MNG LABORATORIES
Neurogenetic Answers™

MNG Exome™

Whole Exome Sequencing (WES) is a genetic test used to identify a heritable cause of a disorder. WES searches through all coding regions of all genes currently identified, thus it has a higher chance to find the cause of a heritable disease.

The MNG Exome™ is the most comprehensive commercially available exome test. We ensure coverage of *all* practice guideline and expert panel reviewed ClinVar pathogenic variants. These include variants located in intronic regions that are not covered by standard exome sequencing. In addition to sequencing, we provide copy number analysis, mitochondrial genome sequencing + deletion and heteroplasmy assessment, and detection of uniparental disomy.

Methodology & Technology

Guaranteed 100% coverage of expert approved ClinVar pathogenic variants, including variants located in intronic regions

Includes **copy number** analysis, **mtDNA** sequencing + deletion and heteroplasmy assessment, and detection of **uniparental disomy**

Assessment of **>99%** of targeted regions, target **200 fold** average coverage, guaranteed **160 fold** average coverage

Variant confirmation via alternative technology

Based on the Agilent SureSelect Human All Exon V6 technology

Data analysis performed by MNG Genome MaNaGer® pipeline

MNG Exome™

Test Offerings

Turnaround time: 2-4 weeks

Test Name	Test Code	Description
MNG Exome™ Trio Sequencing + mtDNA Sequencing	WES001	Proband + up to 2 family members. Trios are preferred for better diagnostic sensitivity. mtDNA analyzed for proband even if the mother's DNA is not available.
MNG Exome™ Additional Family Member	WES002	One or more additional family members can be included when ordering WES001 to increase probability of establishing the correct diagnosis. Depending on findings in the trio, mtDNA may also be used to help in interpreting and reporting.
MNG Exome™ Proband Only Sequencing + mtDNA Sequencing	WES003	We will accept proband only orders when parental or other family member samples are not available.

Note: The MNG Exome™ does not report trinucleotide repeat expansions or genomic rearrangements. We also do not report out carrier status for the proband for diseases unrelated to the condition for which testing is ordered. Whole exome sequencing requires the submission of a signed consent form.

MNG Genome MaNaGer® Pipeline

The value of next-generation sequencing depends on the sensitivity and specificity of the testing as well as clear, user-friendly interpretation and reporting of results. Our unique approach and proprietary process to evaluating the significance of genetic changes centers on the concept of genetic disorders as diseases of molecular systems, rather than individual genes.

15 Years of Sequencing Experience

- Epilepsy
- Movement Disorders
- Neuromuscular
- Neurobehavioral
- Neurometabolic
- Cardiovascular
- Mitochondrial
- Other Inherited Disorders

Proprietary Neurogenetic Database

Genome MaNaGer® Pipeline ensures high specificity & sensitivity of variant detection and interpretation

Expert Decision Making

Functional information about genes and variants obtained from model organisms and *in silico* modeling

First-In-Class Reporting

Reporting variants that are either certain or likely to cause the condition based on available scientific evidence