



The analytical power of WGS is the outcome of its deep analysis of promoter regions, regulatory domains as well as intronic regions of DNA in addition to the coding exons typically covered by other types of tests. The result, is the most powerful test for the identification of genomic variations.

This comprehensive WGS is considered the gold standard in genomic testing and provides invaluable information for the purpose of:

Making a diagnosis: Some genetic conditions can pose a diagnostic challenge that simply cannot be addressed by single-gene testing. WGS offers the most comprehensive solution for a genetic diagnosis.

Precision medicine: Because we are unique from one another, our response to medication differs as well. WGS analyses changes in DNA to identify medication that aligns with your DNA, avoiding the time-consuming and frustrating trial-and-error approach to treatment for diseases such as cancer, diabetes, heart disease, etc.

Disease prevention: Genetic predisposition for diseases are encoded in DNA. WGS can identify such predisposition to inform the implementation of a strategic prevention plan based on your DNA.

HELPING UNDERSTAND GENETIC MYSTERIES



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WHOLE GENOME SEQUENCING

THE MOST COMPREHENSIVE GENETIC TEST

Whole Genome Sequencing (WGS), based on Next-Generation Sequencing technology, is the most superior test in genomics with the power to analyze up to 98% of the human DNA.

Do we need a Trio?

- 'Trio WGS' is an extremely powerful diagnostics tool that involves testing a patient as well as their parent, in parallel, in order to make accurate correlations between genetic findings and clinical presentation.
- The most important application of Trio WGS is the identification of a genetic diagnosis in the commonly seen clinical scenario of a suspected genetic presentation in the absence of family history.
- It is cost-effective, time saving, has the highest detection-rate and clinical meaningfulness.



Technology

- Mapmygenome's WGS is performed using Next-Generation Sequencing (Illumina HiSeq X, 150PE sequencing), with a coverage of 30X*
- WGS at Mapmygenome offers maximum coverage of the human genome ~ 95-98% of the genome
- Our service includes the comprehensive bioinformatics analysis (with sequence alignment, variant calling and annotation), clinical analysis and a detailed report
- If required, our Genetic Counselor's are available to you for technical assistance and report interpretation

*Does not include Sanger confirmation

Reporting

- WGS findings are categorized into Pathogenic, Likely pathogenic, Variant of uncertain significance (VUS), Likely benign and Benign variants in accordance with American College of Medical Genetics and Genomics (ACMG) guidelines
- Primary findings and incidental findings are reported.
- Genetic Counseling is provided

Stauropoulos et al, 2016

Population studied: Inherited diseases in children (neurological diseases/ congenital anomalies)

Recommended WGS as a 'primary test' in pediatric medicine diagnostics; WGS has '4 fold' higher diagnostic rate (34%) than chromosomal microarray analysis (8%) and '>2 fold' increase when compared to microarray plus targeted gene sequencing (13%).

Mapmygenome is a molecular diagnostic company with a technical expertise of 16+ years in the domains of genomics, genetic counseling and molecular genetics big data analysis. We provide a one-stop solution for all genetic services to clinicians. Our teams comprise of biotechnologists, statisticians, genetic counselors, scientists, bioinformaticians, and medical counselors. Our advisory panel has expert scientists and doctors of different medical disciplines.