



**GENETICS PROGRAM
MOLECULAR GENETICS LABORATORY TESTS**

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| Hematological disorders |
| Familial Thrombophilia Factor V Leiden and Factor II c.*97G>A |
| Hereditary Hemochromatosis H63D and C282Y |
| Infertility |
| Fragile X-associated Premature Ovarian Insufficiency (FXPOI) |
| Y-chromosome Microdeletion |
| Intellectual disability/Autism/Developmental delay |
| Fragile X Syndrome (FXS) |
| SNP Array |
| Myeloid Neoplasms (AML/APL and MDS, MPN, MDS/MPN) |
| Myeloid Neoplasms NGS Panel (42) - <i>ABL1, ASXL1, BCOR, BCORL1, BRAF, CALR, CBL, CEBPA, CUX1, CSF3R, DDX41, DNMT3A, EZH2, ETV6, FLT3, IDH1, IDH2, JAK2, GATA2, KIT, KMT2A, KRAS, MPL, NF1, NPM1, NRAS, PHF6, PPM1D, PTPN11, PRPF8, RAD21, RUNX1, SETBP1, SH2B3, SF3B1, SRSF2, STAG,2 TET2, TP53, U2AF1, WT1, ZRSR2</i> |
| Neurological disorders |
| C9orf72-related Disorders |
| Dentatorubral Pallidoluysian Atrophy (DRPLA) |
| Fragile X-associated Tremor/Ataxia syndrome (FXTAS) |
| Friedreich Ataxia (FRDA) |
| Huntington Disease (HD) |
| Oculopharyngeal Muscular Dystrophy (OPMD) |
| Spinal and Bulbar Muscular Atrophy (SBMA) |
| Spinocerebellar Ataxia (SCA) - SCA1, SCA2, SCA3, SCA6, SCA7, SCA8, and SCA17 |
| Pharmacogenetics |
| <i>DPYD</i> Genotyping |
| Prenatal and Neonatal |
| Aneuploidy of Chromosomes 13, 18, 21, X and Y |
| SNP Array |
| Molar Pregnancy |
| Miscellaneous |
| Maternal Cell Contamination (MCC) |
| Specimen Misidentification |
| Uniparental Disomy of Chromosomes 14 and 15 (UPD14 and UPD15) |

| Hereditary Cancer |
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| Mutation Analysis - Familial or Tumour findings |
| Ashkenazi Jewish Founder Panel (9 variants) - <i>APC</i> (c.3920T>A), <i>BRCA1</i> (c.68_69del; c.5266dup), <i>BRCA2</i> (c.5946del), <i>CHEK2</i> (c.1283C>T), <i>GREM1</i> (40 kb upstream dup), <i>MSH2</i> (c.1906G>C), <i>MSH6</i> (c.3959_3962del; c.3984_3987dup) |
| Hereditary Breast/Ovarian/Prostate Cancer Panel (19) - <i>ATM</i> , <i>BARD1</i> , <i>BRCA1</i> , <i>BRCA2</i> , <i>BRIP1</i> , <i>CDH1</i> , <i>CHEK2</i> , <i>EPCAM</i> , <i>HOXB13</i> , <i>MLH1</i> , <i>MSH2</i> , <i>MSH6</i> , <i>PALB2</i> , <i>PMS2</i> , <i>PTEN</i> , <i>RAD51C</i> , <i>RAD51D</i> , <i>STK11</i> , <i>TP53</i> |
| Hereditary Endometrial Cancer Panel (10) - <i>BRCA1</i> , <i>BRCA2</i> , <i>EPCAM</i> , <i>MLH1</i> , <i>MSH2</i> , <i>MSH6</i> , <i>PMS2</i> , <i>POLD1</i> , <i>POLE</i> , <i>PTEN</i> |
| Hereditary GI (Lynch syndrome, Gastric, Pancreas, Polyposis) Cancer Panel (31) - <i>APC</i> , <i>ATM</i> , <i>BMPR1A</i> , <i>BRCA1</i> , <i>BRCA2</i> , <i>CDH1</i> , <i>CDKN2A</i> , <i>CHEK2</i> , <i>CTNNA1</i> , <i>EPCAM</i> , <i>GALNT12</i> , <i>GREM1</i> , <i>MLH1</i> , <i>MLH3</i> , <i>MSH2</i> , <i>MSH3</i> , <i>MSH6</i> , <i>MUTYH</i> , <i>NTHL1</i> , <i>PALB2</i> , <i>PMS2</i> , <i>POLD1</i> , <i>POLE</i> , <i>PTEN</i> , <i>RNF43</i> , <i>RPS20</i> , <i>SDHB</i> , <i>SDHD</i> , <i>SMAD4</i> , <i>STK11</i> , <i>TP53</i> |
| Hereditary Breast/Ovarian/Prostate/GI Cancer Panel (36) - <i>APC</i> , <i>ATM</i> , <i>BARD1</i> , <i>BMPR1A</i> , <i>BRCA1</i> , <i>BRCA2</i> , <i>BRIP1</i> , <i>CDH1</i> , <i>CDKN2A</i> , <i>CHEK2</i> , <i>CTNNA1</i> , <i>EPCAM*</i> , <i>GALNT12</i> , <i>GREM1</i> , <i>HOXB13</i> (G84E), <i>MLH1</i> , <i>MLH3</i> , <i>MSH2</i> , <i>MSH3</i> , <i>MSH6</i> , <i>MUTYH</i> , <i>NTHL1</i> , <i>PALB2</i> , <i>PMS2</i> , <i>POLD1</i> , <i>POLE</i> , <i>PTEN</i> , <i>RAD51C</i> , <i>RAD51D</i> , <i>RNF43</i> , <i>RPS20</i> , <i>SDHB</i> , <i>SDHD</i> , <i>SMAD4</i> , <i>STK11</i> , <i>TP53</i> |
| Lynch Syndrome Panel (5) - <i>EPCAM</i> , <i>MLH1</i> , <i>MSH2</i> , <i>MSH6</i> , <i>PMS2</i> |
| Gastric Cancer Panel (17) - <i>APC</i> , <i>ATM</i> , <i>BRCA1</i> , <i>BRCA2</i> , <i>CDH1</i> , <i>CTNNA1</i> , <i>EPCAM</i> , <i>MLH1</i> , <i>MSH2</i> , <i>MSH6</i> , <i>PALB2</i> , <i>PMS2</i> , <i>SDHB</i> , <i>SDHD</i> , <i>SMAD4</i> , <i>STK11</i> , <i>TP53</i> |
| Pancreatic Adenocarcinoma Panel (12) - <i>ATM</i> , <i>BRCA1</i> , <i>BRCA2</i> , <i>CDKN2A</i> , <i>EPCAM</i> , <i>MLH1</i> , <i>MSH2</i> , <i>MSH6</i> , <i>PALB2</i> , <i>PMS2</i> , <i>STK11</i> , <i>TP53</i> |
| Polyposis Panel (18) - <i>APC</i> , <i>BMPR1A</i> , <i>EPCAM</i> , <i>GREM1</i> , <i>MLH1</i> , <i>MLH3</i> , <i>MSH2</i> , <i>MSH3</i> , <i>MSH6</i> , <i>MUTYH</i> , <i>NTHL1</i> , <i>PMS2</i> , <i>POLD1</i> , <i>POLE</i> , <i>PTEN</i> , <i>SMAD4</i> , <i>STK11</i> , <i>TP53</i> |
| Familial Gastrointestinal Stromal Tumor Panel (7) - <i>KIT</i> , <i>PDGFRA</i> , <i>SDHA</i> , <i>SDHAF2</i> , <i>SDHB</i> , <i>SDHC</i> , <i>SDHD</i> |
| Familial Melanoma Panel (7) - <i>BAP1</i> , <i>BRCA2</i> , <i>CDK4</i> , <i>CDKN2A</i> , <i>MITF</i> , <i>POT1</i> , <i>PTEN</i> |
| Familial Renal Cancer Panel (15) - <i>BAP1</i> , <i>FH</i> , <i>FLCN</i> , <i>MET</i> , <i>MITF</i> , <i>PTEN</i> , <i>SDHA</i> , <i>SDHAF2</i> , <i>SDHB</i> , <i>SDHC</i> , <i>SDHD</i> , <i>TP53</i> , <i>TSC1</i> , <i>TSC2</i> , <i>VHL</i> |
| Hereditary Pheochromocytoma and Paraganglioma Panel (12) - <i>FH</i> , <i>MAX</i> , <i>MEN1</i> , <i>NF1</i> , <i>RET</i> , <i>SDHA</i> , <i>SDHAF2</i> , <i>SDHB</i> , <i>SDHC</i> , <i>SDHD</i> , <i>TMEM127</i> , <i>VHL</i> |
| CNS Tumor Panel (20) - <i>APC</i> , <i>EPCAM</i> , <i>LZTR1</i> , <i>MLH1</i> , <i>MSH2</i> , <i>MSH6</i> , <i>NF1</i> , <i>NF2</i> , <i>PMS2</i> , <i>POLE</i> , <i>POT1</i> , <i>PTCH1</i> , <i>PTEN</i> , <i>SMARCB1</i> , <i>SMARCE1</i> , <i>SUFU</i> , <i>TP53</i> , <i>TSC1</i> , <i>TSC2</i> , <i>VHL</i> |
| Soft Tissue Sarcoma Panel (12) - <i>APC</i> , <i>ATM</i> , <i>BRCA1</i> , <i>BRCA2</i> , <i>CHEK2</i> , <i>EPCAM</i> , <i>MLH1</i> , <i>MSH2</i> , <i>MSH6</i> , <i>NF1</i> , <i>PMS2</i> , <i>TP53</i> |
| AXIN2-related Attenuated Familial Adenomatous Polyposis (1) - <i>AXIN2</i> |
| BAP1 Tumour Predisposition Syndrome (1) - <i>BAP1</i> |
| Birt-Hogg-Dube Syndrome (1) - <i>FLCN</i> |
| Carney Complex (1) - <i>PRKAR1A</i> |
| CHRPE, CMV Thyroid, Desmoid (2) - <i>APC</i> , <i>MUTYH</i> |
| DICER-associated Syndrome (1) - <i>DICER1</i> |

| Hereditary Cancer continued |
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| Dysplastic Nevus Syndrome (2) - <i>CDK4, CDKN2A</i> |
| Familial Isolated Pituitary Adenoma (1) - <i>AIP</i> |
| Hereditary Hyperparathyroidism (1) - <i>CDC73, MEN1</i> |
| Hereditary Leiomyomatosis and Renal Cell Cancer (1) - <i>FH</i> |
| Hereditary Lung Cancer - <i>EGFR: T790M; V834I; V769M</i> |
| Li-Fraumeni Syndrome (1) - <i>TP53</i> |
| MEN1 Syndrome (2) - <i>MEN1, CDKN1B</i> |
| Multiple Endocrine Neoplasia Type 2 (1) - <i>RET</i> |
| Neurofibromatosis Type 1 (1) - <i>NF1</i> |
| Gorlin Syndrome (Nevoid Basal Cell Carcinoma Syndrome) (2) - <i>PTCH1, SUFU</i> |
| Nijmegen Breakage Syndrome (1) - <i>NBN</i> |
| Peutz-Jeghers Syndrome (1) - <i>STK11</i> |
| PTEN Hamartoma Tumour Syndrome (1) - <i>PTEN</i> |
| Rare Polyposis Genes (2) - <i>GALNT12, RPS20</i> |
| Retinoblastoma (1) - <i>RB1</i> |
| Rhabdoid Predisposition Syndrome (2) - <i>SMARCA4, SMARCB1</i> |
| Schwannomatosis - <i>NF2, LZTR1, SMARCB1</i> |
| Sessile Serrated Polyposis Cancer Syndrome (1) - <i>RNF43</i> |
| Small Cell Carcinoma of the Ovary, Hypercalcemic Type (1) - <i>SMARCA4</i> |
| Tuberous Sclerosis (2) - <i>TSC1, TSC2</i> |
| Von Hippel-Lindau Syndrome (1) - <i>VHL</i> |