variantyx

Genomic Unity® Ataxia Repeat Expansion 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.

Genomic Unity® Ataxia Repeat Expansion Analysis is an effective test for diagnosis of ataxias caused by tandem repeat expansion of 12 different loci. For comprehensive coverage of ataxia-related genes in addition to these repeat expansion loci, consider Genomic Unity® Ataxia 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 repeat expansion genes
- ✓ Del/dup analysis of ataxia repeat expansion 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

- 99.5% sensitivity
- 99.5% 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.

Genes analyzed

ATN1, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, CACNA1A, FXN, NOP56, PPP2R2B, TBP