Genomic (Biomarker) Testing for Lung Cancer

What are genes and what role do they play in lung cancer?

A gene is part of your DNA, found at the center of every cell in your body. Gene damage is called a mutation. The damage be from an outside influence (environmental) or inherent (you were born with it). The gene changes (alterations) that we find in lung cancer are not inherent. You did not get them from your parents and you should not pass them down to your children.

This damage can lead to a gene not working right and the cell growing out of control. These cells can then become a tumor. When the genes of lung cells are mutated, it can lead to lung cancer.

Certain mutated genes (also called biomarkers) have been linked directly to lung cancer. Recent advances in genomic testing (or molecular testing) have changed how some cancers are treated. Medications called targeted therapies target mutated genes found in some cases of lung cancer.

How do they test my genes?

If your healthcare provider thinks you may have lung cancer, you will likely have a biopsy done. During a biopsy, a pulmonologist or surgeon will take a piece of the tumor for testing in a lab. A pathologist looks at the sample of lung cells under a microscope.

The pathologist will classify the cells by histology (small cell lung cancer or non-small lung cancer). This biopsy sample may also be used for biomarker testing. Lung cancers that are classified as non-small cell lung cancer (NSCLC) with a subtype called adenocarcinoma, will undergo this biomarker testing. Providers do not routinely test other types (small cell, squamous cell carcinoma) because they are highly unlikely to have one of these targetable mutations.

Biomarker testing can be done early in your treatment plan (at diagnosis), or later on in your treatment. Some larger cancer center laboratories can do this testing, but more often the sample is sent to a commercial laboratory for testing. There is also a blood test that can detect these genomic mutations, and often, oncologists may send both the tumor tissue biopsy AND a blood test to look for these mutations.

If you had lung cancer in the past and it comes back (recurrence), your provider might recommend testing your tumor for biomarkers. This can be done even if you have already had chemotherapy and/or radiation in the past.

What is included in the biomarker pathology report?

Once your cells have been tested in the lab after a biopsy, the pathologist will write a pathology report. This report will have the specifics of your cancer (type, size, lymph nodes). The results of any biomarker testing that is done on your cancer cells might be in the initial pathology report, or they might be in a separate report later.

Your biomarker report may include:

- A list of biomarkers that were tested on your tumor sample, and if that biomarker was found or not.
- Recommended medication or medications that may work against that biomarker.
- The contact information for the pathologist who wrote your report.

It can take up to 4 weeks to get your biomarker testing results. Your provider will receive a copy of these reports. It is a good idea to ask for a copy for your records.
How does knowing my biomarkers help?

There may be medications used to treat tumors with these specific biomarkers. Biomarkers can also help predict the chance that cancer will come back after surgery. This information may be used to decide if further treatment is needed.

Mutated genes linked to lung cancer include:

- EGFR (epidermal growth factor receptor).
- ALK (anaplastic lymphoma kinase).
- ROS1 (ROS proto-oncogene 1, receptor tyrosine kinase).
- BRAF (B-RAF proto-oncogene, serine/threonine kinase).
- NTRK (Neurotrophic receptor tyrosine kinase 1).
- KRAS (Kirsten rat sarcoma viral oncogene homolog).
- HER2 (human epidermal growth factor receptor 2).
- RET (RET proto-oncogene).
- MET (MET proto-oncogene).
- FGFR (Fibroblast growth factor receptor).
- PIK3CA (Phosphatidylinositol-4,5-bisphosphate 3-kinase catalytic subunit alpha).

If there is not enough tissue from the biopsy for the pathologist to test for all of these biomarkers, or if it is not safe for you to have another biopsy done, your provider may take a blood sample. In advanced lung cancer, the DNA from dead cancer cells may be found in the blood. This blood test (sometimes called liquid biopsy) can test for specific biomarkers but is not always as reliable as testing on a tissue biopsy.

What treatments are available based on my biomarker testing?

Some cancer treatments work against certain mutated genes. The mutated genes that have FDA-approved medications are:

- EGFR.
- ALK.
- ROS1.
- BRAF.
- NTRK.
- KRAS.
- RET.
- MET.

Biomarker testing can tell your provider a lot about your cancer. Even if there is not an FDA-approved treatment for a mutation, there is the possibility of future medications to target that gene. Clinical trials are important in furthering our knowledge of this disease. It is through clinical trials that we know what we do today about biomarkers, and many exciting new therapies are currently being tested. Talk to your healthcare provider about participating in clinical trials in your area. You can also explore currently open clinical trials using the OncoLink Clinical Trials Matching Service.