**لیست ژن های مربوط به بیماری ها که توسط کمپانی CeGaT آلمان انجام می گردد**

(List of genes that the CeGaT offers for genetic testing)

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| --- | --- | --- | --- | --- | --- |
| **Gene** | **Disease** | **Step1** | **Step2** | **Step3** | **Kb** |
| **15q11-13** | Angelman Syndrome | Deletion and duplication analysis |  |  | 26700 |
| **15q11-13** | Prader-Willi Syndrome | Deletion and duplication analysis |  |  | 26700 |
| **AAAS** | Achalasia-addisonianism-alacrimia syndrome | Sequencing of all coding exons of the gene |  |  | 1.6 |
| **ABCA3** | Pulmonary surfactant metabolism dysfunction type 3 (ABCA3 deficiency) | Sequencing of all coding exons of the gene |  |  | 5.1 |
| **ABCA3** | Surfactant metabolism dysfunction, pulmonary, 3 | Sequencing of all coding exons of the gene |  |  | 5.1 |
| **ABCA4** | Age-Dependent Macula Degeneration | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 6.8 |
| **ABCA4** | Autosomal Recessive Retinitis Pigmentosa | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 6.8 |
| **ABCA4** | Cone-Rod-Dystrophy | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 6.8 |
| **ABCA4** | Stargardt Disease | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 6.8 |
| **ABCB4** | Cholestasis, intrahepatic, of pregnancy, 3 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 3.9 |
| **ABCB4** | Cholestasis, progressive familial intrahepatic 3 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 3.9 |
| **ABCB4** | Gallbladder disease 1 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 3.9 |
| **ABCD1** | Adrenoleukodystrophy | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.2 |
| **ABHD12** | Polyneuropathy, Deafness, Ataxia, Retinitis Pigmentosa and Cataract | Sequencing of all coding exons of the gene |  |  | 1.2 |
| **ABHD5** | Chanarin-Dorfman syndrome | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **ACADS** | Short Chain Acyl-CoA Dehydrogenase Deficiency | Sequencing of all coding exons of the gene |  |  | 1.2 |
| **ACTA1** | Congenital Fiber-Type Disproportion Myopathy | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **ACTA1** | Nemaline Myopathy | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **ACTA2** | AORTIC ANEURYSM, FAMILIAL THORACIC 6; AAT6 | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **ACTA2** | MOYAMOYA DISEASE 5; MYMY5 | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **ACTA2** | MULTISYSTEMIC SMOOTH MUSCLE DYSFUNCTION SYNDROME | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **ACTG2** | Visceral myopathy | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **ACVRL1** | Telangiectasia, hereditary hemorrhagic, type 2 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.5 |
| **ADAR** | Aicardi-Goutieres syndrome 6 | Sequencing of all coding exons of the gene |  |  | 3.7 |
| **ADAR** | Dyschromatosis symmetrica hereditaria | Sequencing of all coding exons of the gene |  |  | 3.7 |
| **ADCY5** | Dyskinesia, familial, with facial myokymia | Sequencing of all coding exons of the gene |  |  | 3.8 |
| **ADSL** | Adenylosuccinate Lyase Deficiency | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.5 |
| **AGA** | Aspartylglycosaminuria | Sequencing of all coding exons of the gene |  |  | 1 |
| **AGL** | Glycogen Storage Disease  Type III | Sequencing of all coding exons of the gene |  |  | 4.6 |
| **AGXT** | Hyperoxaluria, Primary, Type 1 | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 1.2 |
| **AHI1** | Joubert Syndrome | Sequencing of all coding exons of the gene |  |  | 3.6 |
| **AIPL1** | Cone-Rod-Dystrophy | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.2 |
| **AIPL1** | Leber congenital amaurosis 4 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.2 |
| **AIPL1** | Retinitis pigmentosa, juvenile | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.2 |
| **AKT3** | Hemimegalencephaly | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **AKT3** | Hemimegalencephaly | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **ALDH4A1** | Hyperprolinemia, Type II | Sequencing of all coding exons of the gene |  |  | 1.7 |
| **ALDH5A1** | Succinic semialdehyde dehydrogenase deficiency | Sequencing of all coding exons of the gene |  |  | 1.6 |
| **ALDH7A1** | Pyridoxine-Dependent Epilepsy | Sequencing of all coding exons of the gene |  |  | 1.6 |
| **ALDOB** | Fructose intolerance | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.1 |
| **ALG12** | CDG-Syndrome 1G | Sequencing of all coding exons of the gene |  |  | 1.5 |
| **ALG2** | CDG-Syndrome 1I | Sequencing of all coding exons of the gene |  |  | 1.3 |
| **ALG3** | CDG-Syndrome 1D | Sequencing of all coding exons of the gene |  |  | 1.3 |
| **ALG6** | CDG-Syndrom 1C | Sequencing of all coding exons of the gene |  |  | 1.5 |
| **ALG8** | CDG Syndrome 1H | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.6 |
| **ALG9** | CDG-Syndrome 1L | Sequencing of all coding exons of the gene |  |  | 1.9 |
| **ALMS1** | Alstrom Syndrome | Sequencing of all coding exons of the gene |  |  | 12.5 |
| **ALPL** | Hypophosphatasia, adult | Sequencing of all coding exons of the gene |  |  | 1.6 |
| **ALPL** | Hypophosphatasia, childhood | Sequencing of all coding exons of the gene |  |  | 1.6 |
| **ALPL** | Hypophosphatasia, infantile | Sequencing of all coding exons of the gene |  |  | 1.6 |
| **ALPL** | Odontohypophosphatasia | Sequencing of all coding exons of the gene |  |  | 1.6 |
| **ALS2** | Cardiac Diseases | Sequencing of all coding exons of the gene |  |  | 5 |
| **AMACR** | Alpha-Methylacyl-CoA Racemase Deficiency | Sequencing of all coding exons of the gene |  |  | 1.2 |
| **AMPD1** | Myoadenylate Deaminase Deficiency | Sequencing of all coding exons of the gene |  |  | 2.3 |
| **AMT** | Glycine Encephalopathy | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.2 |
| **ANG** | Cardiac Diseases | Sequencing of all coding exons of the gene |  |  | 0.4 |
| **ANO10** | Spinocerebellar Ataxia, autosomal-recessive 10, SCAR10 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2 |
| **ANO3** | Dystonia 24 | Sequencing of all coding exons of the gene |  |  | 2.9 |
| **AP2S1** | Hypocalciuric hypercalcemia, familial, type III | Sequencing of all coding exons of the gene |  |  | 0.4 |
| **APOA1** | Amyloidosis | Sequencing of all coding exons of the gene |  |  | 0.8 |
| **APOA1** | Amyloidosis, 3 or more types | Sequencing of all coding exons of the gene |  |  | 0.8 |
| **APOA1** | ApoA-I deficiency | Sequencing of all coding exons of the gene |  |  | 0.8 |
| **APOA2** | Apolipoprotein A-II deficiency | Sequencing of all coding exons of the gene |  |  | 0.3 |
| **APOA5** | Hyperchylomicronemia, late-onset | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **APP** | Alzheimer Dementia | Deletion and duplication analysis; Sequencing of hotspots | Deletion and duplication analysis; Deletion and duplication analysis |  | 2.3 |
| **APTX** | Ataxia with Oculomotor Apraxia 1 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1 |
| **AR** | Androgen Insensitivity Syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis | Fragment length analysis | 2.8 |
| **AR** | Spinal and Bulbar Muscular Atrophy | Sequencing of all coding exons of the gene | Deletion and duplication analysis | Fragment length analysis | 2.8 |
| **ARFGEF2** | Periventricular nodular Heterotopia | Sequencing of all coding exons of the gene |  |  | 5.4 |
| **ARHGEF9** | Hyperekplexia | Sequencing of all coding exons of the gene |  |  | 1.6 |
| **ARSA** | Metachromatic Leukodystrophy | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.5 |
| **ARSB** | Mucopolysaccharidosis type VI (Maroteaux-Lamy) | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.6 |
| **ARSI** | Spastic Paraplegia Type 66 | Sequencing of all coding exons of the gene |  |  | 1.7 |
| **ARX** | Agenesis of Corpus Callosum with Abnormal Genitalia | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.7 |
| **ARX** | Partington X-Linked Mental Retardation Syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.7 |
| **ARX** | West-Syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.7 |
| **ARX** | West-Syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.7 |
| **ARX** | X-Linked Lissencephaly with Ambiguous Genitalia | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.7 |
| **ARX** | X-Linked Lissencephaly with Ambiguous Genitalia | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.7 |
| **ASAH1** | Farber lipogranulomatosis | Sequencing of all coding exons of the gene |  |  | 1.2 |
| **ASAH1** | Spinal muscular atrophy with progressive myoclonic epilepsy | Sequencing of all coding exons of the gene |  |  | 1.2 |
| **ASPM** | Primary Autosomal Recessive Microcephaly Type 5 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 10.4 |
| **ASXL1** | Bohring-Opitz Syndrome | Sequencing of all coding exons of the gene |  |  | 4.6 |
| **ATF6** | Achromatopsia 7 | Sequencing of all coding exons of the gene |  |  | 2 |
| **ATL1** | Neuropathy, hereditary sensory, type ID | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.7 |
| **ATL1** | Spastic paraplegia 3A, autosomal dominant | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.7 |
| **ATM** | Ataxia Teleangiectatica (AT) | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 9.2 |
| **ATP13A2** | Kufor-Rakeb Syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 3.5 |
| **ATP1A2** | Familial hemiplegic Migraine Type 2 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 3.1 |
| **ATP1A3** | Rapid Onset Dystonia with Parkinsonism | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 3.1 |
| **ATP2A1** | Brody myopathy | Sequencing of all coding exons of the gene |  |  | 3 |
| **ATP2A2** | Darier-White Disease | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 3.1 |
| **ATP2C1** | Hailey-Hailey disease | Sequencing of all coding exons of the gene |  |  | 2.9 |
| **ATP6AP2** | Mental retardation, X-linked, with epilepsy | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **ATP6V0A4** | Renal tubular acidosis, distal, autosomal recessive | Sequencing of all coding exons of the gene |  |  | 2.5 |
| **ATP6V1B1** | Renal tubular acidosis with deafness | Sequencing of all coding exons of the gene |  |  | 1.5 |
| **ATP7A** | ATP7A-Related Copper Transport Disorders | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 4.5 |
| **ATP7A** | Menkes Disease | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 4.5 |
| **ATP7B** | Wilson Disease | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 4.4 |
| **ATP8A2** | Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4 | Sequencing of all coding exons of the gene |  |  | 3.6 |
| **ATP8B1** | Cholestasis, benign recurrent intrahepatic | Sequencing of all coding exons of the gene |  |  | 3.8 |
| **ATP8B1** | Cholestasis, intrahepatic, of pregnancy, 1 | Sequencing of all coding exons of the gene |  |  | 3.8 |
| **ATP8B1** | Cholestasis, progressive familial intrahepatic 1 | Sequencing of all coding exons of the gene |  |  | 3.8 |
| **ATRX** | Alpha-Thalassemia X-Linked Intellectual Disability Syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 7.5 |
| **ATXN1** | Spinocerebellar Ataxia 1 | Repeat expansion analysis |  |  | 2.4 |
| **ATXN2** | Spinocerebellar Ataxia 2 | Repeat expansion analysis |  |  | 3.9 |
| **ATXN3** | Machado-Joseph Disease | Repeat expansion analysis |  |  | 1.1 |
| **ATXN7** | Spinocerebellar Ataxia 7 | Repeat expansion analysis |  |  | 2.8 |
| **B9D1** | Meckel Syndrome | Sequencing of all coding exons of the gene |  |  | 0.8 |
| **B9D2** | Meckel Syndrome | Sequencing of all coding exons of the gene |  |  | 0.5 |
| **BBS1** | Bardet-Biedl Syndrome | Sequencing of all coding exons of the gene |  |  | 1.8 |
| **BBS2** | Bardet Biedl Syndrome | Sequencing of all coding exons of the gene |  |  | 2.2 |
| **BCKDHA** | Maple Syrup Urine Disease Type 1A | Sequencing of all coding exons of the gene |  |  | 1.3 |
| **BCKDHB** | Maple Syrup Urine Disease | Sequencing of all coding exons of the gene |  |  | 1.2 |
| **BEST1** | Autosomal Dominant  Retinitis Pigmentosa | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.8 |
| **BEST1** | Autosomal Recessive Retinitis Pigmentosa | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.8 |
| **BFSP2** | Cataract, autosomal dominant | Sequencing of all coding exons of the gene |  |  | 1.2 |
| **BICD2** | Spinal muscular atrophy | Sequencing of all coding exons of the gene |  |  | 2.6 |
| **BSCL2** | Lipodystrophy, congenital generalized, type 2 | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **BSCL2** | Neuropathy, distal hereditary motor, type V | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **BSCL2** | Silver spastic paraplegia syndrome | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **BTD** | Biotinidase deficiency | Sequencing of all coding exons of the gene |  |  | 1.6 |
| **C10orf11** | Albinism, oculocutaneous, type VII | Sequencing of all coding exons of the gene |  |  | 0.6 |
| **C12orf65** | Combined Oxidative Phosphorylation Deficiency 7 | Sequencing of all coding exons of the gene |  |  | 0.5 |
| **C1QA** | C1q DEFICIENCY; C1QD | Sequencing of all coding exons of the gene |  |  | 0.7 |
| **C1QC** | C1q DEFICIENCY; C1QD | Sequencing of all coding exons of the gene |  |  | 0.7 |
| **C1QTNF5** | Late-Onset Retinal Degeneration | Sequencing of all coding exons of the gene |  |  | 0.7 |
| **C9orf72** | Cardiac Diseases | Repeat expansion analysis |  |  | 1.4 |
| **CABP4** | Congenital Stationary Night-Blindness | Sequencing of all coding exons of the gene |  |  | 0.8 |
| **CACNA1A** | Episodic Ataxia - Type 2 | Sequencing of all coding exons of the gene | Deletion and duplication analysis | Repeat expansion analysis | 7.5 |
| **CACNA1A** | Familial hemiplegic Migraine Type 1 | Sequencing of all coding exons of the gene | Deletion and duplication analysis | Repeat expansion analysis | 7.5 |
| **CACNA1A** | Paroxysmal Familial Ataxia | Sequencing of all coding exons of the gene | Deletion and duplication analysis | Repeat expansion analysis | 7.5 |
| **CACNA1A** | Spinocerebellar Ataxia Type 6 | Sequencing of all coding exons of the gene | Deletion and duplication analysis | Repeat expansion analysis | 7.5 |
| **CACNA1B** | Dystonia 23 | Sequencing of all coding exons of the gene |  |  | 7 |
| **CACNA1F** | Cone-Rod-Dystrophy | Sequencing of all coding exons of the gene |  |  | 5.9 |
| **CACNA1F** | Congenital Stationary Night-Blindness | Sequencing of all coding exons of the gene |  |  | 5.9 |
| **CACNA1H** | Childhood absence Epilepsy | Sequencing of all coding exons of the gene |  |  | 7.1 |
| **CACNA1H** | Epilepsy, childhood absence, susceptibility to, 6 | Sequencing of all coding exons of the gene |  |  | 7.1 |
| **CACNB4** | Episodic Ataxia - Type 2 | Sequencing of all coding exons of the gene |  |  | 1.6 |
| **CACNB4** | Idiopathic generalised Epilepsy (IGE) | Sequencing of all coding exons of the gene |  |  | 1.6 |
| **CACNB4** | Juvenile Myoclonus Epilepsy (JME) | Sequencing of all coding exons of the gene |  |  | 1.6 |
| **CAPN3** | Limb-Girdle Muscular Dystrophies, autosomal recessive | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.5 |
| **CASK** | CASK-Related X-Linked Mental Retardation | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 2.8 |
| **CASK** | FG-Syndrome | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 2.8 |
| **CASR** | Autosomal Dominant Hypocalcemia | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 3.3 |
| **CASR** | Familial hypocalciuric Hypercalcemia, Type 1 | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 3.3 |
| **CASR** | Neonatal Severe Primary Hyperparathyroidism | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 3.3 |
| **CCDC65** | Ciliary dyskinesia, primary, 27 | Sequencing of all coding exons of the gene |  |  | 1.5 |
| **CCND2** | Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3 | Sequencing of all coding exons of the gene |  |  | 0.9 |
| **CD96** | C syndrome | Sequencing of all coding exons of the gene |  |  | 1.8 |
| **CDHR1** | Cone-Rod-Dystrophy | Sequencing of all coding exons of the gene |  |  | 2.6 |
| **CDHR1** | Retinitis Pigmentosa | Sequencing of all coding exons of the gene |  |  | 2.6 |
| **CDK5RAP2** | Primary Autosomal Recessive Microcephaly Type 3 | Sequencing of all coding exons of the gene |  |  | 5.7 |
| **CDKL5** | Epileptic Encephalopathy, Early Infantile, 2 | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 3.1 |
| **CDKL5** | Rett Syndrome, Congenital Variant | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 3.1 |
| **CDKL5** | West-Syndrome | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 3.1 |
| **CDKL5** | West-Syndrome | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 3.1 |
| **CDON** | Holoprosencephaly | Sequencing of all coding exons of the gene |  |  | 3.9 |
| **CEP63** | Seckel Syndrome | Sequencing of all coding exons of the gene |  |  | 2.1 |
| **CFH** | Age-Dependent Macula Degeneration | Sequencing of all coding exons of the gene |  |  | 3.7 |
| **CFH** | Atypical Hemolytic-Uremic Syndrome | Sequencing of all coding exons of the gene |  |  | 3.7 |
| **CFH** | Basal Laminar Drusen | Sequencing of all coding exons of the gene |  |  | 3.7 |
| **CFTR** | Congenital bilateral absence of vas deferens | Sequencing of hotspots | Sequencing of all coding exons of the gene | Deletion and duplication analysis | 4.4 |
| **CFTR** | Cystic fibrosis | Sequencing of hotspots | Sequencing of all coding exons of the gene | Deletion and duplication analysis | 4.4 |
| **CHCHD10** | Frontotemporal dementia and/or amyotrophic lateral sclerosis 2 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 0.5 |
| **CHCHD10** | Myopathy, isolated mitochondrial, autosomal dominant | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 0.5 |
| **CHCHD10** | Spinal muscular atrophy, Jokela type | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 0.5 |
| **CHD2** | Epileptic encephalopathy, childhood-onset | Sequencing of all coding exons of the gene |  |  | 5.5 |
| **CHD7** | CHARGE Syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 9 |
| **CHD7** | Hypogonadotropic hypogonadism | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 9 |
| **CHD7** | Hypogonadotropic hypogonadism 5 with or without anosmia | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 9 |
| **CHKB** | Congenital Muscular Dystrophy | Sequencing of all coding exons of the gene |  |  | 1.2 |
| **CHM** | Choroidal Sclerosis | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 2 |
| **CHMP2B** | Frontotemporal Dementia | Sequencing of all coding exons of the gene |  |  | 0.6 |
| **CHN1** | Duane Syndrome | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **CHRNA1** | Multiple pterygium syndrome, lethal type | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **CHRNA1** | Myasthenic Syndrome | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **CHRNA2** | Nocturnal frontal lobe Epilepsy | Sequencing of all coding exons of the gene |  |  | 1.6 |
| **CHRNA4** | Nocturnal frontal lobe Epilepsy | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.9 |
| **CHRNB1** | Congenital Myasthenic Syndromes | Sequencing of all coding exons of the gene |  |  | 1.5 |
| **CHRNB2** | Nocturnal frontal lobe Epilepsy | Sequencing of all coding exons of the gene |  |  | 1.5 |
| **CHRNB3** | Cardiac Diseases | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **CHRND** | Multiple pterygium syndrome, lethal type | Sequencing of all coding exons of the gene |  |  | 1.6 |
| **CHRND** | Myasthenic Syndrome | Sequencing of all coding exons of the gene |  |  | 1.6 |
| **CIZ1** | Dystonia 23 | Sequencing of all coding exons of the gene |  |  | 2.7 |
| **CLCN1** | Myotonia Congenita | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 3 |
| **CLCN5** | Dent disease | Sequencing of all coding exons of the gene |  |  | 2.5 |
| **CLCN5** | Hypophosphatemic rickets | Sequencing of all coding exons of the gene |  |  | 2.5 |
| **CLCN5** | Nephrolithiasis, type I | Sequencing of all coding exons of the gene |  |  | 2.5 |
| **CLCN5** | Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis | Sequencing of all coding exons of the gene |  |  | 2.5 |
| **CLN3** | Neuronal Ceroid-Lipofuscinosis | Sequencing of all coding exons of the gene |  |  | 1.3 |
| **CLN3** | Neuronal Ceroid-Lipofuscinosis | Sequencing of all coding exons of the gene |  |  | 1.3 |
| **CLN3** | Neuronal Ceroid-Lipofuscinosis | Sequencing of all coding exons of the gene |  |  | 1.3 |
| **CLN5** | Neuronal Ceroid-Lipofuscinoses | Sequencing of all coding exons of the gene |  |  | 1.2 |
| **CLN6** | Neuronal Ceroid-Lipofuscinoses | Sequencing of all coding exons of the gene |  |  | 0.9 |
| **CLN8** | Neuronal Ceroid-Lipofuscinoses | Sequencing of all coding exons of the gene |  |  | 0.9 |
| **CLPB** | 3-METHYLGLUTACONIC ACIDURIA WITH CATARACTS, NEUROLOGIC INVOLVEMENT, AND NEUTROPENIA; MEGCANN | Sequencing of all coding exons of the gene |  |  | 2.1 |
| **CLRN1** | Usher Syndrome Type 3 | Sequencing of all coding exons of the gene |  |  | 0.7 |
| **CNGA3** | Achromatopsia | Sequencing of all coding exons of the gene |  |  | 2.1 |
| **CNGB1** | Retinitis Pigmentosa | Sequencing of all coding exons of the gene |  |  | 3.8 |
| **CNGB3** | Achromatopsia | Sequencing of hotspots | Sequencing of all coding exons of the gene | Deletion and duplication analysis | 2.4 |
| **CNGB3** | Stargardt Disease | Sequencing of hotspots | Sequencing of all coding exons of the gene | Deletion and duplication analysis | 2.4 |
| **CNTNAP2** | Pitt-Hopkins-Like Syndrome 1 | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 4 |
| **COASY** | Neurodegeneration with brain iron accumulation (NBIA) | Sequencing of all coding exons of the gene |  |  | 1.8 |
| **COASY** | Neurodegeneration with brain iron accumulation 6 | Sequencing of all coding exons of the gene |  |  | 1.8 |
| **COG6** | Congenital disorder of glycosylation, type IIl | Sequencing of all coding exons of the gene |  |  | 2 |
| **COG7** | Congenital disorder of glycosylation, type IIe | Sequencing of all coding exons of the gene |  |  | 2.3 |
| **COL10A1** | Metaphyseal Chondrodysplasia, Schmid Type | Sequencing of all coding exons of the gene |  |  | 2 |
| **COL18A1** | Knobloch Syndrome Type I | Sequencing of all coding exons of the gene |  |  | 4.6 |
| **COL2A1** | Kniest Dysplasia | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 4.5 |
| **COL2A1** | Stickler Syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 4.5 |
| **COL4A1** | Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps | Sequencing of all coding exons of the gene |  |  | 5 |
| **COL4A1** | Brain small vessel disease with or without ocular anomalies | Sequencing of all coding exons of the gene |  |  | 5 |
| **COL4A1** | Porencephaly 1 | Sequencing of all coding exons of the gene |  |  | 5 |
| **COL4A1** | Retinal arteries, tortuosity of | Sequencing of all coding exons of the gene |  |  | 5 |
| **COL4A3** | Alport syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 5 |
| **COL4A3** | Hematuria, benign familial | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 5 |
| **COL6A3** | Bethlem myopathy 1 | Sequencing of all coding exons of the gene |  |  | 9.5 |
| **COL6A3** | Dystonia 27 | Sequencing of all coding exons of the gene |  |  | 9.5 |
| **COL6A3** | Ullrich congenital muscular dystrophy 1 | Sequencing of all coding exons of the gene |  |  | 9.5 |
| **COL9A1** | Multiple epiphyseal Dysplasia | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.8 |
| **COL9A1** | Stickler Syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.8 |
| **COL9A2** | Multiple epiphyseal Dysplasia | Sequencing of all coding exons of the gene |  |  | 2.1 |
| **COL9A3** | Multiple epiphyseal Dysplasia | Sequencing of all coding exons of the gene |  |  | 2.1 |
| **COLEC11** | 3MC syndrome 2 | Sequencing of all coding exons of the gene |  |  | 0.9 |
| **COMP** | Multiple epiphyseal Dysplasia | Sequencing of all coding exons of the gene |  |  | 2.3 |
| **COMP** | Pseudoachondroplasia | Sequencing of all coding exons of the gene |  |  | 2.3 |
| **COPA** | Autoimmune Interstitial Lung, Joint and Kidney Disease | Sequencing of all coding exons of the gene |  |  | 3.7 |
| **COQ8A** | Coenzyme Q10 Deficiency | Sequencing of all coding exons of the gene |  |  | 1.9 |
| **COQ9** | Coenzyme Q10 Deficiency | Sequencing of all coding exons of the gene |  |  | 1 |
| **CP** | Aceruloplasminemia | Sequencing of all coding exons of the gene |  |  | 3.2 |
| **CPA1** | Hereditary pancreatitis (Carboxypeptidase A1; CPA1 gene) | Sequencing of all coding exons of the gene |  |  | 1.3 |
| **CPT2** | Carnitine Palmitoyltransferase II Deficiency | Sequencing of all coding exons of the gene |  |  | 2 |
| **CRB1** | Leber Congenital Amaurosis 8 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 4.2 |
| **CRB1** | Retinitis Pigmentosa | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 4.2 |
| **CRX** | Leber Congenital Amaurosis 7 | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 0.9 |
| **CRX** | Retinitis Pigmentosa | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 0.9 |
| **CSF1R** | Leukoencephalopathy with spheroids | Sequencing of hotspots | Sequencing of all coding exons of the gene |  | 2.9 |
| **CSF2RA** | Pulmonary surfactant metabolism dysfunction type 4 (mutation in the GMCSF receptor α chain; CSF2RA gene) | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 2.6 |
| **CSTB** | Progressive Myoclonus Epilepsy with Ataxia | Repeat expansion analysis | Sequencing of all coding exons of the gene | Deletion and duplication analysis | 0.3 |
| **CSTB** | Progressive Myoclonus Epilepsy with Ataxia | Repeat expansion analysis | Sequencing of all coding exons of the gene | Deletion and duplication analysis | 0.3 |
| **CTRC** | Pancreatitis, chronic, susceptibility to | Sequencing of all coding exons of the gene |  |  | 0.8 |
| **CTSD** | Neuronal Ceroid-Llipofuscinosis | Sequencing of all coding exons of the gene |  |  | 1.2 |
| **CTSF** | Neuronal Ceroid-Lipofuscinosis | Sequencing of all coding exons of the gene |  |  | 1.5 |
| **CTSF** | Neuronal Ceroid-Lipofuscinosis | Sequencing of all coding exons of the gene |  |  | 1.5 |
| **CTSF** | Neuronal Ceroid-Lipofuscinosis | Sequencing of all coding exons of the gene |  |  | 1.5 |
| **CYP19A1** | Aromatase deficiency | Sequencing of all coding exons of the gene |  |  | 1.5 |
| **CYP19A1** | Aromatase excess syndrome | Sequencing of all coding exons of the gene |  |  | 1.5 |
| **CYP27A1** | Cerebrotendinous Xanthomatosis | Sequencing of all coding exons of the gene |  |  | 1.6 |
| **DARS** | Hypomyelination with brainstem and spinal cord involvement and leg spasticity | Sequencing of all coding exons of the gene |  |  | 1.5 |
| **DARS2** | Leukoenzephalopathie mit Hirnstamm- und Rückenmarkbeteiligung | Sequencing of all coding exons of the gene |  |  | 1.9 |
| **DBT** | Maple Syrup Urine Disease | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **DCC** | Mirror movements 1 | Sequencing of all coding exons of the gene |  |  | 4.3 |
| **DCTN1** | Perry Syndrome | Sequencing of all coding exons of the gene |  |  | 3.8 |
| **DCX** | DCX-Related Disorders | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.3 |
| **DES** | Cardiomyopathy, dilated, 1I | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **DES** | Muscular dystrophy, limb-girdle, type 2R | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **DES** | Myopathy, myofibrillar, 1 | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **DES** | Scapuloperoneal syndrome, neurogenic, Kaeser type | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **DFNB31** | DFNB31 Nonsyndromic Hearing Loss and Deafness | Sequencing of all coding exons of the gene |  |  | 2.7 |
| **DGKE** | Nephrotic Syndrome Type 7 | Sequencing of all coding exons of the gene |  |  | 1.7 |
| **DHTKD1** | 2-alpha Aminoadipic 2-Oxoadipic Aciduria | Sequencing of all coding exons of the gene |  |  | 2.8 |
| **DLD** | Dihydrolipoamide Dehydrogenase (E3) Deficiency | Sequencing of all coding exons of the gene |  |  | 1.5 |
| **DLD** | Leigh Syndrome | Sequencing of all coding exons of the gene |  |  | 1.5 |
| **DLD** | Maple Syrup Urine Disease | Sequencing of all coding exons of the gene |  |  | 1.5 |
| **DMD** | Becker muscular dystrophy | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 11.1 |
| **DMD** | Dilated Cardiomyopathy | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 11.1 |
| **DMD** | Dilated Cardiomyopathy | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 11.1 |
| **DMD** | Duchenne muscular dystrophy | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 11.1 |
| **DMP1** | Hypophosphatemic rickets, AR | Sequencing of all coding exons of the gene |  |  | 1.5 |
| **DNAI1** | Ciliary dyskinesia, primary, 1, with or without situs inversus | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.1 |
| **DNAJC5** | Neuronal Ceroid-Lipofuscinoses | Sequencing of all coding exons of the gene |  |  | 0.6 |
| **DOK7** | Myasthenic Syndrome | Sequencing of all coding exons of the gene |  |  | 1.5 |
| **DPAGT1** | Congenital disorder of glycosylation, type Ij | Sequencing of all coding exons of the gene |  |  | 1.2 |
| **DRD2** | Myclonus Dystonia | Sequencing of all coding exons of the gene |  |  | 1.3 |
| **DUOX2** | Thryoid dyshormonogenesis 6 | Sequencing of all coding exons of the gene |  |  | 4.6 |
| **DUOXA2** | Thyroid dyshormonogenesis 5 | Sequencing of all coding exons of the gene |  |  | 1 |
| **DYSF** | Muscular dystrophy, limb-girdle, type 2B | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 6.4 |
| **DYX1C1** | Ciliary dyskinesia, primary, 25 | Sequencing of all coding exons of the gene |  |  | 1.3 |
| **ECHS1** | Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency | Sequencing of all coding exons of the gene |  |  | 0.9 |
| **EFEMP1** | Doyne Honeycomb Retinal Dystrophy | Sequencing of all coding exons of the gene |  |  | 1.5 |
| **EFHC1** | Juvenile Myoclonus Epilepsy (JME) | Sequencing of all coding exons of the gene |  |  | 1.9 |
| **EFHC1** | Myoclonic epilepsy, juvenile, susceptibility to, 1 | Sequencing of all coding exons of the gene |  |  | 1.9 |
| **EFNB1** | Craniofrontonasal Syndrome | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 1 |
| **EGF** | Hypomagnesemia 4, renal | Sequencing of all coding exons of the gene |  |  | 3.6 |
| **EGR2** | Charcot-Marie-Tooth Neuropathy Type 1D | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **EGR2** | Charcot-Marie-Tooth Neuropathy Type 4E | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **ELANE** | Neutropenia, cyclic | Sequencing of hotspots | Sequencing of all coding exons of the gene |  | 0.8 |
| **ELANE** | Neutropenia, severe congenital 1, autosomal dominant | Sequencing of hotspots | Sequencing of all coding exons of the gene |  | 0.8 |
| **ELOVL4** | Stargardt Disease | Sequencing of all coding exons of the gene |  |  | 0.9 |
| **EMX2** | Schizencephaly | Sequencing of all coding exons of the gene |  |  | 0.8 |
| **EMX2** | Schizencephaly | Sequencing of all coding exons of the gene |  |  | 0.8 |
| **ENG** | Telangiectasia, hereditary hemorrhagic, type 1 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2 |
| **EOMES** | Microcephaly Syndrome | Sequencing of all coding exons of the gene |  |  | 2.1 |
| **EP300** | Rubinstein-Taybi syndrome 2 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 7.2 |
| **EPM2A** | Lafora Disease | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 1 |
| **ERCC1** | Xeroderma Pigmentosum | Sequencing of all coding exons of the gene |  |  | 1 |
| **ERCC2** | Xeroderma Pigmentosum | Sequencing of all coding exons of the gene |  |  | 2.3 |
| **ERCC4** | Xeroderma Pigmentosum | Sequencing of all coding exons of the gene |  |  | 2.8 |
| **ERCC5** | Xeroderma Pigmentosum | Sequencing of all coding exons of the gene |  |  | 3.6 |
| **ERCC6** | Age-Dependent Macula Degeneration | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 4.5 |
| **ERCC6** | Cockayne Syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 4.5 |
| **ERCC8** | Cockayne Syndrome | Sequencing of all coding exons of the gene |  |  | 1.2 |
| **ETFA** | Multiple Acyl-CoA Dehydrogenase Deficiency | Sequencing of all coding exons of the gene |  |  | 1 |
| **ETFA** | Multiple Acyl-CoA Dehydrogenase Deficiency | Sequencing of all coding exons of the gene |  |  | 1 |
| **ETFB** | Multiple Acyl-CoA Dehydrogenase Deficiency | Sequencing of all coding exons of the gene |  |  | 1 |
| **ETFB** | Multiple Acyl-CoA Dehydrogenase Deficiency | Sequencing of all coding exons of the gene |  |  | 1 |
| **ETFDH** | Multiple Acyl-CoA Dehydrogenase Deficiency | Sequencing of all coding exons of the gene |  |  | 1.9 |
| **ETFDH** | Multiple Acyl-CoA Dehydrogenase Deficiency | Sequencing of all coding exons of the gene |  |  | 1.9 |
| **ETHE1** | Ethylmalonic encephalopathy | Sequencing of all coding exons of the gene |  |  | 0.8 |
| **EXOSC3** | Pontocerebellar Hypoplasia, Type 1B | Sequencing of all coding exons of the gene |  |  | 0.8 |
| **EXT1** | Multiple Exostoses | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.2 |
| **EXT2** | Multiple Exostoses | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.3 |
| **F12** | Angioedema, hereditary, type III | Sequencing of all coding exons of the gene |  |  | 1.8 |
| **F12** | Factor XII deficiency | Sequencing of all coding exons of the gene |  |  | 1.8 |
| **F12** | Hereditary angioedema type III (mutation in exon 9 of the F12 gene) | Sequencing of all coding exons of the gene |  |  | 1.8 |
| **F2** | Prothrombin mutation | Sequencing of hotspots | Sequencing of all coding exons of the gene |  | 1.9 |
| **F5** | Factor V Leiden mutation / APC resistance | Sequencing of hotspots | Sequencing of all coding exons of the gene |  | 6.7 |
| **FA2H** | Fatty Acid Hydroxylase-Associated Neurodegeneration | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.1 |
| **FA2H** | Spastic Paraplegia 35 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.1 |
| **FAH** | Tyrosinemia | Sequencing of all coding exons of the gene |  |  | 1.3 |
| **FAM58A** | STAR syndrome | Sequencing of all coding exons of the gene |  |  | 0.7 |
| **FANCC** | FANCONI ANEMIA, COMPLEMENTATION GROUP C; FANCC | Sequencing of all coding exons of the gene |  |  | 1.7 |
| **FAS** | Autoimmune lymphoproliferative syndrome, type IA | Sequencing of all coding exons of the gene |  |  | 1 |
| **FAS** | Squamous cell carcinoma, burn scar-related, somatic | Sequencing of all coding exons of the gene |  |  | 1 |
| **FGA** | Hereditary amyloidosis (FGA and TTR gene) | Sequencing of hotspots |  |  | 2.6 |
| **FGD1** | Aarskog-Scott syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.9 |
| **FGD1** | Mental retardation, X-linked syndromic 16 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.9 |
| **FGF14** | Spinocerebellar ataxia 27 | Sequencing of all coding exons of the gene |  |  | 0.8 |
| **FGF23** | Hypophosphatemic rickets, autosomal dominant | Sequencing of all coding exons of the gene |  |  | 0.8 |
| **FGF23** | Osteomalacia, tumor-induced | Sequencing of all coding exons of the gene |  |  | 0.8 |
| **FGF23** | Tumoral calcinosis, hyperphosphatemic, familial | Sequencing of all coding exons of the gene |  |  | 0.8 |
| **FGF8** | Holoprosencephaly | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 0.7 |
| **FGF8** | Hypogonadotropic hypogonadism | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 0.7 |
| **FGFR2** | Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis | Sequencing of all coding exons of the gene |  |  | 2.5 |
| **FGFR2** | Apert syndrome | Sequencing of all coding exons of the gene |  |  | 2.5 |
| **FGFR2** | Beare-Stevenson cutis gyrata syndrome | Sequencing of all coding exons of the gene |  |  | 2.5 |
| **FGFR2** | Bent bone dysplasia syndrome | Sequencing of all coding exons of the gene |  |  | 2.5 |
| **FGFR2** | Craniofacial-skeletal-dermatologic dysplasia | Sequencing of all coding exons of the gene |  |  | 2.5 |
| **FGFR2** | Craniosynostosis, nonspecific | Sequencing of all coding exons of the gene |  |  | 2.5 |
| **FGFR2** | Crouzon Syndrome | Sequencing of all coding exons of the gene |  |  | 2.5 |
| **FGFR2** | Jackson-Weiss syndrome | Sequencing of all coding exons of the gene |  |  | 2.5 |
| **FGFR2** | LADD syndrome | Sequencing of all coding exons of the gene |  |  | 2.5 |
| **FGFR2** | Pfeiffer syndrome | Sequencing of all coding exons of the gene |  |  | 2.5 |
| **FGFR2** | Saethre-Chotzen Syndrome | Sequencing of all coding exons of the gene |  |  | 2.5 |
| **FGFR2** | Scaphocephaly and Axenfeld-Rieger anomaly | Sequencing of all coding exons of the gene |  |  | 2.5 |
| **FGFR2** | Scaphocephaly, maxillary retrusion, and mental retardation | Sequencing of all coding exons of the gene |  |  | 2.5 |
| **FGFR3** | Achondroplasia | Sequencing of hotspots | Sequencing of all coding exons of the gene |  | 2.4 |
| **FGFR3** | CAMPTODACTYLY, TALL STATURE, AND HEARING LOSS SYNDROME; CATSHLS | Sequencing of hotspots | Sequencing of all coding exons of the gene |  | 2.4 |
| **FGFR3** | Crouzon Syndrome | Sequencing of hotspots | Sequencing of all coding exons of the gene |  | 2.4 |
| **FGFR3** | Crouzon Syndrome with Acanthosis Nigricans | Sequencing of hotspots | Sequencing of all coding exons of the gene |  | 2.4 |
| **FGFR3** | Hypochondroplasia | Sequencing of hotspots | Sequencing of all coding exons of the gene |  | 2.4 |
| **FGFR3** | Lacrimo-Auriculo-Dento-Digital Syndrome | Sequencing of hotspots | Sequencing of all coding exons of the gene |  | 2.4 |
| **FGFR3** | Muenke Syndrome | Sequencing of hotspots | Sequencing of all coding exons of the gene |  | 2.4 |
| **FGFR3** | Saddan Syndrome | Sequencing of hotspots | Sequencing of all coding exons of the gene |  | 2.4 |
| **FGFR3** | THANATOPHORIC DYSPLASIA, TYPE I; TD1 | Sequencing of hotspots | Sequencing of all coding exons of the gene |  | 2.4 |
| **FGFR3** | THANATOPHORIC DYSPLASIA, TYPE II; TD2 | Sequencing of hotspots | Sequencing of all coding exons of the gene |  | 2.4 |
| **FLNA** | Periventricular nodular Heterotopia | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 7.9 |
| **FLT4** | Hemangioma, capillary infantile, somatic | Sequencing of all coding exons of the gene |  |  | 4.1 |
| **FLT4** | Hereditary lymphedema type IA (mutation in exons 17-25 of the FTL4 gene) | Sequencing of all coding exons of the gene |  |  | 4.1 |
| **FLT4** | Lymphedema, hereditary, IA | Sequencing of all coding exons of the gene |  |  | 4.1 |
| **FLVCR1** | Posterior Column Ataxia with Retinitis Pigmentosa | Sequencing of all coding exons of the gene |  |  | 1.7 |
| **FLVCR2** | Fowler Syndrome | Sequencing of all coding exons of the gene |  |  | 1.6 |
| **FOLR1** | Neurodegeneration due to Cerebral Folate Transport Deficiency | Sequencing of all coding exons of the gene |  |  | 0.8 |
| **FOXC1** | Axenfeld-Rieger Syndrome | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 1.7 |
| **FOXC1** | Peters Anomaly | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 1.7 |
| **FOXE1** | Bamforth-Lazarus Syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.1 |
| **FOXG1** | Rett Syndrome, Congenital Variant | Sequencing of all coding exons of the gene |  |  | 1.5 |
| **FRAS1** | FRAS1-Related Fraser Syndrome | Sequencing of all coding exons of the gene |  |  | 12 |
| **FREM1** | BIFID NOSE WITH OR WITHOUT ANORECTAL AND RENAL ANOMALIES; BNAR | Sequencing of all coding exons of the gene |  |  | 6.5 |
| **FREM1** | MANITOBA OCULOTRICHOANAL SYNDROME; MOTA | Sequencing of all coding exons of the gene |  |  | 6.5 |
| **FREM1** | TRIGONOCEPHALY 2; TRIGNO2 | Sequencing of all coding exons of the gene |  |  | 6.5 |
| **FSCN2** | Retinitis Pigmentosa | Sequencing of all coding exons of the gene |  |  | 1.6 |
| **FSHB** | Hypogonadotropic hypogonadism 24 without anosmia | Sequencing of all coding exons of the gene |  |  | 0.4 |
| **FTL** | Hyperferritinemia-cataract syndrome (mutation in the IRE of the FTL gene) | Sequencing of all coding exons of the gene |  |  | 0.5 |
| **FTL** | Neurodegeneration with brain iron accumulation (NBIA) | Sequencing of all coding exons of the gene |  |  | 0.5 |
| **FUS** | Cardiac Diseases | Sequencing of all coding exons of the gene |  |  | 1.6 |
| **FZD4** | Familial Exudative Vitreoretinopathy | Sequencing of all coding exons of the gene |  |  | 1.6 |
| **G6PC** | Glycogen Storage Disease Type Ia | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **G6PC3** | Dursun syndrome | Sequencing of all coding exons of the gene |  |  | 1 |
| **G6PC3** | Dursun syndrome | Sequencing of all coding exons of the gene |  |  | 1 |
| **G6PC3** | Neutropenia, severe congenital 4, autosomal recessive | Sequencing of all coding exons of the gene |  |  | 1 |
| **GAA** | Glycogen storage disease II | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 2.9 |
| **GABRA1** | Juvenile Myoclonus Epilepsy (JME) | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **GABRB3** | Childhood absence Epilepsy | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.4 |
| **GABRD** | Epilepsy, generalized, with febrile seizures plus, type 5, susceptibility to | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **GABRD** | Generalised Epilepsy with febrile seizures plus (GEFS+) | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **GABRD** | Juvenile Myoclonus Epilepsy (JME) | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **GABRG2** | Childhood absence Epilepsy | Sequencing of all coding exons of the gene |  |  | 1.5 |
| **GALC** | Krabbe Disease | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.1 |
| **GALNT3** | Tumoral calcinosis | Sequencing of all coding exons of the gene |  |  | 1.9 |
| **GAMT** | Guanidinoacetate Methyltransferase Deficiency | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 0.8 |
| **GARS** | Charcot-Marie-Tooth disease, type 2D | Sequencing of all coding exons of the gene |  |  | 2.2 |
| **GARS** | Neuropathy, distal hereditary motor, type V | Sequencing of all coding exons of the gene |  |  | 2.2 |
| **GATA1** | GATA1-Related Anemia with Thrombocytopenia | Sequencing of all coding exons of the gene |  |  | 1.2 |
| **GATA2** | Emberger syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.4 |
| **GATA2** | Immunodeficiency 21 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.4 |
| **GATA6** | Atrial septal defect 9 | Sequencing of all coding exons of the gene |  |  | 1.8 |
| **GATA6** | Atrioventricular septal defect 5 | Sequencing of all coding exons of the gene |  |  | 1.8 |
| **GATA6** | Pancreatic agenesis and congenital heart defects | Sequencing of all coding exons of the gene |  |  | 1.8 |
| **GATA6** | Persistent truncus arteriosus | Sequencing of all coding exons of the gene |  |  | 1.8 |
| **GATA6** | Tetralogy of Fallot | Sequencing of all coding exons of the gene |  |  | 1.8 |
| **GBA** | Parkinson Syndrome | Sequencing of all coding exons of the gene |  |  | 1.6 |
| **GBE1** | Glycogen Storage Disease Type IV | Sequencing of all coding exons of the gene |  |  | 2.1 |
| **GCDH** | Glutaricacidemia Type 1 | Sequencing of all coding exons of the gene |  |  | 1.3 |
| **GCH1** | Dopa-responsive Dystonia DYT5 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 0.8 |
| **GCK** | Diabetes mellitus, gestational | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.4 |
| **GCK** | Diabetes mellitus, noninsulin-dependent, late onset | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.4 |
| **GCK** | Diabetes mellitus, permanent neonatal | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.4 |
| **GCK** | Hyperinsulinemic hypoglycemia, familial, 3 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.4 |
| **GCK** | MODY, type II | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.4 |
| **GCNT2** | Cataracts, autosomal recessive | Sequencing of all coding exons of the gene |  |  | 1.2 |
| **GCSH** | Glycine Encephalopathy | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 0.5 |
| **GDAP1** | Charcot-Marie-Tooth Neuropathy Type 2H | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **GDAP1** | Charcot-Marie-Tooth Neuropathy Type 2K | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **GDAP1** | Charcot-Marie-Tooth Neuropathy Type 4A | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **GFAP** | Alexander Syndrome | Sequencing of all coding exons of the gene |  |  | 1.3 |
| **GFER** | Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay | Sequencing of all coding exons of the gene |  |  | 0.6 |
| **GJA1** | Hypoplastic Left Heart Syndrome | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **GJA1** | Oculodentodigital Dysplasia | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **GJA1** | Syndactyly, Type III | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **GJB1** | Charcot-Marie-Tooth X-linked | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 0.9 |
| **GJB2** | DFNA 3 Nonsyndromic Hearing Loss and Deafness | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 0.7 |
| **GJB2** | DFNA 3 Nonsyndromic Hearing Loss and Deafness | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 0.7 |
| **GJB2** | DFNB 1 Nonsyndromic Hearing Loss and Deafness | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 0.7 |
| **GJB2** | Vohwinkel Syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 0.7 |
| **GJB3** | GJB3-Related Erythrokeratodermia Variabilis | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 0.8 |
| **GJB6** | DFNA 3 Nonsyndromic Hearing Loss and Deafness | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 0.8 |
| **GJB6** | DFNA 3 Nonsyndromic Hearing Loss and Deafness | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 0.8 |
| **GJC2** | Pelizaeus-Merzbacher-Like Disease 1 | Sequencing of all coding exons of the gene |  |  | 1.3 |
| **GJC2** | Spastic Paraplegia 44 | Sequencing of all coding exons of the gene |  |  | 1.3 |
| **GLA** | Fabry Disease | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.3 |
| **GLB1** | GM1-Gangliosidosis Type 1 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2 |
| **GLB1** | Mucopolysaccharidosis Type 4B | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2 |
| **GLDC** | Glycine Encephalopathy | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 3.1 |
| **GLE1** | Lethal Arthrogryposis with anterior horn cell disease (LAAHD) | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.1 |
| **GLE1** | Lethal Congenital Contracture Syndrome 1 (LCCS1) | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.1 |
| **GLI2** | Culler-Jones syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 4.8 |
| **GLI2** | Holoprosencephaly-9 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 4.8 |
| **GLI3** | Greig Syndrome | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 4.7 |
| **GLRA1** | Hyperekplexia | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.4 |
| **GLRB** | Hyperekplexia | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.5 |
| **GLUL** | Glutamine deficiency, congenital | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **GNA11** | Hypocalcemia, autosomal dominant 2 | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **GNA11** | Hypocalciuric hypercalcemia, type II | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **GNAQ** | Capillary malformations, congenital, 1, somatic, mosaic | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **GNAQ** | Sturge-Weber syndrome, somatic, mosaic | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **GNAS** | Acromegaly, somatic | Sequencing of all coding exons of the gene |  |  | 3.1 |
| **GNAS** | ACTH-independent macronodular adrenal hyperplasia | Sequencing of all coding exons of the gene |  |  | 3.1 |
| **GNAS** | McCune-Albright syndrome, somatic, mosaic | Sequencing of all coding exons of the gene |  |  | 3.1 |
| **GNAS** | Osseous heteroplasia, progressive | Sequencing of all coding exons of the gene |  |  | 3.1 |
| **GNAS** | Pseudohypoparathyroidism Ia | Sequencing of all coding exons of the gene |  |  | 3.1 |
| **GNAS** | Pseudohypoparathyroidism Ib | Sequencing of all coding exons of the gene |  |  | 3.1 |
| **GNAS** | Pseudohypoparathyroidism Ic | Sequencing of all coding exons of the gene |  |  | 3.1 |
| **GNAS** | Pseudopseudohypoparathyroidism | Sequencing of all coding exons of the gene |  |  | 3.1 |
| **GNAT2** | Achromatopsia | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **GNPTAB** | Mucolipidosis | Sequencing of all coding exons of the gene |  |  | 3.8 |
| **GNPTG** | Mucolipidosis | Sequencing of all coding exons of the gene |  |  | 0.9 |
| **GNRHR** | Hypogonadotropic hypogonadism | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1 |
| **GNS** | Mucopolysaccharidosis type IIID | Sequencing of all coding exons of the gene |  |  | 1.7 |
| **GOSR2** | Epilepsy, progressive myoclonic 6 | Sequencing of all coding exons of the gene |  |  | 0.6 |
| **GPHN** | Hyperekplexia | Sequencing of all coding exons of the gene |  |  | 2.3 |
| **GPHN** | Molybdenum cofactor deficiency C | Sequencing of all coding exons of the gene |  |  | 2.3 |
| **GPR143** | Ocular Albinism  x-linked | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.2 |
| **GPSM2** | Chudley-McCullough syndrome | Sequencing of all coding exons of the gene |  |  | 2.1 |
| **GRIN2A** | Epilepsy with neurodevelopmental defects | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 4.4 |
| **GRN** | Frontotemporal Dementia | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.8 |
| **GSN** | Amyloidosis, Finnish type | Sequencing of all coding exons of the gene |  |  | 2.3 |
| **GSS** | Glutathione synthetase deficiency | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **GUCA1A** | Cone-Rod-Dystrophy | Sequencing of all coding exons of the gene |  |  | 0.6 |
| **GUCY2D** | Leber Congenital Amaurosis | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 3.3 |
| **HAX1** | Neutropenia, severe congenital 3, autosomal recessive | Sequencing of all coding exons of the gene |  |  | 0.8 |
| **HCN4** | Sick Sinus Syndrome | Sequencing of all coding exons of the gene |  |  | 3.6 |
| **HCRT** | Narcolepsy 1 | Sequencing of all coding exons of the gene |  |  | 0.4 |
| **HDAC8** | Cornelia de Lange Syndrome | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **HEATR2** | Ciliary dyskinesia, primary, 18 | Sequencing of all coding exons of the gene |  |  | 2.6 |
| **HESX1** | Septooptic dysplasia | Sequencing of all coding exons of the gene |  |  | 0.6 |
| **HEXA** | Hexosaminidase A Deficiency | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.6 |
| **HEXA** | Tay-Sachs Disease | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.6 |
| **HFE** | Hemochromatosis type 1 (C282Y and H63D mutation; complete HFE gene analysis on request) | Sequencing of hotspots | Sequencing of all coding exons of the gene | Deletion and duplication analysis | 1 |
| **HFE** | Hereditary Hemochromatosis | Sequencing of hotspots | Sequencing of all coding exons of the gene | Deletion and duplication analysis | 1 |
| **HFE2** | Hemochromatosis | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.3 |
| **HGSNAT** | Mucopolysaccharidosis type IIIC (Sanfilippo C) | Sequencing of all coding exons of the gene |  |  | 1.9 |
| **HGSNAT** | Retinitis pigmentosa 73 | Sequencing of all coding exons of the gene |  |  | 1.9 |
| **HINT1** | Neuromyotonia and axonal neuropathy, autosomal recessive | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 0.4 |
| **HNF1A** | Maturity-Onset Diabetes of the Young Type 3 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.9 |
| **HNF4A** | Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.4 |
| **HNF4A** | MODY, type I | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.4 |
| **HOGA1** | Hyperoxaluria, Primary, Type 3 | Sequencing of all coding exons of the gene |  |  | 1 |
| **HOXD13** | Brachydactyly, type D | Sequencing of all coding exons of the gene |  |  | 1 |
| **HOXD13** | Brachydactyly, type E | Sequencing of all coding exons of the gene |  |  | 1 |
| **HOXD13** | Brachydactyly-syndactyly syndrome | Sequencing of all coding exons of the gene |  |  | 1 |
| **HOXD13** | Syndactyly, type V | Sequencing of all coding exons of the gene |  |  | 1 |
| **HOXD13** | Synpolydactyly with foot anomalies | Sequencing of all coding exons of the gene |  |  | 1 |
| **HOXD13** | Synpolydactyly, type II | Sequencing of all coding exons of the gene |  |  | 1 |
| **HRAS** | Costello Syndrome | Sequencing of all coding exons of the gene |  |  | 0.6 |
| **HSD11B1** | Cortisone reductase deficiency 2 | Sequencing of all coding exons of the gene |  |  | 0.9 |
| **HSD17B10** | 17-beta-hydroxysteroid dehydrogenase X deficiency | Sequencing of all coding exons of the gene |  |  | 0.8 |
| **HSD17B10** | Mental retardation, X-linked syndromic 10 | Sequencing of all coding exons of the gene |  |  | 0.8 |
| **HSD17B4** | Peroxisomal Bifunctional Enzyme Deficiency | Sequencing of all coding exons of the gene |  |  | 2.3 |
| **HSD17B4** | Perrault syndrome 1 | Sequencing of all coding exons of the gene |  |  | 2.3 |
| **HSD3B7** | Bile acid synthesis defect, congenital, 1 | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **HSF4** | Cataracts, autosomal recessive | Sequencing of all coding exons of the gene |  |  | 1.5 |
| **HTRA1** | CARASIL syndrome | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **HTT** | Huntington Disease, HD | Fragment length analysis |  |  | 9.4 |
| **IDS** | Mucopolysaccharidosis Type II | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.7 |
| **IL1RAPL1** | Mental retardation, X-linked 21/34 | Sequencing of all coding exons of the gene |  |  | 2.1 |
| **IL36RN** | Psoriasis 14, pustular | Sequencing of all coding exons of the gene |  |  | 0.5 |
| **IRF6** | Van der Woude-Syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.4 |
| **ISCU** | Myopathy with lactic acidosis (hereditary) | Sequencing of all coding exons of the gene |  |  | 0.5 |
| **ISPD** | Congenital Muscular Dystrophy | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.4 |
| **IYD** | Thyroid dyshormonogenesis 4 | Sequencing of all coding exons of the gene |  |  | 0.9 |
| **JAG1** | Alagille syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 3.7 |
| **JAG1** | Tetralogy of Fallot | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 3.7 |
| **JAGN1** | Neutropenia, servere congenital, 6, autosomal recessive | Sequencing of all coding exons of the gene |  |  | 0.6 |
| **JAK2** | JAK2-Related Budd-Chiari Syndrome | Sequencing of all coding exons of the gene |  |  | 3.4 |
| **JPH3** | Huntington disease-like 2 (HDL2) | Fragment length analysis |  |  | 2.2 |
| **KCNA1** | Episodic Ataxia - Type 1 | Sequencing of all coding exons of the gene |  |  | 1.5 |
| **KCNC3** | Spinocerebellar Ataxia Type13 | Sequencing of all coding exons of the gene |  |  | 2.3 |
| **KCND3** | Brugada syndrome 9 | Sequencing of all coding exons of the gene |  |  | 2 |
| **KCND3** | Spinocerebellar ataxia 19 | Sequencing of all coding exons of the gene |  |  | 2 |
| **KCNE1** | Jervell and Lange-Nielsen Syndrome (Long-QT-Syndrome) | Sequencing of all coding exons of the gene |  |  | 0.4 |
| **KCNE1** | Romano-Ward Syndrome (Long-QT-Syndrome) | Sequencing of all coding exons of the gene |  |  | 0.4 |
| **KCNE3** | Brugada syndrome 6 | Sequencing of all coding exons of the gene |  |  | 0.3 |
| **KCNJ10** | Enlarged vestibular aqueduct, digenic | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **KCNJ10** | SESAME syndrome | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **KCNJ2** | Andersen syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.3 |
| **KCNJ2** | Atrial fibrillation, familial, 9 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.3 |
| **KCNJ2** | Atrial fibrillation, familial, 9 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.3 |
| **KCNJ2** | Short QT syndrome 3 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.3 |
| **KCNJ5** | Hyperaldosteronism, familial, type III | Sequencing of all coding exons of the gene |  |  | 1.3 |
| **KCNJ8** | Cantu Syndrome | Sequencing of all coding exons of the gene |  |  | 1.3 |
| **KCNK18** | Migraine, with or without aura, susceptibility to, 13 | Sequencing of all coding exons of the gene |  |  | 1.2 |
| **KCNMA1** | Generalised Epilepsy with paroxysmal Dyskinesia | Sequencing of all coding exons of the gene |  |  | 3.7 |
| **KCNQ2** | Benign neonatal Epilepsy | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 2.6 |
| **KCNQ3** | Benign neonatal Epilepsy | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.6 |
| **KCNT1** | Epileptic Encephalopathy | Sequencing of all coding exons of the gene |  |  | 3.7 |
| **KCNT1** | Nocturnal frontal lobe Epilepsy | Sequencing of all coding exons of the gene |  |  | 3.7 |
| **KCNV2** | Retinal Cone Dystrophy 3B | Sequencing of all coding exons of the gene |  |  | 1.6 |
| **KCTD17** | Dystonia 26, myoclonic | Sequencing of all coding exons of the gene |  |  | 0.9 |
| **KCTD7** | Epilepsy, progressive myoclonic 3, with or without intracellular inclusions | Sequencing of all coding exons of the gene |  |  | 0.9 |
| **KIAA1549** | Retinitis Pigmentosa | Sequencing of all coding exons of the gene |  |  | 5.9 |
| **KIF1A** | Mental retardation, autosomal dominant 9 | Sequencing of all coding exons of the gene |  |  | 5.4 |
| **KIF1A** | Neuropathy, hereditary sensory, type IIC | Sequencing of all coding exons of the gene |  |  | 5.4 |
| **KIF1A** | Spastic paraplegia 30 | Sequencing of all coding exons of the gene |  |  | 5.4 |
| **KIF1B** | Charcot-Marie-Tooth disease, type 2A1 | Sequencing of all coding exons of the gene |  |  | 5.3 |
| **KIF5A** | Spastic Paraplegia 10 | Sequencing of all coding exons of the gene |  |  | 3.1 |
| **KIT** | Gastrointestinal stromal tumor, familial | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.9 |
| **KIT** | Germ cell tumors | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.9 |
| **KIT** | Leukemia, acute myeloid | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.9 |
| **KIT** | Mast cell disease | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.9 |
| **KIT** | Piebaldism | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.9 |
| **KMT2A** | Leukemia, myeloid/lymphoid or mixed-lineage | Sequencing of all coding exons of the gene |  |  | 11.9 |
| **KMT2A** | Wiedemann-Steiner syndrome | Sequencing of all coding exons of the gene |  |  | 11.9 |
| **KRAS** | Breast cancer, somatic | Sequencing of all coding exons of the gene |  |  | 0.6 |
| **KRAS** | Noonan syndrome | Sequencing of all coding exons of the gene |  |  | 0.6 |
| **KRT12** | Meesmann corneal dystrophy | Sequencing of all coding exons of the gene |  |  | 1.5 |
| **KRT16** | Pachyonychia congenita 1 | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **KRT16** | Palmoplantar keratoderma, nonepidermolytic, focal | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **KRT5** | Epidermolysis Bullosa Simplex, Dowling-Meara Type | Sequencing of all coding exons of the gene |  |  | 1.8 |
| **KRT5** | Epidermolysis Bullosa Simplex, generalized | Sequencing of all coding exons of the gene |  |  | 1.8 |
| **KRT5** | Epidermolysis Bullosa Simplex, localized | Sequencing of all coding exons of the gene |  |  | 1.8 |
| **KRT6C** | Palmoplantar keratoderma, nonepidermolytic, focal or diffuse | Sequencing of all coding exons of the gene |  |  | 1.7 |
| **L1CAM** | Corpus callosum, partial agenesis of | Sequencing of all coding exons of the gene |  |  | 3.8 |
| **L1CAM** | CRASH syndrome | Sequencing of all coding exons of the gene |  |  | 3.8 |
| **L1CAM** | Hydrocephalus due to aqueductal stenosis | Sequencing of all coding exons of the gene |  |  | 3.8 |
| **L1CAM** | Hydrocephalus with congenital idiopathic intestinal pseudoobstruction | Sequencing of all coding exons of the gene |  |  | 3.8 |
| **L1CAM** | Hydrocephalus with Hirschsprung disease | Sequencing of all coding exons of the gene |  |  | 3.8 |
| **L1CAM** | MASA syndrome | Sequencing of all coding exons of the gene |  |  | 3.8 |
| **L2HGDH** | L-2-Hydroxyglutaric Aciduria | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 1.4 |
| **LAMA1** | Poretti-Boltshauser syndrome | Sequencing of all coding exons of the gene |  |  | 9.2 |
| **LAMA2** | Muscular dystrophy, congenital | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 9.4 |
| **LARS** | Infantile liver failure syndrome 1 | Sequencing of all coding exons of the gene |  |  | 3.5 |
| **LBR** | Greenberg Dysplasia | Sequencing of all coding exons of the gene |  |  | 1.8 |
| **LDLR** | Autosomal dominant familial hypercholesterolemia (LDL receptor mutation) | Sequencing of all coding exons of the gene |  |  | 2.6 |
| **LEP** | Obesity, morbid, due to leptin deficiency | Sequencing of all coding exons of the gene |  |  | 0.5 |
| **LGI1** | Temporal lobe Epilepsy | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.7 |
| **LHCGR** | Leydig cell adenoma, somatic, with precocious puberty | Sequencing of all coding exons of the gene |  |  | 2.1 |
| **LHCGR** | Leydig cell hypoplasia with hypergonadotropic hypogonadism | Sequencing of all coding exons of the gene |  |  | 2.1 |
| **LHCGR** | Leydig cell hypoplasia with pseudohermaphroditism | Sequencing of all coding exons of the gene |  |  | 2.1 |
| **LHCGR** | Luteinizing hormone resistance, female | Sequencing of all coding exons of the gene |  |  | 2.1 |
| **LHCGR** | Precocious puberty, male | Sequencing of all coding exons of the gene |  |  | 2.1 |
| **LHON** | Leber optic atrophy | Sequencing of hotspots |  |  | 2.9 |
| **LHX3** | Pituitary hormone deficiency, combined, 3 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.2 |
| **LMNA** | Cardiomyopathy, dilated, 1A | Sequencing of all coding exons of the gene |  |  | 2 |
| **LMNA** | Charcot-Marie-Tooth disease, type 2B1 | Sequencing of all coding exons of the gene |  |  | 2 |
| **LMNA** | Emery-Dreifuss muscular dystrophy 2, AD | Sequencing of all coding exons of the gene |  |  | 2 |
| **LMNA** | Emery-Dreifuss muscular dystrophy 3, AR | Sequencing of all coding exons of the gene |  |  | 2 |
| **LMNA** | Heart-hand syndrome, Slovenian type | Sequencing of all coding exons of the gene |  |  | 2 |
| **LMNA** | Hutchinson-Gilford progeria | Sequencing of all coding exons of the gene |  |  | 2 |
| **LMNA** | Lipodystrophy, familial partial, 2 | Sequencing of all coding exons of the gene |  |  | 2 |
| **LMNA** | Malouf syndrome | Sequencing of all coding exons of the gene |  |  | 2 |
| **LMNA** | Mandibuloacral dysplasia | Sequencing of all coding exons of the gene |  |  | 2 |
| **LMNA** | Muscular dystrophy, congenital | Sequencing of all coding exons of the gene |  |  | 2 |
| **LMNA** | Muscular dystrophy, limb-girdle, type 1B | Sequencing of all coding exons of the gene |  |  | 2 |
| **LMNA** | Restrictive dermopathy, lethal | Sequencing of all coding exons of the gene |  |  | 2 |
| **LMNB1** | Leukodystrophy, Adult-Onset, Autosomal Dominant | Deletion and duplication analysis | Sequencing of all coding exons of the gene |  | 1.8 |
| **LMX1B** | Nail-patella syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.2 |
| **LPIN1** | Autosomal recessive acute recurrent Myoglobinuria | Sequencing of all coding exons of the gene |  |  | 2.9 |
| **LRAT** | Leber Congenital Amaurosis | Sequencing of all coding exons of the gene |  |  | 0.7 |
| **LRAT** | Retinitis Pigmentosa | Sequencing of all coding exons of the gene |  |  | 0.7 |
| **LRP5** | Familial Exudative Vitreoretinopathy | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 4.8 |
| **LRP5** | Hyperostosis Corticalis Generalisata, Benign Form of Worth, with Torus Palatinus | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 4.8 |
| **LRP5** | Osteoporosis Pseudoglioma Syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 4.8 |
| **LRP5** | Van Buchem Disease, Type 2 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 4.8 |
| **LRRC6** | Ciliary dyskinesia, primary, 19 | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **LRRK2** | Parkinson Syndrome | Sequencing of hotspots |  |  | 7.6 |
| **LYST** | Chediak-Higashi syndrome | Sequencing of all coding exons of the gene |  |  | 11.4 |
| **LYZ** | Amyloidosis, renal | Sequencing of all coding exons of the gene |  |  | 0.4 |
| **MAGEL2** | Schaaf-Yang syndrome |  |  |  | 3.8 |
| **MAN2B1** | Alpha-Mannosidosis | Sequencing of all coding exons of the gene |  |  | 3 |
| **MAPK10** | Epileptic encephalopathy, Lennox-Gastaut type | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **MAPT** | Frontotemporal Dementia | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.3 |
| **MARS2** | Spastic ataxia 3 | Sequencing of all coding exons of the gene |  |  | 1.8 |
| **MASP1** | 3MC syndrome 1 | Sequencing of all coding exons of the gene |  |  | 2.2 |
| **MATN3** | Multiple epiphyseal Dysplasia | Sequencing of hotspots | Sequencing of all coding exons of the gene |  | 1.5 |
| **MBD5** | Mental retardation, autosomal dominant 1 | Deletion and duplication analysis | Sequencing of all coding exons of the gene |  | 4.5 |
| **MC1R** | Albinism, oculocutaneous, type II, modifier of | Sequencing of all coding exons of the gene |  |  | 1 |
| **MC1R** | Melanoma, cutaneous malignant, 5 | Sequencing of all coding exons of the gene |  |  | 1 |
| **MC1R** | UV-induced skin damage | Sequencing of all coding exons of the gene |  |  | 1 |
| **MCOLN1** | Mucolipidosis IV | Sequencing of all coding exons of the gene |  |  | 1.7 |
| **MED13L** | Transposition of the great arteries, dextro-looped 1 | Sequencing of all coding exons of the gene |  |  | 6.6 |
| **MED17** | Microcephaly, postnatal progressive, with seizures and brain atrophy | Sequencing of all coding exons of the gene |  |  | 2 |
| **MEF2C** | Mental Retardation, Stereotypic Movements, Epilepsy, and/or Cerebral Malformations | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 1.5 |
| **MEFV - HS1** | FAMILIAL MEDITERRANEAN FEVER, AUTOSOMAL DOMINANT | Sequencing of hotspots | Sequencing of all coding exons of the gene |  | 1.8 |
| **MEFV - HS1** | FAMILIAL MEDITERRANEAN FEVER, AUTOSOMAL RECESSIVE | Sequencing of hotspots | Sequencing of all coding exons of the gene |  | 1.8 |
| **MEFV - HS2** | FAMILIAL MEDITERRANEAN FEVER, AUTOSOMAL DOMINANT | Sequencing of hotspots | Sequencing of all coding exons of the gene |  | 1.6 |
| **MEFV - HS2** | FAMILIAL MEDITERRANEAN FEVER, AUTOSOMAL RECESSIVE | Sequencing of hotspots | Sequencing of all coding exons of the gene |  | 1.6 |
| **MEFV - HS3** | FAMILIAL MEDITERRANEAN FEVER, AUTOSOMAL DOMINANT | Sequencing of hotspots | Sequencing of all coding exons of the gene |  | 0.8 |
| **MEFV - HS3** | FAMILIAL MEDITERRANEAN FEVER, AUTOSOMAL RECESSIVE | Sequencing of hotspots | Sequencing of all coding exons of the gene |  | 0.8 |
| **MEN1** | Hyperparathyroidism 1 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.8 |
| **MEN1** | Multiple Endocrine Neoplasia Type 1 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.8 |
| **MEOX1** | Klippel-Feil syndrome 2 | Sequencing of all coding exons of the gene |  |  | 0.8 |
| **MFN2** | Charcot-Marie-Tooth Neuropathy Type 2A2 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.3 |
| **MFSD8** | Neuronal Ceroid-Lipofuscinosis | Sequencing of all coding exons of the gene |  |  | 1.6 |
| **MFSD8** | Neuronal Ceroid-Lipofuscinosis | Sequencing of all coding exons of the gene |  |  | 1.6 |
| **MFSD8** | Neuronal Ceroid-Lipofuscinosis | Sequencing of all coding exons of the gene |  |  | 1.6 |
| **MGAT2** | CDG-Syndrome 2A | Sequencing of all coding exons of the gene |  |  | 1.3 |
| **MGME1** | Mitochondrial DNA depletion syndrome 11 | Sequencing of all coding exons of the gene |  |  | 1 |
| **MID1** | Opitz GBBB Syndrome 1 (X-linked) | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 2 |
| **MITF** | Tietz Syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.6 |
| **MITF** | Waardenburg Syndrome Type IIA | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.6 |
| **MOCS1** | Molybdenum Cofactor Deficiency | Sequencing of all coding exons of the gene |  |  | 1.2 |
| **MPDZ** | Nonsyndromic Hydrocephalus, autosomal recessive 2 | Sequencing of all coding exons of the gene |  |  | 6.1 |
| **MPI** | Congenital disorder of glycosylation, type Ib | Sequencing of all coding exons of the gene |  |  | 1.3 |
| **MPL** | Myelofibrosis with myeloid metaplasia, somatic | Sequencing of all coding exons of the gene |  |  | 1.9 |
| **MPL** | Thrombocythemia 2 | Sequencing of all coding exons of the gene |  |  | 1.9 |
| **MPL** | Thrombocytopenia, congenital amegakaryocytic | Sequencing of all coding exons of the gene |  |  | 1.9 |
| **MPZ** | Charcot-Marie-Tooth Neuropathy Type 2I | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 0.7 |
| **MPZ** | Charcot-Marie-Tooth Neuropathy Type 2J | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 0.7 |
| **MRE11** | Ataxia Teleangiectatica (AT) | Sequencing of all coding exons of the gene |  |  | 2.1 |
| **MSH3** | Endometrial carcinoma, somatic | Sequencing of all coding exons of the gene |  |  | 3.4 |
| **MSX1** | Orofacial Cleft 5 | Sequencing of all coding exons of the gene |  |  | 0.9 |
| **MSX1** | Tooth Agenesis, Selective, 1 | Sequencing of all coding exons of the gene |  |  | 0.9 |
| **MSX1** | Witkop Syndrome | Sequencing of all coding exons of the gene |  |  | 0.9 |
| **mtDNA** | Mitochondriopathies | Whole DNA sequencing |  |  | 16.6 |
| **MTM1** | X-Linked Centronuclear Myopathy | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.8 |
| **MTTP** | Abetalipoproteinemia | Sequencing of all coding exons of the gene |  |  | 2.7 |
| **MUSK** | FETAL AKINESIA DEFORMATION SEQUENCE; FADS | Sequencing of all coding exons of the gene |  |  | 2.6 |
| **MUSK** | MYASTHENIC SYNDROME, CONGENITAL, 9, ASSOCIATED WITH ACETYLCHOLINE RECEPTOR DEFICIENCY; CMS9 | Sequencing of all coding exons of the gene |  |  | 2.6 |
| **MUTYH** | Colorectal adenomatous polyposis | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.7 |
| **MVK** | Hyper-IgD syndrome | Sequencing of hotspots | Sequencing of all coding exons of the gene |  | 1.2 |
| **MVK** | Mevalonic aciduria | Sequencing of hotspots | Sequencing of all coding exons of the gene |  | 1.2 |
| **MVK** | Porokeratosis 3, disseminated superficial actinic | Sequencing of hotspots | Sequencing of all coding exons of the gene |  | 1.2 |
| **MYBPC3** | Dilated Cardiomyopathy | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 3.8 |
| **MYBPC3** | Dilated Cardiomyopathy | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 3.8 |
| **MYCN** | Feingold syndrome | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **MYH11** | Aortic aneurysm, familial thoracic 4 | Sequencing of all coding exons of the gene |  |  | 5.9 |
| **MYH3** | Arthrogryposis, distal, type 2A | Sequencing of all coding exons of the gene |  |  | 5.8 |
| **MYH3** | Arthrogryposis, distal, type 2B | Sequencing of all coding exons of the gene |  |  | 5.8 |
| **MYH3** | Freeman-Sheldon Syndrome | Sequencing of all coding exons of the gene |  |  | 5.8 |
| **MYH7** | Dilated Cardiomyopathy | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 5.8 |
| **MYH7** | Dilated Cardiomyopathy | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 5.8 |
| **MYH7** | Left ventricular noncompaction 5 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 5.8 |
| **MYH7** | MYH7-Related Myosin Storage Myopathy | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 5.8 |
| **MYH9** | Deafness, autosomal dominant 17 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 5.9 |
| **MYH9** | Epstein syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 5.9 |
| **MYH9** | Fechtner syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 5.9 |
| **MYH9** | Macrothrombocytopenia and progressive sensorineural deafness | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 5.9 |
| **MYH9** | May-Hegglin anomaly | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 5.9 |
| **MYH9** | Sebastian syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 5.9 |
| **NAGLU** | Charcot-Marie-Tooth disease, axonal, type 2V | Sequencing of all coding exons of the gene |  |  | 2.2 |
| **NAGLU** | Mucopolysaccharidosis type IIIB (Sanfilippo B) | Sequencing of all coding exons of the gene |  |  | 2.2 |
| **NDE1** | Lissencephaly 4 (with microcephaly) | Sequencing of all coding exons of the gene |  |  | 1 |
| **NDE1** | Microhydranencephaly | Sequencing of all coding exons of the gene |  |  | 1 |
| **NDP** | Retinopathy | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 0.4 |
| **NELFA** | Wolf-Hirschhorn Syndrome | Sequencing of all coding exons of the gene |  |  | 1.6 |
| **NF1** | Familial Spinal Neurofibromatosis | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 8.5 |
| **NF1** | Neurofibromatosis 1 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 8.5 |
| **NF1** | Neurofibromatosis-Noonan Syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 8.5 |
| **NF1** | Watson Syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 8.5 |
| **NF2** | Neurofibromatosis 2 | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 1.8 |
| **NFIX** | Marshall-Smith Syndrome | Sequencing of all coding exons of the gene |  |  | 1.5 |
| **NFIX** | Sotos Syndrome | Sequencing of all coding exons of the gene |  |  | 1.5 |
| **NGF** | Hereditary Sensory and Autonomic Neuropathy Type V | Sequencing of all coding exons of the gene |  |  | 0.7 |
| **NHEJ1** | SCID | Sequencing of all coding exons of the gene |  |  | 0.9 |
| **NHLRC1** | Lafora Disease | Sequencing of all coding exons of the gene |  |  | 1.2 |
| **NIPBL** | Cornelia de Lange Syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 8.4 |
| **NKX2-1** | Benign Chorea | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.2 |
| **NKX2-5** | Congenital Hypothyroidism | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1 |
| **NLGN3** | Asperger syndrome susceptibility, X-linked 1 | Sequencing of all coding exons of the gene |  |  | 2.5 |
| **NLGN3** | Autism susceptibility, X-linked 1 | Sequencing of all coding exons of the gene |  |  | 2.5 |
| **NLGN4X** | Asperger syndrome susceptibility, X-linked 2 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.5 |
| **NLGN4X** | Autism susceptibility, X-linked 2 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.5 |
| **NLGN4X** | Mental retardation, X-linked | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.5 |
| **NLRC4** | Autoinflammation with infantile enterocolitis | Sequencing of all coding exons of the gene |  |  | 3.1 |
| **NLRC4** | Familial cold autoinflammatory syndrome 4 | Sequencing of all coding exons of the gene |  |  | 3.1 |
| **NLRP3** | Chronic Infantile Neurological Cutaneous and Articular Syndrome | Sequencing of hotspots | Sequencing of all coding exons of the gene |  | 3.1 |
| **NLRP3** | Familial Cold Autoinflamma- tory Syndrome (FCAS) | Sequencing of hotspots | Sequencing of all coding exons of the gene |  | 3.1 |
| **NLRP3** | Muckle-Wells Syndrome | Sequencing of hotspots | Sequencing of all coding exons of the gene |  | 3.1 |
| **NME8** | Ciliary dyskinesia, primary, 6 | Sequencing of all coding exons of the gene |  |  | 1.8 |
| **NOD2** | Blau syndrome/early-onset sarcoidosis (mutation in exon 4 of the NOD2 gene) | Sequencing of all coding exons of the gene |  |  | 3.1 |
| **NOD2** | Inflammatory bowel disease 1 (analysis of the three most frequent predisposing mutations in the NOD2 gene) | Sequencing of all coding exons of the gene |  |  | 3.1 |
| **NOL3** | Myoclonus, familial cortical | Sequencing of all coding exons of the gene |  |  | 0.8 |
| **NOTCH3** | CADASIL | Sequencing of all coding exons of the gene |  |  | 7 |
| **NPC1** | Niemann-Pick Disease  Type C | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 3.8 |
| **NPC2** | Niemann-Pick Disease  Type C | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 0.5 |
| **NPHP1** | Joubert Syndrome | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 2.2 |
| **NPHP1** | Senior-Loken syndrome 1 | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 2.2 |
| **NPHP3** | Meckel Syndrome | Sequencing of all coding exons of the gene |  |  | 4 |
| **NPHP4** | Senior-Loken syndrome 4 | Sequencing of all coding exons of the gene |  |  | 4.3 |
| **NR2E3** | Goldmann-Favre-Syndrome | Sequencing of all coding exons of the gene |  |  | 1.2 |
| **NR2E3** | Retinitis Pigmentosa | Sequencing of all coding exons of the gene |  |  | 1.2 |
| **NRXN1** | Pitt-Hopkins-Like Syndrome 2 | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 4.6 |
| **NSD1** | Sotos Syndrome | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 8.1 |
| **NSD1** | Weaver Syndrome 1 | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 8.1 |
| **NYX** | Congenital Stationary Night-Blindness | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **OAT** | Gyrate atrophy of choroid and retina with or without ornithinemia | Sequencing of all coding exons of the gene |  |  | 1.3 |
| **OCA2** | Albinism, brown oculocutaneous | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.5 |
| **OCA2** | Albinism, oculocutaneous, type II | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.5 |
| **OCRL** | Dent disease 2 | Sequencing of all coding exons of the gene |  |  | 2.7 |
| **OCRL** | Lowe syndrome | Sequencing of all coding exons of the gene |  |  | 2.7 |
| **OFD1** | Joubert Syndrome | Sequencing of all coding exons of the gene |  |  | 3 |
| **OFD1** | Oral-Facial-Digital Syndrome Type 1 | Sequencing of all coding exons of the gene |  |  | 3 |
| **OFD1** | Simpson-Golabi-Behmel Syndrome Type 2 | Sequencing of all coding exons of the gene |  |  | 3 |
| **OGDH** | Alpha-ketoglutarate dehydrogenase deficiency | Sequencing of all coding exons of the gene |  |  | 3.1 |
| **OPA1** | Optic Atrophy | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 3 |
| **OPA3** | Optic Atrophy | Sequencing of all coding exons of the gene |  |  | 0.5 |
| **OPHN1** | X-Linked Mental Retardation with Cerebellar Hypoplasia and Distinctive Facial Appearance | Sequencing of all coding exons of the gene |  |  | 2.4 |
| **OPTN** | Glaucoma, Open Angle | Sequencing of all coding exons of the gene |  |  | 1.7 |
| **ORAI1** | Immunodeficiency 9 | Sequencing of all coding exons of the gene |  |  | 0.9 |
| **ORAI1** | Myopathy, tubular aggregate, 2 | Sequencing of all coding exons of the gene |  |  | 0.9 |
| **ORC4** | Meier-Gorlin Syndrome 2 | Sequencing of all coding exons of the gene |  |  | 1.3 |
| **ORC6** | Meier-Gorlin Syndrome 3 | Sequencing of all coding exons of the gene |  |  | 0.8 |
| **OTOF** | DFNB 1 Nonsyndromic Hearing Loss and Deafness | Sequencing of all coding exons of the gene |  |  | 6 |
| **OTX2** | Microphthalmia, syndromic 5 | Sequencing of all coding exons of the gene |  |  | 0.9 |
| **OTX2** | Retinal Dystrophy, Early-Onset, and Pituitary Dysfunction | Sequencing of all coding exons of the gene |  |  | 0.9 |
| **P2RX7** | Chronic lymphocytic leukemia (CLL) | Sequencing of all coding exons of the gene |  |  | 1.8 |
| **PABPN1** | Oculopharyngeal Muscular Dystrophy | Sequencing of all coding exons of the gene |  |  | 0.9 |
| **PAFAH1B1** | Lissencephaly | Deletion and duplication analysis | Sequencing of all coding exons of the gene |  | 1.2 |
| **PAH** | Phenylketonuria | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.4 |
| **PALB2** | Hereditary Breast Cancer | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 3.6 |
| **PANK2** | Neurodegeneration with brain iron accumulation (NBIA) | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.7 |
| **PARK2** | Juvenile Parkinson Syndrome | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 1.4 |
| **PARK7** | Juvenile Parkinson Syndrome | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 0.6 |
| **PAX6** | Aniridia | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 1.3 |
| **PAX6** | Cataract with late-onset corneal dystrophy | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 1.3 |
| **PAX6** | Coloboma of optic nerve | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 1.3 |
| **PAX6** | Coloboma, ocular | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 1.3 |
| **PAX8** | Congenital Hypothyroidism | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.4 |
| **PCDH19** | Epileptic Encephalopathy | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 3.4 |
| **PDE4D** | Acrodysostosis 2, with or without hormone resistance | Sequencing of all coding exons of the gene |  |  | 2.4 |
| **PDE6C** | Achromatopsia | Sequencing of all coding exons of the gene |  |  | 2.6 |
| **PDE6H** | Retinal Cone Dystrophy 3A | Sequencing of all coding exons of the gene |  |  | 0.3 |
| **PDHA1** | Pyruvate Dehydrogenase (PDH)-Deficiency | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 1.3 |
| **PEX1** | Peroxisome Biogenesis Disorders (PBD) | Sequencing of all coding exons of the gene |  |  | 3.9 |
| **PEX1** | Zellweger Syndrome | Sequencing of all coding exons of the gene |  |  | 3.9 |
| **PEX10** | Zellweger Syndrome | Sequencing of all coding exons of the gene |  |  | 1 |
| **PEX12** | Zellweger Syndrome | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **PEX2** | Zellweger Syndrome | Sequencing of all coding exons of the gene |  |  | 0.9 |
| **PEX26** | Zellweger Syndrome | Sequencing of all coding exons of the gene |  |  | 0.9 |
| **PEX5** | Zellweger Syndrome | Sequencing of all coding exons of the gene |  |  | 2 |
| **PEX6** | Zellweger Syndrome | Sequencing of all coding exons of the gene |  |  | 2.9 |
| **PEX7** | Peroxisome Biogenesis Disorders (PBD) | Sequencing of all coding exons of the gene |  |  | 1 |
| **PFN1** | Cardiac Diseases | Sequencing of all coding exons of the gene |  |  | 0.4 |
| **PGAM2** | Glycogen Storage Disease X | Sequencing of all coding exons of the gene |  |  | 0.8 |
| **PGK1** | Phosphoglycerate kinase 1 deficiency | Sequencing of all coding exons of the gene |  |  | 1.3 |
| **PHEX** | Hypophosphatemic rickets, X-linked dominant | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.3 |
| **PHF6** | Borjeson-Forssman-Lehmann Syndrome | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **PHYH** | Refsum disease | Sequencing of all coding exons of the gene |  |  | 1 |
| **PIGA** | Paroxysomal Nocturnal Hemoglobinuria | Sequencing of all coding exons of the gene |  |  | 1.5 |
| **PIK3R2** | Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1 | Sequencing of all coding exons of the gene |  |  | 2.2 |
| **PINK1** | Juvenile Parkinson Syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.7 |
| **PITX2** | Axenfeld-Rieger Syndrome | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 1 |
| **PITX2** | Peters Anomaly | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 1 |
| **PKHD1** | Polycystic kidney and hepatic disease | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 12.2 |
| **PKLR** | Pyruvate kinase deficiency | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.7 |
| **PLA2G6** | Neurodegeneration with brain iron accumulation (NBIA) | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.4 |
| **PLA2G6** | Parkinsonism and Dystonia | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.4 |
| **PLCB1** | Epileptic Encephalopathy, Early Infantile, 12 | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 3.7 |
| **PLCB1** | Epileptic Encephalopathy, Early Infantile, 12 | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 3.7 |
| **PLCG2** | Autoinflammation, antibody deficiency, and immune dysregulation syndrome | Deletion and duplication analysis | Sequencing of all coding exons of the gene |  | 3.8 |
| **PLCG2** | Familial cold autoinflammatory syndrome 3 | Deletion and duplication analysis | Sequencing of all coding exons of the gene |  | 3.8 |
| **PLEC** | Epidermolysis bullosa simplex with muscular dystrophy | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 14.1 |
| **PLEC** | Epidermolysis bullosa simplex with nail dystrophy | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 14.1 |
| **PLEC** | Epidermolysis bullosa simplex with pyloric atresia | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 14.1 |
| **PLEC** | Epidermolysis bullosa simplex, Ogna type | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 14.1 |
| **PLEC** | Muscular dystrophy, limb-girdle, type 2Q | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 14.1 |
| **PLP1** | Pelizaeus-Merzbacher disease | Deletion and duplication analysis | Sequencing of all coding exons of the gene |  | 0.8 |
| **PLP1** | Spastic paraplegia 2, X-linked | Deletion and duplication analysis | Sequencing of all coding exons of the gene |  | 0.8 |
| **PMM2** | CDG-Syndrome 1A | Sequencing of all coding exons of the gene |  |  | 0.7 |
| **PMP22** | Charcot-Marie-Tooth Neuropathy Type 1A | Deletion and duplication analysis | Sequencing of all coding exons of the gene |  | 0.5 |
| **PMP22** | Hereditary Neuropathy with liability to pressure palsies | Deletion and duplication analysis | Sequencing of all coding exons of the gene |  | 0.5 |
| **PNKD** | Paroxysmal nonkinesiogenic  Dyskinesia 1 PNKD1 | Sequencing of all coding exons of the gene |  |  | 1.2 |
| **PNKP** | Epileptic encephalopathy, early infantile, 10 | Sequencing of all coding exons of the gene |  |  | 1.6 |
| **PNPLA3** | Susceptibility to nonalcoholic fatty liver disease (PNPLA3 gene) | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **PNPO** | Pyridoxamine 5-prime-phosphate oxidase deficiency | Sequencing of all coding exons of the gene |  |  | 0.8 |
| **POLE** | FILS syndrome | Sequencing of all coding exons of the gene |  |  | 6.9 |
| **POLG** | Alpers Syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 3.7 |
| **POLG** | Mitochondrial recessive Ataxia Syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 3.7 |
| **POLG** | POLG-Related Ataxia Neuropathy Spectrum Disorders | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 3.7 |
| **POLG** | Progressive external  Ophthalmoplegia | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 3.7 |
| **POLG2** | Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4 | Sequencing of all coding exons of the gene |  |  | 1.5 |
| **POLR1C** | Leukodystrophy, hypomyelinating, 11 | Sequencing of all coding exons of the gene |  |  | 1 |
| **POLR1C** | Treacher Collins syndrome 3 | Sequencing of all coding exons of the gene |  |  | 1 |
| **POLR1D** | Treacher Collins syndrome 2 | Sequencing of all coding exons of the gene |  |  | 0.4 |
| **POLR3A** | Leukodystrophy, hypomyelinating, 7 | Sequencing of all coding exons of the gene |  |  | 4.2 |
| **POU1F1** | Pituitary hormone deficiency, combined, 1 | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 1 |
| **PPM1K** | Maple syrup urine disease, mild variant | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **PPT1** | Neuronal Ceroid-Lipofuscinoses | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 0.9 |
| **PQBP1** | Renpenning Syndrome 1 | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 0.8 |
| **PRF1** | Hemophagocytic lymphohistiocytosis, familial, 2 | Sequencing of all coding exons of the gene |  |  | 1.7 |
| **PRF1** | Lymphoma, non-Hodgkin | Sequencing of all coding exons of the gene |  |  | 1.7 |
| **PRICKLE1** | Progressive Myoclonus Epilepsy with Ataxia | Sequencing of all coding exons of the gene |  |  | 2.5 |
| **PRICKLE1** | Progressive Myoclonus Epilepsy with Ataxia | Sequencing of all coding exons of the gene |  |  | 2.5 |
| **PRKAR1A** | Carney Complex | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.1 |
| **PRKCG** | Spinocerebellar ataxia 14 | Sequencing of all coding exons of the gene |  |  | 2.1 |
| **PRKRA** | Dystonia 16 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 0.9 |
| **PRKRA** | Early Onset Dystonia with Parkinsonism | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 0.9 |
| **PRNP** | Genetic Prion Disease | Sequencing of all coding exons of the gene |  |  | 0.8 |
| **PROC** | THROMBOPHILIA DUE TO PROTEIN C DEFICIENCY, AUTOSOMAL DOMINANT; THPH3 | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **PROK1** | Hirschsprung Disease | Sequencing of all coding exons of the gene |  |  | 0.3 |
| **PROM1** | Cone-Rod-Dystrophy | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.6 |
| **PROP1** | Pituitary hormone deficiency, combined, 2 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 0.7 |
| **PRPF31** | Retinitis Pigmentosa | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.5 |
| **PRPH2** | Adult-Onset Vitelliform Macular Dystrophy | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1 |
| **PRPH2** | Cone-Rod-Dystrophy | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1 |
| **PRPH2** | Patterned Dystrophy of Retinal Pigment Epithelium | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1 |
| **PRPH2** | Retinitis Pigmentosa | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1 |
| **PRPS1** | Charcot-Marie-Tooth Neuropathy X Type 5 | Sequencing of all coding exons of the gene |  |  | 1 |
| **PRPS1** | DFNX1 (DFN2) Nonsyndromic Hearing Loss and Deafness | Sequencing of all coding exons of the gene |  |  | 1 |
| **PRRT2** | Familial Paroxysmal Kinesigenic Dyskinesia | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.2 |
| **PRSS1** | Pancreatitis, hereditary | Sequencing of all coding exons of the gene |  |  | 0.7 |
| **PRSS1** | Trypsinogen deficiency | Sequencing of all coding exons of the gene |  |  | 0.7 |
| **PSAP** | Encephalopathy due to prosaposin deficiency | Sequencing of all coding exons of the gene |  |  | 1.6 |
| **PSAP** | Gaucher disease | Sequencing of all coding exons of the gene |  |  | 1.6 |
| **PSAP** | Metachromatic Leukodystrophy | Sequencing of all coding exons of the gene |  |  | 1.6 |
| **PSEN1** | Alzheimer Dementia | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.4 |
| **PSEN2** | Alzheimer Dementia | Sequencing of all coding exons of the gene |  |  | 1.3 |
| **PSMB8** | Autoinflammation, lipodystrophy, and dermatosis syndrome (Nakajo-Nishimura or CANDLE syndrome; PSMB8 gene) | Sequencing of all coding exons of the gene |  |  | 0.8 |
| **PSTPIP1** | Pyogenic sterile arthritis, pyoderma gangrenosum, and acne | Sequencing of all coding exons of the gene |  |  | 1.3 |
| **PTCH1** | Holoprosencephaly | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 4.3 |
| **PTCH2** | Basal cell carcinoma, somatic | Sequencing of all coding exons of the gene |  |  | 3.6 |
| **PTCH2** | Basal cell nevus syndrome | Sequencing of all coding exons of the gene |  |  | 3.6 |
| **PTCH2** | Medulloblastoma, desmoplastic | Sequencing of all coding exons of the gene |  |  | 3.6 |
| **PTDSS1** | Lenz-Majewski hyperostotic dwarfism | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **PTEN** | Bannayan-Riley-Ruvalcaba syndrome | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 1.2 |
| **PTEN** | Cowden syndrome 1 | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 1.2 |
| **PTEN** | Lhermitte-Duclos syndrome | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 1.2 |
| **PTEN** | Macrocephaly/Autism Syndrome | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 1.2 |
| **PTH** | HYPOPARATHYROIDISM, AUTOSOMAL DOMINANT | Sequencing of all coding exons of the gene |  |  | 0.3 |
| **PTH** | HYPOPARATHYROIDISM, AUTOSOMAL RECESSIVE | Sequencing of all coding exons of the gene |  |  | 0.3 |
| **PTPN11** | LEOPARD syndrome 1 | Sequencing of all coding exons of the gene |  |  | 1.8 |
| **PTPN11** | Leukemia, juvenile myelomonocytic, somatic | Sequencing of all coding exons of the gene |  |  | 1.8 |
| **PTPN11** | Metachondromatosis | Sequencing of all coding exons of the gene |  |  | 1.8 |
| **PTPN11** | Noonan syndrome 1 | Sequencing of all coding exons of the gene |  |  | 1.8 |
| **PTRF** | Kongenitale generalisierte Lipodystrophie mit Muskeldystrophie | Sequencing of all coding exons of the gene |  |  | 1.2 |
| **PTS** | 6-Pyruvoyltetrahydropterin Synthase Deficiency | Sequencing of all coding exons of the gene |  |  | 0.4 |
| **PYGM** | Glycogen Storage Disease Type V | Sequencing of all coding exons of the gene |  |  | 2.5 |
| **QDPR** | Dihydropteridine reductase deficiency | Sequencing of all coding exons of the gene |  |  | 0.7 |
| **RAB3GAP1** | Warburg Micro Syndrome 1 | Sequencing of all coding exons of the gene |  |  | 3 |
| **RAD21** | Cornelia de Lange Syndrome | Sequencing of all coding exons of the gene |  |  | 1.9 |
| **RAD51** | Mirror movements 2 | Sequencing of all coding exons of the gene |  |  | 1 |
| **RAD51D** | RAD51D-Related Familial Susceptibility to Breast-Ovarian Cancer | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1 |
| **RAI1** | Smith-Magenis-Syndrome (SMS) | Sequencing of all coding exons of the gene |  |  | 5.7 |
| **RAPSN** | Myasthenic Syndrome | Sequencing of all coding exons of the gene |  |  | 1.2 |
| **RARS2** | Pontocerebellar Hypoplasia Type 6 | Sequencing of all coding exons of the gene |  |  | 1.7 |
| **RBBP8** | Jawad Syndrome | Sequencing of all coding exons of the gene |  |  | 2.7 |
| **RBBP8** | Seckel Syndrome | Sequencing of all coding exons of the gene |  |  | 2.7 |
| **RBM10** | TARP syndrome | Sequencing of all coding exons of the gene |  |  | 2.8 |
| **RBP3** | Retinitis Pigmentosa | Sequencing of all coding exons of the gene |  |  | 3.7 |
| **REEP1** | Spastic Paraplegia 31 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 0.6 |
| **REPS1** | Neurodegeneration with brain iron accumulation (NBIA) | Sequencing of all coding exons of the gene |  |  | 2.4 |
| **RHO** | Night blindness, congenital stationary, autosomal dominant 1 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1 |
| **RHO** | Retinitis pigmentosa 4, autosomal dominant or recessive | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1 |
| **RHO** | Retinitis punctata albescens | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1 |
| **RLBP1** | Bothnia Retinal Dysfunction | Sequencing of all coding exons of the gene |  |  | 1 |
| **RLBP1** | Cone-Rod-Dystrophy | Sequencing of all coding exons of the gene |  |  | 1 |
| **RLBP1** | Fundus albipunctatus | Sequencing of all coding exons of the gene |  |  | 1 |
| **RLBP1** | Retinis Punctata | Sequencing of all coding exons of the gene |  |  | 1 |
| **RNASEH2B** | Aicardi-Goutieres syndrome 2 | Sequencing of all coding exons of the gene |  |  | 0.9 |
| **RNASET2** | Leukoencephalopathy, cystic, without megalencephaly | Sequencing of all coding exons of the gene |  |  | 0.8 |
| **ROBO3** | Horizontal Gaze Palsy and Scoliosis | Sequencing of all coding exons of the gene |  |  | 4.2 |
| **ROGDI** | Kohlschutter-Tonz Syndrome | Sequencing of all coding exons of the gene |  |  | 0.9 |
| **RP1** | Retinitis Pigmentosa | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 6.5 |
| **RP1L1** | Occult Macular Dystrophy (OCMD) | Sequencing of all coding exons of the gene |  |  | 7.2 |
| **RP2** | Retinitis Pigmentosa | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 1.1 |
| **RPE65** | Autosomal Dominant  Retinitis Pigmentosa | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.6 |
| **RPE65** | Autosomal Recessive Retinitis Pigmentosa | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.6 |
| **RPE65** | Leber Congenital Amaurosis 2 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.6 |
| **RPGR** | Retinitis Pigmentosa | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 3.5 |
| **RRM2B** | Mitochondrial DNA Depletion Syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.1 |
| **RS1** | X-Linked Juvenile Retinoschisis | Sequencing of all coding exons of the gene |  |  | 0.7 |
| **RSPH1** | Ciliary dyskinesia, primary, 24 | Sequencing of all coding exons of the gene |  |  | 0.9 |
| **RUNX2** | Cleidocranial dysplasia | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.6 |
| **RUNX2** | Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.6 |
| **SAA1** | Susceptibility to amyloidosis (SAA1 amino acids 70 and 75 encoded by exon 3) | Sequencing of hotspots |  |  | 0.4 |
| **SACS** | Autosomal Recessive Spastic Ataxia Charlevoix-Saguenay | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 13.7 |
| **SALL1** | Townes-Brocks branchiootorenal-like syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 4 |
| **SALL1** | Townes-Brocks syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 4 |
| **SALL4** | Duane-radial ray syndrome | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 3.2 |
| **SALL4** | IVIC syndrome | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 3.2 |
| **SAMHD1** | Aicardi-Goutieres syndrome 5 | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 1.9 |
| **SAMHD1** | Chilblain lupus 2 | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 1.9 |
| **SCN1A** | Familial hemiplegic Migraine Type 3 | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 6 |
| **SCN1A** | Generalised Epilepsy with febrile seizures plus (GEFS+) | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 6 |
| **SCN1B** | Generalised Epilepsy with febrile seizures plus (GEFS+) | Sequencing of all coding exons of the gene |  |  | 0.8 |
| **SCN2A** | Generalised Epilepsy with febrile seizures plus (GEFS+) | Sequencing of all coding exons of the gene |  |  | 6 |
| **SCN3A** | Cryptogenic pediatric partial epilepsy | Sequencing of all coding exons of the gene |  |  | 6 |
| **SCN3A** | Focal epilepsy | Sequencing of all coding exons of the gene |  |  | 6 |
| **SCN3A** | Grand mal epilepsy | Sequencing of all coding exons of the gene |  |  | 6 |
| **SCN4A** | Myasthenic Syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 5.5 |
| **SCN4A** | Paramyotonia Congenita | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 5.5 |
| **SCN8A** | Epileptic Encephalopathy, Early Infantile, 13 | Sequencing of all coding exons of the gene |  |  | 5.9 |
| **SCN9A** | Congenital Indifference to Pain, Autosomal Recessive | Sequencing of all coding exons of the gene |  |  | 5.9 |
| **SCN9A** | Generalised Epilepsy with febrile seizures plus (GEFS+) | Sequencing of all coding exons of the gene |  |  | 5.9 |
| **SCN9A** | Inherited Erythromelalgia | Sequencing of all coding exons of the gene |  |  | 5.9 |
| **SCN9A** | Paroxysmal Extreme Pain Disorder | Sequencing of all coding exons of the gene |  |  | 5.9 |
| **SCNN1B** | Pseudohypoaldosteronism Type 1 | Sequencing of all coding exons of the gene |  |  | 1.9 |
| **SCNN1G** | Pseudohypoaldosteronism Type 1 | Sequencing of all coding exons of the gene |  |  | 2 |
| **SCP2** | Leukoencephalopathy with dystonia and motor neuropathy | Sequencing of all coding exons of the gene |  |  | 1.6 |
| **SDHAF2** | Paragangliomas 2 | Sequencing of all coding exons of the gene |  |  | 0.5 |
| **SDHB** | Cowden-Syndrome | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 0.8 |
| **SDHB** | Hereditary Paraganglioma-Pheochromocytoma Syndromes | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 0.8 |
| **SDHB** | Paraganglioma and Gastric Stromal Sarcoma | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 0.8 |
| **SDHD** | Cowden-Syndrome | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 0.5 |
| **SDHD** | Hereditary Paraganglioma-Pheochromocytoma Syndromes | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 0.5 |
| **SDHD** | Paraganglioma and Gastric Stromal Sarcoma | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 0.5 |
| **SEMA3E** | CHARGE Syndrome | Sequencing of all coding exons of the gene |  |  | 2.3 |
| **SEPT9** | Amyotrophy, hereditary neuralgic | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.8 |
| **SEPT9** | Leukemia, acute myeloid, therapy-related | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.8 |
| **SEPT9** | Ovarian carcinoma | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.8 |
| **SERPINC1** | Thrombophilia due to Antithrombin III Deficiency | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 1.4 |
| **SERPING1** | Angioedema, hereditary, types I and II | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.5 |
| **SERPING1** | Complement component 4, partial deficiency of | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.5 |
| **SETBP1** | Schinzel-Giedion Midface Retraction Syndrome | Sequencing of all coding exons of the gene |  |  | 4.8 |
| **SETX** | Cardiac Diseases | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 8 |
| **SETX** | Spinocerebellar Ataxia with Axonal Neuropathy Type 2 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 8 |
| **SFTPB** | Pulmonary surfactant metabolism dysfunction type 1 (Surfactant protein B deficiency; SFTPB gene) | Sequencing of all coding exons of the gene |  |  | 1.2 |
| **SFTPB** | Surfactant metabolism dysfunction, pulmonary, 1 | Sequencing of all coding exons of the gene |  |  | 1.2 |
| **SFTPC** | Surfactant metabolism dysfunction, pulmonary, 2 | Sequencing of all coding exons of the gene |  |  | 0.6 |
| **SGCE** | Myclonus Dystonia | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.4 |
| **SGSH** | Mucopolysaccharidosis Type IIIA | Sequencing of all coding exons of the gene |  |  | 1.5 |
| **SHH** | Holoprosencephaly-3 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.4 |
| **SHH** | Microphthalmia with coloboma 5 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.4 |
| **SHH** | Schizencephaly | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.4 |
| **SHH** | Schizencephaly | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.4 |
| **SHH** | Single median maxillary central incisor | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.4 |
| **SIL1** | Marinesco-Sjögren-Syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.4 |
| **SIX3** | Holoprosencephaly | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1 |
| **SIX3** | Schizencephaly | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1 |
| **SIX3** | Schizencephaly | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1 |
| **SLC12A3** | Gitelman Syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 3.1 |
| **SLC12A5** | Epileptic encephalopathy, early infantile, 34 | Sequencing of all coding exons of the gene |  |  | 3.4 |
| **SLC16A12** | Cataract, juvenile, with microcornea and glucosuria | Sequencing of all coding exons of the gene |  |  | 1.6 |
| **SLC17A5** | Salla Disease | Sequencing of all coding exons of the gene |  |  | 1.5 |
| **SLC19A3** | Biotin-Responsive Basal Ganglia Disease | Sequencing of all coding exons of the gene |  |  | 1.5 |
| **SLC1A3** | Episodic Ataxia Type 6 | Sequencing of all coding exons of the gene |  |  | 1.6 |
| **SLC22A5** | Carnitine deficiency, systemic primary | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.7 |
| **SLC25A15** | Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome | Sequencing of all coding exons of the gene |  |  | 0.9 |
| **SLC25A19** | Progressive demyelinating neuropathy | Sequencing of all coding exons of the gene |  |  | 1 |
| **SLC25A22** | Epileptic Encephalopathy, Early Infantile, 3 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1 |
| **SLC25A3** | Mitochondrial Phosphate Carrier Deficiency | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **SLC26A2** | Multiple epiphyseal Dysplasia | Sequencing of all coding exons of the gene |  |  | 2.2 |
| **SLC26A4** | Pendred Syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.3 |
| **SLC26A5** | DFNB61 Nonsyndromic Hearing Loss and Deafness | Sequencing of all coding exons of the gene |  |  | 2.2 |
| **SLC2A1** | GLUT1 Deficiency Syndrome | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 1.5 |
| **SLC2A10** | Arterial tortuosity syndrome | Sequencing of all coding exons of the gene |  |  | 1.6 |
| **SLC30A10** | Hypermanganesemia with dystonia, polycythemia, and cirrhosis | Sequencing of all coding exons of the gene |  |  | 1.5 |
| **SLC35A1** | Congenital disorder of glycosylation, type IIf | Sequencing of all coding exons of the gene |  |  | 1 |
| **SLC35D1** | Schneckenbecken dysplasia | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **SLC36A2** | Iminoglycinuria | Sequencing of all coding exons of the gene |  |  | 1.5 |
| **SLC3A1** | Cystinuria | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 2.1 |
| **SLC40A1** | Hereditary Hemochromatosis | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.7 |
| **SLC45A2** | Oculocutaneous Albinism Type 4 | Sequencing of all coding exons of the gene |  |  | 1.6 |
| **SLC46A1** | Hereditary Folate Malabsorption | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **SLC4A1** | Ovalocytosis | Sequencing of all coding exons of the gene |  |  | 2.7 |
| **SLC4A1** | Renal tubular acidosis, distal, AD | Sequencing of all coding exons of the gene |  |  | 2.7 |
| **SLC4A1** | Renal tubular acidosis, distal, AR | Sequencing of all coding exons of the gene |  |  | 2.7 |
| **SLC4A1** | Spherocytosis, type 4 | Sequencing of all coding exons of the gene |  |  | 2.7 |
| **SLC5A5** | Thyroid dyshormonogenesis 1 | Sequencing of all coding exons of the gene |  |  | 1.9 |
| **SLC5A6** | Association with brain, immune, bone, and intestinal dysfunction in a young child | Sequencing of all coding exons of the gene |  |  | 1.9 |
| **SLC6A1** | MYOCLONIC-ATONIC EPILEPSY; MAE | Sequencing of all coding exons of the gene |  |  | 1.8 |
| **SLC6A20** | Hyperglycinuria | Sequencing of all coding exons of the gene |  |  | 1.8 |
| **SLC6A20** | Iminoglycinuria | Sequencing of all coding exons of the gene |  |  | 1.8 |
| **SLC6A3** | Parkinsonism-dystonia, infantile | Sequencing of all coding exons of the gene |  |  | 1.9 |
| **SLC6A5** | Hyperekplexia | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.4 |
| **SLC6A8** | Cerebral creatine deficiency syndrome 1 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.9 |
| **SMAD4** | Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.7 |
| **SMAD4** | Myhre syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.7 |
| **SMAD4** | Polyposis, juvenile intestinal | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.7 |
| **SMC1A** | Cornelia de Lange Syndrome | Sequencing of all coding exons of the gene |  |  | 3.7 |
| **SMC3** | Cornelia de Lange Syndrome | Sequencing of all coding exons of the gene |  |  | 3.7 |
| **SMPD1** | Niemann-Pick Disease Type A / Type B | Sequencing of all coding exons of the gene; Sequencing of all coding exons of the gene | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  | 1.9 |
| **SMS** | X-linked mental Retardation, Snyder-Robinson Type | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **SNAP29** | CEDNIK syndrome | Sequencing of all coding exons of the gene |  |  | 0.8 |
| **SNCA** | Parkinson Syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 0.4 |
| **SOX10** | Waardenburg Syndrome Type 2E | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.4 |
| **SOX10** | Waardenburg Syndrome Type 4C | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.4 |
| **SOX3** | Mental retardation, X-linked, with isolated growth hormone deficiency | Sequencing of all coding exons of the gene |  |  | 1.3 |
| **SOX3** | Panhypopituitarism, X-linked | Sequencing of all coding exons of the gene |  |  | 1.3 |
| **SPAST** | Spastic Paraplegia 4 | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 1.9 |
| **SPATA5** | Epilepsy, hearing loss and mental retardation syndrome | Sequencing of all coding exons of the gene |  |  | 2.7 |
| **SPG11** | Spastic paraplegia 11 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 7.3 |
| **SPG7** | Spastic Paraplegia 7 | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 2.4 |
| **SPINK1** | Pancreatitis, hereditary | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 0.2 |
| **SPINK1** | Tropical calcific pancreatitis | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 0.2 |
| **SPR** | Dopa-Responsive Dyst. due to Sepiapterin Red. Deficiency | Sequencing of all coding exons of the gene |  |  | 0.8 |
| **SPRED1** | Legius syndrome | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 1.3 |
| **SPTAN1** | Epileptic Encephalopathy, Early Infantile, 5 | Sequencing of all coding exons of the gene |  |  | 7.4 |
| **SPTLC1** | Neuropathy, hereditary sensory and autonomic, type IA | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **SRCAP** | Floating-Harbor syndrome | Sequencing of all coding exons of the gene |  |  | 9.7 |
| **SRPX2** | Rolandic Epilepsy | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **ST3GAL3** | Epileptic encephalopathy, early infantile, 15 | Sequencing of all coding exons of the gene |  |  | 1.3 |
| **ST3GAL3** | Mental retardation, autosomal recessive 12 | Sequencing of all coding exons of the gene |  |  | 1.3 |
| **STAT3** | AUTOIMMUNE DISEASE, MULTISYSTEM, INFANTILE-ONSET, 1; ADMIO1 | Deletion and duplication analysis | Sequencing of all coding exons of the gene |  | 2.3 |
| **STAT3** | HYPER-IgE RECURRENT INFECTION SYNDROME, AUTOSOMAL DOMINANT | Deletion and duplication analysis | Sequencing of all coding exons of the gene |  | 2.3 |
| **STK11** | Peutz-Jeghers syndrome | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 1.3 |
| **STUB1** | Spinocerebellar ataxia, autosomal recessive 16 | Sequencing of all coding exons of the gene |  |  | 0.9 |
| **STX16** | Pseudohypoparathyroidism Ib | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1 |
| **STXBP1** | Epileptic Encephalopathy, Early Infantile, 4 | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 1.8 |
| **SUFU** | Basal cell nevus syndrome | Sequencing of all coding exons of the gene |  |  | 1.5 |
| **SUFU** | Medulloblastoma, desmoplastic | Sequencing of all coding exons of the gene |  |  | 1.5 |
| **SUOX** | Sulfite oxidase deficiency | Sequencing of all coding exons of the gene |  |  | 1.6 |
| **SYN1** | Epilepsy, X-linked, with variable learning disabilities and behavior disorders | Sequencing of all coding exons of the gene |  |  | 2.1 |
| **SYNGAP1** | Mental retardation, autosomal dominant 5 | Sequencing of all coding exons of the gene |  |  | 4 |
| **TARDBP** | Cardiac Diseases | Sequencing of all coding exons of the gene |  |  | 1.2 |
| **TBK1** | Frontotemporal dementia and/or amyotrophic lateral sclerosis 4 | Sequencing of all coding exons of the gene |  |  | 2.2 |
| **TBP** | Spinocerebellar Ataxia 17 | Repeat expansion analysis |  |  | 1 |
| **TCF4** | Pitt-Hopkins Syndrome | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 2.3 |
| **TCOF1** | Treacher Collins Syndrome | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 4.5 |
| **TECTA** | Deafness, autosomal dominant 8/12 | Sequencing of all coding exons of the gene |  |  | 6.5 |
| **TECTA** | Deafness, autosomal recessive 21 | Sequencing of all coding exons of the gene |  |  | 6.5 |
| **TEK** | Venous malformations, multiple cutaneous and mucosal | Sequencing of all coding exons of the gene |  |  | 3.4 |
| **TG** | Thyroid dyshormonogenesis 3 | Sequencing of all coding exons of the gene |  |  | 8.3 |
| **TGIF1** | Holoprosencephaly | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.2 |
| **TH** | Dopa-responsive Dystonia THD | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.6 |
| **THAP1** | Primary Dystonia DYT6 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 0.6 |
| **THRB** | THYROID HORMONE RESISTANCE, GENERALIZED, AUTOSOMAL DOMINANT; GRTH | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **THRB** | THYROID HORMONE RESISTANCE, GENERALIZED, AUTOSOMAL RECESSIVE; GRTH | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **THRB** | THYROID HORMONE RESISTANCE, SELECTIVE PITUITARY; PRTH | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **TIA1** | Welander distal myopathy | Sequencing of all coding exons of the gene |  |  | 1.2 |
| **TIMM8A** | Deafness Optic Atrophy Syndrome | Sequencing of all coding exons of the gene |  |  | 0.3 |
| **TIMM8A** | Deafness-Dystonia-Syndrome Mohr-Tranebjaerg-Syndrome | Sequencing of all coding exons of the gene |  |  | 0.3 |
| **TK2** | Mitochondrial DNA depletion syndrome 2 (myopathic type) | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 0.8 |
| **TMEM106B** | Frontotemporal Dementia | Sequencing of all coding exons of the gene |  |  | 0.8 |
| **TMEM126A** | Optic Atrophy Type 7 | Sequencing of all coding exons of the gene |  |  | 0.6 |
| **TMEM165** | CDG-Syndrome 2K | Sequencing of all coding exons of the gene |  |  | 1 |
| **TMEM43** | Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy | Sequencing of all coding exons of the gene |  |  | 1.2 |
| **TNFRSF1A** | Familial autosomal dominant periodic fever (TRAPS; mutation in exons 2, 3, 4, and 6 of the TNFRSF1A gene) | Sequencing of hotspots | Sequencing of all coding exons of the gene |  | 1.4 |
| **TNFRSF1A** | Periodic fever, familial | Sequencing of hotspots | Sequencing of all coding exons of the gene |  | 1.4 |
| **TNNT2** | Cardiomyopathy, familial hypertrophic, 2 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 0.9 |
| **TNNT2** | Cardiomyopathy, familial restrictive, 3 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 0.9 |
| **TNNT2** | Dilated Cardiomyopathy | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 0.9 |
| **TNNT2** | Dilated Cardiomyopathy | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 0.9 |
| **TOMM40** | Alzheimer Disease, late-onset | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **TOR1A** | Primary Dystonia DYT1 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1 |
| **TPM2** | Nemaline Myopathy | Sequencing of all coding exons of the gene |  |  | 0.9 |
| **TPP1** | Neuronal Ceroid-Lipofuscinoses | Sequencing of all coding exons of the gene |  |  | 1.7 |
| **TRAPPC10** | Holoprosencephaly | Sequencing of all coding exons of the gene |  |  | 3.8 |
| **TRAPPC2** | Spondyloepiphyseal Dysplasia | Sequencing of all coding exons of the gene |  |  | 0.5 |
| **TREM2** | Nasu-Hakola disease | Sequencing of all coding exons of the gene |  |  | 0.7 |
| **TREX1** | Aicardi-Goutieres syndrome 1, dominant and recessive | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **TREX1** | Chilblain lupus | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **TREX1** | Vasculopathy, retinal, with cerebral leukodystrophy | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **TRPM1** | Congenital Stationary Night-Blindness | Sequencing of all coding exons of the gene |  |  | 4.9 |
| **TRPS1** | Langer-Giedion Syndrome | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 3.9 |
| **TRPS1** | Trichorhinophalangeal Syndrome Type I | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 3.9 |
| **TRPS1** | Trichorhinophalangeal Syndrome Type III | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 3.9 |
| **TRPV4** | Charcot-Marie-Tooth Neuropathy Type 2C | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.6 |
| **TRPV4** | Hereditary motor and sensory Neuropathy Type 2C | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.6 |
| **TSC1** | Tuberous Sclerosis 1 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 3.5 |
| **TSC2** | Tuberous Sclerosis 2 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 5.4 |
| **TSEN54** | Pontocerebellar Hypoplasia Type 2A | Sequencing of hotspots | Sequencing of all coding exons of the gene |  | 1.6 |
| **TSHB** | HYPOTHYROIDISM, CONGENITAL, NONGOITROUS, 4; CHNG4 | Sequencing of all coding exons of the gene |  |  | 0.4 |
| **TSHR** | Hyperthyroidism, familial gestational | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.3 |
| **TSHR** | Hyperthyroidism, nonautoimmune | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.3 |
| **TSHR** | Hypothyroidism, congenital, nongoitrous, 1 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.3 |
| **TSPAN12** | Familial Exudative Vitreoretinopathy | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 0.9 |
| **TTC8** | Bardet Biedl Syndrome | Sequencing of all coding exons of the gene |  |  | 1.5 |
| **TTC8** | Retinitis Pigmentosa | Sequencing of all coding exons of the gene |  |  | 1.5 |
| **TTR** | Hereditary transthyretin-related Amyloidosis | Sequencing of all coding exons of the gene |  |  | 0.4 |
| **TUBA1A** | Lissencephaly | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **TUBA8** | Polymicrogyria with Optic Nerve Hypoplasia | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **TUBB2B** | Polymicrogyria, Asymmetric | Sequencing of all coding exons of the gene |  |  | 1.3 |
| **TUBB3** | Congenital Fibrosis of the Extraocular Muscles | Sequencing of all coding exons of the gene |  |  | 1.4 |
| **TUBB4A** | Dystonia 4, torsion, autosomal dominant | Sequencing of all coding exons of the gene |  |  | 1.3 |
| **TUBB4A** | Leukodystrophy, hypomyelinating, 6 | Sequencing of all coding exons of the gene |  |  | 1.3 |
| **TWIST1** | Saethre-Chotzen Syndrome | Sequencing of all coding exons of the gene |  |  | 0.6 |
| **TWNK** | Progressive external  Ophthalmoplegia | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.1 |
| **TYR** | Oculocutaneous Albinism Type 1 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.6 |
| **TYROBP** | Nasu-Hakola disease | Sequencing of all coding exons of the gene |  |  | 0.3 |
| **TYRP1** | Albinism, oculocutaneous, type III | Sequencing of all coding exons of the gene |  |  | 1.6 |
| **UBE3A** | Angelman Syndrome | Sequencing of all coding exons of the gene |  |  | 2.6 |
| **UBQLN2** | Frontotemporal dementia and/or amyotrophic lateral sclerosis | Sequencing of all coding exons of the gene |  |  | 1.9 |
| **UNC13D** | Hemophagocytic lymphohistiocytosis, familial, 3 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 3.3 |
| **UPK3A** | Congenital anomalies of kidney and urinary tract, susceptibility to; CAKUT | Sequencing of all coding exons of the gene |  |  | 0.9 |
| **UPK3A** | Possible Association with Congenital Anomalies of the Kidney and Urogenital Tract | Sequencing of all coding exons of the gene |  |  | 0.9 |
| **UPK3A** | Renal adysplasia | Sequencing of all coding exons of the gene |  |  | 0.9 |
| **USH2A** | Usher Syndrome Type 2A | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 15.6 |
| **USP8** | Possible association with spastic paraplegia | Sequencing of all coding exons of the gene |  |  | 3.4 |
| **USP8** | Spastic Paraplegia 59 | Sequencing of all coding exons of the gene |  |  | 3.4 |
| **VAPB** | Cardiac Diseases | Sequencing of all coding exons of the gene |  |  | 0.7 |
| **VCAN** | Wagner Syndrome | Sequencing of all coding exons of the gene |  |  | 10.2 |
| **VCP** | Cardiac Diseases | Sequencing of all coding exons of the gene |  |  | 2.4 |
| **VHL** | Familial Erythrocytosis 2 | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 0.6 |
| **VHL** | Von Hippel-Lindau Syndrome | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 0.6 |
| **VLDLR** | Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.6 |
| **VPS13A** | Chorea-acanthocytosis | Sequencing of all coding exons of the gene |  |  | 9.5 |
| **VPS13B** | Cohen Syndrome | Sequencing of all coding exons of the gene; Deletion and duplication analysis |  |  | 12.1 |
| **VPS35** | Parkinson disease 17 | Sequencing of all coding exons of the gene |  |  | 2.4 |
| **VRK1** | Pontocerebellar Hypoplasia Type 1 | Sequencing of all coding exons of the gene |  |  | 1.2 |
| **WDR35** | Cranioectodermal dysplasia 2 | Sequencing of all coding exons of the gene |  |  | 3.5 |
| **WDR35** | Short-rib thoracic dysplasia 7 with or without polydactyly | Sequencing of all coding exons of the gene |  |  | 3.5 |
| **WDR45** | Neurodegeneration with brain iron accumulation (NBIA) | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.1 |
| **WDR62** | Microcephaly 2, primary, autosomal recessive, with or without cortical malformations | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 4.6 |
| **WDR73** | Galloway-Mowat syndrome | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **WDR81** | Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2 | Sequencing of all coding exons of the gene |  |  | 5.8 |
| **WFS1** | Wolfram Syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 2.7 |
| **WHSC1** | Wolf-Hirschhorn Syndrome | Sequencing of all coding exons of the gene |  |  | 4.1 |
| **WISP3** | Progressive Pseudorheumatoid Arthropathy of Childhood | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **WNT4** | Mullerian aplasia and hyperandrogenism | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **WNT4** | SERKAL syndrome | Sequencing of all coding exons of the gene |  |  | 1.1 |
| **XIAP** | Lymphoproliferative syndrome, X-linked, 2 | Sequencing of all coding exons of the gene |  |  | 1.5 |
| **XK** | McLeod Neuroacanthocytosis Syndrome | Sequencing of all coding exons of the gene |  |  | 1.3 |
| **XPR1** | Basal ganglia calcification, idiopathic, 6 | Sequencing of all coding exons of the gene |  |  | 2.1 |
| **XYLT1** | Desbuquois dysplasia 2 | Sequencing of all coding exons of the gene |  |  | 2.9 |
| **ZC4H2** | Wieacker-Wolf syndrome | Sequencing of all coding exons of the gene |  |  | 0.7 |
| **ZEB2** | Mowat Wilson Syndrome | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 3.6 |
| **ZFPM2** | Tetralogy of Fallot | Sequencing of all coding exons of the gene |  |  | 3.5 |
| **ZFYVE26** | Spastic Paraplegia 15 | Sequencing of all coding exons of the gene |  |  | 7.6 |
| **ZIC1** | Craniosynostosis 6 | Sequencing of all coding exons of the gene | Deletion and duplication analysis |  | 1.3 |
| **ZIC2** | Holoprosencephaly | Deletion and duplication analysis; Sequencing of all coding exons of the gene |  |  | 1.6 |
| **ZIC4** | Dany Walker Malformation | Sequencing of all coding exons of the gene |  |  | 1.2 |