



# MNGenome®

Whole genome sequencing is a novel diagnostic tool used to identify deep intronic regions with known pathogenic variants by sequencing the entire human genome. Approximately 20% of known pathogenic disease causing variants are outside the exon boundaries, which are undetected using exome sequencing. The MNGenome® has the ability to detect mitochondrial depletion, deletion, heteroplasmy, single nucleotide resolution CNV, and repeat expansions. It is a comprehensive approach to a diagnosis of genetic disorders and is the best option to end your patients' diagnostic odyssey.

The MNGenome® can be used if other methods such as WES and CMA failed to provide diagnostic or prognostic insight into a patient's condition, or suggest a therapeutic approach. MNGenome® will increase the probability of a correct diagnosis and suitable treatment options.

## Methodology & Technology

>99% of mappable regions of the genome are sequenced at >30X coverage

Turnaround time of 2-6 weeks

Pathogenic **SNV detection** across the entire genome

Mitochondrial **sequencing, depletion, and deletion** analysis with heteroplasmy assessment

Single nucleotide resolution **copy number analysis** with junction identification

Uniparental disomy detection

**Repeat expansion detection** with confirmation by alternative technology

**STAT option of ≤2 weeks available**

# MNGenome<sup>®</sup>

## Test Offerings

Turnaround time: 2-6 weeks

STAT option ≤ 2 available

Test Name	Test Code	Description
MNGenome <sup>®</sup> Trio Sequencing	WGS001	Proband + up to 2 family members. Trios are preferred for better diagnostic sensitivity.
MNGenome <sup>®</sup> Proband Only Sequencing	WGS003	We will accept proband only orders when parental or other family member samples are not available.

*Note: We do not report out carrier status for the proband for diseases unrelated to the condition for which testing is ordered. Whole genome sequencing requires the submission of a signed consent form.*

Sample Type	Sample Collection Kits
<b>Preferred</b> <ul style="list-style-type: none"><li>• Blood</li></ul>	<b>Alternative</b> <ul style="list-style-type: none"><li>• Muscle</li><li>• Buccal Swab</li><li>• Fibroblasts</li><li>• Extracted DNA</li></ul>
MNG Exome™/MNGenome <sup>®</sup> Kit <i>(Includes 3 EDTA tubes for blood collection for WGS001 and WGS003)</i>	

## MNG Genome MaNaGer<sup>®</sup> 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.

