

Report of the Non-Invasive Chromosome Screening (NICS)

Patient Information


Name	: Mrs. PREETI SINHA	Hospital ID	: LKO0006408
Spouse Name	: MANISH KUMAR	Patient ID	: 10022132491
Date of Birth/Age	: 08/08/1988	Sample ID	: 2300020329
Referring Doctor	: DR. PAWAN YADAV	Sample collected on	: 27/01/2023
Embryologist	: Mr. Lovkush Kumar Verma	Sample received on	: 06/02/2023 14:45
Sample Type	: Spent Blastocyst Culture Media	Report released on	: 21/02/2023 12:18

NICS is an advanced pre-implantation genetic screening solution which offers result with high accuracy by avoiding invasiveness and potential embryo harm along with improved test sensitivity and reliability.

Clinical Indications

Mrs. PREETI SINHA has undergone NICS test due to miscarriage and to screen for euploid embryos.

Result Summary

Embryo ID	Results	Interpretation	Transfer Recommendation	Transfer Priority
2300020329 (PM1)	46,XN. -N($\times 1$,mos, $\sim 58\%$), -10q(q11.21 \rightarrow q21.1, ~ 14 Mb, $\times 1$), -18p($\times 1$,mos, $\sim 36\%$). SCA detected	Aneuploid		Not recommended

Patient name:

Patient ID:

Terms Used in Results

- x1: Monosomy
- x2: Disomy
- x3: Trisomy
- Mos: Mosaicism: % (percentage of mosaicism)
- SCA: Sex Chromosome Aneuploidies

Terms Used in Interpretation



• **Euploid:**
No aneuploidy detected within the LOD of the Test



• **LowLevelMosaicism:**
<=50 Mosaicism present for aneuploidies (specific percentage for each is given in the result)



• **Aneuploid:**
Sample shows Aneuploid for specific chromosomes with LOD of Test

Transfer Recommendation



• Recommended for Embryo Transfer



• Transfer with counselling support about increased risk for aneuploidies

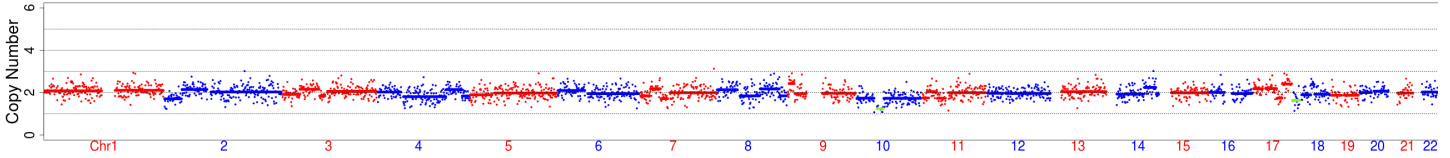


• Not Recommended for transfer

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CNV Chart for Embryo: 2300020329 (PM1)



Ch.	1	2	3	4	5	6	7	8	9	10	11	12
CNV (+/-)	NA	NA	NA	NA	NA	NA	NA	NA	NA	-	NA	NA
Region & Size	NA	NA	NA	NA	NA	NA	NA	NA	NA	q arm	NA	NA
% Mosaic	NA	NA	NA	NA	NA	NA	NA	NA	NA	0	NA	NA

Ch.	13	14	15	16	17	18	19	20	21	22	SCA
CNV (+/-)	NA	NA	NA	NA	NA	-	NA	NA	NA	NA	-
Region & Size	NA	NA	NA	NA	NA	p arm	NA	NA	NA	NA	W.C
% Mosaic	NA	NA	NA	NA	NA	~36	NA	NA	NA	NA	~58

W.C.: Whole Chromosome, N: Normal, +: Gain, -: Loss, NA: No Aneuploidy, 0: Absence of Mosaicism

Results	Details	Interpretation	Transfer recommendation	Transfer Priority
46,XN. -N(×1,mos,~58%), -10q(q11.21 → q21.1,~14Mb,×1), -18p(×1,mos,~36%). SCA detected	Mosaic sex chromosomal aneuploidy detected. Mosaic monosomy of chromosome 10q and 18p detected.	Aneuploid	×	Not recommended

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Test background

IVF embryos are prone to chromosomal aneuploidies. Chromosomal aneuploidies could cause early pregnancy loss or other abnormalities among IVF pregnancies. The occurrence of chromosomal abnormalities in embryos is substantially higher in patients with advanced maternal age, parents with recurrent pregnancy loss, or those who carry chromosomal aberrations such as translocations; all of which result in inferior clinical pregnancy outcomes.

Non-Invasive Screening of embryos in IVF-ICSI (In vitro fertilization- Intracytoplasmic Sperm Injection) for chromosomal aneuploidies can be performed for all 23 chromosomes.

NICS

- NICS is the **non-invasive next-generation sequencing chromosome screening** test introduced by Lilac Insights in elite partnership with Yikon Genomics. This novel technique enables pre-implantation genetic testing to be performed in an **easier, safer and most efficient way**.
- NICS, helps to identify the embryos that are most suitable for transfer during IVF-ICSI to give you the best chance of implantation and a successful pregnancy.
- This test works by identifying embryos with extra or missing chromosomes or parts of chromosomes.
- Embryos with chromosomal abnormalities may fail to implant, or may lead to miscarriage or the birth of a child with a chromosomal conditions like Down syndrome, Patau Syndrome & Edward's syndrome etc.

Methodology

- Non-Invasive Chromosome Screening (NICS) is a screening method of aneuploidies in ICSI- IVF Embryos using cell-free embryonic (cf)-DNA. (cf)-DNA is a mixture of DNA from Inner Cell Mass (ICM) & Trophectoderm (TE) Cells in at the Blastocysts stage (day-5). Spent Culture medium was collected in the sample collection kits as per specifications [1, 2] and shipped to the lab for analysis.
- Pre-PCR and Whole Genome Amplification (WGA) by MALBAC PCR was carried out; followed by Library Preparation, quality check (Qubit Fluorometer) then NGS Sequencing using Ion PI™ Chip on an Ion Proton sequencer (Thermo Fisher). Sequencing was carried out so as to generate 1.5 Million raw reads per sample. The data was then analyzed using ChromGo™ Software (from Yikon Genomics) to determine the valid reads, data QC and aneuploidy calling.

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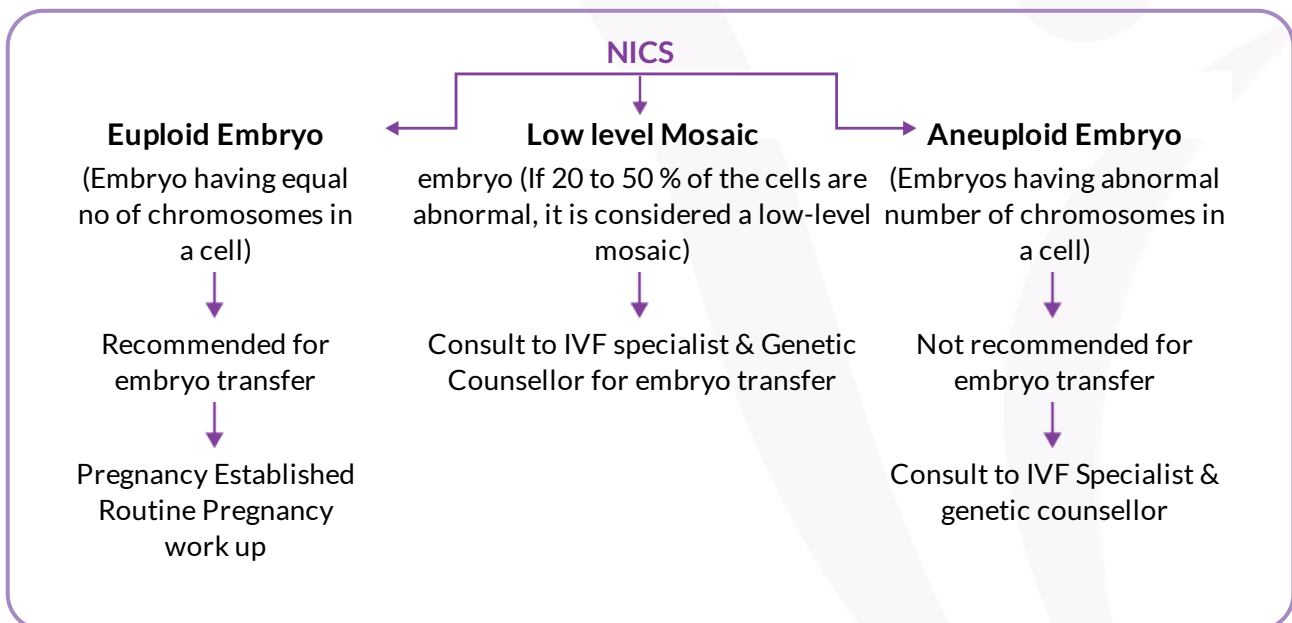
NICS Features

NICS by Lilac Insights, the only Indian Genetic Diagnostic Lab to offer highly efficient next gen test with following features:

- **Non-Invasive:** Biopsy free, hence no potential influence on embryo development.
- **Successfully Validated:**
- **AI Auto-Analysis Module:** Established by our AI study on over 1000 culture medium samples & corresponding embryos, our AI analysis improves the performance of the screening (Sensitivity, specificity, PPV & NPV).
- **Latest Technology:** Elite Partnership with Yikon Genomics, one of the world's largest reproductive health genetic laboratories holding the patent to MALBAC (Multiple Annealing and Looping-Based Amplification Cycle) technology. This technology provides high-sensitivity, high genome coverage, and high uniformity approach to DNA amplification.
- **Expert Team of Genetic Counselling:** Genetic Counsellors at Lilac Insights assists every individual/couple & family to make an informed decision about options available for screening and testing, before and during pregnancy. Also, assist to understand medical and genetic information.

What Next After NICS?

A novel approach to identify the embryos that are most suitable to transfer during IVF-ICSI, to give you the best chance of implantation to boost the success; and to avoid the selection of embryos that are not suitable for a transfer which may lead to implantation/pregnancy failure.



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Disclaimer

- As NICS test is not a confirmatory diagnostic test, the possibility of false positive or false negative results cannot be denied. The results issued for this test does not eliminate the possibility that this pregnancy may be associated with other chromosomal or sub- chromosomal abnormalities, birth defects and other complications.
- It is assumed that the details provided along with the sample are correct.
- The manner in which this information is used to guide patient care is the responsibility of the healthcare provide, including advising for the need for genetic counselling or additional/ subsequent diagnostic testing.
- Some samples may not yield interpretable/reportable results.
- NICS may also result in no embryos being recommended for transfer.
- NICS cannot detect triploidy.
- NICS cannot detect balanced translocations and inversions.
- Low level mosaicism (<50%) is also reported and needs to be correlated clinically. This maybe caused due to cf- DNA from TE cells.
- Pre and post NICS genetic counseling is strongly recommended.
- NICS cannot detect CNVs less than 10 Mb in size*.
 - a. The NICS Test has been performed to obtain a resolution of 10 Mb per chromosome for each sample, however this is based on the number of valid reads generated per sample.
 - b. The Valid reads generated for each sample analyzed is given in the table below along with reporting resolution:

Sample ID	Valid Reads	Reporting Resolution
2300020329 (PM1)	470721	p/q arm

Pallavi Kadam

**Scientific Officer
Genomics**

Dr. Madhavi Pulsakar

**Dr. Madhavi Pulsakar, Ph.D.
General Manager: Genomics**

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References

1. Non-invasive pre-implantation genetic testing for aneuploidy in spent medium may be more reliable than trophectoderm biopsy. Lei Huang, Berhan Bogale, Yaqiong Tang, Sijia Lu, Xiaoliang Sunney Xie, Catherine Racowsky. Proceedings of the National Academy of Sciences Jul 2019, 116 (28) 14105-14112; DOI: 10.1073/pnas.1907472116
2. Fang, R., Yang, W., Zhao, X. et al. Chromosome screening using culture medium of embryos fertilized in vitro: a pilot clinical study. J Transl Med 17, 73 (2019). <https://doi.org/10.1186/s12967-019-1827-1>
3. *Non-invasive pre-implantation genetic testing for aneuploidy in spent medium may be more reliable than trophectoderm biopsy Lei Huang, Berhan Bogale, Yaqiong Tang, Sijia Lue, Xiaoliang Sunney Xie, and Catherine Racowsky.

For more information, visit our website at: [www.lilacinsights.com/\(NICS-FAQS\)](http://www.lilacinsights.com/(NICS-FAQS))