PreSENTIA®
hereditary cancer

Detailed genetic analysis of hereditary cancer



MEDICOVER
GENETICS

WHAT IS **PreSENTIA**?

PreSENTIA is a genetic test for hereditary cancer risk assessment. It identifies inherited genetic alterations associated with hereditary cancer syndromes, helping to determine an individual's predisposition to developing specific cancers.

CLINICAL UTILITY

PreSENTIA identifies clinically actionable inherited genetic alterations associated with hereditary cancer syndromes, enabling the detection of individuals with an increased genetic predisposition to cancer.

The test supports the implementation of evidence-based preventive and risk-reduction strategies aimed at decreasing future cancer risk. These may include enhanced surveillance, prophylactic interventions, and other personalised management approaches.

By providing insights into an individual's genetic risk profile, PreSENTIA helps inform tailored screening and surveillance recommendations, facilitating earlier detection and timely clinical intervention.

The results support informed clinical decision-making and optimal patient management by guiding risk assessment, genetic counselling, and long-term care planning.

Identification of pathogenic variants can inform subsequent cascade testing in relatives, supporting appropriate genetic counselling, surveillence, and preventive measures to be offered to family members.

WHO IS THIS TEST FOR?

The patient's medical family history, their ethnicity, and any physical findings linked to cancer should always be taken into consideration. PreSENTIA is recommended for individuals:

- with personal or family history of a hereditary cancer syndrome
- who have been diagnosed with cancer at an unusually young age
- with multiple types of primary tumours
- who are already diagnosed with cancer or are suspected of having a germline mutation with cancer susceptibility
- with rare cancer types
- with unusual cases of a specific cancer type (i.e. breast cancer in men)
- who belong to a specific racial or ethnic group (i.e. Ashkenazi Jewish)
- with suspicion of having a cancer syndrome (i.e. Li-Fraumeni syndrome and others)

The above information was obtained from professional bodies including National Cancer Institute, American College of Obstetricians and Gynecologists (ACOG) and American Society of Clinical Oncology (ASCO).

PreSENTIA PANELS

PreSENTIA offers **19 panels** designed to identify clinically actionable genetic alterations in genes associated with hereditary cancer predisposition. Comprehensive full exonic coverage* is utilised to maximise the detection of genetic alterations that may contribute to cancer development. Genetic alterations tested include single nucleotide variants (**SNVs**), insertions and deletions (**INDELs**), and copy number alterations (**CNAs**).

The genes tested also cover **49 hereditary cancer syndromes** associated with an increased cancer risk, including Lynch syndrome, Li-Fraumeni syndrome, Familial adenamatous polyposis, Peutz-Jeghers syndrome, Fanconi anaemia syndrome and others.

Pan-Cancer	86 genes
Breast & Gynaecological	27 genes
Breast / Gynaecological guidelines based	22 genes
Breast high-risk	7 genes
BRCA1 / BRCA2	2 genes
Colorectal	27 genes
Colorectal high-risk	16 genes
Colorectal non-polyposis	8 genes
Colorectal polyposis syndrome	12 genes
Gastric / GIST	24 genes
Myelodysplastic syndrome / Leukaemia	31 genes
Prostate	19 genes
Pancreatic	21 genes
Renal	23 genes
Skin (XP-associated)	9 genes
Familial melanoma	13 genes
Paraganglioma / Phaeochromocytoma	10 genes
Parathyroid	4 genes
Thyroid	11 genes

**Exceptions on regions containing repeats, sequences of high homology such as pseudogene and segmental deletions, or extreme GC-content.*

RECOMMENDATIONS FROM PROFESSIONAL BODIES

"A hereditary cancer risk assessment is the key to identifying patients and families who may be at increased risk of developing certain types of cancer."

ACOG, 2019[1]

"It is generally recommended that, when possible, a family member with cancer have genetic counselling and testing first, to identify with more certainty if the cancer in the family is due to an inherited genetic variant."

National Cancer Institute[2]

CASE STUDY

18-year-old male with a clinical diagnosis of polyposis (benign) and no family history of polyposis

PreSENTIA Colorectal Polyposis Syndrome results

- A gross genomic deletion of the entire coding sequence and promoter 1A of the APC
- This deletion is associated with familial adenomatous polyposis (FAP), an autosomal dominant disorder
- Medical management recommendations include: annual colonoscopy, endoscopic evaluation, chemoprevention (in selected patients), duodenal surveillance, ultrasound for thyroid cancer, small bowel visualisation

PreSENTIA accurately identified the inherited genetic alterations associated with increased cancer risk, while offering a clinical management plan tailored to the patient.

1. *Hereditary Cancer Syndromes and Risk Assessment: ACOG Committee Opinion Summary, Number 793. Obstet Gynecol. 2019 Dec;134(6):1366-1367. doi: 10.1097/AOG.0000000000003563. PMID: 31764755.*
2. *Genetic Testing for Inherited Cancer Susceptibility Syndromes. National Cancer Institute. Retrieved 07 July 2023 from https://www.cancer.gov/about-cancer/causes-prevention/genetics/genetic-testing-fact-sheet*

OUR **PROPRIETARY** TECHNOLOGY PLATFORM

Our technology platform has been thoroughly assessed and validated for its precision.

TARGETED TECHNOLOGY AND NOVEL BIOINFORMATICS

PreSENTIA is based on a novel, **Target Capture Enrichment Technology** which utilises Next Generation Sequencing, and has been thoroughly validated for its accuracy and precision. Combined with novel **bioinformatic pipelines**, PreSENTIA provides accurate detection of genetic variants even at low allele frequencies. Our high read-depth analysis enables for increased sensitivity and specificity providing reliable results.

GENETIC ALTERATION DETECTION

Multi-engine analysis incorporating innovative bioinformatic pipelines analyse the sequencing data. These enable **accurate detection** of genetic alterations, even at low levels.

FULL EXONIC COVERAGE

PreSENTIA screens for **all coding regions*** of the genes tested, and targets adjacent non-coding sequence, increasing the chances of identifying any pathogenic or likely pathogenic mutations:

- SNVs
- INDELs
- CNVs

BENEFITS OF **PreSENTIA**

VALUABLE
Identifies genetic mutations linked to cancer

SAFE
Non-invasive sample collection

RELIABLE
Proven technology with robust results

THOROUGH
Full exonic coverage*

Exceptions apply, for details visit **www.medicover-genetics.com*

WHAT WILL THE **REPORT** TELL ME?



CLINICALLY SIGNIFICANT VARIANT DETECTED
Pathogenic and likely pathogenic variants are reported following the American College of Medical Genetics and Genomics (ACMG) classification guidelines.

NO CLINICALLY SIGNIFICANT VARIANT DETECTED
No disease-causing genetic variant has been identified in the genes tested.

VARIANTS OF UNCERTAIN SIGNIFICANCE
A genetic change has been detected, but it is currently unknown whether that change is associated with cancer development.

Detailed interpretation of the results is provided. The variant summary as well as the clinical significance of the genetic alterations identified is reported, along with recommendations on the next steps after PreSENTIA.

Variant re-evaluation can also be requested by healthcare providers at defined timeframes.

WHAT CAN I DO AFTER **PreSENTIA**?

- Inform your patient about the results of PreSENTIA and recommend genetic counselling
- Recommend screening tests at key interval timepoints for early cancer detection
- Recommend prophylactic measures, if applicable
- Recommend testing for immediate family members who might be at risk

HOW TO ADMINISTER **PreSENTIA**

Recommend the appropriate **PreSENTIA** panel to your patient

Collect a buccal swab from your patient



Send the sample to **Medicover Genetics**



The samples will be analysed at **Medicover Genetics** laboratories

The results will be sent to you within 2-3 weeks from sample receipt

CONTACT US

If you have additional questions or concerns, please contact us at **info.genetics@medicover.com**

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