iBio SCIENCE | PÉCSI TUDOMÁNYEGYETEM SZENTÁGOTHAI JÁNOS KUTATÓKÖZPONT

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.

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WORKFLOW

01 **CONTACT**
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02 **SAMPLE DISPATCH**
DNA/RNA, liquid or tumor biopsy sample

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.)

iBio DIAGNOSTICS

GENETIC TESTING SOLUTIONS FOR INHERITED DISEASES

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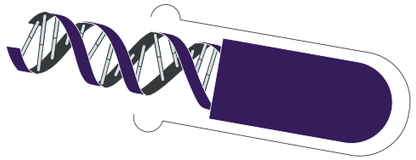
GENE PANELS AND WHOLE EXOME SEQUENCING

- Our various gene panels include clinically relevant and validated genes. All of our tests include both SNP and deletion/duplication (CNV) analysis.
- Whole Exome Sequencing (WES) is offered when a specific genetic test is not available. WES is a comprehensive genetic test that identifies DNA changes in the entire protein-coding region of the genome.
- Gene panels and 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.

TURN AROUND TIME FOR ALL OUR PANELS AND WES:
2-3 WEEKS FROM SAMPLE ARRIVAL

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 affec- ting 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.



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P/N	DESCRIPTION	TURNAROUND TIME
IBDGX001	iBio Single gene analysis	within 2 weeks
IBDGX002	iBio small panel (2-25 genes)	within 2 weeks
IBDGX003	iBio medium panel (26-125 genes)	within 2 weeks
IBDGX004	iBio large panel (from 125 genes)	within 2 weeks
IBDGX005	iBio whole exome	within 2 weeks
IBDGX006	iBio whole exome duo	within 3 weeks
IBDGX007	iBio whole exome trio	within 3 weeks
IBDGX008	iBio whole exome quad	within 3 weeks
IBDGX009	iBio additional family member (quad)	within 3 weeks
IBDGX010	iBio expand to exome	within 2 weeks
IBDGX011	iBio expand to exome duo	within 3 weeks
IBDGX012	iBio expand to exome trio	within 3 weeks
IBDGX013	iBio expand to exome quad	within 3 weeks
IBDGX014	iBio whole exome re-analysis	within 2 weeks
IBDGX015	iBio variant testing (1-10 variants)	within 3 weeks
IBDGXRTRN	Return of remaining samples	

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, deletions and CNVs.
- Variant classification and annotation (e.g. ClinVar, COSMIC, HGMD, dbSNP, Varsome)and generation of clinical report.

All our panels and whole exome analysis include the detection of single nucleotide variants (SNVs), insertions and deletions (indels) and copy number variations (CNVs) using high-quality next-generation sequencing technology and the proprietary bioinformac data analysis pipelines.