GC Genome
The Frontier Company in Genomic Diagnostics

International Requisition Form
Hereditary(Rare) Diseases

Barcode

* All reqired fields MUST be filledin.

Patient Information			
***First Name**		***Last Name**	
***Date of Birth**	D D / M M / Y Y Y Y	***Sex**	□ M □ F
City / State / Country		***Primary Ethnicity (Choose one)**	□ African □ Asian □ Caucasian □ Hispanic □ Others

Physician Information			
***Clinic/Hospital Name**		***Department**	
***Name**		**E-mail**	

Specimen Information			
***Collection Date**	D D / M M / Y Y Y Y	**Sample Type**	□ EDTA 3ml □ G-card (Blood paper)

* Please complete the family information for the Trio and follow-up family test.(Page3)

NGS-Exome Sequencing	
Diagnostic Exome Sequencing (DES)	☐ Proband ☐ Duo ☐ Trio
Whole Exome Sequencing (WES)	☐ Proband ☐ Duo ☐ Trio
Diagnostic Genome Sequencing (DGS)	☐ Proband ☐ Trio

NGS - Hereditary (Rare) Disease Panel							
☐	MLPA	☐	MS - MLPA	☐	CYP21A2	☐	SMN1, SMN2 del/dup

Other Genetic Test: Please note any previous genetic test results
(Ex: ACADS gene, negative)

1. I am aware a completed requisition form, and the consent of a physician is required in order to conduct a genetic test. 2. I acknowledge to have received and understood information about the purpose, scope, and limitations of the test. 3. I consent to personal information and specimen being transferred and processed for the performance of the requested test. 4. I understand genetic variants unrelated to the reason of the test may be found, and I wish to be informed of these incidental findings.	□ Yes

Date	D D / M M / Y Y Y Y	Name of Patient		Signature	

1. I confirm that the patient has given his/her consent for the provision of personal information and specimen for genetic testing. 2. I have explained the purpose, scope, and limitation of the test to the patient and have answered to all of his/her questions regarding the test.	□ Yes

Date	D D / M M / Y Y Y Y	Name of Physician		Signature	

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Clinical Patient Information

Please tick(V) relevant clinical symptoms (more than 5) as well as the degree of significance (+/++/+++).

Age of Manifestation : _____

GROWTH	+	++	+++	NEUROLOGIC	+	++	+++	ENDOCRINE	+	++	+++
Decreased body weight				Seizures				Hyperparathyroidism			
Failure to thrive				Spastic paraplegia				Hypothyroidism			
Feeding difficulties				Spasticity				**KIDNEY**	+	++	+++
Growth delay				Structural brain anomaly				Chronic kidney disease			
Obesity				**SKELETAL**	+	++	+++	Focal glomerulonephrosis			
Overgrowth				Arachnodactyly				Hydronephrosis			
Short stature				Arthrogryposis				Nephrolithiasis			
Tall stature				Brachydactyly				Nephrotic syndrome			
DEVELOPMENT	+	++	+++	Camptodactyly				Polycystic kidney dysplasia			
Developmental regression				Contracture				Proteinuria			
Learning disability				Osteopetrosis				Renal cyst			
Mental retardation				Polydactyly				Renal malformation ( )			
Motor delay				Recurrent fracture				**GENITOURINARY**	+	++	+++
Speech delay				Scoliosis				Abnormal hormone level ( )			
CRANIOFACIAL	+	++	+++	Skeletal dysplasia ( )				Ambiguous genitalia			
Blue sclerae				Syndactyly				Amenorrhea			
Cleft lip/palate				Vertebral anomaly ( )				Cryptorchidism			
Coarse facial features				**MUSCLE/JOINT**	+	++	+++	Delayed puberty			
Craniosynostosis				Hypotonia				Hypogonadism			
Depressed nasal bridge				Joint hypermobility				Hypospadias			
Downslanted palpebral fissures				Joint laxity				Precocious puberty			
Dysostosis				Multiple joint contractures				Premature ovarian failure			
Hirsutism				Muscle atrophy				**DERMATOLOGIC**	+	++	+++
Long philtrum				Muscle weakness				Abnormal blistering of the skin			
Low-set ears				Muscular dystrophy				Abnormality of the nail ( )			
Macrocephaly				Myopathy				Anhidrosis			
Macroglossia				Myotonia				Cafe-au-lait spot			
Microcephaly				Rhabdomyolysis				Hyperextensible skin			
Microdontia				Rigidity				Hyperpigmentation			
Micrognathia				**CARDIOVASCULAR**	+	++	+++	Hypertrichosis			
Midface retrusion				Abnormal heart morphology ( )				Hypopigmentation			
Short neck				Abnormal heart valves ( )				Hypotrichosis			
Others ( )				Aortic root dilatation				Ichthyrosis			
EYES	+	++	+++	Arrhythmia				Neurofibromatosis			
Anhidria				Atrial fibrillation				Sparse hair			
Cataract				Atrial septal defect				**HEMATOLOGIC**	+	++	+++
Coloboma				Bradycardia				Abnormal bleeding			
Cone-rod dystrophy				Brugada syndrome				Abnormal thrombosis			
Corneal dystrophy				Dilated cardiomyopathy				Abnormality of coagulation ( )			
Glaucoma				Hypertrophic cardiomyopathy				Anemia			
Microphthalmia				Long QT syndrome				Bone marrow failure			
Nystagmus				Vantricular septal defect				Neutropenia			
Opthalmoplegia				**RESPIRATORY**	+	++	+++	Pancytopenia			
Optic atrophy				Pulmonary hypertension				Thrombocytopenia			
Ptosis				Pulmonary hypoplasia				**METABOLIC**	+	++	+++
Retinal dystrophy				Recurrent upper respiratory tract infections				Abnormal newborn screen			
Retinitis pigmentosa				Respiratory insufficiency				Aminoacidopathies			
Strabismus				**GASTROINTESTINAL/LIVER**	+	++	+++	Carbohydrate disorders			
Visual impairment				Abnormality of intrahepatic bile duct ( )				Congenital disorders of glycosylation			
EAR	+	++	+++	Acute hepatitis				Fatty acid oxidation defects			
Abnormality of the ear ( )				Cholelithiasis				Hyperammonemia			
Hearing impairment				Cholestasis				Hypoglycemia			
NEUROLOGIC	+	++	+++	Diarrhea				Ketosis			
Amyotrophic lateral sclerosis				Hepatic cysts				Lactic acidosis			
Ataxia				Hepatic failure				Lysosomal storage disorders			
Autism				Hepatic fibrosis				Organic acidemias			
Behavioral abnormality ( )				Hepatomegaly				**IMMUNE**	+	++	+++
Chorea				Hirschsprung disease				Immunodeficiency			
Dementia				Inguinal hernia				Recurrent bacterial infections			
Dystonia				Jaundice				Recurrent fungal infections			
Encephalopathy				Pancreatitis				Recurrent viral infections			
Epilepsy				Splenomegaly				**OTHERS**	+	++	+++
Hypertonia				Umbilical hernia				Abnormal electrolyte level ( )			
Hypotonia				**ENDOCRINE**	+	++	+++	Cancer ( )			
Leukodystrophy				Adrenal hyperplasia				Hydrops			
Neuropathy				Diabetes mellitus				IUGR			
Parkinsonism				Dyslipidemia				Premature birth			
				Hyperinsulinemia							

Clinical Diagnosis / Genes for Test : Please note any genes of interest regarding clinical diagnosis or symptoms of the patient.

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Family History

Please tick(V) the appropriate boxes prior to test requisition.

1. Please tick if either parent shows similar clinical symptoms to that of the patient. YES (☐ Father ☐ Mother) ☐ NO

1-1. If YES, please write the clinical symptoms that apply.

2. Please tick if any siblings shows similar clinical symptoms to that of the patient. ☐ YES (Relationship:) ☐ NO

2-1. If YES, please write the clinical symptoms that apply.

Pedigree

I		☐ Male ◯ Female ◇ Sex Unknown ■ ● ◆ Affected ☐ ◯ ◇ Unaffected
II		
III		

Follow-up Family test (Exome sequencing)

☐	DES family test (Sanger)	☐	WES family test (Sanger)
☐	T-CNV (Targeted CNV detection)	☐	Familial mutation (Sanger-NGS panel family test)

Variant(s) Detected	By HGVS* Naming			(Ex: ACADS gene, c.312G>T)		
Family Info 1	Name		Relationship	☐ Father ☐ Mother ☐ Other ( )	Sex	☐ M ☐ F
	Date of Birth	D D / M M / Y Y Y Y	Sample Collection Date	D D / M M / Y Y Y Y	Disease related to patient symptoms	☐ No ☐ Yes
	Other Specifications					
Family Info 2	Name		Relationship	☐ Father ☐ Mother ☐ Other ( )	Sex	☐ M ☐ F
	Date of Birth	D D / M M / Y Y Y Y	Sample Collection Date	D D / M M / Y Y Y Y	Disease related to patient symptoms	☐ No ☐ Yes
	Other Specifications					
Family Info 3	Name		Relationship	☐ Father ☐ Mother ☐ Other ( )	Sex	☐ M ☐ F
	Date of Birth	D D / M M / Y Y Y Y	Sample Collection Date	D D / M M / Y Y Y Y	Disease related to patient symptoms	☐ No ☐ Yes
	Other Specifications					

*HGVS : Human Genome Variation Society