

# Laboratory Header

## Report of Molecular Genetic Analysis for Breast/Ovarian Cancer

<i>Patient's name:</i>	**	<i>Referred by:</i>	**
	**	<i>Unit:</i>	**
<i>DOB:</i>	**	<i>Pedigree:</i>	**
<i>*NHS Number:</i>		<i>Clinical Genetics</i>	
<i>Unit no.:</i>	**	<i>contact:</i>	**
<i>Date of request:</i>	**		
<i>Reason for study:</i>	BRCA2 mutation analysis.		
<i>Prior risk:</i>	** has a strong family history of breast cancer and is affected herself. **'s risk of having a BRCA mutation is unspecified. ** has previously been screened for mutations in parts of BRCA1 and BRCA2 and none were identified (see report dated **).		

### Results and Interpretation:

<i>Name</i>	<i>Lab No</i>	<i>Analysis</i>	<i>Result</i>	<i>Lab Ref</i>
**	**	BRCA2 ex 21 sequencing	[c.8951_8952delinsGT; c.8964_8972del] heterozygote	**

Analysis indicates that \*\* is heterozygous for the sequence variant [c.8951\_8952delinsGT; c.8964\_8972del] in exon 21 of BRCA2. We are unable to confirm if this occurs as a single variant or as separate variants on different chromosomes. This variant is not listed on the BIC database<sup>1</sup> and as far as we are aware has not been previously reported. The splice site prediction programme<sup>2</sup> predicts that this variant creates an alternative splice donor site. If alternative splicing does not take place, the mutation causes the protein change V2908G and D2913\_A2915del. This evidence would suggest that this sequence variant is pathogenic.

Analysis of other family members may be useful to determine if the disease is segregating with this change. Predictive testing may then be available on the basis that the sequence change is pathogenic.

### Summary:

**\*\* is heterozygous for the [c.8951\_8952delinsGT; c.8964\_8972del] sequence variant.**

**Reported:** \_\_\_\_\_

**Authorised:** \_\_\_\_\_

**Date:**

*Please Note: All reports depend upon the diagnosis of affected individuals, identification of samples and biological relationships of the individuals being correct.. \*NHS numbers are important patient identifiers and should be provided with every sample.*

*Notes: Analysis of exon 21 was with bi-directional sequencing. BRCA2 Accession number U43746. <sup>1</sup> Breast cancer information core database registers BRCA1 and BRCA2 mutations (<http://research.nhgri.nih.gov/bic/>?). <sup>2</sup>The Splice site prediction programme is available online at [http://www.fruitfly.org/seq\\_tools/splice.html](http://www.fruitfly.org/seq_tools/splice.html)*