alamya health

Research Long-Read Genome Sequencing

Alamya Health, in conjunction with our laboratory partners, offers long-read (LR) research grade whole genome sequencing for clinical research participants. A research report will be provided along with raw data files (with some limitations). Turnaround times for test results are dependent on the specific requirements of the research project. *This is not a CLIA validated test and is not a stand-alone clinical diagnostic test.*

Using the Oxford Nanopore technology, the Generation Beyond test includes long-read whole genome sequencing, mitochondrial genome sequencing, DNA methylation profiling, and long-read transcriptome sequencing (cDNA).

Alamya Health reports leverage multiple data streams to provide meaningful results to support your project.

Sequencing

The Generation Beyond test detects single nucleotide variants (SNVs), small insertions and deletions (indels) in regions of the genome inaccessible by short-read technologies, short tandem repeat expansions, structural variants (insertions, inversions, and translocations), and copy number variants (CNVs). Basecalling, mapping, variant calling, and phasing are performed using the EPI2ME pipeline developed by Oxford Nanopore Technologies (https://github.com/epi2me-labs/wf-human-variation).

Transcriptome sequencing

The transcriptome sequencing uses cDNA derived from whole blood RNA depleted for hemoglobin mRNA and ribosomal RNAs. Canonical and modified base calling is performed with Dorado. Mapping, transcript assembly, and gene- and transcript-level quantification is performed using the EPI2ME pipeline developed by Oxford Nanopore Technologies (https://github.com/epi2me-labs/wf-transcriptomes).

Ordering and costs

For testing requests, please contact: sales@alamyahealth.com or call (628) 252-5243.

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