

Test Name : Comprehensive Hereditary Cancer Gene Panel (SNVs, InDels & Large Deletions/Duplications)

Report Details

Sample ID / Order ID: 9623545/1560701
 Date of Order 2025-12-16 12:28:07
 Booking:
 Date of Report: 2nd January 2026
 Report Status: Final

Specimen Information

Sample Type: Peripheral Blood in EDTA (Purple Top)
 Date of Sample 15th December 2025
 Collection:
 Date of Sample 16th December 2025
 Receipt:

Ordering Clinician

Referring Clinician: Dr. Ashwin Oomen Philips,
 Hospital Name: Amala Institute Of Medical Sciences
 Serviced By: NA

Clinical Indication

Clinical Symptoms: Metastatic carcinoma, breast.

Family History: Sister - Breast cancer.
 Aunt - ?Endometrial cancer.

Result

Variant of uncertain significance (VUS) was detected by NGS.

No clinically significant deletions/duplications were detected by NGS/MLPA.

Primary Findings

Gene [#] (Transcript)	Variant Type	Variant	Zygosity	Disease (OMIM)	Inheritance	Classification\$
ATM (+) (ENST00000675843.1)	SNV	c.6624C>A p.His2208Gln MISSENSE	Heterozygous 71X(49.3%)	{Breast cancer, susceptibility to} (OMIM#114480)	Autosomal dominant	Uncertain Significance

ATM (c.6624C>A: p.His2208Gln): Uncertain Significance

Nucleotide change: chr11:108325361:C>A

Protein change: p.His2208Gln

cDNA change: c.6624C>A

Transcript ID: ENST00000675843.1

Zygosity: Heterozygous

Total depth: 71x

Variant Consequence: MISSENSE

Location: Exon 46

Variant Allele Depth: 35x

Variant Evidences:

- The observed variation is documented as variant of uncertain significance in hereditary cancer-predisposing syndrome in the ClinVar database [1051738].
- It lies in the FAT domain of the ATM_HUMAN protein [PF02259].
- The p.His2208Gln variant has not been reported in 1000 genomes, gnomAD (v3.1), gnomAD (v2.1), and topmed databases and has a minor allele frequency of 0.002% in our internal database.
- The *in silico* prediction[#] of the variant is damaging by LRT and Mutation Taster-2 tools.
- The reference codon is conserved across species.

OMIM Phenotype: Susceptibility to breast cancer (OMIM#114480) is caused by mutations in the ATM gene (OMIM#607585).

Based on the above evidence^{\$}, this ATM variation is classified as a variant of uncertain significance and has to be carefully correlated with the clinical symptoms.

Additional Information



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- No other SNV(s)/INDELS, Repeat expansions or CNV(s) that warrants to be reported were detected. All the genes covered in this assay have been screened for the given clinical indications. To view the coverage of all genes [Click here](#). Test methodology details of this assay are given in the appendix.
- Genetic test results are reported based on the recommendations of American College of Medical Genetics and Genomics (ACMG) [PMID: [25741868](#), [31690835](#), [32906214](#)].
- A negative genetic test result does not preclude the genetic basis of the individual's personal (and/or family) history of the disease which could also be due to variation in a genomic region not covered by the test or due to the inherent technical limitations of the test. Please write an email to genetic.counseling@medgenome.com in case you need assistance for genetic counselling. For any further technical queries please write an email to techsupport@medgenome.com

Recommendations

- Validation of the variant(s) by Sanger sequencing is recommended to rule out false positives.
- Sequencing the variant(s) in the parents and the other affected and unaffected members of the family is recommended to confirm significance. For more details on who should be tested, kindly consult your clinician or <https://diagnostics.medgenome.com/brca-genetic-testing-india>.**
- Genetic counselling is recommended to discuss the implications of the test results. Kindly email us at genetic.counselling@medgenome.com for post-test counselling session.
- Kindly consult with your clinician to discuss the surveillance measures and disease management options.



Lakshmi Mahadevan, PhD

Principal Scientist

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Test Methodology

NGS

Targeted gene sequencing: Selective capture and sequencing of the protein coding regions of the genome/genes included in the assay, is performed. Mutations identified in the exonic regions are generally of clinical relevance compared to variations that occur in non-coding regions. Targeted sequencing represents a cost-effective approach to detect variants present in multiple/large genes in an individual. In this hereditary cancer panel, in addition to complete coding segment (CDS), promoter regions of relevant genes and other critical reported/pathogenic variations in the non-coding / coding regions that are documented in the ClinVar, HGMD, BRCA Exchange and LOVD databases mapping to the targeted genes are also included [PMID: [26582918](#), PMID: [28349240](#), PMID: [30586411](#), PMID: [21520333](#)]. Additionally, there is an enhanced CNV coverage for better detection of CNVs. This panel provides a comprehensive and robust approach to identify SNV's, Indels and CNVs through single test.

DNA extracted from blood was used to perform targeted gene capture using a custom capture kit. The libraries were sequenced to mean >80-100X coverage on Illumina sequencing platform. We follow the GATK best practices framework for identification of variants in the sample using *Sentieon* (v201808.07) [<https://europepmc.org/article/PPR/PPR28504>]. The sequences obtained are aligned to human reference genome (GRCh38.p13) using *Sentieon* aligner [PMID: [20080505](#)] and analyzed using *Sentieon* for removing duplicates, recalibration and re-alignment of indels. *Sentieon* haplotype caller has been used to identify variants which are relevant to the clinical indication. Gene annotation of the variants is performed using *VEP* program [PMID: [20562413](#)] against the Ensembl release 99 human gene model [PMID: [29155950](#)]. Copy number variants (CNVs) are detected from targeted sequence data using the *ExomeDepth* (v1.1.10) method, a coverage-based approach [PMID: [22942019](#)]. This algorithm detects rare CNVs based on comparison of the read-depths of the test data with the matched aggregate reference dataset and the overall sensitivity of CNV surveillance through *ExomeDepth* was found to be 97% [PMID: [28378820](#)]. In our internal validation experiments on MLPA verified samples, >80-90% sensitivity was achieved for detecting CNVs.

Clinically relevant mutations were annotated using published variants in literature and a set of diseases databases - ClinVar, OMIM (updated on 11th May 2020), GWAS, HGMD (v2020.2), LOVD, BRCA Exchange and SwissVar [PMID: [265829183](#), PMID: [28349240](#), PMID: [30586411](#), PMID: [21520333](#), PMID: [17357067](#), PMID: [24316577](#), PMID: [20106818](#)]. Common variants are filtered based on allele frequency in 1000Genome Phase 3, gnomAD (v3.1), EVS, dbSNP (v151), 1000 Japanese Genome and our internal Indian population database [PMID: [26432245](#), PMID: [11125122](#), PMID: [32461654](#), PMID: [26292667](#), <https://esp.gs.washington.edu/drupal/>, <https://www.nature.com/articles/ncomms9018>]. Non-synonymous variants effect is calculated using multiple algorithms such as *PolyPhen-2*, *SIFT*, *MutationTaster2* and *LRT*. Only non-synonymous, splice site and critical non-coding variants found in the analysed genes were used for clinical interpretation. Silent variations that do not result in any change in amino acid in the coding region are not reported.

MLPA

Digital MLPA is a semi-quantitative technique that is used to determine the relative copy numbers and detect specific (point) mutations. Like conventional MLPA, it is based on the sample DNA-dependent generation of ligated probe products, followed by PCR amplification of ligated probes by a single PCR primer pair. Digital MLPA generates PCR amplicons that are quantified using Illumina NGS platforms. Sequencing is used to determine the read numbers of each digital MLPA probe amplicon. *Coffalyser* digital MLPA software from MRC Holland is used for the analysis of digital MLPA data. *Coffalyser* digital MLPA automatically recognizes digital MLPA sequence reads from FASTQ files and directly uses them for analysis. *Coffalyser* digital MLPA then prepares two clear reports for every sample indicating both the quality of the run and the aberrations found.

The SALSA digital MLPA D001 Hereditary Cancer Panel 1, assay intended for the detection of exon deletions or duplications in the oncogene screening, which are associated with hereditary predisposition for formation of breast, ovarian, colorectal, gastric, prostate, pancreatic or endometrial tumors, or for melanoma. A total number of approximately 690 probes is included in this D001 Probemix, this consists of 558 probes detecting copy number alterations involved in hereditary cancer (Appendix 1). Five mutation-specific probe(s), which will only generate reads when that particular mutation is present (Appendix 1). Three wild type specific probes, which detect the wild type sequence of a particular mutation. More than 120 control probes and fragments: these include probes for sample identification, probes for the detection of errors or deviations when performing digital MLPA assays, impurities in and fragmentation of the DNA samples, ligase and polymerase activity and extent of hybridization.

Copy number variation (CNV) ratios of below 0.7 or above 1.3 are indicative of a deletion (copy number change from two to one) or duplication (copy number change from two to three), respectively. A CNV ratio of 0.0 indicates a homozygous/hemizygous deletion, 0.35 to 0.65 indicates heterozygous deletion, 1.35 to 1.55 indicates heterozygous duplication and 1.7 to 2.2 indicates homozygous/hemizygous duplication. A ratio between 0.80 to 1.20 indicates a normal copy number status.

Coverage Statistics

Coverage statistics provide an overview of the sequencing depth and coverage uniformity across the target regions.

Average sequencing depth (x)	Average on-target sequencing depth (x)	Percentage target base pairs covered		
		0x	>=5x	>=20x
132	130.25	0.17	99.71	98.92

QC Statistics

QC Statistics provides key metrics on the quality of the sequencing data obtained.

Total data generated (Gb)	Total reads aligned (%)	Data ? Q30 (%)
0.76	99.99	97.82

ACMG Classification Criteria

\$The classification of the variants is done based on American College of Medical Genetics as described below [PMID: [25741868](#)].

Variant	A change in a gene. This could be disease causing (pathogenic) or not disease causing (benign).
Pathogenic	A disease-causing variant in a gene which can explain the patient's symptoms has been detected. This usually means that a suspected disorder for which testing had been requested has been confirmed.
Likely Pathogenic	A variant which is very likely to contribute to the development of disease however, the scientific evidence is currently insufficient to prove this conclusively. Additional evidence is expected to confirm this assertion of pathogenicity.
Variant of Uncertain Significance	A variant has been detected, but it is difficult to classify it as either pathogenic (disease causing) or benign (non-disease causing) based on current available scientific evidence. Further testing of the patient or family members as recommended by your clinician may be needed. It is probable that their significance can be assessed only with time, subject to availability of scientific evidence.

#The transcript used for clinical reporting generally represents the canonical transcript (MANE Select), which is usually the longest coding transcript with strong/multiple supporting evidence. However, clinically relevant variants annotated in alternate complete coding transcripts could also be

reported. Variants annotated on incomplete, and nonsense mediated decay transcripts will not be reported.

#The in-silico predictions are based on Variant Effect Predictor (v104), [SIFT version - 5.2.2; PolyPhen - 2.2.2; LRT version (November 2009); CADD (v1.6); Splice AI; dbNSFPv4.2] and MutationTaster2 predictions are based on NCBI/Ensembl 66 build (GRCh38 genomic coordinates are converted to hg19 using UCSC LiftOver and mapped to MT2). Diseases databases used for annotation includes ClinVar (updated on 17042023), OMIM (updated on 01092023), HGMD (v2023.1), LOVD (Nov-18), DECIPIER (population CNV) and SwissVar.

Limitations

- Genetic testing is an important part of the diagnostic process. However, genetic tests may not always give a definitive answer. In some cases, testing may not identify a genetic variant even though one exists. This may be due to limitations in current medical knowledge or testing technology. Accurate interpretation of test results may require knowing the true biological relationships in a family. Failing to accurately state the biological relationships in the {my/my child's} family may result in incorrect interpretation of results, incorrect diagnoses, and/or inconclusive test results.
- Test results are interpreted in the context of clinical findings, family history and other laboratory data. Only variations in genes potentially related to the proband's medical condition are reported. Rare polymorphisms may lead to false negative or positive results. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.
- Structural variants such as deletions/duplications (CNVs) reported through NGS assay need to be confirmed by orthogonal method to rule out the possibility of false positives. Translocations, repeat expansions and chromosomal rearrangements are not detected in this assay.
- Genetic testing is highly accurate. Rarely, inaccurate results may occur for various reasons. These reasons include, but are not limited to, mislabelled samples, inaccurate reporting of clinical/medical information, rare technical errors or unusual circumstances such as bone marrow transplantation, blood transfusion; or the presence of change(s) in such a small percentage of cells that may not be detectable by the test (mosaicism).
- The variant population allele frequencies and in-silico predictions for GRCh38 version of the Human genome is obtained after lifting over the coordinates from hg19 genome build. The existing population allele frequencies (1000Genome, gnomAD-Exome) are currently available for hg19 genome version only. This might result in some discrepancies in variant annotation due to the complex changes in some regions of the genome.

Disclaimers

- Low coverage was observed in certain regions of the *RB1* gene. This is likely to be due to the genomic complexity in these regions.
- Interpretation of variants in this report is performed to the best knowledge of the laboratory based on the information available at the time of reporting. The classification of variants can change over time and MedGenome cannot be held responsible for this. Please feel free to contact MedGenome Labs (techsupport@medgenome.com) in the future to determine if there have been any changes in the classification of any variations. Re-analysis of variants in previously issued reports in light of new evidence is not routinely performed but may be considered upon request, provided the variant is covered in the current panel.
- It is also possible that a pathogenic variant is present in a gene that was not selected for analysis and/or interpretation in cases where insufficient phenotypic information is available.
- Genes with pseudogenes, paralog genes and genes with low complexity may have decreased sensitivity and specificity of variant detection and interpretation due to inability of the data and analysis tools to unambiguously determine the origin of the sequence data in such regions.
- Incidental or secondary findings (if any) that meet the ACMG guidelines [[PMID: 27854360](#)] can be given upon request.
- The report shall be generated within turnaround time (TAT), however, such TAT may vary depending upon the complexity of test(s) requested. MedGenome under no circumstances will be liable for any delay beyond afore mentioned TAT.
- It is hereby clarified that the report(s) generated from the test(s) do not provide any diagnosis or opinion or recommends any cure in any manner. MedGenome hereby recommends the patient and/or the guardians of the patients, as the case may be, to take assistance of the clinician or a certified physician or doctor, to interpret the report(s) thus generated. MedGenome hereby disclaims all liability arising in connection with the report(s).
- In a very few cases genetic test may not show the correct results, e.g. because of the quality of the material provided to MedGenome. In case where any test provided by MedGenome fails for unforeseeable or unknown reasons that cannot be influenced by MedGenome in advance, MedGenome shall not be responsible for the incomplete, potentially misleading or even wrong result of any testing if such could not be recognised by MedGenome in advance.
- This is a laboratory developed test and the development and the performance characteristics of this test was determined by MedGenome. The sensitivity of an NGS assay to detect large deletions/duplications of >10 bp or copy number variations (CNV) is 80-90%. The CNVs detected are recommended to be confirmed by alternate method.
- Due to inherent technology limitations of an NGS assay, not all bases of the exome can be covered by this test. Accordingly, variants in regions that are not covered may not be identified and/or interpreted. Therefore, it is possible that certain variants are present in one or more of the genes analysed but have not been detected. The variants not detected by the assay that was performed may/may not impact

the phenotype.

- The mutations detected in the NGS assay have not been validated/confirmed by Sanger sequencing.
- In most populations, the major cause of genetic defects in the genes covered by the *D001* probemix are small (point) mutations, most of which will not be detected by using an MLPA probemix.
- Digital MLPA cannot detect any changes that lie outside the target sequence of the probes and will not detect most copy number neutral inversions or translocations. Even when digital MLPA did not detect any aberrations, the possibility remains that biological changes in that gene or chromosomal region do exist but remain undetected.
- Sequence changes (e.g. SNPs, point mutations, small indels) in the target sequence detected by an MLPA probe can cause false positive results. Mutations/SNPs (even when >20 nt from the probe ligation site) can reduce the probe read count by preventing ligation of the probe oligonucleotides or by destabilizing the binding of a probe oligonucleotide to the sample DNA.
- The boundaries (positions of start and end) of the deletion/duplication detected, cannot be deciphered by digital MLPA.
- Variants in untranslated region, promoters and deep intronic regions are not analyzed in the MLPA test.

APPENDIX

Digital MLPA assay covering hereditary Cancer genes:

Position (hg38)	Gene	NM sequence and LRG	# probes / # exons	Gene length (kb)	Remarks
1p34.1	<i>MUTYH</i>	NM_001128425.1 LRG_220	16/16	11.2	
2p21	<i>EPCAM</i>	NM_002354.2 LRG_215	5/9	17.7	exon 7-9 covered; three probes between <i>EPCAM</i> & <i>MSH2</i>
2p21	<i>MSH2</i>	NM_000251.2 LRG_218	26/16	80.1	Includes two probes for recurrent 10 Mb 2p inversion
2p16.3	<i>MSH6</i>	NM_000179.2 LRG_219	19/10	23.9	
2q35	<i>BARD1</i>	NM_000465.4 LRG_297	15/11	84.1	
3p22.2	<i>MLH1</i>	NM_000249.3 LRG_216	24/19	57.5	
3p21.1	<i>BAP1</i>	NM_004656.2 LRG_529	15/17	9.0	
3p13	<i>MITF</i>	NM_198159.3	1 probe		c.952G>A (p.E318K)
5q22.2	<i>APC</i>	NM_000038.4 NM_001127510.1 NM_001127511.1; LRG_130	34/18	138.7	
7p22.1	<i>PMS2</i>	NM_000535.5 LRG_161	29/15	35.9	Includes one intron 7 SVA insertion specific probe
8q21.3	<i>NBN</i>	NM_002485.4 LRG_158	18/16	51.3	
9p21.3	<i>CDKN2A</i>	NM_058195.3 NM_000077.4 LRG_011	13/4	26.7	
10q23.2	<i>BMPR1A</i>	NM_004329.3 LRG_298	18/13	168.5	
10q23.31	<i>PTEN</i>	NM_000314.4 LRG_311	23/9	105.3	Includes three flanking probes in the <i>KLLN</i> gene
11q22.3	<i>ATM</i>	NM_000051.3 LRG_135	66/63	146.3	
12q14.1	<i>CDK4</i>	NM_000075.2 LRG_490	9/8	4.2	

Position (hg38)	Gene	NM sequence and LRG	# probes / # exons	Gene length (kb)	Remarks
12q24.33	<i>POLE</i>	NM_006231.3 LRG_789	1 probe		Wildtype sequence at c.1270C>G mutation (p.Leu424Val)
13q13.1	<i>BRCA2</i>	NM_000059.3 LRG_293	42/27	84.2	Includes one probe for exon 3 ALU insertion c.156_157
15q13.3	<i>SCG5</i> <i>GREM1</i>	NM_001144757.2 NM_013372.6	6 probes	Region covered ~68kb	Detects recurrent 40-kb duplication
16p12.2	<i>PALB2</i>	NM_024675.3 LRG_308	18/13	38.2	Includes one flanking probe in the <i>DCTN5</i> gene
16q22.1	<i>CDH1</i>	NM_004360.3 LRG_301	20/16	98.2	
17p13.1	<i>TP53</i>	NM_000546.5 LRG_321	14/11	19.1	
17q12	<i>RAD51D</i>	NM_002878.3 LRG_516	11/10	20.1	
17q21.31	<i>BRCA1</i>	NM_007294.3 LRG_292	38/23	81.2	Exon numbering different from LRG/NG sequence
17q22	<i>RAD51C</i>	NM_058216.3 LRG_314	11/9	41.3	Includes one flanking probe in the <i>TEX14</i> gene
17q23.2	<i>BRIP1</i>	NM_032043.2 LRG_300	22/20	184.4	Includes one flanking probe in the <i>INTS2</i> gene
18q21.2	<i>SMAD4</i>	NM_005359.5 LRG_318	17/12	54.8	
19p13.3	<i>STK11</i>	NM_000455.4 LRG_319	17/10	22.6	
22q12.1	<i>CHEK2</i>	NM_007194.3 LRG_302	18/15	54.1	Includes one probe for 1100delC mutation and one flanking probe in the <i>HSCB</i> gene

Appendix

GENES COVERED IN THE NGS ASSAY

<i>PRF1</i>	<i>CDKN1B</i>	<i>CDK4</i>	<i>NTHL1</i>	<i>CDH1</i>
<i>FH</i>	<i>SBDS</i>	<i>ERCC5</i>	<i>SDHC</i>	<i>FANCE</i>
<i>TYR</i>	<i>SMARCB1</i>	<i>CHEK1</i>	<i>SMARCA4</i>	<i>ENG</i>
<i>CD82</i>	<i>SUFU</i>	<i>NBN</i>	<i>WT1</i>	<i>FAN1</i>
<i>XPA</i>	<i>MLH1</i>	<i>RNF139</i>	<i>HRAS</i>	<i>TP53</i>
<i>MLH3</i>	<i>PTEN</i>	<i>RNF43</i>	<i>LZTR1</i>	<i>MITF</i>
<i>HNF1A</i>	<i>FANCL</i>	<i>SETD2</i>	<i>APC*</i>	<i>SMAD4</i>
<i>STK11</i>	<i>TGFBR2</i>	<i>MEN1</i>	<i>FANCA</i>	<i>MRE11</i>

<i>ELAC2</i>	<i>CDKN2A</i>	<i>EXT1</i>	<i>RECQL</i>	<i>PPP2R2A</i>
<i>NSD1</i>	<i>ERCC3</i>	<i>PTCH1</i>	<i>HNF1B</i>	<i>RECQL4</i>
<i>MSH3</i>	<i>RAD51B</i>	<i>MET</i>	<i>RAD51D</i>	<i>BLM</i>
<i>ABRAXAS1</i>	<i>SDHB</i>	<i>MAX</i>	<i>RINT1</i>	<i>RNASEL</i>
<i>FANCI</i>	<i>BRCA2</i>	<i>NF2</i>	<i>CUL2</i>	<i>SRGAP1</i>
<i>DDX41</i>	<i>RAD54L</i>	<i>CEP57</i>	<i>FANCF</i>	<i>TSC2</i>
<i>AR</i>	<i>FLCN</i>	<i>XRCC3</i>	<i>ERCC4</i>	<i>AIP</i>
<i>ESR2</i>	<i>KIT</i>	<i>PRKAR1A</i>	<i>BRCA1</i>	<i>EXT2</i>
<i>RAD50</i>	<i>CTNNA1</i>	<i>GATA2</i>	<i>DIS3</i>	<i>AKT1</i>
<i>PAX5</i>	<i>GPC3</i>	<i>HOXB13</i>	<i>NOTCH1</i>	<i>KIF1B</i>
<i>PDGFRA</i>	<i>DIS3L2</i>	<i>BMPR1A</i>	<i>PALLD</i>	<i>CHEK2</i>
<i>TERT*</i>	<i>MUTYH</i>	<i>FANCC</i>	<i>TSC1</i>	<i>BARD1</i>
<i>TMEM127</i>	<i>SMARCE1</i>	<i>BRIP1</i>	<i>RET</i>	<i>XPC</i>
<i>RUNX1</i>	<i>SLX4</i>	<i>SDHD</i>	<i>MED12</i>	<i>NF1</i>
<i>PALB2</i>	<i>CDC73</i>	<i>FANCB</i>	<i>RB1</i>	<i>CDK12</i>
<i>RAD51C</i>	<i>SDHA</i>	<i>DDB2</i>	<i>VHL</i>	<i>SPOP</i>
<i>WRN</i>	<i>BAP1</i>	<i>SLC45A2</i>	<i>EGFR</i>	<i>MSH2</i>
<i>EPCAM</i>	<i>CYLD</i>	<i>DICER1</i>	<i>CBL</i>	<i>PRSS1</i>
<i>RHBDF2</i>	<i>SDHAF2</i>	<i>CEBPA</i>	<i>AXIN2</i>	<i>PTCH2</i>
<i>ERCC2</i>	<i>BUB1B</i>	<i>MXI1</i>	<i>PHOX2B</i>	<i>PBRM1</i>
<i>GALNT12</i>	<i>FANCG</i>	<i>FANCD2</i>	<i>ALK</i>	<i>POLD1</i>
<i>EZH2</i>	<i>PMS2</i>	<i>MSR1</i>	<i>MSH6</i>	<i>PMS1</i>
<i>POT1</i>	<i>ATM</i>	<i>CDKN1C</i>	<i>POLE</i>	<i>ERBB2</i>
<i>FANCM</i>	<i>XRCC2</i>	<i>GFAP</i>		

*Promoter regions of these genes are also analysed.

[^]In addition to complete CDS coverage in these genes, critical non-coding variants reported as pathogenic in clinical databases are also analysed in this assay.

END OF REPORT
