intelliGene

A healthy life by Nesiller...

WGS / WES / CES
Request Consent Form

Patient Information

Name & Surname _____

Gender ☐ Female ☐ Male

Birth Date _____

Address _____

_____

Phone Number _____

E-mail _____

Physician Information

Name & Surname _____

Organization _____

Department _____

Address _____

_____

Phone Number _____

E-mail _____

Requested Test ☐ Whole Genome Sequencing (WGS) ☐ Whole Exome Sequencing(WES) ☐ Clinical Exome Sequencing (CES)

Type of Study

Solo Analysis ☐

Duo Analysis ☐ Fill in the fields below for family members

Trio Analysis ☐ Fill in the fields below for family members

Sample Information

Sample Collection Date/Time_____

Sample Collection Place _____

Sample Type ☐ EDTA Blood(2x4 ml) ☐ DNA (3-5 µg) ☐ Other.....

Family Members' Information	Birth Date	Health Condition
Father's Name & Surname		
Mother's Name & Surname		
Sibling 1		
Sibling 2		

Clinical Indication / Findings: Fill out for each patient

Age of Onset of Findings _____

Are the mother and father blood relatives? ☐ Yes ☐ No

If related, what is the degree of relation?_____

Specify if there are any other affected relatives_____

Suspected Diseases

Suspected inheritance model

☐ Autosomal recessive ☐ Affected proband

☐ Autosomal dominant ☐ Unaffected proband

☐ X-linked

☐ Unknown

Previous genetic test results, if available, should be provided

Transplantation (Bone marrow, tissue, stem cells) ☐ Yes (Please specify) ☐ No

ÖZEL nesiller GENETİK HASTALIKLAR DEĞERLENDİRME MERKEZİ

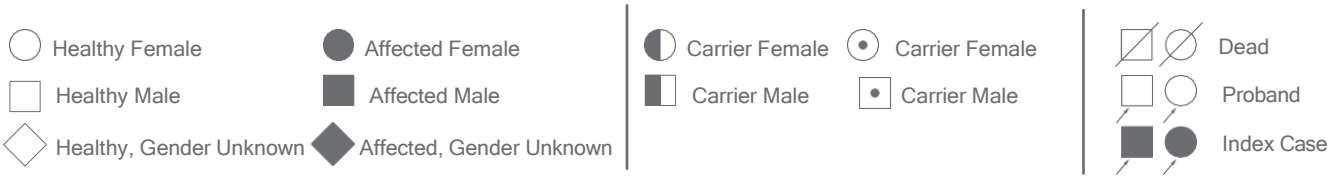
intelliGene

A healthy life by Nesiller...

WGS / WES / CES
Request Consent Form

Family Tree (Please fill out if a family history is available)

Pedigree Symbols



ÖZEL nesiller
GENETİK HASTALIKLAR DEĞERLENDİRME MERKEZİ

intelliGene

A healthy life by Nesiller...

WGS / WES / CES
Request Consent Form

Clinical Findings

NEUROLOGY

Behavioral Disorders
- ☐ Autism
- ☐ Attention Deficit
- ☐ Psychiatric Disorders

Brain Imaging
- ☐ Cortical Developmental Disorder
- ☐ Myelination Abnormality
- ☐ Corpus Callosum Agenesis
- ☐ Brain Atrophy
- ☐ Cerebellar Hypoplasia
- ☐ Heterotopia
- ☐ Holoprosencephaly
- ☐ Hydrocephalus
- ☐ Leukodystrophy
- ☐ Lissencephaly

Movement Disorders
- ☐ Ataxia
- ☐ Chorea
- ☐ Dystonia
- ☐ Parkinsonism

Neuromuscular Disorders
- ☐ Hyperreflexia
- ☐ Muscle Hypertonia
- ☐ Muscle Hypotonia
- ☐ Spasticity

Seizure
- ☐ Febrile Seizure
- ☐ Fokal Seizure
- ☐ Generalized Seizures

Other
- ☐ Craniosynostosis
- ☐ Dementia
- ☐ Encephalopathy
- ☐ Macrocephaly
- ☐ Microcephaly
- ☐ Stroke
- ☐ Intellectual Disability
- ☐ Developmental Delay

DERMATOLOGY AND MUSCULOSKELETAL

Skeleton
- ☐ Limb Disorders
- ☐ Spine Disorders
- ☐ Skeletal System Disorders
- ☐ Joint Hypermobility
- ☐ Multiple Joint Contractures
- ☐ Polydactyly
- ☐ Scoliosis
- ☐ Syndactyly
- ☐ Bunion / Clubfoot

Skin
- ☐ Nail Disorders
- ☐ Skin Pigmentation Disorders
- ☐ Hyperextensive Skin
- ☐ Ichthyosis

MOUTH AND EAR
- ☐ Cleft Lip
- ☐ Cleft Palate
- ☐ Conductive Hearing Loss
- ☐ External Ear Malformations
- ☐ Hypodontia
- ☐ Sensorineural Hearing Loss

GÖZ
- ☐ Blepharospasm
- ☐ Cataract
- ☐ Coloboma
- ☐ Glaucoma
- ☐ Microphthalmia
- ☐ Nystagmus
- ☐ Ophthalmoplegia
- ☐ Optic Atrophy
- ☐ Ptosis
- ☐ Retinitis Pigmentosa
- ☐ Retinoblastoma
- ☐ Strabismus
- ☐ Visual Impairment

METABOLISM
- ☐ Abnormal Creatine Kinase
- ☐ Decreased Serum Carnitine
- ☐ Hyperalaninemia
- ☐ Hypoglycemia
- ☐ Increased CSF Lactate Level
- ☐ Ketosis
- ☐ Lactic Acidosis
- ☐ Organic Acidemia

ENDOCRİNE
- ☐ Diabetes
- ☐ Hyperparathyroidism
- ☐ Hyperthyroidism
- ☐ Hypoparathyroidism
- ☐ Hypothyroidism

CARDIOVASCULAR
- ☐ Angioedema
- ☐ Aort Dilatation
- ☐ Arrhythmia
- ☐ Atrial Septal Defect
- ☐ Aortic Coarctation
- ☐ Dilated Cardiomyopathy
- ☐ Hypertension
- ☐ Hypertrophic Cardiomyopathy
- ☐ Hypotension
- ☐ Lymphedema
- ☐ Heart Malformations
- ☐ Myocardial Infarction
- ☐ Stroke
- ☐ Fallot's Tetralogy
- ☐ Vasculitis
- ☐ Ventricular Septal Defect

REPRODUCTIVE
- ☐ Genital Organ Anomaly
- ☐ Abnormal Internal Genitalia
- ☐ Hypogonadism
- ☐ Hypospadias
- ☐ Infertility

PRENATAL AND DEVELOPMENTAL
- ☐ Facial Anomaly
- ☐ Growth Tension
- ☐ Hemihypertrophy
- ☐ Hydrops Fetalis
- ☐ Intrauterine Growth Retardation
- ☐ Oligohydramnios
- ☐ Excessive Growth
- ☐ Polyhydramnios
- ☐ Preterm Birth
- ☐ Short Stature
- ☐ Height Length

HEMATOLOGY
- ☐ Hemoglobin Anomaly
- ☐ Coagulation Anomaly
- ☐ Anemia
- ☐ Neutropenia
- ☐ Pancytopenia
- ☐ Splenomegaly
- ☐ Thrombocytopenia

URINARY
- ☐ Renal Morphology Anomaly
- ☐ Urinary System Anomaly
- ☐ Hydronephrosis
- ☐ Renal Agenesis
- ☐ Renal Cyst
- ☐ Renal Tubular Function Disorder

ONCOLOGY
- ☐ Adenomatous Polyp
- ☐ Breast Carcinoma
- ☐ Colorectal Carcinoma
- ☐ Leukemia
- ☐ Myelofibrosis
- ☐ Lung Neoplasm
- ☐ Skin Neoplasm
- ☐ Skin Neoplasm
- ☐ Paraganglioma
- ☐ Pheochromocytoma

Notes:

ÖZEL nesiller
GENETİK HASTALIKLAR DEĞERLENDİRME MERKEZİ

intelliGene

A healthy life by Nesiller...

WGS / WES / CES
Request Consent Form

Yapılacak Test ile ilgili Bilgiler

What is WGS / WES / CES analysis?

The genome refers to the complete hereditary material (DNA) of an organism. Exome, on the other hand, is a term used to define all the protein-coding sequences within the genome, specifically the exons. The exome consists of the DNA sequences of genes that code for proteins necessary for the proper functioning of the body. The regions outside the exome play a role in regulating gene activity. Changes in the DNA sequence c an lead to diseases. It is known that most disease-causing DNA mutations occur in protein-coding regions, i.e., exons. However, mutations outside these regions can also cause diseases. Whole Genome Sequencing (WGS) identifies changes across the entire genome, while Whole Exome Sequencing (WES) identifies changes in all protein-coding regions. Clinical Exome Sequencing (CES) identifies changes in protein-coding exons of more than 4,900 genes that are clinically associated in the literature.

Result Reports: In WGS / WES / CES analysis, your results are compared and matched with the reference human genome. However, it is important to note that a definitive diagnosis may not always be reached based on the obtained data. At our genetic diagnostic center, accr edited with ISO 15189 Medical Laboratory Accreditation, the analysis results are carefully evaluated, with data analysis being supplemented by clinical and bioinformatics databases. The results of WGS / WES / CES analysis can be concluded in three possible ways:

Positive: A positive result indicates that disease-causing variant(s) associated with the patient's clinical symptoms have been detected, based on current scientific data.

Negative: A negative result indicates that no disease-causing variant(s) associated with the patient's clinical symptoms have been identified, according to current scientific data. A negative result does not imply that there is no genetic cause for the patient's symptoms.

Uncertain: An uncertain result indicates that variant(s) with unclear clinical significance (VUS) that could potentially be associated with the patient's symptoms have been identified. There is insufficient scientific and medical information to determine whether these variants cause disease. To better understand the impact of an uncertain variant, testing of family members may be necessary, followed by re-analysis with updated scientific literature. The responsibility for requesting new tests (such as for the parents or other family members) and analysis lies with the treating clinician and requires separate approval. Re-evaluation of the data may incur additional costs.

Incidental Findings: In WGS / WES / CES testing, multiple genes are analyzed simultaneously, which may result in the identification of genetic changes that are unrelated to the patient's current symptoms and signs. These types of changes are referred to as incidental findings. These findings can have significant implications for the health of the patient and their family members. The American College of Medical Genetics and Genomics (ACMG) recommends that laboratories report such findings detected in all genes tested. These genes may be associated with certain cancer syndromes, specific types of heart diseases, high cholesterol, and susceptibility to complications from anesthesia. Incidental findings are only investigated and reported with the patient's consent. The analysis of incidental findings identified in index cases in family members requires separate approval.

Use of Parental Samples in WGS / WES / CES Analysis – Limitations of Tests:

WGS / WES / CES analyses may not fully cover all targeted regions due to current technical limitations, and pathogenic variants may not be completely excluded. Additionally, these tests do not analyze every type of genetic change. Therefore, it is possible that the test may not detect the responsible genetic change for the patient's clinical symptoms. The interpretation of results is based on current genetic data, and future analyses with updated information may lead to changes in the results. Accurate clinical information and family history are crucial for correctly interpreting the results of WGS / WES / CES tests. If the provided information is incorrect and/or incomplete, misinterpretation of results may occur. It should be noted that the genetic test is only for the disease/indication specified by your doctor in this form, and the analysis and reporting are done to explain the reason for referral. Test results may indicate the need for additional tests. The genetic test involves a series of laboratory procedures, which may lead to false-positive or false-negative results due to simple errors such as labeling mistakes or clerical errors. It is the doctor's responsibility, in the context of clinical evaluation, to request re-analysis or a repeat test if there is a discrepancy between the report and the clinic, and this is important.

WGS / WES / CES Consent Section

In order to legally carry out genetic analyses, our Genetic Diagnostic Center requires your consent. Written consent must be obtained from the patient or their parent/legal representative, and the form must be signed for the WGS / WES / CES analysis. This form becomes valid once signed by the patient and indicates that the patient acknowledges and approves the content of this document. The form is then delivered after being sig ned by the physician. Your doctor has recommended performing the WGS / WES / CES analysis to clarify the diagnosis/symptoms indicated in this form for you (or for someone under your care or supervision).

Test to be Conducted: WGS / WES / CES Analysis

Sample Type: The genetic test aims to investigate hereditary material (DNA). The sample used for this purpose is typically a peripheral blood sample, which can be obtained by drawing 5-10 ml of blood from the veins in the arm using a needle. Known health risks are generally minimal and may include slight bruising, mild pain, and very rarely, the risk of infection or nerve-vessel damage due to the traumatic effects of the needle. If the DNA sample is insufficient or if confirmation of the diagnosis requires additional testing, a new sample may be necessary. If sufficient DNA products cannot be obtained or the desired quality is not reached, a new sample may be requested.

Turnaround Time: The estimated turnaround time for test results, as provided on our website or by your referring doctor or staff, represents the average time under normal circumstances. However, due to patient-specific variations, the need for further investigation, or collaboration with other laboratories (domestic or international), the average time may be exceeded.

intelliGene

A healthy life by Nesiller...

WGS / WES / CES
Request Consent Form

Storage Consent

Your remaining blood sample will be stored during the legal process and will be discarded afterward. The obtained DNA sample and data will be kept for 5 years. The DNA sample may be used for future genetic testing if needed. The data obtained will also be important for clinical follow-up.

For example, without the need for a new sample or test, the analysis can be repeated or expanded using the existing data.

☐ I accept

☐ I do not consent. Once all legal obligations have been fulfilled, I request that my samples be properly disposed of.

Usage Consent

Test results serve as a crucial resource for healthcare professionals, researchers, and scientists in the investigation of genetic disorders, as well as in the advancement of diagnostic and therapeutic strategies. In this context, personal data will be anonymized and/or encrypted. You may request the destruction of non-anonymous test data at any time. Following anonymization, the rights to the data and materials will be held by the Nesiller Genetic Center. I hereby provide my consent for my test results to be stored and utilized in the database for scientific purposes, including the enhancement and facilitation of disease identification and the provision of statistical data

☐ I accept

☐ I do not consent. My samples should only be stored for the purpose of additional studies that I have approved.

Incidental Findings: I consent to the communication and/or reporting of incidental findings unrelated to my condition, as identified by my doctor, to me or my doctor. (Incidental findings are not examined in prenatal samples.)

☐ I accept. ☐ I do not accept

Report Delivery Preference: All genetic data is personal and cannot be shared with third parties. At our center, only the physician who requested the test receives an email with preliminary information. With your consent, your final report can be delivered to the requesting physician, the physician(s) who are clinically following you, or to you or another person you authorize. To do so, you must indicate your authorization request in writing.

Authorization for Receiving Information Related to the Results: Considering the new gene and variant information that will be added to the literature, I authorize my data to be re-analyzed and for me to be contacted for genetic counseling

☐ Yes, I would like my data to be re-evaluated every 6 months

☐ Yes, I would like my data to be re-evaluated once a year.

☐ No, I do not want to receive counseling services

[1] A discounted price is applied for genetic counseling sessions. Please inquire for more information.

Patient or Legal Representative's Consent ☐ **I confirm that I have read and understood**

I have received information about the WGS / WES / CES analysis test, including a written explanation, which I have read and understood. I have been fully informed about the technical aspects and limitations of the analysis. I understand that there may be false positive/negative results, and that retesting and/or re-analysis may be required. I also understand that there may be a need for re-sampling or additional samples. I was given enough time to ask questions and make a decision, and all medical terms were explained to me. I am aware that I can request further information at any time.

I acknowledge that I am responsible for matters related to genetic diagnosis. I confirm that I am accepting the genetic diagnosis freely, without any threats or undue pressure, and I consent to the 'WGS / WES / CES analysis' being performed on myself and/or my unborn child

	Patient / Parent	Responsible Physician
Name & Surname	_____	_____
Date	_____	_____
Signature		