PREIMPLANTATION GENETIC SCREENING (PGS) BY NGS

Patient Information		Sample Information		Clinic Information
Chart number:		**Date biopsy:**	04/09/2017	**Client:**
Patient name:		**Date of receipt:**	05/09/2017	
Partner name:		**Date of completion:**	08/09/2017	**Doctor:**
Indication:	F/H of Down Syndrome			

METHODOLOGY:

Library preparation:

- Ion ReproSeq™ PGS kit (Thermo Fisher Scientific, Inc., MA, USA).

Rapid NGS (Next Generation Sequencing) (Thermo Fisher Scientific, Inc., MA, USA):

- Ion Reporter software v 5.0 for data analysis.
- ReproSeq Low-pass whole-genome aneuploidy workflow v 1.0: bioinformatics tool used by the above mentioned software to detect 24 chromosomes aneuploidies from a single whole-genome sample with low coverage (minimum 0.01x). Normalization is done using the bioinformatics baseline ReproSeq Low-Coverage Whole-Genome Baseline (5.0) generated from multiple normal samples.

RESULTS:

Embryo	Cell Type	Results	Transference
RJ3	Trophectoderm	No DNA detected	NO
RJ5	**Trophectoderm**	**NORMAL**	**YES**
RJ8	**Trophectoderm**	**NORMAL**	**YES**
RJ9	Trophectoderm	Abnormal: +4	NO
RJ10	**Trophectoderm**	**NORMAL**	**YES**
RJ12	Trophectoderm	No DNA detected	NO
RJ13	**Trophectoderm**	**NORMAL**	**YES**
RJ15	Trophectoderm	Abnormal: +6, +18p	NO

In case of pregnancy prenatal diagnosis is recommended.

INTERPRETATION:

Ion Reporter software generates a graph representing the copy number variation (CNV) of the sample analyzed compared to the reference bioinformatics baseline. An embryo is considered as normal when it has no deviations from the reference bioinformatics baseline for any of the 24 chromosomes. An embryo is considered as abnormal by the presence of aneuploidy when there are points that are diverted into the upper (gain +) or lower part (loss -) of the graph. The presence of aneuploidies for more than five chromosomes on the same specimen is interpreted as a complex abnormal embryo. Reason for 'No DNA detected' can be either no cell present in the analysis tube or poor quality sample. Poor quality sample can be correlated with poor quality embryo. Embryo transfer is not recommended in any cases. This test allows detection of chromosomal aneuploidies in all human chromosomes including sex chromosomes (chromosomes X and Y). BUT SEXING STATUS OF THE ANALYSED EMBRYOS WILL NEVER BE REVEALED IN THE REPORT EITHER IN ANY OTHER WAY TO THE PATIENTS NOR TO THE CLINICIANS.

LIMITATIONS:

PGS for aneuploidy does not analyze specific genes and cannot detect conditions caused by single gene mutations, such as sickle cell anemia, cystic fibrosis, or Tay-Sachs disease. PGS cannot detect structural rearrangements in which there is a balanced (normal) amount of genetic material.With this technique it is not possible to identify deletions and duplications smaller than the limit of resolution of the platform used, mosaic aneuploidy in low grade and, defects affecting the complete set of chromosomes (haploidy, triploidy). PGS cannot detect all potential birth defects. There is a 3-5% risk in the general population of birth defects. These may be caused by genetic and/or non-genetic etiologies.

Validation studies performed by Igenomix demonstrate that there is approximately chance of a misdiagnosis, either by a false negative or a false positive result. Because of the chance of misdiagnosis, the inability to detect mosaicism, and the investigational nature of PGS, ongoing pregnancies resulting from PGS for aneuploidy during IVF should always be followed by prenatal diagnosis, either through chorionic villus sampling at weeks or amniocentesis at 15-18 weeks to confirm a chromosomally normal fetus.

Signature

Sample name, PhD

Biologist

Signature

Sample name, PhD

Laboratory Manager

Note: The results shown in this test are subjected to the veracy of the information provided by the clinic (embryo ID) and to the proper introduction of the corresponding cell into the analysis tube. In case of pregnancy, prenatal diagnosis is recommended to confirm preimplantation diagnosis. Don't hesitate to contact IGENOMIX for any doubt you may have (Contact no. 011-66517933/34 Email: info.india@igenomix.com)