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

Family history of cancer related to the *BRCA1* gene.

ABOUT THE TEST

Certain alterations to a gene called *BRCA1* increase the risk of developing particular kinds of cancers. We specifically tested for the gene alteration previously identified in your family member (c.4963T>C).

YOUR RESULT: The gene alteration that had been identified in your family member was NOT found in your sample

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 the alteration to the *BRCA1* gene that was previously found in your family. This means that you are not at increased risk of developing cancers associated with this gene alteration.

As you have not inherited this alteration in *BRCA1*, you cannot pass this alteration on to your children.

You do not need to do anything as a result of this test, but if you are concerned you can speak with your GP about what these results mean for your overall level of risk. If you have a family history of cancers that are unrelated to *BRCA1*, you can ask your GP whether a referral to the Clinical Genetics Service is appropriate.

NEXT STEPS

- **You do not need to do anything as a result of this test, but if you have questions about it, speak with your GP.**
- **You should continue to receive any cancer screening recommended by your GP.**

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.

You are advised to report any changes or concerning symptoms to your GP.

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	Familial variant NOT detected		c.4963T>C p.(Ser1655Pro)	17: 43070951	Pathogenic

*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 you are unsure whether the family history can be attributed to this alteration, please contact the Clinical Genetics Service for further professional advice or consider a referral.**

FULL INTERPRETATION

This patient has a family history of *BRCA1* gene-related cancer. The familial pathogenic variant is c.4963T>C in the *BRCA1* breast/ovarian cancer susceptibility gene which is predicted to result in an abnormal *BRCA1* protein, p.(Ser1655Pro). This test was specific for the known familial variant.

Fluorescent sequencing analysis of this patient's DNA has NOT detected the familial *BRCA1* pathogenic variant. Therefore, this patient's risk of developing *BRCA1* related cancers associated with this variant is reduced.

TEST METHODOLOGY

Fluorescent sequencing analysis using Mutation Surveyor of part of the *BRCA1* gene confirmed the presence of the familial pathogenic variant in a DNA sample from an affected family member. We cannot completely exclude the possibility that a SNP under the primer binding site could cause a false negative result in this test. Nomenclature according to GenBank accession numbers NM_007294.3 (*BRCA1*) where +1 is the A of the ATG translation initiation codon.

References

Girardi, F., Barnes, D.R., Barrowdale, D., Frost, D., Brady, A.F., Miller, C., Henderson, A., Donaldson, A., Murray, A., Brewer, C. and Pottinger, C., 2018. Risks of breast or ovarian cancer in BRCA1 or BRCA2 predictive test negatives: findings from the EMBRACE study. Genetics in medicine, 20(12), p.1575.

http://openaccess.sgul.ac.uk/109703/1/gim201844.pdf