

INHERITED CANCER GENETIC TESTING

PATIENT DETAILS

(In BLOCK letters)

Full Name

D D M M Y Y Y Y Y Y M M

DOB / / Age / Gender M F Ethnicity

E-mail ID Contact No.

REFERRING CLINICIAN

(In BLOCK letters)

Clinician Name

Hospital

E-mail ID Contact No.

SAMPLE DETAILS

Sample Type

Blood (4 ml-EDTA) DNA [1000 ng (20 ul x 50 ng)] Others

Please indicate here if this sample needs a state/urgent report (Rush charge may apply)

CLINICAL DIAGNOSIS

Clinical details / Pedigree

(Please provide detailed clinical information including type of cancer, age at diagnosis, family history of cancer & investigations performed)

(Relevant documents can be emailed to contact@ncmgglobal.com)

Neuberg Centre for Genomic Medicine (NCGM)

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TEST REQUESTED

1. BRCA1 & BRCA2 NGS NGS + MLPA

2. Cancer Panels

Sr. No.	Cancer Type	Genes Covered
<input type="checkbox"/>	Breast Cancer	<i>BRCA1, BRCA2, ATM, BARD1, BRIP1, CDH1, CHEK2, NBN, NF1, PALB2, PTEN, RAD51C, RAD51D, STK11, TP53</i>
<input type="checkbox"/>	Ovarian Cancer	<i>BRCA1, BRCA2, ATM, BRIP1, MLH1, MSH2, MSH6, PMS2, EPCAM, NBN, PALB2, RAD51C, RAD51D, STK11, DICER1, SMARCA4, MRE11A, BARD1</i>
<input type="checkbox"/>	Prostate Cancer	<i>BRCA1, BRCA2, HOXB13, MLH1, MSH2, MSH6, TP53, PMS2, EPCAM, NBN, CHEK2, ATM</i>
<input type="checkbox"/>	Pancreatic Cancer	<i>ATM, BRCA1, BRCA2, CDKN2A, MLH1, MSH2, MSH6, EPCAM, PALB2, STK11, TP53</i>
<input type="checkbox"/>	Colon Cancer	<i>MLH1, MSH2, MSH6, PMS2, EPCAM, APC, BMPR1A, MUTYH, PTEN, STK11, SMAD4, TP53, GREM1, POLD1, POLE1, AXIN2, NTHL1, MSH3</i>
<input type="checkbox"/>	Endometrial Cancer	<i>MLH1, MSH2, MSH6, PMS2, EPCAM, PTEN, TP53, STK11</i>
<input type="checkbox"/>	Gastric Cancer	<i>APC, BMPR1A, CDH1, CTNNA1, STK11, MLH1, MSH2, MSH6, SMAD4, SDHA, SDHB, SDHC, SDHD, KIT</i>
<input type="checkbox"/>	Thyroid Cancer	<i>RET, APC, PTEN, PRKAR1A, DICER1, TP53, CHEK2</i>
<input type="checkbox"/>	Chromosomal Breakage Syndrome	<i>BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, MAD2L2, PALB2, RAD51, RAD51C, RFX3, SLX4, UBE2T, XRCC2, LIG4, MYSM1, NBN, NHEJ1, ATM, BLM</i>
<input type="checkbox"/>	Predictive Hereditary Cancer Panel	<i>BRCA1, BRCA2, TP53, PALB2, CDH1, PTEN, BRIP1, ATM, CHEK2, NF1, RAD51C, RAD51D, STK11, MLH1, MSH2, MSH6, PMS2, EPCAM, APC, MUTYH, BMPR1A, RNF43, SMAD4, MEN1, RET, RB1, TSC1, TSC2, VHL, SDHB, SDHD, NBN, NF2, SDHC, POLD1, POLE, GREM1, NTHL1, MSH3, AXIN2, GALNT12</i>
<input type="checkbox"/>	Comprehensive Cancer Gene Panel	<i>AIP, AKT1, ALK, APC, AR, ATM, ATR, AXIN1, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, BTNL2, BUB1B, CASR, CD82, CDC73, CDH1, CDK4, CDKN1B, CDKN1C, CDKN2A, CEBPA, CEP57, CFTR, CHEK2, CPA1, CTNNA1, CYLD, DDB2, DICER1, DIS3L2, EGFR, ELAC2, ENG, EPCAM, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, EXT1, EXT2, EZH2, FAM175A, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FGFR2, FH, FLCN, GALNT12, GATA2, GPC3, GREM1, HNF1A, HNF1B, HOXB13, HRAS, KIT, LIG4, LSP1, LZTR1, MAP3K1, MAX, MC1R, MEN1, MET, MIF, MLH1, MLH3, MRE11A, MSH2, MSH3, MSH6, MSR1, MUTYH, MXI1, NBN, NF1, NF2, NSD1, NTHL1, PALB2, PALLD, PDGFRA, PHOX2B, PIK3CA, PMS1, PMS2, POLD1, POLE, POLH, POT1, PRF1, PRKAR1A, PRSS1, PTCH1, PTEN, RAD50, RAD51C, RAD51D, RASAL1, RB1, RECQL4, RET, RHBDF2, RINT1, RNASEL, RNF43, RSP20, RUNX1, SBDS, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLX4, SMAD4, SMARCA4, SMARCB1, SMARCE1, SPINK1, SPRED1, STK11, SUFU, TERT, TGFB1, TGFB2, TMEM127, TOX3, TP53, TSC1, TSC2, VHL, WRN, WTI, XPA, XPC, XRCC2, ZFXH3</i>

Patient Name: _____ Guardian Name: _____
(In case of minor)

Information on Inherited Cancer Genetic Testing :

5-10% cancer cases are hereditary in nature. Variations in certain genes often increase the risk for certain cancers. Genetic tests are recommended by your referring clinician with an aim to identify these disease causing variations either in genes with respect to the patient's symptoms and/or family history.

Next Generation Sequencing (NGS) based testing allows simultaneous assessment of multiple genes.

The various tests included in this category are:

1) BRCA1 & BRCA2 by NGS

Analysis is limited to BRCA1 and BRCA2 genes. Test results can help in deciding risk reducing measures and medical management. The test fit result could bene in assessing the risk of close blood relatives.

2) BRCA1 & BRCA2 by NGS + MLPA

Analysis of BRCA1 and BRCA2 genes for single nucleotide variations and copy number variations.

3) Organ specific and Comprehensive cancer panel

Genes evaluated based on personal and family history of cancer.

Variant Interpretation and Test Results

Variants are interpreted and scored according to a proprietary algorithm ORION Seek which incorporates the criteria defined by the American College of Medical Genetics. Since the ACMG criteria are not purely objective, inter laboratory variation in classification is known to occur. Similarly, variant classification may change over time, subject to accumulation of scientific information. Hence, it is requested to contact the laboratory for any new updates periodically, especially before contemplating prenatal testing or screening of "at risk" relatives. Variant predisposing an individual to inherited cancers has been identified. This may have implications to other family members as well.

Expected Test Results

Positive : Variant predisposing an individual to inherited cancers has been identified. This may have implications to other family members as well.

Negative : No variants related to patient phenotype or family history were detected (refer to test limitations).

Variants of Uncertain Significance : Implies detection of a variant whose significance is not known as of now Re-classification may be possible after accumulation of further variant specific/related data in medical literature. It is recommended to contact the laboratory for periodic of review variant classification especially before considering extended carrier screening.

Limitations of Genetic Testing

- A negative test result does not always exclude genetic basis to your condition or predisposition/ risk for developing a genetic disease in the future. Additional testing may be required in case of a negative report. In some cases the test may not detect a variation even though present in a protein coding area because of limitation in technology/scientific information.
- The current technology does not standardly analyze intronic variants, non-variant, splice nucleotides, repeat expansions & methylation abnormalities. Similarly coverage of gene promoters region may not be uniform or universal
- Copy number variations are known in *EPCAM* and *GREM1*. The Boland inversion is also known in *MSH2* gene. These variations however cannot be detected by the current test methodology.
- This test does not detect any copy number variations, balanced chromosomal rearrangements and large deletions/duplications.
- The accuracy of genetic test results is dependent of the information provided with relation to biological relation, clinical history and sample collection and transport. Contamination may interfere with results. In rare case due to insufficient DNA quantity or quality, a repeat sample may be required. The laboratory usually ensures timely dispatch of reports, however certain unanticipated delays may occur for which the laboratory cannot be held liable.
- The reports are released to your referring clinician as well as the patient/guardian (in case of minor). Since genetic test results are confidential, reports/ information regarding the results will not be released to any other person / clinician unless consent is provided by the patient

I have read and understood have been explained the above in language of my understanding and permit NCGM to perform the recommended genetic analysis.

I understand that the data derived from my genetic testing may be stored indefinitely as a part of the laboratory database. This data is always stored in de-identified form. I understand my de-identified data/sample may be used for research collaborations as well as scientific presentations and publications.

Name:

Signature:

Relationship to Patient:

Date, Time and Place:

Clinician Name & Signature:

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