

FAMILY HISTORY

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Parents karyotype report: Done Not Done

Karyotype findings

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ATTESTATION

I attest that the information given in this form is true and this patient has been informed about the diagnostic procedure & tests.

Sign of wife a Husband

Signature

Note: Tissue: Skin or solid tissue obtained by sterile biopsy should be placed in normal saline inside a sterile container. Place it in a box with cool packs and transport to the Laboratory.

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Testing of Product of Conception (POC) by Next Generation Sequencing (NGS)

What is POC testing by NGS?

- Humans have 46 chromosomes. An extra or a lesser number of chromosomes may result in early miscarriage. Chromosomal abnormalities contribute to around 50% cases of pregnancy losses in the first trimester.
- Testing of product of conception by next generation sequencing helps identify if any chromosomal abnormalities were responsible for miscarriage.
- Testing of product of conception by next generation sequencing offers multiple advantages as compared to conventional karyotype.

Neuberg Centre for Genomic Medicine (NCGM)

Near GTPL House, Opp. Armedia, Sindhu Bhavan Road, Bodakdev, Ahmedabad 380059
Phone: +91-6357244307, 079-61618111 | Email: contact@ncgmglobal.com | Web: www.ncgmglobal.com

Advantages:

- In karyotype, around 70% of the tissue cultures fail to grow resulting in no results. However, analysing a POC sample by NGS method does not require cell culture.
- Risk of altered results due to maternal cell contamination is reduced. The accuracy of detection of chromosomal aneuploidies is >99% by NGS.
- Accurate results are obtained in 98% of cases.
- The results are obtained within 2 weeks.

Limitations:

- The NGS technology cannot detect balanced translocations
- The test can only detect chromosomal abnormalities. A separate test needs to be performed to identify point mutations.
- Maternal cell contamination can interfere with the results in case of blood stained tissue. A separate test needs to be performed to rule out maternal cell contamination.

Turn Around Time (TAT):

- The results are expected within 2 weeks. The laboratory usually ensures timely dispatch of reports, however certain un-anticipated delays may occur for which the laboratory must not be held liable for. Delay in TAT/ requirement for the repeat sample will be informed in a week's time after the sample receipt.

Patient consent:

- I understand that the data derived from my genetic testing may be de-identified and stored indefinitely as part of a laboratory database.
- I understand my de-identified data/ sample may be used for quality control, test development and laboratory improvement purposes which also include research, scientific presentations & publications.
- I have read and understood / have been explained the above information in the language of my understanding and permit the Neuberg Centre For Genomic Medicine to perform the recommended genetic analysis.
- I have had an opportunity to ask questions to my healthcare provider regarding this test, including the reliability of the test results, risk and the alternatives prior to giving my informed consent.

Name:

Signature:

Relationship to the expectant mother:

Date, Time and Place:

Clinician name and signature:

References:

- Tamura, Yuki, et al. "Chromosomal copy number analysis of products of conception by conventional karyotyping and next generation sequencing." *Reproductive medicine and biology* 20.1 (2021): 71-75.
- Robberecht, C., Schuddinck, V., Fryns, JP. et al. Diagnosis of miscarriages by molecular karyotyping: Benefits and pitfalls. *Genet Med* 11, 646–654 (2009). <https://doi.org/10.1097/GIM.Ob013e3181abc92a>
- American College of Obstetricians and Gynecologists. "Committee on Practice Bulletins—Obstetrics; Committee on Genetics; Society for Maternal–Fetal Medicine." *Practice bulletin* 162: 976-78.

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