variantyx

Genomic Unity® Ataxia Analysis

Overview

Ataxias are a group of neurological conditions most often related to degeneration of the cerebellum, the area of the brain responsible for coordinating movement.

With its single method approach to detecting and analyzing tandem repeat expansions, structural variants and small sequence changes, Genomic Unity® Ataxia Analysis is an effective test for diagnosis of ataxias. It provides comprehensive coverage of ataxia-related genes in addition to the ataxia repeat expansion loci that are the focus of Genomic Unity® Repeat Expansion Analysis.

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 ataxia associated genes
- ✓ Del/dup analysis of ataxia associated genes
- ✓ Ataxia STR analysis: *ATN1, ATXN1, ATXN2, ATXN3, ATXN7, ATXN8OS, ATXN10, CACNA1A, FXN, NOP56, PPP2R2B, TBP*

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 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.

Genes analyzed

AFG3L2, ANO10, APTX, ATM, ATN1, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, CACNA1A, CACNA1G, CACNB4, COQ8A, EEF2, ELOVL4, ELOVL5, FGF14, FLVCR1, FXN, GRM1, ITPR1, KCNA1, KCNC3, KCND3, MME, MRE11, MTPAP, NOP56, PDYN, POLG, PPP2R2B, PRKCG, SACS, SETX, SIL1, SLC1A3, SNX14, SPTBN2, STUB1, SYNE1, SYT14, TBP, TDP1, TGM6, TMEM240, TTBK2, TTPA, TWNK, VAMP1