SAPIENS

PATIENT:		DOCTOR
Full Name: **BARCODE / PROTOCOL NO:**		**Full Name:**
Sex: M ☐ F ☐ **Date of Birth:**		**Telephone Number:** **E-mail:**
Mother's Name:	**Father's Name:**	**Institution:**
Passport No: **E-Mail:** **Telephone Number:** **Address:**		**Stamp:** **SAMPLE COLLECTION DATE / TIME:** __ / __ /____ - ___:___ **SAMPLE TYPE:** Peripheral blood: ☐ Tissue: ☐ Bone marrow: ☐ Other: ☐
TEST / PROCEDURE / ASSESSMENT:		
CLINICAL INFORMATION: *(Attach clinical summary and supporting documents if needed)*		

GENETIC TESTING INFORMED CONSENT FORM

This form documents that you have been informed, in accordance with the relevant provisions of the "Regulation on Medical Laboratory Services and Advanced Technology Medical Laboratory Practices," regarding the genetic test, analysis, or evaluation deemed necessary by your physician for yourself and/or an individual for whom you bear legal responsibility. For the procedure to be performed for the purpose of genetic testing, analysis, or evaluation, a suitable sample will be required to obtain the necessary genetic material. This sample will then be processed and undergo various stages (such as obtaining a sufficient cell population through cell culture, isolating DNA, and amplifying specific genes on the DNA, etc.) to prepare it for analysis. A results report will be prepared following the analysis; this report will be drafted in appropriate medical language, in accordance with international standards and regulations, to assist in making the necessary clinical decisions. The accuracy of the results obtained will be limited by factors such as the type of data evaluated through the analysis, the characteristics and limitations of the technical method applied, and the level of information provided by current literature. You may receive "genetic counseling" from a specialist physician to discuss matters such as receiving information about the procedure, planning your and your family's follow-up and treatment process regarding the results, and the reliability of the obtained results; in this case, your physician may refer you accordingly. Please do not forget to request counseling from authorized personnel regarding any topics you do not understand. References to publications, regulations, and other sources used to support some of the information in this form are cited as footnotes within the text, and links to these references—along with their bibliographic details—are provided at the end of the form.

What Type of Sample Will Be Needed?

Depending on the indication, different types of samples are required for genetic testing, such as 5–10 mL of blood, 1 cm² of tissue, or 20 mL of amniotic fluid. Your doctor will determine the type of sample to be used for the test and how it will be collected. The suitability of the sample is crucial for performing the test and ensuring the reliability of the results. Special samples—such as amniotic fluid, chorionic villus sampling (CVS), miscarriage tissue, and tissue biopsies—may need to be collected by your doctor under specific conditions, with a separate informed consent form and after you have been fully informed. The samples collected may be used up after the tests are performed.

What Is the Process and What Are the Characteristics of the Genetic Testing/Analysis/Evaluation to Be Conducted?

Medical genetic analyses can be classified into three main disciplines: conventional cytogenetic and molecular cytogenetic analyses, and molecular genetic analyses.

1. Conventional Cytogenetics (Karyotyping Test, Chromosome Analysis): Structural and numerical abnormalities of the chromosomes are evaluated. This analysis can be performed using a sample such as amniotic fluid collected during the prenatal period, as well as a blood sample, bone marrow aspiration material, etc. For conventional examination, dividing cells are required to make the chromosomes visible and allow for examination. In cases where direct examination is not possible, cell or tissue culture is used to obtain this population of cells. Some analyses, however, require direct preparation. The material collected for analysis may not contain sufficient or suitable cells and tissue; existing cells may not proliferate; or contamination may make analysis impossible. Even if cells proliferate, the quality of the resulting chromosomes may not be sufficient for analysis.

In such cases, it may be necessary to collect a new sample. Occasionally, obtaining a new sample may not be possible. The most significant technical limitation of the conventional method is its susceptibility to false-positive or false-negative results due to low resolution. Some structural abnormalities may go undetected, and "mosaicism"—indicating the presence of more than one chromosomal configuration—may not be identified due to the small number of cells examined. It may be beneficial to supplement chromosome analysis with additional methods.

2. Molecular Cytogenetics (FISH and Array Tests): The method known as FISH involves labeling specific regions of chromosomes with fluorescent dyes for examination. This method provides information only about the targeted region. Molecular karyotyping (array karyotyping, chromosomal microarray) is a method that uses specialized chips designed to simultaneously screen for DNA deletions, duplications, and copy number variations that are too small to be detected by standard chromosome analysis. The results obtained through array karyotyping vary depending on the resolution of the chip used.

3. Molecular Genetic Analyses: These are methods used to analyze the DNA sequence. Following DNA extraction, a target region or gene can be amplified and analyzed using molecular methods. These analyses can be performed using the methods listed below;

DNA can be directly visualized using the gel electrophoresis method. The DNA sequence can be read using capillary electrophoresis. Using next-generation sequencing (NGS);

- Panel tests allow multiple genes to be analyzed simultaneously.
- Clinical exome sequencing enables all 4,500 genes associated with identified genetic diseases to be read simultaneously.
- Whole exome sequencing enables approximately 20,000 genes encoded by DNA to be sequenced.
- Whole genome sequencing enables the entire DNA sequence, consisting of 3 billion base pairs, to be screened.The nature and volume of the data to be obtained will vary depending on the scope of the examination and the technical characteristics and limitations of the methods used. For the same reasons, there is also a possibility of obtaining false-positive and false-negative results with this type of analysis.

Is Genetic Testing Always Diagnostic?

Genetic analyses are not performed solely for the purpose of establishing a diagnosis, confirming a diagnosis, or determining the genetic basis of an existing disease or predisposition. Sometimes they are intended to inform treatment decisions; other times, they aim to determine the prognosis, assess the course of the disease, and estimate the likelihood of recurrence. Some genetic tests—such as preimplantation genetic screening to check embryos obtained through assisted reproductive technologies for chromosomal abnormalities before they are transferred to the uterus, and non-invasive prenatal screening based on cell-free fetal DNA testing, which is used primarily to screen for trisomy 21 in pregnant women—may be entirely qualify as "screening tests." However, even if this is the purpose of the test, it may not always be diagnostic. The fact that the method used is technologically advanced and yields more data does not guarantee that the genetic test will provide a definitive diagnosis. Although the scope of the test is very broad due to technological limitations, it may not provide the ability to examine every targeted region with equal coverage. Because information regarding certain genes and regions outside of genes is not yet sufficient, changes in the tested DNA may not be associated with a disease. The information obtained from the test results may need to be reanalyzed in light of technological and scientific advances and/or supported by additional tests. The decision to request reanalysis and/or additional tests is the responsibility of the attending physician. However, correlating laboratory data with other data—particularly clinical findings—is of great importance in genetic diagnosis and follow-up.

Situations Requiring Processing at Multiple Centers and What You Need to Know About Your Data

All of our genetic information is classified as sensitive personal data. We comply with the relevant regulatory provisions regarding the protection, storage, and sharing of this data with third parties. You may choose not to have a copy of your results sent to anyone, including the physician who requested the test, as indicated in the consent section. Otherwise, only the physician who requested the test may be sent a preliminary notification via email, and your physician(s) can access your results through the hospital's automation system. You may change this preference at a later time. The hand-signed results report is delivered in person to the patient, parent, or legal guardian. If you give your consent or submit a signed request, your hand-signed report may also be sent via mail or other means to the physician(s) who requested the test and are clinically monitoring you, to you, or to another person you authorize. From time to time, solutions for your genetic test may be provided through inter-laboratory collaboration in accordance with applicable laws. The collaborating laboratory may be located within the country or abroad. In some cases, you may be required to fill out an additional consent form. Responsibility for your results is shared by the laboratory providing the service and our center, which obtained your consent. Additionally, the security of your data and sample is our responsibility. However, particularly in cases where testing or analysis must be conducted abroad, by providing your consent, you acknowledge that potential delays and other similar technical or medical issues may arise due to reasons beyond the sender's control. For some tests, while part of the process is conducted at our center, data and sample transfers—whether between laboratories or via cloud-based systems—may be necessary for purposes such as equipment use or data processing. In the event of logistical issues that may arise during the transportation process prior to the sample's delivery to the laboratory (such as sample spoilage, loss, damage, or delays rendering the sample unfit for analysis, etc.), it may be necessary to collect a new sample or repeat the test. In such cases, the costs associated with retesting, sending a new sample, and related expenses are the responsibility of the patient/applicant or the requesting institution. Since the logistics process is beyond the laboratory's direct control, the laboratory cannot be held responsible for any problems that may arise during the transportation process. Based on the legal grounds specified in Articles 5/2(c) and 6/3 of the Personal Data Protection Law,and in accordance with the Regulation on Genetic Disease Evaluation Centers[1], your personal data may be shared in a confidential manner for the purposes of conducting examination, diagnosis, and treatment services; sample acceptance and registration; wet laboratory analysis of the sample; (bioinformatics), reporting, the shipment of samples to other centers, and the billing of summary tests. Genetic Disease Assessment Centers operate under the District and Provincial Health Directorates and the Ministry of Health. Public authorities may occasionally request data from these centers. Additionally, financial support required for testing may be provided by the Social Security Institution (SGK) and its affiliated agencies. In cases required by law or mandated by the authorities, the sharing of test results may be necessary.

Test Results and Reporting, Need for Additional Tests and Retesting

At our center, tests are processed as quickly as possible, and estimated turnaround times are based on average processing times. However, due to circumstances specific to the patient or the laboratory, or the need for additional tests and further evaluation, your results may be available sooner or later than indicated.

Sometimes, the results obtained may not be sufficient to reach a definitive conclusion. For this reason, your report may recommend confirmatory testing, repeat analysis, an additional test, or referral to a specialized center. In such cases, you may be required to provide a new sample or pay an additional fee for the new test. The result stated in the genetic test report may need to be reanalyzed at a later date, taking clinical evaluation into account. This ensures that evaluations and analyses can be conducted using the most up-to-date information, and the report remains current. At the center, reports, electronic records, and their backups, along with the informed consent forms obtained from patients prior to genetic testing and samples, are stored under appropriate conditions for the duration required by legal obligations.

Information Management

The SG Genetic Disorders Assessment Center is responsible for the management of all patient information obtained or generated during the course of laboratory operations, in accordance with legally enforceable agreements. The management of patient information includes confidentiality and privacy. Our laboratory notifies the client and/or patient in advance regarding any information it intends to disclose to the public. Except for information that the client and/or patient has made public, or when agreed upon between the laboratory and the patient (for example, to respond to complaints), all other information is considered confidential and is treated as such.

Disclosure of Information

The SG Genetic Disorders Evaluation Center informs the relevant patient about any information that is disclosed when legally required to do so or when authorized by contractual provisions, provided that such disclosure is not prohibited by law. Information about a patient received from a source other than the patient (e.g., a complainant, a regulatory authority) is kept confidential by the laboratory. The identity of the source is kept confidential by the laboratory and is not shared with the patient unless agreed upon with the source.

PATIENT INFORMATION NOTICE REGARDING THE PROCESSING OF PERSONAL DATA

As Sapiens Genetics and Health Services, Inc., in accordance with the Law on the Protection of Personal Data No. 6698 ("KVKK"), and in our capacity as the Data Controller, within the framework of our disclosure obligations under the KVKK, and taking into account the data security of our patients and their relatives, as well as the importance we place on the privacy of private life and the protection of fundamental rights and freedoms, we would like to inform you regarding the purposes of processing your personal data, the legal grounds, the method of collection, to whom it may be disclosed, and the rights granted to you under the KVKK..

1. What Personal Data Do We Process? We collect various types of personal data to provide healthcare services to our patients in accordance with the principles and conditions set forth in the KVKK and other relevant legislation (such as the Regulation on Personal Health Data). The following data pertaining to our patients is collected from patients themselves, healthcare institutions and organizations, or their relatives for the purpose of conducting medical diagnosis, examination, treatment, and care services. Your identification information (e.g., your first name, last name, Turkish ID number, passport number, date of birth, gender), and your contact information (e.g., your address, phone number, email address); if you are under 18 years of age, guardian identification information (e.g., first name, last name), contact information (e.g., phone number), your health data (e.g., data from private health insurance or social security institutions for the financing and planning of health services, data specified in the consent form you may provide in the event of a medical intervention, your patient protocol number, your test results, examination data, check-up information, information regarding your health and sexual life, and incidental findings), physical premises security information (e.g., camera system footage and recordings), your feedback, thank-you notes, and complaint data, as well as your legal proceedings data are processed.

2. For What Purposes Do We Process Your Personal Data?

Your Personal Data and Special Category Personal Data listed above are processed in accordance with the Regulation on Private Healthcare Institutions Providing Outpatient Diagnosis and Treatment, the Basic Law on Health Services No. 3359, the Decree-Law No. 663 on the Organization and Duties of the Ministry of Health and Its Affiliated Institutions, the Public Health Law No. 1593, the Patient Rights Regulation, the Regulation on Personal Health Data, and all other relevant legislation—to fulfill all our current and future legal obligations arising therefrom; to protect public health; to provide preventive medicine, medical diagnosis, treatment, and care services; to plan and manage health services and their financing; providing you with information regarding your appointment if you have scheduled one, issuing invoices, carrying out risk management and quality improvement activities, responding to any questions or complaints you may have regarding our health services, ensuring financial reconciliation

, responding to requests from the Ministry of Health and other public institutions and organizations in accordance with applicable legislation, measuring patient satisfaction following your receipt of healthcare services and enhancing patient satisfaction, contacting you for informational purposes regarding our services and conducting promotional activities, and the procurement of medications or medical devices. Your personal data will be processed, recorded, stored, retained, and classified in accordance with Articles 5 and 6 of Law No. 6698 and will be accessible only to doctors and staff bound by a duty of confidentiality. To provide information to prosecutors' offices, courts, and relevant public officials upon request and as required by law regarding matters of public safety and any legal disputes that may arise; when necessary, such information may be transferred to authorized public institutions and organizations, medical secretaries, domestic and international laboratories with which we collaborate for medical diagnosis, private insurance companies, law firms, the e-nabız system, the Medula system, and financial advisors, within the scope of the purposes specified above.

3.What Are the Methods Used to Collect Your Personal Data and the Legal Grounds for Such Collection?

Your personal data specified above is collected based on the legal grounds set forth in Articles 5 and 6 of Law No. 6698, namely: (i) where explicitly provided for by law; (ii) where necessary for the data controller to fulfill its legal obligations; (iii) where necessary for the legitimate interests of the data controller, provided that this does not infringe upon the fundamental rights and freedoms of the data subject; (iv) the protection of public health, the provision of preventive medicine, medical diagnosis, treatment, and care services, and the planning and management of health services and their financing, (v) the data subject's explicit consent—for the purposes specified in this Privacy Notice;

a) In compliance with the law and the principles of good faith, b) Provided that the data is accurate and, where necessary, up-to-date, c) For specific, explicit, and legitimate purposes, d) In a manner that is relevant, limited, and proportionate to the purpose for which it is processed, e) Retained for the period prescribed by applicable legislation or as necessary for the purpose for which they are processed, and may be processed, recorded, stored, retained, classified, and transferred through automated means (e.g., camera recordings) or non-automated means as part of any data recording system (e.g., forms filled out by you).

4. What Are Your Rights Regarding the Processing of Personal Data, and How Can You Exercise These Rights?

Article 20 of the Constitution establishes that everyone has the right to be informed about personal data concerning them. Article 11 of the Personal Data Protection Law (KVKK) lists the rights of data subjects under the law.

You may submit requests regarding your rights under the KVKK in writing to the "Data Controller, Sapiens Genetics and Health Services Incorporated," or via a registered electronic mail (KEP) address, secure electronic signature, mobile signature, or the email address previously provided to the data controller by the data subject and registered in the data controller's system.

Data Controller: Sapiens Genetics and Health Services Inc.

Address: Maslak Mh., Eski Büyükdere St., İz Plaza, Block No. 9/6, Sarıyer, Istanbul **Email:** info@sapiens.com.tr

INFORMED CONSENT FORM

This form becomes valid upon signing the consent section and confirms that your physician has provided the necessary information, that you have read and understood the information in the form, and that you approve it as appropriate. You do not need to initial each page separately, except for the consent section. Your consent constitutes a declaration that you have read and understood all pages, discussed any parts you did not understand, received satisfactory answers, and have given your consent. You must keep a copy of the signed form for your records; this will be in your best interest. There are four separate consent sections. The first concerns the continued storage of your sample or data after the legal retention periods have expired, provided it is appropriate; the second concerns the use of your data in statistical studies—following anonymization and complete removal of your identifying information—for which ethical committee approval has also been obtained; the third concerns the reporting of incidental findings; and the fourth pertains to the conduct of the test itself. You retain the right to withdraw your consent at any time; you may contact us to notify us in writing of your change in preference and ensure that your data is processed according to your new preference. When giving your informed consent, please remember that your genetic data also constitutes the personal data of all individuals related to you by blood.

Consent for the Use of Analytical Data and Specimens

Test results and patient specimens are an important research resource for physicians, scientists, and universities. To advance the treatment of diseases and improve our country's statistics, it is essential to grant the right to use this information for research, development, and statistical purposes—provided that your personal information is kept confidential, anonymized, and encrypted. The use of anonymized information falls under the center's authority. Provided that your identifying information remains confidential, your analysis data may be shared anonymously on scientific platforms. Your samples and data may be used as control samples. In addition to your consent, approval from the university's ethics committee will also be required.

Incidental Findings

In particular, when the DNA code is read using clinical exome sequencing, whole-exome sequencing, and whole-genome sequencing—especially with next-generation sequencing technologies—or when copy number variations across the genome are detected via chromosomal microarray analysis, diseases and predispositions that have not yet been identified may emerge in addition to the genes targeted for diagnostic purposes. In accordance with the principle that genetic data must not be used for purposes other than those for which it was collected, findings unrelated to the tested condition are not reported; however, upon request, information may be shared regarding genes or diseases that are treatable and/or preventable through appropriate preventive medical services. This authorization will be updated in accordance with relevant guidelines[2] and used—and information shared—only with the individual's consent. Following the identification of these incidental findings, it may be necessary for family members to undergo testing and receive genetic counseling.

Consent for the Processing and Transfer of Data Pursuant to the Personal Data Protection Law No. 6698

In accordance with the Personal Data Protection Law No. 6698 ("Law") and the relevant Regulation on Genetic Disease Assessment Centers, within the framework set forth as the data controller, and in accordance with the Basic Law on Health Services No. 3359, the Population Planning Law No. 2827, the General Public Health Law No. 1593, the Law on the Procurement, Storage, Transplantation, and Transfer of Organs and Tissues No. 2238, the regulations of the Ministry of Health, and other applicable legislation, your explicit consent is required for the retention and processing of your personal data.

My personal and sensitive personal data may be processed to the extent necessary for the performance of the contract, as explicitly provided by law, or as required to conduct genetic testing, analysis, or evaluation; however, my personal data related to health and sexual life may only be processed to the extent necessary for the protection of public health, preventive medicine, the provision of medical diagnosis, treatment, and care services, and the planning and management of health services and their financing, and to the extent necessary for these purposes; otherwise, I consent to the storage, processing, and transfer of such data in accordance with the provisions set forth in the Informed Consent Form for Genetic Testing,

☐ I give my consent.	☐ I do not give my consent.
☐ Incidental findings may be reported.	☐ Incidental findings may not be reported.
☐ May be used as anonymized data/sample, with identifying information removed, for statistical/control purposes in a study approved by the Ethics Committee.	☐ May not be used for a study approved by the Ethics Committee.
☐ I have read and understood the information notice. I have asked my questions. I know that I can request further consultation when necessary.	

I consent to my report being sent to the e-mail address ..... or being provided to ..... .

I asked the physician questions regarding my concerns about my condition, the risks, the procedures to be performed and the available options, and communicated all my thoughts. I was convinced by the answers I received. As a result of the information provided, I have been adequately informed. I GIVE MY CONSENT. Please write in your own handwriting: "I have read, understood and approved."

.....

Name Surname:

Date/Time:

Signature:

Name and Surname of the Patient or Legal Representative:

Signature of the Patient or Legal Representative:

Reason Why the Patient Was Not Informed and the Patient's Relative Was Informed:

☐ Patient is unconscious ☐ Emergency ☐ Patient is under 18 years of age

☐ Other: .....

Interpreter's Name and Surname (if required):

Signature:

SG GENETIC DISEASES DIAGNOSIS AND EVALUATION CENTER

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