SAPIENS

CLINICAL GENETIC TEST REQUEST FORM

PROTOCOL NO / BARCODE:

PATIENT:		**PHYSICIAN:**
Name and surname:		**Name and surname:**
Gender: Male ☐ Female ☐ **Date of Birth:**		**Phone number:** **E-mail:**
Mother name:	**Father name:**	**Institution:**
ID number: **E-mail:** **Phone number:** **Address:**		**Stamp:** **SAMPLE TAKEN AND DATE OF SAMPLE COLLECTION:** **Date of Receipt:** **Time:** Peripheral Blood ☐ Bone marrow ☐ Tissue ☐ Other ☐

CLINICAL INFORMATION:

(If necessary, use new paper or attach the epicrisis printout and other necessary documents to the request sheet)

IMPORTANT NOTE:

- Genetic tests are subject to consent. It is a legal requirement to obtain the consent of the patient and his/her guardian for those under 18 years of age.
- You can access the all forms at https://www.sapiensgenetics.com/guides-forms/ or request them from our center.
- It is recommended that genetic examinations be carried out with genetic counseling before and after the test. For this purpose, you can make an appointment at our center.

ÖZEL SB GENETİK HASTALIKLAR DEĞERLENDİRME MERKEZİ
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MOLECULAR GENETIC TESTS	
☐ WES Whole Exome Sequencing	☐Gilbert's disease UGT1A1 5' TA repeat count
☐ WES & Mitochondrial Disease Panel	☐5-ALFA Reductase Deletion Duplication Analysis
☐ WES DUO Whole Exome Sequencing Duo	☐MEN1 Gene Sequence Analysis
☐ WES TRIO Whole Exome Sequencing Trio	☐AMH Sequence Analysis
☐ WES TRIO PLUS Whole Exome Sequencing Trio Plus	☐AR (Androgen Receptor) Gene Analysis
☐ CES Clinical Exome Sequencing	☐Congenital Adrenal Hyperplasia (CYP21A2 gene)
☐ Hereditary Disease Panel	☐SOX9 mutations
☐ Familial Mediterranean Fever (FMF) MEFV Whole Gene Analysis	☐Silver Russel 11p15 hypomethylation analysis
☐ Factor II Prothrombin G20210A Mutation Analysis	☐MODY NGS Panel
☐ Factor V G1691A Mutation Analysis	☐GLUD1 Gene Sequence Analysis
☐ MTHFR C677T Mutation Analysis	☐Alpha-Thalassemia (HBA Gene) Mutation Screening
☐ Fragile X syndrome Analysis	☐Beta Thalassemia (HBB Gene) Sequence Analysis
☐ Y chromosome microdeletion analysis	☐Alpha-Thalassemia Deletion/Duplication analysis
☐ F13 V34L Polymorphism Analysis	☐Bernard Soulier Syndrome Type A (GP1BA Gene Sequence Analysis)
☐ FV Cambridge Mutation Analysis	☐Bernard Soulier Syndrome Type B (GP1BB Gene Sequence Analysis)
☐ MTHFR A1298C Mutation Analysis	☐Bernard Soulier Syndrome Type C (GP9 Gene Sequence Analysis)
☐ Noonan Syndrome - Rasopathy Panel	☐HLA-B27
☐ Mitochondrial Disease Panel	☐HLA-B51 (PCR)
☐ Coffin Lowry Syndrome (RSK2 Gene)	☐Achondroplasia FGFR3 sequence analysis
☐ DMD (Duchenne Muscular Dystrophy)	☐Marfan (FBN1 Gene) Sequence Analysis
☐ Lowe Syndrome (OCRL1 Gene) Sequence Analysis	☐Osteogenesis Imperfecta Panel
☐ Parkinson type 9, Kufor Rakeb Disease	☐Cystic Fibrosis CFTR Whole Gene Sequence Analysis
☐ Kennedy Disease Genetic Testing	☐CIAS1 Sequence Analysis
☐ LHON Gene Sequence Analysis	☐FGFR1 sequence analysis
☐ SOS1 Gene Sequence Analysis	☐FGFR2 sequence analysis
☐ TUBB3 Mutation Analysis	☐Fucosidosis (FUCA1) Gene Sequence Analysis
☐ Spinal Muscular Atrophy Screening	☐Kartagener (CILD1 Sequence Analysis)
☐ Spinocerebellar ataxia (SCA) type1	☐Multiple Endocrine Neoplasia Type2A RET Gene Analysis
☐ Spinocerebellar ataxia (SCA) type 2	☐HLA-B5701
☐ Spinocerebellar ataxia (SCA) type3	☐Nijmegen Syndrome Del657 Analysis
☐ Spinocerebellar ataxia (SCA) type6	☐TWIST1 Gene Sequence Analysis
☐ Spinocerebellar ataxia (SCA) type7	☐Known Single Gene Point Mutation Analysis
☐ Spinocerebellar ataxia (SCA) type 8	☐Interferon Beta-1 (IFNB1) Gene Mutation screening with sequence analysis
☐ Spinocerebellar ataxia (SCA) type 10	☐PRG4 gene sequence analysis
☐ Spinocerebellar ataxia (SCA) type 12	☐Familial Mediterranean Fever (FMF) Frequent Mutation Analysis
☐ Spinocerebellar ataxia (SCA) type 17	☐Silver Russell matUPD7

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☐ Myotonic Dystrophy Triple Repeat Analysis	☐RHD genotype determination in peripheral blood
☐ LIMB GIRDLE Sarcoglycanopathy Deletion Screening-1	☐RET Gene Exon 8, 10, 11, 13, 14, 15 and 16 Sequence Analysis
☐ LIMB GIRDLE Sarcoglycanopathy Deletion Screening-2	☐Rubinstein Taybi Syndrome full genome sequence analysis
☐ LIMB GIRDLE Sarcoglycanopathy Deletion Screening-3	☐Sotos NSD1 gene sequence analysis
☐ LIMB GIRDLE Sarcoglycanopathy Deletion Screening-4	☐SALL4 gene analysis
☐ Mitochondrial DNA deletion Screening-1	☐WISKOTT-ALDRICH Syndrome (WAS Gene) Sequence Analysis
☐ Mitochondrial DNA deletion Screening-2	☐ACEI/D polymorphism
☐ Mitochondrial DNA deletion Screening-3	☐Alpha-1 antitrypsin genotype determination (M, S, Z allele)
☐ Spinal Muscular Atrophy SMN1-2 Gene deletion duplication analysis	☐CHN1 gene analysis
☐ Charcot Marie Tooth 1A (PMP22)	☐HOXA1 mutations
☐ Huntington Mutation Analysis	☐KIF2A gene analysis
☐ PTEN Deletion Duplication Analysis	☐MFRP gene mutation
☐ RETT Syndrome (MECP2) Deletion Analysis	☐Pompe Disease Sequence Analysis
☐ DMD Whole Gene Deletion Duplication Screening/Carrier Test Screening	☐Thrombophilia Panel of 2 (FII, FV)
☐ FOXG1 Syndrome	☐CYP2D6 Mutation Analysis
☐Selectin S128R Polymorphism Analysis	

MOLECULAR GENETIC TESTS	
☐ APO E Genotyping	☐ Plasminogen Activator Inhibitor-1 Polymorphism (PAI)
☐ Plavix Activity (CYP2C19)	☐ Warfarin Drug Resistance
☐ GALT Deletion Duplication Analysis	☐ Thrombophilia - Cardiovascular Risk Panel -12 parameters.
☐ Fanconi Bickel Syndrome (SLC2A2 gene)	☐ ACTN3 R577X mutation analysis
☐ Phenylketonuria (PAH) Gene Sequence Analysis	☐ PON1 Polymorphism Analysis (163T>A, 575A>G)
☐ Galactosemia (GALT gene) Sequence Analysis	☐ Friedreich Ataxia Mutation Analysis
☐ Gaucher (GBA) Gene Sequence Analysis	☐ CYP21A2 Deletion Duplication Analysis
☐ Matachromatic Leukodystrophy (ARSA1 gene)	☐ GCH1 sequence analysis
☐ Smith-Lemli-Opitz Syndrome	☐ Hereditary Fructose Intolerance Test
☐ Tay-Sacs Disease Genetic Test (HEXA)	☐ osteochondrodysplasia
☐ Phenylketonuria mutation screening	☐ SURF1 sequence analysis
☐ Glutaric Acidemia Type I Genetic Testing	☐ Wolman (LIPA) Gene Sequence Analysis
☐ MONSTER DISEASE genetic analysis (ASPA gene)	☐ AVPR2 Gene Sequence Analysis
☐ Fabry Syndrome (GLA) Sequence Analysis	☐ HUNTER IDS gene sequence analysis
☐ Hemochromatosis (HFE 2 Mutation)	☐ PAX6 gene analysis
☐ Pompe Disease Sequence Analysis	☐ PROP1 Sequence Analysis
☐ Purine Nucleoside Phosphorylase Insufficiency	☐ CONNEXIN26 mutations
☐ SCA Panel Type 1-2-3-6-7	☐ MC4R Gene Sequence Analysis
☐ HbS E6V Mutation Analysis	☐ MYOC Gene Sequence Analysis
☐ Alzheimer's disease APP exon 16 and 17 genetic analysis, Alzheimer's Type 1	☐ Norrie's Disease

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MOLECULAR CYTOGENTIC - FISH TESTS	
☐ Prader-Willi Syndrome (FISH)	☐ Single Chromosome Painting Fish
☐ Miller-Dieker Syndrome (Fish)	☐ Single Chromosome Subtelomeric Fish
☐ Isolated Lissencephaly Sequence (FISH)	☐ Velocardiofacial Syndrome (FISH)
☐ Digeorge Syndrome (FISH)	☐ Williams Syndrome (FISH)
☐ Cri Du Chat Syndrome (FISH)	☐ Wolf-Hirschhorn Syndrome (FISH)
☐ Angelman Syndrome (FISH)	☐ Gender Determination XY Chromosome (FISH)
☐ Rubinstein Taybi Syndrome (Del 16p13.3) FISH	☐ Kallman's Syndrome (FISH)
☐ Prader-Willi Syndrome(FISH)	☐ Determination of SRY (FISH)
☐ SHOX Deletion (FISH)	☐ XIST Gene Deletion (FISH)
☐ Smith-Magenis Syndrome (Fish)	☐ Aneuploidy Screening (Rapid Fish)
☐ Sotos Syndrome (Fish)	☐ Subtelomeric Scanning (Fish)
☐ Steroid Sulfatase (STS) Deficiency (FISH)	
MOLECULAR CYTOGENETICS - MICROARRAY TESTS	
☐ Chromosomal Microarray - SNP - 300K	☐ Chromosomal Microarray - 850K
CYTOGENETICS - CHROMOSOME TESTS	
☐ Chromosome analysis from skin biopsy material	☐ Chromosome analysis in peripheral blood
☐ SCE Test	☐

***For tests and research projects not listed here, please contact us.**
international@sapiens.com.tr

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Clinical Information, Family History, Examination History (It will be useful to mark compatible findings in the Phenotype Information section.)

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PHENOTYPE INFORMATION

Perinatal History	Growth	Behavioral/Psychiatric	Cognitive/Developmental
☐Prematurity ☐Increased Nt/Cystic Hygroma ☐Iugr ☐Oligohydramnios ☐Polyhydramnios	☐Asymmetric Growth ☐Failure To Thrive ☐Obesity ☐Overgrowth ☐Short Stature ☐Tall Stature	☐Adhd ☐Autism Spectrum Disorder ☐Oppositional-Defiant Disorder ☐Obsessive-Compulsive Disorder ☐Psychiatric Diagnosis	☐Intellectual Disability/Mr. ☐Mild ☐Moderate ☐Severe ☐Engine Delay ☐Speech Delay ☐Developmental Regression
Craniofacial	**Gastrointestinal**	**Genitourinary**	**Skin/Hair/Dental**
☐Cleft Lip ☐Cleft Palate ☐Craniosynostosis ☐Dysmorphic Features ☐Ear Malformation ☐Macrocephaly ☐Microcephaly ☐Synophrys	☐Anal Atresia ☐Gastroschisis ☐Hirschsprung Disease ☐Liver Failure ☐Omphalocele ☐Pyloric Stenosis ☐Tracheoesophageal Fistula	☐Ambiguous Genitalia ☐Clitoromegaly ☐Cryptorchidism ☐Hydronephrosis ☐Hypogonadism ☐Hypospadias ☐Nephrotic Syndrome ☐Renal Agenesis ☐Renal Malformation ☐Renal Tubulopathy	☐Abnormal Fingernails ☐Abnormal Hair ☐Abnormal Skin ☐Dental Anomalies ☐Hemangioma ☐Hyperpigmentation ☐Hypopigmentation
Neuromuscular	**Musculoskeletal**	**Ophthalmologic**	**Cardiovascular**
☐Ataxia ☐Autonomic Dysfunction ☐Cerebral Palsy ☐Dementia ☐Dystonia ☐Encephalopathy ☐Hypotonia ☐Muscle Weakness ☐Peripheral Neuropathy ☐Seizures ☐Spasticity Stroke/Tias ☐Structural Brain Anomaly	☐Club Foot ☐Contractures ☐Diaphragmatic Hernia ☐Foot Deformity ☐Joint Laxity ☐Limb Anomaly ☐Oligodactyly ☐Polydactyly ☐Scoliosis ☐Skeletal Dysplasia ☐Syndactyly ☐Vertebral Anomaly	☐Aniridia ☐Blindness ☐Cataracts ☐Coloboma ☐Microphthalmia ☐Myopia ☐Ophthalmoplegia ☐Optic Atrophy ☐Ptosis ☐Retinitis Pigmentosa	☐Aortic Dilatation/Dissection ☐Arrhythmia ☐Arterial Dilatation/Dissection ☐Atrial Septal Defect ☐Av Canal Defect ☐Bicuspid Aortic Valve ☐Coarctation Of The Aorta ☐Cardiomyopathy ☐Hypoplastic Left Heart ☐Pulmonic Stenosis ☐Tetralogy of Fallot ☐Ventricular Septal Defect
Hearing	**Hematologic/Immunologic**	**Endocrine**	**Metabolic/Mitochondrial**
☐Sensorineural Hearing Loss ☐Conductive Hearing Loss ☐Mixed Hearing Loss	☐Anemia ☐Immunodeficiency ☐Iron Deficiency ☐Neutropenia ☐Pancytopenia ☐Thrombocytopenia	☐Adrenal Abnormality ☐Diabetes, Type I ☐Diabetes, Type II ☐Gonadal Abnormality ☐Hypothalamic Abnormality ☐Parathyroid Abnormality ☐Pituitary Abnormality ☐Thyroid Abnormality	☐Abnormal Cpk ☐Abnormal Plasma Carnitine/Acylcarnitine ☐Elevated Pyruvate ☐Elevated Alanine ☐Hypoglycemia ☐Ketosis ☐Lactic Acidosis ☐Organic Aciduria ☐Ragged Red Fibers

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