

Concepto NIPT is a non-invasive prenatal test, which is a genetic screening test pregnant women can take from week 10 of their pregnancy. The Concepto NIPT test determines the risk of the baby having the most common chromosome abnormalities.

## Properties and limitations of Concepto NIPT

- NIPT stands for Non-Invasive Prenatal Testing which determines the risk of the baby having trisomy 21, trisomy 18 or trisomy 13, sex chromosome abnormalities and gender. It is also possible to detect other chromosomal aneuploidies and genetic abnormalities, listed on the right side of this page which are offered under Concepto Absolute package, suitable for singleton pregnancies only. Gender information may also be reported at the patient's request.\*
- Although the Concepto NIPT test is highly accurate for identification of trisomies 21, 18 and 13, Concepto NIPT is NOT a diagnostic test. The test, like many others, has limitations that include the degree of false positives and false negatives results. This means that there is possibility of the presence of chromosomal abnormalities despite the negative result of the test (the result is termed as "false negative"). The result of the test may be positive for certain chromosomal abnormalities, when these are not present (the result is termed as "false positives"). Possible reasons for false positive or false negative results include: presence of mosaicism (fetus, placenta, or mother), balanced or unbalanced translocations, inversions, duplications, deletions, Robertson translocation, chromosomal aneuploidies, or maternal chromosome markers. In the case of a known maternal chromosomal abnormality listed above, the sample is not suitable for Concepto NIPT testing. A sample can be taken if the mother has recovered metastatic cancer and no tumour DNA is present in the blood.
- Concepto NIPT detects trisomies T21, T18, and T13 with a sensitivity and specificity higher than 99%. The detection rate of sex chromosome aneuploidies is 99.99%. The analysis can determine the sex of the fetus with 99.53% reliability. This information cannot be used to diagnose sexually transmitted diseases or diseases linked to sex chromosomes. Some diseases on the list of del/dup syndromes can also be caused by other genetic reasons, NIPT only detects and analyses the specific fragments according to authorized databases.
- The test is also suitable for twin pregnancy (Only with basic trisomies), egg donor and IVF pregnancies. The test evaluation in such cases is not yet possible, as the number of aneuploidies in such pregnancies is limited.
- The performance of the Concepto NIPT analysis may be affected by therapy with heparin and heparin analogues. Patients who have received a blood transfusion within one year prior to testing date, are not eligible for the Concepto NIPT test. If the mother had transplant surgery or stem cell therapy, the sample is not appropriate for Concepto NIPT testing. In case of cellular immunotherapy where exogenous DNA is introduced or human serum albumin therapy, at least 4 weeks have to pass from the last accepted therapy before collecting the blood sample.
- In cases of "vanishing twin syndrome" the samples can be accepted only if the syndrome was detected before 8th gestational week of pregnancy and at least 8 weeks have passed since its detection.
- Prior to testing, you should consult with a qualified healthcare provider as to whether any of the above listed conditions apply to you and/or advise your healthcare provider if you are already aware that any of the above listed conditions apply to you. Test results should always be interpreted in the context of other clinical and family information. Never make decisions related to your pregnancy without first consulting your doctor.
- In case that the patient has BMI>40 the failure rate of the Concepto NIPT test can be increased due to a lower fetal fraction.
- Fetal fraction below 3.5% can lead to the failure of analysis and no official result can be provided. Official results with 3.5% fetal fraction and above have specificity and sensitivity as indicated.
- Concepto NIPT test results do not exclude the presence of all abnormalities of chromosomes and birth defects.
- Also, with the test we cannot detect all abnormalities caused by chromosomal polyploidy (triploid, tetraploid, etc.), chromosomal balanced translocation, inversion, ring, UPD, monogenic/polygenic disease, etc. This test cannot exclude the fetal mosaic chromosomal diseases.

NIPT Standard	NIPT Advance	NIPT Absolute
Down's Syndrome (Trisomy 21)	Down's Syndrome (Trisomy 21)	Triple X syndrome
Edward's Syndrome (Trisomy 18)	Edward's Syndrome (Trisomy 18)	Trisomy 21, 18 and 13
Patau's Syndrome (Trisomy 13)	Patau's Syndrome (Trisomy 13)	Trisomy 21, 18 and 13
Gender- Male (XY) Female (XX) - (Optional)	Gender- Male (XY) Female (XX) - (Optional)	Sex chromosome aneuploidies* - X(O) - Turners Syndrome
Sex chromosome anomalies:	Sex chromosome anomalies:	XXY - Klinefelter's syndrome
Turner's Syndrome (X(O))	Turner's Syndrome (X(O))	XXY - Jacob's syndrome and XXX - Triple X syndrome
Klinefelter's syndrome (XXY)	Klinefelter's syndrome (XXY)	High Demand 92 Microdeletions
Jacob's syndrome (XXY)	Jacob's syndrome (XXY)	
Triple X syndrome (XXX)	Triple X syndrome (XXX)	
	High Demand 6 Microdeletions	

- \* Non-Invasive Prenatal Testing for Trisomy 21, 18 and 13 – Clinical Experience from 146,958 Pregnancies, Wei Wang et al, Journal of Ultrasound in Obstetrics and Gynecology
- BMC Med Genomics. 2012 Dec 1;5:57. doi: 10.1186/1755-8794-5-57.
- Ultrasound Obstet Gynecol. 2014 Jul;44(1):17-24. doi: 10.1002/uog.13361
- J Matern Fetal Neonatal Med. 2014 Dec;27(18):1829-33. doi:10.3109/14767058.2014.885942
- Ultrasound Obstet Gynecol. 2015 May;45(5):530-8. doi: 10.1002/uog.14792
- From in-house data. Internal analysis shows a sensitivity of over 90% (cfDNA9.5%) in selected del/dup syndromes with abnormal size over 3 Mb

### Performance statistics:

SYNDROME	SENSITIVITY	SPECIFICITY
Trisomy 21	99.17%	99.95%
Trisomy 18	99.24%	99.95%
Trisomy 13	>99.9%	99.96%
CVI	>10 Mb >99.9%	99.97%
CVI	<10 Mb >99.9%	99.86%
Fetal sex	99.53%	99.20%
Sex Chromosome Aneuploidies		
XXY	>99.9%	99.6%
XXY	>99.9%	99.6%
XXX	>99.9%	99.6%
X(O)	>99.9%	99.6%

Generally, resample rates are 2.18%, and no-call rate 0.069%.

CONCEPTO NIPT  
Fast. Accurate. Accessible.



6 MICRO DELETIONS

CONCEPTO NIPT  
Fast. Accurate. Accessible.



92 SYNDROMES OF DELETION AND DUPLICATION

## Concepto NIPT test result information

- Your test results will be sent to the healthcare provider at which you ordered the Concepto NIPT test typically within 5 working days from receiving in the lab. In a small number of cases the sample must be reanalyzed and you may experience a mild delay in receiving your report.
- LOW RISK** means there is a very low chance of the baby having an abnormal number of chromosomes for the conditions tested for.
- HIGH RISK** indicates the baby has an increased chance of having one of the genetic conditions tested for. In the case of HIGH RISK RESULTS, further invasive testing, such as amniocentesis, is required to confirm the test results.
- RESAMPLE REQUIRED** – In a small number of cases (around 2.18% of all samples received) we are unfortunately unable to analyze the fetal DNA in enough detail in order to provide you with a result. In such cases we require a new blood sample in order to run a new test. There is no additional cost for resampling.
- NO CALL** means that we have been unable to detect a result despite resampling. The incidence of this happening is extremely low at only 0.069% of all samples received. In such cases, a refund of the Concepto NIPT test will be issued.

## Concepto NIPT Data Protection

- As a patient, I am aware that this is informative document provided according to the General Data Protection Regulation (GDPR), the Controller of my personal data is Concepto NIPT. The data will only be shared with patient consent to our genetic consultants when high risk results occur.
- By signing the consent on Page 2, I expressly agree and give permission for your personal data included in this test request form (including, without limitation, my name, address, information about my pregnancy, and other relevant information), as well as my blood sample, to be sent to Concepto NIPT Limited (including its partners and affiliates) for the purpose of performing the Concepto NIPT test. In the event I withdraw my consent or request not to receive the results of the test, Concepto NIPT Limited will use commercially reasonable efforts to promptly destroy my blood sample in compliance with applicable laws and regulations, and Concepto NIPT Limited's standard protocols for sample destruction. I agree that in the event Concepto NIPT Limited performs the NIPT test selected on this form, Concepto NIPT Limited may store my personal data (including the test results) and remaining sample (if any) for the applicable legally required time period.
- I am informed that I have the right to request access, correction, deletion, or restriction of the processing of my personal data from the Controller of personal data to object to the processing or transfer of data at any time without prejudice to the legality of data processing. I can object the processing of personal data, however in such case I can exercise any of my rights by writing an email to support@conceptoclinic.co.uk .

## Informed Consent

- I understand that gender information will be provided to me based on my request.
  - I understand that this test is not intended to provide a final diagnosis and should, in case of a high-risk result, not be relied on as sole evidence for a diagnostic conclusion. I understand that the sensitivity rate of this test is not 100% and may, due to the biological, technical or any other difficulties, result in a 'false positive' or a 'false negative' result.
  - I understand that the chance of a false positive or false negative result cannot be completely excluded and can be increased due to: vanishing twin pregnancy, abnormal reproductive history, heparin medication, BMI>40, the history of benign or malignant tumor. With my signature I confirm that I will still take this test and be willing to take responsibilities for the relevant risk.
  - I understand that the result of the test does not eliminate the possibility of other genetic abnormalities that are not tested with the Concepto NIPT test.
  - I understand that the results of the Concepto NIPT test are not a diagnosis or confirmation that the fetus has or does not have chromosomal aneuploidies, but the results of the Concepto NIPT test provide an assessment of the risk of genetic abnormalities in the fetus.
  - I understand that in case of BMI>40 there is a higher chance for failure of the analysis due to lower fetal fraction.
  - During the consultation before the test and before signing this consent statement, I have had enough time and opportunity to get answers to all my questions regarding the Concepto NIPT test and I understand all the information.
  - I agree to provide accurate information about all previous tests such as ultrasound/other screening/diagnostic tests performed in this pregnancy.
  - I consent to have my test results sent to the undersigned healthcare provider, or their place of business, to an address provided by them.
  - I confirm that I know a Concepto healthcare employee may contact me about the outcome in case of the high-risk result.
  - I confirm that I am fully capable of business (able to enter in contracts). If not, on my behalf, this statement is signed by a legal guardian.
  - I confirm that I am currently not aware of having any blood-borne infectious disease.
- I am aware that I can ask the chosen doctor who provides me with NIPT counseling not to inform me of the results of the test;
- I am aware that a refund for the services rendered is not possible, except in the case of especially justified and demonstrated unforeseen circumstances as mutually agreed upon by all stakeholders;
- I have fully read and understood the statement and consent to the processing of personal data
- I have read and understood the features, limitations, and risks of the Concepto NIPT test;
- I give express consent to the processing of personal data by the company Concepto NIPT for the performance of the Concepto NIPT test.
- The undersigned patient and healthcare professional confirm that the above-mentioned contents are true, and the doctor has fully fulfilled the obligation to inform about what the test covers and what its limitations and requirements are.

**Patient Information:**

Name

Phone

Address

Date of Birth (DD/MM/YY)

Barcode (Please stick the barcode provided with your kit)

Email address (result will be emailed to this address)

Sample Date and time (DD/MM/YY HH:MM)

**Partner Information:**

Partner (Clinic) Name

Partner Location

Email Address

Phone

Address (with Post Code)

**Test Details**

NIPT Test type	Includes
NIPT Standard <input type="checkbox"/>	Trisomy 21, 18 and 13, Sex chromosome aneuploidies* – X(0) –Turners Syndrome, XXY – Klinefelter’s syndrome, XYY– Jacob’s syndrome and XXX– Triple X syndrome.
NIPT Advance <input type="checkbox"/>	Trisomy 21, 18 and 13, Sex chromosome aneuploidies* – X(0) –Turners Syndrome, XXY – Klinefelter’s syndrome, XYY– Jacob’s syndrome and XXX– Triple X syndrome. 6 Microdeletions
NIPT Absolute <input type="checkbox"/>	Trisomy 21, 18 and 13, Sex chromosome aneuploidies* – X(0) –Turners Syndrome, XXY – Klinefelter’s syndrome, XYY– Jacob’s syndrome and XXX– Triple X syndrome. 92 Microdeletions

Fetal sex\* to be reported  Yes  No

\*In case of twin pregnancies, male results apply for one or both fetuses, female results apply to both fetuses.

Twin pregnancies are only eligible for basic trisomies.

Gender Test  Blue or  pink



**Limitation:** BlueOrPink Gender Test can not be performed in case of vanishing/ demised twin


Gestational age \_\_\_\_\_ weeks \_\_\_\_\_ days as on \_\_\_\_\_ (sample date in DD/MM/YY HH:MM)

Number of fetuses (Select any one)  1  2 IVF Pregnancy  Yes  No

Ultrasound findings (If applicable)

**You should have completed 10 weeks of pregnancy for NIPT, or 6 weeks for the Blue or Pink gender test.**


By signing below, I consent that I have read, understood and followed the informed consent section, in its entirety. I understand all the materials and products provided by Concepto NIPT Limited (Concepto NIPT) are for information purposes only. I understand that Concepto NIPT Limited is not liable for any actions that might be taken by myself or anyone else as a result of using this service. I agree to release, indemnify and hold harmless the company, and all of its employees and stakeholders from all liabilities associated with products & services purchased. I have read and/or have had read to me the information regarding Concepto tests. I have read, understood, and/or received an explanation of the nature, purpose, duration, and foreseeable effects and what I will be expected to do. My questions have been answered satisfactorily.



PATIENT/LEGAL GUARDIAN: \_\_\_\_\_

Date (DD/MM/YY HH:MM): \_\_\_\_\_

Signature: \_\_\_\_\_



Healthcare provider: \_\_\_\_\_

Signature: \_\_\_\_\_