

**GENETICS PROGRAM
MOLECULAR GENETICS LABORATORY TESTS**

Prenatal and Neonatal
Aneuploidy of Chromosomes 13, 18, 21, X and Y
SNP microarray
Molar pregnancy
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) <ul style="list-style-type: none"> - SCA1, SCA2, SCA3, SCA6, SCA7, SCA8, and SCA17
Intellectual disability/Autism/Developmental delay
Fragile X syndrome (FXS)
SNP microarray
Hematological disorders
Familial Thrombophilia <ul style="list-style-type: none"> Factor V Leiden and Factor II c.*97G>A
Hereditary Hemochromatosis <ul style="list-style-type: none"> H63D and C282Y
Infertility
Fragile X-associated Premature Ovarian Insufficiency (FXPOI)
Y-chromosome Microdeletion
Miscellaneous
Maternal Cell Contamination (MCC)
Specimen Misidentification
Uniparental Disomy of Chromosomes 14 and 15 (UPD14 and UPD15)
Hereditary Cancer
Mutation Analysis <ul style="list-style-type: none"> - Familial or Tumour findings
Ashkenazi Jewish Founder Panel (9 variants) <ul style="list-style-type: none"> - APC (c.3920T>A), BRCA1 (c.68_69del; c.5266dup), BRCA2 (c.5946del), CHEK2 (c.1283C>T), GREM1 (40 kb upstream dup), MSH2 (c.1906G>C), MSH6 (c.3959_3962del; c.3984_3987dup)
Hereditary Breast/Ovarian/Prostate Cancer Panel (19) <ul style="list-style-type: none"> - ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, HOXB13, MLH1, MSH2, MSH6, PALB2, PMS2, PTEN, RAD51C, RAD51D, STK11, TP53

Hereditary Cancer continued
Hereditary Endometrial Cancer Panel (10) - <i>BRCA1, BRCA2, EPCAM, MLH1, MSH2, MSH6, PMS2, POLD1, POLE, PTEN</i>
Hereditary GI (Lynch syndrome, Gastric, Pancreas, Polyposis) Cancer Panel (28) - <i>APC, ATM, BMPR1A, BRCA1, BRCA2, CDH1, CDKN2A, CHEK2, CTNNA1, EPCAM, GREM1, MLH1, MLH3, MSH2, MSH3, MSH6, MUTYH, NTHL1, PALB2, PMS2, POLD1, POLE, PTEN, SDHB, SDHD, SMAD4, STK11, TP53</i>
Lynch Syndrome Panel (5) - <i>EPCAM, MLH1, MSH2, MSH6, PMS2</i>
Gastric Cancer Panel (17) - <i>APC, ATM, BRCA1, BRCA2, CDH1, CTNNA1, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, SDHB, SDHD, SMAD4, STK11, TP53</i>
Pancreatic Adenocarcinoma Panel (12) - <i>ATM, BRCA1, BRCA2, CDKN2A, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, STK11, TP53</i>
Polyposis Panel (18) - <i>APC, BMPR1A, EPCAM, GREM1, MLH1, MLH3, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, SMAD4, STK11, TP53</i>
Familial Gastrointestinal Stromal Tumor Panel (7) - <i>KIT, PDGFRA, SDHA, SDHAF2, SDHB, SDHC, SDHD</i>
Familial Melanoma Panel (7) - <i>BAP1, BRCA2, CDK4, CDKN2A, MITF, POT1, PTEN</i>
Familial Renal Cancer Panel (15) - <i>BAP1, FH, FLCN, MET, MITF, PTEN, SDHA, SDHAF2, SDHB, SDHC, SDHD, TP53, TSC1, TSC2, VHL</i>
Hereditary Pheochromocytoma and Paraganglioma Panel (12) - <i>FH, MAX, MEN1, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL</i>
CNS Tumor Panel (20) - <i>APC, EPCAM, LZTR1, MLH1, MSH2, MSH6, NF1, NF2, PMS2, POLE, POT1, PTCH1, PTEN, SMARCB1, SMARCE1, SUFU, TP53, TSC1, TSC2, VHL</i>
Soft Tissue Sarcoma Panel (12) - <i>APC, ATM, BRCA1, BRCA2, CHEK2, EPCAM, MLH1, MSH2, MSH6, NF1, PMS2, 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, MUTYH</i>
DICER-associated Syndrome (1) - <i>DICER1</i>
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>

Hereditary Cancer continued
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>