

Name	: Ms. DUMMY	Centre Details	: ---
Age	: x y z Yrs.Sex: Female	Accession.ID	: ---
Collection Date	: ---	Referred By	: SELF
Received Date	: 09/Dec/2025 08:08PM	Report Date	: 31/Dec/2025 10:25 PM
Registration Date	: 09/Dec/2025 08:08PM	Ref.No/TRF.No	:

OncHereditary Cancer Panel

Sample Type: Whole Blood EDTA

Clinical History

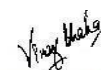
Status: Affected
 Type of cancer: Carcinoma left breast
 Investigations: Histopathology report (22-11-2025) – Invasive carcinoma of no special type (ductal), grade II, NBR score of 6.
 Pathological staging– pT2N3a

Test Results and Interpretation

NO SIGNIFICANT VARIANT RELATED TO PHENOTYPE DETECTED.
 MLPA is recommended for deletion/duplication analysis of BRCA1 and BRCA2 genes

Test Information

1. Clinical correlation as well as reverse phenotyping is recommended for all reports.
2. Genetic counseling for accurate interpretation of test results is recommended.
3. The reported findings are based on NGS analysis.
4. Analysis includes both single nucleotide (SNV) as well as copy number variant analysis (CNV).
5. Copy number variants when detected are included in the report.
6. Since CNV analysis is performed on a comparative basis, a negative result does not exclude the presence of a CNV.
7. The CNV pipeline is not validated for <3 exon copy number variants wherein detection is influenced by the underlying gene region and structure.
8. Variant calling (SNV and CNV) may be limited in low covered regions as well as in regions of low complexity and in pseudogenes.
9. Synonymous variants (not affecting splice site) as well as intronic variants are usually not reported.
10. Analysis and reporting is focussed on the provided phenotype and based on relevant HPO (Human Phenotype Ontology) terms as well as on genes associated with provided phenotype.
11. A genotype based analysis is also performed when the above yields negative results but reporting is limited to genes wherein current available evidence suggests a possible association with the provided phenotype.



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 Head- Molecular Biology
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Test Information

12. It may not be possible to fully resolve certain details about variants, such as mosaicism, phasing, or mapping ambiguity.
13. Disease descriptions are included from OMIM, Genereviews and PUBMED indexed articles as and where applicable.
14. The test methodology currently does not detect large deletions/duplications, triplet repeat expansions and epigenetic changes. The test also does not include analysis of predictors for multifactorial, polygenic and/or complex diseases.
15. Phenotype variability may be due to modifying genetic/non-genetic factors and is not a part of the current analysis.
16. Candidate genes and genes with limited evidence are designated as genes of uncertain significance and all variants detected therein are classified as variants of uncertain significance.
17. Typically only variants at a depth of $\geq 10X$ are reported. Lower depth variants may be false positives.
18. Detected variants in low complexity regions as well as variants at low depth and relatively low VAF should be reconfirmed by an alternate methodology.
19. CNV confirmation via MLPA or Exon array is recommended for copy number variants involving a single gene as well as for CNV $< 400kb$. Only large constitutional CNV can be tested via other array platforms.
20. Variant depth/Total depth has been mentioned in summary of the variants.
21. Parental/Maternal testing as applicable is recommended for variants when detected for phasing (where applicable)
22. Segregation analysis (testing of multiple affected as well as unaffected members) of detected variants (if any) is recommended. Variant classification is subject to change after segregation.
23. ACMG secondary findings as well as carrier status of variants are only provided when requested.
24. Within carrier screening risk factors/alleles as well as hypomorphic variants may not be included
25. Interobserver as well as inter-laboratory variation is known with respect to variant classification due to the subjective nature of the provided criteria. Though the laboratory follows the updated recommendations provided by the ClinGen SVI as well as ACMG, independent assessment of variant classification by the referring clinician is recommended before decision making.
26. For prenatal samples analysis is limited to provided clinical phenotypes utilizing relevant HPO terms. Typically variants of uncertain significance are only introduced if the respective gene has been associated with the observed fetal phenotype. In solo fetal exomes, variants of uncertain deemed to be disease causing based on genotype characteristics may be included for further evaluation by the referring clinician. In prenatal scenarios trio fetal testing is strongly recommended to allow better interpretation of detected variants.
27. If the above results do not correlate completely with patient phenotype, additional testing is advised based on clinician's discretion.
28. Typically, heterozygous variants of uncertain significance in genes associated with autosomal recessive disorder are not reported in the proband. Such variants are included if relevant to phenotype in carrier screening.
29. A negative report does not exclude a genetic disorder due to inherent limitations of the assay design.
30. On the background of whole exome sequencing, analysis is limited to provided indications/ requested testing. Hence analysis is limited to a single gene if the same has been requested.
31. As a part of knowledge sharing initiative, all reported variants are submitted in de-identified form in the ClinVar database.
32. Extracted DNA if available after requisitioned testing will be stored as per recommendations. Please note that DNA may degrade over time and this may affect the quality of the stored sample.
33. Collected blood samples are not stored
34. Prenatal samples (AF/CVS/cord blood) are not stored. Extracted DNA if available is stored as acknowledged above.
35. As per PCPNDT fetal gender is not revealed.
36. Maternal cell contamination is recommended for prenatal and POC (product of conception) to ensure accuracy of test results. The same is performed only when requested and on the availability of the maternal sample.
37. Discrepant maternal cell contamination results may rise with use of donor gamete and hence information regarding the same should be provided to the laboratory.
38. Reproductive decision making is not recommended based on variants of uncertain significance.
39. Raw data can be transferred on request and after due consent/assent from the involved patient/ family. Additional charges will be applicable for the same.
40. The test performed by the laboratory with the assumption that the sample belongs to the person herewith mentioned in the requisition form and appropriate consent as well as prenatal counseling including test information has been provided by the referring clinician.
41. Repeat sampling may be required in case of gender discrepancy (unless the same can be attributed to an underlying scientific reason) as well as in rare cases where DNA/data quality prevents further analysis.



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Test Information

42. Reanalysis of data is recommended as deemed necessary by the referring clinician. Additional charges will be applicable for the same.

Technical Notes

Methodology - Massively Parallel Sequencing (Next Generation Sequencing): Genomic DNA from the submitted specimen was enriched for the complete coding regions and splice site junctions of genes listed below using a custom bait- capture system. Paired End Sequencing was performed with 2x100/2x150 chemistry. Reads were assembled and were aligned to reference sequences based on NCBI RefSeq transcripts and human genome build GRCh37/UCSC hg19. Data was filtered and analyzed to identify variants of interest and interpreted in the context of a single most damaging, clinically relevant transcript for the purpose of the report, indicated as a part of variant details. Enrichment and analysis focus on the coding sequence of the indicated transcripts, 5-10bp of flanking intronic sequence, and other specific genomic regions demonstrated to be causative of disease at the time of assay design. Deletion and duplication analysis is performed in cases when indicated but detected variations need to be confirmed by an alternate methodology. Sequence and copy number variants are reported according to the Human Genome Variation Society (HGVS).

Laboratory reporting protocol: The analysis is based on the provided phenotype: relevant HPO terms, curated gene panels and relevant literature is assessed for phenotype based analysis. Variant reporting is limited to exon regions and upto 10 basepairs within exon intron boundaries. Previously reported deep intronic and non coding variants will be included when detected at a depth more than 10X . Variant reporting is performed at a minimum depth of 10X. The gnomAD variant frequency reflects the liftover of hg38 to hg19.

For Mitochondrial Genome Sequencing (if requested): Only phenotype-related Pathogenic and Likely Pathogenic variations reported in the MitoMap database as well as literature are reported. Haplogroups are not analyzed. A list of variants other than the above are available on request. Analyzed genes include:MT-ND1, MT-ND2, MT-ND3, MT-ND4L, MT-ND4, MT-ND5, MT-ND6, MT-CYB, MT-CO1, MT-CO2, MT-CO3, MT-ATP6, MT-ATP8, MT-RNR2, MT-RNR1, MT-RNR2, MT-TA, MT-TR, MT-TN, MT-TD, MT-TC, MT-TE, MT-TQ, MT-TG, MT-TH, MT-TI, MT-TL1, MT-TL2, MT-TK, MT-TM, MT-TF, MT-TP, MT-TS1, MT-TS2, MT-TT, MT-TW, MT-TY, MT-TV.

Tools and Databases employed for analysis: Clinvar, OMIM, HGMD, UCSC genome browser, Uniprot, Ensembl, dbSNP, gnomAD, ExAC, Pubmed, Dgap, icgc, Kaviar, various bioinformatics analysis, predictive tools and disease specific databases used as available and appropriate. Such tools/databases would be mentioned wherever used.

REVEL: The REVEL score for an individual missense variant can range from 0 to 1, with higher scores reflecting greater likelihood that the variant is disease-causing.

CADD: The variants with scores above 20 are predicted to be among the 1.0% most deleterious possible substitutions in the human genome.

Bioinformatics pipeline version: 15.10.8

Gene Coverage

Indication Based Analysis:

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
AAGAB	100%	ABRAXAS1	100%	ACD	100%	AIP	100%
AKR1B1	100%	AKT1	100%	ALK	100%	AMER1	100%
ANKRD26	99.3%	ANTXR1	99.8%	APC	100%	AQP5	100%
AR	100%	AREG	96.4%	ARTN	100%	ASXL1	100%

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Gene Coverage

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ATAT1	100%	ATG2B	100%	ATM	100%	ATP2C1	100%
ATR	100%	ATRX	100%	AURKAIP1	100%	AXIN1	100%
AXIN2	100%	BACH1	100%	BAP1	100%	BARD1	100%
BLM	100%	BMPR1A	100%	BRAF	100%	BRCA1	100%
BRCA2	100%	BRIP1	100%	BTNL2	100%	BUB1B	100%
CASR	100%	CAST	100%	CBL	100%	CD151	100%
CD70	100%	CD82	100%	CDC73	100%	CDH1	100%
CDK12	100%	CDK4	100%	CDKN1B	100%	CDKN1C	94.1%
CDKN2A	100%	CDSN	100%	CEBPA	100%	CEL	94.6%
CEP57	100%	CFH	100%	CFTR	100%	CHEK1	100%
CHEK2	100%	COL17A1	100%	COL7A1	100%	CPA1	100%
CSTA	100%	CTC1	100%	CTNNA1	100%	CTNNB1	100%
CTSC	100%	CYLD	100%	DDB2	100%	DDX41	100%
DGCR8	100%	DICER1	100%	DIS3L2	100%	DKC1	100%
DLST	100%	DNAJC21	100%	DSG1	100%	DSG2	100%
DSG4	100%	DSP	100%	DSPP	100%	DST	100%
EFL1	100%	EFNA1	100%	EGFR	100%	EGLN1	100%
ELAC2	100%	ELANE	100%	ENG	100%	ENPP1	98%
EPAS1	100%	EPCAM	100%	ERBB2	100%	ERC2	100%
ERCC1	100%	ERCC2	100%	ERCC3	100%	ERCC4	100%
ERCC5	100%	ERCC6L2	100%	ETV6	100%	EXO1	100%
EXPH5	100%	EXT1	100%	EXT2	100%	EZH2	100%
FAM111B	100%	FANCA	100%	FANCB	100%	FANCC	100%
FANCD2	100%	FANCE	100%	FANCF	100%	FANCG	100%
FANCI	100%	FANCL	100%	FANCM	100%	FAP	100%

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Gene Coverage

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
FBXW7	100%	FERMT1	100%	FGFR2	100%	FH	100%
FLCN	100%	FZR1	100%	G6PC3	100%	GALNT12	100%
GATA1	100%	GATA2	100%	GEN1	100%	GFI1	100%
GJB6	100%	GLMN	100%	GPC3	100%	GPR101	100%
GREM1	100%	GRIP1	100%	GSKIP	100%	HAVCR2	100%
HAX1	100%	HNF1A	100%	HNF1B	100%	HOXB13	100%
HRAS	100%	IKZF1	100%	ITGA3	100%	ITGA6	100%
ITGB4	100%	JUP	100%	KANK2	100%	KDM3B	100%
KIF1B	100%	KIT	100%	KITLG	100%	KLHL24	100%
KRAS	100%	KRT1	100%	KRT10	90.5%	KRT14	100%
KRT16	100%	KRT17	100%	KRT5	100%	KRT6A	100%
KRT6B	100%	KRT6C	100%	KRT9	100%	LAMA3	100%
LAMA4	100%	LAMB3	100%	LAMC2	100%	LDLR	100%
LGALS12	100%	LIG4	100%	LORICRIN	100%	LOXL2	100%
LSP1	100%	LZTR1	100%	MAD2L2	100%	MAGI1	100%
MAP2K1	100%	MAP2K2	100%	MAP3K1	100%	MAX	100%
MBD4	100%	MC1R	100%	MCPH1	100%	MECOM	100%
MEN1	100%	MET	100%	MITF	100%	MLH1	100%
MLH3	100%	MLLT1	99.3%	MMAB	100%	MMP1	100%
MRE11	100%	MRPL23	100%	MRPL36	100%	MSH2	100%
MSH3	100%	MSH6	100%	MSR1	100%	MUTYH	100%
MXI1	100%	MYCN	100%	MYSM1	100%	NAF1	100%
NBN	100%	NCOA2	100%	NF1	99.7%	NF2	100%
NHEJ1	100%	NHP2	100%	NOP10	100%	NQO2	100%
NRAS	100%	NSD1	100%	NSMCE3	100%	NSUN2	100%

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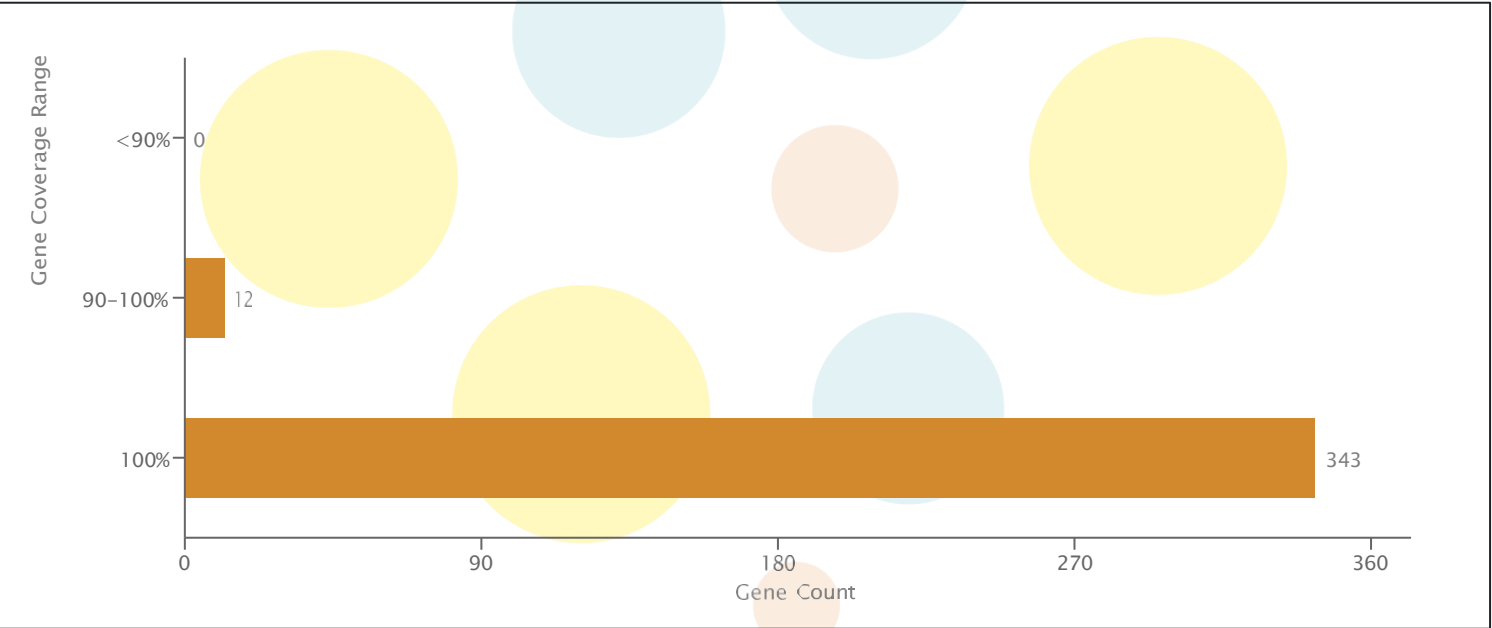
Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
NT5C3A	100%	NTHL1	100%	NYNRIN	100%	PALB2	100%
PALLD	100%	PARN	100%	PAX5	100%	PDGFRA	100%
PHOX2B	100%	PIK3CA	100%	PKP1	100%	PLEC	100%
PMS1	100%	PMS2	100%	POLD1	100%	POLE	100%
POLH	100%	POLR1G	100%	POMP	100%	POT1	100%
PPM1D	100%	PPP2R2A	100%	PRF1	100%	PRKAR1A	100%
PROC	100%	PRSS1	100%	PRSS58	100%	PTCH1	100%
PTEN	100%	PTPN11	100%	RAB43	100%	RAD50	100%
RAD51	100%	RAD51B	99.7%	RAD51C	100%	RAD51D	100%
RAD54L	100%	RAF1	100%	RASA2	100%	RASAL1	100%
RB1	100%	RECQL	100%	RECQL4	100%	REST	100%
RET	100%	RFWD3	100%	RHBDF2	100%	RINT1	100%
RIT1	100%	RNASE3	100%	RNASEL	100%	RNF2	100%
RNF43	100%	RNMT	100%	RPL11	100%	RPL15	100%
RPL17	100%	RPL23	100%	RPL26	100%	RPL27	100%
RPL31	100%	RPL35A	100%	RPL5	100%	RPL9	100%
RPS10	100%	RPS17	100%	RPS19	100%	RPS20	100%
RPS24	100%	RPS26	100%	RPS27	100%	RPS28	100%
RPS29	100%	RPS7	100%	RRAS	100%	RSPO1	100%
RTEL1	100%	RUNX1	100%	SAMD9	100%	SAMD9L	100%
SBDS	100%	SDHA	100%	SDHAF2	100%	SDHB	100%
SDHC	100%	SDHD	100%	SERPINB7	100%	SERPINB8	100%
SHOC2	100%	SIX1	100%	SIX2	100%	SLC12A3	100%
SLC25A11	100%	SLURP1	100%	SLX4	100%	SMAD4	100%
SMAD9	100%	SMARCA4	100%	SMARCB1	100%	SMARCE1	100%

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Gene Coverage

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
SOS1	100%	SOS2	100%	SPINK1	100%	SPRED1	100%
SRP72	100%	STK11	100%	SUFU	100%	TAT	100%
TERT	100%	TESC	100%	TGFB1	100%	TGFBR2	100%
TGM5	100%	TINF2	100%	TMC6	100%	TMEM127	100%
TNIP1	100%	TOX3	100%	TP53	100%	TPSAB1	100%
TRIM28	99%	TRIM37	100%	TRIP13	100%	TRPV3	100%
TSC1	100%	TSC2	100%	TSR2	100%	UBE2T	100%
VHL	100%	WRAP53	100%	WRN	100%	WT1	100%
XPA	100%	XPC	100%	XRCC2	100%	XRCC3	100%
ZCCHC8	100%	ZFHX3	100%	ZNF395	100%		

Gene Coverage Distribution



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QC Metrics

Total aligned reads	99.95 %
Total reads	53.82 (M)
Total data generated	7.99 (Gb)
Total reads which passed mapping quality cutt-off	7.61 (Gb)

Disclaimer:

1. The report is validated by third party lab
2. Test results are dependent on the quality of the sample received by the lab
3. Tests are performed as per schedule given in the test listing and in any unforeseen circumstances, report delivery may be delayed
4. Test results may show interlaboratory variations
5. All dispute and claims are subjected to local jurisdiction only. Clinical correlation advised
6. Test results are not valid for medico legal purposes
7. For all queries, feedback, suggestions, and complaints, please contact customer care support +0124 665 0000



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