CGC genetics

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REQUEST FORM

PRENATAL SCREENING

*MANDATORY FIELDS

ORDERING PHYSICIAN

PHYSICIAN'S NAME* | DEPARTMENT

MEDICAL CENTER* | PHONE

EMAIL* | CITY | COUNTRY

PATIENT INFORMATION

NAME* | DATE OF BIRTH* (dd/mm/yyyy) | MEDICAL RECORD NO.

PREGNANT WEIGHT* | ETHNIC GROUP | GENDER*

Kgs | feminino

BILING INFORMATION (if receipt is issued on patient's behalf):

VAT No. | ADDRESS | ZIP CODE | CITY | COUNTRY

TEST*

- ☐ **COMBINED 1ST TRIMESTER** — NT + NB + PAPP-A + freeβhCG (11w – 13w+6d)
- ☐ **EARLY COMBINED SCREENING** — PAPP-A + freeβhCG (9w – 11w)] + [NT + NB (11w – 13w+6d)
- ☐ **2ND TRIMESTER** — AFP + freeβhCG (14-22 weeks)
- ☐ **NTD only** — AFP (14-22 weeks)

COLLECTION DATA*

DATE AND TIME OF COLLECTION (dd/mm/yyyy) ____/____/____ ____ :____

LOCATION OF COLLECTION ____

LMP (dd/mm/yyyy) ____/____/____

GESTATIONAL AGE AT TIME OF COLLECTION:

by LMP ____ weeks, ____ days.

by ULTRASOUND ____ weeks, ____ days.

ULTRASOUND DATA*

ULTRASOUND DATE (dd/mm/yyyy) ____/____/____

GESTACIONAL AGE (US) ____ weeks, ____ days.

NO. OF FETUSES ____ **CHORIONICITY** ____

CRL ____ mm **NT** ____,____ mm

NASAL BONE ☐ present ☐ absent ☐ not evaluated

CLINICAL INFORMATION*

	YES	NO
Other screening in this gestation	☐	☐
Pregnant woman was insulin-dependent before pregnancy	☐	☐
If not, is she presently being treated with insulin?	☐	☐
Smoker	☐	☐
Loss of blood in this pregnancy	☐	☐
Previous chromosomal studies	☐	☐
Family history of Down Syndrome	☐	☐
Valproic Acid / anticonvulsants in this pregnancy	☐	☐
Previous child with Neural Tube Defects (NTD)	☐	☐
Family history of non-NTD abnormalities	☐	☐

	YES	NO
MEDICALLY ASSISTED REPRODUCTION	☐	☐

EXTRACTION DATE (dd/mm/yyyy) ____/____/____

METHOD | EMBRYO TRANSFER (dd/mm/yyyy)

____ | ____/____/____

Donor age | Donor DOB (dd/mm/yyyy)

____ | ____/____/____

I certify that (i) the patient (or legal representative) has agreed to have this testing performed, by signing the patient informed consent, (ii) the patient informed consent is in agreement with the legal requirements and that (iii) I am providing CGC Genetics all relevant medical information indispensable for the testing to be performed.

I certify that (i) I was informed about the benefits, risks and limitations of the test to be performed, (ii) I put all the questions that I consider relevant and understood the answers provided and (iii) that I understand that a normal result does not guarantee a normal baby. I give authorization to proceed with the requested genetic test and the use of the sample exclusively to this end.

☐ **I agree** ☐ **I do not agree**

The sample can also be used for scientific research purposes. ☐ *I agree* ☐ *I do not agree*

PHYSICIAN'S SIGNATURE ____
(mandatory)

Date: ____/____/____

PATIENT'S SIGNATURE ____
(to be used in case no other informed consent is available)

Date: ____/____/____

PRENATAL SCREENING

This is a brief description about Prenatal Screening test. In case any questions remain, please consult your attending physician.

Combined First Trimester Screening (Ultrasound and Biochemical)

- Performed between 11 weeks and 13 weeks + 6 days of pregnancy, the combined screening combines the ultrasound information with biochemical measurements in maternal blood.
- The ultrasound examination precisely evaluates gestational age and nuchal translucency (NT).
- In maternal blood sample, two elements are quantified – free fraction of Human Chorionic Gonadotropin (free ß-hCG) and Pregnancy-Associated Plasma Protein A (PAPP-A) – which are found in the blood of all pregnant women.
- Ultrasound and biochemical results are then used to calculate a specific risk for Down Syndrome (trisomy 21), trisomy 18 and trisomy 13.
- Accuracy of Combined First Trimester Screening: Combined 1st Trimester Screening (NT+freeß-hCG+PAPP-A) detects 90% - 92% of babies with Down syndrome and may detect other anomalies.

Early Combined Screening

- Early Combined Screening is another version of Combined 1st Trimester Screening, in which marker evaluation is performed as early as possible: blood collection and biochemical measurements are performed between 9 and 11 weeks; and ultrasound data is taken into account for risk calculation at 11-12 weeks.
- This combination allows to pull the best efficiency of each marker (detection rate of 93% to 97%) and still be possible to provide the result right after ultrasound examination, which may occur in a medical setting, thus shortening the planning process in cases of increased risk.

Second Trimester Prenatal Screening

- Performed between 14 and 22 weeks.
- The sample is analyzed to quantify two chemical elements designated as Alpha-fetoprotein (AFP) and Human Chorionic Gonadotropin ß-hCG, which are present in the blood of all pregnant women.
- The results allow to calculate the risk for Down syndrome, trisomy 18, trisomy 13 and NTDs.
- Accuracy of Second Trimester Prenatal Screening: 2nd trimester screening detects > 98% of babies with anencephaly, 90% with spina bifida and 80% with Down syndrome.

When Prenatal Screening shows an increased risk

- 3 to 5% of women have an increased risk result.
- In case screening results shows an increased risk, you should consider other screening and/or diagnostic methods.
- Your doctor will always explain the results to you, suggesting which subsequent tests to perform (such as Non-Invasive Prenatal Test, detailed ultrasound or prenatal diagnosis, which requires amniocentesis or chorionic villus biopsy for fetal chromosomal analysis).
- As an alternative to invasive diagnosis (*i.e.*, amniocentesis or chorionic villus) it is now possible to perform, as early as 10 weeks of pregnancy, a non-invasive prenatal test, such as TOMORROW Prenatal test, that from a maternal blood sample, detects trisomy 21, 18 and 13, identifies fetal gender and aneuploidies in sex chromosomes (Monosomy X, XXX; XXY, XYY). TOMORROW Prenatal Test has sensitivity above 99% for trisomy 21.

When Prenatal Screening shows a reduced risk

- In order to increase detection rate, it is now possible to perform, as early as 10 weeks of pregnancy, a non-invasive prenatal test, such as TOMORROW Prenatal test, that from a maternal blood sample, detects trisomy 21, 18 and 13, identifies fetal gender and aneuploidies in sex chromosomes (Monosomy X, XXX; XXY, XYY). TOMORROW Prenatal Test has sensitivity above 99% for trisomy 21.

Important notice

Prenatal screening test result indicating a reduced risk does not exclude the possibility of the baby having Down syndrome, trisomy 18 or any other chromosomal anomaly, neither it eliminates the possibility of congenital defects, mental retardation or any other disorder not traceable by Prenatal Screening. It only indicates that there is a decreased risk for the described anomalies. If you have any questions, please consult with your physician or contact CGC Genetics.