DÜZEN LABORATORIES GROUP
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Promissium NIPT
REQUEST ve CONSENT FORM

Promissium NIPT
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GENETIC DISEASES EVALUATION CENTER

Patient's Information

Name Surname: ....

Identification #: _ _ _ _ _ _ _ _ _ _ _ Phone: + _ _ _ (_ _ _) _ _ _ - _ _ _ _

National ID or Passport # or any other officially recognized ID — country code — area code

Date of Birth (DD/MM/YYYY): _ _ / _ _ / _ _ _ _ E-mail: ....

Physician's Information

Referring Physician / Laboratory: ....

E-mail: .... Phone: + _ _ _ (_ _ _) _ _ _ - _ _ _ _

country code — area code

Requested Test ☐ 6837 Promissium NIPT [*basic*] ☐ 6838 Promissium NIPT [*genome-wide*]

Clinical Indication(s)

- ☐ Advanced maternal age
- ☐ High-risk prenatal biochemical screening result (*Please attach result of your biochemical screening test result*)
- ☐ Chromosomal abnormality in the previous child
- ☐ Translocation carrier in the family
- ☐ Chromosomal abnormality suspicion in the fetal ultrasonography (*Please attach copy of your ultrasonography report*)

Sampling Date (DD/MM/YYYY): _ _ / _ _ / _ _ _ _ Time: _ _ : _ _ AM/PM

Blood taken (10 ml) from the mother between the 10th and 18th weeks of her pregnancy should be immediately put into *Streck*™ tubes that can be obtained from our laboratory. The tube should be labeled with a barcode containing patient's name, surname and national identification number. The tube containing the blood sample must be delivered to our laboratory within 96 hours at a temperature range of 6℃ - 24℃ together with the filled and signed request/consent form. The tube used provides maximum stability for 96 hours at the specified temperature range, stability may decrease if this period is exceeded.

Information Regarding to Pregnancy

Pregnancy: ☐ Single embryo ☐ Twin pregnancy ☐ Multiple pregnancy

! In twin pregnancies confidence intervals of the test vary. For other factors affecting sensitivity and specificity please refer to back page.

IVF Pregnancy: ☐ Yes ☐ No

Gestation period: _ _ weeks _ day(s)

Gestational age estimation: ☐ Last menstrual period (LMP) ☐ Ultrasonography ☐ IVF

Last Menstrual Period (LMP): _ _ / _ _ / _ _ _ _ (DD / MM / YYYY) Estimated Due Date (EDD): _ _ / _ _ / _ _ _ _ (DD / MM / YYYY)

I acknowledge that I have read, understood, and agreed with the ***informed consent form*** on the back page. I declare that all my questions about the subject have been properly answered and in addition to the situations mentioned in here, I have been given full and clear information about all known issues, tests, methods, causes, and results regarding to this test, and all my doubts have been removed. I acknowledge that my peripheral blood sample can be taken and used for the purposes specified in the ***informed consent form*** on the back page.

I have been informed that my results cannot be shared with third parties and institutions without my explicit consent in accordance with the Regulation on Genetic Diseases Evaluation Centers, accordingly, I request that my results be sent to the e-mail address I have specified in my own handwriting below and shared with my doctor, whose contact information I have provided in the *Patient Information* section above.

E-mail: _

Date: _ _ / _ _ / _ _ _ _ Patient's Signature:

I declare that my patient read the "***informed consent form***" on the back page; that I answered all my patient's questions about the limitations, results and method of application of the Non-Invasive Prenatal Screening Test (NIPT), as well as the conditions specified here and that I have informed her about all related matters.

Date: _ _ / _ _ / _ _ _ _ Physician's Signature:

INFORMED CONSENT FORM

This form has been prepared to inform you about issues on taking a peripheral blood sample from you to perform the NON-INVASIVE PRENATAL TEST (NIPT), which is explained in detail below, and on storing, examining, evaluating this material, and transferring obtained results to you. To comply with legal obligations, this NIPT test can only be performed if you give your consent and therefore, you must read the information below, know the reasons and consequences of the examinations and studies to be carried out, and give your approval beforehand.

AIM OF THE TEST – The main purpose of this test is to determine risks for trisomy 21 (Down syndrome), trisomy 18 (Edwards syndrome), trisomy 13 (Patau syndrome) and aneuploidies in the sex chromosomes in our Promissium NIPT [*basic*]; and besides these for rare autosomal aneuploidies, deletions and duplications of over 7 Mb in autosomal chromosomes, in our Promissium NIPT [*genome-wide*].

TESTING INFORMATION – This test is performed on an Illumina NextSeq 550Dx next generation DNA sequencing platform using the IVD-CE certified Illumina VeriSeq™ V2 Kit. With our "*basic*" test, 99.9% of trisomy 21, trisomy 18 and trisomy 13 cases can be detected. With our "*genome-wide*" test, in addition to these trisomies, 96.4% of rare autosomal aneuploidy cases, and 74.1% of autosomal partial deletion and duplication (≥7 Mb) cases can be detected. Polyploidies, balanced chromosomal anomalies, and non-chromosomal genetic diseases are not covered by this test.

First, extracellular DNA (cfDNA) - which is found in small amounts in the blood taken from the mother, is separated. This cfDNA contains DNA fragments of both the mother and the baby. Then, this DNA is sequenced on the next generation sequencing platform to obtain sequences for all DNA fragments present. These sequences are then compared with the reference human genome and analyzed in terms of both length and genomic coordinates with the help of an algorithm. The ratio and quality of the baby's DNA in the blood of the mother is obtained statistically. If the quality, ratio and amount are appropriate, the targeted chromosomal anomalies are evaluated using certain statistical models. Depending on the quality, amount and ratio of obtained DNA, this statistical model cannot make a precise evaluation within reliable limits under certain conditions. In this case, the result is reported as invalid, and a new sample may be asked from the mother.

CONDITIONS THAT AFFECT THE RELIABILITY OF TEST RESULTS – In following cases, results may not be given and/or the reliability of the given result is low: multiple pregnancy (except twin pregnancy), maternal organ transplantation, maternal malignancy, maternal blood/bone marrow transplantation, immunotherapy or stem cell therapy within the last 3 months, maternal or fetoplacental mosaicism, *intrauterine ex* or loss of one of the twins.

TEST RESULTS – The report is given as "*high risk*" or "*low risk*" separately for each of the chromosomal diseases screened. Issues related with the baby that can be detected as a result of the examination are ONLY limited to the specified chromosomal diseases and do not cover all possible genetic diseases. A "*low risk*" result for the screened disease does not necessarily guarantee that the baby is healthy for this disease. If a "*high risk*" result is given for any of the chromosomal diseases screened, this does not necessarily mean that the baby has this disease. In these pregnancies, a definitive diagnostic test is recommended and for a final diagnosis chromosome analysis from amniocentesis or chorionic villus material is required.

CONSENT - I have read the above explanations to inform me regarding the risk determination of my unborn baby for chromosomal diseases specified in the test descriptions and screened within the scope of the test which is the reason for my application to the laboratory. I acknowledge and declare that:

- ❑ **I know the purpose of the test;**
- ❑ **I have been informed about whether the peripheral blood draw required for the test will harm my health, my child and/or my baby's health and that I have accepted, in advance, this procedure with my free will and consent;**
- ❑ **I accept in advance that the results to be obtained from the test may have false positive and false negative values;**
- ❑ **There is a possibility that the tests performed may not yield conclusive or correct results due to scientific and/or environmental factors;**
- ❑ **The test may not give conclusive results to determine the risk and if happens, the peripheral blood can be taken again, and I give my consent and accept possible delays that may occur for this reason;**
- ❑ **Side effects such as pain, ache and infection may be seen in the application area or body due to the peripheral blood collection and such ailments are natural due to medical intervention, therefore I will not make any complaints about such issues that may arise;**
- ❑ **My samples/results can be kept by the laboratory for future scientific purposes such as research, training or test standardization under the strict condition that my results will not be shared with any person, institution or organization, except for scientific research to develop medical genetics, and will be kept confidential under all circumstances, and my personal information is protected;**
- ❑ **For married couples, it is their own choice to learn individual test results alone or with their partner.**

As a result; I acknowledge that I have read, understood, and agreed with the above-mentioned points. I declare that all my questions about the subject have been properly answered and in addition to the situations mentioned in here, I have been given full and clear information about all known issues, tests, methods, causes, and results regarding to this test, and all my doubts have been removed. I acknowledge that my peripheral blood sample can be taken and used for the purposes specified in the informed consent form above.