PORTFOLIO OF GENETIC TESTS

***CellGenetics* laboratory offers wide portfolio of genetic tests:**

Hereditary thrombophilia:

Basic hereditary thrombophilia profile:

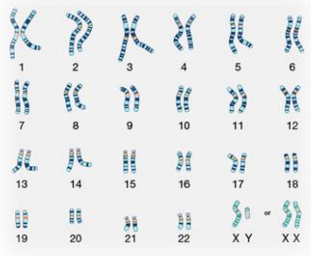
- *F2* (20210G>A)
- *F5* Leiden (1691G>A)
- *MTHFR* (677C>T)
- *PAI-1* (4G/5G)

Expanded hereditary thrombophilia profile:

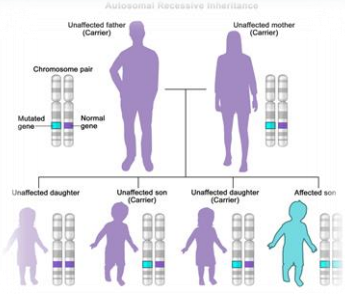
- *F2* (20210G>A)
- *F5* Leiden (1691G>A)
- *F5* R2 (3980A>G)
- *MTHFR* (677C>T)
- *MTHFR* (1298A>C)
- *PAI-1* (4G/5G)
- *F13* (Val34Leu)
- *PROCR*

Reproductive health:

- Karyotyping
- Y chromosome azoospermia factor region microdeletions (AZFa, AZFb, AZFc)
- *CFTR* gene mutations carrier status (absence of vas deferens)
- *FMR1* gene mutations carrier status (fragile X-associated primary ovarian insufficiency)



Carrier status:



- Cystic fibrosis: common mutations in the *CFTR* gene in Bulgaria
- Phenylketonuria: common mutations in the *PAH* gene in Bulgaria
- β-Thalassemia: mutations in the *HBB* gene
- Spinal muscular atrophy (deletions ex 7.8 in *SMN1*)
- Duchenne and Becker dystrophy: deletions, duplications in the dystrophin gene (*DMD*)
- Three nucleotide repeat expansion: Huntington's chorea (*HTT*)
- Postnatal screening for microdeletion syndromes: DiGeorge, Wolf-Hirschhorn, Cri du Chat, Prader-Willi, Angelman, 1p36 deletion, etc.
- Postnatal screening for subtelomeric deletions and duplications
- Single variant carrier screen by Sanger sequencing in families with known mutation
- Confirmation of single variant carrier status by Sanger sequencing in families with known mutation
- Screening for carriers of pathological variants in genes associated with rare recessive diseases

Non-invasive prenatal tests:

- Non-invasive fetal RhD blood group genotyping (mother/fetus Rh compatibility)
- Non-invasive paternity test
- Non-invasive prenatal screening for monogenic diseases

Invasive prenatal tests:

- Prenatal DNA fragment analysis: chromosomes 13, 18, 21, X, Y
- Prenatal diagnoses of monogenic diseases (known mutation in the family)
- ChromoSeq® - Genome-wide DNA Analysis:
  - ✓ Complete/partial aneuploidies
  - ✓ Unbalanced translocations
  - ✓ Microdeletions and microduplications
  - ✓ Deletions and duplications in specific genes

DNA analysis of abortion material:

- DNA fragment analysis of abortion material: chromosomes 13, 18, 21, X, Y
- DNA fragment analysis of abortion material: chromosomes 15, 16 and 22
- DNA fragment analysis of abortion material: chromosomes 13, 15, 16, 18, 21, 22, X, Y
- ChromoSeq® - Genome-wide DNA Analysis:
  - ✓ Complete/partial aneuploidies
  - ✓ Unbalanced translocations
  - ✓ Microdeletions and microduplications
  - ✓ Deletions and duplications in specific genes

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