

Genetic testing and targeted therapy

When diagnosed with lung cancer your doctor will do a variety of diagnostic tests to understand the biology of your tumour and to guide your treatment. One of these tests is genetic testing that looks at the genes that are causing your cancer. Genetic testing provides a more comprehensive picture of your lung cancer and helps your oncologist develop and guide the most appropriate treatment plan for you. Genetic testing is carried out if you have non-small cell lung cancer.

What is genetic testing?

Genetic testing can have many names, including mutation testing, genomic testing, genetic panel testing, biomarker testing, NGS (next generation sequencing) or molecular testing. Essentially, all these terms refer to the same process. Cells from a biopsy of your lung cancer are examined under a microscope and have a range of tests carried out upon them. These tests determine if there is a mutation present by the way the cells look, behave and react to the tests.

Somatic vs germline

Gene mutations found in lung cancer are almost always somatic. Somatic mutations, which are acquired during an individual's lifetime, are specific to tumour cells while germline mutations are inherited and present in all cells of the body. Therefore, the genes mentioned in this document will not be passed onto children. As an example, BRCA gene found in breast cancer can be passed onto children.

Why is genetic testing important?

A high percentage of lung cancers carry some form of mutation. Mutations can occur regardless of smoking status but is often higher in young people who have never smoked. A mutation can possibly occur in cancer due to multiple factors which may be environmental or genetic, or the exact cause may not be known. The process of genetic testing can take several weeks to fully complete. Taking the time to get the results is important. Testing for mutations can help your oncologist to better understand the type of cancer you have, and to identify which treatment may be most effective and appropriate for you. Biomarkers, such as PD-L1 will also be determined through testing, which can also help to determine treatment. Common mutations associated with lung cancer include:

- EGFR - epidermal growth factor receptor
- ALK - anaplastic lymphoma kinase
- ROS1 - Reactive Oxygen Species 1
- KRAS - Kirsten rat sarcoma viral oncogene homolog

Research is continually being undertaken in lung cancer to learn more about the role mutations play. Check with your oncologist if your biopsy sample has had genetic testing and ask for the results to be explained to you.

What happens if a mutation is found?

The presence of a mutation gives your oncologist information about your cancer, and possibly how it will behave. Depending on what type of mutation is found, there may be a specific treatment, called targeted therapy suitable for you.

What is targeted therapy?

As it sounds, targeted therapy is a treatment which precisely targets a particular type of cancer cell or mutation.

How does targeted therapy work?

Targeted therapy is able to attack cancer cells, while minimising harm to healthy cells. It is only active, or effective, on the cell or genetic material that it has been designed to treat/target. Targeted therapy may affect the cancer cells it targets through different methods such as:

- blocking or turning off signals that tell cancer cells to grow and divide
- preventing the cells from living longer than normal
- destroying cancer cells.

In lung cancer, targeted therapy may come in the form of a tablet or capsule. Some may be taken daily for many months or years while they are effective in treating your lung cancer. However, targeted therapies do have some limitations. When taking targeted treatment for metastatic disease, we know at some point the treatment will stop working. The targeted cancer cell can mutate again or become resistant to treatment – meaning it may work as expected when you first start on the treatment and then it may stop working. It is also possible that side effects from the treatment are not able to be tolerated. If this happens often a “drug holiday” is taken for you to recover. The treatment may then start again at the same or lower dose to see how you tolerate it. If this is still not tolerated, then another type of targeted therapy may be tried or another treatment option explored, for example, immunotherapy or chemotherapy. Many factors determine which treatment is the best for you. Your oncologist will discuss the risks and benefits involved in all your options.

Are there side effects with targeted therapy?

Whilst specifically targeting cancer cells, targeted therapies do have the potential to cause side effects, which can vary in type and severity. Common side effects can include skin reactions, changes in the way your bowel works, fatigue, joint aches and pains. They are usually able to be better managed when you report them early to your treating healthcare team. Ask your treating healthcare team about what possible side effects you should be on the lookout for and what to do in the event you experience them.

What if there is no genetic mutation or no targeted therapy?

It is possible that your lung cancer will not carry a mutation, or it may have one for which there is no specific targeted therapy currently available. If this is the case and another treatment is required, your oncologist may offer chemotherapy, immunotherapy or a combination of these two treatments together. At all times, your oncologist will consider your entire health picture and recommend what is the most appropriate treatment for you.



Looking ahead

Greater understanding of mutations in cancer cells has changed the way cancer is treated, resulting in the development of many therapies that target specific cancer cells and mutations. Some mutations found in lung cancer are also identified in other types of cancers. This has helped in the ongoing improvement and development of treatments and outcomes through broader cancer research. It is possible that new targeted treatment options may become available in the future, after your initial diagnosis and treatment. Speak with your oncologist about all your options, including availability of clinical trial for treatment.



**Scan to connect with a
Lung Cancer Specialist Nurse
or call 1800 654 301.**

Note to reader: This information is intended as a general guide only and is not intended or implied to be a substitute for professional medical advice or treatment. While all care is taken to ensure accuracy at the time of publication, always consult with your doctor about matters that affect your health.