

# MyWES<sup>®</sup> (WES) & iGenome<sup>®</sup> (WGS) with NGS



**Whole genome  
sequencing results  
delivered in 5 weeks**

Next Generation Sequencing  
for the Diagnosis of Genetic Disorders



**Genekor**

Committed to Biotechnological Innovation

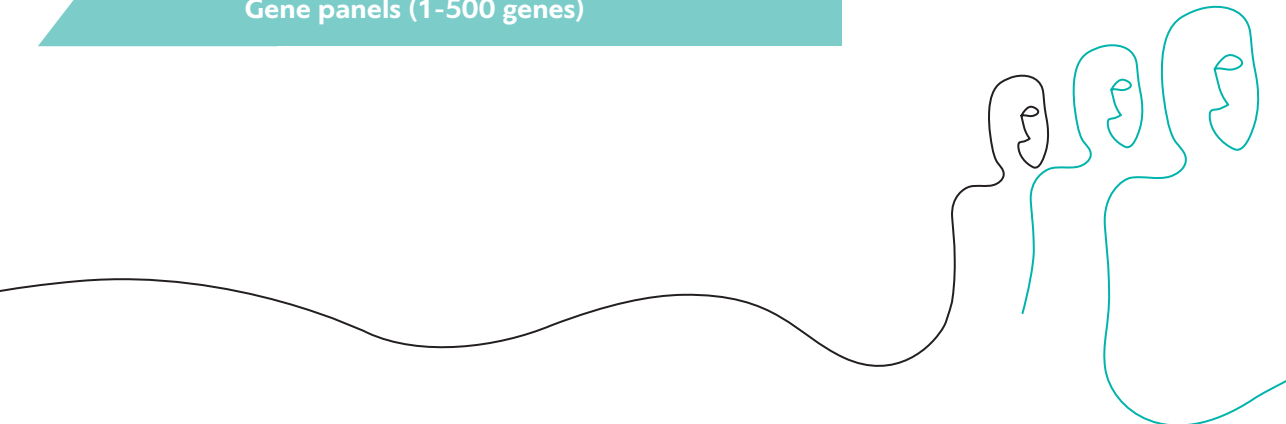
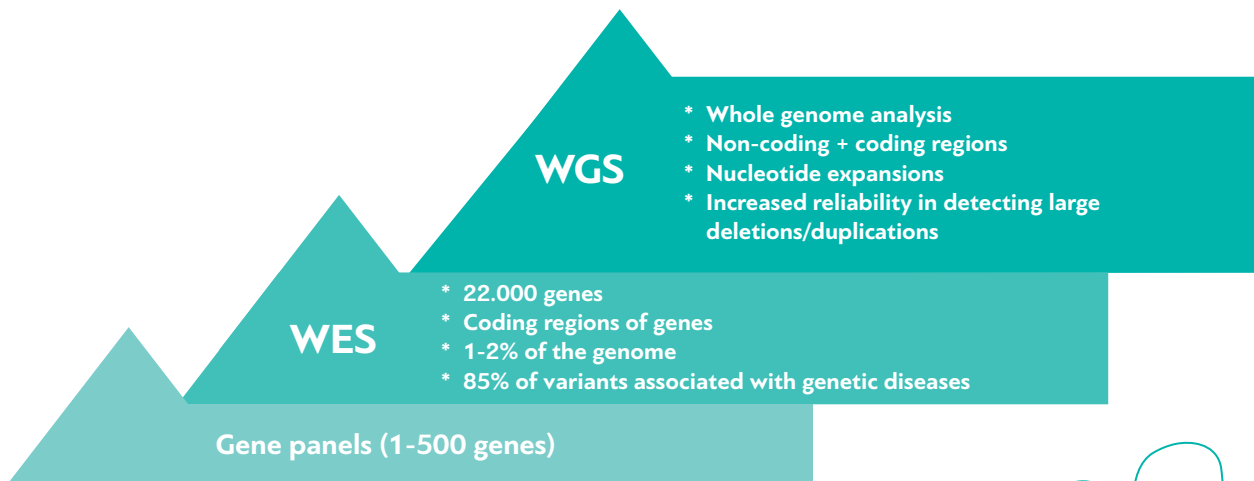
# Genetic Analyses and Clinical Utility

## MyWES<sup>®</sup> – Whole Exome Sequencing (WES)

- Nuclear
- Complete sequencing of the coding regions of the genome (exons) – 22,000 genes
- 85% of the genetic basis of hereditary diseases is located in the coding regions
- Clinical utility – Databases and Clinical Guidelines

## iGenome<sup>®</sup> – Whole Genome Sequencing (WGS)

- Nuclear/Mitochondrial DNA
- Complete sequencing uniform coverage of the entire genome
- Higher sensitivity for detecting large structural changes/nucleotide expansions
- Possibility for future reanalysis



Test Features	MyWES®	iGenome®
Types of Variants	<ul style="list-style-type: none"> <li>- SNVs/indels/CNVs in coding regions and coding – noncoding region boundaries</li> <li>- mitochondrial genome</li> </ul>	<p><b>WHOLE GENOME</b></p> <ul style="list-style-type: none"> <li>- SNVs/indels/CNVs in coding and noncoding regions boundaries</li> <li>- mitochondrial genome</li> <li>- nucleotide expansions</li> </ul>
Depth of Analysis	>100X	30-40X
Turnaround Time	20 working days	25 working days
Possibility for Future Reanalysis	✓	✓
Secondary Findings: 84 Clinically Significant Gens from the ACMG SF v3.3 List Based on International Guidelines	✓	✓

## In which cases do we choose iGenome® (WGS)?



- Complex phenotype e.g. Syndromic clinical presentation
- Differential diagnosis – Unspecified clinical diagnosis
- Previous genetic testing (gene panel) without a positive result, but a genetic basis is still suspected

**The use of genetic testing contributes to confirming the diagnosis and optimizing therapeutic management in areas such as:**

- Neurology (Cerebrum DX® ► neurodevelopmental disorders, epilepsy, neurodegenerative diseases, etc.)
- Cardiology (Cordis DX® ► cardiomyopathies, arrhythmogenic syndromes, aortopathies)
- Diseases with suspected genetic background (endocrinological, metabolic, pediatric, rare diseases, etc.)
- Preconception carrier screening - OIKOGENE®
- Screening of asymptomatic individuals to assess the risk of developing medically actionable genetic conditions.
- Pharmacogenomic analyses

# Clinical utility of using parental samples in WGS – Trio Analysis

The diagnostic value is higher, more accurate, and faster in Trio analysis, where the sequencing of parental samples is performed simultaneously with that of the child being tested.

## Why choose GENEKOR for Genetic testing?

**Genekor is the only laboratory in Greece with the technological infrastructure and expertise to perform Whole Genome Sequencing (WGS) analyses.**

Advanced Next-Generation Sequencing (NGS) MGI DNBSEQ T7

- Speed – Whole genome analysis results delivered in 5 weeks!
- Depth of analysis
- Reliability
- Clinical laboratory geneticists with years of experience for result interpretation

*Before testing, genetic counseling is provided for the most accurate evaluation of the clinical phenotype and to determine the appropriate type of analysis to use.*

*For more information, you may contact the Genekor scientific team at +30 210 60321388.*

*Clinical Laboratory Geneticist & Head of the Inherited Diseases Department: Dr. Dimitra Bouzarelou*

### Accreditations & Certifications



**Clinical Reliability:** ISO 15189:2022 (Cert. No. 822)\* & CAP Accredited Laboratory.



**Data Security:** Certified according to ISO/IEC 7001:2022 for Information Security Management.



**Quality Management:** ISO 9001:2015 Certified Quality Management System.



**Method Reliability:** MyWES® is performed using CE-IVD marked reagents/software, in compliance with IVDR.

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