How nucleoLIS Ē.finity can Support Whole Genome Sequencing from Start to Finish

By Austin McGarrett

Over the past several years, whole genome sequencing has expanded beyond its origins as a research tool. With whole genome sequencing, an individual's DNA strands can now be analyzed for any evidence of genetic disorders. This, therefore, enhances the diagnostic capabilities of healthcare providers, helping them make a more accurate diagnosis and treatment recommendations in the management of their patients.

Whole genome sequencing, however, is an intricate and complex process that must be performed according to a predefined workflow. Failure to adhere strictly to the workflow may lead to inaccurate results which, if not rectified, may negatively impact patient care. Having a laboratory information system (LIS) uniquely designed to support the workflow of whole genome sequencing can help in several ways to maintain the integrity of the process and ensure the validity of the results.

At Psychē, we have developed nucleoLIS Ē.finity to support and maintain the workflow for whole genome sequencing as well as cytogenetic testing. Using our software, you can rest assured that the genome sequence workflow was strictly followed and that the final results are valid. Discussed below are the various steps involved in whole genome sequencing and how our nucleoLIS Ē.finity package can help assist each of these steps.

1) Test request and specimen collection

An order request must be placed by a submitting physician before whole genome sequencing can be initiated. The requesting provider must provide the patient's demographics, a reason for the request, type of sequencing analysis needed, preferred specimen type, and other relevant details. The process cannot begin until all the necessary information has been provided.

Our nucleoLIS Ē.finity application has an order entry template where all the prerequisite information can be entered. It also provides direction regarding the collection as well as storage requirements of the specimen needed for the process using special alerts for users.

2) DNA isolation and library preparation

Once the specimen has been collected - blood, bone marrow, tissue, or formalin-fixed paraffin-embedded (FFPE) tissue – the next step is to extract and isolate the genomic DNA which is then quantified. Following that, the DNA extract is then fragmented into small pieces using specialized equipment; chemicals are then added which tag the ends of the DNA fragments.
Using nucleoLIS Ē.finity, you can keep tabs on the number of DNA fragments, average fragment length, and number of polymerase chain reaction (PCR) cycles. You can also do quality control during this step by documenting the kit version used as well as the reagent lot numbers.

3) Cluster generation and sequencing

With cluster generation, the DNA fragments are bound to flow cells where they are copied thousands of times, a process that is known as amplification. Once amplification has been completed, the DNA is then sequenced; the DNA nucleotides (A, C, T, G) are matched with the DNA fragments on the flow cells. The A nucleotide matches with its T counterpart while the G nucleotide pairs with the C nucleotide. Matching pairs trigger signals that inform the instrument which

nucleotides matched. During this step in the process, our nucleoLIS Ē.finity application can be used for flow cell identification, protocol information, and quality control.

4) DNA analysis

After DNA sequencing, the next step is to compare the DNA with the reference genome, a human genome recognized worldwide as the genome standard. Genomic differences, as well as variants, are looked for and identified; several base algorithms are used during this process. nucleoLIS Ē.finity can run the necessary metrics needed for the DNA analysis; it can also identify which algorithms to be used as well as the algorithm parameters.

5) Result reporting

This is the final step of the whole genome sequencing process. The work performed, the metrics used, and the test results are documented and prepared in a format that can be readily understood by the requesting provider as well as other relevant parties. Our nucleoLIS Ē.finity application has report templates that allow for easy and standardized documentation of all the necessary result information.

For more information about nucleoLIS Ē.finity, contact us today.