

***Trust Logo***

**<GLH region name>**

**NHS Genomic Laboratory Hub**

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| ***Head of Department***  *Name* |  | *Local Genetics Service*  *Local Trust*  *Address*  *Address*  *Post Code*  *Web site address* |
| General Enquiries: *telephone contact*  Email: *generic email address* |
|  | | |

**GENOMIC LABORATORY REPORT**

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| Dr xxx | **Patient Name:** | **Jane DOE** |
| Consultant | Gender: | Female |
| <<Hospital address>> | Date of Birth: | 14 Jan 1968 |
| NHS No: | 123 456 7890 |
| Hospital No: | NK |
| Your ref: | GC12345 |

**Reason for testing**

Diagnostic testing. <<Referral reason>>. Patient phenotype / HPO terms

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| --- |
| **Result summary** |
| **A hereditary (germline) genetic cause for the patient’s cancer has not been identified** |

**Result**

No pathogenic variants were detected in the genes in this panel.

**Implications**

This testing excludes >99% of pathogenic variants in these genes. This result reduces the chance of, but does not completely exclude, a diagnosis of hereditary breast and ovarian cancer.

Date issued: <AUTHORISEDDATE> Authoriser: Clinical Scientist

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**TECHNICAL INFORMATION**

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| **Sequence analysis** | **Dosage analysis** |
| No pathogenic variants detected | No copy number variants detected |

**Test methodology**

1. Genes screened in the panel: *BRCA1; BRCA2;* *PALB2, ATM, CHEK2* (all coding exons & exon-intron boundaries). **For *ATM* & *CHEK2* genes only clearly truncating variants (nonsense, frameshift, ±1/2 splice & CNVs) in these genes, plus the *ATM* c.7271T>G p.(Val2424Gly) pathogenic missense variant, are reported.**
2. Methodology including sensitivity, CNV detection, Bioinformatics pipeline etc e.g. Enrichment method: Agilent SureSelect Custom Design, sequenced on the Illumina NextSeq platform with a sensitivity of at least 95%.The target regions of selected transcripts is covered to a minimum read depth of 30x.
3. Screening for large deletions and duplications is performed using comparative depth of coverage of NGS data. Deletions/duplications are confirmed by Multiplex Ligation-Dependent Probe Amplification (MRC-Holland).
4. Limits of detection e.g.NGS technical sensitivity may be reduced for genes with pseudogenes or paralogs, and copy-number variation >xx nucleotides.
5. Variant classification according to the American College of Medical Genetics and Genomics (ACMG)1, Association for Clinical Genomic Science (ACGS) 2020 guidelines2 and Cancer Variant Interpretation Group-UK gene specific and consensus specification for Cancer Susceptibility Genes3 (<https://www.cangene-canvaruk.org/canvig-uk>; <http://www.canvaruk.org/>) and ClinGen Hereditary Breast, Ovarian and Pancreatic Cancer Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for ATM Version 1.1 Expert Panel Page: <https://www.clinicalgenome.org/affiliation/50039>4

1Richards *et al.* (2015) Genetics in Medicine 17:405-24. (PMID 25741868)

2www.acgs.uk.com/quality/best-practice-guidelines

3 Garrett et al (2020) J Med Genet (PMID: 32170000); <https://www.cangene-canvaruk.org/canvig-uk>;

4 <https://clinicalgenome.org/site/assets/files/7451/clingen_hbop_acmg_specifications_atm_v1_1.pdf>

1. Only relevant results are shown; full details of methods and results, including benign/likely benign variants and variants of uncertain clinical significance, are stored on file and are available on request.

**Sample details**

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| Your lab ref: | 122001180 |  |  |
| Sample ID: | 1234567 | Sample collected: | 05 Jun 2020 |
| Sample type: | DNA from peripheral blood | Sample received: | 05 Jun 2020 |