

***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**

Predictive testing. <<Referral reason>>

|  |
| --- |
| **Result summary** |
| **Familial <likely> pathogenic <*GENE*> variant NOT detected** |

**Result**

Sequence/<dosage> analysis has shown no evidence of the familial <likely> pathogenic <*GENE*> variant.

**Implications**

This individual is **not** at significantly increased risk of developing familial <*GENE>*-associated cancer. Their residual risk for cancer susceptibility is dependent on their personal and familial history.

This individual’s descendants are not at risk for the familial <likely> pathogenic variant.

Date issued: <AUTHORISEDDATE> Authoriser: Clinical Scientist

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**TECHNICAL INFORMATION**

**Familial variant details**

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| Gene | Zygosity | HGVS description | Location: GRCh37 (hg37) | \*Classification  |
| *GENE* | **Variant NOT detected in <PATIENTFULLNAME>** | NM\_xxx c.xxT>C p.(Xxx) | ChrX(GRCh37):g.xxxxxxA>G | <Likely> Pathogenic |

**Test methodology**

1. METHODOLOGY e.g. Genomic DNA Sanger sequencing with direct chromatogram check & >95% sensitivity
2. Only clinically relevant results are shown; full details of methods and results, including benign/likely benign variants and variants of uncertain clinical significance with limited evidence for pathogenicity are available on request.
3. Variant classification according to the American College of Medical Genetics and Genomics (ACMG)1 and Association for Clinical Genomic Science (ACGS) 2020 guidelines2 and Cancer Variant Interpretation Group-UK 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)

2[www.acgs.uk.com/quality/best-practice-guidelines](file:///%5C%5CSJUHBB%5CDATA%5CDEPT%5CYRGS%5Cdnalabs%5CDarwin%5CReport%20footnotes%20%26%20templates%5CACGS%20BRCA%20report%20templates%5CProposed%20predictive%20templates%5Cwww.acgs.uk.com%5Cquality%5Cbest-practice-guidelines)

3 Garrett et al (2020) J Med Genet (PMID: 32170000)

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

**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 |