BABYSEQ NIPT INFORMED CONSENT FORM

The purpose of this information text is to inform you about the BabySEQ test (a NIPT test). Before providing healthcare services, it is our obligation to inform you and obtain your consent, and it is your right to receive this information. Therefore, we kindly ask you to read this document carefully, complete the required fields, and contact us through any of the indicated communication channels if you have questions before or after the test.

Patient Name and Surname:

ID Number:

1 – What Is the BABYSEQ Test and When Is It Performed?

BabySEQ is a non-invasive screening test used to screen for chromosomal abnormalities. It is primarily used to screen for numerical chromosomal abnormalities, including Trisomy 21 (Down syndrome), Trisomy 18, and Trisomy 13, as well as numerical changes in all chromosomes. When the extended panel is selected, it also provides the opportunity to screen for deletions and duplications above a certain size threshold.

While some chromosomal disorders are incompatible with life (such as Trisomy 13 and Trisomy 18), others may be associated with intellectual and physical developmental delays. This test does not detect single-gene disorders, triploidy, congenital malformations, balanced rearrangements, or low-level mosaicism. However, in countries like ours where consanguineous marriages are relatively common, the prevalence of single-gene disorders is also high. Therefore, the BabySEQ Rare and BabySEQ XL tests have been developed to screen for single-gene disorders as well.

In these panels, testing is performed on both the mother and the father. If a risk to the fetus is identified based on parental testing, you and your physician will be contacted, and the identified condition(s) will be investigated in a fetal sample.

In the BabySEQ Rare test, SMA, Cystic Fibrosis, and Fragile X syndrome are screened. In the BabySEQ XL test, more than one thousand diseases for which the parents are carriers and that may pose a risk to the fetus are screened. In both tests, conditions that arise for the first time in the fetus and are not carried by the parents (autosomal dominant de novo mutations) cannot be detected.

When should this test be performed?

The test can be performed from the 10th week of pregnancy onward. However, it is recommended that the test be performed after the ultrasound (USG) examination conducted by your physician at the 11th week. If increased nuchal translucency is detected on ultrasound relative to gestational age, an invasive diagnostic test may be required instead of this screening test. In such cases, you may consult our physicians for further planning.

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

As with all medical tests, the BabySEQ test has a margin of error; its results are not definitive. Although highly sensitive, NIPT is a screening test, not a diagnostic test. It cannot replace diagnostic procedures such as chorionic villus sampling (CVS), amniocentesis, or cordocentesis. Depending on the BabySEQ result, additional analyses or confirmatory tests may be required.

Due to technical factors or biological reasons such as certain maternal diseases, fetal or maternal mosaicism, organ transplantation, recent full blood transfusion, or a vanishing twin, false negative or false positive results may occur. The effectiveness of the test decreases in twin or higher-order multiple pregnancies. Triploidy cannot be detected due to technical limitations. Low fetal DNA fraction reduces reliability. Maternal obesity or a high body mass index (BMI) is a common cause of low fetal DNA fraction.

For the BabySEQ Basic Panel and Extended Panel, there are three possible test results:

a) High risk:

Indicates an increased probability of a numerical chromosomal abnormality in the fetus for the chromosomes specified in the report or an increased likelihood of a deletion or duplication in the indicated genomic region. Confirmation with an invasive diagnostic test such as CVS or amniocentesis is required. (Applicable to BabySEQ Basic Panel and Extended Panel.)

b) Low risk:

Indicates that the risk of the screened conditions has been significantly reduced compared to the pre-test risk. The degree of risk reduction varies depending on the condition. Even if the result is low risk, if fetal ultrasound anomalies are detected during pregnancy follow-up or if there are other concerns about the baby's health, your physician may recommend fetal karyotyping or another genetic test. In such cases, we also recommend consulting our center.

c) No result due to low fetal DNA fraction:

This indicates that the laboratory was unable to obtain a result due to insufficient DNA. In such cases, the likelihood of a chromosomal abnormality in the fetus or the placenta may be slightly higher than average. When this outcome occurs, two options may be offered: repeating the test using a second sample (free of charge) to clarify the results, or proceeding directly with an invasive diagnostic procedure such as Chorionic Villus Sampling (CVS), amniocentesis, or cordocentesis.

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Possible Test Results for BabySEQ Rare and BabySEQ XL

In the BabySEQ Rare and BabySEQ XL tests, diseases for which the parents are carriers and that may pose a risk to the fetus are evaluated using blood samples obtained from both the mother and the father. If a fetal risk is identified in these tests, you and your physician will be contacted, and the disease(s) identified as risky will be investigated in a fetal sample obtained via CVS or from amniotic fluid. In the BabySEQ Rare test, SMA, Cystic Fibrosis, and Fragile X are screened; in BabySEQ XL, more than one thousand diseases for which the parents are carriers and that may pose a risk to the fetus are screened. The report you receive will include three possible results:

a) Carrier status has been identified in the couple that may pose a risk to the fetus.

The variant or variants* detected in both partners, as well as variants located on the mother's X chromosome that may cause disease risk in a male fetus, will be stated in the report. The presence of these variants does not necessarily mean that the fetus will be affected. When an increased risk is identified, the family and the physician are informed, and depending on the degree of risk, chorionic villus sampling or amniocentesis may be performed to determine whether the fetus is affected. No additional fee will be charged for these confirmatory tests.

b) No carrier status has been identified in the couple that may pose a risk to the fetus.

No carrier status that would pose a fetal risk has been detected in either partner. When this result is obtained, routine pregnancy follow-up is recommended. However, this test cannot eliminate the risk for all genetic diseases, although it significantly reduces the risk. This test does not reduce the risk related to de novo mutations that may occur in the fetus.

c) The test needs to be repeated because sufficient DNA could not be obtained due to technical reasons.

Insufficient DNA could be obtained from the sample collected for testing. This is a rare technical issue and requires a new sample to be collected.

Note: Only pathogenic (genetic changes that are definite or near-definite causes of disease) and likely pathogenic variants (changes with a >95% probability of being disease-causing) are reported.

3 – What Other Information Should I Know About the Test?

3.1. The stated turnaround times are approximate; results may be reported sooner or later.

3.2. After testing, any remaining DNA samples may be stored indefinitely for test validation or training purposes. After completion of the requested test(s), you may apply to the laboratory to request destruction of your blood and DNA samples. You may also indicate this in the approval section of this form. Refusing permission for your sample to be used for validation or training does not affect your test result.

3.3. In some cases, the detected findings may need to be supported by additional "diagnostic tests." If so, you will be contacted and informed about these tests. If you choose not to undergo the recommended additional tests, diagnostic success may decrease. For tests with high costs, additional payment may be required.

3.4. If deemed medically necessary, part or all of the testing may be sent to another center.

3.5. In rare cases, you may need to submit or provide a second material/sample. If no result can be obtained, the test may be repeated.

3.6. If you do not provide your sample at our center and it is delivered by cargo/courier, you will be notified by message once the sample arrives at our center. Payment information is also provided in that message. After you make the payment to the provided account and share the receipt, your testing process will be initiated. During this period, sample security is ensured at our center.

3.7. If you withdraw from the test after making payment, we may refund the remaining amount excluding the expenses incurred up to that stage, depending on the progress of the test; however, if a significant portion of the test has already been completed, a refund may not be possible.

3.8. Some data obtained from the tests may not be reported for scientific or ethical reasons.

3.9. Your test results will be stored in Intergen records.

3.10. If your sample was not collected by Intergen, Intergen is not responsible for whether the material was collected correctly, the suitability of the material, or the information provided by other centers.

3.11. If our physicians determine that a different test than the one requested by your physician would be more suitable to establish a diagnosis, you and/or your physician will be informed and the test deemed appropriate will be recommended in order to protect your interests and to avoid the need for repeat sampling. Our center reserves the right to decline tests that are not medically appropriate.

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3.12. If you request, your test result may be sent to you via communication channels such as WhatsApp, Gmail, Hotmail, etc. However, we would like to inform you that these applications are of foreign origin, which means that the transmitted message may reach the servers of companies located abroad, i.e., it may be transferred outside the country. Ensuring the confidentiality of the data by the companies that own these applications is not under the control of our center; you may access their privacy policies on their own websites.

3.13. As a licensed healthcare institution, in order to perform genetic testing, we are required to obtain your identity, contact, health, and genetic data. In accordance with our obligation of confidentiality, we protect this data and ensure its privacy. Unless you provide consent or there is a legal obligation, we do not share your data with third parties/institutions. If you give consent, we may use your data in studies conducted for scientific purposes, provided that your personal data is anonymized and your right to privacy and personal rights are protected. You have the right to learn whether your personal data is processed; to request information if your personal data has been processed; to learn the purpose of processing and whether it is used in accordance with that purpose; to know the third parties to whom your personal data is transferred domestically or abroad; to request correction if your personal data is processed incompletely or incorrectly; to request the deletion or destruction of your personal data and to request that such actions be notified to third parties to whom the personal data has been transferred; to object to an outcome against you that may arise as a result of analysis of the processed data exclusively through automated systems; and to request compensation if you suffer damages due to unlawful processing of your personal data. We will respond to your applications within 30 days.

CONSENT FORM

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 period of time to read the written information. I understand the purpose of the test, its results, reliability, the reporting time, and the processes related to the processing of my personal data."

Dear Patient; in accordance with the statement above, if you consent to the genetic test being performed on your material under the conditions specified in the information form, please write **"I consent to the genetic test being conducted."** in the blank space below:

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(As any medical intervention in the field of healthcare may only be performed after obtaining informed consent, it will not be possible for us to perform the test if you do not provide consent.)

2 – Consent Form for Additional Testing if Medically Necessary (To be completed by the patient)

Dear Patient; as explained in the information form, in genetic studies, if medically necessary to establish a diagnosis, additional tests that may assist in diagnosis may be performed, or it may be considered during the process that a test different from the one specified may be more appropriate. If you accept this, please mark the box below stating "I accept." If you do not accept additional testing, please mark the box stating "I do not accept." Please note that if you do not undergo the recommended additional tests, diagnostic success may decrease. If additional tests require payment, our team will inform you.

□ ***I accept.***

□ ***I do not accept.***

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3 – Scientific Research Consent Form (To be completed by the patient)

Dear Patient; as Intergen Genetic Diseases Evaluation Center, in order to protect and improve public health, we may conduct scientific studies by anonymizing the health and genetic data under our responsibility. In these studies, we take all necessary administrative and technical measures to ensure the confidentiality of private life and the protection of personal data.

Under these conditions, please indicate whether you consent to the use of the data you have shared with us for scientific research by marking one of the boxes below:

□ **I consent.**

□ **I do not consent.**

4 – Consent Form for the Delivery of Test Results (To be completed by the patient)

Dear Patient; please mark the box corresponding to the method by which you would like us to deliver your test results. In case of any changes in your contact information, please inform us as soon as possible. If you do not wish your report to be shared with your physician/referring institution, please indicate this below.

In communications established with us through foreign-origin applications such as WhatsApp, Gmail, and similar platforms whose servers are located abroad, we provide the following information regarding the processing of your personal data:

When a message or email is sent to our center via foreign-origin applications, as a healthcare institution bound by confidentiality, we protect all general and sensitive personal data belonging to you—such as identity, contact, health, and genetic data—that may be obtained through such messages or emails, and we do not share them with third parties unless there is a legal obligation. However, sending a message or email via such applications means that the content of the message, including your general and sensitive personal data, is also shared with the commercial company in the country where the application developer is located; in other words, your data may be transferred abroad. In such cases, ensuring the confidentiality of data obtained by these applications is not under the control of our center.

□ **Please send my results to my phone via a messaging application.**

Phone number to which the test report will be sent:

Messaging application to be used: □ WhatsApp

□ **Other applications:**

(Data transfer abroad may occur when using foreign-origin messaging applications.)

□ **Please send my results to my email address.**

Email address:

(Data transfer abroad may occur when using foreign-origin email services.)

□ **Other:**

I approve the sharing of my test results with the following person(s):

Name and Surname:

Relationship:

This Information Form and Explicit Consent Form have been signed between Intergen and the Patient in order to determine the terms and conditions of the service to be provided by Intergen and to obtain the Patient's informed consent.

PATIENT	**ON BEHALF OF INTERGEN**
Full Name:	Full Name:
Date:	Date:
Signature:	Signature:

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