The following consent should be reviewed with the referring physician or the specialist in genetics, prior to signing

Referring Centre		Barcode	

Introduction to SANCO Prenatal Test

What is SANCO Prenatal Test?

SANCO Prenatal Test is a genetic screening test for pregnant women. The test determines very precisely the risk of trisomy and other selected chromosomal anomalies of the fetus. The test poses no risk to the mother or baby and is highly sensitive, being able to detect 99% of the cases of the most common chromosomal trisomies. This examination does not replace invasive testing (such as amniocentesis), but in most cases helps to avoid it. **SANCO Test** can be done from 10 weeks of gestation onwards.

How does SANCO Test work?

During pregnancy, fetal DNA, originated mainly from the placenta, circulates in the mother's blood. This genetic material is most of the time equal to the fetal genome and can be used to quantify the risk of a chromosomal anomaly. The **SANCO Test** is based on the analysis of placental DNA extracted from a blood sample (10 ml) collected from the mother.

SANCO Test is a screening test:

Sensitivity, specificity and accuracy of the **SANCO Test** are very high. However, the **SANCO Test** is a screening test. This means that not all affected pregnancies will be detected during examination, and if the **SANCO Test** results in a high risk for a disease, a confirmatory invasive test will be required for the final diagnosis (preferentially amniocentesis).

What can be evaluated with the SANCO Test?

Non-invasive prenatal genetic tests, such as **SANCO Test**, are performed mainly to exclude the presence of the most common trisomies (extra copies of chromosomes 21, 18 and 13). **SANCO** are genome-wide tests, which means that all the chromosomes and several other sub-chromosomal selected abnormalities can also be evaluated.

Condition	Description	SANCO sensitivity rate*
Trisomy 21	An additional, third copy, of chromosome 21 causes the disease commonly known as Down Syndrome, which is the most common genetic cause of intellectual disability; in Down's syndrome, other abnormalities may also be observed, such as heart disorders, hearing and visual impairments, immunity disorders and gastro-intestinal disorders.	>99.9%
Trisomy 18 and 13	An extra copy of chromosome 18 causes Edwards syndrome, and of chromosome 13 causes Patau syndrome. Many babies with these syndromes are miscarried or stillborn and of those babies born alive, most die before one year of age due to severe birth defects. Babies who survive beyond one year have serious developmental problems.	T18: >99.9% T13: >99.9%
Sex chromosome abnormalities	The test determines the risk of sex chromosomal aneuploidies (XXX, XXY, XYY and X monosomy). These abnormalities are less severe but more common than autosomal trisomies. Except monosomy X (Turner syndrome) there is no risk of miscarriage; most affected children are expected to be physically and intellectually healthy. They may require medical attention of an endocrinologist and other specialists and in adulthood they are at risk of infertility.	X monosomy 90.5%
Fetus gender	Described as male / female	99.9%
Rare autosomal aneuploidies	Abnormal number (aneuploidy) of 1-12, 14, 15-17, 19, 20, 22 chromosomes, is a disorder so serious, that its presence in the fetus causes miscarriage in the first trimester. Thus, if such abnormality is detected in the **SANCO,** it is usually limited to the placenta, but still may rarely be associated with fetal mosaicism, pregnancy complications, poor fetal growth, and an increased risk of miscarriage. Sometimes the cause of a high risk result is cancer in the pregnant woman.	96.4%
All (over 430) autosomal deletions and duplications ≥7 Mbp **SANCO**	Such large genetic errors (7 million base pairs) occur randomly and very rarely, but, because of the extent of the damage, they have severe clinical consequences. The location, size and type of the lesion (deletion or duplication) determines the occurrence and severity of various clinical symptoms, which most often include intellectual disability, delayed psychomotor development and birth defects.	74.1%

* VeriSeq™ NIPT Solution v2 Package Insert Illumina, Inc. 2019;

Test result

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Possible SANCO Test results:

A low risk result (negative - no aberration detected) means the result of the chromosome examination does not differ from the standards and there is a very low risk of the fetus having the anomaly in question.

A high risk result (positive - aberration detected) means high probability, but not certainty, that the fetus is affected by the indicated disease. In this type of results, the positive predictive value will be reported, if known, i.e. information about the probability of the presence in the fetus of an anomaly found in the **SANCO test**. The rarer the defect, the lower the positive predictive value of the result. Any high risk result requires genetic counselling.

An "**Unable to perform testing" result** means that it was not possible to perform the **SANCO Prenatal Test**. Sometimes it is necessary to repeat the test or re-draw the blood to obtain a result. A new sample will be requested at no extra cost, but it increases the time of waiting for the result. For that reason, this possible result should be considered in the planning of prenatal testing. If the **SANCO test** cannot be performed even from the second sample, a report on the inability to perform the testing will be issued and the test cost will be reimbursed. Test cancellation during its performance or refusal of subsequent blood draw do not entitle to reimbursement of any costs.

Advantages and disadvantages of the genome-wide screening approach:

The genome-wide screening allows risk assessment of genetic abnormalities, which may cause miscarriage, IUGR or low birth weight; sometimes it may even result in the early detection of cancer in a pregnant woman. If you choose the **SANCO** genome-wide type of screening, there is a small probability (up to 0.5%) of an unusual chromosomal defect result, in addition to common trisomies. The identified changes may thus impact the health of the fetus or the mother. With such rare, positive **SANCO test** results, a confirmatory invasive testing (ie amniocentesis) will be required to verify if the anomaly is of fetal origin. It may turn out that the cause of this positive **SANCO test** result is not a fetal disease, but, for example, the onset of cancer or placental mosaicism; further testing would be recommended.

Consider the benefits and possible disadvantages of the whole-genome analysis (need for further, possibly unnecessary testing), and make a choice about the scope of testing, if needed, with the help of your physician.

The genome-wide **SANCO test** can be performed in singleton and twin pregnancies being the result, in twins, reported for both fetuses combined and may be slightly less accurate than for a singleton pregnancy.

What the test does not detect:

Hereditary and monogenic diseases resulting from a "small" gene damage, such as cystic fibrosis or spinal muscular atrophy;
Multigenic and multifactorial diseases or malformations such as cleft lip, epilepsy, autism;
Small deletions and duplications, below the resolution of the method (7 Mbp for **SANCO**);
Aneuploidy of sex chromosomes in twin pregnancies;
Polyploidy (like triploidy) and **balanced translocations**
Fetal mosaic disease when the fetal chromosomal defect does not occur in sufficient percentage of placental cells.

Contraindications to testing:

ABSOLUTE:

(1) **less than 10 completed weeks of pregnancy** (2) **Cancer** (3) **bone marrow or organ transplantation** (4) **blood transfusion** in the last 3 months (5) **vanishing twin in multiple pregnancy with twin demise after 8th week of pregnancy** (6) **allogenic stem cell therapy (**7) systemic **lupus** erythematosus

Consult **a specialist** in case of:

(1) finding **birth defects** in an anatomy scan (ultrasound)
(2) **high risk of trisomy** in the first trimester combined test

Consult **the SANCO helpline** if:

(1) one **twin died** in multiple pregnancy (the vanishing twin syndrome)
(2) you are a known **carrier of a chromosomal anomaly**
(3) you were vaccinated with **your partner's lymphocytes**
(4) you are undergoing **immunotherapy**

NIPT testing, as recommended by the Polish Society of Human Genetics and the Polish Society of Gynecologists and Obstetricians, should not be chosen as a replacement for invasive based testing when a risk >1:100 is derived from the first trimester combined test.

The low risk test result does not exclude other genetic disorders or birth defects that are not assessed by the SANCO test.

This information is for you, take it home.

SANCO Non Invasive Prenatal Test

REQUEST FORM v05.0 (02-2025)

Copy for the laboratory [Genomed]

Genomed

Referring Centre		Barcode	

PATIENT INFORMATION

First name

Surname

Phone No:

ID/Social Security No.

Date of birth: DD / MM / YYYY

Weight: kg Height: cm

e-mail:

TEST INDICATIONS

- ☐ Maternal anxiety
- ☐ Advanced maternal age (>35 years)
- ☐ Positive serum screening (fill in below)
- ☐ Abnormal ultrasound (fill in below)
- ☐ Pregnancy failures: ☐ miscarriage ☐ pregnancy/child with chromosomal aberration (please specify) .....
- ☐ *In vitro* fertilization
- ☐ Chromosomal aberration carried by the mother or the father (please specify) of examined fetus.....
- ☐ Family history of a genetic disease (please specify): .....

Additional remarks: ....

ie chronic disorders, medications, vanishing twin

Pregnancy: ☐ Singleton ☐ Twin Date of delivery: DD / MM / YYYY

Chorionicity (in twin pregnancy):

☐ DC/DA: dichorionic, diamniotic ☐ MC/DA: monochorionic, diamniotic ☐ MC/MA: monochorionic, monoamniotic

Nuchal Translucency (NT): mm βhCG: MoM PAPP-A: MoM Other markers: .....

Prior screening test: ☐ First trimester combined PAPP-A test ☐ 1st trimester ultrasound ☐ 2nd trimester ultrasound ☐ Other ☐ None

Risk assessment: T21 1: T18 1: T13 1:

TEST REQUESTED

oraz

Maximum 5 working days

☐ **SANCO standard**

- Trisomies 21, 18, 13 + Fetal sex
- Sex chromosome aneuploidies (analysis not available for twin pregnancies)

oraz

Maximum 5 working days

☐ **SANCO expanded (genome-wide)**

- Trisomies 21, 18, 13 + Fetal sex
- Sex chromosome aneuploidies (analysis not available for twin pregnancies)
- Analysis of the number (aneuploidy) of all autosomal chromosomes
- Panel of of all autosomal deletions and duplications ≥7 million base pairs (over 430 deletions and duplications, including 1p36 deletion, 12p duplication, Cri-du-chat syndrome (5p), 16p11.2-p12.2 deletion, Pallister-Killian syndrome (12p isochromosome), 2q33.1 deletion, Cat Eye syndrome (22pter-22q11 tetrasomy), 16p11.2-p12.2 duplication)

☐ Patient does **NOT WANT** to know the fetus/fetuses' sex

Gestational Age at Draw: weeks days Date of blood collection: DD / MM / YYYY time

Doctor's signature: Blood drawn by:

SANCO Non Invasive Prenatal Test

Copy for Laboratory [Genomed]

Genomed

Patient Consent Statement for SANCO Prenatal Test

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Non-invasive prenatal testing (NIPT), based on the analysis of cell-free DNA from maternal blood, is not a diagnostic test, but a screening test. Therefore, test results cannot be used as the sole basis for diagnosis. Extensive further testing is necessary to verify the result to avoid irreversible decisions regarding the pregnancy. It applies to both, low and high risk reports. Your doctor will discuss with you which tests can provide an unambiguous result.

I fully understand the screening test limitations outlined in this consent form.

1. I have received explanations with regard to the disease(s) being tested for, and diagnostic significance of the genetic test(s) to be performed, from the referring physician (in accordance with art. 9, par. 2 of the Act of 6 November 2008 on Patients' Rights and the Ombudsman for Patient's Rights).
   OR
2. I declare that, exercising my right under Article 9.4 of the aforementioned Act, I have consciously waived, prior to giving consent for the above-mentioned test, the right to obtain from the physician the information referred to in Article 9.2 of the aforementioned Act, concerning the diagnostic significance of the planned genetic test and the nature of the disease that the test is intended to detect.
3. I understand that the **SANCO Prenatal Test** is a genome-wide testing, but the result of the test, to which I agree, will cover only the conditions selected in the Request form and as such may not include information about all aberrations possible to detect by genome-wide testing.
4. I understand that **SANCO test** result, regardless of the scope, will be available **within 5 working days,** counted from the first day after the day when the NZOZ Genomed laboratory receives a qualitatively and quantitatively adequate sample.
5. I acknowledge that my test result will be sent to the referring healthcare provider via an Internet service immediately upon receipt. I understand that my **SANCO Prenatal Test** results, due to medical consequences, should be reported by my physician and that **any high risk result requires a genetic counselling session**.
6. I understand that screening test results **are NOT diagnostic results,** and that a high-risk result should be **confirmed by further testing, as per my doctor's recommendation**.
7. I understand that the ability to perform the **SANCO test** depends on many factors, not all of which can be predicted or eliminated. If it is not possible to evaluate the risk of trisomy 21, 18 and 13, the test will be repeated at no costs or refunded by Genomed. I may be asked for another blood sample and therefore wait longer for mu result.
8. I have been informed that, in case of a "high risk" result of trisomy 21, 18 or 13, **Genomed will reimburse the cost of confirmatory invasive testing** (if it is not covered by the medical healthcare system), including: amniocentesis, chorionic villus sampling (CVS) or cordocentesis, and analytical genetic procedures, such as GTG karyotype, fluorescence in situ hybridization (FISH) or equivalent, up to 300 EUR/1200 PLN. The reimbursement will require the original invoice issued to the Patient by the institution performing the procedure, or will be done by paying an invoice issued directly to Genomed S.A., and after providing a copy / duplicate of the test result. For any high risk result, Genomed may offer, at its own expense, specialist consultation or confirmatory testing from invasively or postnatally collected material or may support confirmatory diagnostic testing by other means.
9. I agree to provide accurate and relevant **information regarding pregnancy and delivery** and all previous tests, such as anatomy scan or other screening or diagnostic testing performed in this pregnancy. I understand and accept that my physician or NZOZ Genomed laboratory staff may contact me for such information.
10. I consent to the **anonymous** storage and use of my sample left after **SANCO** test for the purpose of quality assurance and to improve the diagnostics and treatment of genetic diseases.

(if both boxes are left blank, consent shall be assumed) ☐ **YES** ☐ **NO**

Date:	**Name:**	**Signature:**
DD / MM / YYYY		

This is a copy for the laboratory. Ask for a photocopy or take photo, if you want to take this form home