

GENETIC TEST INFORMATION FORM

The purpose of this informative text is: (To be marked by the physician and the following blank filled in.)

- ☐ Long Read Sequencing (LRS) ☐ WES ☐ WGS ☐ Molecular genetic test ☐ Pregnarisk (Panel Test) ☐ Cytogenetic test
☐ Other

To inform you about the test to be conducted within the scope of It is an obligation for us and a right for you to provide this information and obtain your consent before offering health services. Therefore, we kindly ask you to read the text carefully, fill in the blanks, and communicate your questions before or after the test.

1- What Are Genetic Tests, When Are They Conducted?

LRS (Long-Read Whole Genome Sequencing): Short read tests like WES and WGS may not fully detect large changes, repeated regions, or complex variants in DNA. Besides its success in diagnosing these changes, this new technology also contributes to diagnosis by identifying some epigenetic diseases and methylation disorders. In other words, alongside the data obtained by short-read whole genomes, it can elucidate chromosomal changes (such as translocation, inversion, deletion), repeat sequence diseases, accurate interpretation of HLAs, and all situations requiring haplotype analysis (i.e., separately visualizing the genes inherited from our mother and father). Therefore, "Long-read whole genome" has started to be used for patients who cannot be diagnosed with other methods.

WES (Whole Exome Sequencing): It involves examining the coding regions of about 20,000 genes. It is conducted to perform a general screening in undiagnosed diseases, to screen genes collectively if multiple genes are involved in diagnosed diseases, and sometimes to understand whether there are additional diseases with clinical outcomes alongside the main disease. It can also be done to determine disease carrierships without having a disease.

WGS (Whole Genome Sequencing): Conducted for the same purposes as WES, but it is a more comprehensive test as it includes coding and non-coding parts of genes and significant portions of non-genic regions, and its diagnosis rate is higher for some disease groups.

Molecular genetic test: If there is a specific preliminary diagnosis, it is conducted to investigate the gene or genes causing that disease and to detect the genetic change causing the disease. The same gene can be investigated with one or multiple methods, and it may be necessary to switch from one to another during the study. Sometimes, it may be necessary to take samples from other family members to interpret the result. This situation can only be decided while the study is ongoing.

Cytogenetic test: Chromosomes, which are the packaged form of genes and DNA, are examined. It can be studied from blood, bone marrow, and miscarriage material. Additionally, studies can be conducted from the water taken from the womb during pregnancy, a piece of the placenta, and rarely from the baby's blood. Results are usually provided within the specified time frames, but rarely, additional material may be needed. Sometimes, additional tests outside the specified ones may be required to elucidate the change according to the result. This situation can only be decided while the study is ongoing. If such a need arises during your test, you will be informed.

Pregnarisk (PANEL TEST): Recommended for couples planning to marry and have children and for couples planning preimplantation genetic diagnosis with IVF. It is a panel test that examines the loss, gain, and repeat number in certain genes along with exome sequencing and chromosome analysis of partners. Families and babies at risk of having issues during pregnancy are identified, and results are reported in detail. Some changes of "unknown clinical significance" according to literature data are also reported in these studies. In this case, the change and risks are explained in detail in the report.

Other (Please write detailed information about the test below.):

2- What Do Test Results Mean, Is There a Margin of Error, Can It Be Considered Completely Reliable?

If the test results are consistent with clinical findings, they support the diagnosis or reduce the likelihood of some diagnoses. In other words, it is to support the clinical diagnosis. The ultimate clinical evaluation is essential. Clinical evaluation can be done by a genetic specialist or a specialist physician in other fields or a general practitioner, or together by these physicians, depending on the nature of the disease. Therefore, we may need to have consultations with your doctors during your studies. Like every test in the medical field, genetic tests have a margin of error, and the results are not definitive. The margin of error varies depending on the nature of the disease, the resolution of the test, the quality of the material, the correct and sufficient flow of information, and many similar reasons.

3- What Are Other Information I Need to Know?

1. The time given for the tests is approximate; results may be available in a shorter or longer time.
2. Your increased DNA samples after the test may be stored indefinitely for test validation or educational purposes. You can request the destruction of your blood and DNA samples by contacting the laboratory after the completion of the requested test above. You can also indicate this in the approval section of this form. Not allowing your sample to be used for test validation or educational purposes does not affect your test result.
3. Sometimes, the changes found in the test result may need to be supported with additional "diagnostic tests." In this case, you will be contacted and informed about these tests. If you do not have the recommended additional tests conducted, the success of the diagnosis may decrease. Additional payments may be required for those tests with high costs.

4. If deemed medically necessary, some or all of the tests may be sent to another center.
5. Rarely, you may need to send or provide a second material/sample. If results cannot be obtained, the test may be repeated.
6. If your sample is not given at our center and arrives by cargo or courier, you will be notified by message that the sample has reached our center. Payment information is also provided in the message. After making the payment to the account details provided to you and sharing the receipt, your processes will begin. Meanwhile, sample security is ensured at our center.
7. If you decide to cancel the test after making the payment, we may refund the remaining amount except for the expenses incurred up to that point, but if a significant part of the test has been completed, a refund is not possible.
8. Some data obtained from tests may not be reported for scientific or ethical reasons.
9. Your test results will be stored in Intergen records.
10. If your material was not taken by Intergen, Intergen is not responsible for whether the material was taken correctly, its suitability, or information provided at other centers.
11. If our center's physicians determine that another test is more suitable for diagnosis than the test requested by your physician, information will be provided to you and/or your physician to protect your benefit as a patient and to prevent you from having to provide a new sample, and the test deemed appropriate will be recommended. Our center reserves the right to refuse tests that are not medically appropriate.
12. Upon request, your test result can be sent to you via communication methods like WhatsApp, Gmail, Hotmail, etc. However, please be informed that these applications are of foreign origin, meaning the message sent reaches the servers of companies based abroad, i.e., it is transferred abroad. The confidentiality of the data ensured by the application owner companies is not under our control, and you can access their privacy policies on their websites.
13. As a licensed healthcare institution, we need to obtain identity, contact, health, and genetic data to perform genetic tests. We protect and ensure the confidentiality of this data due to our obligation to maintain confidentiality. Unless there is consent or a legal obligation, we do not share your data with third parties/institutions. If you consent, we may use your personal data for scientific studies we conduct, provided that it is anonymized and your right to privacy and personal rights are protected. Pursuant to Article 11 of the Personal Data Protection Law No. 6698, you have the right to: learn whether your personal data is processed, request information if your personal data has been processed, learn the purpose of processing your personal data and whether they are used in accordance with this purpose, know the third parties to whom your personal data is transferred at home or abroad, request correction if your personal data is processed incompletely or inaccurately, request the deletion or destruction of your personal data within the conditions set forth in Article 7 of the Law, request notification of the actions taken in accordance with subparagraphs (d) and (e) to third parties to whom your personal data have been transferred, object to a result arising against you by analyzing the processed data exclusively through automated systems, and request compensation for damage arising from the unlawful processing of your personal data. We will respond to your applications within 30 days.
14. In the event of the laboratory's closure, acquisition, or merger, the continuous availability and integrity of stored patient samples and records will be ensured in accordance with the Sample Storage Durations List and the Record Storage Durations List in compliance with legal regulations and the practices of the Ministry of Health of the Republic of Turkey. In the event of the closure, acquisition, or merger of our center, written or verbal information will be provided to patients/institutions/physicians. Subsequently, the secure transfer and storage of samples and records will be ensured by us.

CONSENT FORMS

1- Genetic Test Consent Form (To be completed by the patient.)

"I have read the information form about the genetic test. I have been verbally informed about the test by my physician, my questions have been answered, and I have been given a reasonable time to read the written information. I understand the purpose, results, reliability, reporting time, and processing of my personal data."

Dear Patient; if you consent to the genetic test on your material under the conditions specified in the information form, please write

"I CONSENT TO THE TEST" in the space below:

.....
(Any intervention in the health field can only be carried out after obtaining informed consent, so if you do not consent, we cannot perform the test.)

2- Additional Test Consent Form If Medically Necessary (To be completed by the patient.)

Dear Patient; as explained in the information form, additional tests that may assist in diagnosis can be performed or another test may be evaluated during the process if medically necessary for a diagnosis in genetic studies. Additionally, secondary findings that may emerge as a result of the tests besides the findings that led you to apply will be reported if deemed medically necessary. If you accept this, please check "I agree" from the boxes below. If you do not accept additional tests, please check "I do not agree" from the boxes below. We would like to note that not undergoing the recommended additional tests may decrease diagnostic success. If additional tests are chargeable, our team will inform you.

☐ I agree. ☐ I do not agree.

3- Scientific Research Consent Form (To be filled by the patient.)

Dear Patient; As the Intergen Genetic Disorders Evaluation Center, we can conduct scientific studies by anonymizing the health and genetic data we hold to protect and improve public health. We take all necessary administrative and technical measures to ensure the privacy of personal data and confidentiality of private life in these studies. Under these conditions, please indicate whether you consent to the use of the data you share with us in scientific studies by checking one of the boxes below:

- ☐ I consent.
☐ I do not consent.

4- Consent Form for Sending Test Results (To be filled by the patient.)

Dear Patient; Please check the box for the method you want us to use to deliver your test results to you. Notify us as soon as possible if there are any changes to the information. If you do not want your report shared with your doctor/referring institution, please specify below. In communications with us through foreign-origin applications such as WhatsApp, Gmail, whose servers are abroad, we provide the following information to inform you about the processing of your personal data.

When a message or email is sent to our center through foreign-origin applications, we protect all general and special personal data that can be obtained from this message or email, such as identity, communication, health, genetics, as a healthcare organization under the obligation of confidentiality, and do not share them with third parties unless legally required. However, sending a message or email through these applications means that, in addition to our center, the general and special personal data within the message or email content are shared with the trade company in the country where the application developer is located, thus transferring your data abroad. In this case, the confidentiality of the data obtained by the applications is not under the control of our center.

I approve the sharing of my test results with the following individuals:

Name Surname: Relationship Status:

- ☐ Send to my phone via messaging application.
Phone to send the test report:.....

Preferred messaging application: ☐ WhatsApp

☐ Other Application:

(There may be a transfer of data abroad with foreign-origin messaging applications.)

- ☐ Send to my email address.

Email address:

(There may be a transfer of data abroad with foreign-origin emails.)

☐ Other:

The above information form and open consent form are signed between Intergen and the Patient to determine the terms and conditions of the service to be provided by Intergen and to obtain the Patient's consent by informing them about this.

If the person to be tested has undergone a bone marrow transplant, please check the appropriate box below.

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PATIENT Full Name: Date: Signature:	If the patient is under 18 years of age or lacks the capacity to give informed consent, the Legal Representative's: Full Name: Signature: Date:	ON BEHALF OF INTERGEN Full Name: Date: Signature:
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