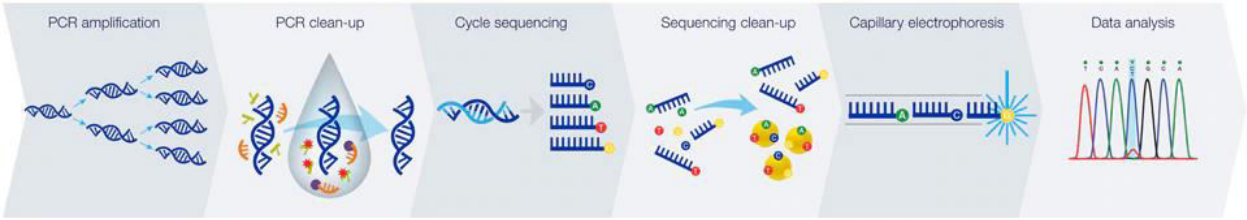
Applied Biosystems SeqStudio Genetic Analyzer

Applied Biosystems SeqStudio Genetic Analyzer is a low-throughput, easy-to-use, and convenient bench top system that delivers gold-standard Sanger sequencing technology and fragment analysis.

Workflow:



DNA sequencing is a laboratory method used to determine the order of the bases within the DNA. Differences in the sequence of these 3 billion base pairs in the human genome lead to each person's unique genetic makeup. In medicine, DNA sequencing is used for a range of purposes, including diagnosis and treatment of diseases. In general, sequencing allows healthcare practitioners to determine if a gene or the region that regulates a gene contains changes, called variants or mutations that are linked to a disorder.

When considering or undergoing genetic testing, it is important to seek help in interpreting these results from a genetics expert such as a medical geneticist or genetic counselor to better understand the test results, implications of the results, and any potential risk of having or passing a genetic condition on to your children.

How is DNA sequencing used?

There are a wide variety of medical applications for DNA sequencing. These techniques can be used to test one gene or several genes to help diagnose medical conditions. Some examples include:

- **Targeted sequencing**-sequencing of select variants or areas within a gene's exons (the segments of DNA that code for proteins). When there is a known effect of certain types of changes to one or more genes, it may help guide medical care to only test for these known changes. One example is testing a tissue biopsy sample from a melanoma to determine whether or not the cells have a mutation (disease-causing variant) in the *BRAF* gene. A mutation in *BRAF* is found in more than 50% of melanomas, and people with advanced melanoma that have *BRAF* mutations may respond to drugs that target these mutations, a treatment referred to as targeted cancer therapy. Targeted drugs work differently than standard chemotherapy and may have fewer side effects.
- **Mutations** (disease-causing variants) in this gene cause Marfan syndrome, a disorder that affects the connective tissue that makes up many parts of the body, including bones, muscles, ligament, blood vessels, and heart valves.
- **Multi-gene panel sequencing**-sequencing parts or all of several genes to detect mutations (disease-causing variants) that can cause a genetic disorder. An example of this is a panel to test for mutations in the *MLH1, MSH2, MSH6, PMS2* and *EPCAM* genes. Mutations in these genes can cause Lynch syndrome, an inherited disorder that increases the risk of many types of cancer, especially colon cancer and endometrial cancer.
- ***BCR-ABL1* mutations**- impart resistance to tyrosine kinase inhibitors currently available for treatment of chronic myelogenous leukaemia.

SeqStudio Genetic Analyzer advances, a broader number of applications for these techniques will continue to make their way into clinical and laboratory testing settings.