Hereditary and Cancer Genetic Testing

Genetic Testing for Hereditary Cancers

Genetic testing for hereditary cancers provides life-changing insights for patients and their families. By identifying genetic variants associated with cancer risk, it guides personalized screening, prevention, and treatment strategies.

Next-Generation Sequencing (NGS) Panels allow comprehensive analysis of multiple genes simultaneously, including those relevant for hereditary cancers and syndromes with overlapping clinical features. This ensures early detection, improved diagnostic accuracy, and better-informed healthcare decisions.

Benefits of Hereditary Cancer Genetic Testing

- **Early Detection:** Identify individuals at increased risk before symptoms appear.
- **Family Impact:** Helps at-risk relatives take preventive measures or pursue early screening.
- **Personalized Treatment:** Guides therapy selection based on genetic findings.
- **Improved Diagnosis:** Detects rare or overlapping syndromes that may otherwise be missed.
- **Ongoing Updates:** Continuous refinement of test interpretation as genetics advances.

Key Features of NGS Panels

- **Comprehensive Coverage:** Includes all clinically relevant genes and variants, coding and non-coding.
- **High Sensitivity & Specificity:** Detects SNVs, InDels, and larger CNVs accurately.
- **Advanced Bioinformatics:** Sophisticated algorithms and curated databases for precise variant interpretation.
- **Variant Reclassification Program:** Clinicians receive updates on new findings for previously tested variants.
- **Fast Turnaround:** Results typically available within 15 business days.

Clinical Utility

- Identifies cancer predisposition genes for informed risk assessment.
- Supports differential diagnosis in patients with overlapping symptoms.
- Detects variants affecting treatment response to optimize therapy.
- Includes high-resolution CNV analysis to capture deletions/duplications (5–10% of disease-causing variants).

Available Panels and Tests

BRCA1 & BRCA2 Screening

- **TAT:** 15 business days | **Genes:** 2
- **Purpose:** Detects hereditary mutations in BRCA1 and BRCA2, responsible for 5–10% of breast cancers in women.
- **Method:** NGS including CNV analysis | **Coverage:** ≥99% at ≥20x
- **Sample:** EDTA Blood 5 ml

Key Benefits:

- Identifies high-risk individuals for targeted surveillance.
- Supports early preventive interventions for patients and relatives.

Breast Cancer Screening

- **TAT:** 15 business days | **Genes:** 28
- **Purpose:** Detects mutations in BRCA1, BRCA2, and other breast cancer-associated genes (ATM, BRIP1, CHEK2, PALB2, etc.) which have also been associated with increased cancer risk.
- **Method:** NGS including CNV analysis | **Coverage:** ≥99% at ≥20x
- **Sample:** EDTA Blood 5 ml

Key Benefits:

- Comprehensive detection of hereditary breast cancer variants.
- Enables proactive risk management for patients and family members.

Cancer Screening

- **TAT:** 15 business days | **Genes:** 67
- **Purpose:** has been carefully selected based on its risk potential in the development of one or more of the following cancers: breast, ovarian, colorectal, gastric, thyroid, endometrial, pancreatic, melanoma, renal, and prostate. This panel is appropriate for patients with a positive personal history of early-onset cancer, rare cancer, bilateral cancer, or multiple primary cancers.
- **Method:** NGS including CNV analysis | **Coverage:** ≥99% at ≥20x
- **Sample:** EDTA Blood 5 ml

Key Benefits:

- Identifies hereditary cancer syndromes.
- Guides personalized surveillance and treatment strategies.

Comprehensive Cancer Screening

- **TAT:** 15 business days | **Genes:** 118
- **Purpose:** the most extensive cancer panel, covering a large number of cancer-associated genes. Carefully selected based on its risk potential in the development of one or more of the following cancers: breast, ovarian, colorectal, gastric, thyroid, endometrial, pancreatic, melanoma, renal, and prostate, among others.
- **Method:** NGS including CNV analysis | **Coverage:** ≥99% at ≥20x
- **Sample:** EDTA Blood 5 ml

Key Benefits:

- Provides the most complete hereditary cancer risk evaluation.
- Maximizes diagnostic yield and clinical utility.