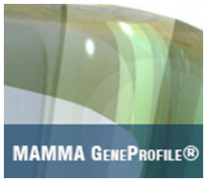
GeneProfile®:Massive sequencing at the service of hereditary disease diagnosis.



MAMMA GENE PROFILE®

Simultaneous study of BRCA1/BRCA2 genes by massive sequencing

MAMA GeneProfile® is a novel genetic study for **Hereditary Breast and Ovarian Cancer** (HBOC), which simultaneously studies **BRCA1 and BRCA2** by parallel massive sequencing. This panel may be complemented with an MLPA Study.

Cancer Disease	*Gene Panel Description*
Hereditary Breast and Ovarian Cancer	**2 genes panel**: BRCA1 and BRCA2. (MLPA included)

ONCO GENE PROFILE®

Simultaneous study of 80 genes by massive sequencing

This panel includes the genes associated with Affects of Multiple Cancer and of Familial Aggregation. Specific panels are available for Hereditary Breast and Ovarian Cancer (HBOC), as are panels to study Familial Polyposis Adenomatous (FAP) and Hereditary non-polyposis colorectal cancer (HNPCC). All these may be complemented with an MLPA Study.

Cancer Disease	Gene Panel Description
Hereditary Breast and Ovarian Cancer	**13 genes panel:** BRCA1, BRCA2, RAD51C, CDH1, TP53, PTEN, STK11, PALB2, RAD51D, BRIP1, XRCC2, ERCC4 , ATM.
Hereditary Nonpolyposis Colon Cancer	**4 genes panel:** MLH1, MSH2, MSH6, PMS1
Hereditary Colorectal Adenomatous Polyposis	**2 genes panel:** APC, MUTYH
Juvenile Polyposis Syndrome	**3 genes panel:** SMAD14, BMPR1A, PTEN
PTEN Hamartoma Tumor Syndrome (PHTS)	**1 gene panel:** PTEN
Peutz-Jeghers Syndrome	**1 gene panel:** STK11
Hereditary Diffuse Gastric Cancer	**1 gene panel:** CDH1
Li-Fraumeni Syndrome	**1 gene panel:** TP53
Familial Melanoma	**1 gene panel:** p16 (CDKN2A)
Prostate Cancer	**1 gene panel:** HOXB13
Gastric Cancer	**9 genes panel:** MLH1, MSH2, MSH6, PMS1, TP53, CHEK2, BRCA1, BRCA2, APC
Ataxia Telengiectaxia	**1 gene panel:** ATM
Endocrine tumors (MEN2/ CMT, MEN1, Von Hippel Lindau)	**3 genes panel:** RET, MEN1, VHL
Pheochromocytoma / Paraganglioma	**8 genes panel:** SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, KIF1B, MAX
Pancreatic Cancer (Sd Peutz Jeghers, Pancreatitis)	**4 genes panel:** STK11, PRSS1, BRCA2, PALB2
Renal Cancer (clear cell renal cancer, papillary renal cancer, Birt-Hogg-Dube syndrome)	**5 genes panel:** VHL, MET, FLCN, SDHB, SDHD
Congenital Dyskeratosis	**3 genes panel:** DKC1, TERT, NOP10
Congenital Neutropenia	**4 genes panel:** ELA2, GFI1, WAS, HAX1
Fanconi Anemia	**16 genes panel:** FANCA, FANCB, FANCC, FANCD1, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCJ (BRIP1), FANCL, FANCM, FANCN , FANCO , FANCP, FANCQ
Syndroms in pediatric Oncology (Wilms Tumor, Bloom Syndrome, Sotos Syndrome, Simpson-Golabi-Behmel syndrome, Nijmegel Breakaje Syndrome, Xeroderma Pigmentosum)	**14 genes panel:** WT1, BLM, NSD1, GPC3, NBS1, XPA, XPB, XPC, XPD, XPE, XPG, XPF, XPH, XPV

If you want to analyse other genes apart from those we offer in our panels, please contact us so we can propose a personal study. Our system is flexible enough to adapt it to study custom genes panels for research purposes.

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Contact us!

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