iBio - WHOLE EXOME SEQUENCING (WES)

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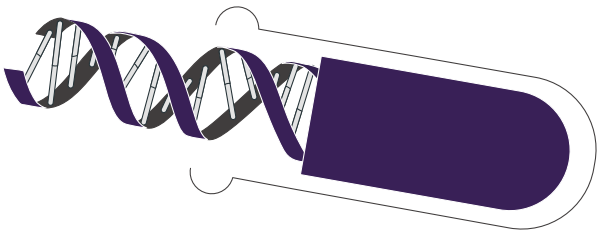
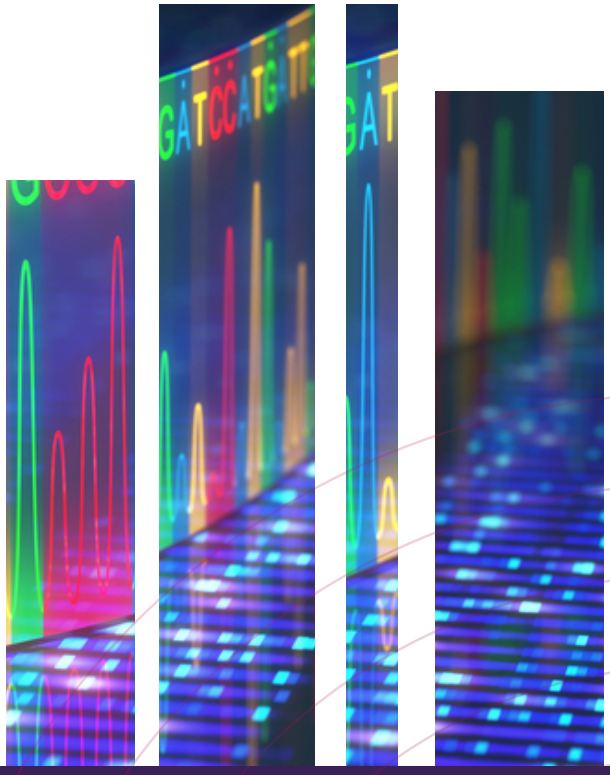
WHOLE EXOME SEQUENCING

- Whole Exome Sequencing (WES) is a comprehensive genetic test that identifies DNA changes in the entire protein-coding region of the genome.
- WES may contribute to diagnosis of rare diseases in less time and at a lower overall cost than traditional DNA testing.
- WES is recommended
  - when symptoms are complex and unspecific
  - when a diagnosis of significant molecular heterogeneity is indicated
  - in case of neurodevelopmental disorders, including intellectual disability, global developmental delay, and autism spectrum disorder.



RARE DISEASES

- Rare diseases are defined in Europe as those that have a prevalence of one in 2000 or less.
- Over 8000 rare diseases are known affecting approximately 6% of the global population.
- 70% of rare diseases are genetically determined, the majority of which may be recognized with WES.
- Without timely, comprehensive genetic testing, patients often have to wait years for the correct diagnosis often following a stressful diagnostic odyssey.



METHOD

- Isolated genomic DNA or peripheral blood sample in EDTA.
- Sequencing of total coding regions as well as 3'/5' UTRs of the entire exon.
- Bioinformatic identification of single nucleotide variants (SNV), short insertions and deletions.
- Variant classification and annotation (e.g. ClinVar, COSMIC, HGMD, dbSNP, Varsome).

COVERAGE: >95% (>500×)

AVERAGE SEQUENCING DEPTH: >1000×

TURNAROUND TIME: 2-3 WEEKS

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iBio SCIENCE

UNIVERSITY OF PÉCS
SZENTÁGOTHAI RESEARCH CENTRE

WORKFLOW

Our mission is advancing scientific research in the fields of **BIOTECHNOLOGY** and **MEDICINE** as well as applying the latest innovative technologies in diagnostics. **IBIOSCIENCE LTD.** in collaboration with **UNIVERSITY OF PÉCS SZENTÁGOTHAI RESEARCH CENTER** provides state-of-the-art next generation sequencing services and expertise for the Hungarian scientific community.

01 **CONTACT**
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02 **SAMPLE DISPATCH**
DNA or peripheral blood

03 **SAMPLE PROCESSING**
paired-end 150 bp sequencing

04 **GENETIC ANALYSIS**
bioinformatic analysis, variant identification and annotation

05 **REPORT**
categorization of variants based on guidelines (pathogenic, likely pathogenic, VUS etc.)

REFERENCES

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