

Whole Exome Sequencing

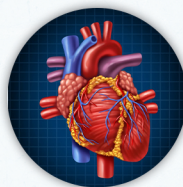
Whole Exome Sequencing is a **Next Generation Sequencing (NGS)** based technique that analyses the protein coding regions of your DNA known as the exons. WES covers 1-2% of the genome containing ~85% of known disease causing variants. WES provides a single, comprehensive test to uncover the genetics of complex conditions.

What Conditions Can WES Detect?

WES identifies mutations linked to hundreds of disorders across key specialties:



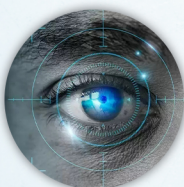
GDD, LD, ASD, Epileptic Seizure,
Neurodegenerative / Neuromuscular
disorders etc.



Cardiomyopathies, MODY ,
Brugada & Long QT syndrome,
Pompe & Gaucher disease, etc.



Polycystic kidney disease, Alport syndrome,
Familial Focal Segmental Glomerulosclerosis,
Von Hippel-Lindau Disease , etc.



Retinitis Pigmentosa,
Leber Congenital Amaurosis,
Achromatopsia. etc.



Familial adenomatous polyposis,
Wilson disease, Gilbert syndrome,
Glycogen storage disease, etc.



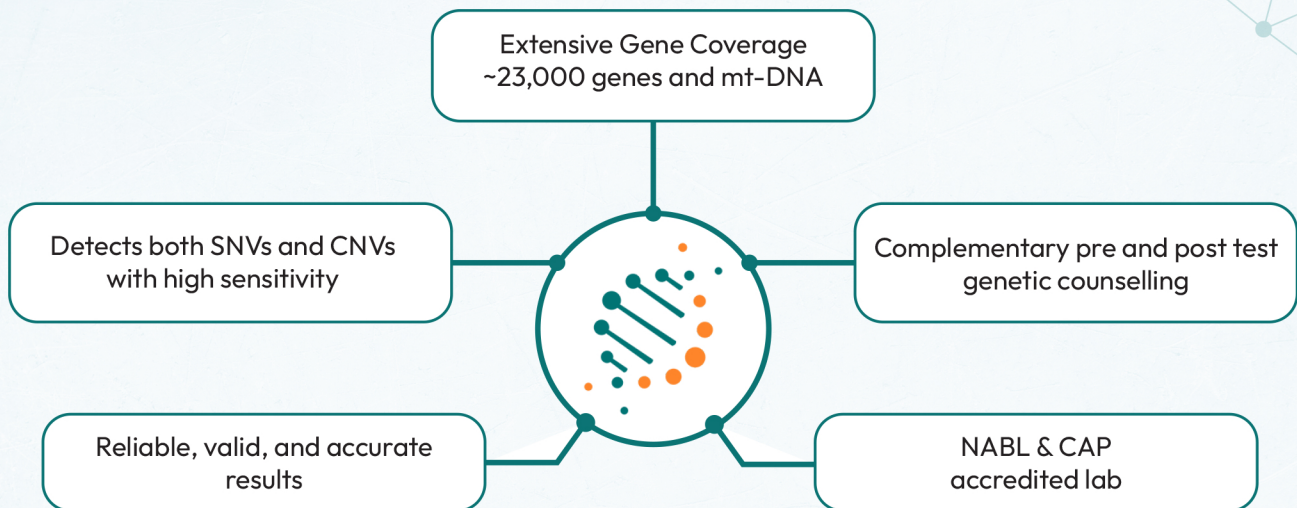
Waardenburg syndrome,
Usher Syndrome, Pendred syndrome,
Treacher Collins syndrome, etc.

Who Should Take WES?

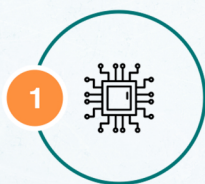
Consider WES if you or your patient:

- Has an undiagnosed underlying genetic disease after standard testing.
- Presents with unclassifiable or heterogeneous symptoms (e.g., developmental delay + speech delay + seizures).
- Seeks a definitive diagnosis to guide disease management and family planning.
- Has a family history of a genetic disorder but the specific mutation is unknown.

Why OneDNA?



Technical Specifications



1
Illumina
NovaSeq 6000



2
99–100%
Sensitivity & Specificity



3
80–100x
Depth



4
~23,000 genes
Analyzed



5
SNVs, CNVs,
mt-DNA mutations



6
TAT
4 Weeks



7
2–3 ml Blood
in EDTA tube

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