GENETIC TEST REPORT

Patient Details:		Test requested by:		Test carried out by:	
Name:	Jane Doe	Name:	Dr Requesta	Laboratory:	Gentest UK
Date of birth:	18 March 1970	Organisation:	Chester Hospital	Telephone:	01223 666555
Sex:	Female	Telephone:	01223 555555	Date received:	26 February 2018
NHS number:	NH00198	Copies to:	Dr A. Nother Dr X. Tra	Date reported:	12 March 2018
Sample type:	Blood			Authorised by:	A Tester

REASON FOR TEST

A test was requested to determine if your cancer is related to alterations in the *BRCA1*, *BRCA2*, or *PALB2* genes.

ABOUT THE TEST

This test looked at genes called *BRCA1*, *BRCA2* and *PALB2*. Specific alterations to these genes increase the risk of developing particular kinds of cancers.

YOUR RESULT: No gene alterations related to an increased risk of cancer were found. This means your cancer is not due to known alterations in the genes we looked at.

WHAT THIS RESULT MEANS FOR YOU

Genes are small sections of your DNA that affect, for example, what you look like, or your chances of developing a specific health condition.

This test did not find any alterations associated with an increased risk of cancer in the *BRCA1*, *BRCA2* or *PALB2* genes. This means that your cancer diagnosis is unlikely to be associated with *BRCA1*, *BRCA2*, or *PALB2*. It is still possible that your diagnosis of cancer has a genetic cause as alterations in other genes or combinations of genes can increase cancer risk. However, many cancers develop spontaneously and are not inherited.

The cancer risk for you and your relatives depends on many factors, including your family history. You can speak with your GP about what these results mean for your overall level of risk. If you have a family history of cancer, your doctor may wish to consider referring you to the Clinical Genetics Service for further advice.

We did not find a gene alteration in your sample, so these results cannot explain why your cancer developed. This means we cannot offer a genetic test to your relatives at this time. Your relatives should seek advice from their GPs to learn more about their risk of developing the same or related cancers.

NEXT STEPS

- **Speak with your doctor about what this result means for you.**
- **If your relatives would like advice about their own risks for cancer, they should speak with their GPs.**

Regardless of the result of this test, everybody is encouraged to be body aware. Both sexes, men and women, should be "chest aware", it's as simple as TLC: Touch, Look and Check. Further information about how to check your chest for signs of breast cancer can be found at:
https://www.nhs.uk/common-health-questions/womens-health/how-should-i-check-my-breasts/

For females, the symptoms of ovarian cancer vary (see https://www.nhs.uk/conditions/ovarian-cancer), but some are similar to Irritable Bowel Syndrome, such as frequent bloating or discomfort in your tummy.

For males, prostate cancer symptoms often relate to urination, such as needing to pee more urgently, or more often. Further prostate cancer symptoms can be found at: https://www.nhs.uk/conditions/prostate-cancer

MORE INFORMATION AND SUPPORT

If you have questions about your test result, talk to the person who requested your test. The contact details of the person who requested your test can be found in the header at the top of this page. Your GP can also phone or write to the East Anglian Clinical Genetics Service on 01223 216446 to make a referral or ask for further advice to help answer your questions.

For more detailed information about genetic tests and *BRCA*, visit the beginner's guide to *BRCA1* and *BRCA2*.

FOR **YOUR RECORDS**

Technical Information

This page provides technical details from the laboratory about the test.

Patient Details:

Name:	Jane Doe		
Date of birth:	18 March 1970	Sample type:	Blood
Sex:	Female	Sample collected:	10 March 2018
NHS number:	NH00198	Sample received:	12 March 2018

The information on this page provides technical details about the test.

TEST DETAILS FOR ORDERING CLINICIAN

Gene	Zygosity	Inheritance	HGVS Description	Location: GRCh38 (hg38)	*Classification
BRCA1	No clinically pathogenic variant detected				Not applicable
BRCA2	No clinically pathogenic variant detected				Not applicable
PALB2	No clinically pathogenic variant detected				Not applicable

*Variants are classified using the current cancer specific modifications of the ACMG guidelines (http://dx.doi.org/10.1136/jmedgenet-2019-106759 Appendix 1)

• **Cancer risk assessment should take into account the family history of cancer. If this patient has a strong family history of cancer, you may wish to consider a referral to the Clinical Genetics Service for further advice.**

FULL INTERPRETATION

DNA from this patient has been screened for pathogenic variants in the *BRCA1*, *BRCA2* and *PALB2* breast/ovarian cancer susceptibility genes by next generation sequencing and for large scale deletions/duplications of the *BRCA1* & *BRCA2* genes by multiplex ligation-dependent probe amplification (MLPA).

No variants currently known to be pathogenic were detected in *BRCA1*, *BRCA2* or *PALB2* nor large scale deletion/duplication detected in *BRCA1* or *BRCA2*.

These results do not exclude the possibility that this patient may have a pathogenic variant in another cancer susceptibility gene.

TEST METHODOLOGY

Next generation sequencing of all coding sequence and exon/intron boundaries of *BRCA1*, *BRCA2* and *PALB2*. In house sample preparation using Nextera XT library construction protocol and sample processing; Sequencing of long range PCR products on Illumina MiSeq.

We cannot completely exclude the possibility that a SNP under the primer binding site could cause a false negative result in this procedure. However, this test has greater than 99% sensitivity for defined variants.

Multiplex ligation dependent probe amplification (MLPA) analysis of *BRCA1* (using kit P002-D1) and *BRCA2* (using kit P090-B1). Nomenclature according to GenBank accession numbers NM_007294.3 (*BRCA1*), NM_000059.3 (*BRCA2*) and NM_024675.3 (*PALB2*) where +1 is the A of the ATG translation initiation codon.