3. **A significant genetic change is found at a higher level:** If a genetic change if found at a level suggesting it might be inherited, you will be offered germline testing. Germline testing, usually done using a blood sample, confirms whether the change is inherited. If confirmed, this may indicate a cancer predisposition syndrome. This information can help identify cancer risks for your relatives and guide screening or prevention strategies for your family.

WHAT IF THE RESULTS ARE NOT RELATED TO MY CURRENT CANCER?

Occasionally, molecular profiling may uncover an inherited genetic change that is unrelated to your current cancer. Even if this result does not affect your treatment, it may be important for your long-term health and may have implications for your family members. Your healthcare team will provide support and guidance to help you and your family understand these findings.

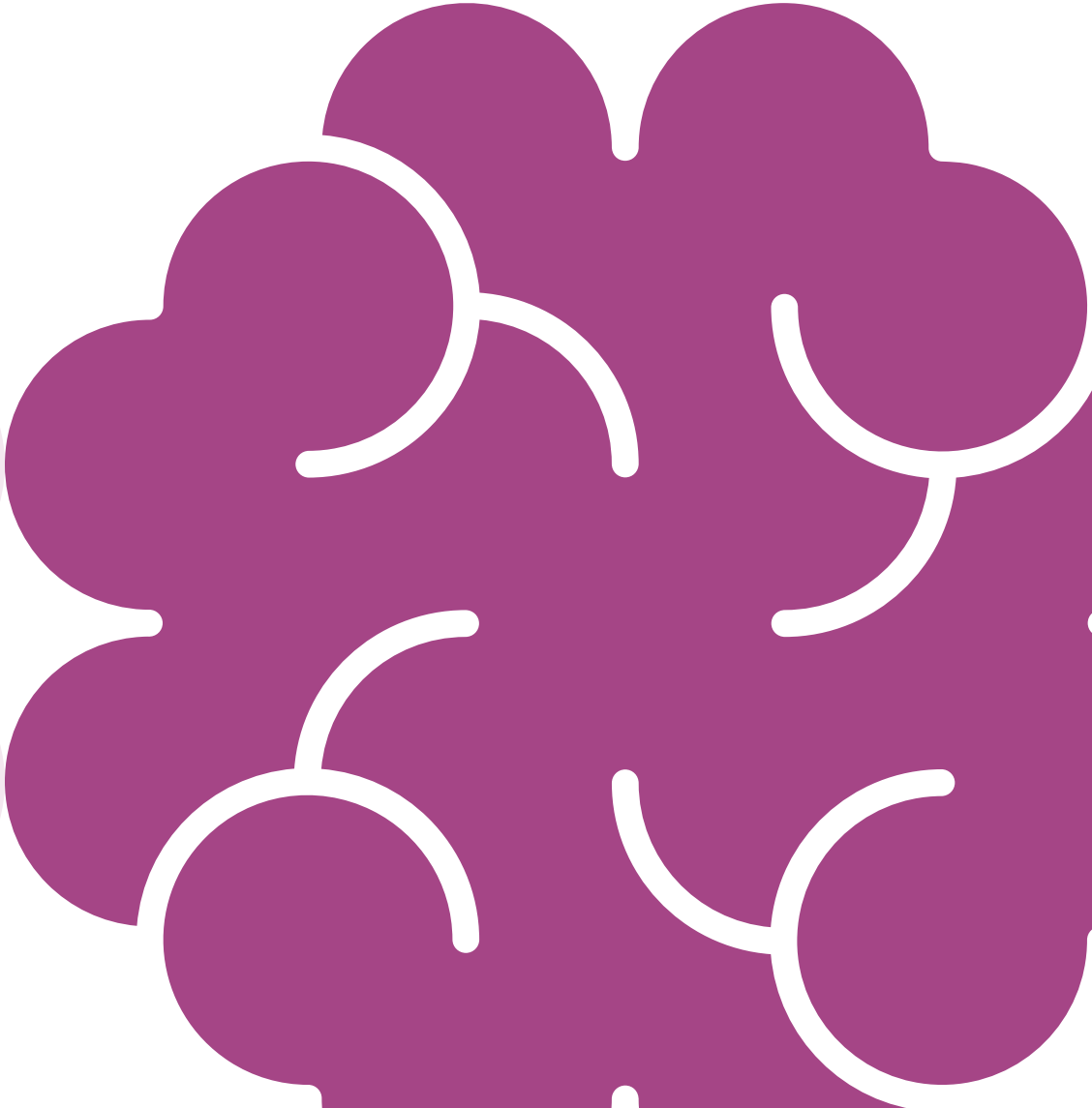
USEFUL LINKS

- https://belfasttrust.hscni.net/service/laboratory-services/clinical-genetics/patient-support/
- https://belfasttrust.hscni.net/service/laboratory-services/clinical-genetics/common-referrals-and-information-for-patients-professionals/

BT24-3292

HSC Belfast Health and Social Care Trust
caring supporting improving together

TUMOUR PROFILING FOR CANCER PATIENTS



TUMOUR PROFILING FOR CANCER PATIENTS

This leaflet provides information for patients and families about molecular profiling, a type of tumour testing. This testing examines the DNA in cancer cells to identify genetic changes (variants) that can guide treatment decisions and provide valuable information for you and your family.

WHY IS MOLECULAR PROFILING IMPORTANT?

Cancer develops because of genetic changes in cells. Many of these changes are acquired during a person's lifetime (called somatic changes) and can affect how the cancer grows and responds to treatment. Molecular profiling helps your doctors understand your cancer better and may guide the use of targeted therapies, precision medicines, or determine your eligibility for clinical trials.

In some cases, the testing may also reveal genetic changes that were inherited (called germline changes). These findings may indicate an underlying cancer predisposition syndrome, which can be important not only for you but also for your relatives, as they might share the same inherited risk.

HOW IS MOLECULAR PROFILING DONE?

Molecular profiling is carried out on tumour tissue obtained during surgery or a biopsy as part of routine pathology testing. No additional procedures are needed for this analysis.

ACQUIRED CHANGES		INHERITED CHANGES
The genetic change is **not** present from birth	**EMBRYO** 	The genetic change **is** present from birth
Genetic changes are present in the tumour but not in other cells	**GENETIC CHANGE** 	The genetic change is present in every cell
Not passed on to children	**FAMILY** 	Can be passed on to children and may affect other family members

WHAT TYPE OF GENETIC CHANGES MIGHT BE IDENTIFIED?

Tumour testing can detect two main types of genetic changes in cancer cells:

1. **Somatic changes:** These are acquired changes that occur only in the tumour cells and are not inherited. They may help guide treatment decisions.
2. **Germline changes:** These are inherited changes present in all cells of the body. They may indicate an increased risk of cancer for you and your family members.

WHICH GENES WILL BE ANALYSED IN MY TUMOUR?

Genes are like "coded instructions" that tell cells how to grow and function. During molecular profiling, we examine specific genes that are linked to an increased risk of cancer and for which screening or risk reducing measures are available.

The table below lists the genes that are routinely analysed in **all tumour types.** Some of these genes may not be directly related to your specific type of cancer or your family history of cancer. Additionally, other genes associated with the particular type of cancer you have may also be analysed during testing.

GENE	CANCER TYPES MOST ASSOCIATED
BRCA1 and PALB2	Breast and Ovarian
BRCA2	Breast, Ovarian and Prostate
MLH1, MSH2, MSH6	Colorectal and Endometrial (womb)

WHAT ARE THE POSSIBLE RESULTS OF MOLECULAR PROFILING?

1. **No significant genetic changes are found:** Even if no significant changes are identified, this information can still help your doctors make treatment decisions. If there is a strong family history of cancer, you may be referred to a clinical genetics service for further evaluation.
2. **A significant genetic change is found at a low level (somatic):** If the change is present only in the tumour and at a low level, it may influence your treatment options. This type of change is unlikely to be inherited, so additional testing for you or your family members is not typically needed.