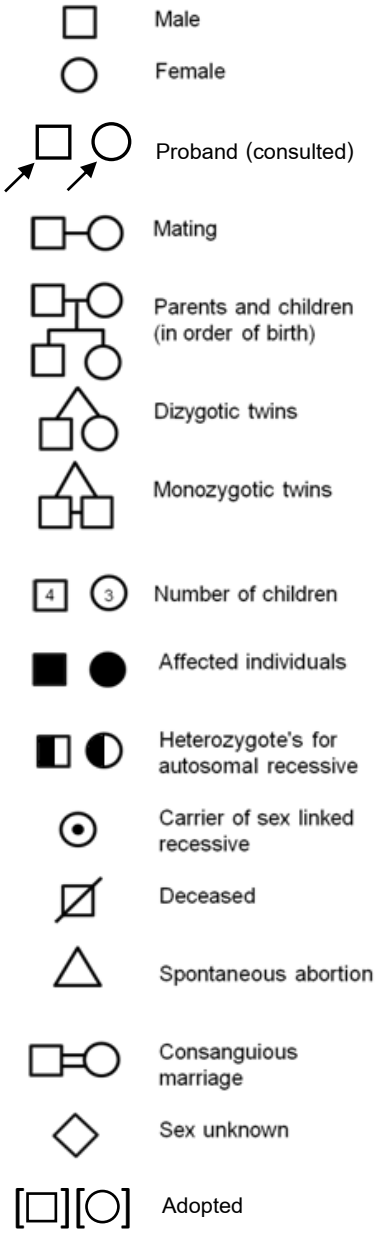
Additional family member 3 relationship……

☐ Affected ……
☐ Unaffected
☐ Unknown

14. Pedigree*

Race/Ethnicity:
Paternal Ancestry: _____
Maternal Ancestry: _____
Consanguinity: ☐ Yes ☐ No

Pedigree Nomenclature



**Please complete at least three-generation pedigree of the proband's family for family history assessment*