

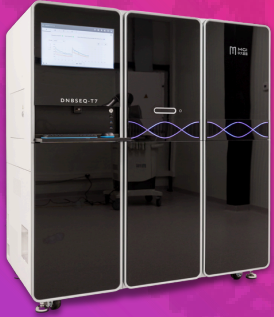
What is Whole Exome Sequencing (WES)?

Whole Exome Sequencing (WES) is a next-generation sequencing (NGS) technique widely used around the world and in our country. It focuses on sequencing the protein-coding regions of the genome, known as the exome.

The exome contains all DNA segments responsible for protein production in humans. Although it represents less than 2% of the entire genome, it includes about 85% of the genetic variants known to be associated with human diseases.

WES is performed to identify variants in the exons. By determining the order of DNA sequences in these regions, it helps detect genetic disorders and provides important information for diagnosis and treatment

Which Device Are We Using?



DNBSEQ-T7

Intergen Diagnostic Algorithm

At Intergen, no diagnosis is made solely by software. Our WES Analysis and Reporting Team of 35 experts, together with our clinical team, carefully evaluate each case. Every WES analysis is reviewed by at least three specialists, each conducting the analysis multiple times.

Using our in-house analysis algorithm combined with a clinical perspective, we focus not on listing every possible variant, but on reporting only those clinically relevant variants, along with their interpretations and, when appropriate, treatment recommendations. Thanks to this comprehensive approach, our diagnostic yield for WES reaches up to 70%, compared to the global average of around 40%.

In addition, the Intergen clinical team provides genetic counseling both before and after testing, supporting patients and their physicians throughout the entire process. Reports are explained directly to the patient and physician, and post-diagnosis management can be organized according to the recommendations of the Intergen clinical team.

WHOLE EXOME SEQUENCING WES

In Summary:

- WES can provide a diagnosis for patients without a current diagnosis.
- It supports differential diagnosis in conditions with multiple possible causes (e.g., epilepsy).
- It can identify additional hidden diseases that may complicate the patient's clinical picture.
- It reveals recurrence risks within families and detects high-risk but asymptomatic individuals (e.g., arrhythmias).
- It has become a routine tool for all chronic diseases

What Does Whole Exome Sequencing (WES) Offer?

Whole Exome Sequencing (WES) is the process of analyzing all the protein-coding regions of an individual's DNA (the exome).

Diagnosis of Genetic Disorders: WES is an effective tool for diagnosing genetic diseases, especially for identifying the causes of rare genetic conditions.

Preventive Medicine: WES can identify diseases that may develop later or are difficult to diagnose under challenging conditions (e.g., familial cancers, bleeding disorders).

- It helps prevent complications caused by unknown diseases in the patient.
- Pre-marital and pre-pregnancy screenings can reduce the risk of affected children and maternal complications.
- WES is considered one of the gold standards in extended newborn screening.

Cancer Research: Lifetime cancer risk is approximately 40–45% in men and 35–38% in women, with about 10% being familial. This means around 4% of the population has a familial form of cancer, with higher rates in those with a positive family history.

- Monitoring familial cancers helps reduce risk.
- Treatment for familial cancers may differ from sporadic cases.
- Screening for familial cancers is important for cancer prevention and in all cancer cases.
- The costs of limited screening (e.g., BRCA1-2) are comparable to those of broader panels, making comprehensive testing worthwhile.

In Which Cases Can I Consider WES?

- Endocrine disorders
- Epilepsy
- Obesity and/or diabetes
- Immune system disorders
- Autoimmune diseases
- Patients with chronic pain
- Suspected autism
- Metabolic disorders
- Patients with a family history of sudden or early death
- Patients with pregnancy loss (miscarriage, recurrent pregnancy loss, stillbirth, birth with anomalies)
- Risks identified during pregnancy monitoring (ultrasound anomalies, abnormal screening test results)
- Patients with a family history of recurrent genetic disorders
- Cancer patients
- Individuals with a family history of recurrent, early-onset, or frequent cancers
- Patients in intensive care
- Newborns in neonatal intensive care
- Patients without a definitive diagnosis
- Patients experiencing side effects from medical treatments
- Patients who do not respond to treatment
- All chronic diseases
- Rheumatologic disorders
- Neurological disorders
- Infertility

Whole Exome Sequencing (WES) Test Workflow

The physician submits a WES request for their patient.

Blood is collected from the patient and first-degree relatives into a purple-capped EDTA tube and shipped to Intergen.

Once the samples arrive at Intergen, the Sample Reception Unit creates the patient record.

Samples with completed registration are sent to the laboratory, and DNA is isolated.

The isolated DNA is prepared by the WES Lab team for the instrument process, and the analysis is initiated.

Once the instrument analysis process is completed, the bioinformatics analysis and data processing begin.

From the moment the patient first visits your clinic until the final report is prepared, detailed clinical genetic counseling is provided to both the physician and the patient. For physicians, the Intergen clinical team supports them throughout the entire process, from selecting the appropriate test for their patient to planning treatment.

After the family study, the final report is sent.

After the preliminary report is sent, the family studies are immediately initiated.

The analyses are submitted for approval to Prof. Dr. Serdar Ceylaner. Once approved, the preliminary report process is initiated.

The prepared data is shared with the WES analysis team and the Medical Genetics Specialist team, and a detailed analysis is initiated.