MEDIPOL
İSTANBUL MEDİPOL UNIVERSITY
GENETIC DISEASES ASSESSMENT CENTER

NEUROLOGY TEST LIST

Please tick the relevant box.
Fill in the Anamnesis & Consent Form on the Back Page.

MOLECULAR GENETIC TESTS	SAMPLE TYPE
☐ Angelman Syndrome Methylation Analysis	Blood[EDTA]
☐ Antithrombin 3 Deficiency - SERPINC1 Full Gene Sequence Analysis	Blood[EDTA]
☐ APO E Genotype Analysis (E2, E3, E4)	Blood[EDTA]
☐ ARX Gene Associated Diseases - ARX Full Gene Sequence Analysis	Blood[EDTA]
☐ Ataxia Telangiectasia - ATM Full Gene Sequence Analysis	Blood[EDTA]
☐ CADASIL Disease - NOTCH3 (Exon 3-6) Sequence Analysis	Blood[EDTA]
☐ Canavan Syndrome - ASPA Full Gene Sequence Analysis	Blood[EDTA]
☐ Charcot-Marie-Tooth Neuropathy X Associated-GJB1 (CX32) Full Gene Sequence Analysis	Blood[EDTA]
☐ Cohen Syndrome - VPS13B (COH1) Mutation Analysis	Blood[EDTA]
☐ Congenital Disorders of Glycosylation Type 1A (CDG1A)-PMM2 Gene Analysis	Blood[EDTA]
☐ DMD/BMD Deletion/Duplication Analysis	Blood[EDTA]
☐ Dravet Syndrome - SCN1A Full Gene Sequence Analysis	Blood[EDTA]
☐ Dystonia Type 1 - TOR1A Full Gene Sequence Analysis	Blood[EDTA]
☐ Factor V H1299R Mutation Analysis	Blood[EDTA]
☐ Fragile X Syndrome - FMR1 CGG Repeat Genetic Analysis	Blood[EDTA]
☐ Galactosemia - GALT Full Gene Sequence Analysis	Blood[EDTA]
☐ Gaucher Disease - GBA Full Gene Sequence Analysis	Blood[EDTA]
☐ Glutaric Aciduria Type 1 - GCDH Full Gene Sequence Analysis	Blood[EDTA]
☐ GM1 Gangliosidosis Type 1 - GLB1 Full Gene Sequence Analysis	Blood[EDTA]
☐ GNE Associated Myopathy - GNE Full Gene Sequence Analysis	Blood[EDTA]
☐ Hallervorden Spatz Syndrome-PANK2 Full Gene Sequence Analysis	Blood[EDTA]
☐ Hereditary Spastic Paraplegia Type 4-SPASTIN (SPAST) Full Gene Sequence Analysis	Blood[EDTA]
☐ HLA B5 (B51, B52) PCR	Blood[EDTA]
☐ Huntington's Disease - HD CAG Repeat Genetic Analysis	Blood[EDTA]
☐ Joubert Syndrome Type 10, 12 - OFD1, KIF7 Full Gene Sequence Analysis	Blood[EDTA]
☐ Known Mutation Analysis	Blood[EDTA]
☐ Krabbe Disease - GALC Full Gene Sequence Analysis	Blood[EDTA]
☐ Kufor Rakeb Disease - ATP13A2 Full Gene Sequence Analysis	Blood[EDTA]
☐ Lafora Disease - EPM2A Full Gene Sequence Analysis	Blood[EDTA]
☐ Leigh Syndrome - SURF1 Full Gene Sequence Analysis	Blood[EDTA]
☐ Limb-Girdle Muscular Dystrophy 2D, 2E-SGCA, SGCB Full Gene Sequence Analysis	Blood[EDTA]
☐ Maple Syrup Urine Disease Type 1A, 1B - BCKDHA, BCKDHB Gene Analysis	Blood[EDTA]
☐ Metachromatic Leukodystrophy Infantile Type-ARSA Full Gene Sequence Analysis	Blood[EDTA]
☐ Molasses - MT-TL1 Mutation Analysis	Blood[EDTA]
☐ Myofibrillary Myopathy 4-ZASP (LDB 3) Full Gene Sequence Analysis	Blood[EDTA]
☐ Myotonic Dystrophy Type 1 - DMPK CTG Repeat Genetic Analysis	Blood[EDTA]
☐ NCL 1 Infantile Type - PPT 1 Mutation Analysis	Blood[EDTA]
☐ NCL2 Late Infantile Type - TPP1 Mutation Analysis	Blood[EDTA]
☐ Nemaline Myopathy 1 - TPM3 Full Gene Sequence Analysis	Blood[EDTA]
☐ Neurofibromatosis Type 1, 2 - NF1, NF2 Full Gene Sequence Analysis	Blood[EDTA]
☐ Noonan Syndrome - PTPN11 (PTP2) Full Gene Sequence Analysis	Blood[EDTA]
☐ Phenylketonuria (PKU) - PAH Full Gene Sequence Analysis	Blood[EDTA]
☐ Prion Disease - PRNP Full Gene Sequence Analysis	Blood[EDTA]
☐ Rett Syndrome - MECP2 Full Gene Sequence Analysis	Blood[EDTA]
☐ Roberts Syndrome - ESCO2 Full Gene Sequence Analysis	Blood[EDTA]
☐ Sepiapterin Reductase Deficiency - SPR Full Gene Sequence Analysis	Blood[EDTA]
☐ Smith Lemli Opitz (SLO) Syndrome-DHCR7 Full Gene Sequence Analysis	Blood[EDTA]
☐ Snyder Robinson Syndrome - SMS Full Gene Sequence Analysis	Blood[EDTA]
☐ Sotos Syndrome - NSD1 Full Gene Sequence Analysis	Blood[EDTA]
☐ Spinal Muscular Atrophy - SMN1 Carrier Analysis	Blood[EDTA]
☐ Spinal Muscular Atrophy - SMN1 Deletion Analysis (7,8)	Blood[EDTA]
☐ Spinocerebellar Ataxia Panel 1 (SCA 1, 2, 3, 6, 7)	Blood[EDTA]
☐ Spinocerebellar Ataxia Panel 2 (SCA 8, 10, 12, 17)	Blood[EDTA]
☐ Tuberous Sclerosis Type 1, 2 - TSC1, TSC2 Full Gene Sequence Analysis	Blood[EDTA]
☐ Von Hippel Lindau Syndrome - VHL Full Gene Sequence Analysis	Blood[EDTA]
☐ Wilson's Disease - ATP7B Sequence Analysis	Blood[EDTA]
☐ Wolfram Syndrome - WFS1 Full Gene Sequence Analysis	Blood[EDTA]
☐ X-Associated Adrenoleukodystrophy - ABCD1 Full Gene Sequence Analysis	Blood[EDTA]

THROMBOPHIA PANEL	SAMPLE TYPE
☐ Factor V Leiden G1691A Mutation Analysis	Blood[EDTA]
☐ Prothrombin G20210A Mutation Analysis	Blood[EDTA]
☐ MTHFR Mutation Analysis (C677T)	Blood[EDTA]
☐ MTHFR Mutation Analysis (A1298C)	Blood[EDTA]
☐ PAI-SERPINE1 Mutation Analysis (4G&5G)	Blood[EDTA]
☐ Factor V H1299R Mutation Analysis	Blood[EDTA]

CYTOGENETIC / MOLECULAR CYTOGENETIC TESTS	SAMPLE TYPE
☐ 1P36 Deletion FISHAnalysis	Blood[EDTA]
☐ Angelman Syndrome (del 15q11-q13) FISH Analysis	Blood[HEP]
☐ ARRAY CGH (Molecular Karyotyping)	Blood[HEP]
☐ Chromosome Analysis	Blood[HEP]
☐ Cri Du Chat Syndrome (del 5p15.2) FISH Analysis	Blood[HEP]
☐ DiGeorge Syndrome / VCF Syndrome FISH Analysis	Blood[HEP]
☐ FISH Analysis of Ichthyosis Steroid Sulfatase (del Xp22.32-STS)	Blood[HEP]
☐ N-MYC Neuroblastoma (2p24.1) FISH Analysis	Blood[HEP]
☐ Prader Willi / Angelman Syndrome FISH Analysis	Blood[HEP]
☐ Smith Magenis Syndrome (del 17p11.2-RAI1) FISH Analysis	Blood[HEP]
☐ Sotos Syndrome (del 5q35-NSD1) FISH Analysis	Blood[HEP]
☐ Williams Syndrome (del 7q11.23) FISH Analysis	Blood[HEP]
☐ Wolf Hirschhorn Syndrome - WHCR FISH Analysis	Blood[HEP]

MOLECULAR MICROBIOLOGY TESTS	SAMPLE TYPE
☐ Influenza A, B Virus RNA PCR	Swap (NF), BAL, Sputum, CSF
☐ Brucella DNA PCR	CSF, Tissue, Blood[EDTA]
☐ CMV DNA PCR Quantitative	CSF, Urine, Blood[EDTA]
☐ HSV Type 1 DNA PCR	CSF, Swap, Blood[EDTA]
☐ HSV Type 2 DNA PCR	CSF, Swap, Blood[EDTA]
☐ Meningitis Panel (PCR)	CSF, Body Fluid
☐ Rubella RNA PCR	Blood[EDTA]
☐ TBC DNA PCR	BAL, Sputum, CSF, Urine
☐ Treponema Palladium DNA PCR	Urine, Swap, Thin Prep
☐ Varicella Zoster Virus (VZV) DNA PCR	CSF, Blood[EDTA]

NEXT GENERATION SEQUENCING (NGS) PANELS	SAMPLE TYPE
☐ Aicardi Goutieres Syndrome Screening Panel (7 Genes)	Blood[EDTA]
☐ Alzheimer's Disease/Frontotemporal Dementia Screening Panel (57 Genes)	Blood[EDTA]
☐ Amyloid-Related Diseases Screening Panel (24 Genes)	Blood[EDTA]
☐ Amyotrophic Lateral Sclerosis Screening Panel (38 Genes)	Blood[EDTA]
☐ Ataxia Gene Panel (579 Genes)	Blood[EDTA]
☐ Autism Spectrum Disorders Gene Panel (199 Genes)	Blood[EDTA]
☐ Charcot-Marie-Tooth (CMT) Disease Screening Panel (39 Genes)	Blood[EDTA]
☐ Comprehensive Dystonia Screening Panel (265 Genes)	Blood[EDTA]
☐ Comprehensive Muscular Dystrophy/Myopathy Screening Panel (296 Genes)	Blood[EDTA]
☐ Contracture Screening Panel (Fetal Akinesia, Arthrogryposis) (197 Genes)	Blood[EDTA]
☐ Dementia Gene Panel (137 Genes)	Blood[EDTA]
☐ Emery-Dreifuss Muscular Dystrophy Gene Panel (6 Genes)	Blood[EDTA]
☐ Epilepsy Gene Panel (434 Genes)	Blood[EDTA]
☐ Epileptic Encephalopathy Gene Panel (66 Genes)	Blood[EDTA]
☐ Episodic Ataxia Panel (10 Genes)	Blood[EDTA]
☐ Generalized Myoclonic Epilepsy Gene Panel (61 Genes)	Blood[EDTA]
☐ Holoprosencephaly Gene Panel (51 Genes)	Blood[EDTA]
☐ Hyperekplexia Panel A.D. & A.R. (10 Genes)	Blood[EDTA]
☐ Infantile Spasm Gene Panel (17 Genes)	Blood[EDTA]
☐ Leukodystrophy and Leukoencephalopathy Gene Panel (142 Genes)	Blood[EDTA]
☐ Limb-Girdle Muscular Dystrophy Gene Panel (22 Genes)	Blood[EDTA]
☐ Lissencephaly Gene Panel (95 Genes)	Blood[EDTA]
☐ Metabolic Diseases Gene Panel (492 Genes)	Blood[EDTA]
☐ Mitochondrial Diseases Gene Panel (241 Genes)	Blood[EDTA]
☐ Movement Disorders Gene Panel (1310 Genes)	Blood[EDTA]
☐ Muscle Disorders Gene Panel (1756 Genes)	Blood[EDTA]
☐ Myasthenia Gene Panel (76 Genes)	Blood[EDTA]
☐ Myofibrillar Myopathy Gene Panel (11 Genes)	Blood[EDTA]
☐ Nemaline Myopathy Gene Panel (11 Genes)	Blood[EDTA]
☐ Neuroid Ceroid Lipofuscinosis (NCL) Gene Panel (11 Genes)	Blood[EDTA]
☐ Neuropathy Gene Panel (467 Genes)	Blood[EDTA]
☐ Parkinson's Disease / Parkinsonism Screening Panel (105 Genes)	Blood[EDTA]
☐ Polymicrogyry Gene Panel (103 Genes)	Blood[EDTA]
☐ Septo Optic Dysplasia Gene Panel (15 Genes)	Blood[EDTA]
☐ Spastic Paraplegia Screening Panel (116 Genes)	Blood[EDTA]
☐ Spinal Muscular Atrophy Gene Panel (33 Genes)	Blood[EDTA]
☐ Clinical Exome Sequence Analysis (6699 Genes)	Blood[EDTA]
☐ Whole Exome Sequence Analysis (WES)	Blood[EDTA]
☐ Whole Genome Sequence Anlaysis (WGS)	Blood[EDTA]
☐ Whole Mitochondrial (mtDNA) DNA Sequence Analysis	Blood[EDTA]

Kavacık District, Ekinciler Street No: 19
Beykoz 34810/Istanbul/Turkey

Web : medigen.medipol.edu.tr
e-mail : genetik@medipol.edu.tr

Tel : 0216 681 15 15