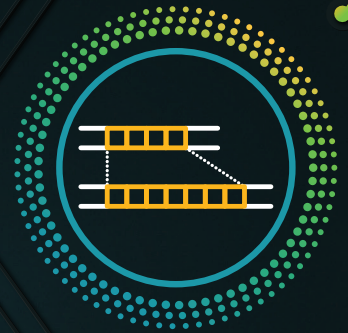


Simultaneous Analysis of Up to 23 Repeat Expansion Loci. All in 1 Run.

Streamline Your Repeat Expansion Research
with EGM and OhmX™



Current methods for detecting repeat expansion disorders face significant technical and workflow challenges, making it difficult to efficiently scale. Electronic genome mapping (EGM) on the OhmX Platform represents an innovative approach to addressing many of these challenges in a single run on the OhmX Analyzer. In as little as 4 days, allele-specific repeat sizing up to 23 loci can be determined using the RepX Analysis Pipeline on the Nabsys Navigator™ cloud platform.

How It Works

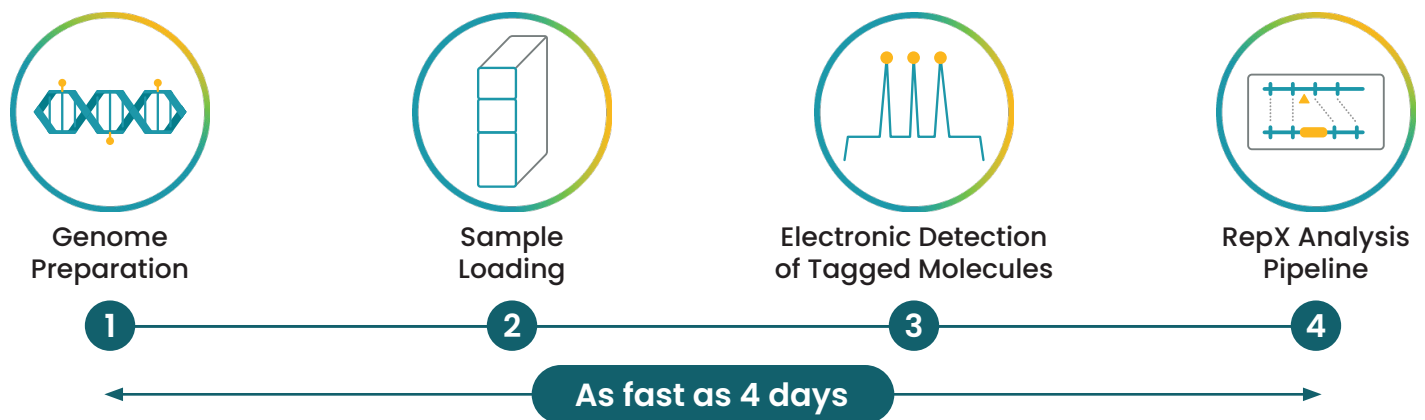


Figure 1. Schematic representation of RepX repeat expansion analysis pipeline workflow on the OhmX Platform

Key Benefits

- **Amplification-Free**
No amplification or ligation steps, preserves the original HMW DNA
- **23 Loci Covered in One Assay**
Spans ataxias, ALS/FTD, epilepsy, neurodevelopmental and neuromuscular disorders in one assay avoiding tedious locus by locus testing
- **Cost-Effective**
Approaches PCR costs per locus assayed
- **Capture Repeat Instability**
Single molecule sensitivity enables measurement of repeat lengths for every molecule to characterize variation within expanded alleles

Supported Applications

- **Confirmation of Sequencing Results**
Orthogonal confirmation and sizing of expansions called by short- or long-read sequencing, or by repeat-calling software such as ExpansionHunter.
- **Front-Line Specialty Screen**
A single EGM run across ataxia, neuromuscular, and other ambiguous neurogenetic presentations, in place of sequential locus-by-locus testing.
- **Research and Pharma**
Genetic characterization, qualification, and trial recruitment. Consistent repeat sizing across a cohort of samples.

Example Data

FMRI (Fragile X) repeat expansion

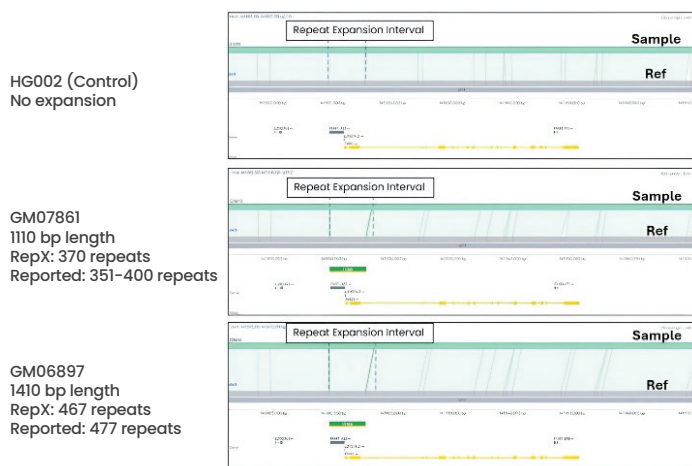


Figure 2. *FMRI* CGG expansion detection in cell lines. Reference hg38 carries 20 CGG repeats. A 1,100 bp insertion was detected in GM07861 (reported 351-400 repeats, 993-1,140 bp) and an 1,400bp insertion in GM06897 (reported 477 repeats, 1,371 bp)

Concordance to gel sizing in *FXN* (Friedreich's ataxia)

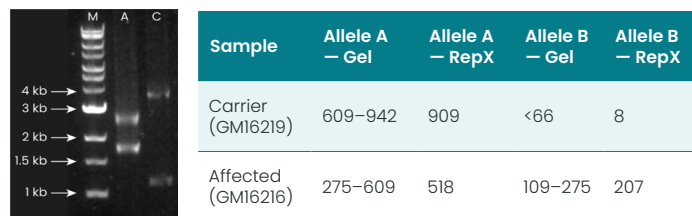


Figure 3. *FXN* expansion detection in cell lines. (Left) Gel image of PCR products for alleles in affected (A) and carrier (C) samples, with affected allele sizes of ~2.5kb and ~1.6kb, and carrier allele sizes of ~3.7kb and ~1.2kb. (Right) Estimated repeat sizing by gel and RepX for each allele, demonstrating general agreement between methods



See How RepX Analysis Fits In Your Repeat Expansion Workflow

Request a demonstration of RepX Analysis in Nabsys Navigator, or download the *FXN* and *FMRI* technical notes at nabsys.com/applications/repX

DMPK (Myotonic dystrophy type 1) repeat expansion

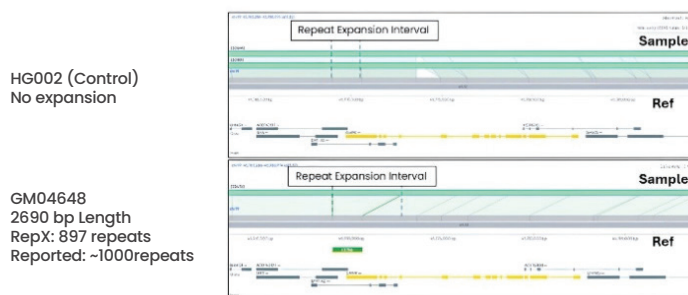


Figure 4. *DMPK* CTG expansion detection in GM04648. The HG002 control shows no expansion across the repeat expansion interval. A 2,690 bp insertion was detected in GM04648 (RepX estimate 897 repeats; reported ~1,000 repeats).

Disease Gene	Gene
Friedreich's ataxia	<i>FXN</i>
Spinocerebellar ataxia type 10	<i>ATXN10</i>
Spinocerebellar Ataxia type 31	<i>BEAN1</i>
Spinocerebellar Ataxia type 36	<i>NOP56</i>
CANVAS / late-onset ataxia	<i>RFC1</i>
Parkinson's Disease/Multiple System Atrophy	<i>FGF14</i>
Myotonic dystrophy type 1	<i>DMPK</i>
Myotonic dystrophy type 2	<i>CNBP</i>
Oculopharyngodistal myopathy 1	<i>LRP12</i>
Oculopharyngodistal myopathy 1	<i>GIPC1</i>
Frontotemporal dementia (FTD)/ALS	<i>C9ORF72</i>
Frontotemporal lobar degeneration (FTLD)	<i>GOLGA8A</i>
Unverricht-Lundborg disease	<i>CSTB</i>
Familial adult myoclonic epilepsy type 1	<i>SAMD12</i>
Familial adult myoclonic epilepsy type 2	<i>STARD7</i>
Familial adult myoclonic epilepsy type 3	<i>MARCHF6</i>
Familial adult myoclonic epilepsy type 6	<i>TNRC6A</i>
FRAXE intellectual disability	<i>AFF2</i>
FRA12A intellectual disability	<i>DIP2B</i>
Fragile X syndrome / FXTAS / FXPOI	<i>FMRI</i>
Glutaminase deficiency with neurodevelopmental disorder	<i>GLS</i>
Fragile site FRA11B	<i>CBL</i>
Baratela-Scott syndrome	<i>XYLT1</i>

Table 1. List of repeat expansion loci supported by RepX Analysis Pipeline.