REQUEST FORM: NON-INVASIVE PRENATAL TEST (NIPT)

SAMPLE INFORMATION	
Hospital or laboratory Label	Strict preanalytical conditions apply (see below) Date Time

LNS BARCODE LABEL
LNS label

LNS LUXEMBOURG

LABORATOIRE NATIONAL DE SANTE
NATIONAL CENTER OF GENETICS

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Forms available at www.lns.lu

PHYSICIAN REQUESTING THE TEST

Surname and first name of the doctor requesting the test

Address and country

Telephone / direct line

Fax

Date of request

Signature / Stamp

PATIENT

Birth name

First name

Married name

Sex

Date of birth

National identification number

Address and country

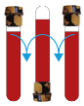
Patient covered by the CNS ☐ Yes ☐ No

*If not covered by the CNS, the patient will receive an invoice from the laboratory, which they may pass on to their insurance company, where applicable.

Copies to be sent to [Only the doctor/healthcare professional who has requested the test may give the results to the patient.]

BLOOD SAMPLE: PREANALYTICAL CONDITIONS

The mother's blood must be collected in a **Streck tube**.

Take 2 STRECK tubes	Fill the tubes	Invert 10 times	Transport: Temperature and duration
			May be kept at ambient temperature for a maximum period of 48 hours 

PATIENT INFORMATION

Personal details:

Patient Age | Pre-pregnancy weight ..... kg | Height | BMI

If the patient's BMI is >35, taking the sample at 16 weeks of gestation onwards reduces the risk of a 'no call' result due to low foetal fraction (Livergood et al., AJOG, supplement 2017).

Relevant personal or family history: .....

Type of pregnancy: ☐ natural ☐ through IVF

Date of beginning of pregnancy or expected date of delivery : (determined by measuring CRL, except in the case of IVF) : I_I_I / I_I_I / I_I_I_I_I

12-week ultrasound scan: (or attach report)
☐ Not performed
☐ No abnormalities found on ultrasound scan
☐ Abnormalities found on ultrasound scan, please specify:..... (or attach report)

Number of embryos:
☐ Singleton pregnancy: CRL: ..... mm NT: ..... mm
☐ Vanishing twin
☐ Twin pregnancy ☐ Dichorionic - Diamniotic ☐ Monochorionic - Diamniotic ☐ Monochorionic - Monoamniotic

Twin 1 (twin A): CRL: ..... mm NT: ..... mm Twin 2 (twin B): CRL: ..... mm NT: ..... mm

Treatments or clinical situations which may affect the result:
☐ Blood transfusion ☐ Immunotherapy ☐ Specific treatment :
☐ Organ transplant, stem cell transplant or bone marrow transplant ☐ Cancer .....

CLINICAL INFORMATION

PLEASE NOTE: this test must not be performed when nuchal translucency is >3.5 mm or when other abnormalities have been identified on the ultrasound scan

☐ low-risk population screening
☐ Age of mother >35 years old at the end of the pregnancy
☐ Maternal serum marker screening risk >1/1000 (please attach report)
☐ Previous pregnancy involving aneuploidy – in the patient or a 1st degree relative
☐ Balanced Robertsonian translocation in one of the parents involving chromosome 13 or 21.....
☐ Other (please contact the laboratory): .....

FORM-CYTGEN-PREANALYTICAL-EN_RequestFormNIPT Version 3.0

INFORMATION FOR PATIENTS ON NON-INVASIVE PRENATAL TESTING (NIPT)

What is NIPT?

Non-invasive prenatal testing (NIPT) is a genetic test which screens for chromosomal abnormalities in foetuses, primarily trisomies 13, 18 and 21 from a blood sample of the mother. One of the aims is to minimize the number of invasive procedures.

This technique is based on the sequencing of free-floating DNA called cell-free foetal DNA (cffDNA), in the mother's blood. cffDNA is mainly composed of DNA derived from the mother's cells, but also contains DNA from placental cells (cytotrophoblastic cells), which reflects the chromosomal makeup of the foetus. High-throughput sequencing allows for the accurate measurement of cffDNA, and thus assessment whether an extra chromosome 21 (trisomy 21) is present in the foetus. The technique also allows the detection of other chromosomal anomalies

The NIPT offered by The National Center of Genetics is a whole-genome approach provided Illumina®: Veriseq NIPT solution V2, CE-IVD certified. Results and performance of this technique are very high: the sensitivity of detection for trisomy 21,18 or 13 is around 99% . Nevertheless, NIPT remains a screening method that cannot exclude at 100% the presence of a trisomy 13, 18,21 in the foetus even if the result is negative. Further details about the performances of the method are available on our website : https://lns.lu/departement/genetique/nipt/

How does the test work in practice?

You are free to decide whether to do NIPT testing or not. NIPT can only be carried out after your 12-week ultrasound scan. Nuchal translucency should be normal. NIPT may be requested by your doctor, ideally from **12 weeks of gestation onwards and never before 10 weeks of gestation.**

The test should be requested following a consultation during which you will have had the chance to ask your doctor any questions you may have about the test. That way, you can make an informed decision based on the knowledge of the test reliability and limitations. The doctor who has requested the test will ask you to provide written consent for the test. The result of your NIPT will be sent out within 10 days only to the doctor who has requested in 10 days. The test will never be sent to you directly. NIPT is currently free of charge for patients covered by the CNS.

Results

- **If the result is negative**: the amount of foetal DNA from chromosome 13, 18 or 21 is normal and no foetal trisomy has been detected. The pregnancy should be monitored as planned.
- **If the result is positive**: the amount of foetal DNA from chromosome 13, 18 or 21 is abnormally high and there is a high probability that the foetus has one of these trisomies. A diagnostic test to confirm the result should be performed. This involves analysing the foetal chromosomes based on an amniotic fluid sample (more rarely a chorionic villus sample). Both are invasive procedures, which carry a low risk of miscarriage (0.1%).
  In very rare cases, NIPT can detect other anomalies besides trisomy 13, 18 or 21. That could be another trisomy, a loss or a gain of a part of a chromosome > 7 Mb, or a chromosomal anomaly from maternal origin. These anomalies would be reported only in case of medical interest for the ongoing pregnancy or the patient.
  - **If the result is inconclusive:** on very rare occasions (< 1 % of patients), a reliable result cannot be obtained. Depending on your clinical situation, you may be advised to repeat the test.

Limitations of NIPT

- Antenatal screening for sex chromosomes aneuploidies is not recommended by different national guidelines: these anomalies are therefore not covered by this NIPT screening.
- Although NIPT is very reliable, there are very rare cases of false positives. In those situations, the abnormality is present in the placenta but not in the foetus (cffDNA is derived from the placenta). This phenomenon is known for any chromosomal anomaly but more common for anomalies other than trisomy 13, 18 and 21. For that reason, **NIPT remains a screening test** only and a positive result should always be confirmed by foetal chromosome analysis on amniotic fluid punction.
- There are also very rare cases of false negatives, where the abnormality is present in the foetus but not in the placenta. This is why NIPT is not a substitute for foetal karyotyping or prenatal ultrasound scans.
- The reliability of NIPT is lower in twin pregnancies.
- NIPT is not intended for the the detection of balanced chromosomal abnormalities (translocations, inversions), mosaic chromosomal abnormalities, microdeletions or microduplications syndromes. This does not allow detection of point mutations or other genetics events underlying monogenic disorders. NIPT does not replace invasive foetal genetic analysis in case of ultrasound findings.

CONSENT

I have understood the degree of reliability and limitations of NIPT. I have had the opportunity to ask my doctor any questions I have on this screening test and these have been answered in a clear and satisfactory manner. I understand that and consent to my personal data being stored for medical purposes only. I understood that the results is sent solely to my doctor.

Based on the above, I hereby consent to having NIPT performed under the conditions outlined above.

I would like to know the sex of my child.	☐ Yes ☐ No
I consent that my sample and my personal data may be used (anonymously) for the purpose of clinical research, confirmatory testing or method validation by the laboratory.	☐ Yes ☐ No

Date and place : …… Signature of the patient: …….. Signature of the physician: ……