igenomix
PIONEERS IN REPRODUCTIVE GENETICS

GENETIC DIAGNOSIS OF PRODUCT OF CONCEPTION (POC)

Patient Information		Sample Information		Clinic Information
Patient ID	PAT-000005186	**Sample type**	Biopsy	**Clinic**
Internal Reference	POC-18M0001/POC 18-02	**Reception date**		**Referral physician**
Patient Name		**Results date**		
Date of birth		**Gestagional age:**	8 weeks	
Gender	Female	**Gestation type**	Singleton	

Methodology:

Fetal versus maternal tissue is identified whenever possible. The specimen is cleaned, 3 dissections obtained and DNA is extracted with Qiagen QIAamp DNA extraction kits. Short tandem Repeat (STR) AmpFISTR Identifiler Plus (Life technologies) protocol is used to detect or rule out maternal cell contamination and polyploidies. Genetic testing for Products of Conception (POC) is done using a Next Generation Sequencing technology with the PGM (Personal Genome Machine) or S5 PGS Assay (Next Generation Sequencing) for 24 chromosomes to determine numerical chromosome abnormalities. The Ion ReproSeq PGS kit for 24 chromosomes aneuploidy screening (Thermo Fisher Scientific, USA) is used. Data analysis is performed using the Ion Reporter Software (Thermo Fisher Scientific, USA), that generates a graph representing the copy number variation (CNV) of the sample analyzed compared to a reference bioinformatics baseline generated from multiple normal samples.

Interpretation

Ion Reporter software generates a graph representing the copy number variation (CNV) of the sample analyzed compared to the reference bioinformatics baseline. A sample is considered as normal when it has no deviations from the reference bioinformatics baseline for any of the 24 chromosomes. A sample 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. With this technique is not possible to identify deletions and duplications smaller than the limit of resolution of the platform used, balanced structural abnormalities, mosaic aneuploidy in low grade, and some defects affecting the complete set of chromosomes, such us triploidy 69,XXX (if maternal origin) and tetraploidy 92,XXXX or 92,XXXY.

Results:

Internal reference	NGS results
POC-18M0001/POC18-02	**Abnormal:+22**

The sample analyzed is identified as an abnormal with trisomy on chromosome 22. As the abortive tissue was chromosomally abnormal this could be the reason for the early abortion. To avoid the same situation in future we recommend the patient to undergo an IVF cycle with Preimplantation Genetic Screening (PGS). Using PGS by NGS, only chromosomally normal embryos will be transferred to the maternal uterus.

Signature

Sample name, PhD

Biologist

Signature

Sample name, PhD

Laboratory Manager