



Genomic Unity[®] Case Study

Shifting the diagnostic paradigm with a whole genome platform

Biallelic *RFC1* expansion confirms clinical CANVAS diagnosis in 70-year-old male

Clinical presentation

A 70-year-old male with a suspected clinical diagnosis of CANVAS presented with multiple symptoms including:

- Progressive ataxia with onset in late 50's
- Progressive sensory neuropathy including decreased lower extremity vibration and position sense bilaterally, positive Romberg test
- Dysphagia
- Vision changes

Previous genetic testing

No previous genetic testing was reported.



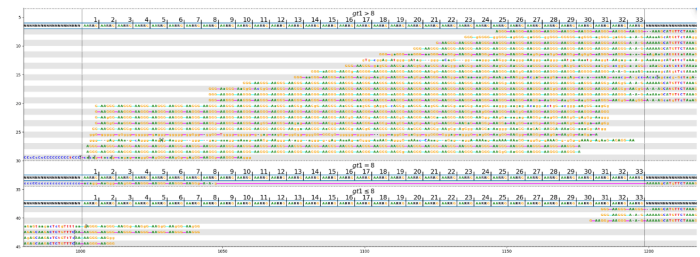
Genomic Unity[®] Comprehensive Ataxia Analysis was ordered as first-line testing because of its ability to identify all relevant variant types in a single test, including repeat expansions in 17 ataxia genes.

Results and interpretation

Varietyx **Genomic Unity[®] Comprehensive Ataxia Analysis** identified two fully expanded *RFC1* alleles with >250 AAGGG repeats.

The expansions were confirmed internally using Oxford Nanopore long read sequencing.

Diagnosis: Cerebellar ataxia, neuropathy, vestibular areflexia syndrome (CANVAS)



Uniform data from WGS clearly identifies the biallelic expansion.

The Variantyx difference

Why was this biallelic *RFC1* expansion identified by Genomic Unity® Comprehensive Ataxia Analysis, when it would have been missed by other ataxia tests?

- ✓ Repeat expansions can not be detected by standard genetic tests, including panels and exomes.

Variantyx genome analysis detects all major variant types in a single test including small sequence changes, structural variants, repeat expansions and mitochondrial variants - which is critical for first-line testing of ataxia patients.

- ✓ Repeat expansions can be detected by PCR and Southern blotting, but *RFC1* would have to be specifically targeted. Very few available ataxia tests are able to overcome the challenges of this locus.

Variantyx genome analysis detects repeat expansions in >35 genes, including *RFC1*. Variantyx is one of the first labs to offer *RFC1* testing.

Variantyx tests that would have identified this variant

Genomic Unity® Whole Genome Analysis | Genomic Unity® Exome Plus Analysis | Genomic Unity® Movement Disorders Analysis | Genomic Unity® Comprehensive Ataxia Analysis | Genomic Unity® Ataxia Repeat Expansion Analysis | Genomic Unity® Neuromuscular Disorders Analysis | Genomic Unity® Neuropathies Analysis

Bring the power of whole genome sequencing to your practice today.

To get started, contact your Clinical Sales Specialist and visit us online at [variantyx.com](https://www.variantyx.com).