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New INSIGHTS

Neuberg DIAGNOSTICS | CENTER FOR GENOMIC MEDICINE

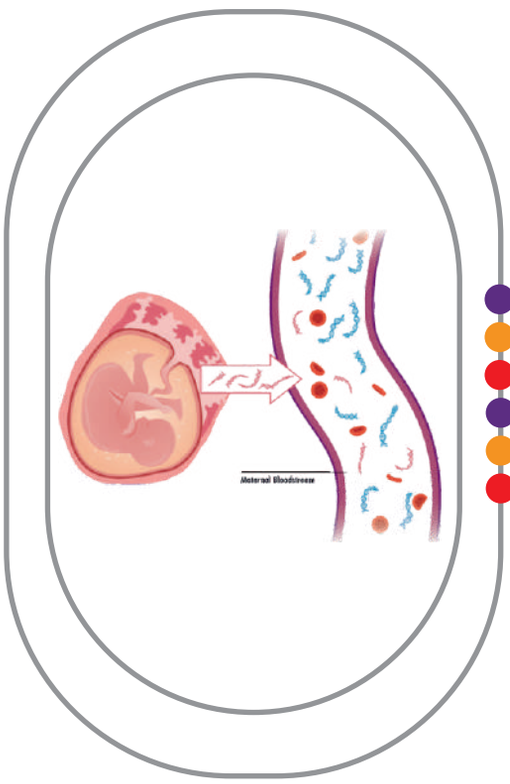
CHROME

The most preferred non-invasive prenatal test

Non-Invasive Prenatal Screening (NIPS)

Serial number : 022 Edition : 1. 2022

INTRODUCTION:



01 Cell- free DNA fragments (cfDNA) - short fragments of DNA, which can be found circulating in the mother's blood.

02 Fetal cfDNA comprises 3-13% of total DNA in maternal plasma[1]

03 NIPS evaluates cfDNA released by cells from the placenta[1]

04 Highly sensitive as compared to traditional maternal serum screening and is non-invasive: No risk of miscarriage[1]

05 NIPS can screen for common chromosomal aneuploidies, rare chromosomal aneuploidies and some of the common chromosomal microdeletions.[1]

06 Currently, routine prenatal screening for aneuploidies of chromosome 13,18 and 21 is recommended by American College of Obstetricians and Gynecologists [2].

NIPS IN PRENATAL CARE:

NIPS has been accepted as routine prenatal care owing to its high specificity, sensitivity and detection rate with minimum false positive rate.

RECOMMENDED FOR

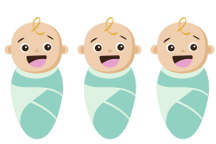
Advanced maternal age

Abnormal maternal serum screen results

Previous pregnancy with chromosomal abnormalities

In every pregnancy to rule out common chromosomal aneuploidies.

NOT SUITABLE FOR



Multiple Gestations (more than 2 fetuses)/ Vanishing Twins [3]

Fetal Anomalies on Utrasound/Known Genetic Anomalies that cannot be diagnosed with NIPS [3]

Gestational age <9 weeks

If an expectant mother has recently (upto 3 months ago) received a blood transfusion, stem cell therapy, or organ transplantation.[3]

VALIDATED FOR

Singleton/ Twin Pregnancy

IVF/Donor Egg or Surrogate

Gestational Age as early as 9 weeks

Fetal Fraction as low as 2% in singleton pregnancy and 4% in twin pregnancy.

About CHROME NIPS:

Chrome NIPS uses Whole Genome Sequencing (WGS) technology on Illumina platform. It is supported by in-house customized bioinformatics pipeline to screen fetal chromosomal aneuploidies.

NCGM offers below listed testing options for NIPS

CHROME Focus

Targeted analysis for aneuploidies of five common chromosomes 13 / 18 /21 /X / Y

CHROME Comprehensive

Analysis for aneuploidy detection of all 23 pairs of chromosomes

CHROME Plus

Analysis of all 23 pairs of chromosomes for aneuploidy and common microdeletion syndromes:
1.DiGeorge (22q11.2),
2.Angelman (15q11.2),
3.Prader-willi (15q11.2),
4.Cri-du-chat(5p),
5.Wolf Hirschhorn (4p),
6.1p36 deletion

Turn Around Time (TAT):

After the receipt of the sample, NIPS results are expected within 5-7 working days*.

*The laboratory usually ensures timely dispatch of reports; however certain un-anticipated delays may occur for which the laboratory should not be held liable for. Delay in TAT/ requirement for the repeat sample will be informed within 7 working days after the sample receipt.

Sample Details :

Sample Requirements:

- A 10 ml maternal blood sample in a cell-free DNA (cfDNA) collection tube (Streck tube).

Sample Rejection Criterion:

- Sample not received in cfDNA tube
- Hemolyzed sample
- Inadequate blood sample

Documents Required:

- Test requisition form (TRF) and Form G
- Ultrasound report
- Maternal serum screening report (If available)

Expected test outcomes:

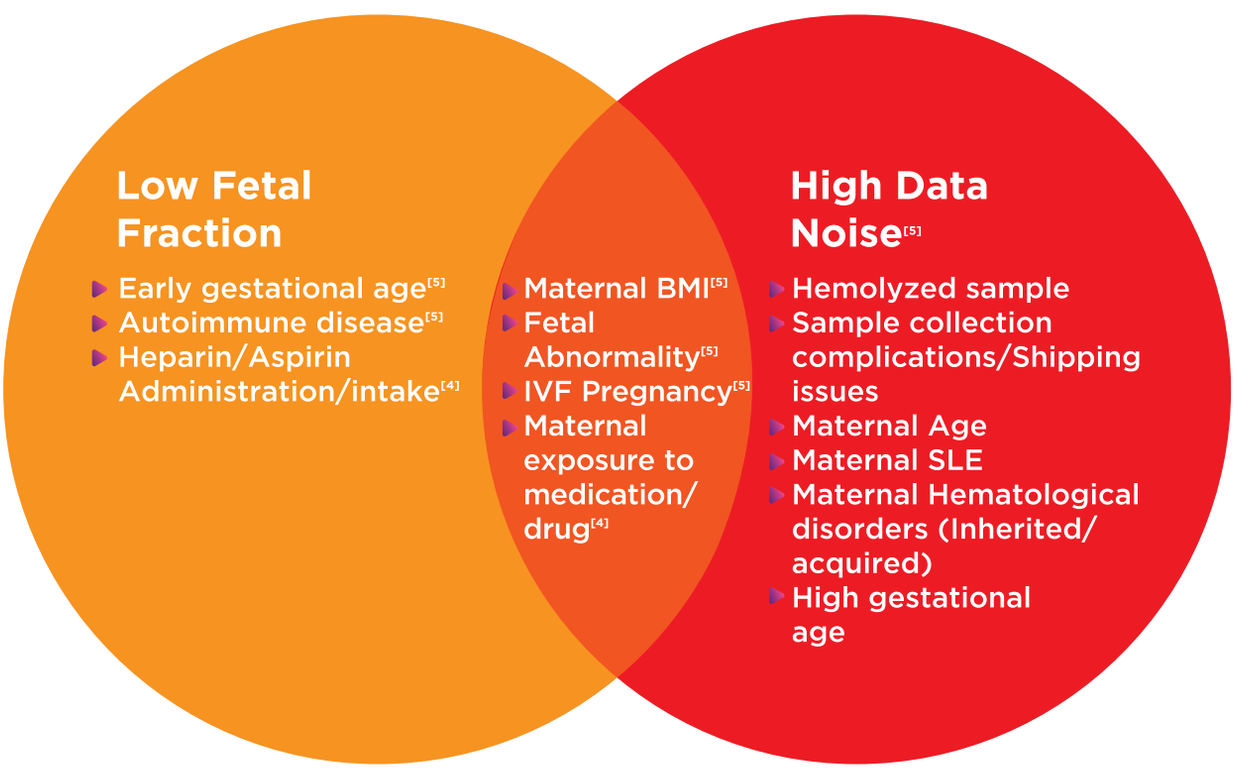
Low Risk
- Substantially decreases the risk of targeted aneuplodies
- Follow up ultrasound examinations recommended

High Risk
- Increased risk for the reported chromosomal abnormality
- Amniocentesis is strongly recommended to confirm the findings.

No call results - Repeat Sampling (<1%)
- Laboratory could not generate the results with the provided sample and repeat sampling is required.
- Reasons could be low fetal fraction/High data noise

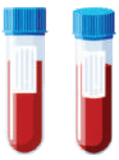
Invasive Testing Recommended
- Even after repeat sampling, same issues persist for NIPS failure

Factors affecting NIPS results:



Body mass index (BMI) Systemic lupus erythematosus (SLE)

CHROME highlights



Tested Samples
> 35,000

Screens
All **23**
chromosomes

Performed
As early as **9** weeks

Fetal fraction
As low as **2%**

Sensitivity/Specificity
>99%
for T21, 18 and 13

Turnaround Time
5-7
working days

Test Limitations:

- The NIPS CHROME FOCUS is validated for chromosomes 21,18,13, X and Y in singleton and twin gestations at gestational age of atleast 9 weeks.
- The NIPS CHROME FOCUS is not intended, neither validated for diagnosis nor for use in pregnancies with more than two fetuses, mosaicism, partial chromosomal aneuploidy, translocations or maternal aneuploidy.
- The NIPS CHROME FOCUS is a screening test and the positive predictive value is not 100%. Hence confirmation of high risk results is recommended by invasive testing.
- A LOW RISK test result reduces the risk of fetal aneuploidy but it does not ensure an unaffected fetus. It also does not negate the possibility that the fetus may be affected with sub-chromosomal abnormalities, gene defects and birth defects. Need for an invasive testing may arise later in pregnancy.
- False positive and false negative results are known. Factors affecting test accurancy include confined placental mosaicism, maternal neoplasms and low fetal fraction.
- The lower limit of detection for singleton pregnancies is at fetal fraction of 2%. The lower limit of detection for twin pregnancies is at fetal fraction of 4%
- The CHROME - NIPS is a CAP (College of American Pathologists) accredited test.

DISCLAIMER! : NIPS is a screening test, a low risk does not exclude the above evaluated disorders.

References:

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PARTNERS IN HEALTH

DR. SHIVA MURARKA

Senior Scientist (Molecular Genetics)
PhD Reproductive Sciences
shiva.murarka@ncgmglobal.com

DR. UDHAYA KOTECHA

Head of Division Inherited Disorders (NGS)
Clinical Geneticist
M.D. Pediatrics
Fellowship in Medical Genetics
udhaya.kotecha@ncgmglobal.com

DR. SHEETAL SHARDA

Director - Clinical Genomics
Development & Implementation
M.D. Pediatrics
D.M. Medical Genetics
sheetal.sharda@ncgmglobal.com

DR. PARTH SHAH

Senior Advisor
MD (Hematology and Medical Oncology)
parth.shah@neubergdiagnostics.com

DR. SANDIP SHAH

Consultant Pathologist
M.D (Pathology & Bacteriology)
Laboratory Director
drsandip@neubergdiagnostics.com

FOR MORE DETAILS, CONTACT US AT

079 61618111
079 40408181

ncgmglobal.com

Neuberg DIAGNOSTICS

• India • UAE • South Africa • USA