An Illustrated Guide to Blood Biopsies at the Broad Institute of MIT and Harvard

Researchers have traditionally studied patients' cancers by surgically sampling a piece of the tumor, providing clues to effective treatments. Unfortunately, this invasive procedure only provides a snapshot at one point in time, even though tumors evolve and mutate in response to therapy. Also, in some patients, many tumors exist and an individual surgical biopsy may not reflect the genetics of all of them equally.

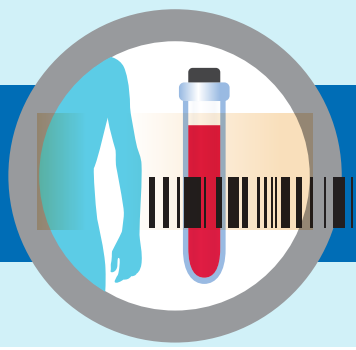
Today, scientists around the world are developing new techniques, called blood biopsies, that may allow researchers to understand tumor genomics over time, all from simple blood tests. Tumors often shed bits of DNA into the bloodstream. By identifying the genomic sequences encoded in these DNA fragments, researchers can gain an accurate look at the latest genetic code of a patient's cancer.

Some blood biopsy technologies focus on profiling a select set of tumor mutations. At the Broad Institute, scientists and collaborators have developed a way to use blood biopsies to sequence the entire genome of a patient's cancer in a scalable and cost-effective manner.

The Broad Institute is using blood biopsies to detect and discover the mutations that confer sensitivity and resistance to therapy. This information will help inform better therapeutic strategies both now and into the future.

Here is how we perform this technique:

1

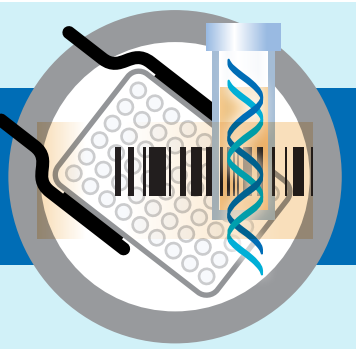


The cancer patient's blood is drawn at a hospital and anonymized with a barcode that is entered into a computer system for tracking.

2

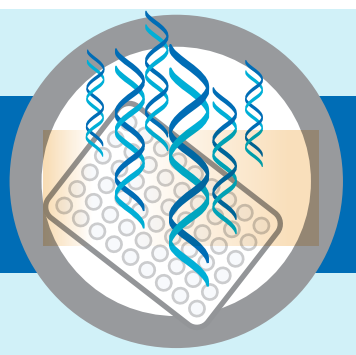
The vials of blood arrive at the Broad Institute, where scientists separate the blood plasma (containing the DNA of interest) from the cells by spinning the blood in a centrifuge. The plasma is identified with a second barcode, tracked in the same system.

3



The plasma is fed into a robotic system that will automatically and consistently extract the DNA of interest.The DNA is then stored in another barcoded tube.

4



The DNA is quantified and prepared for sequencing at the Broad Institute's Genomics Platform.

5

A few rounds of sequencing provides a high-level view of how much tumor DNA there is in the blood — a potential indicator of the seriousness of the disease. By finding the mutations unique to that tumor, doctors could someday choose the most appropriate treatment.

BROAD INSTITUTE

The Broad Institute has processed approximately 3,000 samples in this way since 2015. Researchers are using the information to help understand how tumors mutate and grow resistant to treatment.