# SERVICES OFFERED BY THE LABORATORY

- DNA isolation from various biologic materials:
  - venous blood
  - biopsy material
  - amniotic fluid, etc.
- DNA qualitative and quantitative detection
- Sequencing:
  - direct DNA fragment sequence
  - microsatellite analysis/fragment length analysis
- Genotyping
  - analysis of both rare and frequent polymorphisms
- Design of synthetic oligonucleotides (primers)
- Determination of genetic markers characterizing various populations
- Genetic detection of individual's sex in variously collected samples
- Data statistical analysis of the acquired genetic data
- Individual research plan development for multiple genetic studies





IEGULDĪJUMS TAVĀ NĀKOTNĒ



Prospect was printed by ERDF Project 2010/0200/2DP/2.1.1.2.0/10/APIA/VIAA/006

### Rīga Stradiņš university

#### STUDY DIRECTIONS

- Study of various monogenic genetic disorders:
  - Cystic fibrosis (mucoviscidosis)
  - Hereditary hemochromatosis
  - Wilson's disease
  - Gilbert's syndrome
  - Alpha 1-antitrypsin deficiency, etc.
- Study of various multifactorial disorders:
  - Coronary heart disease
  - Cleft lip and palate, etc.
- Study of genes involved in pharmacogenetics:
  - Cytochrome P450 system encoding genes
  - NAT2 encoding gene, etc.
- Study of sex-related disorders

#### **CONTACTS**

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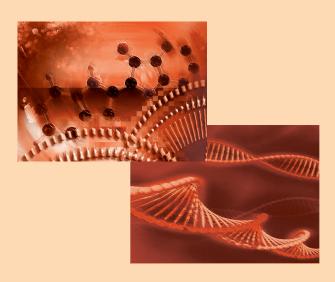


LABORATORY
OF MOLECULAR
GENETICS

#### ABOUT THE LABORATORY

Scientific Laboratory of Molecular Genetics of Rīga Stradiņš University has been registered in the Register of Medical Treatment Establishments of the Republic of Latvia (No. 019218301).

Laboratory participates in international external quality control schemes: CF Network (Cystic Fibrosis Network), RFB (Reference Institute for Bioanalytics), EMQN (European Molecular Genetics Quality Network), etc.



The laboratory has co-operation partners both in Latvia (Latvian Biomedical Research and Study Centre (Latvian Genome Database), Riga Eastern Clinical University Hospital, Children's Clinical University Hospital, etc.) and abroad (Institute of Molecular Genetics of Russian Academy of Sciences, Russia; Hospital of Lithuanian University of Health Sciences Kaunas Clinics, Lithuania; etc.).

#### **DNA DIAGNOSTICS**

## LABORATORY PERFORMS DNA DIAGNOSTICS OF THE FOLLOWING DISEASES\*

MALE INFERTILITY	
Y chromosome microdeletion	AZFa, AZFb, AZFc region deletions in Y chromosome
Congenital bilateral absence of vas deference (CBAVD)	F508del mutation and IVS8(T)5 variant in the <i>CFTR</i> gene

HEREDITARY LIVER DISEASES			
Wilson's disease	H1069Q mutation in the <i>ATP7B</i> gene		
	Selective screening of most frequent mutations in the <i>ATP7B</i> gene		
	Full sequencing of the <i>ATP7B</i> gene		
Hereditary hemochromatosis	C282Y, H63D mutations in the <i>HFE</i> gene		
Gilbert's syndrome	(TA) <sub>7</sub> mutation in the <i>UGT1A1</i> gene		
Alpha 1-antitripsyn deficiency	PIZ and PIS mutations in the SERPINA1 gene		

DEEP VEIN THROMBOSIS				
Determination of inherited thrombophilia mutation panel	R506Q mutation in the Factor V Leiden gene			
	A20210G mutation in the prothrombin encoding gene			
	C677T and A1298C mutations in the MTHFR gene			

<sup>\*</sup> within the scope of state-paid services, paid services, and contracts concluded



HEREDITARY METABOLIC DISEASES				
	F508del mutation in the <i>CFTR</i> gene			
Cystic fibrosis (mucoviscidosis)	Selective screening of most frequent mutations in the <i>CFTR</i> gene			
	Full sequencing of the CFTR gene			
Ornithine transcar- bamylase deficiency	Full sequencing of the <i>OTC</i> gene			
Gaucher's disease	N370S and L444P mutations in the <i>GBA</i> gene			

OTHER INHERITED DISEASES		
COPD	PIZ and PIS mutations in the SERPINA1 gene	
Arrhythmogenic right ventricular dysplasia	Full sequencing of the <i>PSK2</i> gene	
Lactose intolerance	-13910 C/T mutation in the <i>MCM6</i> gene	
Ahondroplasia	G380R mutation in the <i>FGFR3</i> gene	
Hypochondroplasia	N540K mutation in the FGFR3 gene	
Isolated growth hormone deficiency	Full sequencing of the <i>GH1</i> gene	