ENDOCRINE & METABOLISM DISORDERS TEST CATALOGUE

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

MEDIPOL UNV ISTANBUL MEDIPOL UNIVERSITY

CEQAS | EMQN The European Molecular Genetics Quality Network | QCMD | CAP ACCREDITED

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

Methods Used in Our Center with Comprehensive Test Lists:

- 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

ENDOCRINE & METABOLISM DISORDERS TEST LIST

MOLECULAR GENETIC TESTS

Test Name	Gene Name	Test Methods	Sample
11 Beta Hydroxylase Deficiency	CYP11B1	Whole Gene Analysis	Blood[EDTA]
17 Beta Hydroxysteroid Dehydrogenase X Deficiency	HSD17B10	Whole Gene Analysis	Blood[EDTA]
21 Hydroxylase Deficiency	CYP21A2	Whole Gene Analysis	Blood[EDTA]
21 Hydroxylase Deficiency	CYP21A2	Deletion & Duplication Analysis	Blood[EDTA]
3 Hydroxyacyl CoA Dehydrogenase Deficiency	HADHA	Whole Gene Analysis	Blood[EDTA]
5 Alpha Reductase Deficiency	SRD5A2	Whole Gene Analysis	Blood[EDTA]
Alkaptonuria	HGD (HGO)	Whole Gene Analysis	Blood[EDTA]
Alstrom Syndrome	ALMS1	Whole Gene Analysis	Blood[EDTA]
Amyloidosis	TTR	Whole Gene Analysis	Blood[EDTA]
Androgen Insensitivity Syndrome	AR	Whole Gene Analysis	Blood[EDTA]
Apert Syndrome	FGFR2	Whole Gene Analysis	Blood[EDTA]
Arginase Deficiency	ARG1	Whole Gene Analysis	Blood[EDTA]
Aromatase Deficiency Syndrome, AROD	CYP19A1	Mutation Analysis (1558C>T)	Blood[EDTA]
Autoimmune Polyendocrinopathy Syndrome	AIRE	Whole Gene Analysis	Blood[EDTA]
Bardet Biedl Syndrome	BBS1	Whole Gene Analysis	Blood[EDTA]
Bardet Biedl Syndrome	BBS10	Whole Gene Analysis	Blood[EDTA]
Bardet Biedl Syndrome	BBS2	Whole Gene Analysis	Blood[EDTA]
Berardinelli Seip Congenital Lipodystrophy Type 1	AGPAT2	Whole Gene Analysis	Blood[EDTA]
Berardinelli Seip Congenital Lipodystrophy Type 2	BSCL2	Whole Gene Analysis	Blood[EDTA]
Biotinidase Deficiency	BTD	Whole Gene Analysis	Blood[EDTA]
Canavan Disease	ASPA	Whole Gene Analysis	Blood[EDTA]
Carbamoyl Phosphate Synthetase Deficiency	CPS1	Whole Gene Analysis	Blood[EDTA]
Cerebrotendinous Xanthomatosis	CYP27A1	Whole Gene Analysis	Blood[EDTA]
Chanarin Dorfman Syndrome	ABHD5	Whole Gene Analysis	Blood[EDTA]
Citrullinemia Type 1	ASS1	Whole Gene Analysis	Blood[EDTA]
Combined Pituitary Hormone Deficiency	LHX4	Whole Gene Analysis	Blood[EDTA]
Combined Pituitary Hormone Deficiency Type 1	POU1F1	Whole Gene Analysis	Blood[EDTA]
Combined Pituitary Hormone Deficiency Type 2	PROP1	Whole Gene Analysis	Blood[EDTA]
Congenital Adrenal Hypoplasia	NROB1 (DAX1)	Whole Gene Analysis	Blood[EDTA]
Congenital Hypothyroidism Type 2	PAX8	Whole Gene Analysis	Blood[EDTA]

ENDOCRINE & METABOLISM DISORDERS TEST LIST

MOLECULAR GENETIC TESTS

Test Name	Gene Name	Test Methods	Sample
Congenital Lipoid Adrenal Hyperplasia	STAR	Whole Gene Analysis	Blood^{EDTA}
Cystic Fibrosis	CFTR	Whole Gene Analysis	Blood^{EDTA}
Cystic Fibrosis	CFTR	Deletion & Duplication Analysis	Blood^{EDTA}
D-2-Hydroxy Glutaric Aciduria	D2HGDH	Whole Gene Analysis	Blood^{EDTA}
Fabry Disease	GLA	Whole Gene Analysis	Blood^{EDTA}
Familial Hypercholesterolemia	LDLR	Whole Gene Analysis	Blood^{EDTA}
Familial Hyperinsulinemic Hypoglycemia 5	INSR	Whole Gene Analysis	Blood^{EDTA}
Familial Hyperlipidemia	LPL	Whole Gene Analysis	Blood^{EDTA}
Familial Hypertriglyceridemia	APOA5	Whole Gene Analysis	Blood^{EDTA}
Familial Hypertriglyceridemia	LIPI	Whole Gene Analysis	Blood^{EDTA}
Familial Isolated Hypoparathyroidism	CASR	Whole Gene Analysis	Blood^{EDTA}
Familial Medullar Thyroid Carcinoma	RET	Whole Gene Analysis	Blood^{EDTA}
Fanconi Bickel Syndrome	GLUT2	Whole Gene Analysis	Blood^{EDTA}
Fructose Intolerance	ALDOB	Whole Gene Analysis	Blood^{EDTA}
Fructose-1,6-Bisphosphatase Deficiency	FBP1	Whole Gene Analysis	Blood^{EDTA}
Galactosemia	GALT	Whole Gene Analysis	Blood^{EDTA}
Gaucher Disease	GBA	Whole Gene Analysis	Blood^{EDTA}
Glucocorticoid Resistance	NR3C1	Whole Gene Analysis	Blood^{EDTA}
Glucose Transporter Defect Syndrome Type 1	SLC2A1 (GLUT1)	Whole Gene Analysis	Blood^{EDTA}
Glutaric Aciduria Type 1	GCDH	Whole Gene Analysis	Blood^{EDTA}
Glycogen Storage Disease Type 1A	G6PC	Whole Gene Analysis	Blood^{EDTA}
Glycogen Storage Disease Type 1B	SLC37A4	Whole Gene Analysis	Blood^{EDTA}
Glycogen Storage Disease Type 2	GAA	Whole Gene Analysis	Blood^{EDTA}
Glycogen Storage Disease Type 3	AGL	Whole Gene Analysis	Blood^{EDTA}
Glycogen Storage Disease Type 4	GBE1	Whole Gene Analysis	Blood^{EDTA}
Glycogen Storage Disease Type 6	PYGL	Whole Gene Analysis	Blood^{EDTA}
Glycogen Storage Disease Type 9A	PHKA2	Whole Gene Analysis	Blood^{EDTA}
Glycogen Storage Disease Type 9C	PHKG2	Whole Gene Analysis	Blood^{EDTA}
GM1 Gangliosidosis Type 1	GLB1	Whole Gene Analysis	Blood^{EDTA}
GNE Gene Analysis	GNE	Whole Gene Analysis	Blood^{EDTA}

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MOLECULAR GENETIC TESTS

Test Name	Gene Name	Test Methods	Sample
Gonadotropin Releasing Hormone Deficiency, Isolated	GNRHR	Whole Gene Analysis	Blood^{EDTA}
HDR Syndrome	GATA3	Whole Gene Analysis	Blood^{EDTA}
Hemochromatosis - (18 Mutations)	HFE	Mutation Analysis	Blood^{EDTA}
HMG CoA Lyase Deficiency	HMGCL	Whole Gene Analysis	Blood^{EDTA}
Holt Oram Syndrome	TBX5	Whole Gene Analysis	Blood^{EDTA}
Homocystinuria - (844ins68bp)	CBS	Mutation Analysis	Blood^{EDTA}
Hyper lgD Syndrome	MVK	Whole Gene Analysis	Blood^{EDTA}
Hypercalcemia, Infantile	CYP24A1	Whole Gene Analysis	Blood^{EDTA}
Hyperphenylalaninemia BH4 Deficient Type A	PTS	Whole Gene Analysis	Blood^{EDTA}
Hypomagnesemia with Secondary Hypocalcemia	TRPM6	Mutation Analysis	Blood^{EDTA}
Hypoparathyroidism, Familial	GCMB	Whole Gene Analysis	Blood^{EDTA}
Hypophosphatemia	PHEX	Whole Gene Analysis	Blood^{EDTA}
Insulin Like Growth Factor 1	IGF1	Whole Gene Analysis	Blood^{EDTA}
Krabbe Disease	GALC	Whole Gene Analysis	Blood^{EDTA}
Krabbe Disease - (Exons 11 - 17)	GALC	Deletion Analysis	Blood^{EDTA}
L-2-Hydroxyglutaric Aciduria	L2HGDH	Whole Gene Analysis	Blood^{EDTA}
Lecithin Cholesterol Acyltransferase Deficiency	LCAT	Whole Gene Analysis	Blood^{EDTA}
Leigh Syndrome	SURF1	Whole Gene Analysis	Blood^{EDTA}
Liddle Syndrome	SCNN1B	Whole Gene Analysis	Blood^{EDTA}
Lowe Syndrome	OCRL	Whole Gene Analysis	Blood^{EDTA}
Malignant Hyperthermia	RYR1	Whole Gene Analysis	Blood^{EDTA}
Maple Syrup Urine Disease Type 1A	BCKDHA	Whole Gene Analysis	Blood^{EDTA}
Maple Syrup Urine Disease Type 1B	BCKDHB	Whole Gene Analysis	Blood^{EDTA}
Maple Syrup Urine Disease Type 2	DBT	Whole Gene Analysis	Blood^{EDTA}
McCune Albright Syndrome	GNAS	Whole Gene Analysis	Blood^{EDTA}
Medium Chain Acyl Coenzyme A Dehydrogenase Deficiency	ACADM	Whole Gene Analysis	Blood^{EDTA}
MELAS Genetic Analysis	MT-TL1	Whole Gene Analysis	Blood^{EDTA}
Methylmalonic Acidemia	MMAA	Whole Gene Analysis	Blood^{EDTA}
Methylmalonic Acidemia	MMAB	Whole Gene Analysis	Blood^{EDTA}
Methylmalonic Acidemia	MUT	Whole Gene Analysis	Blood^{EDTA}

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MOLECULAR GENETIC TESTS

Test Name	Gene Name	Test Methods	Sample
MODY Type 1	HNF4A	Whole Gene Analysis	Blood^{EDTA}
MODY Type 2	GCK	Whole Gene Analysis	Blood^{EDTA}
MODY Type 2	GCK	Deletion & Duplication Analysis	Blood^{EDTA}
MODY Type 3	HNF1A	Whole Gene Analysis	Blood^{EDTA}
MODY Type 4	IPF1 (PDX1)	Whole Gene Analysis	Blood^{EDTA}
MODY Type 5	HNF1B	Whole Gene Analysis	Blood^{EDTA}
MODY Type 5	HNF1B	Deletion & Duplication Analysis	Blood^{EDTA}
MTHFR Mutation Analysis (A1298C)	MTHFR	Mutation Analysis	Blood^{EDTA}
MTHFR Mutation Analysis (C677T)	MTHFR	Mutation Analysis	Blood^{EDTA}
Mucolipidosis Type 2	GNPTAB	Whole Gene Analysis	Blood^{EDTA}
Mucopolysaccharidosis Type 1	IDUA	Whole Gene Analysis	Blood^{EDTA}
Mucopolysaccharidosis Type 2	IDS	Whole Gene Analysis	Blood^{EDTA}
Mucopolysaccharidosis Type 3A	SGSH	Whole Gene Analysis	Blood^{EDTA}
Multiple Acyl-CoA Dehydrogenase Deficiency	ETFA	Whole Gene Analysis	Blood^{EDTA}
Multiple Acyl-CoA Dehydrogenase Deficiency	ETFB	Whole Gene Analysis	Blood^{EDTA}
Multiple Acyl-CoA Dehydrogenase Deficiency	ETFDH	Whole Gene Analysis	Blood^{EDTA}
Multiple Endocrine Neoplasia Type 1	MEN1	Whole Gene Analysis	Blood^{EDTA}
Multiple Endocrine Neoplasia Type 2A, 2B	RET	Whole Gene Analysis	Blood^{EDTA}
Multiple Endocrine Neoplasia Type 4	CDKN1B	Whole Gene Analysis	Blood^{EDTA}
Multiple Sulfatase Deficiency	SUMF1	Whole Gene Analysis	Blood^{EDTA}
N-Acetylglutamate Synthase Deficiency	NAGS	Whole Gene Analysis	Blood^{EDTA}
NCL1 Infantile Type	PPT1	Mutation Analysis	Blood^{EDTA}
NCL2 Late Infantile Type	PPT1	Mutation Analysis	Blood^{EDTA}
NCL2 Late Infantile Type	TPP1	Mutation Analysis	Blood^{EDTA}
NCL3 Juvenile Type	CLN3	Deletion Analysis	Blood^{EDTA}
Nephropatic Cystinosis	CTNS	Whole Gene Analysis	Blood^{EDTA}
Neutral Lipid Storage Disease with Myopathy	PNPLA2	Whole Gene Analysis	Blood^{EDTA}
Niemann Pick Disease Type A, B	SMPD1	Whole Gene Analysis	Blood^{EDTA}
Niemann Pick Disease Type C	NPC1	Whole Gene Analysis	Blood^{EDTA}
Niemann Pick Disease Type C2	NPC2	Whole Gene Analysis	Blood^{EDTA}

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MOLECULAR GENETIC TESTS

Test Name	Gene Name	Test Methods	Sample
Nonketotic Hyperglycinemia	GLDC	Whole Gene Analysis	Blood^{EDTA}
Nonketotic Hyperglycinemia	GLDC	Deletion & Duplication Analysis	Blood^{EDTA}
Noonan Syndrome	PTPN11 (PTP2)	Whole Gene Analysis	Blood^{EDTA}
NR5A1 Gene Analysis	NR5A1	Whole Gene Analysis	Blood^{EDTA}
Obesity	MC4R	Whole Gene Analysis	Blood^{EDTA}
Ornithine Transcarbomylase Deficiency	OTC	Whole Gene Analysis	Blood^{EDTA}
Osteogenesis Imperfecta Type 1, 2A, 3, 4	COL1A1	Whole Gene Analysis	Blood^{EDTA}
Osteogenesis Imperfecta Type 1, 2A, 3, 4	COL1A2	Whole Gene Analysis	Blood^{EDTA}
Osteogenesis Imperfecta	CRTAP	Whole Gene Analysis	Blood^{EDTA}
Osteogenesis Imperfecta	LEPRE1	Whole Gene Analysis	Blood^{EDTA}
Paraganglioma Type 1	SDHD	Whole Gene Analysis	Blood^{EDTA}
Paraganglioma Type 3	SDHC	Whole Gene Analysis	Blood^{EDTA}
Paraganglioma Type 4	SDHB	Whole Gene Analysis	Blood^{EDTA}
Pendred Syndrome	FOXI1	Whole Gene Analysis	Blood^{EDTA}
Pendred Syndrome	KCNJ10	Whole Gene Analysis	Blood^{EDTA}
Pendred Syndrome	SLC26A4	Whole Gene Analysis	Blood^{EDTA}
Pendred Syndrome	SLC26A4	Deletion & Duplication Analysis	Blood^{EDTA}
Phenylketonuria	PAH	Whole Gene Analysis	Blood^{EDTA}
Pierre Robin Syndrome	SOX9	Whole Gene Analysis	Blood^{EDTA}
Polycystic Liver Disease	PRKCSH & SEC63	Whole Gene Analysis	Blood^{EDTA}
Primary Hyperoxaluria Type 1	AGXT	Whole Gene Analysis	Blood^{EDTA}
Primary Hyperoxaluria Type 2	GRHPR	Whole Gene Analysis	Blood^{EDTA}
Primary Systemic Carnitine Deficiency	SLC22A5	Whole Gene Analysis	Blood^{EDTA}
Primary Systemic Carnitine Deficiency	SLC22A5	Deletion & Duplication Analysis	Blood^{EDTA}
Propionic Acidemia	PCCA	Whole Gene Analysis	Blood^{EDTA}
Propionic Acidemia	PCCB	Whole Gene Analysis	Blood^{EDTA}
Pyridoxine Deficiency	ALDH7A1	Whole Gene Analysis	Blood^{EDTA}
Nonketotic Hyperglycinemia	GLDC	Whole Gene Analysis	Blood^{EDTA}
Nonketotic Hyperglycinemia	GLDC	Deletion & Duplication Analysis	Blood^{EDTA}
Sandhoff Disease	HEXB	Whole Gene Analysis	Blood^{EDTA}

ENDOCRINE & METABOLISM DISORDERS TEST LIST

MOLECULAR GENETIC TESTS

Test Name	Gene Name	Test Methods	Sample
Septooptic Dysplasia	HESX1	Whole Gene Analysis	Blood[EDTA]
Short Chain Acyl CoA Dehydrogenase Deficiency	ACADS	Whole Gene Analysis	Blood[EDTA]
SHOX Gene Analysis	SHOX	Whole Gene Analysis	Blood[EDTA]
Shwachman - Diamond Syndrome	SBDS	Whole Gene Analysis	Blood[EDTA]
Smith Lemli Opitz Syndrome	DHCR7	Whole Gene Analysis	Blood[EDTA]
SRY Gene Analysis	SRY	PCR	Blood[EDTA]
Succinic Semialdehyde Dehydrogenase Deficiency	ALDH5A1	Whole Gene Analysis	Blood[EDTA]
Tangier Disease	ABCA1	Whole Gene Analysis	Blood[EDTA]
Tay Sachs Syndrome	HEXA	Whole Gene Analysis	Blood[EDTA]
Thyroid Hormone Resistance	THRB	Whole Gene Analysis	Blood[EDTA]
TPO Gene Analysis	TPO	Whole Gene Analysis	Blood[EDTA]
TSH Receptor	TSHR	Whole Gene Analysis	Blood[EDTA]
Tyrosinemia Type 1	FAH	Whole Gene Analysis	Blood[EDTA]
Very Long Chain Acyl CoA Dehydrogenase Deficiency	ACADVL	Whole Gene Analysis	Blood[EDTA]
Wilson Disease - (13 Exons)	ATP7B	Targeted Exons Analysis	Blood[EDTA]
Wolfram Syndrome	WFS1	Whole Gene Analysis	Blood[EDTA]
X Linked Hypohidrotic Ectodermal Dysplasia Type 1	EDA	Whole Gene Analysis	Blood[EDTA]

MOLECULAR CYTOGENETIC TESTS

Test Name	Gene Name	Test Methods	Sample
Array CGH	All Chromosomes	Microarray	Blood[EDTA]
Choromosome X, Y Analysis	Chromosome X,Y	FISH Analysis	Blood[HEP]
Prader Willi/Angelman Syndrome	15q11-q13 del	FISH Analysis	Blood[HEPA]
SRY Analysis	SRY	FISH	Blood[HEP]

ENDOCRINE & METABOLISM DISORDERS TEST LIST

NEXT GENERATİON SEQUENCİNG (NGS) PANELS

Test Name	Gene Name	Sample
Whole Exome Sequencing (WES)	Whole Exome	Blood^{EDTA}
46,XY Disorders of Sex Development / Complete Gonadal Dysgenesis Sequencing Panel	AKR1C2, AMH, AMHR2, AR, ARX, ATRX, B3GALTL, CYB5A, CYP11A1, CYP17A1, DHCR7, DHH, DYNC2H1, GATA4, HCCS, HSD17B3, LHCGR, MAMLD1, MAP3K1, NR5A1, OPHN1, SOX9, SRD5A2, SRY, WT1, ZFPM2	Blood^{EDTA}
Abnormal / Ambiguous Genıtalia Sequencing Panel	AKR1C2, AR, ARX, ATRX, B3GALTL, BCOR, BMP4, CDKN1C, CEP41, CHD7, CREBBP, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP19A1, CYP21A2, DHCR24, DHCR7, DHH, DNMT3B, DYNC2H1, ESCO2, FAM58A, FAT4 FEZF1, FIG4, FRAS1, FREM2, GATA4, GRIP1, HCCS, HOXA13, HSD17B3, HSD3B2, ICK, IL17RD, IRF6, KAL1, KISS1R, LHCGR, LMNA, MAP3K1, MKKS, MKS1, NEK1, NR0B1, NR5A1, NSMF, OPHN1, POR, PTPN11, RIPK4, ROR2, RSPO1, SALL1, SCARF2, SEMA3A, SETBP1, SOX9, SPECC1L, SRD5A2, SRY, STAR, TBX15, TCTN3, TSPYL1, UBR1, WDR60, WNT4, WNT7A WT1, ZFPM2	Blood^{EDTA}
Chronic Pancreatitis Panel	CASR, CFTR, CTRC, PRSS1, SPINK1	Blood^{EDTA}
Endocrine Disorders Sequencing Panel	ABCC8, AGPAT2, AKT2, BLK, BMP15, BSCL2, CHD7, CIDEC, CISD2, CYP17A1, CYP19A1, EIF2AK3, FGF8, FGFR1, FIGLA, FOXP3, FSHR, GATA6, GCK, GDF9, GLIS3, GNRH1, GNRHR, HADH, HNF1A, HNF1B, HNF4A, IER3IP1, INS, INSR, KCNJ11, KISS1, KISS1R, KLF11, LHCGR, LMNA, NEUROD1, NOBOX, NR0B1, NR5A1, NSMF, PAX4, PDX1, POR, PPARG, PROK2, PROKR2, PSMC3IP, PTF1A, PTRF, RFX6, SLC2A2, TAC3, TACR3, TBC1D4, WFS1, ZMPSTE24	Blood^{EDTA}
Glycogen Storage Diseases	AGL, ALDOA, ENO3, G6PC, GAA, GBE1, GYG1, GYS1, LAMP2, LDHA, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PYGL, PYGM, SLC2A2, SLC37A4	Blood^{EDTA}

ENDOCRINE & METABOLISM DISORDERS TEST LIST

NEXT GENERATİON SEQUENCİNG (NGS) PANELS

Test Name	Gene Name	Sample
Hypogonadotropic Hypogonadism (HH) and Related Disorders NGS Panel	ANOS1, CHD7, FGF8, FGFR1, GNRH1, GNRHR, KISS1, KISS1R, NR0B1, NSMF, PROKR2, TAC3, TACR3	Blood^{EDTA}
Hypospadias Sequancing Panel	AR, ARX, ATRX, B3GALTL, BCOR, BMP4, CDKN1C, CREBBP, CUL7, CYP11A1, CYP21A2, DHCR7, DNMT3B, EFNB1, EPG5, ESCO2, EVC, EVC2, FAT4, FBXL4, FGF10, FGFR1, FGFR2, FGFR3, FIG4, FLNA, FRAS1, FREM2, GLI3, GPC3, GRIP1, HBA1, HCCS, HNF1B, HOXA13, HSD3B2, IRF6, MAMLD1, MAP3K1, MED12, MID1, MKKS, NR5A1, PCNT, PDE4D, PEX1, PITX2, PTDSS1, PTPN11, RBBP8, SALL1, SETBP1, SOX2, SPECC1L, SRD5A2, TMEM70, TP63, UBR1, WDR35, WNT7A, WT1, ZEB2	Blood^{EDTA}
Kallmann Syndrome Panel	ANOS1, CHD7, FGF8, FGFR1, GNRH1, GNRHR, KISS1, KISS1R, NSMF, PROKR2, SEMA3A, TAC3, TACR3	Blood^{EDTA}
Multiple Endocrine Neoplasias / Paraganglioma / Pheochromacytoma Panel	CDKN1B, MAX, MEN1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL	Blood^{EDTA}
Pancreatitis Panel	CFTR, CPA1, CTRC, PRSS1, SPINK1	Blood^{EDTA}
Premature Ovarian Failure Sequencing Panel	BMP15, CYP17A1, CYP19A1, FIGLA, FSHR, GDF9, LHCGR, NOBOX, NR5A1, POR, PSMC3IP	Blood^{EDTA}
Thyroid Cancer Genetic Panel	BRAF V600E, KRAS Codon 12/13, NRAS Codon 61, HRAS Codon 12/13/61, RET / PTC1, RET / PTC3, PAX8 / PPARg	Paraffin Blocks

WHOLE EXOME SEQUENCING (WES)

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

KAVACIK MH. EKİNCİLER CAD. NO:19 34810 - BEYKOZ-ISTANBUL / TURKEY
Call: +90 (216) 681 15 15 | Email: genetik@medipol.edu.tr | Web: genetik.medipol.edu.tr