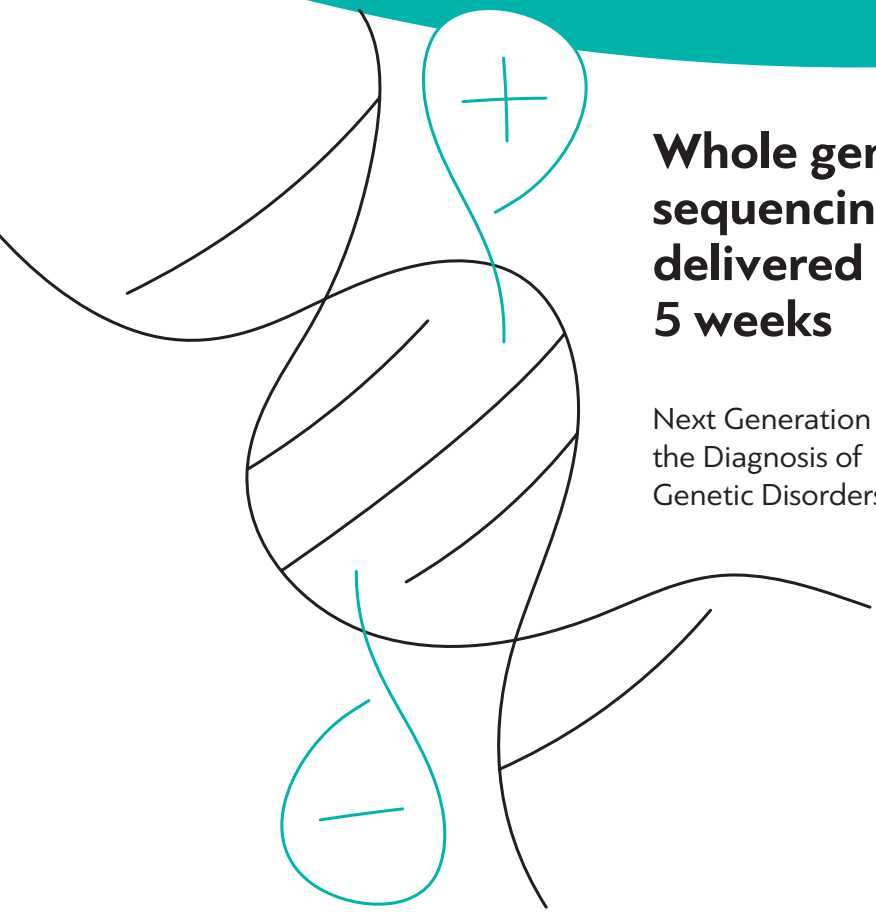
MyWES (WES) & iGenome (WGS) with NGS



Whole genome sequencing results delivered in 5 weeks

Next Generation Sequencing for the Diagnosis of Genetic Disorders

Genekor

Committed to Biotechnological Innovation

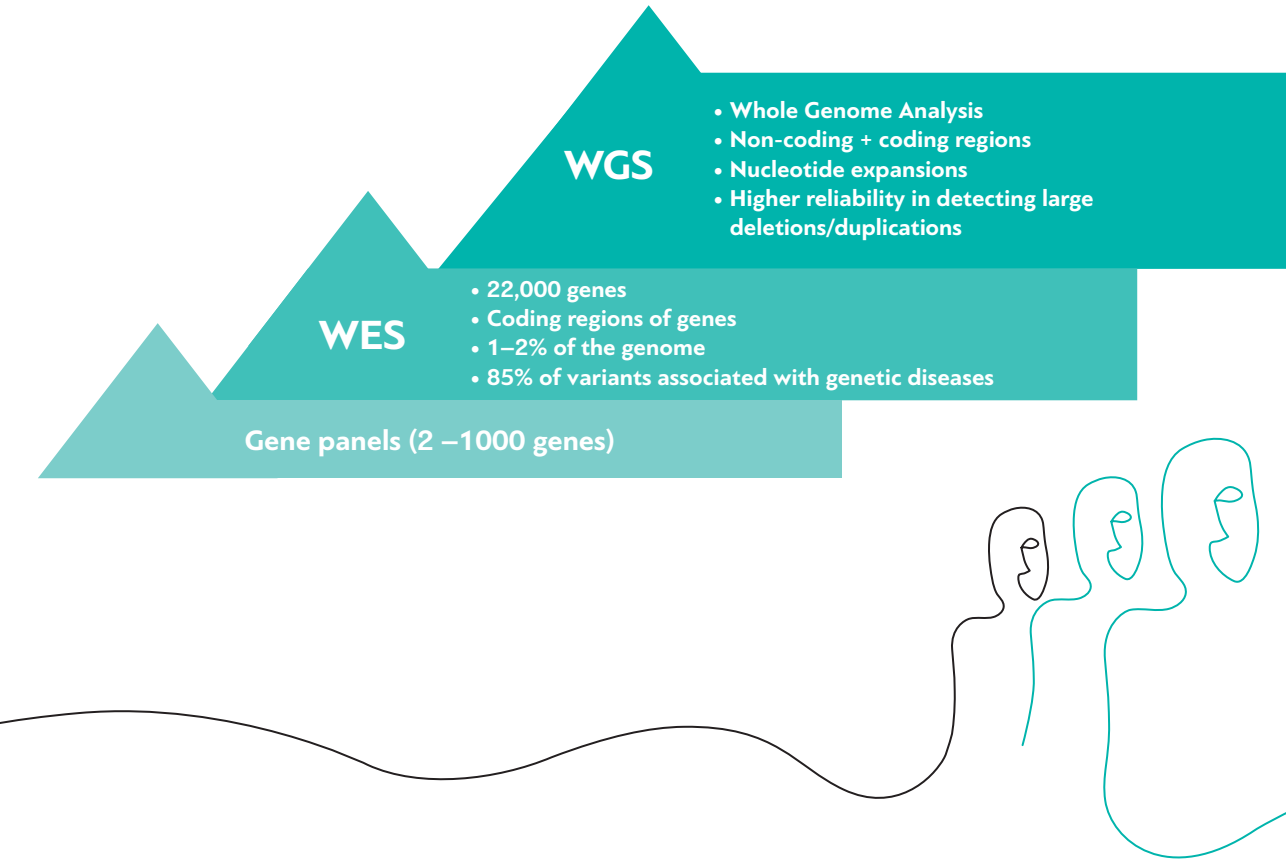
Genetic Analyses and Clinical Utility

MyWES - Whole Exome Sequencing-WES

- Nuclear / Mitochondrial DNA
- 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

Whole Genome Sequencing-WGS

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



Test Features	Whole Exome sequencing-WES	Whole Genome sequencing-WGS
Types of Variants	SNVs/indels/CNVs in coding regions and coding – noncoding region boundaries of the mitochondrial genome	**WHOLE GENOME** SNVs/indels/CNVs in coding and noncoding regions of the mitochondrial genome, nucleotide expansions
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	√	√
Pharmacogenomics		√

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 (WES) – neurodevelopmental disorders, epilepsy, neurodegenerative diseases, etc.)
- Cardiology (Cordis DX (WES) – 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 genetic risk of developing certain medically actionable genetic diseases
- 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 has one of the top technological infrastructure and expertise to perform Whole Genome Sequencing analyses in Europe.

Advanced Next-Generation Sequencing (NGS) Technology – 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.

Clinical Laboratory Geneticist & Head of Inherited Diseases Dpt: Dr. Dimitra Bouzarelou

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