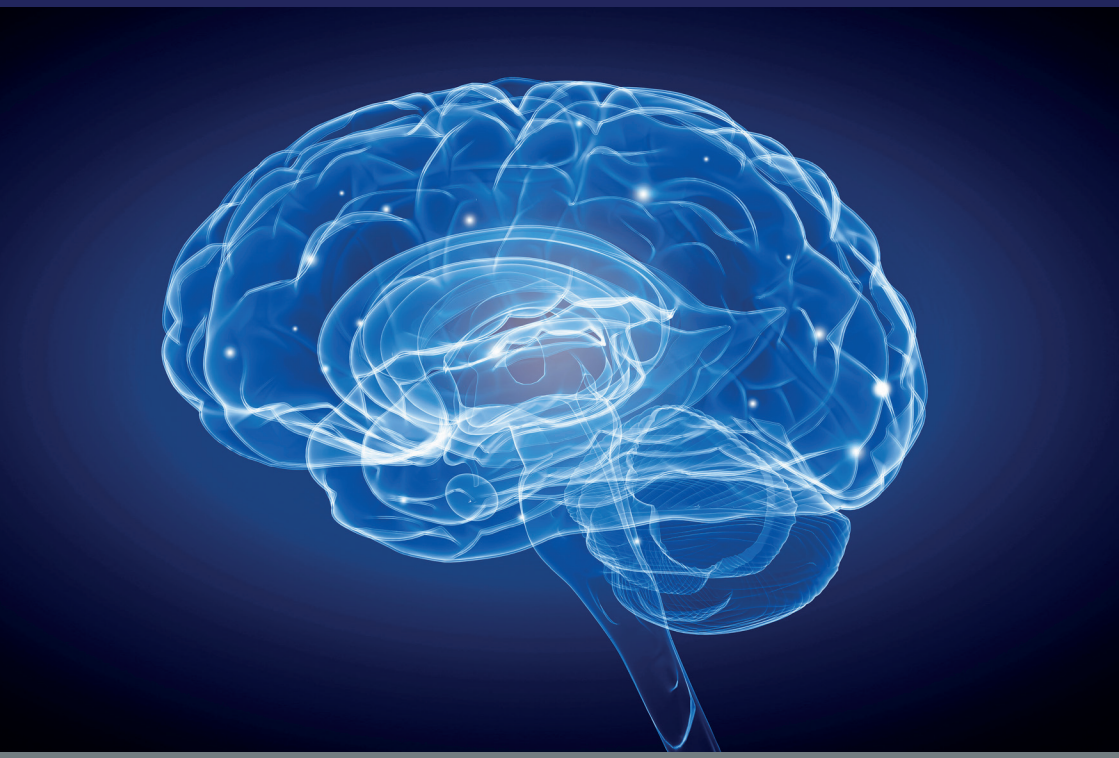
NEUROLOGY TEST CATALOG



MOLECULAR GENETIC TESTS | MOLECULAR CYTOGENETIC TESTS | NEXT GENERATION SEQUANCING PANELS

MEDIPOL UNV
ISTANBUL MEDIPOL UNIVERSITY

CEQAS Cytogenomic External Quality Assessment Service | EMQN The European Molecular Genetics Quality Network | QCMD Quality Control for Molecular Diagnostics | CAP ACCREDITED COLLEGE of AMERICAN PATHOLOGISTS

ISTANBUL MEDIPOL UNIVERSITY
CENTER for GENETIC DIAGNOSIS
'Single Address for All Genetic Tests'

İMU Center for Genetic Diagnosis:

EMQN The European Molecular Genetics Quality Network

CEQAS Cytogenetic External Quality Assessment Service

CAP ACCREDITED COLLEGE of AMERICAN PATHOLOGISTS

QCMD Quality Control for Molecular Diagnostics

- Next Generation Sequencing
- Conventional Cytogenetics
- PCR
- Array CGH
- Repeat Expansion Analysis
- Molecular Microbiology
- Deletion & Duplication Analysis (MLPA)
- Non-Invasive Prenatal Screening
- Geographic Origin Determination
- First Generation (Sanger) Sequencing
- Molecular Cytogenetics
- Real-Time PCR
- Gene-Chip
- Prenatal Analysis
- HI-SKY Karyotyping
- Aneuploidy testing with FISH
- Clinical Exome
- Liquid Biopsy

İMU Center for Genetic Diagnosis:

Our diagnostic center is located at Istanbul Medipol University Kavacık Campus within REMER (Research Center for Regenerative and Restorative Medicine; www.remer.medipol.edu.tr), a newly built 100 Million-USD world class biomedical research facility. It provides access to state-of-the-art laboratory accommodation with the latest top-of-the-range technology and equipment. There is a stimulating mix of internationally established clinical and basic scientists investigating human physiology, pathogenesis of diseases and novel therapeutic approaches using a wide range of methodologies. Core facilities include:

- Cell Culture and Sorter Laboratories
- Molecular Biology and Histology
- Full Animal House
- Department of Proteomics, Genomics and Bioinformatics
- Advanced Microscopy, Electrophysiology and Optogenetics Laboratories
- Dental Research and Biomechanical Laboratories
- Pharmaceutical Chemistry and Pharmacognosy Laboratories
- Neuroscience, Microsurgery and Analytical Chemistry Units

IMU Center for Genetic Diagnosis serves for screening, diagnosis, prevention, genetic counseling and follow-up of single gene and chromosomal disorders, multifactorial diseases, teratogenic disorders and congenital malformations. In addition invasive/non-invasive prenatal and pre-implantation genetic diagnosis and tests for personalized medicine are performed in our center.

Departments in our Center:

- Molecular Genetics
- Cytogenetics
- Molecular Cytogenetics
- Molecular Infections
- Genetic Counseling

Membership to International Quality Assurance Programs:

IMU Center for Genetic Diagnosis gives a special importance to top quality service. In this framework, we are a member of international quality control programs in the fields of Cytogenetics and Molecular Genetics.

Detailed Analyses with Latest Technology:

Next Generation Sequencing:

- Whole Genome
- Whole Exome Sequencing
- Fixed Panels (Ready to Use Panels)
- Custom Panel
- Chip Sequencing
- Methylation Sequencing
- Metagenomics (Shotgun Sequencing or 16S-18S Amplicon Sequencing)
- Whole Transcriptome (RNA-seq)
- mRNA gene expression
- Targeted RNA gene expression
- Small RNA profiling

Sanger Sequencing:

- De Novo Sequencing
- SNP genotyping analysis
- Fragment analysis

Real Time PCR Studies:

- SNP Detection-Polymorphism
- Gene expression quantitation
- Viral quantitation
- Detection of pathogens
- Genotyping-melting-curve analysis
- Detection of DNA damage (microsatellite instability)
- Methylation detection
- Minimal residual disease (MRD) monitoring
- Monitoring of chimerism after BMT
- Prenatal diagnosis in single cell isolated from mother's blood
- Prenatal diagnosis of hemoglobinopathies

NEUROLOGY TEST LIST

MOLECULAR GENETIC TESTS

Test Name	Gene Name	Test Methods	Sample
Alexander Disease	GFAP	Alexander Disease	Blood^{EDTA}
Alpers Huttenlocher Syndrome	POLG	Alpers Huttenlocher Syndrome	Blood^{EDTA}
ALS Type 1	ANG	ALS Type 1	Blood^{EDTA}
ALS Type 1	FUS	ALS Type 1	Blood^{EDTA}
ALS Type 1	SOD1	ALS Type 1	Blood^{EDTA}
ALS Type 1	TARDBP	ALS Type 1	Blood^{EDTA}
ALS Type 4	SETX	ALS Type 4	Blood^{EDTA}
ALS Type 4	SETX	ALS Type 4	Blood^{EDTA}
Alstrom Syndrome	ALMS1 (Exon 8,10,16)	Alstrom Syndrome	Blood^{EDTA}
Alstrom Syndrome	ALMS1	Alstrom Syndrome	Blood^{EDTA}
Alzheimer's Disease Type 1	APP (Exon 16,17)	Alzheimer's Disease Type 1	Blood^{EDTA}
Alzheimer's Disease Type 1	APP	Alzheimer's Disease Type 1	Blood^{EDTA}
Alzheimer's Disease Type 2	APOE (E2,E3,E4)	Alzheimer's Disease Type 2	Blood^{EDTA}
Alzheimer's Disease Type 3	PSEN1	Alzheimer's Disease Type 3	Blood^{EDTA}
Alzheimer's Disease Type 4	PSEN2	Alzheimer's Disease Type 4	Blood^{EDTA}
Amyloidosis	TTR (2 Mutations)	Amyloidosis	Blood^{EDTA}
Amyloidosis	TTR	Amyloidosis	Blood^{EDTA}
Amyotrophic Lateral Sclerosis	ANG	Amyotrophic Lateral Sclerosis	Blood^{EDTA}
Amyotrophic Lateral Sclerosis	FUS	Amyotrophic Lateral Sclerosis	Blood^{EDTA}
Amyotrophic Lateral Sclerosis	SOD1	Amyotrophic Lateral Sclerosis	Blood^{EDTA}
Amyotrophic Lateral Sclerosis	TARDBP	Amyotrophic Lateral Sclerosis	Blood^{EDTA}
Angelman Syndrome	UBE3A	Angelman Syndrome	Blood^{EDTA}
Angelman Syndrome	UBE3A	Angelman Syndrome	Blood^{EDTA}
Angelman Syndrome	UBE3A	Angelman Syndrome	Blood^{EDTA}
ARX	ARX	ARX	Blood^{EDTA}
Ataxia Telangiectasia Disease	ATM	Ataxia Telangiectasia Disease	Blood^{EDTA}
Ataxia With Vitamin E Deficiency	TTPA	Ataxia With Vitamin E Deficiency	Blood^{EDTA}
Atypical Hemolytic Uremic Syndrome Type 6	THBD	Atypical Hemolytic Uremic Syndrome Type 6	Blood^{EDTA}
Bilateral Frontoparietal Polymicrogyria	GPR56	Bilateral Frontoparietal Polymicrogyria	Blood^{EDTA}
Cadasil	NOTCH3 (Exon 3,6)	Cadasil	Blood^{EDTA}
Cardiofaciocutaneous Syndrome	BRAF (EXON 6,11-16)	Cardiofaciocutaneous Syndrome	Blood^{EDTA}

NEUROLOGY TEST LIST

MOLECULAR GENETIC TESTS

Test Name	Gene Name	Test Methods	Sample
Cardiofaciocutaneous Syndrome	BRAF	Whole Gene Analysis	Blood^{EDTA}
Cardiofaciocutaneous Syndrome	KRAS	Whole Gene Analysis	Blood^{EDTA}
Cardiofaciocutaneous Syndrome	MAP2K1 & MAP2K2 (Exon 2,3)	Sequence Analysis	Blood^{EDTA}
Cardiofaciocutaneous Syndrome	MAP2K1 & MAP2K2	Whole Gene Analysis	Blood^{EDTA}
Carnitine Palmitoyltransferase II Deficiency (CPT II)	CPT2	Whole Gene Analysis	Blood^{EDTA}
Cerebrotendinous Xanthomatosis	CYP27A1	Whole Gene Analysis	Blood^{EDTA}
Charcot Marie Tooth Neuropathy Type 1A	PMP22	Deletion & Duplication Analysis	Blood^{EDTA}
Charcot Marie Tooth Neuropathy Type 1B	MPZ	Whole Gene Analysis	Blood^{EDTA}
Charcot Marie Tooth Neuropathy Type 1C	LITAF	Whole Gene Analysis	Blood^{EDTA}
Charcot Marie Tooth Neuropathy Type 1D	EGR2	Whole Gene Analysis	Blood^{EDTA}
Charcot Marie Tooth Neuropathy Type 1E	PMP22	Whole Gene Analysis	Blood^{EDTA}
Charcot Marie Tooth Neuropathy Type 2A1	KIF1B	Whole Gene Analysis	Blood^{EDTA}
Charcot Marie Tooth Neuropathy Type 2A2	MFN2	Whole Gene Analysis	Blood^{EDTA}
Charcot Marie Tooth Neuropathy Type 2B1	LMNA	Whole Gene Analysis	Blood^{EDTA}
Charcot Marie Tooth Neuropathy Type 2B	EAB7	Whole Gene Analysis	Blood^{EDTA}
Charcot Marie Tooth Neuropathy Type 2D	GARS	Whole Gene Analysis	Blood^{EDTA}
Charcot Marie Tooth Neuropathy Type 2E,1F	NEFL	Whole Gene Analysis	Blood^{EDTA}
Charcot Marie Tooth Neuropathy Type 2I,2J	MPZ	Whole Gene Analysis	Blood^{EDTA}
Charcot Marie Tooth Neuropathy Type 2K	GDAP1	Whole Gene Analysis	Blood^{EDTA}
Charcot Marie Tooth Neuropathy Type 2L	HSPB8	Whole Gene Analysis	Blood^{EDTA}
Coffin Lowry Syndrome	RPS6KA3	Whole Gene Analysis	Blood^{EDTA}
Congenital Disorders Of Gylcosylation Type 1A	PMM2	Whole Gene Analysis	Blood^{EDTA}
Congenital Disorders Of Gylcosylation Type 1B	MPI	Whole Gene Analysis	Blood^{EDTA}

NEUROLOGY TEST LIST

MOLECULAR GENETIC TESTS

Test Name	Gene Name	Test Methods	Sample
Congenital Myasthenic Syndrome	CHRNB1	Whole Gene Analysis	Blood^{EDTA}
Congenital Myasthenic Syndrome	CHRND	Whole Gene Analysis	Blood^{EDTA}
Congenital Myasthenic Syndrome	CHRNE	Whole Gene Analysis	Blood^{EDTA}
Congenital Myasthenic Syndrome	COLQ	Whole Gene Analysis	Blood^{EDTA}
Congenital Myasthenic Syndrome	DOK7	Whole Gene Analysis	Blood^{EDTA}
Congenital Myasthenic Syndrome	MUSK	Whole Gene Analysis	Blood^{EDTA}
Congenital Myasthenic Syndrome	RAPSN	Whole Gene Analysis	Blood^{EDTA}
DJ-1 Related Parkinson Disease	PARK7	Whole Gene Analysis	Blood^{EDTA}
Duchenne and Becker Muscular Dystrophy	DMD-BMD (18 Exons)	Deletion Analysis	Blood^{EDTA}
Duchenne and Becker Muscular Dystrophy	DMD-BMD (79 Exons)	Deletion & Duplication Analysis	Blood^{EDTA}
Duchenne and Becker Muscular Dystrophy	DMD-BMD	Whole Gene Analysis	Blood^{EDTA}
Dystonia 1	TOR1A	Whole Gene Analysis	Blood^{EDTA}
Episodic Ataxia Type 1	KCNA1	Whole Gene Analysis	Blood^{EDTA}
Facioscapulohumeral Muscular Dystrophy	D4Z4	Deletion Analysis	Blood^{EDTA}
Fragile X Seyndrome (CGG Repeat)	FMR1	Repeat Analysis	Blood^{EDTA}
Friedreich Ataxia (GAA Repeat)	FXN	Repeat Analysis	Blood^{EDTA}
Friedreich Ataxia	FXN	Whole Gene Analysis	Blood^{EDTA}
Frontotemporal Dementia	GRN	Whole Gene Analysis	Blood^{EDTA}
Frontotemporal Dementia With Parkinsonism 17	MAPT	Whole Gene Analysis	Blood^{EDTA}
Gaucher Disease	GBA	Whole Gene Analysis	Blood^{EDTA}
GM1 Gangliosidisis Type 1	GLB1	Whole Gene Analysis	Blood^{EDTA}
GNE	GNE	Whole Gene Analysis	Blood^{EDTA}
Griscelli Seyndrome	MYO5A	Whole Gene Analysis	Blood^{EDTA}
Hallervarden Spatz Syndrome	PANK2	Whole Gene Analysis	Blood^{EDTA}
Hemiplegic Migraine Type 2	ATP1A2	Whole Gene Analysis	Blood^{EDTA}
Hereditary Spastic Paraplegia 3	SPG3A	Whole Gene Analysis	Blood^{EDTA}
Hereditary Spastic Paraplegia 4	SPASTIN (SPAST)	Whole Gene Analysis	Blood^{EDTA}
Huntington Disease	CAG	Repeat Analysis	Blood^{EDTA}

NEUROLOGY TEST LIST

MOLECULAR GENETIC TESTS

Test Name	Gene Name	Test Methods	Sample
Hyperphenylalaninemia C Defiency	QDPR	Whole Gene Analysis	Blood^{EDTA}
Hypokalemic Periodic Paralysis Type 1	CACNA1S	Whole Gene Analysis	Blood^{EDTA}
Hypomagnesemia With Secondary Hypocalcemia	TRPM6	Mutation Analysis	Blood^{EDTA}
Infantile Neuroaxonal Dystrophy	PLA2G6	Whole Gene Analysis	Blood^{EDTA}
Joubert Syndrome Type 10	OFD1	Whole Gene Analysis	Blood^{EDTA}
Joubert Syndrome Type 12	KIF7	Whole Gene Analysis	Blood^{EDTA}
Kennedy Disease (AR)	CAG	Repeat Analysis	Blood^{EDTA}
Kufor Rakeb Disease	ATP13A2	Whole Gene Analysis	Blood^{EDTA}
Leber Hereditary Optic Neuropathy-mtDNA	NADH (10 Mutations)	Mutation Analysis	Blood^{EDTA}
Leber Hereditary Optic Neuropathy-mtDNA	NADH (3 Mutations)	Mutation Analysis	Blood^{EDTA}
Leigh Syndrome	SURF1	Whole Gene Analysis	Blood^{EDTA}
Lesch Nyhan Syndrome	HPRT1	Deletion & Duplication Analysis	Blood^{EDTA}
Lesch Nyhan Syndrome	HPRT1	Whole Gene Analysis	Blood^{EDTA}
Leukoencephalopathy With Vanishing White Matter	EIF2B1, B2, B3, B4&B5	Whole Gene Analysis	Blood^{EDTA}
Leukoencephalopathy With Vanishing White Matter	EIF2B5	Whole Gene Analysis	Blood^{EDTA}
Limb Girdle Muscular Dystrophy Type 1C	CAV3	Whole Gene Analysis	Blood^{EDTA}
Limb Girdle Muscular Dystrophy Type 2A	CAPN3	Whole Gene Analysis	Blood^{EDTA}
Limb Girdle Muscular Dystrophy Type 2B	DYSF	Whole Gene Analysis	Blood^{EDTA}
Limb Girdle Muscular Dystrophy Type 2C	SGCG	Whole Gene Analysis	Blood^{EDTA}
Limb Girdle Muscular Dystrophy Type 2D	SGCA (p.Arg77Cys)	Mutation Analysis	Blood^{EDTA}
Limb Girdle Muscular Dystrophy Type 2D	SGCA	Whole Gene Analysis	Blood^{EDTA}
Limb Girdle Muscular Dystrophy Type 2E	SGCB	Whole Gene Analysis	Blood^{EDTA}
LRRK2 Realated Parkinson Disease	LRRK2 (Exons 31, 42)	Mutation Analysis	Blood^{EDTA}
MELAS Genetic Analysis	MT-TL1 (3 Mutations)	Mutation Analysis	Blood^{EDTA}

NEUROLOGY TEST LIST

MOLECULAR GENETIC TESTS

Test Name	Gene Name	Test Methods	Sample
MELAS Genetic Analysis	MT-TL1	Whole Gene Analysis	Blood^{EDTA}
Menkes Disease	ATP7A	Whole Gene Analysis	Blood^{EDTA}
Mowat Wilson Syndrome	ZEB2	Whole Gene Analysis	Blood^{EDTA}
Mucopolysaccharidosis Type 3A	SGSH	Whole Gene Analysis	Blood^{EDTA}
Multiminicore Disease	SEPN1	Whole Gene Analysis	Blood^{EDTA}
Multiple Acyl-CoA Dehydrogenase Deficiency	ETFA	Whole Gene Analysis	Blood^{EDTA}
Myoclonic Dystonia	SGCE	Whole Gene Analysis	Blood^{EDTA}
Mytotonia Congenita	CLCN1	Whole Gene Analysis	Blood^{EDTA}
Myotonic Dystrophy Type 1	DMPK (CTG)	Repeat Analysis	Blood^{EDTA}
Myotonic Dystrophy Type 2	CNBP	Mutation Analysis	Blood^{EDTA}
NAIP	NAIP	Whole Gene Analysis	Blood^{EDTA}
Narcolepsy	HLA-DQA1&HLA-DQB1 (Type DQB1-0602&DQA1-0102		Blood^{EDTA}
NARP Syndrome	MTATP6	Mutation Analysis	Blood^{EDTA}
NCL2 Late Infantile Type	TPP1	Whole Gene Analysis	Blood^{EDTA}
Nemaline Myopathy 1	TPM3	Whole Gene Analysis	Blood^{EDTA}
Neurofibromatosis Type 1	NF1	Whole Gene Analysis	Blood^{EDTA}
Neurofibromatosis Type 2	NF2	Whole Gene Analysis	Blood^{EDTA}
Neutral Lipid Storage Disease With Myopathy	PNPLA2	Whole Gene Analysis	Blood^{EDTA}
Noonan Syndrome Tip 1	PTPN11	Whole Gene Analysis	Blood^{EDTA}
NPHP1	NPHP1	Whole Gene Analysis	Blood^{EDTA}
NTRK1	NTRK1	Whole Gene Analysis	Blood^{EDTA}
Opitz G/BBB Syndrome, X Linked	MID1	Whole Gene Analysis	Blood^{EDTA}
Ornithine Transcarbamylase Deficiency	OTC	Whole Gene Analysis	Blood^{EDTA}
Osteopetrosis Type 1	TCIRG1	Whole Gene Analysis	Blood^{EDTA}
Osteopetrosis Type 3 (AR)	CA2	Whole Gene Analysis	Blood^{EDTA}
Pallister Hall Syndrome	GLI3	Whole Gene Analysis	Blood^{EDTA}
PARK2 Related Juvenile Parkinson Disease	PARK2	Whole Gene Analysis	Blood^{EDTA}
Pelizaeus Merzbacher Disease	PLP1	Deletion & Duplication Analysis	Blood^{EDTA}
Pelizaeus Merzbacher Disease	PLP1	Whole Gene Analysis	Blood^{EDTA}

NEUROLOGY TEST LIST

MOLECULAR GENETIC TESTS

Test Name	Gene Name	Test Methods	Sample
Pelizaeus Merzbacher Like Disease	GJC2	Whole Gene Analysis	Blood^{EDTA}
Peroxisome Biogenesis Disorders	PEX1	Mutation Analysis	Blood^{EDTA}
Peroxisome Biogenesis Disorders	PEX1	Whole Gene Analysis	Blood^{EDTA}
Phenylketonuria (PKU)	PAH	Mutation Analysis	Blood^{EDTA}
Phenylketonuria (PKU)	PAH	Whole Gene Analysis	Blood^{EDTA}
PINK1 Related Parkinson Disease	PINK1	Whole Gene Analysis	Blood^{EDTA}
Polyglucosan Body Disease	GBE1	Whole Gene Analysis	Blood^{EDTA}
Prion Disease	PRNP	Whole Gene Analysis	Blood^{EDTA}
Progressive Myoclonic Epilepsy Type 1	CSTB	Whole Gene Analysis	Blood^{EDTA}
Progressive Myoclonic Epilepsy Type 2A	EPM2A	Whole Gene Analysis	Blood^{EDTA}
Progressive Myoclonic Epilepsy Type 2B	NHLRC1	Whole Gene Analysis	Blood^{EDTA}
Protein C Deficiency Syndrome	PROC	Whole Gene Analysis	Blood^{EDTA}
Pyridoxine Deficiency	ALDH7A1	Whole Gene Analysis	Blood^{EDTA}
Rett Syndrome	MECP2	Deletion & Duplication Analysis	Blood^{EDTA}
Rett Syndrome	MECP2 (6 Mutation)	Mutation Analysis	Blood^{EDTA}
Rett Syndrome	MECP2	Whole Gene Analysis	Blood^{EDTA}
Roberts Syndrome	ESCO2	Whole Gene Analysis	Blood^{EDTA}
Sandhoff Disease	HEXB	Whole Gene Analysis	Blood^{EDTA}
SCN1B	SCN1B	Whole Gene Analysis	Blood^{EDTA}
SCN9A	SCN9A	Whole Gene Analysis	Blood^{EDTA}
Segawa Syndrome (AD)	GCH1	Whole Gene Analysis	Blood^{EDTA}
Segawa Syndrome (AR)	TH	Whole Gene Analysis	Blood^{EDTA}
Silver Syndrome	BSCL2	Whole Gene Analysis	Blood^{EDTA}
Sjogren Larsson Syndrome	ALDH3A2	Whole Gene Analysis	Blood^{EDTA}
Sjogren Larsson Syndrome	ALDH3A2	Whole Gene Analysis	Blood^{EDTA}
Smith Lemli Opitz Syndrome	DHCR7	Whole Gene Analysis	Blood^{EDTA}
SNCA Related Parkinson Disease	SNCA	Whole Gene Analysis	Blood^{EDTA}
Spastic Ataxia Of Charlevaix Saguenay (AR)	SACS	Whole Gene Analysis	Blood^{EDTA}
Spastic Paraplegia Type 11	SPG11	Whole Gene Analysis	Blood^{EDTA}

NEUROLOGY TEST LIST

MOLECULAR GENETIC TESTS

Test Name	Gene Name	Test Methods	Sample
Spinal Musculer Atrophy	SMN1 (Exon 7,8)	Deletion Analysis	Blood^{EDTA}
Spinal Musculer Atrophy	SMN1	Whole Gene Analysis	Blood^{EDTA}
Spinocerebellar Ataxia Type 1	ATXN1 CAG	Repeat Analysis	Blood^{EDTA}
Spinocerebellar Ataxia Type 2	ATXN2 CAG	Repeat Analysis	Blood^{EDTA}
Spinocerebellar Ataxia Type 3	ATXN3 CAG	Repeat Analysis	Blood^{EDTA}
Spinocerebellar Ataxia Type 6	CACNA1A CAG	Repeat Analysis	Blood^{EDTA}
Spinocerebellar Ataxia Type 7	ATXN7 CAG	Repeat Analysis	Blood^{EDTA}
Spinocerebellar Ataxia Type 8	ATXN80S	Mutation Analysis	Blood^{EDTA}
Spinocerebellar Ataxia Type 10	ATXN10	Mutation Analysis	Blood^{EDTA}
Spinocerebellar Ataxia Type 12	PPP2R2B CAG	Repeat Analysis	Blood^{EDTA}
Spinocerebellar Ataxia Type 17	TBP CAA/CAG	Repeat Analysis	Blood^{EDTA}
Succinic Semialdehyde Dehydrogenase Deficiency	ALDHSA1	Whole Gene Analysis	Blood^{EDTA}
Tay Sachs Disease	HEXA	Whole Gene Analysis	Blood^{EDTA}
Trichothiodyrtrophy Dehydrogenase Deficiency	ALDH5A1	Whole Gene Analysis	Blood^{EDTA}
Troyer Syndrome	SPG20	Whole Gene Analysis	Blood^{EDTA}
Tuberosis Sclerosis	TSC1	Whole Gene Analysis	Blood^{EDTA}
Tuberosis Sclerosis	TSC2	Whole Gene Analysis	Blood^{EDTA}
Ullrich Congenital Muscular Dystrophy	COL6A1	Whole Gene Analysis	Blood^{EDTA}
Ullrich Congenital Muscular Dystrophy	COL6A2	Whole Gene Analysis	Blood^{EDTA}
Ullrich Congenital Muscular Dystrophy	COL6A3	Whole Gene Analysis	Blood^{EDTA}
Van Der Knaap Disease	MLC1	Whole Gene Analysis	Blood^{EDTA}
Van Der Knaap Disease	MLC1	Whole Gene Analysis	Blood^{EDTA}
Very Long Chain Acyl CoA Dehydrogenase Deficiency	ACADVL	Whole Gene Analysis	Blood^{EDTA}
Waardenburg Syndrome Type 1	PAX3	Whole Gene Analysis	Blood^{EDTA}
Waardenburg Syndrome Type 2	MITF	Whole Gene Analysis	Blood^{EDTA}
Waardenburg Syndrome Type 4	EDN3	Whole Gene Analysis	Blood^{EDTA}
Waardenburg Syndrome Type 4	EDNRB	Whole Gene Analysis	Blood^{EDTA}
Waardenburg Syndrome Type 4	SOX10	Whole Gene Analysis	Blood^{EDTA}
Walker Warburg Syndrome	FKRP	Whole Gene Analysis	Blood^{EDTA}

NEUROLOGY TEST LIST

MOLECULAR GENETIC TESTS

Test Name	Gene Name	Test Methods	Sample
Walker Warburg Syndrome	FKTN	Whole Gene Analysis	Blood^{EDTA}
Walker Warburg Syndrome	LARGE	Whole Gene Analysis	Blood^{EDTA}
Walker Warburg Syndrome	POMGNT1	Whole Gene Analysis	Blood^{EDTA}
Walker Warburg Syndrome	POMT1	Whole Gene Analysis	Blood^{EDTA}
Walker Warburg Syndrome	POMT2	Whole Gene Analysis	Blood^{EDTA}
Warburg Micro Syndrome Type 1	RAB3GAP1	Whole Gene Analysis	Blood^{EDTA}
Warburg Micro Syndrome Type 2	RAB3GAP2	Whole Gene Analysis	Blood^{EDTA}
Warburg Micro Syndrome Type 3	RAB18	Whole Gene Analysis	Blood^{EDTA}
Wilson Disease	ATP7B	Whole Gene Analysis	Blood^{EDTA}
Wolfram Syndrome	WFS1	Whole Gene Analysis	Blood^{EDTA}
X-Linked Adrenoleukodystrophy	ABCD1	Whole Gene Analysis	Blood^{EDTA}

THROMBOPHILIA PANEL

Test Name	Sample
Factor V Leiden Mutation Analysis (G1691A)	Blood^{EDTA}
Factor XIII V34L Mutation Anaylsis	Blood^{EDTA}
MTHFR Mutation Analysis (A1298C)	Blood^{EDTA}
MTHFR Mutation Analysis (C677T)	Blood^{EDTA}
PAI-SERPINE1 Mutation Anaylsis	Blood^{EDTA}
Prothrombin (F2) Mutation Anaylsis	Blood^{EDTA}

MOLECULAR CYTOGENETIC TESTS

Test Name	Sample
1p36 del FISH ANALYSIS	Blood^{EDTA}
Angelman Syndrome (del 15q11-q13) FISH Analysis	Blood^{EDTA}
Cri Du Chat Syndrome (del 5p15.2) FISH Analysis	Blood^{EDTA}
Di George Syndrome (10p) FISH Analysis	Blood^{EDTA}
Di George / VCF Syndrome FISH Analysis	Blood^{EDTA}
Ichthyosis Steroid Sulfatase (del Xp22.32-STS) FISH Analysis	Blood^{EDTA}
Neurofibromatosis Type 1 – NF1 (17q11.2 del) FISH Analysis	Blood^{EDTA}
N – MYC Neuroblastoma (2p24.1) FISH Analysis	Blood^{EDTA}
Rubinstein Taybi Syndrome (del 16p13.3) FISH Analysis	Blood^{EDTA}
Smith Magenis Syndrome (del 17p11.2-RAI1) FISH Analysis	Blood^{EDTA}
Sotos Syndrome – NSD1 (del 5q35) FISH Analysis	Blood^{EDTA}
Subtelomeric FISH Analysis	Blood^{EDTA}
Williams Syndrome (del 7q11.23) FISH Analysis	Blood^{EDTA}
Wolf Hirschhorn Syndrome – WHCR FISH Analysis	

WHOLE EXOME SEQUENCING (WES)

	Target	Ekzon Number	10 Target	Coverage
Whole Exome	45mb	214,405	> %80	10X

NEUROLOGY TEST LIST

NEXT GENERATİON SEQUENCİNG (NGS) PANELS

Test Name	Gene Name
Aicardi Goutieres Syndrome Screening Panel	ADAR, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, TREX1
Alzheimer Disease / Frontotemporal Dementia Screening Panel	A2M, ABCA7, ACE, APOE, APP, BIN1, BLMH, C9orf72, CD2AP, CHMP2B, CLU, CR1, CSF1R, CST3, DNMT1, FTL, FUS, GBA, GCDH, GRN, HFE, HLA-DQB1, HSD17B10, ITM2B, MAPK10, MAPT, MLYCD, MPO, MS4A2, MS4A6E, NOS3, PICALM, PLAU, PRNP, PSEN1, PSEN2, SNCA, SNCB, SORL1, SPG21, SQSTM1, TARDBP, TBP, TOMM40, TREM2, UBQLN2, VCP
Amyloid Related Disorders Screening Panel	APOA1, APOA2, APP, B2M, CST3, FGA, GSN, IL31RA, ITM2B, LYZ, MEFV, NLRP12, NLRP3, OSMR, PSEN2, TNFRSF1A, TTR
Amyotrophic Lateral Sclerosis Screening Panel	ALS2, ANG, ATXN2, C9orf72, CHMP2B, DCTN1, ERBB4, FIG4, FUS, MAPT, MATR3, NEFH, OPTN, PARK7, PFN1, PRPH, PSEN1, SETX, SIGMAR1, SOD1, SQSTM1, TARDBP, UBQLN2, VAPB, VCP
Charcot Marie Tooth Disease Screening Panel	AARS, AIFM1, ARHGEF10, BAG3, BSCL2, C12orf65, CTDP1, DNM2, EGR2, FGD4, FIG4, GAN, GARS, GDAP1, GJB1, HK1, HSPB1, HSPB8, INF2, KARS, LITAF, LMNA, LRSAM1, MED25, MFN2, MPZ, MTMR2, NDRG1, NEFL, PMP22, PRPS1, PRX, RAB7A, SBF2, SEPT9, SH3TC2, SLC12A6, TRPV4, YARS
Comprehensive Dystonia Screening Panel	ACTB, ADAR, AFG3L2, AP1S2, APTX, ARSA, ARX, ATM, ATP13A2, ATP1A2, ATP1A3, ATP7B, AUH, C19orf12, CACNA1A, CHMP2B, CP, DCAF17, DDC, DLAT, DRD2, DRD5, EARS2, ERCC6, FA2H, FASTKD2, FBXO7, FOXG1, FOXRED1, FTL, GAMT, GCDH, GCH1, GJA1, HPRT1, HTT, KCNMA1, KCNQ2, L2HGDH, MAPT, MARS2, MAT1A, MCOLN1, MMADHC, MPV17, NPC2, PANK2, PARK2, PDGFB, PDHX, PINK1, PLP1, PNKD, PRKRA, PRRT2, PSEN1, PTEN, PTS, QDPR, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, SCP2, SDHAF1, SGCE, SLC19A3, SLC20A2, SLC2A1, SLC46A1, SLC6A3, SPR, SUCLA2, SUOX, TAF1, TH, THAP1, TIMM8A, TOR1A, TPK1, TREM2, TREX1, UBQLN2, WDR45, ZNF592

NEUROLOGY TEST LIST

NEXT GENERATİON SEQUENCİNG (NGS) PANELS

Test Name	Gene Name
Comprehensive Muscular Dystrophy / Myopathy Screening Panel	AARS2, ACADS, ACADVL, ACTA1, ADCK3, AFG3L2, AGK, AGL, AGRN, AIFM1, ALDOA, AMPD1, ANO5, ATP2A1, B3GNT1, BAG3, BIN1, C10orf2, C12orf65, CACNA1S, CAPN3, CAV3, CFL2, CHAT, CHKB, CHRNA1, CHRND, CHRNE, CLCN1, CNBP, CNTN1, COA5, COL4A1, COL6A1, COL6A2, COL6A3, COLQ, COQ2, COQ9, COX10, COX14, COX6B1, CPT2, CRYAB, DES, DMD, DMPK, DNAJB6, DNM2, DOK7, DUX4, DYSF, EARS2, EMD, ENO3, FARS2, FASTKD2, FHL1, FKRP, FKTN, FLNC, FOXRED1, GAA, GBE1, GFER, GFM1, GFPT1, GNE, GTDC2, GYG1, GYS1, HADHA, HADHB, HCCS, HRAS, HSPG2, ISCU, ISPD, ITGA7, KBTBD13, KIF21A, LAMA2, LAMP2, LARGE, LDB3, LMNA, LPIN1, MATR3, MEGF10, MPV17, MRPL3, MRPS16, MRPS22, MTFMT, MTM1, MTMR14, MTO1, MUSK, MYF6, MYH14, MYH2, MYH7, MYL2, MYOT, NEB, OPA1, PABPN1, PDSS1, PDSS2, PFKM, PGAM2, PGK1, PHKA1, PLEC, PNPLA2, POLG, POLG2, POMGNT1, POMT1, POMT2, PRKAG2, PRKAG3, PTRF, PUS1, PYGM, RAPSN, RRM2B, RYR1, SARS2, SCN4A, SCO1, SCO2, SDHAF1, SEPN1, SEPT9, SGCA, SGCB, SGCD, SGCG, SIL1, SLC22A5, SLC25A4, SLC37A4, SLC52A1, STIM1, SUCLG1, SURF1, SYNE1, SYNE2, TAZ, TCAP, TK2, TNNT1, TPM2, TPM3, TRIM32, TRMU, TSFM, TTN, TUBB3, TUFM, TYMP, VCP, YARS2
Congenital Myasthenic Syndrome Screening Panel	AGRN, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, COLQ, DOK7, GFPT1, MUSK, RAPSN, SCN4A
Epilepsy Gene Panel	AARS, ABAT, ABCC8, ACY1, ADSL, ALDH7A1, ALG1, ALG11, ALG13, ALG3, AMACR, AMT, ARHGEF9, ARX, ASAH1, ATP1A2, ATP1A3, ATP6AP2, ATP7A, ATRX, AUTS2, BOLA3, BTD, CACNA1A, CASK, CDKL5, CHD2, CHRNA2, CHRNA4, CHRNB2, CLDN16, CLDN19, CLN3, CLN5, CLN6, CLN8, CNNM2, CNTNAP2, COQ2, CPA6, CPS1, CPT2, CSTB, CTSD, CUL4B, D2HGDH, DCX, DLAT, DNAJC5, DNM1, DPAGT1, DPM1, DPYD, DYNC1H1, DYRK1A, EGF, EHMT1, EPM2A, FA2H, FARS2, FASN, FGD1, FLNA, FOLR1, FOXG1, FOXRED1, FXYD2, GABRA1, GABRB3, GABRG2, GAMT, GCK, GCSH, GLDC, GLRA1, GLRB, GLUD1, GOSR2, GPC3, GPHN, GRIA3, GRIN1, GRIN2A, GRIN2B, GRN, HADH, HCN1, HDAC4, HLCS, HNRNPU, HSD17B10, HSD17B4, IDH2, IER3IP1, IFIH1, IQSEC2, KANSL1, KCNA1, KCNJ10, KCNJ11, KCNMA1, KCNQ2, KCNQ3, KCTD7, KDM5C, LGI1, LIAS, MBD5, MECP2, MED12, MEF2C, MFSD8, MOCS1, MOCS2, MPDU1, MTHFR, NDUFA1, NDUFA11, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFB3, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFV1, NDUFV2, NEDD4L, NHLRC1, NRXN1, NUBPL, OFD1, OPHN1, PAK3, PC, PCDH19, PDHA1, PDHB, PDP1, PDX1, PEX1, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26, PEX3, PEX5, PEX6, PHF6, PHGDH, PIGA, PIGN, PIGO, PLA2G6, PLCB1, PLP1, PMM2, PNKP, PNPO, POLG, PPP2R1A, PPT1, PQBP1, PRICKLE1, PRICKLE2, PRRT2, RAB39B, RARS2, RNASEH2A, RNASEH2B, RNASEH2C, ROGDI, RPS6KA3, RRM2B, RYR3, SAMHD1, SCARB2, SCN1A, SCN1B, SCN2A, SCN8A, SCN9A, SLC16A1, SLC19A3, SLC25A15, SLC25A22, SLC2A1, SLC6A1, SLC6A8, SLC9A6, SMC1A, SMS, SPTAN1, SRPX2, ST3GAL3, ST3GAL5, STXBP1, SUOX, SYN1, SYNGAP1, SYP, TBC1D24, TBCE, TCF4, TPP1, TREX1, TRPM6, TSC1, UBE3A, WWOX, ZEB2

NEUROLOGY TEST LIST

NEXT GENERATİON SEQUENCİNG (NGS) PANELS

Test Name	Gene Name
Fetal Akinesia, Arthrogryposis, or Contractures Screening Panel	ABCC8, ACTA1, AIMP1, ALG12, ALG2, ALG3, ALG6, ALG8, ALG9, ARX, ASAH1, ATP7A, B4GALT1, BAG3, BIN1, CAPN3, CDC6, CDT1, CHAT, CHRNA1, CHRND, CHRNG, CHST14, CHUK, CLIC2, CNTN1, COG7, COL6A1, COL6A2, COL6A3, CPT2, DHCR24, DOK7, DPAGT1, DPM1, DPM3, DST, EGR2, EMD, ERBB3, ERCC1, ERCC2, ERCC6, ERLIN2, EXOSC3, FAM20C, FBN1, FBN2, FHL1, FKRP, FKTN, FLVCR2, FUCA1, GAD1, GBA, GBE1, GCK, GDAP1, GJB1, GLE1, GLUL, GUSB, HDAC8, HRAS, HSPG2, HUWE1, IDS, IGHMBP2, ISPD, KCNJ11, KLKB1, LAMA2, LAMB1, LARGE, LMNA, MED12, MEGF10, MFN2, MOGS, MPDU1, MPI, MPZ, MTM1, MYBPC1, MYH2, MYH3, MYH8, MYOT, OCRL, ORC1, ORC4, ORC6, PEX7, PGM1, PIP5K1C, PLA2G6, PLEC, PLEKHG5, PLOD3, PLP1, PMM2, POLG, POMGNT1, POMT1, POMT2, PTRF, RAPSN, RARS2, RFT1, RIPK4, RYR1, SEPN1, SEPSECS, SGCG, SKI, SLC12A6, SLC25A19, SLC2A10, SLC39A13, SMN1, SOX10, SPG20, SRD5A3, SYNE1, TMEM165, TNNI2, TNNT1, TNNT3, TPM2, TPM3, TRPV4, TSEN2, TSEN34, TSEN54, UBA1, VIPAS39, VPS33B, VRK1, ZMPSTE24, ZNF335, ZNF592
Hyperekplexia Gene Panel (O.D & O.R.)	ARHGEF9, GLRA1, GLRB, GPHN, SLC6A5
Metabolic Disorders Gene Panel	AASS, ABAT, ABCD1, ABCD4, ABCG5, ABCG8, ABHD12, ABHD5, ACACA, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACAT2, ACO2, ACOX1, ACSF3, ACSL4, ACY1, ADA, ADCY5, ADK, ADSL, AGA, AGK, AGL, AGPAT2, AGPS, AGXT, AHCY, AK1, AK2, AKR1D1, ALAD, ALAS2, ALDH18A1, ALDH2, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, ALOX12B, ALPL, AMACR, AMN, AMPD3, AMT, AP1S1, APOC2, APRT, ARG1, ARSA, ARSB, ASAH1, ASL, ASPA, ASS1, ATIC, ATP6V0A2, ATP7A, ATP7B, ATP8B1, AUH, B3GALNT1, B3GALTL, B3GAT3, B3GNT1, B4GALT1, B4GALT7, BAAT, BCKDHA, BCKDHB, BCMO1, BLVRA, BMP2, BPGM, BTD, C7orf10, CANT1, CAT, CBS, CEL, CERKL, CFTR, CHIT1, CHKB, CHST14, CHST3, CHST6, CHSY1, CLN3, CLN5, CLN6, CLN8, COG1, COG4, COG5, COG6, COG7, COG8, COMT, CP, CPOX, CPS1, CPT1A, CPT2, CTH, CTNS, CTSA, CTSC, CTSD, CTSK, CUBN, CYB5R3, CYP11A1, CYP11B1, CYP11B2, CYP17A1, CYP19A1, CYP1B1, CYP21A2, CYP27A1, CYP27B1, CYP2R1, CYP7B1, D2HGDH, DAO, DBH, DBT, DCXR, DDC, DDOST, DGAT1, DGUOK, DHCR24, DHCR7, DHFR, DHODH, DLD, DMGDH, DNAJC19, DNM1L, DNM2, DNMT1, DNMT3B, DOLK, DPAGT1, DPM1, DPM3, DPYD, DPYS, EBP, ELOVL4, ENO3, EPHX1, EPHX2, ETFA, ETFB, ETFDH, ETHE1, EXT1, EXT2, FA2H, FAH, FBP1, FECH, FH, FKRP, FKTN, FMO3, FOLR1, FTCD, FUCA1, FUT2, FUT6, G6PC, G6PC3, G6PD, GAA, GAD1, GALC, GALE, GALK1, GALNS, GALT, GAMT, GATM, GBA, GBE1, GCDH, GCH1, GCK, GCLC, GCLM, GCSH, GFPT1, GK, GLA, GLB1, GLDC, GLRA1, GLRX5, GLUD1, GLUL, GLYCTK, GM2A, GNE, GNMT, GNPAT, GNPTAB, GNPTG, GNS, GOT1, GPD1, GPD1L, GPHN, GPI, GPX1, GRHPR, GSS, GUSB, GYG1, GYS1, GYS2, H6PD, HADH, HADHA, HADHB, HEXA, HEXB, HFE, HGD, HGSNAT, HIBCH, HK1, HLCS, HMBS, HMGCL, HMGCS2, HMOX1, HOGA1, HPD, HPRT1, HS6ST1, HSD11B1, HSDB11B2, HSD17B10, HSD17B3, HSD17B4, HSD3B2, HSD3B7, HYAL1, IDH2, IDH3B, IDS, IDUA, IMPAD1, IMPDH1, ISPD, IVD, KMT2D, L2HGDH, LAMP2, LARGE, LCAT, LCT, LDHA, LDHB, LFNG, LIPA, LIPC, LMBRD1, LPIN1, LPIN2, LPL, LRAT, LTC4S, MAN1B1, MAN2B1, MANBA, MAOA, MAT1A, MCCC1, MCCC2, MCEE, MCOLN1, MFSD8, MGAT2, MINPP1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MOCS1, MOCS2, MOGS, MPDU1, MPI, MSMO1, MTHFD1, MTHFR, MTM1, MTMR2, MTR, MTRR, MUT, MVK, NAGA, NAGLU, NAGS, NEU1, NMNAT1, NPC1, NPC2, NSD1, NSDHL, NT5C3A, NT5E, OAT, OCRL, OPA3, OTC, OXCT1, PAH, PANK2, PC, PCBD1, PCCA, PCCB, PEPD, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3,

NEUROLOGY TEST LIST

NEXT GENERATİON SEQUENCİNG (NGS) PANELS

Test Name	Gene Name
Metabolic Disorders Gene Panel	PEX5, PEX6, PEX7, PFKM, PGAM2, PGK1, PGM1, PHGDH, PHKA1, PHKA2, PHYH, PIGA, PIGL, PIGM, PIGN, PIGO, PIGV, PIK3CA, PIK3R1, PIK3R2, PIK3R5, PIKFYVE, PIP5K1C, PKLR, PLA2G5, PLA2G6, PLA2G7, PLCB1, PLCB4, PLCD1, PLCE1, PLCG2, PLIN1, PLOD1, PLOD2, PLOD3, PMM2, PNMT, PNP, PNPLA2, PNPLA6, PNPO, POLR3A, POLR3B, POMGNT1, POMT1, POMT2, PPOX, PPT1, PRODH, PRPS1, PSAP, PSAT1, PSPH, PTEN, PTGIS, PTPN11, PTS, PYCR1, PYGL, PYGM, QDPR, RDH12, RDH5, RFT1, RPE65, RPIA, SARDH, SAT1, SCARB2, SCP2, SEPSECS, SGSH, SI, SLC16A1, SLC17A5, SLC22A5, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A38, SLC2A1, SLC2A2, SLC30A10, SLC33A1, SLC35A1, SLC35C1, SLC37A4, SLC39A4, SLC3A1, SLC46A1, SLC52A1, SLC52A3, SLC5A1, SLC5A2, SLC6A8, SLC7A7, SLC7A9, SLCO1B1, SLCO1B3, SMPD1, SMS, SOD1, SPR, SPTLC1, SPTLC2, SRD5A3, ST3GAL3, ST3GAL5, STAR, STS, SUCLA2, SUCLG1, SUMF1, SUOX, TALDO1, TAT, TAZ, TBXAS1, TCIRG1, TCN2, TECR, TH, TK2, TMEM165, TMLHE, TPI1, TPMT, TPP1, TUSC3, TYMP, TYR, TYRP1, UGT1A1, UMPS, UPB1, UROC1, UROD, UROS, XDH
Mitochondrial Gene Panel	AARS2, ABCB7, ACAD8, ACAD9, ACADSB, ACAT1, ACO2, ACSF3, ADAR, ADCK3, AFG3L2, AGK, AIFM1, ANO10, APTX, ATP5E, ATPAF2, BCKDHA, BCKDHB, BCS1L, BOLA3, C10orf2, C12orf65, C19orf12, CHKB, COA5, COQ2, COQ4, COQ5, COQ6, COQ9, COX10, COX14, COX15, COX4I1, COX4I2, COX6B1, COX7A1, COX7A2, CYCS, DARS2, DBT, DGUOK, DLAT, DLD, DNAJC19, DNM1L, EARS2, ECSIT, ELAC2, ETFA, ETFB, ETFDH, ETHE1, FARS2, FASTKD2, FH, FOXRED1, FXN, GARS, GATM, GCDH, GFER, GFM1, GLRX5, GLUD1, HADH, HADHA, HADHB, HARS2, HCCS, HIBCH, HLCS, HMGCL, HMGCS2, HOGA1, HSD17B10, HSPA9, HSPD1, ISCU, KARS, LARS2, LIAS, LRPPRC, MARS2, MCCC2, MFF, MFN2, MPV17, MRPL3, MRPL48, MRPS16, MRPS22, MRRF, MTCH2, MTFMT, MTHFD1L, MTO1, MTPAP, NARS2, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA4, NDUFA6, NDUFA7, NDUFA8, NDUFA9, NDUFAB1, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF7, NDUFB1, NDUFB2, NDUFB3, NDUFB6, NDUFB9, NDUFC2, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS5, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NDUFV3, NFU1, NUBPL, OPA1, OPA3, OXCT1, PANK2, PC, PCK2, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PINK1, POLG, POLG2, POP1, PUS1, PYCR1, RARS2, RRM2B, SARS2, SCO1, SCO2, SDHA, SDHAF1, SDAHF2, SDHB, SDHC, SDHD, SLC19A2, SLC19A3, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A3, SLC25A38, SLC25A39, SLC25A4, SLC3A1, SPG7, SUCLA2, SUCLG1, SURF1, TACO1, TAZ, TFAM, TFB1M, TIMM8A, TK2, TMEM126A, TMEM70, TOMM40, TPK1, TRMU, TSFM, TTC19, TUFM, TYMP, UCP1, UCP2, UCP3, UQCRB, UQCRQ, WFS1, YARS2

NEUROLOGY TEST LIST

NEXT GENERATİON SEQUENCİNG (NGS) PANELS

Test Name	Gene Name
Movement Disorders Gene Panel	ABCB7, ABCD1, ABHD12, ACTB, ADCK3, ADCY5, AFG3L2, ALDH3A2, ANO10, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APTX, ARSA, ARX, ASPA, ATCAY, ATL1, ATM, ATP13A2, ATP1A3, ATP7B, B4GALNT1, BCAP31, BCKDHA, BCKDHB, BSCL2, C10orf2, C12orf65, C19orf12, CA8, CACNA1A, CACNB4, CCT5, COQ2, COQ9, CP, CSTB, CYP27A1, CYP7B1, DBT, DCAF17, DCTN1, DDC, DLAT, DLD, DNMT1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF4G1, ERLIN2, FA2H, FBXO7, FGF14, FLVCR1, FTL, GALC, GAN, GBA, GCDH, GCH1, GFAP, GJC2, GLB1, GOSR2, GPR56, GRM1, HEXB, HPRT1, HSPD1, ITPR1, KCNA1, KCNC3, KCNJ10, KCTD7, KIAA0196, KIAA0226, KIF1A, KIF1C, KIF5A, L1CAM, MARS2, MECP2, MMADHC, MRE11A, MTHFR, MTPAP, MTTP, NIPA1, NKX2-1, NPC1, NPC2, NUP62, OPA1, PANK2, PAX6, PDE8B, PDGFB, PDHA1, PDHX, PDSS1, PDSS2, PDYN, PEX10, PEX7, PHYH, PIK3R5, PLA2G6, PLP1, PMM2, PNKD, PNKP, PNPLA6, POLG, PRKCG, PRKRA, PRRT2, REEP1, RNASEH2A, RNASEH2B, RNASEH2C, RNFI70, RTN2, SACS, SAMHD1, SCN8A, SETX, SGCE, SIL1, SLC12A6, SLC16A2, SLC19A3, SLC1A3, SLC20A2, SLC25A15, SLC2A1, SLC30A10, SLC33A1, SLC6A3, SMPD1, SNCA, SPAST, SPG11, SPG20, SPG21, SPG7, SPR, SPTBN2, SUOX, SYNE1, TAF1, TDP1, TGM6, TH, THAP1, TIMM8A, TMEM67, TOR1A, TREX1, TTBK2, TTC19, TTPA, VCP, VLDLR, VPS13A, WDR45, WDR81, WWOX, ZFYVE26, ZFYVE27, ZNF592
Muscle Disorders Gene Panel	ACADVL, ACTA1, ACVR1, AGL, AGRN, ANO5, ATP2A1, B3GNT1, BAG3, BIN1, CACNA1S, CAPN3, CAV3, CFL2, CHAT, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE, CLCN1, CNTN1, COL12A1, COL6A1, COL6A2, COL6A3, COLQ, CPT2, CRYAB, DAG1, DES, DMD, DNAJB6, DNM2, DOK7, DPAGT1, DPM3, DYNC1H1, DYSF, EMD, ENO3, ERBB3, FHL1, FKRP, FKTN, FLNC, GAA, GBE1, GFPT1, GNE, GYG1, GYS1, HSPG2, IGHMBP2, ISCU, ISPD, ITGA7, KBTBD13, KCNJ2, KLHL9, LAMA2, LAMP2, LARGE, LDB3, LDHA, LMNA, LPIN1, MATR3, MEGF10, MSTN, MTM1, MUSK, MYBPC3, MYF6, MYH2, MYH3, MYH7, MYOT, NEB, ORAI1, PABPN1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PIP5K1C, PLEC, PNPLA2, POMGNT1, POMT1, POMT2, PTRF, PYGM, RAPSN, RYR1, SCN4A, SEPN1, SGCA, SGCB, SGCD, SGCG, STIM1, TCAP, TNNI2, TNNT1, TPM2, TPM3, TRIM32, TRPV4, TTC19, TTN, UBA1, VCP, VIPAS39
Neuropathies HMSN Gene Panel	AARS, AIFM1, ATL1, BSCL2, C10orf2, DCTN1, DNAJB2, DNM2, DNMT1, DYNC1H1, EGR2, FAM134B, FGD4, FIG4, GAN, GARS, GDAP1, GJB1, HSPB1, HSPB3, HSPB8, IGHMBP2, IKBKAP, INF2, KARS, KIF1A, KIF1B, KIF5A, LITAF, LMNA, LRSAM1, MED25, MFN2, MPZ, MTMR2, NDRG1, NEFL, NGF, NTRK1, PLEKHG5, PMP22, PRPS1, PRX, RAB7A, SBF2, SCN10A, SCN9A, SH3TC2, SLC12A6, SLC5A7, SMN1, SPTLC1, SPTLC2, SYT2, TRPV4, TTR, VCP, WNK1, YARS
Parkinson Disease / Parkinsonizm Screening Panel	ADH1C, AFG3L2, APOE, APP, ATP13A2, ATP1A3, ATXN2, BST1, C10orf2, C19orf12, CHCHD10, CLN3, CP, CSF1R, CYP2D6, CYP2E1, DCAF17, DCTN1, DDC, DNAJC5, DNAJC6, EIF4G1, FBXO7, FMR1, FTL, GAK, GBA, GCH1, GIGYF2, GRN, HLA-DRB5, HTRA2, KIF5A, LRRK2, LYST, MAPT, MCCC1, MECP2, MED13, MPV17, NPC1, PARK2, PARK7, PDGFB, PINK1, PITX3, PLA2G6, POLG, PRKRA, PRNP, PSEN1, PTEN, PTS, QDPR, RAB7L1, SLC20A2, SLC6A3, SMPD1, SNCA, SNCB, SPG11, SPR, STK39, SYT11, TAF1, TARDBP, TBP, TH, UCHL1, VPS35, WDR45
Spastic Paraplegia Screening Panel	ABCD1, ACOX1, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ATL1, BSCL2, C12orf65, CCT5, CYP7B1, ERLIN2, FA2H, FBXO7, GAD1, GAN, GJC2, HARS2, HSPD1, KDM5C, KIAA0196, KIF1A, KIF5A, L1CAM, LARS2, MARS2, NIPA1, OPA3, PLP1, PNPLA6, PSEN1, REEP1, RTN2, SLC16A2, SLC19A3, SLC2A1, SLC33A1, SPAST, SPG11, SPG20, SPG21, SPG7, STXBP1, TTR, ZYFVE26, ZFYVE27

MEDİPOL UNV İSTANBUL MEDİPOL ÜNİVERSİTESİ

NÖROLOJİ TEST LİSTESİ

Lütfen İlgili Kutucuğu İşaretleyiniz
Arka Sayfadaki Anamnez & Onam Formunu Doldurunuz

MOLEKÜLER GENETİK TESTLER	NUMUNE TÜRÜ
☐ Alexander Hastalığı - GFAP (DTIP1A4) Tüm Gen Dizi Analizi	Kan EDTA
☐ Antitrombin 3 Eksikliği - SERPINCI Tüm Gen Dizi Analizi	Kan EDTA
☐ APO E Genotip Analizi (E2, E3, E4)	Kan EDTA
☐ ARX Geni ile İlişkili Hastalıklar - ARX Tüm Gen Dizi Analizi	Kan EDTA
☐ Ataksi Telenjiektazi - ATM Tüm Gen Dizi Analizi	Kan EDTA
☐ Bilinen Mutasyon Analizi	Kan EDTA
☐ CADASIL - NOTCH3 (Ekzon 3-6) Dizi Analizi	Kan EDTA
☐ Canavan Sendromu - ASPA Tüm Gen Dizi Analizi	Kan EDTA
☐ Charcot-Marie-Tooth Nöropati X İlişkili - GJB1 (CX32) Tüm Gen Dizi Analizi	Kan EDTA
☐ Cohen Sendromu - VPS13B (COH1) Mutasyon Analizi	Kan EDTA
☐ Distoni Tip 1 - TORIA Tüm Gen Dizi Analizi	Kan EDTA
☐ Dravet Sendromu - SCN1A Tüm Gen Dizi Analizi	Kan EDTA
☐ Duchenne / Becker Musküler Distrofi Delesyon Analizi (79 Ekzon)	Kan EDTA
☐ Duchenne / Becker Musküler Distrofi Taşıyıcılık Analizi	Kan EDTA
☐ Faktör V Cambridge Mutasyon Analizi	Kan EDTA
☐ Faktör V H1299R Mutasyon Analizi	Kan EDTA
☐ Fenilketonüri (PKU) - PAH Mutasyon Analizi (6 Mutasyon)	Kan EDTA
☐ Frajil X Sendromu - FMR1 CGG Tekrarı Genetik Analizi	Kan EDTA
☐ Galaktozemi - GALT Tüm Gen Dizi Analizi	Kan EDTA
☐ Gaucher Hastalığı - GBA Tüm Gen Dizi Analizi	Kan EDTA
☐ Glutarik Asidüri Tip 1 - GCDH Tüm Gen Dizi Analizi	Kan EDTA
☐ GM1 Gangliosidosis Tip 1 - GLB1 Tüm Gen Dizi Analizi	Kan EDTA
☐ GNE İlişkili Miyopati - GNE Tüm Gen Dizi Analizi	Kan EDTA
☐ Hallervorden Spatz Sendromu - PANK2 Tüm Gen Dizi Analizi	Kan EDTA
☐ Herediter Spastik Parapleji Tip 4 - SPASTIN (SPAST) Tüm Gen Dizi Analizi	Kan EDTA
☐ HLA B51 PCR	Kan EDTA
☐ Huntington Hastalığı - HD CAG Tekrarı Genetik Analizi	Kan EDTA
☐ Joubert Sendromu Tip 10 - OFD1 Tüm Gen Dizi Analizi	Kan EDTA
☐ Joubert Sendromu Tip 12 - KIF7 Tüm Gen Dizi Analizi	Kan EDTA
☐ Kufor Rakeb Hastalığı - ATP13A2 Tüm Gen Dizi Analizi	Kan EDTA
☐ Konjenital Glikolizasyon Bozukluğu Tip1A (CDG1A) - PMM2 TGDA	Kan EDTA
☐ Krabbe Hastalığı - GALC Tüm Gen Dizi Analizi	Kan EDTA
☐ Lafora Hastalığı - EPM2A Tüm Gen Dizi Analizi	Kan EDTA
☐ Leigh Sendromu - SURF1 Tüm Gen Dizi Analizi	Kan EDTA
☐ Limb-Girdle Musküler Distrofi 2D - SGCA Tüm Gen Dizi Analizi	Kan EDTA
☐ Limb-Girdle Musküler Distrofi 2E - SGCB Tüm Gen Dizi Analizi	Kan EDTA
☐ Maple Syrup Urine Hastalığı Tip 1A - BCKDHA Tüm Gen Dizi Analizi	Kan EDTA
☐ Maple Syrup Urine Hastalığı Tip 1B - BCKDHB Tüm Gen Dizi Analizi	Kan EDTA
☐ Melas - MT-TL1 Mutasyon Analizi	Kan EDTA
☐ Miyofibriler Miyopati 4 - ZASP (LDB3) Tüm Gen Dizi Analizi	Kan EDTA
☐ Miyotonik Distrofi Tip 1 - DMPK CTG Tekrarı Genetik Analizi	Kan EDTA
☐ NCL1 İnfantil Tip - PPT1 Mutasyon Analizi	Kan EDTA
☐ NCL2 Geç İnfantil Tip - TPP1 Mutasyon Analizi	Kan EDTA
☐ Nemalin Miyopati 1 - TPM3 Tüm Gen Dizi Analizi	Kan EDTA
☐ Noonan Sendromu - PTPN11 (PTP2) Tüm Gen Dizi Analizi	Kan EDTA
☐ Nöroferrinopati - FTL Tüm Gen Dizi Analizi	Kan EDTA
☐ Nörofibromatozis Tip 1 - NF1 Tüm Gen Dizi Analizi	Kan EDTA
☐ Parietal Foramina Tip 2 - ALX4 Tüm Gen Dizi Analizi	Kan EDTA
☐ Prion Hastalığı - PRNP Tüm Gen Dizi Analizi	Kan EDTA
☐ Rett Sendromu - MECP2 Tüm Gen Dizi Analizi	Kan EDTA
☐ Roberts Sendromu - ESCO2 Tüm Gen Dizi Analizi	Kan EDTA
☐ Sepiapterin Redüktaz Eksikliği - SPR Tüm Gen Dizi Analizi	Kan EDTA
☐ Smith Lemli Opitz (SLO) Sendromu - DHCR7 Tüm Gen Dizi Analizi	Kan EDTA
☐ Snyder Robinson Sendromu - SMS Tüm Gen Dizi Analizi	Kan EDTA
☐ Sotos Sendromu - NSD1 Tüm Gen Dizi Analizi	Kan EDTA
☐ Spinal Musküler Distrofi - SMN1 Delesyon Analizi (7,8)	Kan EDTA
☐ Spinal Musküler Distrofi - SMN1 Tüm Gen Dizi Analizi	Kan EDTA
☐ Spinal Musküler Distrofi - SMN1 Taşıyıcılık Analizi	Kan EDTA
☐ Spinoserebellar Ataksi Tip 1 - ATXN1 CAG Tekrarı	Kan EDTA
☐ Spinoserebellar Ataksi Tip 2 - ATXN2 CAG Tekrarı	Kan EDTA
☐ Spinoserebellar Ataksi Tip 3 - ATXN3 CAG Tekrarı	Kan EDTA
☐ Spinoserebellar Ataksi Tip 6 - CACNA1A CAG Tekrarı	Kan EDTA
☐ Spinoserebellar Ataksi Tip 7 - ATXN7 CAG Tekrarı	Kan EDTA
☐ Spinoserebellar Ataksi Tip 8 - ATXN80S Mutasyon Analizi	Kan EDTA
☐ Spinoserebellar Ataksi Tip 10 - ATXN10 Mutasyon Analizi	Kan EDTA

MOLEKÜLER GENETİK TESTLER	NUMUNE TÜRÜ
☐ Spinoserebellar Ataksi Tip 12 - PPP2R2B CAG Tekrarı	Kan EDTA
☐ Spinoserebellar Ataksi Tip 17 - TBP CAA/CAG Tekrarı	Kan EDTA
☐ Tüberoz Skleroz Tip 1 - TSC1 Tüm Gen Dizi Analizi	Kan EDTA
☐ Tüberoz Skleroz Tip 2 - TSC2 Tüm Gen Dizi Analizi	Kan EDTA
☐ Von Hippel Lindau Sendromu - VHL Tüm Gen Dizi Analizi	Kan EDTA
☐ Wilson Hastalığı - ATP7B (13 Ekzon) Dizi Analizi	Kan EDTA
☐ Wolfram Sendromu - WFS1 Tüm Gen Dizi Analizi	Kan EDTA
☐ X İlişkili Adrenolökodistrofi - ABCD1 Tüm Gen Dizi Analizi	Kan EDTA

MOLEKÜLER SİTOGENETİK TESTLER	NUMUNE TÜRÜ
☐ 1p36 Delesyon Sendromu FISH Analizi	Kan EDTA
☐ Angelman Sendromu (del 15q11-q13) FISH Analizi	Kan EDTA
☐ Cri Du Chat Sendromu (del 5p15.2) FISH Analizi	Kan EDTA
☐ Di George Sendromu (10p) FISH Analizi	Kan EDTA
☐ Di George Sendromu / VCF Sendromu FISH Analizi	Kan EDTA
☐ N-MYC Nöroblastoma (2p24.1) FISH Analizi	Kan EDTA
☐ Prader Willi / Angelman Sendromu FISH Analizi	Kan EDTA
☐ Rubinstein Taybi Sendromu (del 16p13.3) FISH Analizi	Kan EDTA
☐ Smith Magenis Sendromu (del 17p11.2-RAI1) FISH Analizi	Kan EDTA
☐ Sotos Sendromu (del 5q35-NSD1) FISH Analizi	Kan EDTA
☐ Williams Sendromu (del 7q11.23) FISH Analizi	Kan EDTA
☐ Wolf Hirschhorn Sendromu - WHCR FISH Analizi	Kan EDTA
☐ İktiyozis Steroid Sülfataz (del Xp22.32-STS) FISH Analizi	Kan EDTA

MOLEKÜLER ENFEKSİYON TESTLERİ	NUMUNE TÜRÜ
☐ Brucella DNA PCR	BOS, Doku, Kan EDTA
☐ CMV DNA PCR KANTİTATİF	A.S., BOS, İdrar, Kan EDTA
☐ HSV Tip 1 DNA PCR	BOS, Serum, Swap, Kan EDTA
☐ HSV Tip 2 DNA PCR	BOS, Serum, Swap, Kan EDTA
☐ İnfluenza A, B Virüs DNA PCR	BOS, Serum, Swap
☐ Rubella RNA PCR	Amniyon sıvısı, Kan EDTA
☐ TBC DNA PCR	BAL, Balgam, BOS
☐ Varicella Zoster Virüs DNA PCR	BOS, Serum, Kan EDTA
☐ Panfungal DNA PCR	BOS, Serum, Kan EDTA
☐ Treponema Palladium DNA PCR	BOS, Serum, Kan EDTA

TROMBOFİLİ PANELİ	NUMUNE TÜRÜ
☐ Faktör V Leiden Mutasyon Analizi	Kan EDTA
☐ Protrombin (F2) Mutasyon Analizi (G20210A)	Kan EDTA
☐ MTHFR Mutasyon Analizi (C677T)	Kan EDTA
☐ MTHFR Mutasyon Analizi (A1298C)	Kan EDTA
☐ Faktör XIII V34L Mutasyon Analizi	Kan EDTA
☐ PAI-SERPINE1 Mutasyon Analizi (4G&5G)	Kan EDTA

YENİ NESİL DİZİLEME PANELLERİ	NUMUNE TÜRÜ
☐ Aicardi Goutieres Sendromu Tarama Paneli (6 Gen)	Kan EDTA
☐ Alzheimer Hastalığı / Frontotemporal Demans Tarama Paneli (47 Gen)	Kan EDTA
☐ Amiloid İlişkili Hastalıklar Tarama Paneli (17 Gen)	Kan EDTA
☐ Amiyotrofik Lateral Skleroz Tarama Paneli (25 Gen)	Kan EDTA
☐ Charcot-Marie-Tooth Hastalığı Tarama Paneli (39 Gen)	Kan EDTA
☐ Epilepsi Gen Paneli (219 Gen)	Kan EDTA
☐ Hareket Bozuklukları Gen Paneli (174 Gen)	Kan EDTA
☐ Hiperekpleksia Paneli O.D. & O.R. (5 Gen)	Kan EDTA
☐ Kapsamlı Distoni Tarama Paneli (85 Gen)	Kan EDTA
☐ Kapsamlı Müsküler Distrofi / Miyopati Tarama Paneli (164 Gen)	Kan EDTA
☐ Kas Bozuklukları Gen Paneli (115 Gen)	Kan EDTA
☐ Konjenital Miyastenik Sendrom Tarama Paneli (12 Gen)	Kan EDTA
☐ Kontraktür Tarama Paneli (Fetal Akinezi, Artrogripozis) (141 Gen)	Kan EDTA
☐ Metabolik Hastalıklar Gen Paneli (498 Gen)	Kan EDTA
☐ Mitokondrial Gen Paneli (206 Gen)	Kan EDTA
☐ Nöropati Paneli (59 Gen)	Kan EDTA
☐ Parkinson Hastalığı / Parkinsonizm Tarama Paneli (71 Gen)	Kan EDTA
☐ Spastik Parapleji Tarama Paneli (47 Gen)	Kan EDTA
☐ Tüm Mitokondrial DNA Dizi Analizi	Kan EDTA
☐ Tüm Ekzom Dizi Analizi (WES)	Kan EDTA

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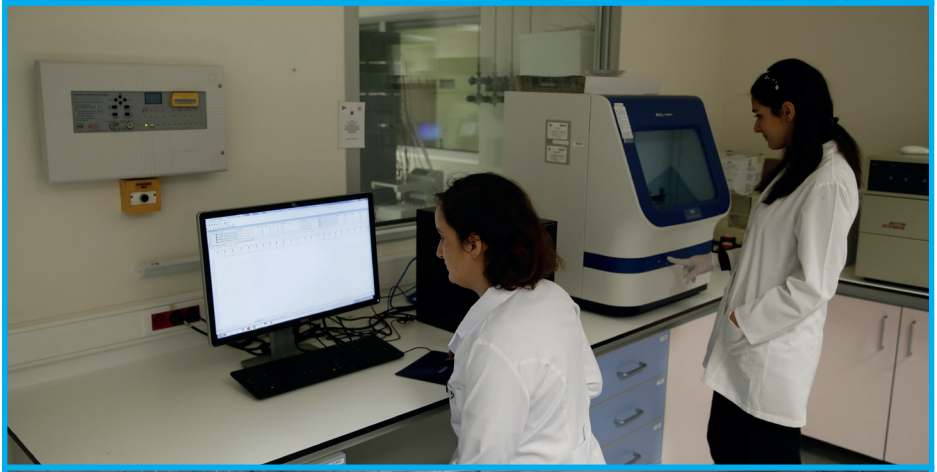


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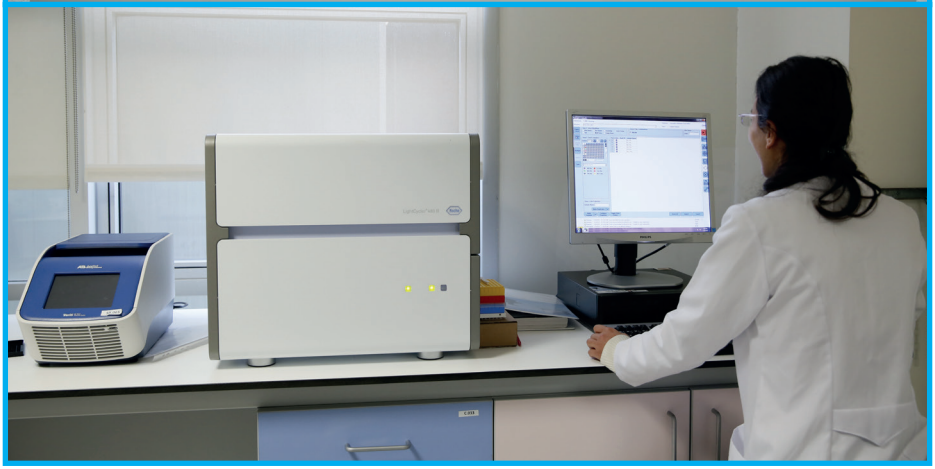

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