

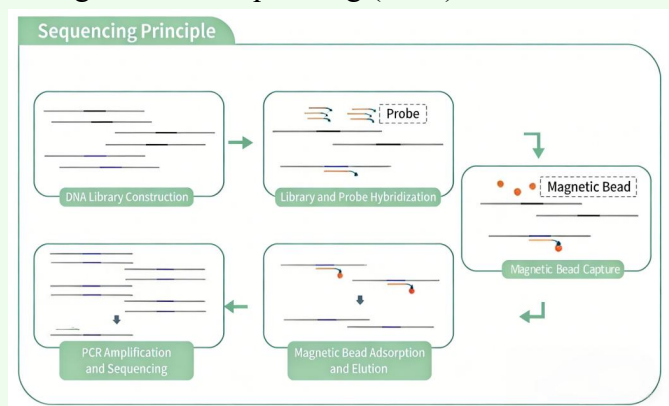
Whole Exome Sequencing (WES)

Product Overview

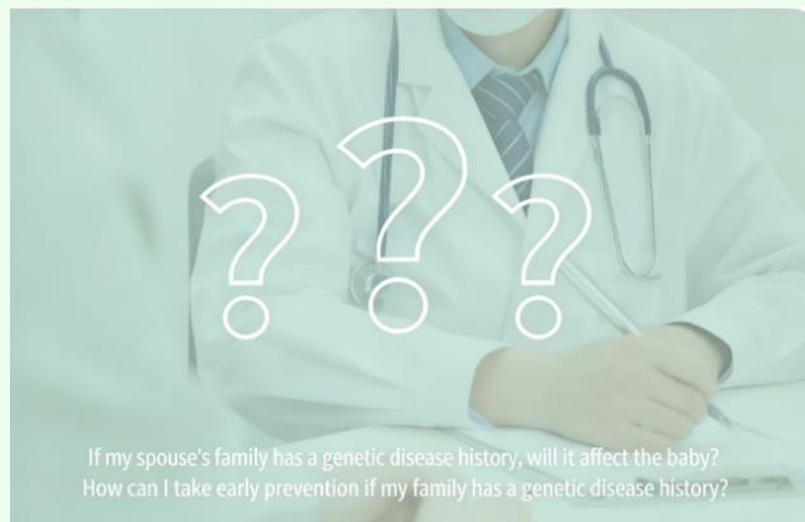
Our Genetic WES platform leverages Next-Generation Sequencing (NGS) and precision capture technology to sequence all exons across **20,000+** human genes. In addition to coding regions, the test detects variants in flanking intronic splice sites and the mitochondrial genome, identifying point mutations and small InDels with high sensitivity. Aligning with the OMIM database, we provide deep clinical insights into **4,000+** genes linked to over **7,000** monogenic conditions. The detection scope spans a wide array of medical specialties, including cardiology, neurology, gastroenterology, nephrology, immunology, hematology, and musculoskeletal medicine.

Methodology

Whole Exome Sequencing (WES) is a cutting-edge genomic analysis technology designed to target and sequence the protein-coding regions of the genome. by leveraging high-efficiency probe capture and next-generation sequencing (NGS).



Why Choose WES?



The exome represents the complete set of all exons within an individual's genome—the specific DNA regions directly responsible for protein coding. Although the human exome comprises over **20,000** genes, it constitutes only **1%–2%** of the entire human genome [1]. Over **85%** of known disease related genetic mutations occur in exonic regions. Notably, the vast majority of known disease-associated genetic mutations are located within these exonic regions.

By leveraging Whole Exome Sequencing (WES) technology, researchers and clinicians can simultaneously sequence all protein-coding regions. This advanced approach enables the precise detection of genetic mutations, polymorphisms, and other hereditary markers linked to disease onset, providing a robust foundation for the clinical auxiliary diagnosis and treatment of genetic disorders.



Sample Type



Solution & Order Information

CYGNUS' complete one-stop solution supports the full experimental workflow, including reagent supply, automated extraction, automated library construction, sequencing and professional data analysis, delivering a seamless and integrated experience .



Order Information

Equipment	Reagent
DNA Extraction +Library Automated Machine	CEXome DNA Library Preparation Kit(48rxn)
Local Data Bioinformatics workstation CYGNUS X1	CEXome Testing Targeted Capture Kit(48rxn)
GS200 Sequencer	Q3C Reagent Kit (PE150) 400M

Reference

[1] Monaghan KKG , Leach NT, pekarekD, prasad Pp, Rose NC;
ACMG professional practice and Guidelines committee.
The use of exome sequencing in prenatal diagnosis:
a points to consider document of the American college of
Medical Genetics and Genomics (ACMG) . Genet MMed. 2020;
22(4):675 - 680 .doi:10 . 1038/s41436 - 019 - 0731 - 7

Clinical Significance

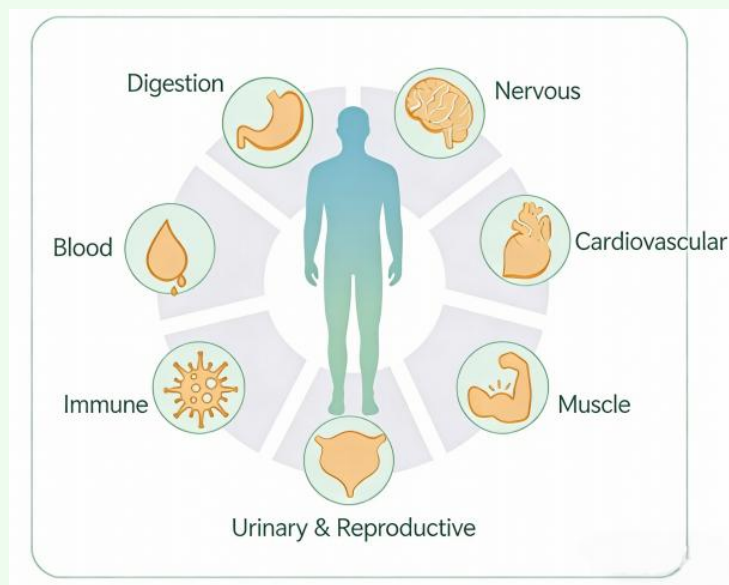
Monogenic Disease Diagnosis: WES facilitates the elucidation of pathogenic mechanisms and inheritance patterns, providing essential diagnostic support for patients with single-gene disorders.

Diagnosis of Complex & Rare Diseases: For patients with atypical clinical features or undiagnosed "medical mysteries," this assay assists in pinpointing the molecular cause of the disease.

Risk Assessment for Families and Offspring: Identifies familial genetic risks to provide reproductive guidance and reduce the incidence of hereditary conditions in future generations.

Personalized Health Management: Enables the development of tailored health management strategies based on the individual's genetic profile to delay disease progression through targeted interventions.

Advancement of Clinical Research: Facilitates the discovery of novel genes or new variants through large-scale case-association studies, driving the evolution of genomic medicine.



Guidelines

ACMG (American College of Medical Genetics and Genomics) Guidelines:

"Standards and Guidelines for the Interpretation of Sequence Variants" (ACMG-AMP, 2015)

"Clinical Practice Guideline for the Use of WES/WGS as a First-Line Diagnostic Test for Congenital Anomalies or Intellectual Disability" (2021)

ACMG Recommendations on Secondary Findings:

ACMG SF v3.2 Guidelines



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