CYPRUS ORGANIZATION FOR THE PROMOTION OF QUALITY
CYPRUS ACCREDITATION BODY

ACCREDITATION CERTIFICATE no. *L061-3*

The Board of Governors
of the Cyprus Organization for the Promotion of Quality,
the National Accreditation Body,
in accordance with the Article 7 of the Law 156(I)/2002

GRANTS ACCREDITATION to

DEPARTMENTS/ LABORATORIES of
THE CYPRUS INSTITUTE OF NEUROLOGY AND GENETICS

in Nicosia

The Departments/Laboratories shown in annexes were assessed according to the Accreditation Criteria for Medical Laboratories, as defined in the Standard

CYS EN ISO 15189:2012

and were found technically competent to carry out the **Tests** included in the Scope of Accreditation which is described in the **Annexes** to this Certificate and is an **integrated part of it. The Scope of Accreditation** can change only after approval from the Cyprus Accreditation Body.

CYS-CYSAB is a signatory of the European co-operation for Accreditation Multilateral Agreement (EA-MLA) for accreditation in this field.

The current Accreditation Certificate, no. ***L061-3*, is issued on the 27th February 2024 and is valid from the 20th June 2022 until the 19th June 2026.**

Accreditation was awarded for the first time on 20th June 2014.

Antonis Ioannou
Director

Date: **27th February 2024**

This laboratory is accredited in accordance with the recognised International Standard ISO 15189:2012. This accreditation demonstrates technical competence for a defined scope and the operation of a laboratory quality management System (ISO-ILAC-IAF Communiqué, January 2015).

Annex

to the Accreditation Certificate no. L061-3 (BG)

SCOPE OF ACCREDITATION

for the

DEPARTMENT OF BIOCHEMICAL GENETICS (BG) OF THE CYPRUS INSTITUTE OF NEUROLOGY AND GENETICS

***Valid as from the 15th of December 2022 until the 19th June 2026**

Materials /Products	Types of examinations	Methods applied / Technical fields
Plasma	Acylcarnitine analysis	In-house method (generation acylcarnitine – butyl esters) on Tandem Mass Spectrometer/ Waters M05.09.BG.10
Blood sample	Determination of Lactate Levels	Enzymatic (Instruchemie Kit) on Thermo Scientific Spectrophotometer M05.09.BG.01
Plasma or Serum	Determination of total Homocysteine levels	Chemiluminescent Microparticle ImmunoAssay on Abbott ARCHITECT i1000SR system M05.09.BG.65
Urine sample	Organic acid analysis	In-house method (oxidation and extraction with ethyl acetate and diethyl ether) on Gas Chromatography-Mass Spectrometer/Agilent M05.09.BG.31
Urine sample	Quantitative determination of Glycosaminoglycans (GAGS)	In-house method (dimethylmethylene blue -DMB) on Thermo Scientific Spectrophotometer M05.09.BG.19
Plasma or Serum	Vitamins A&E	Chromsystems kit on HPLC/ Waters M05.09.BG.80
Plasma or Serum	Plasma/Serum Vitamin B12 measurement on the ArchitecTi system	Chemiluminescence Microparticle immunoassay (CMIA) using kit from Abbott diagnostics Architect i1000SR (Abbott) automated immunoassay analyser M05.09.BG.69

Plasma or Serum	Plasma/Serum Folate measurement on the ArchitecTi system	Chemiluminescence Microparticle immunoassay (CMIA) using kit from Abbott diagnostics Architect i1000SR (Abbott) automated immunoassay analyser M05.09.BG.70
Plasma	Ammonia	Spectrophotometric determination using kit from Instruchemie Thermo Scientific Spectrophotometer M05.09.BG.5
Urine sample	Urine GAGS (Gycosaminoglycans) electrophoresis	One dimensional electrophoresis Titan Gel Chamber EP (Hellena Biosciences) M05.09.BG.21
Plasma, Urine	Amino acid analysis (Alanine, Alloisoleucine, Arginine, Asparagine, Aspartate, Citrulline, Cystine, Glutamine, Glutamic Acid, Glycine, Histidine, Hydroxyproline*, Isoleucine, Leucine, Lysine, Methionine, Ornithine, Phenylalanine, Proline, Serine, Taurine, Threonine, Tryptophan, Tyrosine, Valine)	Ion-exchange chromatography ARACUS M05.09.BG.9

Note: The reference to trade names of the analyser/kit is related to a particular method and protocol

Authorised person to sign the test reports is Dr Petros Petrou
In his absence, Dr Theodoros Georgiou are authorized to sign reports.

Annex

to the Accreditation Certificate no. L061-3 (CG)

SCOPE OF ACCREDITATION

for the

DEPARTMENT OF CYTOGENETICS AND GENOMICS (CG) OF THE CYPRUS INSTITUTE OF NEUROLOGY AND GENETICS

***Valid as from the 15th December 2022 until the 19th June 2026**

Materials /Products	Types of examinations	Methods applied / Technical fields
Clinical Cytogenetics		
Chorionic Villus Sample (CVS), Amniotic Fluid, Peripheral and Fetal Blood, Skin Biopsy, Products of Conception	Detection of chromosome abnormalities (Test Codes 1,2,3,4,5,6, 8)	Cells culturing, harvest and karyotyping by G-banding analysis (Chromosome anaslysis – Qualitative)
Molecular Cytogenetics		
CVS, Amniotic Fluid, Peripheral and Fetal Blood, Skin Biopsy, Products of Conception	Identification/Confirmation/ Characterization of Chromosomal abnormalities by FISH (Test code 20) Analysis of Disease(s)/ Syndrome(s) by FISH (Test code 21)	Qualitative, Detection of location and number of signals by FISH (Fluorescent In Situ Hybridization) analysis with targeted DNA probes.
DNA Analysis		
CVS, Amniotic Fluid, Peripheral and Fetal Blood	Prenatal analysis of FMR1 gene (Fragile X Syndrome) (Test code 60) Postnatal analysis of FMR1 gene (Fragile X Syndrome)-per Individual (Test code 61)	Qualitative, Detection of tri-nucleotide repeats and methylation by PCR and Southern Blot analysis.
Peripheral Blood	Screening of Y(AZF) chromosomal microdeletions (Test code 65)	Qualitative, Detection of Y microdeletion by Multiple PCR analysis.
Peripheral Blood	Molecular analysis of Prader Willi/Angelman Syndrome genomic region 15q11-q13 (Test code 68)	Qualitative, Detection of methylation status, microdeletion of microduplication of the region by MS-MLPA analysis.
CVS, Amniotic Fluid, Peripheral and Fetal Blood	Rapid prenatal analysis of 13,18,21, X,Y aneuploides (Test code 69)	Semi-quantitative detection of aneuploidies by QF-PCR (Quantitative Fluorescent-PCR) analysis.
CVS, Amniotic Fluid, Peripheral and Fetal Blood, Skin Biopsy, Products of Conception	Detection of genomic imbalances with high resolution microarray-CGH (Test code 70)	Semi-quantitative detection of copy number changes by array-CGH (Comparative Genomic Hybridization) analysis.
Trophectoderm biopsy at blastocyst stage.	*Pre-implantation genetic testing (Test Codes 71) - Detection of copy number changes and aneuploideies.	Manufacturer's manual (Illumina Inc.) Next Generation Sequencing (NGS)

DNA from Peripheral Blood	* 1. Targeted panel Analysis-Rett Angelman panel (Test code 110) 2. Clinical Exome Sequencing (CES) for single patient analysis and trio analysis (Test codes111, 112, 113, 114 & 117) 3.Whole Exome Sequencing (WES) for single patient analysis and trio analysis (Test codes 115, 116 & 118) 4. Sanger sequencing for confirmation of variants detected by NGS (Test code 119)	For 1, 2, and 3: Next generation Sequencing For 4: PCR followed by Sanger Sequencing. NextSeq 2000

Authorised persons to sign the test reports are Dr Carolina Sismani, Dr Paola Evangelidou.
In the absence of the above, Ms Nicole Salameh and Mr Angelos Alexandrou are authorized to sign reports for molecular analysis or Ms Nicole Salameh and the analyst are authorized to sign reports for cytogenetic analysis.

SCOPE OF ACCREDITATION

for the

NEUROGENETICS DEPARTMENT (ND) OF THE CYPRUS INSTITUTE OF NEUROLOGY AND GENETICS

***Valid as from the 15th December 2022 until the 19th June 2026.**
****Valid as from the 10th May 2023 until the 19th June 2026.**

Materials /Products	Types of examinations	Methods applied / Technical fields
Molecular Diagnostic Tests		
Peripheral Blood	DNA extraction from Peripheral Blood (Test code 10)	In house salting out procedure using the Gentra Puregene Blood Kit of Qiagen
Extracted DNA from peripheral blood	DEL/DUP test of *PMP22*, *SMN1* and **DMD* genes (Test codes 6.05, 7.01 and 71).	MLPA CE-IVD (Multiple Ligation-Dependent Probe Amplification, using SALSA MLPA KIT -P033, -P021 and -*P034-B2 DMD-1 & -*P035-B1 DMD-2 respectively) and using the ABI *3500xl*/*3130xl* Genetic Analyser.
Extracted DNA from peripheral blood	Repeat analysis of *ATXN1, ATXN2, ATXN3, CACNA1A, ATXN7* and *HTT* genes (Test codes 4, 12, 5, 13, 14, 15 and 2).	In house fragment analysis using the ABI *3500xl*/*3130xl* Genetic Analyser.
Extracted DNA from peripheral blood	Repeat analysis of *FXN* gene (Test codes 3 and 3.01)	In house long-PCR analysis, sequence analysis and TP-PCR analysis using the ABI *3500xl/3130xl* Genetic Analyser.
Extracted DNA from peripheral blood	Molecular analysis of sequence variations in the *MPZ, CX32/GJB1* and *PMP22, TTR, MFN2, NEFL, GDAP1, GARS, EGR2, LMNA, APTX, SOD1, BSCL2, TDP43, LRRK2, GJC2, SPAST, ATL1, REEP1* and *FUS* (Test codes 6.02, 6.03, 6.04, 1.01, 6.06, 6.07, 6.08, 6.09, 6.10, 6.11 and 34, 20, 23, 24, 25, 26, 27, 28, 29, 31 and 32). Detection of *TTR* gene Val30Met variant (Test Code 1).	In house Sequencing analysis using the ABI *3500xl/3130xl* Genetic Analyser. Standard established procedure, manufacturer's manual (ABI).
Extracted DNA from peripheral blood	*Detection of variations in the multiple genes. WES: [Test codes *40.2, **(41, 42, 43, 44, 45, 46, 47, 48, 49, 50, 51, 52, 53, 54, 55, 56, 57, 58, 59, 60, 61, 64, 65, 67 and 69)].	Next Generation sequencing using Illumina Nextseq 550/2000.

Authorised persons to sign the test reports are:

1st signature (permanent)	2nd signature	3rd signature
K. Christodoulou	**A. Georghiou or P. Nikolaou or C. Votsi***	**L. Koutsou or P. Nikolaou or C. Votsi* (if not already signed at the second column) or none**

Annex

to the Accreditation Certificate no. L061-3 (CGTUP)

SCOPE OF ACCREDITATION

for the

DEPARTMENT OF CANCER GENETICS, THERAPEUTICS & ULTRASTRUCTURAL PATHOLOGY (CGTUP) OF THE CYPRUS INSTITUTE OF NEUROLOGY AND GENETICS

Materials/ Products	Types of Tests	Methods applied/ Technical fields
Peripheral Blood	Screening for Mutation detection in the: *BRCA1 (*10), *BRCA2* (12), *TP53* (14), *APC* (16), *MLH1* (19), *MSH2* (21), *PMS1* (23), *PMS2* (25), *MSH6* (27), *KRAS* (29), *PTEN* (41), *STK11* (43), *CHEK2* (45), *ATM* (47), *PALB2* (49), *BRIP1* (51), *CDKN2A* (53), *BRAF* (55), *EGFR* (61), *CKIT* (64), *PDFGRA* (67), *CDH1* (70), *MYH* (72) and *VHL* (74) genes.	In house PCR/Sanger sequencing method and sequence analysis using the ABI *3130xl* Genetic Analyser.
Peripheral Blood and various types of tissues	Detection of the presence/absence of known mutation(s) in the: *BRCA1* (11), *BRCA2* (13), *TP53* (15), *APC* (17), *MLH1* (20), *MSH2* (22), *PMS1* (24), *PMS2* (26), *MSH6* (28), *PTEN* (42), *STK11* (44), *CHEK2* (46), *ATM* (48), *PALB2* (50), *BRIP1* (52), *CDKN2A* (54), *BRAF* (56), *CKIT* (66), *PDFGRA* (69), *CDH1* (71), *MYH* (73) and *VHL* (75) genes. Screening for mutations in exons 18-21 of the *EGFR* gene (62). Screening for mutations in exons 2-4 of the *KRAS* gene (29.1). Screening for mutations in exons 2-4 of the *KRAS* and *NRAS* genes (29.2). Screening for mutations in exons 9, 11, 13 and 17 of the *C-KIT* gene (65). Screening for mutations in exons 12 and 18 of the *PDFGRA* gene (68).	In house PCR/Sanger sequencing method and sequence analysis using the ABI *3130xl* Genetic Analyser or MLPA (Multiple Ligation-Dependent Probe Amplification) using the ABI *3130xl* Genetic Analyser following the manufacturer's protocol (MRC Holland).
Peripheral Blood	Mutation analysis for mutation *CFHR5* internal duplication of exons 2 and 3 (63).	In house PCR
Peripheral Blood	Detection of large rearrangements (duplication(s) or deletion(s) of whole exon(s)) in the *BRCA1* (10, 11), *BRCA2* (12, 13), *MLH1* (19, 20), *MSH2* (21, 22), and *APC* (16, 17), *VHL (74, 75) genes.	MLPA (Multiple Ligation-Dependent Probe Amplification) following the manufacturer's protocol (MRC Holland) and using the ABI *3130xl* Genetic Analyser.
Peripheral Blood and various types of tissues	DNA extraction and storage (Test codes 6 and 7)	In house DNA extraction method using phenol/ chloroform or QIAamp DNA FFPE Tissue Kit (Qiagen) (DNA)

Peripheral Blood and various types of tissues	Detection of Microsatellite instability (MSI) (Test code 18)	In house fluorescent multiplex PCR based method using five mononucleotide repeat markers: BAT25, BAT26, NR21, NR22 and NR24. Amplified fragments are detected using the ABI *3130xl.*
Various types of tissues (Muscle, Kidney, Nerve, Cilia, Liver)	Processing and examination of specimens in the Transmission Electron Microscope used for diagnostic histopathology (Test code 1)	Descriptive report with photographs using JEOL JEM 1010 TEM or *THERMO SCIENTIFIC TALOS L120C
Various types of tissues (Muscle, Kidney, Nerve, Cilia, Liver)	Examination of specimens in the Transmission Electron Microscope used for diagnostic histopathology (Test code 2)	Descriptive report with photographs using JEOL JEM 1010 TEM or *THERMO SCIENTIFIC TALOS L120C
DNA (germline/ somatic)	Mutation detection in BRCA1 and BRCA2 genes Qualitative DNA analyses and evaluation of gene variants (Test code 81)	Multiplex PCR with BRCA MASTR Dx CE-IVD kit (Multiplicom/Agilent) and Next generation sequencing (NGS) with MiSeq Dx (Illumina)
DNA (germline)	Mutation detection in a cancer gene panel Qualitative DNA analyses and evaluation of gene variants (BRCA1, BRCA2, PALB2, CHEK2, BARD1, BRIP1, RAD51C, RAD51D, TP53, MRE11A, RAD50, NBN, FAM175A, ATM, STK11, MEN1, PTEN, CDH1, MUTYH, BLM, XRCC2, MLH1, MSH6, PMS2, MSH2, 3'UTR of EPCAM) (Test Codes 84, 85, 86)	Multiplex PCR with CleanPlex Hereditary Cancer Panel (Paragon Genomics) and Next Generation Sequencing (NGS) with MiSeq (Illumina)

Authorised persons to sign the reports for molecular pathology are Dr. Mihalis Panagiotidis and Dr. Andreas Hadjisavvas. In the absence of any of the aforementioned persons, Dr. Maria Loizidou signs (p.p.).
Authorised persons to sign the reports for electron microscopy are Dr. Mihalis Panagiotidis and Ms Louiza Potamiti. In the absence of any of the abovementioned persons, Dr. Andreas Hadjisavvas signs (p.p.).

Annex

to the Accreditation Certificate no. L061-3 (MV)

SCOPE OF ACCREDITATION

for the

DEPARTMENT OF MOLECULAR VIROLOGY (MV) OF THE CYPRUS INSTITUTE OF NEUROLOGY AND GENETICS

Valid as from the 20th of June 2022 until the 19th of June 2026.
*** Valid as from the 27th of February 2024 until the 19th of June 2026.**

Materials /Products	Types of examinations	Methods applied / Technical fields
Serum, EDTA-blood, CSF, biopsies, urine, stool, nasal, nasopharyngeal or tracheal swabs, BAL, Cervical/vulvar wash, Amniotic Fluid	DNA and RNA Extraction	MagPurix system (ZinexTS) KingFisher Flex
	Virus serology	
	Presence of IgG Antibodies against CMV (Test code 3)	CE-IVD certified ELISA Kit, VIDIA
	Presence of IgM Antibodies against CMV (Test code 4)	CE-IVD certified ELISA Kit, VIDIA
	Presence of IgG Antibodies against EBV NA (nuclear antigen) (Test code 11)	CE-IVD certified ELISA Kit, Euroimmun
	Presence of IgG Antibodies against EBV EA (early antigen) (Test code 11.1)	CE-IVD certified ELISA Kit, Euroimmun
	Presence of IgM Antibodies against EBV (Test code 12)	CE-IVD certified ELISA Kit, Euroimmun
	Level and avidity of IgG and level of IgM Antibodies against Toxoplasma gondii (Test code 17, 17.1, 18)	CE-IVD certified ELISA Kit, NovaTec
	Qualitative Virus DNA detection	
Whole blood (serum), EDTA-blood, Liquor, Biopsies, urine, stool, nasal or tracheal swabs, BAL	Analysis of the presence of viral DNA from VZV, Parvovirus, BK-Virus, Adenovirus (Test codes 47, 71, 76, 63)	In house Real-Time PCR assay on ABI7500A or Quantstudio 5 A & C
Blood, CSF, Urine, Amniotic Fluid	Presence of Toxoplasma gondii DNA (Test code 78)	In house Real-Time qPCR on ABI7500A or Quantstudio 5 A & C
	Quantitative Virus DNA detection	
Whole blood (serum), EDTA-blood, Liquor, Biopsies, urine, stool, nasal or tracheal swabs, BAL, Cervical/vulvar wash	Analysis of the quantity of viral DNA from HSV-1, HSV-2, CMV and EBV (Test codes 44, 45, 41, 43, 46)	In house q-PCR assay on ABI7500A or Quantstudio 5 A & C

(last matrix only for Test Codes 44, 45, 41)		
Qualitative Virus RNA detection		
Whole blood (serum), EDTA-blood, Liquor, Biopsies, urine, stool, nasal or tracheal swabs, BAL, Cervical/vulvar wash	Presence of viral RNA from Enteroviruses and Influenza H1N1(Test codes 48, 77)	In house Real-Time RT-PCR assay on ABI7500A or Quantstudio 5 A & C
Serum, nasal, nasopharyngeal or tracheal swabs, BAL, CSF	Presence of viral RNA from Influenza A, Influenza B and RSV (Test codes 100, 101, 102)	In house Multiplex Real-Time RT-PCR assay on on ABI7500A or Quantstudio 5 A & C
Serum; nasal, nasopharyngeal and tracheal swabs; BAL	Presence of SARS-CoV-2 RNA (Test code 120.1)	In-house RT-qPCR on Quantstudio 5 A to E
	Presence of SARS-CoV-2 RNA (Test code 120.2)	CE-IVD Thermofisher Taqpath RT-qPCR on Quantstudio 5 A to E
Virus Genotyping		
Cervical/vulvar wash, swabs	Presence of and Typing of Human Papillomavirus (Test code 49.1)	In house PCR assay/ Restriction profile analysis, Southern Blot
Cervicovaginal/penile smear wash /swab samples	Detection of 37 Human Papilloma (HPV) Virus types (Test code 49.2)	*Opegen High + Low Papilloma Strip IVD kit (Operon, Spain)

Note: The reference to trade names of the analyser/kit is related to a particular method and protocol

Authorised person to sign the test reports is Dr Christina Christodoulou.
In her absence the scientist Dana Koptides or Jan Richter are authorized to sign reports.

DEPARTMENT OF MOLECULAR GENETICS
THALASSAEMIA (MGT) OF
THE CYPRUS INSTITUTE OF NEUROLOGY AND GENETICS

Valid as from the 20th of June 2022 until the 19th of June 2026
*** Valid as from the 15th of December 2022 until the 19th June 2026**
**** Valid as from the 11th of May 2023 until the 19th June 2026**

Materials /Products	Types of examinations	Methods applied / Technical fields
Peripheral and Cord blood	DNA extraction from Peripheral blood, Cord blood.	– M 05.09.MGT.01-Puregene Blood Core Kit C-Qiagen
Chorionic Villus Sample (CVS)	Microscopic cleaning of CVS tissue. DNA extraction from CVS tissue.	– M 05.09.MGT.02-Nucleospin Tissue Kit-Macherey-Nagel
Maternal plasma	DNA extraction from maternal plasma.	– M 05.09.MGT.25/26-QIAmp Circulating Nucleic acid Kit-Qiagen
Peripheral Blood	DNA extraction from Peripheral blood	– MagCore (HF16) nucleic acid extractor*
DNA (peripheral blood, cord blood, CVS)	• Molecular Analysis of α-thalassaemia. • Molecular Analysis for Prenatal diagnosis (PND) for α-thalassaemia.	– M 05.09.MGT.10-GAP-PCR_a- thalassaemia deletions: --MEDII. – M 05.09.MGT.11-GAP-PCR_a- thalassaemia deletions: -α3.7, ααα. – M 05.09.MGT.12-GAP-PCR_a- thalassaemia deletions: --MEDI α20 5. – M 05.09.MGT.13-GAP-PCR_a- thalassaemia deletions: --SEA, --THAI, --FIL. – M 05.09.MGT.14-GAP-PCR_a- thalassaemia deletions: -a4 2. – M 05.09.MGT.30- Multiplex GAP PCR_-α3.7, ααα, --MEDI, -α20.5. – M 05.09.MGT.16-RED-PCR_a-thalassaemia point mutations: aPolyA1a. – M 05.09.MGT.17-RED-PCR_a-thalassaemia point mutations: aPolyA2a. – M 05.09.MGT.18-RED-PCR_a-thalassaemia point mutations: -α5ntα, αAgrα. – M 05.09.MGT.19-RED-PCR_a-thalassaemia point mutations: Initiation Codon, αIcariaα. – M 05.09.MGT.22-Sanger Sequencing_ a2 and a1-globin genes. ABI3130xl ABI3500xl Genetic Analyzers – M 05.09.MGT.36-MLPA-HBA_Fragment Analysis. ABI3130xl ABI3500xl Genetic Analyzers Thermocycler: PCR1(#3386) PCR2(#3087) PCR3(#4254) PCR5(#2633)

Materials /Products	Types of examinations	Methods applied / Technical fields
DNA (peripheral blood, cord blood, CVS)	Molecular Analysis of β-thalassaemia.	– M 05.09.MGT.03-ARMS_β-thalassaemia point mutations: -IVSI-110. – M 05.09.MGT.04-ARMS_β-thalassaemia point mutations: - IVSI-6. – M 05.09.MGT.05-ARMS_β-thalassaemia point mutations: -IVSI-1. – M 05.09.MGT.06-ARMS_β-thalassaemia point mutations: -IVSII-745. – M 05.09.MGT.07-ARMS_β-thalassaemia point mutations: -C-39. – M 05.09.MGT.08-ARMS_β-thalassaemia point mutations: - 87. – M 05.09.MGT.09-ARMS_β-thalassaemia point mutations: -IVSII-1. – M 05.09.MGT.15-GAP PCR-Hb Lepore_β-thalassaemia deletions. – M 05.09.MGT.23-Sanger Sequencing_β-globin genes. ABI3130xl, ABI3500xl Genetic Analyzers – M 05.09.MGT.36-MLPA-HBB_Fragment Analysis. ABI3130xl ABI3500xl Genetic Analyzers – *M 05.09.MGT.41-Minisequencing Assay for the Detection of 7 HBB Mutations [IVSI-110 G>A, IVSI-I G>A, IVSI-6 T>C, IVSII-745 C>G, cd39 C>T, -87 C>G, and cd6 GAG>GTG (HbS)] based on the SNaPshot minisequencing approach. ABI3130xl ABI3500xl Genetic Analyzers – M 05.09.MGT.32-HBB-STR's Fragment analysis. ABI3130xl ABI3500xl Genetic Analyzers Thermocycler: PCR1(#3386) PCR2(#3087) PCR3(#4254) PCR5(#2633)
		– M 05.09.MGT.33-Real-Time PCR assay using Taqman chemistry for the presence or absence of the known mutation, IVSI-110, ABI 7900HT Fast Real-Time PCR system – M 05.09.MGT.34-Real-Time PCR assay using Taqman chemistry for the presence or absence of the known mutation, IVSI-1, ABI 7900HT Fast Real-Time PCR system – M 05.09.MGT.35-Real-Time PCR assay using Taqman chemistry for the presence or absence of the known mutation, IVSI-6, ABI 7900HT Fast Real-Time PCR system
DNA (Chorionic Villus Sample, CVS and peripheral blood)	Molecular Analysis for Prenatal diagnosis (PND) for β-thalassaemia.	– M 05.09.MGT.32-HBB-STR's Fragment analysis. Thermocycler: PCR1(#3386) PCR2(#3087) PCR3(#4254) PCR5(#2633), ABI3130xl ABI3500xl Genetic Analyzers All the methods stated in the "Molecular Analysis of β-thalassaemia"
Single Blastomere biopsies, 5-10 cell Trophectoderm biopsies	Pre-Implantation Genetic Diagnosis (PGD) for β-thalassaemia	– M 05.09.MGT.37-Protocol_PGD HBB_multiplex PCR – M 05.09.MGT.32-HBB-STR's Fragment analysis All the methods stated in the "Molecular Analysis of β-thalassaemia"
DNA (peripheral blood-maternal plasma)	Non-invasive prenatal determination of fetal Rhesus	– M 05.09.MGT.25-Real-Time PCR assay using Taqman chemistry for RHD genotyping

Materials /Products	Types of examinations	Methods applied / Technical fields
	Non-invasive prenatal determination of fetal sex	– M 05.09.MGT.26-Real-Time PCR assay using Taqman chemistry for fetal sex genotyping
Genomic DNA from Whole Blood	• Detection of mutations related to pathogenic conditions of the whole human exome • Detection of mutations related to pathogenic conditions of the whole human exome (Trio analysis) • Molecular diagnosis of non-malignant haematological diseases, including iron metabolism and heme synthesis disorders (Haem-NGS) • Molecular diagnosis of Hepatological diseases (Hep-NGS) • Molecular diagnosis of Bone diseases (Ost-NGS) • Molecular diagnosis of Oxidative stress (OXIS-NGS)	– M 05.09.MGT.43_NGS-WES/CES, Exome Library Preparation, Data Analysis Illumina® DNA Prep with Exome 2.0 Plus Enrichment **, Reference Guide v03
Genomic DNA from Whole Blood	Diagnosis of Thalassaemia	– M 05.09.MGT.42- Devyser® Thalassemia NGS, Molecular Diagnosis for Thalassaemia with NGS*

Authorised persons to sign the test reports are:
-For Prenatal Diagnosis for thalassaemia: **Carsten Lederer and Thessalia Papasavva. 3rd signature: Miranda Petrou or George Christopoulos or Xenia Feleki.
-Diagnostic reports for thalassaemia: **Carsten Lederer and Thessalia Papasavva. 3rd signature: Miranda Petrou or George Christopoulos or Xenia Feleki.
-For non-invasive prenatal diagnosis: **Carsten Lederer and Thessalia Papasavva. 3rd signature: George Christopoulos or Xenia Feleki
-For PGD: **Carsten Lederer and George Christopoulos or Thessalia Papasavva
-For NGS:
M 05.09.MGT.42- Devyser® Thalassemia NGS, Molecular Diagnosis for Thalassaemia with NGS: **Carsten Lederer, Thessalia Papasavva, Miranda Petrou, Xenia Feleki
M 05.09.MGT.43_NGS-WES/CES, Exome Library Preparation, Data Analysis: **Carsten Lederer, Thessalia Papasavva

Annex

to the Accreditation Certificate no. L061-3 (MGFT)

SCOPE OF ACCREDITATION

for the

DEPARTMENT OF MOLECULAR GENETICS FUNCTION AND THERAPY (MGFT) OF THE CYPRUS INSTITUTE OF NEUROLOGY AND GENETICS

***Valid as from the 15th December 2022 until the 19th June 2026.**
****Valid as from the 31st May 2023 until the 19th June 2026.**

Materials /Products	Types of examinations	Methods applied / Technical fields
Peripheral Blood, CVS, amniotic fluid	**Full mutation and copy number variation (CNV) analysis of the *CFTR* (Cystic Fibrosis Transmembrane Conductance Regulator) gene. (Test code 1.1)	Genetic screening by Next Generation Sequencing (NGS) using the Devyser CFTR kit in the Illumina MiSeq system.
Peripheral Blood	Screening of mutations in exons 2, 3, 5, 10 of the *MEFV* (Mediterranean Fever) gene (Test Code 7.1)	In house DNA Sequencing method in the genetic analyzer ABI 3130 XL and *ABI 3500xl*.
Peripheral Blood	Screening of mutations in exons 10, 11, 13, 14, 15, 16 of the *RET* proto-oncogene (Test Code 11.1)	In house DNA Sequencing method in the genetic analyzer ABI 3130 XL and *ABI 3500xl*.
Peripheral Blood	1. Screening of mutations in exon 2 of *GJB2* gene (Test Code 13.1) 2. Detection of D13S1830 mutation in the *GJB6* gene (Test Code 15.1)	1. In house DNA Sequencing method in the genetic analyzer ABI 3130 XL and *ABI 3500xl*. 2. In-house PCR method.
Peripheral Blood	Screening of point mutations and large deletions or other rearrangements in the *CYP21A2* gene (Test Code 16.1)	In house DNA Sequencing method in the genetic analyzer ABI 3130 XL and *ABI 3500xl*.and Manufacturer's kit for MLPA method (MRC Holland)
Peripheral Blood	Detection of H63D and C282Y mutations in the *HFE* (*H*igh Iron *Fe*) gene (Test Code 10.7)	In house PCR-RLFP method
Peripheral Blood	Screening of mutations in SRD5A2 gene for 5-alpha Reductase Deficiency (Test Code 21.1)	In house DNA Sequencing method in the genetic analyzer ABI 3130 XL and *ABI 3500xl*.
Peripheral Blood, cerebrospinal fluid (CSF)	*Detection of oligoclonal immunoglobulin G (Test code 14)	Manufacturer's kit for Isoelectric focusing on agarose gel using the Sebia HYDRASYS 2 Scan & Focusing system.

Materials /Products	Types of examinations	Methods applied / Technical fields
Peripheral Blood	** 1. Detection of mutations related to pathogenic conditions of the whole human exome (Test code 43) 2. Detection of mutations related to pathogenic conditions of the whole human exome (Trio analysis) (Test code 44) 3. Detection of mutations related to characterized pathogenic conditions (Test code 41) 4. Detection of mutations related to characterized pathogenic conditions (Trio analysis) (Test code 42) 5. Detection of mutations related to Hereditary Recurrent Fevers (HRFs) (Test code 24.3) 6. Detection of mutations related to Hearing Loss (Test code 15.6) 7. Detection of mutations related to Disorders of Sexual Development (Test code 45) 8. Detection of mutations related to Premature and Delayed Puberty (Test code 38.4) 9. Detection of mutations related to Glucose and Insulin Homeostasis (MODY and OBESITY) (Test code 20.7) 10. Detection of mutations related to Thyroid Function (Test code 39.1)	Exome Sequencing by Next Generation Sequencing (NGS) using the Illumina DNA Prep with Enrichment kit and analysed in the Illumina Nextseq500 and Nextseq2000 systems.

Authorised persons to sign the test reports are Leonidas Phylactou and Vassos Neocleous.

***Valid as from the 15th December 2022 until the 19th June 2026.**

***Primary Sample Collection**		
Materials /Products	**Types of examinations**	**Methods applied / Technical fields**
Blood, Urine	1. Primary Sample collection and handling 2. Sample reception 3. Patient registration/Request Form 4. Sample transportation 5. Result transmission	1. I05.04.04.BG, I05.04.04.CG, I05.04.04.CGTUP, I05.04.04.MGFT, I05.04.04.MGT, I05.04.04.MV, I05.04.04.ND, I05.04.09 2. I05.04.01, I05.04.02.BG, I05.04.02.CG, I05.04.02.CGTUP, I05.04.02.MGFT, I05.04.02.MGT, I05.04.02.MV, I05.04.02.ND 3. SOP05.04, F05.04.01.BG, F05.04.01.CG, F05.04.01.CGTUP, F05.04.01.MGFT, F05.04.01.MGT, F05.04.01.MV, F05.04.01.ND 4. F05.04.01.BG, F05.04.01.CG, F05.04.01.CGTUP, F05.04.01.MGFT, F05.04.01.MGT, F05.04.01.MV, F05.04.01.ND 5. I05.08.01.BG, I05.08.01.CG, I05.08.01.CGTUP, I05.08.01.MGFT, I05.08.01.MGT, I05.08.01.MV, I05.08.01.ND

General Remarks

These Annexes refer **only to tests** carried out **in the premises of the Laboratory,** Address: 6 Iroon Avenue, 2371 Nicosia.

Antonis Ioannou
Director

Date: **27th February 2024**