NEXT ERA

Cutting edge genome and epigenome profiling

Alamya Health's Next Era test thoroughly interrogates genomic and epigenomic causes of disease. By integrating short-read (SR) and long-read (LR) whole genome sequencing, DNA methylation analysis, SR transcriptome, and phenome, Next Era maximizes the diagnostic potential.

With a diagnostic rate 20-30% greater than standard of care testing, this comprehensive and in-depth diagnostic testing is for those who have had uninformative or inconclusive prior testing, have complex phenotypes, or wish to avoid prolonged sequential testing.

The turnaround time is 6-12 weeks.

Sequencing

Short-read genome sequencing: The Next Era SR genome is sequenced using MGI or Illumina technology in a CAP/CLIA certified laboratory. This is an established technology that provides highly accurate assessment of some genomic variation in regions of the genome accessible to this methodology.

Long-read genome sequencing: The Next Era LR genome is sequenced using Oxford Nanopore technology. This is a newer technology that provides assessment of additional types of genomic variation and in regions of the genome inaccessible to SR genome sequencing.

DNA methylation profiling: Using Oxford Nanopore technology, DNA methylation profiles detect disorders of DNA methylation, guide interpretation of variants in genes that encode epigenetic regulators, assess imprinted regions, and delineate gene promoter methylation status.

Short-read transcriptome sequencing: RNA-seq, also known as transcriptome sequencing, improves the diagnostic rate depending on the clinical phenotype and tissue sample. RNA-seq aids in prioritizing and resolving variants of unknown significance (VUS) and characterizing variants with the potential to alter gene expression and splicing.

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Reporting

Diagnostic assessment for monogenic or oligogenic predispositions: Alamya Health integrates multiple data streams to provide meaningful results to support healthcare providers and patients in achieving genomic diagnoses. We integrate the sequence and DNA methylation data with the phenome (i.e., patient phenotype [clinical findings], familial inheritance patterns) and knowledge mined from the literature and curated databases to identify probable genetic causes of disease.

Pharmacogenomic (PGx) variants: Genomic variants for which there are prescribing guidelines. The pharmacogenomic variants reported are those that impact commonly prescribed medications.

Comprehensive risk assessment for monogenic (single-gene) predispositions - based on client consent:

Medically actionable variants: Genomic variants for which there are consensus guidelines for surveillance and management. The monogenic predispositions reported are the medically actionable disorders defined by the American College of Medical Genetics and Genomics. This is presently 81 genes.

Carrier status: Genomic variants for which there are risks to offspring if the parents are carriers. The 1,300 genes reported follow the principles promulgated by MacKenzie's Mission (Australian carrier screening program). Carrier testing is primarily used to guide reproductive planning. However, variants detected in a small subset of genes may be associated with health implications for identified carriers and these will be included in the report along with management resources.

Repeat disorders: Testing for a number of disorders associated with expansion or contraction of repeated sequences of DNA. These disorders often involve neurological impairment, which can be progressive. For a number of these disorders, for example Huntington disease, there are not effective treatments.

Polygenic risk score: This testing provides a relative risk for developing a complex health problem by assessing many genetic variants across an individual's genome along with baseline health factors (e.g., the risk for developing cardiovascular disease would be higher for someone in their sixties than for someone in their twenties). Alamya Health's polygenic risk score testing assesses the risk for cardiovascular disease in women and men and for sporadic breast cancer in women. These test results can help to guide health and lifestyle strategies for those with higher risk scores.

Payment information

This test is only available via private pay. Some insurance plans may cover a portion of the test (i.e., SR genome sequencing) and you may wish to review your policy for coverage specifics.

Visit our website at alamyahealth.com for more information on our Next Era test.