

| Test Name  | Sample                | Amount | Method  | Turn Around Time |
|--|-----------------------|--------|---|------------------|
| 11-Beta-Hydroxylase Deficiency Genetic Analysis                                  | Blood <sup>EDTA</sup> | 5 ml   | CYP11B1 Whole Gene Analysis                                 | 2-3 Months       |
| Aarskog Syndrome   | Blood <sup>EDTA</sup> | 5 ml   | FGD1 Whole Gene Analysis                                    | 2 Months         |
| ACE (Angiotensin I-Converting Enzyme) Genotype Analysis                          | Blood <sup>EDTA</sup> | 5 ml   | I/D Genotyping with PCR+RFLP                                | 5 Days           |
| Achondroplasia Mutation Analysis   | Blood <sup>EDTA</sup> | 5 ml   | G1138A and G1138C substitution in FGFR3 with Tetra-ARMS     | 20 Days          |
| Acute Myeloid Leukemia Genetic Testing Responsiveness to Various FLT3 Inhibitors | Blood <sup>EDTA</sup> | 5 ml   | FLT3 Internal Tandem Duplication and D835 Mutation Analysis | 1-2 Weeks        |
| Acute Myeloid Leukemia Genetic Testing Responsiveness to Various FLT3 Inhibitors | Blood <sup>EDTA</sup> | 5 ml   | FLT3 Internal Tandem Duplication In Exon 14                 | 2-3 Weeks        |
| AGT M235T Genotyping Renal Tubular Dysgenesis (RTD)-AGT M235T                    | Blood <sup>EDTA</sup> | 5 ml   | AGT (SERPINA8) Gene M235T Mutation with PCR+RFLP            | 10 Days          |
| Alagille Syndrome  | Blood <sup>EDTA</sup> | 5 ml   | JAG1 Whole Gene Analysis                                    | 2 Months         |
| Alexander Diseases   | Blood <sup>EDTA</sup> | 5 ml   | GFAP Whole Gene Analysis                                    | 3 Months         |
| Alfa Thalassemia   | Blood <sup>EDTA</sup> | 5 ml   | HBA1, HBA2 sequence variations                              | 2 Months         |
| Alkaptonuria Sequence Analysis   | Blood <sup>EDTA</sup> | 5 ml   | HGD Whole Gene Analysis                                     | 2-3 Months       |
| Alpha Thalassemia Mutation Analysis  | Blood <sup>EDTA</sup> | 5 ml   | Deletions and mutations of HBA1, HBA2                       | 45 Days          |
| Alpha Thalassemia X-related Mental Retardation Syndrome                          | Blood <sup>EDTA</sup> | 5 ml   | ATRX Whole Gene and Splice Junctions                        | 3-4 Months       |
| Alpha-1-Antitrypsin Genotyping A1AT Deficiency                                   | Blood <sup>EDTA</sup> | 5 ml   | Pi*Z, Pi*S, Pi*M Mutation Analysis                          | 20 Days          |
| Alport Syndrome Genetic Analysis (Autosomal Recessive or Dominant)               | Blood <sup>EDTA</sup> | 5 ml   | COL4A3 and COL4A4 Whole Gene Analysis                       | 3-4 Months       |
| Alport Syndrome Genetic Analysis (X-linked)                                      | Blood <sup>EDTA</sup> | 5 ml   | COL4A5 Whole Gene Analysis                                  | 3-4 Months       |
| Alport Syndrome Genetic Analysis (X-linked)                                      | Blood <sup>EDTA</sup> | 5 ml   | COL4A5 Del/Dup Analysis                                     | 2 Months         |
| Alzheimer Disease PSEN1 Genetic Analysis, Alzheimer Type 3                       | Blood <sup>EDTA</sup> | 5 ml   | Deletion of exon 9 in PSEN1                                 | 1-2 Months       |
| Alzheimer Disease PSEN1 Genetic Analysis, Alzheimer Type 3                       | Blood <sup>EDTA</sup> | 5 ml   | PSEN1 sequence variants                                     | 1-2 Months       |
| Alzheimer Disease PSEN2 Genetic Analysis, Alzheimer Type 4                       | Blood <sup>EDTA</sup> | 5 ml   | PSEN2 sequence variants                                     | 1-2 Months       |
| Amyotrophic Lateral Sclerosis (ALS) Panel  | Blood <sup>EDTA</sup> | 5 ml   | SOD1, FUS, TARDBP, ANG Whole gene analysis                  | 2-3 Months       |

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| Amyotrophic Lateral Sclerosis (ANG)   | Blood <sup>EDTA</sup> | 5 ml   | ANG Whole gene analysis                             | 2-3 Months       |
| Amyotrophic Lateral Sclerosis (FUS)   | Blood <sup>EDTA</sup> | 5 ml   | FUS Whole gene analysis                             | 2-3 Months       |
| Amyotrophic Lateral Sclerosis (SOD1)  | Blood <sup>EDTA</sup> | 5 ml   | SOD1 Whole gene analysis                            | 2-3 Months       |
| Amyotrophic Lateral Sclerosis (TARDBP)  | Blood <sup>EDTA</sup> | 5 ml   | TARDBP Whole gene analysis                          | 2-3 Months       |
| Androgen Receptor Gene Analysis<br>Testicular Feminization<br>Androgen Insensitivity Syndrome       | Blood <sup>EDTA</sup> | 5 ml   | AR Whole Gene Analysis                              | 1-2 Months       |
| Angelman Syndrome   | Blood <sup>EDTA</sup> | 5 ml   | UBE3A Whole Gene Analysis                           | 3 Months         |
| Angelman Syndrome   | Blood <sup>EDTA</sup> | 5 ml   | 15. Chromosome Paternal UPD (UPD Analysis)          | 2 Months         |
| Antithrombin 3 Deficiency   | Blood <sup>EDTA</sup> | 5 ml   | SERPINC1 Whole Gene Analysis                        | 2-3 Months       |
| Antithrombin 3 Deficiency   | Blood <sup>EDTA</sup> | 5 ml   | SERPINC1 Del/Dup Analysis                           | 2-3 Months       |
| APC Mutation Analysis<br>Adenomatous Polyposis Of The Colon; APC<br>Familial polyposis of the colon | Blood <sup>EDTA</sup> | 5 ml   | APC Gene Sequence Alterations                       | 2 Months         |
| APC Mutation Analysis<br>Adenomatous Polyposis Of The Colon; APC<br>Familial polyposis of the colon | Blood <sup>EDTA</sup> | 5 ml   | Duplication/deletion of one or more exons           | 2 Months         |
| Apert Syndrome Genetic Analysis<br>Apert-Crouzon Disease  | Blood <sup>EDTA</sup> | 5 ml   | FGFR2 - Detecting point mutations (S252W and P253R) | 1-2 Months       |
| APOE Genotyping<br>Alzheimer Disease Risk Factor  | Blood <sup>EDTA</sup> | 5 ml   | PCR&RFLP ε2, ε3, ε4                                 | 15 Days          |
| APP Genetic Analysis<br>Alzheimer Disease Genetic Analysis  | Blood <sup>EDTA</sup> | 5 ml   | APP Gene Whole Gene Analysis                        | 2-3 Months       |
| APP Genetic Analysis<br>Alzheimer Type 1 (AD1)  | Blood <sup>EDTA</sup> | 5 ml   | APP sequence variants in exons 16 and 17            | 1-2 Months       |

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|---|-----------------------|---------|--|------------------|
| Arginase Deficiency<br>Urea Cycle Disorders   | Blood <sup>EDTA</sup> | 5 ml    | ARG1 Whole Gene Sequencing   | 2-3 Months       |
| Aromatase Deficiency Genetic Analysis   | Blood <sup>EDTA</sup> | 5 ml    | CYP19A1 Gene Targeted Mutation Analysis Polymorphism 1558C -> T  | 2 Months         |
| ARPKD Genetic Analysis<br>Autosomal Recessive Polycystic Kidney Disease   | Blood <sup>EDTA</sup> | 5 ml    | First Phase Exons: 3, 5, 9, 14, 16, 20, 21, 22, 30, 32, 33, 34, 36, 37, 39, 43, 50, 54, 55, 57, 58, 59, 62 | 2 Months         |
| ARPKD Genetic Analysis<br>Autosomal Recessive Polycystic Kidney Disease   | Blood <sup>EDTA</sup> | 5 ml    | Second Phase: Other 43 exons   | 2 Months         |
| ARPKD Genetic Analysis<br>Autosomal Recessive Polycystic Kidney Disease   | Blood <sup>EDTA</sup> | 5 ml    | PKHD1 Whole Gene Analysis  | 2-3 Months       |
| ARSACS Genetic Analysis<br>Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay  | Blood <sup>EDTA</sup> | 5 ml    | SACS Whole Gene Analysis   | 3-4 Months       |
| ARX Gene Related Diseases<br>Partington X-Linked Mental Retardation Syndrome,<br>X-Linked West Syndrome<br>(X-linked Infantile Spasm Syndrome<br>Proud Syndrome | Blood <sup>EDTA</sup> | 5 ml    | ARX Whole Gene Analysis  | 2-3 Months       |
| Arylsulfatase A Deficiency  | Blood <sup>EDTA</sup> | 5 ml    | ARSA gene sequence variants  | 2-3 Months       |
| AT1R CC Genotyping<br>Angiotensin II Receptor, Type 1 (AGTR1)<br>Cardiovascular Risk Factor   | Blood <sup>EDTA</sup> | 5 ml    | PCR&RFLP AT1R Gene A1166C Targeted Mutation Analysis   | 20 Days          |
| Ataxia-Telangiectasia Genetic Analysis<br>Louis-Bar Syndrome Genetic Analysis   | Blood <sup>EDTA</sup> | 5 ml    | ATM whole gene analysis  | 6 Months         |
| Ataxia-Telangiectasia Genetic Analysis<br>Louis-Bar Syndrome Genetic Analysis   | Blood <sup>EDTA</sup> | 5-10 ml | ATM ATM Protein Analysis (Western Blot & Radyosensitivite test)  | 4-5 Months       |
| Autism Panel 1- Female  | Blood <sup>EDTA</sup> | 5 ml    | All Known Chromosomal Regions Associated With Autism   | 3-4 Months       |

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|--|-----------------------|--------|---|------------------|
| (Female with Autism Spectrum Disorder and Macrocephaly)                              |                       |        | Spectrum Disorders  |                  |
| Autism Panel 1- Female<br>(Female with Autism Spectrum Disorder and Macrocephaly)    | Blood <sup>EDTA</sup> | 5 ml   | PTEN Whole Gene Sequencing  | 3-4 Months       |
| Autism Panel 1- Female<br>(Female with Autism Spectrum Disorder and Macrocephaly)    | Blood <sup>EDTA</sup> | 5 ml   | MECP2 Whole Gene Sequencing   | 3 Months         |
| Autism Panel 1- Female<br>(Female with Autism Spectrum Disorder and Macrocephaly)    | Blood <sup>EDTA</sup> | 5 ml   | CDKL5 Whole Gene Sequencing   | 4-5 Months       |
| Autism Panel 1- Male<br>(Male with Autism Spectrum Disorder and Macrocephaly)        | Blood <sup>EDTA</sup> | 5 ml   | All Known Chromosomal Regions Associated With Autism Spectrum Disorders | 3-4 Months       |
| Autism Panel 1- Male<br>(Male with Autism Spectrum Disorder and Macrocephaly)        | Blood <sup>EDTA</sup> | 5 ml   | PTEN Whole Gene Sequencing  | 3-4 Months       |
| Autism Panel 1- Male<br>(Male with Autism Spectrum Disorder and Macrocephaly)        | Blood <sup>EDTA</sup> | 5 ml   | CDKL5 Whole Gene Sequencing   | 4-5 Months       |
| Autism Panel 2- Female<br>(Female with Autism Spectrum Disorder and No Macrocephaly) | Blood <sup>EDTA</sup> | 5 ml   | All Known Chromosomal Regions Associated With Autism Spectrum Disorders | 2-3 Months       |
| Autism Panel 2- Female<br>(Female with Autism Spectrum Disorder and No Macrocephaly) | Blood <sup>EDTA</sup> | 5 ml   | MECP2 Whole Gene Sequencing   | 2-3 Months       |
| Autism Panel 2- Female<br>(Female with Autism Spectrum Disorder and No Macrocephaly) | Blood <sup>EDTA</sup> | 5 ml   | CDKL5 Whole Gene Sequencing   | 4-5 Months       |
| Autism Panel 2- Male<br>(Male with Autism Spectrum Disorder and No Macrocephaly)     | Blood <sup>EDTA</sup> | 5 ml   | All Known Chromosomal Regions Associated With Autism Spectrum Disorders | 3-4 Months       |
| Autism Panel 2- Male<br>(Male with Autism Spectrum Disorder and No Macrocephaly)     | Blood <sup>EDTA</sup> | 5 ml   | CDKL5 Whole Gene Sequencing   | 4-5 Months       |
| Bardet-Biedl Syndrome 1 Genetic Analysis   | Blood <sup>EDTA</sup> | 5 ml   | BBS1 Whole gene sequencing  | 2-3 Months       |
| Bardet-Biedl Syndrome 10 Genetic Analysis  | Blood <sup>EDTA</sup> | 5 ml   | BBS10 Whole gene sequencing   | 2-3 Months       |
| Bardet-Biedl Syndrome 2 Genetic Analysis   | Blood <sup>EDTA</sup> | 5 ml   | BBS2 Whole gene sequencing  | 2-3 Months       |
| Barth Syndrome Genetic Analysis  | Blood <sup>EDTA</sup> | 5 ml   | TAZ Whole Gene Analysis   | 2-3 Months       |
| Bartter Syndrome Type 2  | Blood <sup>EDTA</sup> | 5 ml   | KCNJ1 Whole Gene Analysis   | 2-3 Months       |
| Bartter Syndrome Type 3  | Blood <sup>EDTA</sup> | 5 ml   | CLCNKB Whole Gene Analysis  | 6-7 Months       |
| Bartter Syndrome Type 4  | Blood <sup>EDTA</sup> | 5 ml   | BSND Whole Gene Analysis  | 2-3 Months       |
| Bartter Syndrome Type 1  | Blood <sup>EDTA</sup> | 5 ml   | SLC12A1 Whole Gene Analysis   | 6-7 Months       |
| Basal Cell Nevus Syndrome  | Blood <sup>EDTA</sup> | 5 ml   | PTCH1 Whole Gene and Deletion Duplication Analysis                      | 3-4 Months       |

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|--|---|------------|--|------------------|
| Gorlin Syndrome  |   |            |  |                  |
| Beckwith-Wiedemann Syndrome  | Blood <sup>EDTA</sup>                     | 5 ml       | KCNQ1OT1 and H19 methylations  | 3 Months         |
| Beckwith-Wiedemann Syndrome  | Blood <sup>EDTA</sup>                     | 5 ml       | 11p15.5 paternal UPD (methylation analysis)  | 4 Months         |
| Beckwith-Wiedemann Syndrome  | Blood <sup>EDTA</sup>                     | 5 ml       | CDKN1C Whole Gene Analysis   | 3 Months         |
| Bernard-Soulier Syndrome   | Blood <sup>EDTA</sup>                     | 5 ml       | GP1BA Whole Gene Analysis  | 2-3 Months       |
| Bernard-Soulier Syndrome   | Blood <sup>EDTA</sup>                     | 5 ml       | GP1BB Whole Gene Analysis  | 2-3 Months       |
| Bernard-Soulier Syndrome   | Blood <sup>EDTA</sup>                     | 5 ml       | GP9 Whole Gene Analysis  | 2-3 Months       |
| Beta Thalassaemia Whole Gene Analysis  | Blood <sup>EDTA</sup> ,<br>Amniotic Fluid | 5ml, 20 ml | HBB Gene 3 Exon +2 Intron Sequencing   | 3-4 Weeks        |
| Biotinidase Deficiency Genetic Analysis  | Blood <sup>EDTA</sup>                     | 5 ml       | 9 Mutations: D444H, A171T, F403V, G98, 7-BP DEL/3-BP INS, Q456H, R157H, R538C, D252G | 2-3 Months       |
| Biotinidase Deficiency Genetic Analysis  | Blood <sup>EDTA</sup>                     | 5 ml       | BTD Whole Gene Analysis  | 2-3 Months       |
| Birt-Hogg-Dube Syndrome Genetic Analysis   | Blood <sup>EDTA</sup>                     | 5 ml       | Analysis of FLCN gene exon 4 to 14 (exons 1-3 are non coding)                        | 3-4 Months       |
| Birt-Hogg-Dube Syndrome Genetic Analysis   | Blood <sup>EDTA</sup>                     | 5 ml       | FLCN del/dup analysis  | 3-4 Months       |
| Blackfan-Diamond Anemia  | Blood <sup>EDTA</sup>                     | 5 ml       | RPS19 Whole Gene Analysis  | 2-3 Months       |
| Blepharophimosis, Ptosis, and Epicanthus Inversus Syndrome   | Blood <sup>EDTA</sup>                     | 5 ml       | FOXL2 Whole Gene Analysis  | 1 Months         |
| Blepharophimosis, Ptosis, and Epicanthus Inversus Syndrome   | Blood <sup>EDTA</sup>                     | 5 ml       | FOXL2 Deletion Duplication Analysis  | 1 Months         |
| Borjeson-Forsman-Lehmann Syndrome Genetic Analysis   | Blood <sup>EDTA</sup>                     | 5 ml       | PHF6 gene 2-10 Exons Sequencing  | 2-3 Ay           |
| BRCA1 Whole Gene Analysis<br>Familial Breast and Ovarian Cancer Susceptibility                           | Blood <sup>EDTA</sup>                     | 5 ml       | BRCA1 Whole Gene Analysis  | 3 Months         |
| BRCA2 Whole Gene Analysis<br>Familial Breast and Ovarian Cancer Susceptibility                           | Blood <sup>EDTA</sup>                     | 5 ml       | BRCA2 Whole Gene Analysis  | 3 Months         |
| CADASIL<br>Cerebral Autosomal Dominant Arteriopathy<br>with Subcortical Infarcts and Leukoencephalopathy | Blood <sup>EDTA</sup>                     | 5 ml       | Sequence alteration in NOTCH3 exons 3, 4, 5, 6 sequence analysis                     | 1 Months         |

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| Canavan Syndrome Genetic Analysis                                  | Blood <sup>EDTA</sup> | 5 ml   | Y231X, E285A, A305E Mutations in ASPA gene                                      | 2 Months         |
| Canavan Syndrome Genetic Analysis                                  | Blood <sup>EDTA</sup> | 5 ml   | ASPA Whole Gene Analysis  | 2-3 Months       |
| Carbamoylphosphate Synthetase I Deficiency<br>Urea Cycle Disorders | Blood <sup>EDTA</sup> | 5 ml   | CPS1 Whole Gene Sequencing  | 2-3 Months       |
| Cardiofaciocutaneous Syndrome (BRAF)                               | Blood <sup>EDTA</sup> | 5 ml   | BRAF exons 11&15 (codon 461-465, G600E)   | 1-2 Months       |
| Cardiofaciocutaneous Syndrome (BRAF)                               | Blood <sup>EDTA</sup> | 5 ml   | BRAF Exons 6&11-16  | 2-3 Months       |
| Cardiofaciocutaneous Syndrome (BRAF)                               | Blood <sup>EDTA</sup> | 5 ml   | BRAF Whole Gene Sequencing  | 2-3 Months       |
| Cardiofaciocutaneous Syndrome (MAP2K1 & MAP2K2)                    | Blood <sup>EDTA</sup> | 5 ml   | MAP2K1 & MAP2K2 exons 2-3   | 2-3 Months       |
| Cardiofaciocutaneous Syndrome (MAP2K1 & MAP2K2)                    | Blood <sup>EDTA</sup> | 5 ml   | MAP2K1 & MAP2K2 Whole Gene Sequencing   | 2-3 Months       |
| Cardiofaciocutaneous Syndrome Genetic Analysis (KRAS)              | Blood <sup>EDTA</sup> | 5 ml   | KRAS Whole Gene Sequencing  | 2-3 Months       |
| Charcot-Marie-Tooth Neuropathy Type 1A Genetic Analysis            | Blood <sup>EDTA</sup> | 5 ml   | Duplication of PMP22  | 20 Days          |
| Charcot-Marie-Tooth Neuropathy Type 1B Genetic Analysis            | Blood <sup>EDTA</sup> | 5 ml   | MPZ Whole Gene Analysis   | 2-3 Months       |
| Charcot-Marie-Tooth Neuropathy Type 1B Genetic Analysis            | Blood <sup>EDTA</sup> | 5 ml   | RAB7 Gene Sequence Variant  | 2-3 months       |
| Charcot-Marie-Tooth Neuropathy Type 1C Genetic Analysis            | Blood <sup>EDTA</sup> | 5 ml   | LITAF Whole Gene Analysis   | 2-3 Months       |
| Charcot-Marie-Tooth Neuropathy Type 1D Genetic Analysis            | Blood <sup>EDTA</sup> | 5 ml   | EGR2 Whole Gene Analysis  | 2-3 Months       |
| Charcot-Marie-Tooth Neuropathy Type 1E Genetic Analysis            | Blood <sup>EDTA</sup> | 5 ml   | PMP22 Whole Gene Analysis   | 2-3 Months       |
| Charcot-Marie-Tooth Neuropathy Type 2 Genetic Analysis             | Blood <sup>EDTA</sup> | 5 ml   | KIF1B, MFN2, RAB7, LMNA, GARS, NEFL, MPZ, GDAP1, CJB1<br>Gene Sequences Variant | 3-4 months       |
| Charcot-Marie-Tooth Neuropathy Type 2A1 Genetic Analysis           | Blood <sup>EDTA</sup> | 5 ml   | KIF1B Gene Sequence Variant   | 2-3 months       |
| Charcot-Marie-Tooth Neuropathy Type 2A2 Genetic Analysis           | Blood <sup>EDTA</sup> | 5 ml   | MFN2 Gene Sequence Variant  | 2-3 months       |
| Charcot-Marie-Tooth Neuropathy Type 2B1 Genetic Analysis           | Blood <sup>EDTA</sup> | 5 ml   | LMNA Gene Sequence Variant  | 2-3 months       |
| Charcot-Marie-Tooth Neuropathy Type 2D Genetic Analysis            | Blood <sup>EDTA</sup> | 5 ml   | GARS Gene Sequence Variant  | 2-3 months       |
| Charcot-Marie-Tooth Neuropathy Type 2E/1F Genetic<br>Analysis      | Blood <sup>EDTA</sup> | 5 ml   | NEFL Gene And Promoter Region Sequencing  | 2-3 Months       |
| Charcot-Marie-Tooth Neuropathy Type 2E/1F Genetic                  | Blood <sup>EDTA</sup> | 5 ml   | NEFL Gene Sequence Variant  | 2-3 months       |

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| Analysis   |                       |        |   |                  |
| Charcot-Marie-Tooth Neuropathy Type 2I/2J Genetic Analysis                               | Blood <sup>EDTA</sup> | 5 ml   | MPZ Gene Sequence Variant                     | 2-3 months       |
| Charcot-Marie-Tooth Neuropathy Type 2K Genetic Analysis                                  | Blood <sup>EDTA</sup> | 5 ml   | GDAP1 Gene Sequence Variant                   | 2-3 months       |
| Charcot-Marie-Tooth Neuropathy Type 2L Genetic Analysis                                  | Blood <sup>EDTA</sup> | 5 ml   | HSPB8 Gene Sequence Variant                   | 2-3 months       |
| CHARGE Syndrome Genetic Analysis   | Blood <sup>EDTA</sup> | 5 ml   | CHD7 Whole Gene Analysis                      | 2-3 Months       |
| CHARGE Syndrome Genetic Analysis   | Blood <sup>EDTA</sup> | 5 ml   | CHD7 Deletions (8q12.2)                       | 1 Month          |
| Cholestasis, Progressive Familial Intrahepatic 1   | Blood <sup>EDTA</sup> | 5 ml   | ATP8B1 Whole Gene Analysis                    | 5-6 Months       |
| Cholestasis, Progressive Familial Intrahepatic 2   | Blood <sup>EDTA</sup> | 5 ml   | ABCB11 (BSEP) Whole Gene Analysis             | 5-6 Months       |
| Cholestasis, Progressive Familial Intrahepatic 3   | Blood <sup>EDTA</sup> | 5 ml   | ABCB4 (MDR3) Whole Gene Analysis              | 5-6 Months       |
| Citrullinemia Type I<br>Urea Cycle Disorders   | Blood <sup>EDTA</sup> | 5 ml   | ASS1 Whole Gene Sequencing                    | 2-3 Months       |
| Classical Hemochromatosis Genetic Analysis   | Blood <sup>EDTA</sup> | 5 ml   | 2 Mutation (C282Y, H63D)                      | 20 Days          |
| Classical Hemochromatosis Genetic Analysis   | Blood <sup>EDTA</sup> | 5 ml   | 18 Mutation                                   | 20 Days          |
| Classical Hemochromatosis Genetic Analysis   | Blood <sup>EDTA</sup> | 5 ml   | HFE Whole Gene Analysis                       | 2-3 Months       |
| Coffin-Lowry Syndrome  | Blood <sup>EDTA</sup> | 5 ml   | RPS6KA3 Whole Gene Analysis                   | 3 Months         |
| COL1A1/2 Related Osteogenesis Imperfecta<br>Osteogenesis Imperfecta Type I, IIA, III, IV | Blood <sup>EDTA</sup> | 5 ml   | COL1A1 Whole Gene and COL1A2 47 Exon Analysis | 2-3 Months       |
| Combined Immune Deficiency   | Blood <sup>EDTA</sup> | 5 ml   | RAG1 & RAG2 whole gene sequencing             | 2-3 Months       |
| Combined Pituitary Hormone Deficiency 1  | Blood <sup>EDTA</sup> | 5 ml   | POU1F1 (PIT1) Whole Gene Analysis             | 2 Months         |
| Combined Pituitary Hormone Deficiency 2  | Blood <sup>EDTA</sup> | 5 ml   | PROP1 Whole Gene Analysis                     | 2 Months         |
| Congenital Adrenal Hypoplasia<br>21-Hidroksilase Genetic Analysis                        | Blood <sup>EDTA</sup> | 5 ml   | CYP21A2 Target Mutation Analysis              | 1 Months         |
| Congenital Adrenal Hypoplasia<br>21-Hidroksilase Genetic Analysis                        | Blood <sup>EDTA</sup> | 5 ml   | CYP21A2 Sequence Analysis                     | 2 Months         |

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| Congenital Adrenal Hypoplasia 21-Hidroksilase Genetic Analysis   | Blood <sup>EDTA</sup> | 5 ml   | CYP21A2 Sequence Analysis & MLPA                  | 3 Months         |
| Congenital Adrenal Hypoplasia X Linked Adrenal Hyperplasia       | Blood <sup>EDTA</sup> | 5 ml   | NROB1 Whole Gene Analysis                         | 2-3 Months       |
| Congenital Disorders of Glycosylation Type IA                    | Blood <sup>EDTA</sup> | 5 ml   | PMM2 Whole Gene Analysis                          | 2-3 Months       |
| Congenital Disorders of Glycosylation Type IB                    | Blood <sup>EDTA</sup> | 5 ml   | MPI Whole Gene Analysis                           | 2-3 Months       |
| Congenital Disorders of Glycosylation Type IC                    | Blood <sup>EDTA</sup> | 5 ml   | ALG6 Whole Gene Analysis                          | 2-3 Months       |
| Congenital Merosin-Deficient Muscular Dystrophy 1A               | Blood <sup>EDTA</sup> | 5 ml   | LAMA2 Whole Gene Analysis                         | 2-3 Months       |
| Congenital Myasthenic Syndrome Genetic Analysis (CHAT)           | Blood <sup>EDTA</sup> | 5 ml   | CHAT Whole Gene Analysis                          | 2-3 Months       |
| Congenital Myasthenic Syndrome Genetic Analysis (CHRNA1)         | Blood <sup>EDTA</sup> | 5 ml   | CHRNA1 Whole Gene Analysis                        | 2-3 Months       |
| Congenital Myasthenic Syndrome Genetic Analysis (CHRND)          | Blood <sup>EDTA</sup> | 5 ml   | CHRND Whole Gene Analysis                         | 2-3 Months       |
| Congenital Myasthenic Syndrome Genetic Analysis (CHRNE)          | Blood <sup>EDTA</sup> | 5 ml   | CHRNE Whole Gene Analysis                         | 2-3 Months       |
| Congenital Myasthenic Syndrome Genetic Analysis (COLQ)           | Blood <sup>EDTA</sup> | 5 ml   | COLQ Whole Gene Analysis                          | 2-3 Months       |
| Congenital Myasthenic Syndrome Genetic Analysis (DOK7)           | Blood <sup>EDTA</sup> | 5 ml   | DOK7 Target Mutation Analysis $\epsilon$ 1267delG | 2-3 Months       |
| Congenital Myasthenic Syndrome Genetic Analysis (RAPSN)          | Blood <sup>EDTA</sup> | 5 ml   | RAPSN Target Mutation Analysis 1293insG           | 2-3 Months       |
| Congenital Myasthenic Syndrome Genetic Analysis (CHRNA1)         | Blood <sup>EDTA</sup> | 5 ml   | CHRNA1 Whole Gene Analysis                        | 2-3 Months       |
| Congenital Nystagmus 1, X-linked Genetic Analysis                | Blood <sup>EDTA</sup> | 5 ml   | FRMD7 Whole Gene Analysis                         | 2-3 Months       |
| Connexin 32 Charcot-Marie-Tooth, X-Linked Type 1 ( GJB1) (CMTX1) | Blood <sup>EDTA</sup> | 5 ml   | Sequence alteration in GJB1                       | 2 Months         |
| Cornelia de Lange Syndrome 1                                     | Blood <sup>EDTA</sup> | 5 ml   | NIPBL Whole Gene Analysis                         | 2-3 Months       |
| Cornelia de Lange Syndrome 2                                     | Blood <sup>EDTA</sup> | 5 ml   | SMC1A Whole Gene Analysis                         | 2-3 Months       |
| Crigler-Najjar Syndrome Type1, Type2 (CNS1, CNS2)                | Blood <sup>EDTA</sup> | 5 ml   | UGT1A1 Sequence Analysis                          | 2-3 Months       |
| CRTAP Related Osteogenesis Imperfecta                            | Blood <sup>EDTA</sup> | 5 ml   | CRTAP Whole Gene Analysis                         | 2-3 Months       |
| CYP2C19 Genotyping   | Blood <sup>EDTA</sup> | 5 ml   | PCR&RFLP CYP2C19 Genotype *1, *2, *3              | 20 Days          |

## BURC GENETIC DIAGNOSTIC CENTER MOLECULAR GENETIC TEST LIST (2010)

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|--|-----------------------|--------|---|------------------|
| CYP2C9 Genotyping<br>Warfarin, Caumadin Resistance                   | Blood <sup>EDTA</sup> | 5 ml   | PCR&RFLP CYP2C9 Genotype *1, *2, *3                     | 20 Days          |
| CYP2D6 Genotyping  | Blood <sup>EDTA</sup> | 5 ml   | PCR&RFLP CYP2D6 Genotype*1, *2A, *2B, *3, *4, *10       | 20 Days          |
| Cystathionine Beta-Synthetase (CBSins68)<br>Cardiovascular Risk      | Blood <sup>EDTA</sup> | 5 ml   | CBS gene 844ins68bp                                     | 20 Days          |
| Cystic Fibrosis Genetic Analysis                                     | Blood <sup>EDTA</sup> | 5 ml   | Frequent 36 Mutations Analysis in CFTR Gene             | 20 Days          |
| Cystic Fibrosis Genetic Analysis                                     | Blood <sup>EDTA</sup> | 5 ml   | CFTR Gene 8 Exons 500 Mutations Analysis                | 1 Months         |
| Cystic Fibrosis Genetic Analysis                                     | Blood <sup>EDTA</sup> | 5 ml   | CFTR Gene Whole Gene Analysis                           | 3 Months         |
| D-2-Hydroxyglutaric Aciduria<br>Genetic Analysis                     | Blood <sup>EDTA</sup> | 5 ml   | D2HGDH Whole Gene Analysis                              | 4 Months         |
| Dandy Walker Syndrome  | Blood <sup>EDTA</sup> | 5 ml   | 50kb Resolution Genome Screening                        | 1-2 Months       |
| Dandy Walker Syndrome  | Blood <sup>EDTA</sup> | 5 ml   | ZIC1 and ZIC4 Gene Screening                            | 1 Months         |
| Danon Disease Genetic Analysis                                       | Blood <sup>EDTA</sup> | 5 ml   | LAMP2 Whole Gene Analysis                               | 2-3 Months       |
| Dent Disease 1 Genetic Testing                                       | Blood <sup>EDTA</sup> | 5 ml   | CLCN5 Whole Gene Analysis                               | 1-2 Months       |
| Dentatorubral-Pallidoluysian Atrophy (DRPLA) Genetic<br>Analysis     | Blood <sup>EDTA</sup> | 5 ml   | CAG Repeat Analysis in ATN1                             | 1-2 Months       |
| Distal Renal Tubular Acidosis, Autosomal Recessive; RTADR            | Blood <sup>EDTA</sup> | 5 ml   | ATP6V0A4 (ATP6N1B) Whole Gene Analysis                  | 4-5 Months       |
| Distal Renal Tubular Acidosis, With Progressive Nerve<br>Deafness    | Blood <sup>EDTA</sup> | 5 ml   | ATP6V1B1 (ATP6B1) Whole Gene Analysis                   | 4-5 Months       |
| Distoni 1 (DYT 1)<br>Distoni Muskulorum Deformans 1                  | Blood <sup>EDTA</sup> | 5 ml   | TOR1A 904_906 delGAG Targeted Mutation Analysis         | 1-2 Months       |
| DJ-1 Related Parkinson Disease<br>Parkinson Disease 7                | Blood <sup>EDTA</sup> | 5 ml   | PARK7 Whole Gene Analysis                               | 2-3 Months       |
| Dravet Syndrome Genetic Anakysis                                     | Blood <sup>EDTA</sup> | 5 ml   | SCN1A Sequence Variants, Large Deletions and Insertions | 3-6 Months       |
| Duchenne muscular dystrophy (DMD)<br>Becker muscular dystrophy (BMD) | Blood <sup>EDTA</sup> | 5 ml   | Deletion of DMD Gene (18 exons)                         | 1 Months         |

| Test Name  | Sample                              | Amount  | Method  | Turn Around Time |
|--|-------------------------------------|---------|---|------------------|
| Duchenne muscular dystrophy (DMD)<br>Becker muscular dystrophy (BMD) | Blood <sup>EDTA</sup>               | 5 ml    | Quantitative analysis of Deletion and Duplications in DMD Gene (79 Exons) | 2-3 Months       |
| Duchenne muscular dystrophy (DMD)<br>Becker muscular dystrophy (BMD) | Blood <sup>EDTA</sup>               | 5 ml    | Sequencing  | 3-6 Months       |
| Dystonia 1 (DYT 1)<br>Distoni Muskulorum Deformans 1                 | Blood <sup>EDTA</sup>               | 5 ml    | TOR1A Whole Gene Analysis   | 2-3 Months       |
| Dystrophic Epidermolysis Bullosa                                     | Blood <sup>EDTA</sup>               | 5 ml    | COL7A1 selected exons between 73-76                                       | 1 Month          |
| Dystrophic Epidermolysis Bullosa                                     | Blood <sup>EDTA</sup>               | 5 ml    | COL7A1 Whole Gene Analysis  | 2-3 Months       |
| Ehlers Danlos Syndrome Type I  | Skin punch biopsy                   | 5-10 mg | COL5A1 and COL5A2 whole gene analysis                                     | 6-12 Months      |
| Ehlers Danlos Syndrome Type I  | Skin punch biopsy                   | 5-10 mg | COL1A1 and COL1A2 whole gene analysis                                     | 6-12 Months      |
| Ehlers Danlos Syndrome Type II                                       | Skin punch biopsy                   | 5-10 mg | COL5A1 and COL5A2 whole gene analysis                                     | 6-12 Months      |
| Ehlers Danlos Syndrome Type IV, Autosomal Dominant                   | Skin punch biopsy                   | 5-10 mg | COL3A1 whole gene analysis  | 6-12 Months      |
| Ehlers Danlos Syndrome Type VI                                       | Blood <sup>EDTA</sup>               | 5 ml    | PLOD1 whole gene analysis and duplication analysis of exon 10-16          | 2-3 Months       |
| Ehlers Danlos Syndrome Type VII, Autosomal Dominant                  | Blood <sup>EDTA</sup>               | 5 ml    | 5, 6, 7. Exon splicing site of COL1A1 and COL1A2                          | 2-3 Months       |
| ENOS (NOS3 Polymorphism Analysis)<br>Cardiovascular Risk Analysis    | Blood <sup>EDTA</sup>               | 5 ml    | PCR&RFLP NOS3 glu298asp Polymorphisms                                     | 10 Days          |
| Epidermolysis Bullosa Testi  | Skin biopsy                         |         | Immunofluorescence & Electron Microscopy & Mutation Analysis              | -                |
| Epidermolysis Bullosa Testing  | skin biopsy & Blood <sup>EDTA</sup> | 5 ml    | Immunofluorescence & Electron Microscopy                                  | 2-3 Months       |
| Episodic Ataxia Type 2   | Blood <sup>EDTA</sup>               | 5 ml    | CACNA1A Whole Gene Analysis   | 3 Months         |
| Fabry Disease<br>Alpha-Galactosidase A Deficiency                    | Blood <sup>EDTA</sup>               | 5 ml    | GLA Whole Gene Analysis   | 2 Months         |
| Factor 10 Deficiency Genetic Analysis                                | Blood <sup>EDTA</sup>               | 5 ml    | F10 Whole Gene Analysis   | 2-3 Months       |

| Test Name  | Sample                                       | Amount | Method  | Turn Around Time |
|--|--|--------|---|------------------|
| Factor V and Factor VIII Combined Deficiency Analysis  | Blood <sup>EDTA</sup>                        | 5 ml   | LMAN1 & MCFD2 Whole Gene Sequencing             | 3 Months         |
| Factor V Leiden Mutation Analysis  | Blood <sup>EDTA</sup>                        | 5 ml   | PCR&RFLP 1691 G->A Targeted Mutation Analysis   | 5 Days           |
| Familial Hyperlipidemia Genetic Analysis   | Blood <sup>EDTA</sup>                        | 5 ml   | LPL Gene Whole Coding Regions Sequence Analysis | 2-3 Months       |
| Fanconi-Bickel Syndrome  | Blood <sup>EDTA</sup>                        | 5 ml   | GLUT2 (SLC2A2) Whole Gene Analysis              | 2-3 Months       |
| Farber Disease Genetic Analysis  | Blood <sup>EDTA</sup>                        | 5 ml   | ASAHI Whole Gene Analysis                       | 3-4 Months       |
| Feingold Syndrome Genetic Analysis   | Blood <sup>EDTA</sup>                        | 5 ml   | MYCN gene 2-3 exons and 3' UTR region           | 2 Months         |
| Feingold Syndrome Genetic Analysis   | Blood <sup>EDTA</sup>                        | 5 ml   | MYCN Gene Deletion/ Duplication Analysis        | 2-3 Months       |
| Fenilketonuri (PKU)<br>PAH Eksikliği<br>Hiperfenilalaninemi (HPA)  | Blood <sup>EDTA</sup>                        | 5 ml   | PAH Whole Gene analysis                         | 2 Months         |
| FGFR2 Genetic Analysis<br>Apert Syndrome<br>Jackson-Weiss Syndrome<br>Crouzon Syndrome<br>Beare-Stevenson Syndrome | Blood <sup>EDTA</sup>                        | 5 ml   | Targeted Mutation Analysis                      | 1-2 Months       |
| FMF 12 Mutations Analysis  | Blood <sup>EDTA</sup>                        | 5 ml   | PCR+RFLP+ARMS & PCR+Reverse Hybridization       | 20 Days          |
| FMF 40 Mutations Analysis  | Blood <sup>EDTA</sup>                        | 5 ml   | DNA Sequencing                                  | 30 Days          |
| FMF Whole Gene Analysis  | Blood <sup>EDTA</sup>                        | 5 ml   | DNA Sequencing                                  | 2 Months         |
| Focal Segmental Glomerulosclerosis Type 1  | Blood <sup>EDTA</sup>                        | 5 ml   | ACTN4 Whole Gene Analysis                       | 2 Months         |
| Focal Segmental Glomerulosclerosis Type 2  | Blood <sup>EDTA</sup>                        | 5 ml   | TRPC6 Whole Gene Analysis                       | 2 Months         |
| Focal Segmental Glomerulosclerosis Type 3  | Blood <sup>EDTA</sup>                        | 5 ml   | CD2AP Whole Gene Analysis                       | 2 Months         |
| Fragile X (CGG Repeated) STR Analysis  | Blood <sup>EDTA</sup> ,<br>Amniotic<br>Fluid | 5 ml   | Fragile X (CGG Repeated) STR Analysis           | 45 Days          |
| Fragile X Genetic Analysis   | Blood <sup>EDTA</sup>                        | 5 ml   | FMR1 whole gene sequencing                      | 2 Months         |
| Fraser Syndrome  | Blood <sup>EDTA</sup>                        | 5 ml   | FREM2 & FRAS1 15 Mutation Analysis, Microarray  | 2 Months         |

| Test Name  | Sample                | Amount | Method  | Turn Around Time |
|--|-----------------------|--------|---|------------------|
| Fraser Syndrome  | Blood <sup>EDTA</sup> | 5 ml   | 6. Exon of FREM2 Sequencing for Glu1972Lys Mutation               | 2 Months         |
| Friedreich Ataxia Genetic Analysis, FRDA   | Blood <sup>EDTA</sup> | 5 ml   | FXN Targeted Mutation Analysis                                    | 45 Days          |
| Friedreich Ataxia Genetic Analysis, FRDA   | Blood <sup>EDTA</sup> | 5 ml   | FXN Sequencing  | 6-8 weeks        |
| Frontometaphyseal Dysplasia  | Blood <sup>EDTA</sup> | 5 ml   | FLNA Whole Gene Sequencing  | 3-4 Months       |
| Frontotemporal Dementia With Parkinsonism-17 Genetic Analysis                                      | Blood <sup>EDTA</sup> | 5 ml   | MAPT Whole Gene Analysis  | 3-4 Months       |
| Frontotemporal Dementia,GRN-related Genetic Analysis   | Blood <sup>EDTA</sup> | 5 ml   | GRN Whole Gene Analysis   | 3-4 Months       |
| Fructose Intolerance   | Blood <sup>EDTA</sup> | 5 ml   | ALDOB Gene del4E4, A149P, A174D, N334K Targeted Mutation Analysis | 2-3 Months       |
| Fructose-1,6-Bisphosphatase Deficiency Genetic Testing   | Blood <sup>EDTA</sup> | 5 ml   | FBP1 Whole Gene Analysis  | 1 Months         |
| FSH Muscular Dystrophy Genetic Analysis<br>Facioscapulohumeral Muscular Dystrophy Genetic Analysis | Blood <sup>EDTA</sup> | 5 ml   | Targeted Mutation Analysis, 3.3 Kb DNA Motif deletions            | 1-2 Months       |
| G6PD gene sequence analysis  | Blood <sup>EDTA</sup> | 5 ml   | G6PD Whole Gene Analysis  | 2 Months         |
| Galactosemia Genetic Analysis  | Blood <sup>EDTA</sup> | 5 ml   | GALT gene sequence variants                                       | 2-3 Months       |
| Gastric Cancer Genetic Testing   | Blood <sup>EDTA</sup> | 5 ml   | CDH1 Whole Gene Analysis  | 1-2 Months       |
| Gastric Cancer Genetic Testing   | Blood <sup>EDTA</sup> | 5 ml   | CDH1 Whole Gene and Deletion Duplication Analysis                 | 1-2 Months       |
| Gaucher Disease Genetic Analysis   | Blood <sup>EDTA</sup> | 5 ml   | Whole Gene Analysis   | 2-3 Months       |
| Gaucher Genetic Analysis   | Blood <sup>EDTA</sup> | 5 ml   | GBA Gene 6 Common Mutations Analysis                              | 2 Months         |
| Gilbert Syndrome   | Blood <sup>EDTA</sup> | 5 ml   | TA number on promoter region of UGT1A1 Gene                       | 1-2 Months       |
| Gitelman Syndrome  | Blood <sup>EDTA</sup> | 5 ml   | SLC12A3 Whole gene analysis                                       | 3-4 Months       |
| Gitelman Syndrome  | Blood <sup>EDTA</sup> | 5 ml   | SLC12A3 Del/Dup Analysis  | 2-3 Months       |
| GSTM1 Gene Deletion<br>Lung Cancer   | Blood <sup>EDTA</sup> | 5 ml   | PCR&RFLP GSTM1 (+/-)  | 20 Days          |
| GSTT1 Gene Deletion  | Blood <sup>EDTA</sup> | 5 ml   | PCR&RFLP GSTT1 (+/-)  | 20 Days          |
| Hallervorden Spatz Syndrome  | Blood <sup>EDTA</sup> | 5 ml   | PANK2 Sequence Variants   | 2 Months         |

| Test Name  | Sample   | Amount          | Method   | Turn Around Time |
|--|--|-----------------|--|------------------|
| Hemiplegic Migraine 2 Genetic Analysis                                     | Blood <sup>EDTA</sup>  | 5 ml            | ATP1A2 Whole Gene Analysis                               | 3-4 months       |
| Hemochromatosis Type 2 (Juvenile) Genetic Analysis                         | Blood <sup>EDTA</sup>  | 5 ml            | HJV Exon 4 analysis                                      | 2-3 Months       |
| Hemochromatosis Type 2 (Juvenile) Genetic Analysis                         | Blood <sup>EDTA</sup>  | 5 ml            | HJV Whole gene analysis                                  | 2-3 Months       |
| Hemochromatosis Type 2 (Juvenile) Genetic Analysis                         | Blood <sup>EDTA</sup>  | 5 ml            | HAMP Whole Gene Analysis                                 | 2-3 Months       |
| Hemochromatosis Type 2 (Juvenile) Genetic Analysis                         | Blood <sup>EDTA</sup>  | 5 ml            | HAMP and HJV Whole Gene Analysis                         | 2-3 Months       |
| Hemochromatosis Type 3 Genetic Analysis                                    | Blood <sup>EDTA</sup>  | 5 ml            | Mutation Analysis  | 1 Month          |
| Hemochromatosis Type 3 Genetic Analysis                                    | Blood <sup>EDTA</sup>  | 5 ml            | TFR2 Whole Gene Analysis                                 | 2-3 Months       |
| Hemochromatosis, Type 4 Genetic Analysis                                   | Blood <sup>EDTA</sup>  | 5 ml            | SLC40A1 (FPN1) Frequent mutations                        | 1 Ay             |
| Hemochromatosis, Type 4 Genetic Analysis                                   | Blood <sup>EDTA</sup>  | 5 ml            | SLC40A1 (FPN1) Whole Gene Analysis                       | 3-4 Ay           |
| Hemolytic-Uremic Syndrome, Atypical Genetic Testing                        | Blood <sup>EDTA</sup>  | 5 ml            | CFH Whole Gene Analysis                                  | 3-4 Months       |
| Hemophilia A Genetic Analysis  | Blood <sup>EDTA</sup>  | 5 ml            | F8 intron 1 gene inversion                               | 2-3 Months       |
| Hemophilia A Genetic Analysis  | Blood <sup>EDTA</sup>  | 5 ml            | F8 intron 22-A gene inversion                            | 2-3 Months       |
| Hemophilia A Genetic Analysis  | Blood <sup>EDTA</sup>  | 5 ml            | F8 Whole Gene Analysis                                   | 3-5 Months       |
| Hemophilia B Genetic Analysis<br>Christmas Disease<br>Factor IX Deficiency | Blood <sup>EDTA</sup>  | 5 ml            | F9 Whole Gene Analysis                                   | 1-2 Months       |
| Hereditary Multiple Exostosis Genetic Analysis                             | Blood <sup>EDTA</sup>  | 5 ml            | EXT1 Whole Gene Analysis                                 | 4-5 Months       |
| Hereditary Multiple Exostosis Genetic Analysis                             | Blood <sup>EDTA</sup>  | 5 ml            | EXT2 Whole Gene Analysis                                 | 4-5 Months       |
| Hereditary Non-Polyposis Colon Cancer                                      | Blood <sup>EDTA</sup>  | 5 ml            | MLH1 Whole Gene Analysis                                 | 2 Months         |
| Hereditary Non-Polyposis Colon Cancer                                      | Blood <sup>EDTA</sup>  | 5 ml            | MSH2 Whole Gene Analysis                                 | 2 Months         |
| Hereditary Non-Polyposis Colon Cancer                                      | Paraffin Block<br>Tumor<br>Section,<br>Blood <sup>EDTA</sup> | 4-5 µm,<br>5 ml | MSI (Microsatellite Instability test)                    | 3-4 Months       |
| Hereditary Pancreatitis Genetic Analysis                                   | Blood <sup>EDTA</sup>  | 5 ml            | SPINK1 (3. Exon) and PRSS1 (1-3 exons) Sequence Analysis | 2-3 Months       |

| Test Name  | Sample                | Amount | Method  | Turn Around Time |
|--|-----------------------|--------|---|------------------|
| Hereditary Pancreatitis Genetic Analysis                   | Blood <sup>EDTA</sup> | 5 ml   | SPINK1 Whole Gene Analysis  | 2-3 Months       |
| Hereditary Pancreatitis Genetic Analysis                   | Blood <sup>EDTA</sup> | 5 ml   | PRSS1 Whole Gene Analysis   | 2-3 Months       |
| Hereditary Paraganglioma                                   | Blood <sup>EDTA</sup> | 5 ml   | SDHD Gene Sequencing  | 1-2 Months       |
| Hereditary Paraganglioma                                   | Blood <sup>EDTA</sup> | 5 ml   | SDHB Gene Sequencing  | 1-2 Months       |
| Hereditary Paraganglioma                                   | Blood <sup>EDTA</sup> | 5 ml   | SDHC Gene Sequencing  | 1-2 Months       |
| Hereditary Paraganglioma                                   | Blood <sup>EDTA</sup> | 5 ml   | SDHD, SDHB, SDHC Gene Deletions   | 1-2 Months       |
| Hereditary Spastic Paraplegia 3, Autosomal Dominant (SPG3) | Blood <sup>EDTA</sup> | 5 ml   | SPG3A Whole Gene Sequencing and Deletion/Duplication Analysis   | 2-3 Months       |
| Hereditary Spastic Paraplegia 4, Autosomal Dominant (SPG4) | Blood <sup>EDTA</sup> | 5 ml   | SPASTIN (SPAST) Whole Gene Sequencing and Deletion/Duplication Analysis   | 2-3 Months       |
| Hirschsprung Disease Genetic Analysis                      | Blood <sup>EDTA</sup> | 5 ml   | RET whole gene sequencing & Del/dup analysis  | 3-4 Months       |
| HLA B27 Genetic Analysis<br>Ankylosing Spondylitis (AS)    | Blood <sup>EDTA</sup> | 5 ml   | HLA-B27 B*2701-B*2723 alleles   | 5 Days           |
| Huntington Disease Gene Analysis                           | Blood <sup>EDTA</sup> | 5 ml   | CAG trinucleotide repeat expansion of HD gene with aPCR&FS  | 1 Months         |
| Hutchinson-Gilford Progeria Syndrome                       | Blood <sup>EDTA</sup> | 5 ml   | Silent p.Gly608Gly mutation in exon 11 of the LMNA gene   | 1 Months         |
| Hutchinson-Gilford Progeria Syndrome                       | Blood <sup>EDTA</sup> | 5 ml   | LMNA Whole Gene Analysis  | 2-3 Months       |
| Hydroxyglutaric Aciduria Quantitative Analysis             | Urine                 | 5 ml   | Quantification Analysis for L2HGDH and D2HGDH – Mass Spectrometry   | 1-2 Months       |
| Hypochondroplasia(HCH) Genetic Analysis                    | Blood <sup>EDTA</sup> | 5 ml   | FGFR3 gene N540K (C1620A) and N540K (C1620G)Target Mutation Analysis  | 20 Days          |
| Hypochondroplasia(HCH) Genetic Analysis                    | Blood <sup>EDTA</sup> | 5 ml   | FGFR3 gene 7, 8, 9, 10, 13 ve 15. Exon Sequence Analysis  | 2 Months         |
| Hypokalemic Periodic Paralysis Genetic Analysis            | Blood <sup>EDTA</sup> | 5 ml   | CACNA1S Whole Gene Analysis   | 7-8 Months       |
| Hypokalemic Periodic Paralysis Genetic Analysis            | Blood <sup>EDTA</sup> | 5 ml   | <b>CACNA1S 4 Mutation</b><br>(c.1583G>A (R528H), c.1582C>G (R528G), c.3716G>A (R1239H) and c.3715C>G (R1239G))<br>&<br><b>SCN4A 5 Mutation</b><br>(c.2006G>A (R669H), c.2014C>A (R672S), c.2015G>A (R672H), c.2014C>G (R672G), c.2014C>T (R672C)) | 2-3 Months       |

| Test Name  | Sample                | Amount | Method   | Turn Around Time |
|--|-----------------------|--------|--|------------------|
| Hypomagnesemia 3 Genetic Analysis                                    | Blood <sup>EDTA</sup> | 5 ml   | CLDN16 Whole Gene Sequencing   | 1-2 Months       |
| Hypomagnesemia with Secondary Hypocalcemia Genetic Analysis          | Blood <sup>EDTA</sup> | 5 ml   | TRPM6 mutations scanning   | 2 Months         |
| Ichthyosis Vulgaris Genetic Analysis                                 | Blood <sup>EDTA</sup> | 5 ml   | FLG 4 Mutations: pR501X, c.2282del4, pR2447X and p.S3247X                                    | 2 Months         |
| Ichthyosis Vulgaris Genetic Analysis                                 | Blood <sup>EDTA</sup> | 5 ml   | FLG 7 Mutations: pR501X, c.2282del4, c.3702delG, p.E2422X, c.7267delCA, pR2447X and p.S3247X | 2-3 Months       |
| IgVH (Ig VH) Mutation Analysis                                       | Blood <sup>EDTA</sup> | 5 ml   | IgVH (Ig VH) Mutation Analysis   | 1 Months         |
| Imatinib Resistance Analysis   | Blood <sup>EDTA</sup> | 5 ml   | ABL1 Gene Targeted Mutation Analysis (T315I, G250E, Y253H, E355K, F317L, M351T mutations)    | 20 Months        |
| Inclusion Body Myopathy 2 (IBM 2)                                    | Blood <sup>EDTA</sup> | 5 ml   | GNE Whole Gene Analysis  | 2-3 Months       |
| Inclusion Body Myopathy 2 (IBM 2)                                    | Blood <sup>EDTA</sup> | 5 ml   | GNE gene M712T mutation  | 2-3 Months       |
| Incontinentia Pigmenti Deletion Analysis                             | Blood <sup>EDTA</sup> | 5 ml   | Deletion of exons 4-10 of IKBKG (NEMO)   | 2 Months         |
| Incontinentia Pigmenti Deletion Analysis                             | Blood <sup>EDTA</sup> | 5 ml   | IKBKG (NEMO) sequence variations   | 2-3 Months       |
| IPEX Syndrome  | Blood <sup>EDTA</sup> | 5 ml   | FOXP3 Whole Gene Analysis  | 2 Months         |
| JAK2 Gene Mutation, Polisitemia Vera, Thrombocythemia, Myelofibrosis | Blood <sup>EDTA</sup> | 5 ml   | JAK2 Gene V617F mutations with Tetra ARMS  | 20 Days          |
| Jarcho-Levin Syndrome DLL3 Genetic Analysis                          | Blood <sup>EDTA</sup> | 5 ml   | DLL3 Whole Gene Sequencing   | 2 Months         |
| Kallmann Syndrome (FGFR1)  | Blood <sup>EDTA</sup> | 5 ml   | FGFR1 sequence variants  | 2 Months         |
| Kallmann Syndrome (KAL1)   | Blood <sup>EDTA</sup> | 5 ml   | KAL1 sequence variants   | 2 Months         |
| Kallmann Syndrome (PROK2)  | Blood <sup>EDTA</sup> | 5 ml   | PROKR2 sequence variants   | 1 Months         |
| Kallmann Syndrome (PROKR2)   | Blood <sup>EDTA</sup> | 5 ml   | PROK2 sequence variants  | 1 Months         |
| Kennedy's Disease<br>Spinal and Bulbar Muscular Atrophy, SBMA        | Blood <sup>EDTA</sup> | 5 ml   | AR Gene Target Mutation Analysis   | 4-6 Weeks        |
| Kistik Fibrozis (CFTR) 5 T Varyant Analysis                          | Blood <sup>EDTA</sup> | 5 ml   | PCR, CFTR Gene IVS8-(5T) Analysis  | 20 Days          |
| Krabbe Disease Genetic Analysis                                      | Blood <sup>EDTA</sup> | 5 ml   | GALC Whole Gene Analysis   | 2 Months         |

| Test Name   | Sample                | Amount | Method  | Turn Around Time |
|---|-----------------------|--------|---|------------------|
| Kufor Rakeb Disease<br>Parkinson Disease 9                    | Blood <sup>EDTA</sup> | 5 ml   | ATP13A2 Whole Gene Analysis   | 3-4 Months       |
| L-2-Hydroxyglutaric Aciduria Genetic Analysis                 | Blood <sup>EDTA</sup> | 5 ml   | L2HGDH Whole Gene Analysis  | 4 Months         |
| Lafora Disease<br>Myoclonic Epilepsy Of Lafora                | Blood <sup>EDTA</sup> | 5 ml   | NHLRC1 Whole Gene Analysis  | 2 Months         |
| Lafora Disease Genetic Analysis                               | Blood <sup>EDTA</sup> | 5 ml   | EPM2A Whole Gene Analysis   | 2-3 Months       |
| Lamellar Ichthyosis Type 1                                    | Blood <sup>EDTA</sup> | 5 ml   | TGM1 Sequence Variants  | 2-3 Months       |
| Leber Hereditary Optic Neuropathy Mutation Analysis,(LHON)    | Blood <sup>EDTA</sup> | 5 ml   | mtDNA NADH dehydrogenase gene mutations:<br>m.11778G>A, m.14484T>C, m.3460G>A   | 20 Days          |
| Leber's Congenital Amaurosis- Sequencing Panel                | Blood <sup>EDTA</sup> | 5 ml   | Whole gene sequencing of 15 Genes: CRB1, CRX, GUCY2D, LRAT, TULP1, RPE65, RPGRIP1, CEP290, RDH12, LCA5, SPATA7, AIPL1, IMPDH1, IQCB1, RD3 | 2 Months         |
| Leber's Congenital Amaurosis - Target Mutation Panel          | Blood <sup>EDTA</sup> | 5 ml   | 641 mutations in 13 genes (including AIPL1, CRB1, CRX, GUCY2D, LRAT, TULP1, MERTK, RPE65, RPGRIP1, CEP290, RDH12, LCA5, SPATA7)           | 2 Months         |
| Leber's Congenital Amaurosis 2 (LCA 2)                        | Blood <sup>EDTA</sup> | 5 ml   | RPE65 Whole Gene Sequencing   | 2-3 Months       |
| Leigh Syndrome  | Blood <sup>EDTA</sup> | 5 ml   | SURF1 Whole Gene Analysis   | 2 Months         |
| LEPRE1 Related Osteogenesis Imperfecta                        | Blood <sup>EDTA</sup> | 5 ml   | LEPRE1 Whole Gene Sequencing  | 2-3 Months       |
| Lesch - Nyhan Syndrome Genetic Analysis                       | Blood <sup>EDTA</sup> | 5 ml   | HPRT1 Whole Gene Sequencing   | 2-3 Months       |
| Lesch - Nyhan Syndrome Genetic Analysis                       | Blood <sup>EDTA</sup> | 5 ml   | HPRT1 Del/Dup Analysis (MLPA)   | 3-4 Months       |
| Li-Fraumeni Syndrome 1 Genetic Analysis<br>SBLA Syndrome      | Blood <sup>EDTA</sup> | 5 ml   | TP53 Exon 5-8 Sequence Anaysis  | 2 Months         |
| Li-Fraumeni Syndrome 1 Genetic Analysis<br>SBLA Syndrome      | Blood <sup>EDTA</sup> | 5 ml   | TP53 Whole Gene Sequencing  | 3 Months         |
| LIMB-GIRDLE MUSCULAR DYSTROPHY TYPE 1C (LGMD1C)               | Blood <sup>EDTA</sup> | 5 ml   | CAV3 Whole gene analysis  | 1 Month          |
| Limb-girdle Muscular Dystrophy Type 2A & 2B (LGMD2A & LGMD2B) | Blood <sup>EDTA</sup> | 5 ml   | CAPN3 and DYSF Whole gene analyses  | 2-3 Months       |
| Limb-girdle Muscular Dystrophy Type 2B (LGMD2B)               | Blood <sup>EDTA</sup> | 5 ml   | DYSF Whole gene analysis  | 2-3 Months       |

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| Test Name   | Sample                | Amount | Method  | Turn Around Time |
|---|-----------------------|--------|---|------------------|
| Limb-Girdle Muscular Dystrophy Type 2C (LGMD2C)                           | Blood <sup>EDTA</sup> | 5 ml   | SGCG Whole Gene Analysis  | 2 Month          |
| Loeys-Dietz Syndrome Genetic Analysis                                     | Blood <sup>EDTA</sup> | 5 ml   | TGFBR1 Whole Gene Analysis                                      | 1-2 Months       |
| Loeys-Dietz Syndrome Genetic Analysis                                     | Blood <sup>EDTA</sup> | 5 ml   | TGFBR2 Whole Gene Analysis                                      | 1-2 Months       |
| Lowe Syndrome Genetic Analysis  | Blood <sup>EDTA</sup> | 5 ml   | OCRL Whole Gene Analysis  | 2-3 Months       |
| LRRK2 Related Parkinson Disease<br>Parkinson Disease 8                    | Blood <sup>EDTA</sup> | 5 ml   | LRRK2 (PARK8) Sequence Analysis of Selected Exons (31. and 41.) | 2-3 Months       |
| Lung Cancer   | Blood <sup>EDTA</sup> | 5 ml   | EGFR Whole Gene Analysis  | 2-3 Months       |
| Lung Cancer   | Blood <sup>EDTA</sup> | 5 ml   | EGFR 18-21 exons  | 2-3 Months       |
| Lung Cancer   | Blood <sup>EDTA</sup> | 5 ml   | EGFR Deletion Duplication Analysis                              | 2 Months         |
| Lymphedema Distichiasis Syndrome<br>Meige disease                         | Blood <sup>EDTA</sup> | 5 ml   | FOXC2 Whole Gene Sequencing                                     | 3-4 Months       |
| Malignant Hyperthermia Genetic Analysis                                   | Blood <sup>EDTA</sup> | 5 ml   | Selected Exons in RYR1 (20 Exons)                               | 2-3 Months       |
| Malignant Hyperthermia Genetic Analysis                                   | Blood <sup>EDTA</sup> | 5 ml   | RYR1 Whole Gene Analysis  | 2-3 Months       |
| Marfan Syndrome 1   | Blood <sup>EDTA</sup> | 5 ml   | FBN1 Whole Gene Analysis  | 2-3 Months       |
| Marfan Syndrome 2   | Blood <sup>EDTA</sup> | 5 ml   | TGFBR2 Whole Gene Analysis                                      | 1-2 Months       |
| Mastocytosis Genetic Analysis   | Blood <sup>EDTA</sup> | 5 ml   | KIT D816V mutation in exon 17                                   | 1 Months         |
| Meckel-Gruber Syndrome Genetic Analysis                                   | Blood <sup>EDTA</sup> | 5 ml   | MKS1 exon 16-17 Analysis  | 2 Months         |
| Meckel-Gruber Syndrome Genetic Analysis                                   | Blood <sup>EDTA</sup> | 5 ml   | MKS1 Whole Gene Analysis  | 2-3 Months       |
| Medium-Chain Acyl-Coenzyme A<br>Dehydrogenase Deficiency Genetic Analysis | Blood <sup>EDTA</sup> | 5 ml   | ACADM Whole Gene Analysis                                       | 2-3 Months       |
| Medium-Chain Acyl-Coenzyme A<br>Dehydrogenase Deficiency Genetic Analysis | Blood <sup>EDTA</sup> | 5 ml   | ACADM 11. Exon Analysis   | 2-3 Months       |
| Melanoma-Pancreatic Cancer  | Blood <sup>EDTA</sup> | 5 ml   | CDKN2A Whole Gene Analysis                                      | 1 Months         |

| Test Name  | Sample                                | Amount         | Method  | Turn Around Time |
|--|---------------------------------------|----------------|---|------------------|
| MELAS, MTT (MIT) Genetic Analysis<br>Mitochondrial Encephalopathy, Lactic Acidosis,<br>And Stroke-Like Episodes  | Blood <sup>EDTA</sup>                 | 5 ml           | MT-TL1 (MTTL1) frequent 3 mutation<br>(A3243G, T3271C, A3252G) Analysis | 20 Days          |
| MELAS, MTT (MIT) Genetic Analysis<br>Mitochondrial Encephalopathy, Lactic Acidosis,<br>And Stroke-Like Episodes  | Blood <sup>EDTA</sup>                 | 5 ml           | MT-TL1 Whole Gene Analysis  | 2 Months         |
| Menkes Disease Genetic Analysis  | Blood <sup>EDTA</sup>                 | 5 ml           | ATP7A Sequence Alterations  | 3-4 Months       |
| MERRF Syndrome Genetic Analysis  | Blood <sup>EDTA</sup>                 | 5 ml           | A8344G mutation Analysis  | 1-2 Months       |
| MERRF Syndrome Genetic Analysis  | Blood <sup>EDTA</sup>                 | 5 ml           | A8344G, T8356C, G8363A, G8361A Mutation Analysis                        | 1-2 Months       |
| Mitochondrial DNA Deletion Analysis<br>Kearns-Sayre Syndrome<br>Pearson Syndrome<br>Progressive External Ophthalmoplegia (PEO)                                 | Blood <sup>EDTA</sup> /Skin<br>Biopsy | 5 ml,<br>50 mg | Mitochondrial DNA Deletions   | 2 Months         |
| Mitochondrial DNA Scanning Panel<br>MELAS, MERRF, NARP, LHON<br>Kearns-Sayre Syndrome<br>Pearson Syndrome<br>Maternally Transmitted Diabetes-Deafness Syndrome | Blood <sup>EDTA</sup> /Skin<br>Biopsy | 5 ml,<br>50 mg | Multiplex PCR/ASO, ARMS, Southern Blot                                  | 2 Months         |
| Mitochondrial Neurogastrointestinal Encephalopathy   | Blood <sup>EDTA</sup>                 | 5 ml           | ECGF1 (TYMP) (TP) Whole Gene Analysis                                   | 2-3 Months       |
| mRNA MART-1 (Melanoma)   | Blood <sup>EDTA</sup>                 | 5 ml           | Quantitative Analysis   | 20 Days          |
| mRNA MART-1 (Melanoma)   | Blood <sup>EDTA</sup>                 | 5 ml           | Qualitative Analysis  | 20 Days          |
| mRNA MGB (Mammaglobulin)   | Blood <sup>EDTA</sup>                 | 5 ml           | Quantitative Analysis   | 20 Days          |
| mRNA MGB (Mammaglobulin)   | Blood <sup>EDTA</sup>                 | 5 ml           | Qualitative Analysis  | 20 Days          |
| mRNA PSA (Prostate Cancer)   | Blood <sup>EDTA</sup>                 | 5 ml           | Quantitative Analysis   | 20 Days          |
| mRNA PSA (Prostate Cancer)   | Blood <sup>EDTA</sup>                 | 5 ml           | Qualitative Analysis  | 20 Days          |
| mRNA Tyrosinase (Melanoma)   | Blood <sup>EDTA</sup>                 | 5 ml           | Quantitative Analysis   | 20 Days          |

| Test Name   | Sample                | Amount | Method  | Turn Around Time |
|---|-----------------------|--------|---|------------------|
| mRNA Tyrosinase (Melanoma)  | Blood <sup>EDTA</sup> | 5 ml   | Qualitative Analysis                          | 20 Days          |
| MSUD (Maple Syrup Urine Disease) Genetic Analysis   | Blood <sup>EDTA</sup> | 5 ml   | BCKDHA, BCKDHB and DBT Sequence Variants      | 3 Months         |
| MTHFR A1298C Mutation Analysis  | Blood <sup>EDTA</sup> | 5 ml   | PCR & RFLP Targeted Mutation Analysis         | 5 Days           |
| MTHFR C677T Mutation Analysis   | Blood <sup>EDTA</sup> | 5 ml   | PCR & RFLP Targeted Mutation Analysis         | 5 Days           |
| Muckle-Wells Syndrome   | Blood <sup>EDTA</sup> | 5 ml   | CIAS1 (NLRP3) Whole Gene Analysis             | 2-3 Months       |
| Mucopolysaccharidosis Genetic Analysis Type I   | Blood <sup>EDTA</sup> | 5 ml   | IDUA Whole Gene Sequencing                    | 3-4 Months       |
| Mucopolysaccharidosis Genetic Analysis Typell   | Blood <sup>EDTA</sup> | 5 ml   | IDS Whole Gene Sequencing                     | 3-4 Months       |
| Muenke Syndrome Mutation Analysis   | Blood <sup>EDTA</sup> | 5 ml   | P250R substitution in FGFR3                   | 1-2 Months       |
| Multiple Endocrine Neoplasia Type 1 (MEN I)   | Blood <sup>EDTA</sup> | 5 ml   | MEN1 Whole Gene Analysis                      | 2-3 Months       |
| Muscular Dystrophy Type 2A (LGMD2A)   | Blood <sup>EDTA</sup> | 5 ml   | CAPN3 Whole Gene Analysis                     | 2-3 Months       |
| Myoclonic Dystonia  | Blood <sup>EDTA</sup> | 5 ml   | SGCE gene exon 1-7, 9 Analysis                | 1-2 Months       |
| Myotonia Congenita  | Blood <sup>EDTA</sup> | 5 ml   | CLCN1 Whole Gene Sequencing                   | 2-3 Months       |
| Myotonic Dystrophy Type I   | Blood <sup>EDTA</sup> | 5 ml   | DMPK gene STR Analysis                        | 45-60 Days       |
| Myotonic Dystrophy Type II  | Blood <sup>EDTA</sup> | 5 ml   | CNBP (ZNF9) gene STR Analysis                 | 60-75 Days       |
| N-Acetylglutamate Synthase Deficiency<br>Urea Cycle Disorders                                       | Blood <sup>EDTA</sup> | 5 ml   | NAGS Whole Gene Sequencing                    | 2-3 Months       |
| Narcolepsy Genetic Analysis   | Blood <sup>EDTA</sup> | 5 ml   | HCRT gene DQB1*0602 ve DQA1*0102 Alleles      | 1-2 Months       |
| NARP(Neuropaty, ataxia, Retinitis Pigmentosa) Mutation<br>Analysis<br>Leigh Syndrome                | Blood <sup>EDTA</sup> | 5 ml   | MTATP6 gene T8993G Targeted Mutation Analysis | 1 Months         |
| Nephrotic Syndrome 1 Genetic Analysis<br>Congenital, Finnish Nephrosis 1                            | Blood <sup>EDTA</sup> | 5 ml   | NPHS1 Whole Gene Analysis                     | 4-5 Months       |
| Nephrotic Syndrome 2 Genetic Analysis<br>Nephrotic Syndrome, Steroid-Resistant, Autosomal Recessive | Blood <sup>EDTA</sup> | 5 ml   | NPHS2 Whole Gene Analysis                     | 2-3 Months       |

| Test Name  | Sample                | Amount | Method  | Turn Around Time |
|--|-----------------------|--------|---|------------------|
| Neuroferritinopathy<br>Basal Ganglia Disease<br>Hyperferritinemia Cataract Syndrome              | Blood <sup>EDTA</sup> | 5 ml   | FTL Whole Gene Analysis   | 1-2 Months       |
| Neurofibromatosis Type 1 Genetic Analysis  | Blood <sup>EDTA</sup> | 6 ml   | NF1 Whole Gene Analysis   | 4-6 Months       |
| Neurofibromatosis Type 2 Genetic Analysis  | Blood <sup>EDTA</sup> | 5 ml   | NF2 Whole Gene and Deletion/ Duplication Analysis                 | 2-3 Months       |
| Neuronal Ceroid-Lipofuscinoses Infantile Type (INCL)<br>(Santavuori-Haltia)                      | Blood <sup>EDTA</sup> | 5 ml   | PPT1 gene T75P, R122W (c.364A>T) R151X Targeted Mutation Analysis | 20 Days          |
| Neuronal Ceroid-Lipofuscinoses Infantile Type (INCL)<br>(Santavuori-Haltia)                      | Blood <sup>EDTA</sup> | 5 ml   | PPT1 Whole Gene Analysis  | 2-3 Months       |
| Neuronal Ceroid-Lipofuscinoses Juvenile Type (JNCL)<br>(Batten Disease, Spielmeyer-Vogt Disease) | Blood <sup>EDTA</sup> | 5 ml   | CLN3 gene 1Kb Deletion Analysis                                   | 20 Days          |
| Neuronal Ceroid-Lipofuscinoses Late Infantile (LINCL)  | Blood <sup>EDTA</sup> | 5 ml   | TPP1 gene R208X,IVS5-1G>C Targeted Mutation Analysis              | 20 Days          |
| Neuronal Ceroid-Lipofuscinoses Late Infantile (LINCL)  | Blood <sup>EDTA</sup> | 5 ml   | TPP1 Whole Gene Analysis  | 2 Months         |
| Niemann-Pick Disease, Type A - Type B  | Blood <sup>EDTA</sup> | 5 ml   | SMPD1 Whole Gene Analysis   | 2-3 Months       |
| Niemann-Pick disease, Type C1  | Blood <sup>EDTA</sup> | 5 ml   | NPC1 Whole Gene Analysis  | 2 Months         |
| Niemann-Pick disease, Type C2  | Blood <sup>EDTA</sup> | 5 ml   | NPC2 Whole Gene Analysis  | 2 Months         |
| Nijmegen Breakage Syndrome<br>Berlin Breakage Syndrome<br>Seemanova Syndrome                     | Blood <sup>EDTA</sup> | 5 ml   | NBS1 gene 657 del5 Targeted Mutation Analysis                     | 1-2 Months       |
| Nijmegen Breakage Syndrome<br>Berlin Breakage Syndrome<br>Seemanova Syndrome                     | Blood <sup>EDTA</sup> | 5 ml   | NBS1 exon 6 Sequence Analysis                                     | 2 Months         |
| Nijmegen Breakage Syndrome<br>Berlin Breakage Syndrome<br>Seemanova Syndrome                     | Blood <sup>EDTA</sup> | 5 ml   | NBS1 Whole Gene Analysis  | 3 Months         |

| Test Name   | Sample                | Amount  | Method   | Turn Around Time |
|---|-----------------------|---------|--|------------------|
| Non-ketotic Hyperglycinemia (NKH)<br>Glycine Encephalopathy   | Blood <sup>EDTA</sup> | 5 ml    | GLDC Whole Gene Analysis   | 4-5 Months       |
| Non-ketotic Hyperglycinemia (NKH)<br>Glycine Encephalopathy   | Blood <sup>EDTA</sup> | 5 ml    | AMT Whole Gene Analysis  | 4-5 Months       |
| Noonan Syndrome Genetic Analysis  | Blood <sup>EDTA</sup> | 5 ml    | PTPN11 gene 3, 8, 9,13 exons   | 1-2 Months       |
| Noonan Syndrome Genetic Chip Analysis   | Blood <sup>EDTA</sup> | 5 ml    | Noonan Spectrum Chip, PTPN11, KRAS, SOS1, RAF Gene Scanning          | 2 Months         |
| Optic Atrophy, Autosomal Dominant   | Blood <sup>EDTA</sup> | 5 ml    | OPA1 Whole Gene Analysis   | 3-4 Months       |
| Optic Atrophy, Autosomal Dominant   | Blood <sup>EDTA</sup> | 5 ml    | 118 Marker OPA1 Targeted Mutation Analysis                           | 2-3 Months       |
| Ornithine Transcarbamylase Deficiency<br>Urea Cycle Disorders   | Blood <sup>EDTA</sup> | 5 ml    | OTC Whole Gene Sequencing  | 2-3 Months       |
| Osler-Rendu-Weber Syndrome 1; ORW1<br>Hereditary Hemorrhagic Telangiectasia Type 1; HHT1  | Blood <sup>EDTA</sup> | 5 ml    | ENG Whole Gene Analysis  | 2-3 Months       |
| Osler-Rendu-Weber Syndrome 1; ORW1<br>Hereditary Hemorrhagic Telangiectasia Type 1; HHT1  | Blood <sup>EDTA</sup> | 5 ml    | ENG and ACVRL1 Deletion/ Duplication Analysis (MLPA)                 | 2-3 Months       |
| Osler-Rendu-Weber Syndrome 2; ORW2<br>Hereditary Hemorrhagic Telangiectasia Type 2; HHT2  | Blood <sup>EDTA</sup> | 5 ml    | ACVRL1 Whole Gene Analysis   | 2-3 Months       |
| Osteopetrosis with Renal Tubular Acidosis Genetic Analysis<br>Osteopetrosis, Autosomal Recessive 3; OPTB3<br>Carbonic Anhydrase II Deficiency<br>Guibaud-Vainsel Syndrome<br>Marble Brain Disease | Blood <sup>EDTA</sup> | 5 ml    | CA2 Whole Gene Analysis  | 2 Months         |
| Osteoporosis Panel  | punch biopsy          | 5-10 mg | Vitamin D Receptor (VDR), COL1A1, E2 Receptor gene Mutation Analysis | 1-2 Months       |
| OTOF-Related Deafness Genetic Analysis  | Blood <sup>EDTA</sup> | 5 ml    | OTOF Whole Gene Analysis   | 4-5 Months       |
| PAI1 (SERPINE1) Genetic Analysis<br>Cardiovascular Risk   | Blood <sup>EDTA</sup> | 5 ml    | SERPINE1 gene 4G/5G Targeted Mutation Analysis                       | 20 Days          |
| Pancreatic Carcinoma Genetic Analysis   | Blood <sup>EDTA</sup> | 5 ml    | KRAS Whole Gene Analysis   | 2-3 Months       |

| Test Name  | Sample                | Amount | Method  | Turn Around Time |
|--|-----------------------|--------|---|------------------|
| Papillorenal Syndrome<br>Renal Coloboma Syndrome   | Blood <sup>EDTA</sup> | 5 ml   | PAX2 Whole Gene Analysis  | 2-3 Months       |
| Paramyotonia Congenita Of Von Eulenburg  | Blood <sup>EDTA</sup> | 5 ml   | SCN4A Whole Gene Analysis   | 2-3 Months       |
| PARK2-Related Juvenile Parkinsonism  | Blood <sup>EDTA</sup> | 5 ml   | PARK2 (PARKIN) Sequence Variants + Deletions (MLPA)               | 2-3 Months       |
| Pelizaeus-Merzbacher Disease   | Blood <sup>EDTA</sup> | 5 ml   | PLP1 Deletion Duplication Analysis                                | 1 Months         |
| Perforin Gene Mutation Analysis<br>Familial Hemophagocytic Lymphohistiocytosis 2 (FHL 2) | Blood <sup>EDTA</sup> | 5 ml   | PRF1 Whole Gene Analysis  | 1 Months         |
| Periodic Fever, Familial, Autosomal Dominant (TRAPS)<br>Genetic Analysis                 | Blood <sup>EDTA</sup> | 5 ml   | 2-5 Exons of TNFRSF1A   | 2-3 Months       |
| Periodic Fever, Familial, Autosomal Dominant (TRAPS)<br>Genetic Analysis                 | Blood <sup>EDTA</sup> | 5 ml   | TNFRSF1A Whole Gene Analysis                                      | 2 Months         |
| Peutz-Jeghers Syndrome Genetic Analysis  | Blood <sup>EDTA</sup> | 5 ml   | STK11 Whole Gene Analysis   | 2-3 Months       |
| Peutz-Jeghers Syndrome Genetic Analysis  | Blood <sup>EDTA</sup> | 5 ml   | STK11 Del/Dup Analysis (MLPA)                                     | 3-4 Months       |
| Phenylketonuria (PKU) Genetic Analysis<br>PAH Deficiency<br>Hyperphenylalaninemia        | Blood <sup>EDTA</sup> | 5 ml   | 6 Common Mutations (L48S, R158Q, R261Q, G272X, R408W, IVS10nt546) | 30 Days          |
| Pierre-Robin Syndrome Genetic Analysis   | Blood <sup>EDTA</sup> | 5 ml   | SOX9 Whole Gene Analysis  | 2-3 Months       |
| PINK1 Related Parkinson Disease, Parkinson Disease 6                                     | Blood <sup>EDTA</sup> | 5 ml   | PINK1 (PARK6) Whole Gene Analysis                                 | 2-3 Months       |
| Plasminogen Deficiency Type 1 Genetic Analysis   | Blood <sup>EDTA</sup> | 5 ml   | PLG Whole Gene Analysis   | 2-3 Months       |
| Polycystic Kidney Disease - Autosomal Dominant   | Blood <sup>EDTA</sup> | 5 ml   | PKD1 Whole Gene Analysis  | 2-3 Months       |
| Polycystic Kidney Disease - Autosomal Dominant   | Blood <sup>EDTA</sup> | 5 ml   | PKD2 Whole Gene Analysis  | 2-3 Months       |
| Polycystic Kidney Disease - Autosomal Dominant   | Blood <sup>EDTA</sup> | 5 ml   | PKD1 & PKD2 Whole Gene Analysis                                   | 4-5 Months       |
| Polycystic Liver Disease   | Blood <sup>EDTA</sup> | 5 ml   | PRKCSH and SEC63 Sekans Analysis                                  | 2-3 Months       |
| Porphyria Acute Intermittent   | Blood <sup>EDTA</sup> | 5 ml   | HMBS (PBG) Whole Gene Analysis                                    | 3 Months         |
| Prader Willi Syndrome  | Blood <sup>EDTA</sup> | 5 ml   | Methylation Analysis  | 2 Months         |
| Prader Willi Syndrome  | Blood <sup>EDTA</sup> | 5 ml   | UPD (PWCR)  | 2 Months         |

| Test Name   | Sample                | Amount | Method  | Turn Around Time |
|---|-----------------------|--------|---|------------------|
| Primary Ciliary Dyskinesia<br>Kartagener Syndrome                         | Blood <sup>EDTA</sup> | 5 ml   | Analysis of 43 mutation in DNAH5, 18 mutation in DNAI1                  | 2 Months         |
| Primary Hyperoxaluria Type 1  | Blood <sup>EDTA</sup> | 5 ml   | AGXT Whole Gene Analysis  | 2 Months         |
| Primary Hyperoxaluria Type 2  | Blood <sup>EDTA</sup> | 5 ml   | GRHPR Whole Gene Analysis   | 2 Months         |
| Primary Pulmonary Hypertension 1 Genetic Analysis                         | Blood <sup>EDTA</sup> | 5 ml   | BMPR2 Whole Gene Analysis   | 2-3 Months       |
| Primary Pulmonary Hypertension 1 Genetic Analysis                         | Blood <sup>EDTA</sup> | 5 ml   | BMPR2 Deletion/Duplication Analysis (MLPA)                              | 2-3 Months       |
| Prion Disease Genetic Analysis  | Blood <sup>EDTA</sup> | 5 ml   | PRNP Whole Gene Analysis  | 2 Months         |
| Progressive Pseudorheumatoid Arthropathy of Childhood                     | Blood <sup>EDTA</sup> | 5 ml   | WISP3 Whole Gene Analysis   | 2-3 Months       |
| Protein C Deficiency Syndrome   | Blood <sup>EDTA</sup> | 5 ml   | PROC Whole Gene Analysis  | 2-3 Months       |
| Prothrombin Gene Mutation Analysis (Trombophilia Risk)                    | Blood <sup>EDTA</sup> | 5 ml   | PCR+RFLP Targeted Mutation Analysis (G20210G>A)                         | 5 Days           |
| Proto-Oncogene RET Mutation Analysis<br>Ailesel Medüller Tiroid Karsinoma | Blood <sup>EDTA</sup> | 5 ml   | 10,11,13,14,16, exons   | 1-2 Months       |
| Pseudohypoadosteronism Type 1, Autosomal Recessive                        | Blood <sup>EDTA</sup> | 5 ml   | SCNN1A Whole Gene Analysis  | 2-3 Months       |
| Pseudohypoparathyroidism Genetic Analysis, Albright Syndrome              | Blood <sup>EDTA</sup> | 5 ml   | GNAS Whole Gene Sequencing  | 2 Months         |
| Pseudoxanthoma Elasticum  | Blood <sup>EDTA</sup> | 5 ml   | ABCC6 gene R1141X mutation & 23-29 exons deletion Analysis              | 1-2 Months       |
| Pseudoxanthoma Elasticum  | Blood <sup>EDTA</sup> | 5 ml   | ABCC6 Whole Gene Analysis   | 4-5 Months       |
| Retinitis Pigmentosa Autosomal Dominant                                   | Blood <sup>EDTA</sup> | 5 ml   | Microarray Analysis 370 mutation  | 2-3 Months       |
| Retinitis Pigmentosa Autosomal Recessive                                  | Blood <sup>EDTA</sup> | 5 ml   | Microarray Analysis 585 mutation  | 2-3 Months       |
| Rett Syndrome   | Blood <sup>EDTA</sup> | 5 ml   | PCR+RFLP Targeted Mutation Analysis (R168X, R270X, R255X, R294X, T158M) | 20 Days          |
| Rett Syndrome   | Blood <sup>EDTA</sup> | 5 ml   | MECP2 gene deletions (MLPA)   | 3 Months         |
| Rett Syndrome   | Blood <sup>EDTA</sup> | 5 ml   | MECP2 Whole Gene Analysis   | 3 Months         |
| Rheumatoid arthritis Rick Factors   | Blood <sup>EDTA</sup> | 5 ml   | Molecular study of genetic profile HLA DRB1*0401, 0404,                 | 2 Months         |

| Test Name   | Sample                | Amount | Method   | Turn Around Time |
|---|-----------------------|--------|--|------------------|
|   |                       |        | 0301   |                  |
| Rheumatoid arthritis Rick Factors                               | Blood <sup>EDTA</sup> | 5 ml   | SLC22A4 Whole Gene Analysis  | 2-3 Months       |
| Rheumatoid arthritis Rick Factors                               | Blood <sup>EDTA</sup> | 5 ml   | SLC22A4 intron 1 SNP   | 2 Months         |
| Rhizomelic Chondrodysplasia Punctata 1 Genetic Analysis (RCDP1) | Blood <sup>EDTA</sup> | 5 ml   | PEX7 Whole Gene Analysis   | 2-3 Months       |
| Rigid Spine Muscular Dystrophy Multiminicore Disease (MmD)      | Blood <sup>EDTA</sup> | 5 ml   | SEPN1 Whole Gene Analysis  | 2-3 Months       |
| Roberts Syndrome Genetic Analysis                               | Blood <sup>EDTA</sup> | 5 ml   | ESCO2 Whole Gene Analysis  | 2-3 Months       |
| ROR2 Related Robinow Syndrome                                   | Blood <sup>EDTA</sup> | 5 ml   | ROR2 Whole Gene Analysis and deletions (MLPA)                                  | 2 Months         |
| Rubinstein-Taybi Syndrome                                       | Blood <sup>EDTA</sup> | 5 ml   | CREBBP Whole Gene Analysis   | 2-3 Months       |
| Rubinstein-Taybi Syndrome                                       | Blood <sup>EDTA</sup> | 5 ml   | EP300 Whole Gene Analysis  | 2-3 Months       |
| RUSSEL-SILVER Syndrome  | Blood <sup>EDTA</sup> | 6 ml   | Methylation Analysis (Analysis of polymorphic markers on chromosome 7) and UPD | 2-3 Months       |
| Sandhoff Disease  | Blood <sup>EDTA</sup> | 5 ml   | HEXB Whole Gene Analysis & 16kb deletion Analysis                              | 2 Months         |
| Sandhoff Disease  | Blood <sup>EDTA</sup> | 5 ml   | HEXB CGH array del/dup Analysis  | 2 Months         |
| Segawa Syndrome (Otozomal Dominant)                             | Blood <sup>EDTA</sup> | 5 ml   | GCH1 Whole Gene Analysis   | 2-3 Months       |
| Segawa Syndrome (Otozomal Recessive)                            | Blood <sup>EDTA</sup> | 5 ml   | TH Whole Gene Analysis   | 3 Months         |
| Sepiapterin Reductase Deficiency                                | Blood <sup>EDTA</sup> | 5 ml   | SPR Whole Gene Analysis  | 2-3 Months       |
| Septo optic Dysplasia   | Blood <sup>EDTA</sup> | 5 ml   | HESX1 Whole Gene Analysis  | 2-3 Months       |
| Severe Congenital Neutropenia                                   | Blood <sup>EDTA</sup> | 5 ml   | HAX1 Whole Gene Analysis   | 2 Months         |
| Shögren-Larsson Syndrome Genetic Analysis                       | Blood <sup>EDTA</sup> | 5 ml   | ALDH3A2 Whole Gene Analysis  | 2 Months         |
| Shögren-Larsson Syndrome Genetic Analysis                       | Blood <sup>EDTA</sup> | 5 ml   | ALDH3A2 Whole Gene Analysis + 9kb deletion analysis (exon9)                    | 2-3 Months       |
| SHOX related Short Stature                                      | Blood <sup>EDTA</sup> | 5 ml   | SHOX Whole Gene Analysis   | 2-3 Months       |
| Shwachman-Diamond Syndrome Genetic Analysis                     | Blood <sup>EDTA</sup> | 5 ml   | SBDS Whole Gene Analysis   | 2-3 Months       |

| Test Name  | Sample                | Amount | Method  | Turn Around Time |
|--|-----------------------|--------|---|------------------|
| Shwachman-Diamond Syndrome Genetic Analysis  | Blood <sup>EDTA</sup> | 5 ml   | SBDS Common Mutation Analysis                                       | 1-2 Months       |
| Sickle Cell Anemia Carrier Testing   | Blood <sup>EDTA</sup> | 5 ml   | HBB whole gene sequencing   | 2 Weeks          |
| Smith Lemli Opitz Syndrome   | Blood <sup>EDTA</sup> | 5 ml   | DHCR7 Whole Gene Sequencing   | 2 Months         |
| Smith-Magenis Syndrome   | Blood <sup>EDTA</sup> | 5 ml   | RAI1 Whole Gene Analysis  | 2-3 Months       |
| SNCA Related Parkinson Disease<br>Familial Parkinson Disease Type 1  | Blood <sup>EDTA</sup> | 5 ml   | SNCA Whole Gene Sequencing + Deletion Analysis (MLPA)               | 2-3 Months       |
| Sotos Syndrome   | Blood <sup>EDTA</sup> | 5 ml   | NSD1 Whole Gene Sequencing  | 3-4 Months       |
| Spinal Muscular Atrophy (SMA) Carrier Analysis   | Blood <sup>EDTA</sup> | 5 ml   | PCR Based Dosage Analysis (SMN1 exon 7-8 Analysis)                  | 20 Days          |
| Spinal Muscular Atrophy I (SMA I)<br>Spinal Muscular Atrophy II (SMA II)<br>Spinal Muscular Atrophy III (SMA III)<br>Spinal Muscular Atrophy IV (SMA IV) | Blood <sup>EDTA</sup> | 5 ml   | SMN1 gene exon 7-8 Deletion Analysis & NAIP gene exon 5-6 Deletions | 20 Days          |
| Spinocerebellar Ataxia Panel 1 (1, 2, 3, 6, 7 Types)   | Blood <sup>EDTA</sup> | 5 ml   | Ataxin1, Ataxin2, Ataxin3, CACNA1A , Ataxin7 repeat regions         | 1 Months         |
| Spinocerebellar Ataxia Panel 2 (8, 10, 12, 17 Types)   | Blood <sup>EDTA</sup> | 5 ml   | ATXN80S, ATXN10, PPP2R2B, TBP repeat regions                        | 2-3 Months       |
| Spinocerebellar Ataxia Type 1, SCA 1   | Blood <sup>EDTA</sup> | 5 ml   | 100 CAG Repeats in ATXN1  | 20 Days          |
| Spinocerebellar Ataxia Type 10, SCA 10   | Blood <sup>EDTA</sup> | 5 ml   | ATTCT Repeats in ATXN10 Gene  | 2-3 Months       |
| Spinocerebellar Ataxia Type 12, SCA 12   | Blood <sup>EDTA</sup> | 5 ml   | CAG Repeats in PPP2R2B Gene   | 2-3 Months       |
| Spinocerebellar Ataxia Type 14, SCA 14   | Blood <sup>EDTA</sup> | 5 ml   | PRKCG Whole Gene Analysis   | 4-5 Months       |
| Spinocerebellar Ataxia Type 17, SCA 17   | Blood <sup>EDTA</sup> | 5 ml   | CAA/CAG Repeats in TBP Gene   | 2-3 Months       |
| Spinocerebellar Ataxia Type 2, SCA 2   | Blood <sup>EDTA</sup> | 5 ml   | ATXN2 CAG trinucleotide repeat expansions                           | 20 Days          |
| Spinocerebellar Ataxia Type 3, SCA 3   | Blood <sup>EDTA</sup> | 5 ml   | ATXN3 CAG trinucleotide repeat expansions                           | 20 Days          |
| Spinocerebellar Ataxia Type 5, SCA 5   | Blood <sup>EDTA</sup> | 5 ml   | SPTBN2 Gene mutations   | 2 Months         |
| Spinocerebellar Ataxia Type 6, SCA 6   | Blood <sup>EDTA</sup> | 5 ml   | CACNA1A CAG trinucleotide repeat expansions                         | 20 Days          |
| Spinocerebellar Ataxia Type 7, SCA 7   | Blood <sup>EDTA</sup> | 5 ml   | ATXN7 CAG trinucleotide repeat expansions                           | 20 Days          |

| Test Name   | Sample                | Amount | Method   | Turn Around Time |
|---|-----------------------|--------|--|------------------|
| Spinocerebellar Ataxia Type 8, SCA 8  | Blood <sup>EDTA</sup> | 5 ml   | CAGCTG Repeats in ATXN80S Gene                               | 2-3 Months       |
| Spondylocostal Dysostosis Type 2  | Blood <sup>EDTA</sup> | 5 ml   | MESP2 Whole Gene Analysis                                    | 2-3 Months       |
| Sprinting Performance Genetic Testing   | Blood <sup>EDTA</sup> | 5 ml   | ACTN3 R577X Mutation Analysis                                | 2 Week           |
| Stargardt Disease (Otozomal Recessive)  | Blood <sup>EDTA</sup> | 5 ml   | Most common ~500 Mutation on ABCR (ABCA4) Gene               | 2-3 Months       |
| Tay-Sachs Genetic Analysis  | Blood <sup>EDTA</sup> | 5 ml   | HEXA Whole Gene Analysis                                     | 3-4 Months       |
| TBX5 related Holt-Oram Syndrome   | Blood <sup>EDTA</sup> | 5 ml   | TBX5 gene Mutation Analysis                                  | 3 Months         |
| Thrombophilia Panel<br>- Faktör V Leiden Mutation<br>- Protrombin Gene Mutation<br>- MTHFR Gene Mutation    | Blood <sup>EDTA</sup> | 5 ml   | RFLP&PCR Targeted mutation Analysis                          | 5 Days           |
| Thrombotic Thrombocytopenic Purpura, (TTP)<br>Microangiopathic hemolytic anemia<br>Upshaw-schulman Syndrome | Blood <sup>EDTA</sup> | 5 ml   | ADAMTS13 Whole Gene Analysis                                 | 2-3 Months       |
| Thyroid Dyshormonogenesis 2A Genetic Analysis   | Blood <sup>EDTA</sup> | 5 ml   | TPO Whole Gene Analysis                                      | 4-5 Months       |
| TNF Genotyping  | Blood <sup>EDTA</sup> | 5 ml   | TNF- $\alpha$ promoter -238,-308,-488 regions                | 20 Days          |
| TPMT ( Thiopurine S-Methyltransferase) Genotyping   | Blood <sup>EDTA</sup> | 5 ml   | PCR&RFLP&ARMS with TPMT1-2-3A-3B-3C                          | 20 Days          |
| Transthyretin Amyloidosis Genetic Analysis  | Blood <sup>EDTA</sup> | 5 ml   | TTR V30M or V122I Mutations                                  | 1-2 Months       |
| Transthyretin Amyloidosis Genetic Analysis  | Blood <sup>EDTA</sup> | 5 ml   | TTR Whole Gene Analysis                                      | 2-3 Months       |
| TSH Receptor Genetic Analysis (Congenital Hypothyroidism)   | Blood <sup>EDTA</sup> | 5 ml   | TSHR 10. exon  | 1-2 Months       |
| TSH Receptor Genetic Analysis (Congenital Hypothyroidism)   | Blood <sup>EDTA</sup> | 5 ml   | TSHR Whole Gene Analysis                                     | 2 Months         |
| Tuberous sclerosis Type1 and Type2 Genetic Analysis   | Blood <sup>EDTA</sup> | 5 ml   | TSC1 and TSC2 Whole Gene Analysis ve Del/Dup Analysis (MLPA) | 3-4 Months       |
| Ullrich Congenital Muscular Dystrophy   | Blood <sup>EDTA</sup> | 5 ml   | COL6A1 Whole Gene Analysis                                   | 2 Months         |
| Ullrich Congenital Muscular Dystrophy   | Blood <sup>EDTA</sup> | 5 ml   | COL6A2 Whole Gene Analysis                                   | 2 Months         |
| Ullrich Congenital Muscular Dystrophy   | Blood <sup>EDTA</sup> | 5 ml   | COL6A3 Whole Gene Analysis                                   | 2 Months         |
| Ullrich Congenital Muscular Dystrophy   | Blood <sup>EDTA</sup> | 5 ml   | COL6A1 & COL6A2 & COL6A3 Whole Gene Analysis                 | 2-3 Months       |

| Test Name   | Sample                | Amount | Method   | Turn Around Time |
|---|-----------------------|--------|--|------------------|
| Van Der Knaap Disease Genetic Analysis (Megalencephalic Leukoencephalopathy With Subcortical Cysts)               | Blood <sup>EDTA</sup> | 5 ml   | MLC1 Whole Gene Sequencing ve Del/Dup Analysis (MLPA)    | 2-3 Months       |
| VEGF Genotyping   | Blood <sup>EDTA</sup> | 5 ml   | VEGF +813 CC bölgesi                                     | 20 Days          |
| Vitamin D Dependent Rickets   | Blood <sup>EDTA</sup> | 5 ml   | FGF23 Whole Gene Analysis                                | 1-2 Months       |
| Vitamin D Dependent Rickets   | Blood <sup>EDTA</sup> | 5 ml   | PHEX Whole Gene Analysis                                 | 2-3 Months       |
| Von Hippel-Lindau Syndrome  | Blood <sup>EDTA</sup> | 5 ml   | VHL Whole Gene Analysis                                  | 2-3 Months       |
| Von Hippel-Lindau Syndrome  | Blood <sup>EDTA</sup> | 5 ml   | VHL Whole Gene Sequencing and Del/Dup Analysis           | 2-3 Months       |
| Waardenburg Syndrome Type 1 Genetic Analysis  | Blood <sup>EDTA</sup> | 5 ml   | PAX3 gene mutations                                      | 2-3 Months       |
| Waardenburg Syndrome Type 2 Genetic Analysis  | Blood <sup>EDTA</sup> | 5 ml   | MITF Whole Gene Analysis                                 | 2-3 Months       |
| Waardenburg Syndrome Type 4 Genetic Analysis (EDN3)   | Blood <sup>EDTA</sup> | 5 ml   | EDN3 Whole Gene Analysis                                 | 2-3 Months       |
| Waardenburg Syndrome Type 4 Genetic Analysis (EDNRB)  | Blood <sup>EDTA</sup> | 5 ml   | EDNRB Whole Gene Analysis                                | 2-3 Months       |
| Waardenburg Syndrome Type 4 Genetic Analysis (SOX10)  | Blood <sup>EDTA</sup> | 5 ml   | SOX10 Whole Gene Analysis                                | 2-3 Months       |
| Walker-Warburg Syndrome Genetic Analysis  | Blood <sup>EDTA</sup> | 5 ml   | POMT1 Whole gene sequencing                              | 3-4 Months       |
| Walker-Warburg Syndrome Genetic Analysis  | Blood <sup>EDTA</sup> | 5 ml   | POMT2 Whole gene sequencing                              | 3-4 Months       |
| Walker-Warburg Syndrome Genetic Analysis  | Blood <sup>EDTA</sup> | 5 ml   | FKRP Whole gene sequencing                               | 3 Months         |
| Walker-Warburg Syndrome Genetic Analysis  | Blood <sup>EDTA</sup> | 5 ml   | FKTN Whole gene sequencing                               | 3 Months         |
| Walker-Warburg Syndrome Genetic Analysis  | Blood <sup>EDTA</sup> | 5 ml   | LARGE Whole gene sequencing                              | 3 Months         |
| Walker-Warburg Syndrome Genetic Analysis  | Blood <sup>EDTA</sup> | 5 ml   | POMGNT1 Whole gene sequencing                            | 3 Months         |
| Walker-Warburg Syndrome Genetic Analysis  | Blood <sup>EDTA</sup> | 5 ml   | FKTN, LARGE, POMGNT1, POMT1, POMT2 Whole gene sequencing | 4 Months         |
| WAS Related Disorders<br>Wiskott Aldrich Syndrome<br>X-Linked thrombocytopenia<br>X-linked congenital neutropenia | Blood <sup>EDTA</sup> | 5 ml   | WAS Whole Gene Analysis                                  | 2-3 Months       |
| Wilson Disease<br>(Hepatolenticular Degeneration)   | Blood <sup>EDTA</sup> | 5 ml   | DGGE & Sequencing Analysis for ATP7B 13 exon             | 4-5 Months       |

| Test Name  | Sample                | Amount | Method   | Turn Around Time |
|--|-----------------------|--------|--|------------------|
| Wilson Disease<br>(Hepatolenticular Degeneration)                            | Blood <sup>EDTA</sup> | 5 ml   | DGGE & Sequencing Analysis for ATP7B Whole Gene  | 3 Months         |
| Wolfram Syndrome Genetic Analysis  | Blood <sup>EDTA</sup> | 5 ml   | WFS1 Whole Gene DHPLC Sequencing   | 2 Months         |
| X-Linked Adrenoleukodystrophy (X-ADL)<br>Adrenomyeloneuropathy               | Blood <sup>EDTA</sup> | 5 ml   | ABCD1 Whole Gene Analysis  | 2 Months         |
| X-Linked Adrenoleukodystrophy (X-ADL)<br>Adrenomyeloneuropathy               | Blood <sup>EDTA</sup> | 5 ml   | ABCD1 Whole Gene Analysis and Deletion/duplication<br>Analysis                                 | 2 Months         |
| X-Linked Agammaglobulinemia Genetic Analysis                                 | Blood <sup>EDTA</sup> | 5 ml   | BTK gene sequence variants   | 2-3 Months       |
| X-linked Lymphoproliferative Syndrome  | Blood <sup>EDTA</sup> | 5 ml   | SH2D1A Whole Gene Analysis   | 1-2 Months       |
| X-Linked Sideroblastic Anemia Genetic Analysis                               | Blood <sup>EDTA</sup> | 5 ml   | ALAS2 Whole Gene Analysis  | 3-4 Months       |
| Y Chromosome Microdeletions  | Blood <sup>EDTA</sup> | 5 ml   | Multiplex PCR  | 15 Days          |
| Zellweger Syndrome Spectrum Genetic Analysis                                 | Blood <sup>EDTA</sup> | 5 ml   | PEX1 mutations (c.2097dupT, p.Gly843Asp, c.2916delA)   | 1-2 Months       |
| Zellweger Syndrome Spectrum Genetic Analysis                                 | Blood <sup>EDTA</sup> | 5 ml   | PEX1 (exon 13, 15, 18), PEX6 (exon1),<br>PEX12 (exon 2, 3), PEX26 (exon1, 2) Sequence Analysis | 2-3 Months       |
| Zellweger Syndrome Spectrum Genetic Analysis                                 | Blood <sup>EDTA</sup> | 5 ml   | PEX1 Whole Gene Analysis   | 2-3 Months       |
| Zellweger Syndrome Spectrum Genetic Analysis                                 | Blood <sup>EDTA</sup> | 5 ml   | PEX6, PEX12, PEX26 Whole Gene Analysis   | 2-3 Months       |
| Frontometaphyseal Dysplasia Carrier Testing                                  | Blood <sup>EDTA</sup> | 5 ml   | FLNA Targeted Mutation analysis  | 4-5 Weeks        |
| Surfactant Metabolism Dysfunction<br>Surfactant Protein B Deficiency         | Blood <sup>EDTA</sup> | 5 ml   | SFTPB Whole Gene Analysis  | 2-3 Months       |
| Thyroid Hormone Resistance<br>THRB Gene Analysis                             | Blood <sup>EDTA</sup> | 5 ml   | THRB Whole Gene Analysis   | 1-2 Months       |
| Thyroid Hormone Resistance<br>THRB Gene Analysis                             | Blood <sup>EDTA</sup> | 5 ml   | THRB Exon 4 - 10 Sequencing  | 3 - 5 Months     |
| EPCR Genetic Analysis  | Blood <sup>EDTA</sup> | 5 ml   | EPCR Whole Gene Analysis   | 2 Months         |
| <b>Cortisol 11-Beta-Ketoreductase Deficiency</b>                             | Blood <sup>EDTA</sup> | 5 ml   | HSD112B Whole Gene Analysis  | 2 - 3 Months     |
| <b>Surfactant Protein A1/A2 Deficiency</b>                                   | Blood <sup>EDTA</sup> | 5 ml   | SFTPA1/A2 Whole Gene Analysis  | 1 - 2 Months     |
| <b>Surfactant Protein C Deficiency<br/>Surfactant Metabolism Dysfunction</b> | Blood <sup>EDTA</sup> | 5 ml   | SFTPC Whole Gene Analysis  | 2-4 Months       |

| Test Name   | Sample                | Amount | Method  | Turn Around Time |
|---|-----------------------|--------|---|------------------|
| <b>Spastic paraplegia 11</b>  | Blood <sup>EDTA</sup> | 5 ml   | SPG11 Whole Gene Analysis   | 2 – 3 Months     |
| <b>Griscelli Syndrome Type 1</b>  | Blood <sup>EDTA</sup> | 5 ml   | MYO5A exon 19 and 28 sequence analysis  | 2 – 3 Months     |
| <b>Griscelli Syndrome Type 2</b>  | Blood <sup>EDTA</sup> | 5 ml   | RAB27A exon 4,5,6 sequence analysis   | 2 – 3 Months     |
| <b>Pena-Shokeir Syndrome Type 1<br/>Fetal Akinesia Deformation Sequence</b> | Blood <sup>EDTA</sup> | 5 ml   | RAPSN Whole Gene Analysis   | 2-3 Months       |
| <b>Pena-Shokeir Syndrome Type 1<br/>Fetal Akinesia Deformation Sequence</b> | Blood <sup>EDTA</sup> | 5 ml   | DOK7 Whole Gene Analysis  | 2-3 Months       |
| <b>Hypertrophic Cardiomyopathy (CMH or HCM)</b>                             | Blood <sup>EDTA</sup> | 5 ml   | MYH7, MYBPC3, TNNI3, TPM1, TNNT2, MYL2, MYL3, TNNC1, CSRP3, PRKAG2, PLN, and ACTC1 whole gene analysis (Resequencing based on DNA microarray) | 3-4 Months       |
| <b>Long-Chain 3-Hydroxyacyl-Coa Dehydrogenase Deficiency</b>                | Blood <sup>EDTA</sup> | 5 ml   | HADHA Whole Gene Analysis   | 6 – 7 Months     |
| <b>Long-Chain 3-Hydroxyacyl-Coa Dehydrogenase Deficiency</b>                | Blood <sup>EDTA</sup> | 5 ml   | HADHA Gene Common Mutation Analysis (1528G>C & 1132 C>T )   | 1-2 Months       |
| <b>Netherton Syndrome</b>   | Blood <sup>EDTA</sup> | 5 ml   | SPINK5 Whole Gene Analysis  | 2-3 Months       |
| <b>Lysosomal Acid Lipase Deficiency</b>                                     | Blood <sup>EDTA</sup> | 5 ml   | LIPA Whole Gene Analysis  | 2-3 Months       |
| <b>Metaphyseal Chondrodysplasia, Schmid Type</b>                            | Blood <sup>EDTA</sup> | 5 ml   | COL10A1 gene C & N Terminal Domains Analyses  | 4 – 6 Weeks      |
| <b>Metaphyseal Chondrodysplasia, Schmid Type</b>                            | Blood <sup>EDTA</sup> | 5 ml   | COL10A1 Whole Gene Analysis   | 2 – 3 Months     |
| <b>Ichthyosis, X-Linked</b>   | Blood <sup>EDTA</sup> | 5 ml   | STS Whole Gene Analysis   | 2 – 3 Months     |