
WHOLE
GENOME
SEQUENCING
WGS
İNTERGEN
Genetics and
Rare Diseases
Diagnosis Center

Whole Genome Sequencing (WGS) is a DNA sequencing technique that refers to the determination of the complete genetic material, or genome, of an organism. This technique aims to identify the entire DNA sequence of an individual or organism.
In recent years, this technique has begun to be used in the diagnosis of diseases and research to identify genetic variants. Genome sequencing is used to determine drug sensitivities and perform haplotype analyses, especially for patients who cannot be diagnosed with exome sequencing.

In Which Diseases Is Whole Genome Sequencing (WGS) Used for Diagnosis/Detection?
Monogenic Diseases: It is an effective tool in diagnosing genetic diseases caused by a single genetic mutation. It is particularly used to find and understand the causes of rare genetic diseases.
Cancer: Genomic analysis of cancer cells can be performed with WGS. This can be used to determine familial cancer risks, identify cancer types, define potential targets, and personalize treatment strategies.
Genetic Variants and Polymorphisms: WGS is effective in determining numerous mild variants and carrier statuses in individuals. When genetic polymorphisms are evaluated together, it allows for the determination of risk scores for many diseases. It can provide solutions for multifactorial diseases and diseases where the cause cannot be clearly identified.
Carrier Statuses: WGS is an important tool for identifying genetic diseases that an individual may carry. This information allows for early diagnosis of certain diseases that may emerge in later ages and facilitates planning before marriage and childbearing if there is a plan.
Genetic Regions with Rearrangements: WGS can be effective in identifying large-scale changes in genetic structure. Situations such as structural variations, chromosomal changes, and translocations can be detected. (Long-read whole genome sequencing has a higher capture rate for these changes.)
Pharmacogenetic Applications: WGS can be used to identify genetic factors affecting individuals' drug metabolism. This can play a very important role in personalized medicine applications.

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