Patient information sheet: **for those who receive a variant of uncertain significance genetic test result (R208 Panel)**

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What does my genetic testing result mean?

Cancer is a very common condition, with 1 in 2 people diagnosed in their lifetime. Around 5-10% of cases have an underlying inherited genetic cause, whilst the other 90-95% are thought to be due to a combination of random chance, age, lifestyle, environmental factors, and genetic risk factors that we cannot currently test for.

Our genetic code can be thought of as our 'inherited instructions' which tell our body how to grow, develop and function. There are many genetic differences between individuals, most of which are completely harmless and contribute to normal human variation.

Having a 'variant of uncertain significance' (VUS) means that the laboratory have found a change in one of your genes known to be associated with cancer, but do not have enough scientific evidence at present to confirm if it is disease causing or if it is part of normal human variation. For this reason, the clinical significance of the result is unclear, and we are unable to determine whether your cancer diagnosis was caused by an inherited gene change, or by other factors. This result does not rule out, or confirm an underlying genetic cause.

Do I need any further genetic testing?

There would be no clinical benefit of further genetic testing at this time, as the test you have already had analysed the genes that we currently know are associated with breast and ovarian cancer. However, there are continuous efforts being made to better understand the genetics of inherited cancer. We are hopeful that with time we will be able to confirm whether your VUS is associated with increased cancer risks, or if it is part of normal, harmless human variation.

You will be offered an appointment with clinical genetics, who will aim to see you within 9 weeks to discuss your result in greater detail. This is also a good opportunity to address any questions and concerns you may have. Although this result has not provided a definitive genetic explanation for your cancer, sometimes in cases where there is a considerable family history of cancer, increased screening recommendations may be given to yourself and certain relatives to reflect this.

What does this result mean for my blood relatives?

If we are uncertain whether a gene change found is the cause of the cancers in your family, we will not be able to offer genetic testing for this to unaffected relatives. This is because like yourself, we would not know what the result means for them. We may suggest genetic testing for other family members who have developed certain cancers. The results of this may help us to better understand your result. Prior to your appointment with clinical genetics, you may be asked about your family history to help with our assessment.

Contact details for the clinical genetics service

In the meantime, if you have any **urgent** queries, please do not hesitate to contact us.

Email: genetic.medicine@belfasttrust.hscni.net

Tel: 028 9504 8022