Reanalysis and Interpretation for Whole Genome Sequencing (Internal Data)

Test Code	D0502
Test Summary	This test provides reanalysis and interpretation of whole genome data previously sequenced at PerkinElmer Genomics
Turn-Around-Time (TAT)*	2 - 4 weeks
Acceptable Sample Types	
Acceptable Billing Types	Self (patient) Payment Institutional Billing

Test Description

This test involves reanalysis and interpretation of previously generated data from a PerkinElmer Genomics whole genome sequencing test. All variants identified will be analyzed according to American College of Medical Genetics and Genomics (ACMG) guidelines. In addition to SNVs, our WGS analysis will reliably detect CNVs of 3 exons or greater as well as large-scale CNVs such as microdeletions and other gene/chromosomal-level events. CNVs of 1-2 exons may be detected and reported with the recommendation for follow-up testing. Mitochondrial DNA analysis is included. It is recommended that updated clinical notes and phenotypes are provided to aid in the reanalysis.

Test Methods and Limitations

Alignment to the human reference genome (hg19) is performed and annotated variants are identified in the targeted region. Variants are called at a minimum coverage of 8X and an alternate allele frequency of 20% or higher. Single nucleotide variants (SNVs) meeting internal quality assessment guidelines are confirmed by Sanger sequence analysis for records after results are reported. Indels and SNVs may be confirmed by Sanger sequence analysis before reporting at director discretion. This assay cannot detect variants in areas containing large numbers of tandem repeats. Mitochondrial DNA is analyzed using the same pipeline. Copy number variation (CNV) analysis is designed to detect deletions and duplications of three exons or more; in some instances, due to the size of the exons or other factors, not all CNVs may be analyzed. Only CNVs related to phenotype are reported. This assay is not designed to detect mosaicism; possible cases of mosaicism may be investigated at the discretion of the laboratory director. Primary data analysis is performed using Illumina DRAGEN Bio-IT Platform v.2.03. Secondary and tertiary data analysis is performed using PerkinElmer's internal ODIN v.1.01 software for SNVs and Biodiscovery's NxClinical v.4.3 or Illumina DRAGEN Bio-IT Platform v.2.03 for CNV and absence of heterozygosity (AOH).

Detailed Sample Requirements