FARABI MEDICAL LABORATORY

PGT SEX DETERMINATION

Preimplantation Genetic Testing (PGT) Sex Determination by PCR

Overview

Preimplantation Genetic Testing (PGT) for Chromosomal Sex Determination using PCR-based molecular analysis on trophectoderm biopsy samples enables determination of embryo sex prior to transfer and may be clinically indicated in specific reproductive scenarios.

Clinical Indications

PGT for sex determination may be considered in:

- Prevention of X-linked genetic disorders
- Families with known sex-linked pathogenic variants
- Specific medically justified reproductive planning
- Situations where sex selection is legally permitted

Methodology

- Trophectoderm biopsy (Day 3-5 blastocyst)
- DNA extraction from embryonic cells
- PCR amplification of Y-chromosome–specific markers (e.g., SRY gene)
- Internal amplification controls to confirm DNA quality
- Electrophoretic detection and analysis

Result Interpretation:

- Detection of Y-specific sequences → XY embryo
- Absence of Y-specific sequences → XX embryo

Advantages

✓ Rapid turnaround time compatible with IVF workflow
✓ High analytical sensitivity
✓ Requires minimal DNA input
✓ Suitable for integration with other PGT strategies

Important Limitations

- Determines chromosomal sex only
- Does NOT assess whole-chromosome aneuploidy and structural rearrangements
- Does NOT replace PGT-A or PGT-M when clinically indicated
- Mosaicism cannot be fully excluded
- Rare PCR failure or allele dropout may occur

Sample Requirements

- Trophectoderm biopsy (recommended)
- Proper labeling and secure transport conditions required
- Coordination with IVF laboratory prior to biopsy recommended

Turnaround Time

4 to 5 hours and optimized to align with embryo transfer scheduling.

Quality & Reporting

- Performed in a dedicated molecular genetics laboratory
- Validated PCR methodology
- Results reported with interpretation and clinical guidance
- Genetic counseling recommended where appropriate

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