Genomic Unity® Case Study

Shifting the diagnostic paradigm with a whole genome platform

Different diagnosis, same phenotypes

Shared phenotypes	Age	Additional phenotypes	Genomic Unity® finding
Progressive cerebellar ataxia, gait imbalance	70 year old male	Positive Romberg test, vision changes	Biallelic *RFC1* expansion Diagnosis: CANVAS
	67 year old male	Tingling and numbness, nystagmus, hearing loss	*FGF14* expansion Diagnosis: SCA27B
Progressive sensory neuropathy	59 year old female	Eye gaze abnormalities, history of cancer	*ATXN1* expansion Diagnosis: SCA1
Dysphagia and/or dysarthria	73 year old female	Leg spasticity, arm/ neck stiffness	Biallelic *FXN* expansion Diagnosis: Friedreich Ataxia
	67 year old female	Chorea, tremors, memory issues	*NOTCH2NLC* expansion Diagnosis: Neuronal Intranuclear Inclusion Disease

Variantyx

The Variantyx difference

Why was Genomic Unity® Comprehensive Ataxia Analysis an appropriate choice as a first-line test for these patients?

- 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 and repeat expansions. This is critical for ataxia patients where repeat expansions are a likely cause.

- While repeat expansions can be detected by PCR and Southern blotting, each suspected gene would have to be specifically targeted and tested.

  Only Variantyx genome analysis detects repeat expansions in >35 genes, including challenging loci like *RFC1, FGF14* and *NOTCH2NLC*, so you don't have to choose which to focus on.

- Interruptions can impact interpretation of pathogenicity, but PCR and Southern blotting typically only look at overall repeat length.

  Variantyx genome analysis identifies interruptions, allowing intermediate sized alleles without any interruption to be correctly classified as pathogenic, as in this *ATXN1* case.

Variantyx tests that would have identified this variant

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

Variantyx

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