

## Information for those who receive a PTEN positive genetic test result (R208 Panel)

### What does my genetic testing result mean?

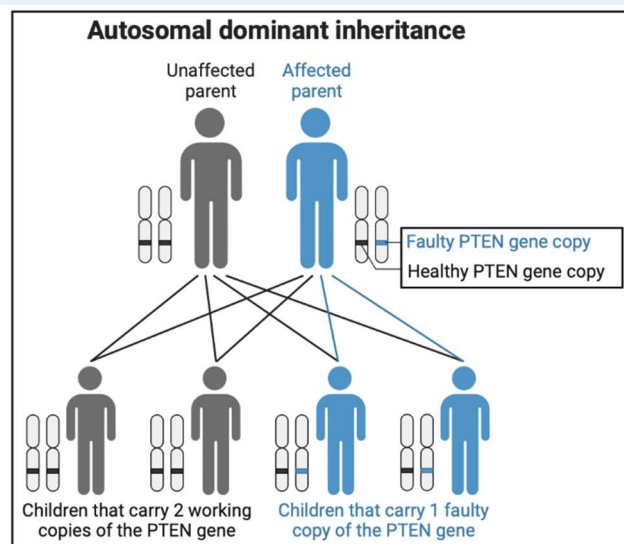
Our genes can be thought of as our 'inherited set of instructions'. They tell our body how to grow, develop and function. The PTEN positive result you have received means that a pathogenic (cancer-causing) change has been identified in 1 of your 2 PTEN gene copies that clearly affects how it works.

Individuals who carry a PTEN pathogenic gene change have an increased lifetime risk of developing several cancers, including breast cancer. Therefore, this result provides a genetic explanation for your cancer diagnosis, and may now be used by your oncology team to guide ongoing treatment and management decisions. Your lifetime risks of developing further primary cancers is greater than the general population due to this result. You may be eligible for additional screening to detect early signs of cancer, and/or risk-reducing surgical options.

### What does this result mean for my blood relatives?

We receive half of our genetic information from each parent. If one parent carries an altered PTEN gene copy, there is a 50% chance that this will be passed on to each child (regardless of their gender), and a 50% chance that the healthy copy is passed on. This is known as **autosomal dominant inheritance**.

Because we share some of our genetic information with our blood relatives, there is a chance they may also carry the same gene change and therefore also have increased risks of developing certain cancers. For this reason, they may be eligible for genetic testing to determine whether they have also inherited it. This will be coordinated through clinical genetics and will be discussed at your follow-up appointment.



### What are the next steps?

You will soon be offered an appointment with clinical genetics. During this, the genetics health professional will discuss in greater detail the specific risks associated with the gene change that has been identified in you, as well as screening and management options, and support available to yourself and your relatives. This is also a good opportunity to discuss any questions and concerns you may have. Prior to your appointment, you will be asked to provide information about your family history:

- To allow tailored risk information to be provided (associated genetic risks vary significantly when family history and individual circumstances are considered)
- To allow us to identify all of those at-risk who should be offered testing (note that we do not contact your family members)

The genetics team can also provide you with information and support to help share this information with the relevant relatives in a way that you feel most comfortable with.

### 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](mailto:genetic.medicine@belfasttrust.hscni.net)

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