Billing code	Indication/test
565471-565482	Aarskog-Scott syndrome
565390-565401	Abacavir toxicity (HLA-B*57:01 genotyping) - Pharmacogenetics
565390-565401	ABCB1 genotyping (c.3435C>T, c.1199G>A) - Transport protein - Pharmacogenetics
565456-565460	Achondrogenesis / Kniest dysplasia / Hypochondrogenesis
565390-565401	Achondroplasia (FGFR3 hot spot mutation - p.Gly380)
565471-565482	Achromatopsia
565456-565460	Acrocapitofemoral dysplasia / Brachydactyly, type A1
565471-565482	ACTH-independent macronodular adrenal hyperplasia 2 / Cushing syndrome
565493-565504	Adams-Oliver syndrome (gene panel)
565552-565563	Adenomatous polyposis, familial (gene panel)
565471-565482	Adrenogenital syndrome
565471-565482	Adrenoleukodystrophy, X-linked
565471-565482	Agammaglobulinemia
565456-565460	Agnathia-otocephaly complex
565471-565482	Alagille syndrome
565471-565482	Albright hereditary osteodystrophy
565456-565460	Algrove syndrome (Triple A syndrome)
565456-565460	Alpha Tryptasemia, hereditary
565390-565401	Alpha-1-antitrypsin deficiency (hot spot mutations)
565471-565482	alpha-globin hemoglobinopathies
565471-565482	Alport autosomal recessive and X-linked and hematuria
565471-565482	Alzheimer disease (gene panel)
565390-565401	Alzheimer Disease, late onset (AD2) / ApoE2, E3, and E4 isoforms
565456-565460	Amyloidosis
565456-565460	Amyotrophic lateral sclerosis (GGGGCC repeat expansion in the C9ORF72 gene)
565493-565504	Amyotrophic Lateral Sclerosis (ALS) (extended gene panel)
565456-565460	Amyotrophic lateral sclerosis (gene panel)
565471-565482	Androgen insensitivity (AR gene)
565493-565504	Aneurysm, Thoracic Aortic, familial (gene panel)
565456-565460	Angelman / Prader Willi Syndrome
565390-565401	Angioedema type III, hereditary (F12 gene - hot spot mutations - p.Thr328Lys; p. Thr328Arg)
565471-565482	Angioedema, hereditary (gene panel)
565471-565482	Aniridia
565471-565482	Anterior segment dysgenesis
565456-565460	Antithrombine III deficiency (thrombophilia) (SERPINC1 gene)
565471-565482	Arrhytmogenic cardiopathy
565456-565460	Arterial Tortuosity Syndrome
565471-565482	Arteriovenous malformation (gene panel)
565493-565504	Ataxia (gene panel)
565530-565541	Ataxia telangiectasia
565493-565504	Atypical Hemolytic Uremic Syndrome (aHUS) (gene panel)
565493-565504	Autism (gene panel)
565471-565482	Autoimmune disease, multisystem, infantile-onset (ADMIO) / Hyper-IgE recurrent infection syndrome
565456-565460	Autoimmune lymphoproliferative syndrome
565471-565482	Autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy (APECED)
565390-565401	Azoo-/oligozoospermia (microdeletion of 3 regions of Y-chromosome AZFa, b and c)
565530-565541	BAP1-related tumor predisposition syndrome (TPDS)
565456-565460	Becker muscular dystrophy / Duchenne muscular dystrophy (deletion/duplication DMD gene)
565471-565482	Becker muscular dystrophy / Duchenne muscular dystrophy (Full sequencing DMD gene)
565390-565401	Becker nevus
565456-565460	Beckwith-Wiedemann syndrome
565471-565482	Beta-globin hemoglobinopathies
565390-565401	Beta-globin hemoglobinopathies (Hot-spot mutations : Sickle cell disease (HBS), hemoglobin C, hemoglobin E or Hemoglobin D)
565471-565482	Bethlem myopathy / Ullrich congenital muscular dystrophy / Myosclerosis Myopathy
565471-565482	Bicuspid aortic valve
565471-565482	Bile Acid Primary Malabsorption
565471-565482	Bile Acid Synthesis Congenital Defect (gene panel)
565530-565541	Birt-Hogg-Dubé syndrome
565471-565482	Blepharophimosis type I /II
565530-565541	Bloom syndrome

565552-565563	Bone marrow failures syndromes with or without organ dysfunction ,inherited
565493-565504	Brain malformations (gene panel)
565552-565563	Breast and Ovarian Cancer, HBOC, Familial (gene panel)
565515-565526	Breast Cancer Trial
565390-565401	Breast cancer, hereditary / Li-Fraumeni syndrome (Hot spot mutation - 1100delC)
565515-565526	Breast or Pancreatic or Prostate Cancer, metastatic
565471-565482	Bronchiectasis
565456-565460	Brugada syndrome
565471-565482	Buschke-Ollendorff / Melorheostosis with Osteopoikilosis
565471-565482	Butyrylcholinesterase deficiency - Pharmacogenetics
565471-565482	Cadasil (exons of EGFL domains (2 - 24))
565390-565401	Canavan disease (hot spot mutation - p.Glu285Ala, p.Tyr231*)
565471-565482	Candidiasis, familial 7 / Immunodeficiency 31A (AD) / Immunodefyciency 31B (AR)
565456-565460	CANVAS disease - repeat in RFC1 gene
565471-565482	Capillary malformation – microcephaly
565493-565504	Cardiac arrhytmia, inherited (gene panel)
565471-565482	Cardiofaciocutaneous syndrome (gene panel)
565471-565482	Cardiomyopathy, dilated
565471-565482	Cardiomyopathy, hypertrophic
565493-565504	Cardiomyopathy: hypertrophic cardiomyopathy, dilated cardiomyopathy, restrictive cardiomyopathy, left ventricular non-compaction cardiomyopathy, arrhythmogenic right ventricular cardiomyopathy (gene panel)
565493-565504	Cardiopathies, hereditary (gene panel)
565530-565541	Carney syndrome
565471-565482	Carnitine Palmitoyl transferase type II
565471-565482	Caroli Disease
565493-565504	Cataract (gene panel)
565456-565460	Cataract, juvenile with microcornea and glucosuria
565471-565482	Catecholaminergic polymorphic ventricular tachycardia (CPVT)
565471-565482	Central Precocious Puberty (5 genes)
565471-565482	Cerebral cavernous malformation (gene panel)
565456-565460	Cerebral folate transport deficiency (2 genes)
565493-565504	Cerebral palsy (gene panel)
565493-565504	Charcot-Marie-Tooth (other than type 1A) (gene panel, IPN panel)
565456-565460	Charcot-Marie-Tooth type 1A / Hereditary Neuropathy with Liability to Pressure Palsies
565471-565482	CHARGE syndrome
565493-565504	Child Interstitial Lung Disease (child - gene panel)
565471-565482	Cholelithiasis, Low Phospholipid associated (LPAC syndrome)
565493-565504	Cholestasis (gene panel)
565471-565482	Cholestasis, progressive familial intrahepatic (gene panel)
565471-565482	Choroideremia
565471-565482	Chronic granulomatous disease, X-linked
565493-565504	Chronic progressive external ophthalmoplegia (CPEO) (Full sequencing of mtDNA genome)
565493-565504	Ciliopathy / polycystic kidney and liver diseases / ADTKD/ nephronophtisis / Bardet-Biedl syndromes and kidney cancers (gene panel)
565493-565504	cleft lip with/whitout cleft palate (virtual gene panel)
565456-565460	Clouston syndrome
565471-565482	Coagulopathies
565390-565401	Coeliac disease (HLA-DQ2, HLA-DQ8) - Pharmacogenetics
565552-565563	Colon carcinoma (hereditary/familial) (gene panel)
565552-565563	Colorectal cancer, hereditary (gene panel)
565456-565460	Combined immunodeficiency (severe), X-linked
565456-565460	Combined pituitary hormone deficiency 1 (CPHD - POU1F1 gene)
565456-565460	Combined pituitary hormone deficiency 2 (CPHD - PROP1 gene)
565471-565482	Congenital Central Hypoventilation Syndrome / Ondine syndrome
565471-565482	Congenital disorder of glycosylation (small panel)
565493-565504	Congenital disorders of glycosylation (extended panel)
565456-565460	Congenital generalized lipodystrophy type 1
565390-565401	Congenital generalized lipodystrophy type 2 / Spastic paraplegia-17 / Hereditary motor neuronopathy type VA / Silver spastic paraplegia syndrome (hot spot mutation - p.Asn88Ser; p.Ser90; p.Arg96His)
565493-565504	Congenital malformation gene panel
565471-565482	Congenital myotonia (Becker-Thomsen disease) (CLCN1 gene)
565552-565563	Constitutional Mismatch Repair Deficiency Syndrome

565456-565460	Contractural arachnodactyly (Arthrogryposis Distal Type 9 / Beals-Hecht syndrome), congenital
565493-565504	Corneal dystrophy (gene panel)
565456-565460	Costello Syndrome- Schimmelpenning syndrome
565530-565541	Cowden disease / PTEN hamartoma tumor syndrome
565493-565504	Craniosynostosis (gene panel)
565390-565401	Craniosynostosis / Apert syndrome (hot spot mutations - exon 7)
565390-565401	Craniosynostosis / Crouzon syndrome (hot spot mutation - exon 9)
565390-565401	Craniosynostosis / Muenke syndrome (hot spot mutation - p.Pro250Arg)
565456-565460	Craniosynostosis syndromes (Apert, Crouzon)
565471-565482	Creatine deficiency by Guanidinoacetate methyltransferase deficiency
565471-565482	Creatine deficiency,X linked
565471-565482	Crigler Najjar Syndrome
565456-565460	Crisponi syndrome
565471-565482	Currarino syndrome
565471-565482	Cutis Laxa / Geroderma osteodysplasticum (gene panel)
565530-565541	Cylindromatosis
565390-565401	CYP2B6*6,*11,*18 genotyping - drug metabolism - Pharmacogenetics
565390-565401	CYP2C19*2,*3,*17 genotyping - drug metabolism - Pharmacogenetics
565390-565401	CYP2C9*2,*3 genotyping - drug metabolism - Pharmacogenetics
565456-565460	CYP2D6 genotyping (full gene sequencing + pseudogene and CNV analysis)- drug metabolism - Pharmacogenetics
565390-565401	CYP3A4*22 genotyping - drug metabolism - Pharmacogenetics
565390-565401	CYP3A5*3 genotyping - drug metabolism - Pharmacogenetics
565471-565482	Cystic Fibrosis (complete CFTR gene)
565353-565364	Cystic Fibrosis / Congenital absence of the vas deferens / CFTR-related disorders (50 hot spot mutations)
565471-565482	Deficiency of Vitamin K-Dependent Clotting Factors
565493-565504	Dementia, young onset (gene panel)
565456-565460	Dentatorubral pallidoluysian atrophy - ATN1 gene CAG repeat expansion
565493-565504	Dermatogenetic panel, severe, rare and hereditary genodermatoses (gene panel)
565493-565504	Developmental disorders: intellectual disability and multiple congenital anomalies (gene panel)
565493-565504	Diabetes neonatal / Maturity onset Diabete of the Young (MODY) / Hyperinsulinism (gene panel)
565390-565401	Diabetes, mitochondrial (hot spot mutation - m.3243A>G, MTTL1 (tRNA-Leu))
565530-565541	Dicer1 tumor predisposition syndrome
565456-565460	Dihydropyrimidine dehydrogenase deficiency (5-fluorouracil (5-FU) toxicity) - DPYD complete gene, Pharmacogenetics
565390-565401	Dihydropyrimidine dehydrogenase deficiency (5-fluorouracil (5-FU) toxicity) - Pharmacogenetics
565493-565504	Disorders of sex development - Primary Ovarian insufficiency - Hypogonadotropic Hypogonadism (gene panel)
565471-565482	Duane-radial ray syndrome
565390-565401	Dysautonomia, familial (FD) (hot spot mutation - c.2204+6T>C)
565493-565504	Dyskeratosis Congenita (gene panel)
565471-565482	Dyslipidemia (gene panel)
565493-565504	Dyslipidemia (gene panel)
565493-565504	Dystonia (gene panel)
565456-565460	Ectodermal dysplasia
565471-565482	Ectopia lentis
565471-565482	Ectrodactyly / cleft lip/palate syndrome type 3 / Ectodermal dysplasia
565456-565460	Ehlers-Danlos Syndrome, vascular type (type IV)
565493-565504	Ehlers-Danlos syndroom, EDS (gene panel)
565471-565482	Ellis-van Creveld syndrome
565471-565482	Emberger syndrome / Immunodeficiency 21
565493-565504	Endocrine Disorders - Hypothyroidism (gene panel)
565552-565563	Endometrial cancer (gene panel)
565456-565460	Enhanced S-Cone Syndrome
565574-565585	Enzymatic dosage Chitotriosidase
565574-565585	Enzymatic dosage Fabry disease
565574-565585	Enzymatic dosage Gaucher disease
565574-565585	Enzymatic dosage MPS1/Hurler syndrome
565574-565585	Enzymatic dosage Pompe disease
565493-565504	Epidermal nevus syndrome (gene panel)

565493-565504	Epidermolysis bullosa (gene panel)
565493-565504	Epilepsy (gene panel)
565493-565504	Epileptic encephalopathies (gene panel)
565471-565482	Episodic ataxia 2
565530-565541	Erythrocytoses, polycythémies, thrombocytoses et neutropénies congénitales (gene panel)
565471-565482	Fabry disease
565471-565482	Facioscapulohumeral muscular dystrophy 1A (D4Z4 repeat)
565390-565401	Facioscapulohumeral Muscular Dystrophy 2 (hypomethylation D4Z4 repeats)
565390-565401	Factor V- cambridge, liverpool and hong kong variant (hot spot mutations - p.Arg334Thr, p.Arg306)
565552-565563	Familial cancer predisposition (gene panel)
565471-565482	Familial Exudative Vitreoretinopathy, autosomal dominant
565493-565504	Familial hemiplegic Migraine (gene panel)
565471-565482	Familial Mediterranean Fever
565390-565401	Fanconi anemia (FANCC) (hot spot mutation - c.456+4A>T)
565552-565563	Fanconi anemia (gene panel)
565456-565460	Feingold syndrome
565493-565504	Fertilisation failure-oocyte maturation arrest-embryonic arrest (gene panel)
565471-565482	filaggrin gene
565456-565460	Floating Harbor
565471-565482	FMR1-premutation instability
565375-565386	Fragile X syndrome and fragile X-associated disorders (FXTAS, FXPOI)
565456-565460	Friedreich ataxia - GAA repeat expansion
565471-565482	FRMD7-related infantile nystagmus / Nystagmus, infantile periodic alternating, X-linked
565471-565482	Frontotemporal lobar degeneration/ Amyotrophy Lateral Sclerosis (gene panel)
565456-565460	Fructosemia (ALDOB gene)
565456-565460	FSHR - Ovarian Hyperstimulation Syndrome
565471-565482	Fukuyama congenital muscular dystrophy
565552-565563	Gastric Cancer (gene panel)
565530-565541	Gastric cancer, diffuse, hereditary
565530-565541	Gastrointestinal stromal tumor
565456-565460	Gaucher disease diagnostic (GBA gene hot spot mutations - p.Asn409Ser; p.Leu483Pro; c.84dupG; c.115+1G>A;)
565471-565482	Gaucher disease diagnostic (GBA gene sequencing)
565471-565482	Generalized Arterial Calcification of Infancy
565493-565504	Genetic disorders of Calcium and Phosphate metabolism (gene panel)
565530-565541	Germline analysis of BRCA1/2 for iPARP treatment
565390-565401	Gilbert disease / Irinotecan sensitivity / Raltegravir toxicity - Pharmacogenetics
565493-565504	Glaucoma (gene panel)
565471-565482	Globozoospermia (DPY19L2 gene)
565471-565482	Glycogen storage disease type 0
565456-565460	Glycogen storage disease type 1a
565471-565482	Glycogen storage disease type 9
565456-565460	GM2-gangliosidosis / Tay-Sachs syndrome diagnostic (HEXA gene hot spot mutations - c.1274_1277dupTATC, c.1421+1G>C and c.805G>A (p.Gly269Ser))
565530-565541	Gorlin syndrome (gene panel)
565471-565482	Hallervorden-Spatz disease (Neurodegeneration with brain iron accumulation type 1) / HARP syndrome (Hypoprebetalipoproteinemia, Acanthocytosis, Retinitis pigmentosa, and Pallidal degeneration)
565456-565460	Hearing loss, Recessive nonsyndromic hearing loss and deafness DFNB (2 genes)
565493-565504	Hearing loss (deafness), (gene panel)
565456-565460	Hearing loss , Frequent hearing deficiency (1st tier)
565456-565460	Hearing loss, Autosomal dominant non-syndromic sensorineural deafness type DFNA9 (COCH partial sequencing)
565471-565482	Hearing loss, Deafness, autosomal dominant 6/14 / Wolfram syndrome
565456-565460	Hearing loss, Deafness, autosomal recessive 1A
565471-565482	Hearing loss, Deafness, X-linked
565471-565482	Hearing loss, STRC gene
565493-565504	Heart defects, structural, congenital (gene panel)
565456-565460	Hemangioma, congenital
565493-565504	Hemochromatosis (gene panel)
565316-565320	Hemochromatosis hereditary type 1 (HFE gene - hot spot mutations - p.Cys282Tyr; p.His63Asp)
565456-565460	Hemochromatosis hereditary type 4 (SLC40A1 gene)

565471-565482	Hemochromatosis hereditary type 2 to type 5 (gene panel)
565456-565460	Hemochromatosis, juvenile (HJV and HAMP genes)
565471-565482	Hemophilia A
565456-565460	Hemophilia A (inversions)
565471-565482	Hemophilia B
565493-565504	Hepatology (gene panel)
565493-565504	Hepatorenal disorders (gene panel)
565471-565482	Hereditary angioneurotic edema (2 genes)
565552-565563	Hereditary Breast and Ovarian Cancer, HBOC (13 genes)
565552-565563	Hereditary cancer (Breast, ovary, colon) (26 genes)
565552-565563	Hereditary cancer (gene panel)
565552-565563	Hereditary cancer panel (gene panel)
565493-565504	Hereditary Hemolytic Anemias due to unknown or doubtful origin (gene panel - 52 genes)
565456-565460	Hereditary Neuropathy with Liability to Pressure Palsies (HNPP)
565552-565563	Hereditary nonpolyposis colorectal cancer / Lynch syndrome (gene panel)
565552-565563	Hereditary Polyposis Panel (gene panel)
565471-565482	Hirschsprung disease
565331-565342	Homocystinuria (hot spot mutation - c.1298A>C)
565331-565342	Homocystinuria (hot spot mutation - c.677C>T)
565456-565460	Huntington disease - CAG repeat expansion
565471-565482	Hydrocephalia, x linked / CRASH (corpus callosum hypoplasia, retardation, adducted thumbs, spastic paraplegia, and hydrocephalus) syndrome (L1CAM gene)
565471-565482	Hypercholanemia, familial (gene panel)
565471-565482	Hypercholesterolemia, Familial (Gene Panel)
565471-565482	Hyperekplexia (gene panel)
565493-565504	Hyperinsulinism (gene panel)
565515-565526	Hypermethylation promoter MLH1
565515-565526	Hypermethylation promoter MLH1 and p.V600 of BRAF1
565471-565482	Hyperoxaluria
565530-565541	Hyperparathyroidism (gene panel)
565456-565460	Hyperthyroidism (familial gestational or familial nonautoimmune, hypothyroidism, thyrotropin) - TSHR
565471-565482	Hypocalciuric hypercalcemia, familial type I or Hypocalcemia or Hypoparathyroidism, familial isolated (CASR gene)
565456-565460	Hypocalciuric hypercalcemia, familial type II
565456-565460	Hypocalciuric hypercalcemia, familial type III
565471-565482	Hypocalciuric Hypercalcemia, Neonatal Severe Hyperparathyroidism, Hypocalcemia
565471-565482	Hypochondroplasia (full sequencing)
565390-565401	Hypochondroplasia (hot spot mutation - p.Asn540Lys)
565456-565460	Hypochondroplasia (hot spot mutations - p.Asn540; p.Ile538; p.Lys650 FGFR3)
565456-565460	Hypogonadism. Male Infertility (LHB gene)
565493-565504	Hypogonadotropic hypogonadism (gene panel)
565390-565401	Hypokalemic periodic paralysis, type 1 (CACNA1S gene hot spot mutations)
565471-565482	Hypoparathyroidism sensorineural deafness and renal disease
565471-565482	Hypophosphatasia
565471-565482	Hypophosphatemic rickets
565471-565482	Hypoplastic pancreas-intestinal atresia-hypoplastic gallbladder (Martinez-Frias) syndrome
565493-565504	Ichthyosis (gene panel)
565456-565460	Immune deficiency with hyperIgM, type 3
565471-565482	Immunodeficiency - Activated PI3K-delta syndrome
565471-565482	Immunodeficiency 30 / Susceptibility to mycobacterial diseases due to complete IL12RB1 deficiency
565471-565482	Immunodysregulation - polyendocrinopathy - enteropathy - X-linked (IPEX) syndrome
565456-565460	Incontinentia pigmenti (IKBKG gene)
565471-565482	Infertility due to oocyte maturation disorders (OMD), preimplantation embryonic lethality (PREMBL), oocyte/zygote/embryo maturation arrest (OZEMA) and/or total fertilization failure (TFF) (gene panel)
565493-565504	Infertility due to sperm defects (gene panel)
565493-565504	Intellectual Disability (gene panel)
565456-565460	Jewish mutation panel (Tay Sachs, Fanconi, Dysautonomia, Canavan) (4 genes; 7 hot spot mutations)
565471-565482	Kabuki syndrome (gene panel)
565471-565482	Kallmann syndrome (ANOS1 gene)
565471-565482	Kallmann syndrome / Hypogonadotropic Hypogonadism (FGFR1 gene)

565456-565460	Kennedy disease (Spinal and bulbar muscular atrophy, SBMA) - CAG repeat expansion
565471-565482	Keratinopathic ichthyosis (epidermolytic ichtyosis, superficial epidermolytic ichthyosis, congenital reticular ichthyosiform erythroderma) (3 genes)
565552-565563	Kidney cancer (renal cell carcinoma) (gene panel)
565493-565504	Kidney Diseases, inherited (Gene Panel)
565493-565504	Leber Congenital Amaurosis - Retinal dystrophy, early onset (gene panel)
565456-565460	Leber hereditary optic neuropathy (LHON) – (DNAJC30 gene)
565456-565460	Leber hereditary optic neuropathy (LHON) (hot spot mutations - p.Ala53Thr in MT-ND1; p.Arg340His in MT-ND4; p.Met64Val in MT-ND6)
565493-565504	Leigh / NARP Syndrome
565456-565460	Leigh syndrome
565530-565541	Leiomyomatosis and renal cell cancer
565471-565482	Leri-Weill dyschondrosteosis / SHOX-related short stature
565493-565504	Leukodystrophy (gene panel)
565456-565460	Leydig cell hypoplasia or Precocious puberty, male-limited
565530-565541	Li-Fraumeni syndrome
565471-565482	Lipodystrophy (2 genes)
565493-565504	Lipodystrophy and/or hyperinsulinism (gene panel)
565471-565482	Lissencephaly (LIS1 gene)
565456-565460	Lissencephaly (Tubulin alpha 1A gene)
565471-565482	Lissencephaly / subcortical band heterotopia
565456-565460	Lissencephaly 3
565390-565401	Long chain 3-hydroxyl-CoA dehydrogenase deficiency (hot spot mutation - p.Glu510Gln)
565471-565482	Long QT syndrome
565471-565482	Lymphoproliferative syndrome, X-linked (SH2D1A gene) / Duncan's disease
565471-565482	Lymphoproliferative syndrome, X-linked (XIAP gene)
565515-565526	Lynch syndrome - MLH1 promoter hypermethylation and BRAF V600E mutation
565530-565541	Lynch-like syndrome
565493-565504	Lysosomal Storage Disease (gene panel)
565456-565460	Macrozoospermia (AURKC gene)
565456-565460	Macular dystrophy
565456-565460	Maffucci syndrome (gene panel)
565456-565460	Male infertility
565493-565504	Malformations of cortical development (gene panel)
565471-565482	Malignant hypertermia
565530-565541	Malignant Mesothelioma (BAP1; CDKN2A genes)
565456-565460	Marfan Syndrome
565456-565460	Maternally inherited deafness
565456-565460	Maternally-inherited diabetes and deafness / Mitochondrial myopathy with reversible cytochrome C oxidase deficiency / mitochondrial tRNA glutamic acid
565471-565482	Maturity onset Diabete of the Young (MODY), type 5 / Renal cysts and diabetes syndrome (gene panel)
565493-565504	Maturity onset Diabete of the Young, MODY (gene panel)
565471-565482	Mc Ardle disease, glycogene storage disease type V
565390-565401	Mc Cune Albright syndrome
565471-565482	Medium chain acyl-CoA dehydrogenase deficiency (MCAD-ACADM gene)
565390-565401	Medium chain acyl-CoA dehydrogenase deficiency (MCAD hot spot mutation - p.Lys329Glu)
565530-565541	Medullary thyroid carcinoma
565530-565541	Medulloblastoma (gene panel)
565552-565563	Melanoma Panel, hereditary (gene panel)
565530-565541	Meningioma (gene panel)
565456-565460	Mental retardation, X-linked, syndromic, Borck type
565493-565504	Metabolic diseases with hepatic disorders (gene panel)
565493-565504	Metabolic disorders including disorders of glycosylation, peroxisomal disorders, organic acidurias, glycogenosis disorders, neurotransmitter disorders (gene panel)
565493-565504	Microphthalmia / Anophthalmia / Coloboma-Anterior Segment Dysgenesis (MAC-ASD) (gene panel)
565456-565460	Microphthalmia, syndromic 5; Retinal dystrophy, early-onset, and pituitary dysfunction
565515-565526	Microsatellite instability analysis
565471-565482	Microspherophakia / Megalocornea / primary congenital Glaucoma / Weill-Marchesani syndrome 3 recessive type
565456-565460	Microtia, hearing impairment, and cleft palate
565456-565460	Mitochondrial complex II deficiency
565456-565460	Mitochondrial complex III deficiency / Bjornstad syndrome / Gracile syndrome

565471-565482	Mitochondrial complex IV deficiency
565456-565460	Mitochondrial complex V (ATP Synthase) deficiency
565493-565504	Mitochondrial disorders (gene panel)
565493-565504	Mitochondrial disorders, mitchondrial DNA based (Full sequencing of mtDNA genome)
565456-565460	Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes (MELAS) (full sequencing)
565390-565401	Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes (MELAS) (hot spot mutation - m.3243A>G)
565493-565504	Mitochondrial NeuroGastroIntestinal Encephalomyopathy (MNGIE) syndrome
565390-565401	Mitochondrial non-syndromic sensorineural deafness with susceptibility to aminoglycoside exposure (hot spot mutation - 1555A-G in MT-RNR1)
565493-565504	Movement Disorders (gene panel)
565493-565504	mtDNA depletion syndrome
565390-565401	MTHFR c.677C>T et c.1298A>C
565456-565460	MUC1-VNTR insertion
565471-565482	Mucolipidosis II and III
565471-565482	Mucopolysaccharidosis (MPS) type I / Hurler-Scheie syndrome
565456-565460	Mucopolysaccharidosis (MPS) type II / Hunter syndrome
565471-565482	Mucopolysaccharidosis (MPS) type IIIA / Sanfilippo A
565471-565482	Mucopolysaccharidosis (MPS) type IIIB, Sanfilippo B
565471-565482	Mucopolysaccharidosis (MPS) type IVA, Morquio A syndrome
565471-565482	Mucopolysaccharidosis (MPS) type IVB, Morquio B syndrome / GM1 gangliosidosis
565471-565482	Mucopolysaccharidosis (MPS) type VII / SLY disease
565456-565460	Mucopolysacharidosis type IIIa (N-sulfoglucosamine sulfohydrolase)
565471-565482	Multiple congenital anomalies-hypotonia-seizures syndrome 1
565530-565541	Multiple endocrine neoplasia (gene panel)
565530-565541	Multiple endocrine neoplasia type 2A and 2B / Familial medullary thyroid carcinoma
565530-565541	Multiple endocrine neoplasia, type 1
565471-565482	Multiple epiphyseal dysplasia
565471-565482	Multiple osteochondromas
565530-565541	Myeloid neoplasms with germline predisposition (Hereditary MDS/Acute Leukemia) (gene panel)
565552-565563	Myeloid/lymphoid neoplasms with germline predisposition
565390-565401	Myhre syndrome (hot spot mutation - p.I500)
565390-565401	Myoadenylate deaminase deficiency (AMPD1 gene hot spot mutation - p.Gln12*)
565456-565460	Myoclonic epilepsy associated with ragged-red fibers (MERFF) (full sequencing) (2nd tier)
565390-565401	Myoclonic epilepsy associated with ragged-red fibers (MERFF) (hot spot mutation - m.8344A>G)
565471-565482	Myoclonic epilepsy of Unverricht and Lundborg / Progressive myoclonus epilepsy /Baltic myoclonic epilepsy
565493-565504	Myopathy (gene panel)
565493-565504	Myopia (early onset high myopia)
565456-565460	Myotonic dystrophy type 2 - CCTG repeat expansion
565471-565482	Nanophthalmos
565390-565401	Narcolepsy (HLA-DQB1*0602 Genotyping) - Pharmacogenetics
565493-565504	Nephrocalcinosis and nephrolithiasis (gene panel)
565493-565504	Nephropathies, hereditary (gene panel)
565471-565482	Nephrotic syndrome - steroid resistant
565493-565504	Nephrotic syndrome, Focal Segmental Glomerulosclerosis (FSGS) , Alport syndrome and podocytopathy (gene panel)
565471-565482	Netherton syndrome (SPINK5 gene)
565456-565460	Neuraminidase deficiency / Sialidosis
565530-565541	Neuroblastoma (2 genes)
565493-565504	Neurodegeneration (gene panel)
565493-565504	Neurodegeneration with Brain Iron Accumulation (gene panel)
565471-565482	Neurodegeneration with iron accumulation in brain 2A & 2B
565493-565504	Neurodevelopmental disorders gene panel
565552-565563	Neuroendocrine tumor (NET) (gene panel)
565552-565563	Neurofibromatosis type 1 / Legius syndrome
565530-565541	Neurofibromatosis type 1 / Legius syndrome (2 genes)
565530-565541	Neurofibromatosis type 2
565530-565541	Neurofibromatosis type I
565493-565504	Neuromuscular disorders (gene panel)
565493-565504	Neuropathy (gene panel)

565390-565401	Neuropathy, Ataxia, and Retinitis Pigmentosa (NARP) syndrome (hot spot mutation - m.8993T>C/G)
565471-565482	Niemann-Pick disease
565530-565541	Nijmegen Breakage Syndrome
565471-565482	Non-cholestatic jaundice with direct bilirubin (gene panel)
565456-565460	Noonan syndrome (Screening PTPN11)
565456-565460	Norrie disease (NDP gene)
565471-565482	Obesity, early onset (gene panel)
565456-565460	Obesity, Monogenic early onset
565493-565504	Obesity (extended gene panel)
565456-565460	Occipital horn syndrome / Distal Spinal Muscular atrophy
565471-565482	Occult macular dystrophy
565493-565504	Ocular albinism and oculocutaneous albinism type 1, 2, 3, 4, 6, 7, 8 (gene panel)
565456-565460	Oculo Dento Digital Dysplasia
565493-565504	Oculocutaneous albinism / Ocular albinism / Isolated nystagmus (gene panel)
565456-565460	Oculopharyngeal Muscular Dystrophy - GCN repeats expansion
565530-565541	Oligodontia-colorectal cancer syndrome
565552-565563	Onco-endocrine pathologies (gene panel)
565471-565482	Opitz G/BBB syndrome, X linked
565493-565504	Optic atrophy (gene panel)
565493-565504	Osteogenesis imperfecta / Osteoporose (gene panel)
565530-565541	Ovarian neoplasms, rare non-epithelial (2 genes)
565471-565482	Overgrowth & vascular anomalies / CLOVES syndrome
565331-565342	Overgrowth & vascular anomalies / Proteus syndrome (c.49G>A (p.Glu17Lys) mutation)
565493-565504	Overgrowth (gene panel)
565552-565563	Pancreatic cancer (gene panel)
565456-565460	Pancreatitis hereditary (PRSS1 gene)
565456-565460	Pancreatitis, hereditary (gene panel-small)
565471-565482	Pancreatitis, hereditary (gene panel)
565530-565541	Paraganglioma-pheochromocytoma (gene panel-small)
565552-565563	Paraganglioma-pheochromocytoma (gene panel)
565552-565563	Parathyroid tumor (gene panel)
565530-565541	Parkinson (gene panel)
565493-565504	Paroxysmal Episodic Disorders (gene panel)
565456-565460	Pearson Syndrome
565493-565504	Pediatric oncopredisposition (gene panel)
565471-565482	Pendred syndrome
565493-565504	Periodic Fever (gene panel)
565471-565482	Periodic paralysis (myotonia) / Paramyotonia congenita (SCN4A gene)
565493-565504	Peripheral neuropathy (gene panel)
565493-565504	Perrault syndrome (gene panel)
565530-565541	Peutz-Jeghers Syndrome
565456-565460	Pharmacogenes analysis
565471-565482	Phenylketonuria
565530-565541	Pituitary adenoma (gene panel)
565471-565482	Polyarteritis nodosa, childhood-onset / Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome
565493-565504	Polycystic kidney disease type 1 and 2
565456-565460	Polymicrogyria, asymmetric
565471-565482	Polymicrogyria, bilateral perisylvian, frontoparietal
565471-565482	Pompe disease, Glycogen storage disease II (GAA gene)
565471-565482	Porencephaly / Hemorrhagic stroke / Cerebral small vessel disease / Idiopathic cerebral white matter lesions / HANAC / Isolated retinal arteriolar tortuosity
565471-565482	Porphyria (gene panel)
565493-565504	Premature ovarian insufficiency (POI) (gene panel)
565493-565504	Primary Arterial Hypertension (gene panel)
565493-565504	Primary cardiac arrhythmias (Atrial fibrillation / Brugada syndome / Catech. polymorphic ventricular tachycardia / Early repolaristion syndrome / Ideopathic ventricular fibrillation / Long QT syndrome / Sick sinus syndrome / Short QT syndrome) (gene pane)
565493-565504	Primary Ciliary Dyskinesia (gene panel)
565493-565504	Primary Electrical disorders / Brugada syndrome / Long QT syndrome (LQT) / Short QT syndrome (SQT) / Arrhythmogenic right ventricular cardiomyopathy (ARVC) / Catecholaminergic polymorphic ventricular tachycardia (CPVT) (gene panel)
565515-565526	Primary familial erythrocytosis or Primary familial congenital polycythemia
565493-565504	Primary immune deficiencies (gene panel)

565471-565482	Primary immune deficiencies / Autoimmune polyendocrine syndrome type 1
565493-565504	Primary lymphedema / fetal hydrops (gene panel)
565471-565482	Progressive external ophthalmoplegia
565530-565541	Prostate cancer
565552-565563	Prostate cancer (gene panel)
565390-565401	Prostate cancer susceptibility (HOXB13 - hot spot mutation p.(Gly84Glu))
565471-565482	Protein S deficiency
565471-565482	Pseudoxanthoma Elasticum
565456-565460	Pseudoxanthoma Elasticum with clotting deficiency
565530-565541	PTEN hamartoma tumor syndrome
565493-565504	Pulmonary Arterial Hypertension / Rendu Osler Weber disease (gene panel)
565493-565504	Pulmonary Fibrosis (gene panel) + rs35705950 of MUC5B gene
565471-565482	Pyruvate dehydrogenase deficiency / X-linked Leigh syndrome
565493-565504	RASopathy (gene panel)
565456-565460	Recurrent metabolic encephalomyopathic crises-rhabdomyolysis-cardiac arrhythmia-intellectual disability syndrome (TANGO2 gene)
565530-565541	Renal carcinoma (genepanel)
565552-565563	Renal cell carcinoma (kidney cancer) (gene panel)
565471-565482	Renal cysts and diabetes syndrome
565493-565504	Renal disease, ens stage ESRD (gene panel)
565493-565504	Renal or urinary tract malformation (CAKUT) (gene panel)
565471-565482	Rendu-Osler-Weber disease (genepanel)
565390-565401	Resistance to clopidrogel - PON1 genotyping (Q192R) - Pharmacogenetics
565456-565460	Resistance to vitamin K antagonists - VKORC1 sequencing (all exons) - Pharmacogenetics
565493-565504	Respiratory disorders (gene panel): non-CF bronchiectasis; pulmonary hypertension; interstitial lung disease
565390-565401	Response to antiviral treatment in hepatitis C - IL28B genotyping (rs8099917 (T>G) + rs12979860 (C>T)) - Pharmacogenetics
565493-565504	Retinal dystrophy / RETNET (gene panel)
565471-565482	Retinitis pigmentosa, X-Linked
565530-565541	Retinoblastoma
565456-565460	Retinoschisis, XL
565471-565482	Rett syndrome
565530-565541	Rhabdoid Tumor Predisposition Syndrome (SMARCA4; SMARCB1 genes)
565530-565541	Rhabdomyosarcoma
565456-565460	Rokitansky syndrome
565456-565460	Saethre-Chotzen syndrome
565471-565482	Sandhoff disease
565390-565401	Schinzel-Giedion midface retraction syndrome
565530-565541	Schwannoma Predisposition Syndrome (gene panel)
565456-565460	Segawa syndrome (GCH1 gene)
565471-565482	Segawa syndrome (TH gene)
565456-565460	Septo-optic dysplasia
565530-565541	Sessile serrated polyposis syndrome (RNF43 gene)
565390-565401	Sex determining region Y
565456-565460	Sex determining region Y
565493-565504	Short stature/ Growth retardation/ (gene panel)
565456-565460	Silver-Russell syndrome
565456-565460	Silver-Russell syndrome (11p15 methylation)
565493-565504	Skeletal dysplasia (gene panel)
565493-565504	Skin disorders (gene panel)
565390-565401	SLCO1B1*1b,*5,*15 genotyping (transport protein) - Pharmacogenetics
565530-565541	Small cell carcinoma of the ovary
565456-565460	Smith Lemli Opitz
565471-565482	Smith Lemli Opitz
565456-565460	Sorsby fundus dystrophy (TIMP3)
565493-565504	Spastic Paraplegia (gene panel)
565471-565482	Spastic paraplegia-4
565456-565460	Spinal muscular atrophy (SMA) type 1 (Werdnig-Hoffmann), type 2, type 3 (Kugelberg-Welander) and type 4
565456-565460	Spinocerebellar ataxia (SCA) type 17 - CAG/CAA repeat expansion
565471-565482	Spinocerebellar ataxia (SCA) types 1, 2, 3, 6, 7 - CAG repeat expansion
565471-565482	Spinocerebellar ataxia (SCA) types 8, 10, 12, 17 - repeat expansion
565390-565401	Spinocerebellar ataxia (type 13)

565456-565460	Spinocerebellar ataxia (type 8, 17) + Dentatorubral pallidoluysian atrophy - repeat expansion
565456-565460	Spondylo-epiphyseal dysplasia
565471-565482	Stargardt disease
565456-565460	Steinert myotonic dystrophy - CTG repeat expansion
565493-565504	Stickler syndrome (gene panel)
565493-565504	Stroke (gene panel)
565390-565401	Sturge-Weber syndrome (gene panel)
565471-565482	Subcortical band heterotopia, lissencephaly
565456-565460	Supravalvular aortic stenosis
565515-565526	Susceptibility to Cutaneous Malignant Melanoma
565390-565401	Susceptibility to pancreatitis induced by thiopurine immunosuppressants
565456-565460	Swachman-Bodian-Diamond syndrome
565471-565482	Synpolydactyly / Brachydactyly
565390-565401	Tay Sachs disease (hot spot mutations - c.1274_1277dupTATC, c.1421+1G>C and c.805G>A (p.Gly269Ser))
565471-565482	Telangiectasia,hereditary hemorrhagic of Rendu Osler and Weber (gene panel)
565493-565504	Telomeropathies (virtual gene panel)
565456-565460	Temple syndrome / Kagami-Ogata Syndrome
565471-565482	Thalassemia Alpha (2 genes)
565456-565460	Thanatophoric dysplasia (hot spot mutations - p.Arg248 / p.Gly370 / p.Ser371 / p.Tyr373 / p.Lys650 / p.X806 in FGFR3 gene)
565456-565460	Thiamine-responsive encephalopathy
565390-565401	Thiopurine S-Methyltransferase deficiency - TPMT genotyping - TPMT*2/ TPMT*3A/3B/3C - Pharmacogenetics
565493-565504	Thoracic Aortic Aneurysm, familial (gene panel)
565456-565460	Thrombophilia due to protein C deficiency (PROC gene)
565493-565504	Thyroid disgenesis (gene panel)
565456-565460	Thyroid hormone receptor resistance (THRB gene)
565390-565401	Torsion dystonia 1 (DYT1) (hot spot mutation - c.907_909delGAG)
565456-565460	TPMT and NUDT15 sequencing - Pharmacogenetics
565471-565482	TRANSTHYRETIN (TTR) Analysis
565456-565460	Transthyretine amyloïdose
565471-565482	Treacher Collins (gene panel)
565493-565504	Trombosis - Hemostasis (gene panel)
565471-565482	Tuberous sclerosis (2 genes)
565493-565504	Tubulopathy (gene panel)
565390-565401	Uniparental disomy
565493-565504	Usher syndrome (gene panel)
565471-565482	Usher syndrome, type IIA
565471-565482	Vas deferens, congenital bilateral aplasia of, X-linked (ADGRG2 gene)
565471-565482	Vascular malformations (somatic)
565471-565482	Vascular mineralisation
565471-565482	Venous malformation (3 genes)
565390-565401	VEXAS Syndrome
565390-565401	Vitamin K antagonists toxicity or dose selection - VKORC1 genotyping (-1639G>A + 1173C>T) - Pharmacogenetics
565471-565482	Vitelliform Macular Dystrophy
565456-565460	V-maf Musculoaponeurotic fibrosarcoma oncogen homolog B
565530-565541	Von Hippel Lindau syndrome
565493-565504	Von Willebrand disease
565493-565504	Waardenburg syndrome (gene panel)
565471-565482	Waardenburg Syndrome types I and III
565456-565460	WAGR Syndrome
565471-565482	Weill-Marchesani syndrome
565456-565460	WHIM (warts, hypogammaglobulinemia, infections, and myelokathexis) syndrome
565530-565541	Wilms tumor (DICER1; WT1 genes)
565471-565482	Wilson Disease
565471-565482	Wiskott-Aldrich syndrome
565456-565460	X-chromosome inactivation
565390-565401	Zygosity (medical)