

Report of Preimplantation Genetic Testing- Aneuploidy (PGT-A)

Patient Information

Name: Mrs. Swarnambha S	Age/Gender: 41 Yrs/Female
Referring Physician: Dr. Snehadarshini Karanth	Referring centre: Indira IVF Hospital Pvt. Ltd- Bangalore
Embryologist: Ms. Manisha	Hospital ID: NGB0000011
Specimen Type: Embryo Biopsy	Date of collection: 23/11/2024
Sample ID: 2400258973	Date received: 20/12/2024
Patient ID: 1102435605	Report Date: 31/12/2024
Embryo biopsy ID/s: SL1	

Clinical Reason

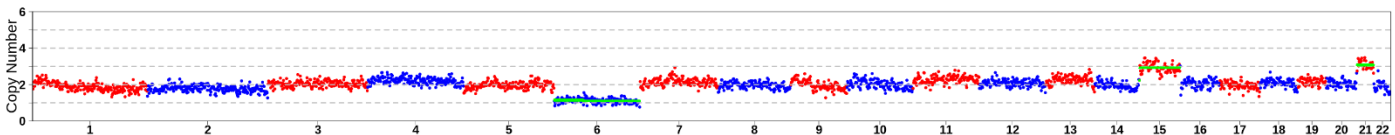
The embryo biopsy sample **SL1** is being evaluated for the chromosomal aneuploidies due to advanced maternal age, using PGT-A.

Results & Interpretation

The embryo biopsy sample **SL1** is not recommended for embryo transfer based on the PGT-A results.

Sample Id/ Embryo ID	Karyotype	Interpretation	Recommendation for Transfer
2400258973 [SL1]	47, XN -6 (x1), +15 (x3), +21 (x3)	Aneuploid	Not Recommended Based on PGT-A results

2400258973 [SL1]



PGT-A details

Transfer Recommendations	<p>Short Summary explaining transfer recommendations</p> <p>The transfer recommendations are based solely on the results of the aneuploidies or chromosomal anomalies analysed in this report.</p> <p>This does not take into consideration any other genetic defects such as small deletions, duplication, mutations or epigenetic defects ('small' refers to <10MB in size specific to the particular chromosome).</p> <p>However, the final decision to transfer a particular embryo is that of the clinician and the embryologist based on their clinical and histological correlation.</p>
Analysis Performed	Preimplantation Genetic Testing of the embryo biopsy sample for the chromosomal aneuploidies using ChromGo analysis software.
Method of Analysis	PGS Aneuploidy using Yikon PGS Kit with the Ion S5 System.

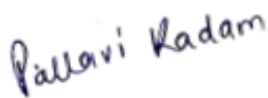
Interpretation:

The results have identified **Monosomy of chromosome 6, Trisomy of chromosome 15 and Trisomy of chromosome 21** in embryo biopsy sample **SL1**.

These results indicate that the embryo biopsy sample **SL1 is not recommended** for transfer based on the PGT-A result.

Recommendations

1. Clinical correlation and Genetic counseling is recommended.



Pallavi Kadam

Lab Incharge - NGS



Dr. Durgadatta Tosh

Head – NGS Division

PRECAUTIONS: Although all precautions are taken during DNA tests, the currently available data indicate that the technical error rate of all types of DNA analysis is approximately 2%. It is important that all clinicians or persons requesting DNA diagnostic tests are aware of these data before acting upon these result.

Disclaimers

- The PGT- A with NGS procedure is used to detect Aneuploidy only in the sample(s) provided.
- Based on the findings, it will be recommended whether or not the embryo from which the samples were taken is/ are suitable or implantation.

- A “Normal” result indicates that no Aneuploidy was detected in that sample within the limits of the present test. An “Abnormal” result means that one or more Aneuploidy has been detected.
- Although the test provides transfer recommendation based on the presence or absence of Aneuploidy; the findings should be correlated clinically by the physician in charge before deciding to carry out the embryo transfer.
- In some cases, following PGT-A by NGS, no Euploid (unaffected) embryos may be found in the samples provided. In such cases, transfer of affected embryos is not recommended.
- If the laboratory is unable to diagnose the genetic status of all provided embryos; in such cases transfer of undiagnosed embryos is not recommended.
- Segmental mosaic chromosomal aneuploidies could be due to poor embryo quality or an artifact appeared during biopsy procedure.
- Some of the clinical indications (inclusion criteria) for PGT-A are recurrent miscarriage, repeated implantation failure and advanced maternal age. While the exclusion criteria are low total antral follicle count and poor embryo quality.
- In accordance to the Pre-Conception and Pre-Natal Diagnostic Techniques (PCPNDT) Act, 1994, Govt. of India; the sex of the sample analyzed will not be disclosed in the report.

References

1. Jonatan B. *et al.*, Risk of Misdiagnosis Due to Allele Dropout and False-Positive PCR Artifacts in Molecular Diagnostics. *The Journal of Molecular Diagnostics*, Volume 17, Issue 5, 505 – 514.
2. Rycke D. M. *et al.*, Preimplantation Genetic Testing for Monogenic Disorders, *Genes* 2020, 11, 871.
3. Masset H. *et al.*, Multi-centre evaluation of a comprehensive preimplantation genetic test through haplotyping-by-sequencing, *Human Reproduction*, Volume 34, Issue 8, August 2019, Pages 1608–1619.
4. PGDIS Guidelines: https://www.pgdis.org/docs/newsletter_071816.html
5. Viotti M. Preimplantation Genetic Testing for Chromosomal Abnormalities: Aneuploidy, Mosaicism, and Structural Rearrangements. *Genes (Basel)*. 2020 May 29;11(6):602.
6. ENSEMBL: <http://www.ensembl.org>
7. Amberger J. S. *et al.*, OMIM.org: leveraging knowledge across phenotype–gene relationships, *Nucleic Acids Research*, Volume 47, Issue D1, 08 January 2019, Pages D1038–D1043,

*****End of the Report*****