Hereditary(Rare) Diseases

Requisition and Consent Form

Barcode

*All required fields MUST be filled in.

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*	□ 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 WB 3.0 ml □ G-card (Blood paper)

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

NGS-Exome Sequencing			
Diagnostic Exome Sequencing (DES)	□ Proban	□ Duo	□ Trio
Whole Exome Sequencing (WES)	□ Proban	□ Duo	□ Trio
Diagnostic Genome Sequencing (DGS)	□ Proban		□ Trio
NGS - Hereditary(Rare) Disease Panel			
☐ CMA	☐ SMN1, SMN2 del/dup		

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

*Please note any genes of interest regarding clinical diagnosis or symptoms of the patient.
*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							

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		

* Family test is possible only when DES, WES, or DGS tests are requested.

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