Genetic tests for reproduction

PRECONCEPTION

Purpose: Reproductive risk assessment and infertility investigation

Testing material: Peripheral blood

KARYOTYPE
Chromosomal rearrangements or sex chromosome aneuploidies

CARRIER SCREENING
Risk assesment for severe recessive conditions

FRAGILE X PREMUTATION ANALYSIS
For Premature Ovarian Insufficiency

CYSTIC FIBROSIS TESTING
For Obstructive Azoospermia

Y CHROMOSOME MICRODELETIONS
For Azoospermia and Oligospermia

PREIMPLANTATION

Purpose: Identification of genetic and chromosomal abnormalities in embryos

Testing material: Biopsied cells
(day 3, 5-7 of embryo development)

Preimplantation Genetic Testing (PGT)

PGT-A (for aneuploidies)
Detection of chromosomal aneuploidies

PGT-SR (for structural rearrangements)
Detection of chromosome imbalances

PGT-M (for monogenic disorders)
Detection of specific genetic variants associated with monogenic disorders

PRENATAL

Purpose: Fetal genetic evaluation during pregnancy, especially for high-risk cases

Non-invasive prenatal testing (NIPT)
Testing material: maternal peripheral blood, 1st trimester
Screen for chromosomal alterations/microdeletions/microduplications

Invasive prenatal test
Testing material: placental tissue, amniotic fluid or fetal blood

i. Chromosomal analysis
ii. Copy number variations (CNV) and single nucleotide polymorphisms (SNP)
iii. Gene sequencing