CASE STUDY

18-year-old male with a clinical diagnosis of polyposis (benign)

The patient was diagnosed with a benign form of polyposis without any indication of family history.

PreSENTIA Colorectal Polyposis Syndrome was then performed to identify potential germline mutations linked with polyposis. The test identified:

- A gross genomic **deletion** of the entire coding sequence and promoter 1A of the **APC** gene
- This deletion is associated with **familial adenomatous polyposis (FAP)**

PreSENTIA results led to:

- Accurate identification of the inherited genetic alterations associated with increased cancer risk
- Clinical management plan tailored to the patient
- Medical management recommendations, including:
  - Annual colonoscopy
  - Endoscopic evaluation
  - Chemoprevention (in selected patients)
  - Duodenal surveillance
  - Ultrasound for thyroid cancer

CLINICAL UTILITY

PreSENTIA accurately identified the genetic alteration that is associated with FAP, an autosomal dominant disorder. If not identified early or treated, FAP increases the risk of developing colorectal cancer in the future. PreSENTIA enabled an early treatment plan that will benefit the patient.

ABOUT MEDICOVER GENETICS

Medicover Genetics is a leading innovator in genetic diagnostics, laboratory enablement, and clinical testing, with over 25 years of experience supporting healthcare systems worldwide. The company offers a comprehensive portfolio of genetic testing services and clinical counselling, supported by certified diagnostic products and platforms.At the core of its offering is the proprietary Technology Transfer Platform—an end-to-end genomics solution that enables partner laboratories worldwide to perform high-precision genetic tests in-house. Medicover Genetics' CE-marked, IVDR-certified portfolio, developed through decades of clinical and medical expertise, includes VERACITY™, a non-invasive prenatal test (NIPT), and TarCET™, a suite of clinical assays addressing hereditary cancers, cardiovascular and metabolic disorders, infertility, neonatal conditions, and other critical health areas. Driven by continuous research and development, Medicover Genetics is actively expanding into advanced areas such as liquid biopsy for therapy selection and minimal residual disease (MRD) testing. Its laboratories are CAP and ISO 15189 accredited, CLIA, ISO 9001, and ISO 13485 certified, and comply with GMP and GCP, ensuring the highest quality standards. With a presence across Europe, Asia, and Africa, Medicover Genetics collaborates closely with laboratories and clinicians to deliver scalable, high-impact genetic solutions that support personalized medicine and enable informed clinical decision-making. www.medicover-genetics.com

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MKT-ONC-PS-GEN-DR-EN-V03

PreSENTIA®
hereditary cancer

Genes and genetic alterations



MEDICOVER GENETICS

WHAT IS PreSENTIA?

PreSENTIA is a genetic test for hereditary cancer. It identifies hereditary genetic alterations which are implicated in hereditary cancer syndromes and increase the risk of developing cancer in the future.

CLINICAL UTILITY

PreSENTIA is designed to identify the genetic alterations which can predispose to cancer development in the future

- Better classification of the genetic variants
- Identify individuals and family members who are at high risk of developing cancer
- Take early prophylactic measures to minimize the risk of developing cancer
- Identify the best clinical management

LIST OF HEREDITARY CANCER SYNDROMES TESTED

Hereditary cancer susceptibility genes can be associated with hereditary cancer syndromes which increase the risk of developing cancer in the future. The table below indicates the syndromes tested in PreSENTIA.

PreSENTIA HEREDITARY CANCER SYNDROMES TESTED	
Ataxia-telangiectasia syndrome	Li-Fraumeni syndrome
BAP1 mutation associated disease	Li-Fraumeni syndrome 2
Constitutional mismatch repair syndrome	Lynch syndrome
DICER 1 syndrome	Multiple endocrine neoplasia type 1
Familial adenomatous polyposis / Attenuated familial adenomatous polyposis	Multiple endocrine neoplasia type 2
Fanconi anemia syndrome	MUTYH-associated polyposis syndrome
Hereditary breast & ovarian cancer syndrome	Peutz-Jeghers syndrome
Hereditary diffuse gastric syndrome	Polymerase proofreading associated syndrome
Hereditary mixed polyposis syndrome	PTEN hamartoma syndrome
Hereditary melanoma-pancreatic cancer syndrome	Retinoblastoma
Hereditary paraganglioma – pheochromocytoma syndrome	Von-Hippel Lindau syndrome
Juvenile polyposis syndrome	Xeroderma pigmentosum syndrome

FEATURES AND SPECIFICATIONS



Target Capture Enrichment Technology



Genetic alterations: SNVs, INDELs, CNAs



Sample type: Buccal swab



TAT: 2-3 weeks

LIST OF GENES AND GENETIC ALTERATIONS TESTED

PreSENTIA examines different genetic alterations including single nucleotide variants (**SNVs**), insertions and deletions (**INDELs**), and copy number alterations (**CNAs**) via next generation sequencing (NGS). PreSENTIA targets **full coding exon regions***. The following table indicates the type of genetic alterations covered by PreSENTIA in the different panels.

PANEL	SNVs/INDELs, CNAs
Pan-Cancer *62 genes*	*AKT1, ATM, BARD1, BRAF, BRCA1, BRCA2, BRIP1, CHEK2, CTNNB1, DICER1, EGFR, ERBB2, ERBB3, ESR1, FBXW7, FOXA1, FOXL2, GATA3, KIT, KRAS, MAP3K1, MLH1, MRE11, MSH2, MSH6 MTOR, NBN, NRAS, PALB2, PIK3CA, PIK3CB, PMS2, POLE, PTEN, RAD51C, RAD51D, RAF1, RET, RUNX1, SMAD4, TP53*
Breast/Gynecological *26 genes*	*ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, DICER1, EPCAM†, MLH1, MRE11, MSH2, MSH6, MUTYH, NBN, PALB2, PMS2, POLD1, POLE, PTEN, RAD50, RAD51C, RAD51D, SMARCA4, STK11, TP53*
Breast/Gynecological Guidelines-Based *19 genes*	*ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM†, MLH1, MSH2, MSH6, NBN, PALB2, PMS2, PTEN, RAD51C, RAD51D, TP53, STK11*
Breast High-Risk *7 genes*	*BRCA1, BRCA2, CDH1, PALB2, PTEN, STK11, TP53*
BRCA1 / BRCA2 *2 genes*	*BRCA1, BRCA2*
Colorectal *17 genes*	*APC, BMPR1A, CDH1, CHEK2, EPCAM†, GREM1†, MLH1, MSH2, MSH6, MUTYH, PMS2, POLD1, POLE, PTEN, SMAD4, STK11, TP53*
Colorectal High-Risk *10 genes*	*APC, BMPR1A, EPCAM†, MLH1, MSH2, MSH6, MUTYH, PMS2, SMAD4, STK11*
Colorectal Non-Polyposis *5 genes*	*EPCAM†, MLH1, MSH2, MSH6, PMS2*
Colorectal Polyposis Syndrome *7 genes*	*APC, BMPR1A, MUTYH, POLD1, POLE, SMAD4, STK11*
Myelodysplastic Syndrome Leukemia *24 genes*	*ATM, BRCA1, BRCA2, BRIP1, EPCAM†, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, MLH1, MSH2, MSH6, PALB2, PMS2, RAD51C, SLX4, TP53*
Gastric *14 genes*	*APC, BMPR1A, CDH1, EPCAM†, MLH1, MSH2, MSH6, PMS2, SDHB, SDHC, SDHD, SMAD4, STK11, TP53*
Prostate *15 genes*	*ATM, BRCA1, BRCA2, CHEK2, EPCAM†, HOXB13‡, MLH1, MSH2, MSH6, NBN, PALB2, PMS2, PTEN, RAD51D, TP53*
Pancreatic *17 genes*	*APC, ATM, BRCA1, BRCA2, BMPR1A, CDK4, CDKN2A, (CDKN2A$^{p16(INK4A)}$, CDKN2A$^{p14(ARF)}$), EPCAM†, MEN1, MLH1, MSH2, MSH6, PALB2, PMS2, SMAD4, STK11, TP53*
Renal *13 genes*	*BAP1, EPCAM†, MLH1, MSH2, MSH6, PMS2, PTEN, SDHAF2, SDHB, SDHC, SDHD, TP53, VHL*
Skin (XP-Associated) *9 genes*	*DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, POLH, XPA, XPC*
Familial Melanoma *7 genes*	*BAP1, BRCA2, CDK4, CDKN2A (CDKN2A$^{p16(INK4A)}$, CDKN2A$^{p14(ARF)}$), PTEN, RB1, TP53*
Paraganglioma/ Pheochromocytoma *6 gene*	*RET, SDHAF2, SDHB, SDHC, SDHD, VHL*
Parathyroid *1 gene*	*MEN1*
Thyroid *1 gene*	*RET*

†SNVs/INDELs are not covered ‡CNAs are not covered

**Exceptions on regions containing repeats, sequences of high homology (pseudogenes and segmental duplications) or high GC-content*