



Genomic Unity[®] Cardiac Channelopathies Analysis

Overview

Genomic Unity[®] Cardiac Channelopathies Analysis is an appropriate test for patients with symptoms of cardiac channelopathies.

Method

PCR free whole genome sequencing (WGS) is used as the underlying NGS technology. Its consistent read depth across >98% of the genome enables identification of multiple variant types from a single patient sample.

Proprietary algorithms optimized for each variant type are used to perform discrete in-silico analyses of the data which are brought together for collective interpretation, providing a complete genetic picture.

Rigorously trained variant scientists interpret all variant types in the context of the patient's phenotype and generate a unified clinical report.

Included analyses

- ✓ Sequencing analysis of cardiac channelopathy genes
- ✓ Del/dup analysis of cardiac channelopathy genes

Genes analyzed

ANK2, CASQ2, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, RYR2, SCN5A

Test performance

Highly uniform sequencing depth

- 30X mean mappable coverage
- >98% of nucleotides covered at ≥8x
- >99% of HGMD and ClinVar annotated variants covered at ≥8x

Highly sensitive detection of SNVs and indels up to 50 bp

- 99.7% sensitivity
- 99.6% positive predictive value

Highly sensitive detection of structural variants

- 96% clinical sensitivity
- In most cases, the exact genomic coordinates (the breakpoints) of the structural variant can be determined

Accepted sample types

- Blood - optimally 5ml
- gDNA - 5µg minimum
- Saliva

Turnaround time

6-8 weeks after sample receipt

Reflex option

In the case that Genomic Unity[®] Ataxia Repeat Expansion Analysis does not identify causal variants, the option is given to reflex up to Genomic Unity[®] Whole Genome Analysis which looks more broadly for causal variants across all genes.

The reflex option is offered for a nominal patient pay price when not covered by the patient's insurance.