Annexe 1 - Bijlage 1

RIZIV code	Full name
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
565493-565504	Achondrogenesis type 2 / Hypochondrogenesis / Spondyloepimetaphyseal dysplasia congenita, Strudwick type / Platyspondylic dysplasia, Torrance type / Stickler syndrome type 1/ Spondyloepiphyseal dysplasia congenita / Czech dysplasia/ Kniest dysplasia
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
565471-565482	Acyl-CoA dehydrogenase 9 deficiency
565493-565504	Adams-Oliver syndrome
565552-565563	Adenomatous Polyposis, familial (gene panel)
565530-565541	Adenomatous Polyposis, familial type 4, Autosomal Recessive
565515-565526	Adenomatous Polyposis, familial, Autosomal Recessive (NTHL1 gene)
565471-565482	Adrenogenital syndrome
565471-565482	Adrenoleukodystrophy (X-linked)
565471-565482	Agammaglobulinemia
565456-565460	Agnathia-otocephaly complex
565493-565504	Alagille syndrome (gene panel)
565471-565482	Alanyl-tRNA synthetase 2, mitochondrial
565471-565482	Albright hereditary osteodystrophy
565456-565460	Algrove syndrome (Triple A syndrome)
565390-565401	Alpha Tryptasemia, Hereditary
565390-565401	Alpha-1-antitrypsin deficiency (2 hot sopt mutations / p.Glu366Lys; p.Glu288Val)
565471-565482	alpha-globin hemoglobinopathies
565471-565482	Alport autosomal recessive and X-linked and hematuria (3 genes)
565471-565482	Alzheimer disease (gene panel)
565390-565401	Alzheimer Disease, late onset (AD2) / ApoE2, E3, and E4 isoforms
565456-565460	Amyloidosis hereditary / Dystransthyretinemic hyperthyroxinemia
565456-565460	Amyloïdosis, TTR gene
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)
565456-565460	Angelman / Prader Willi Syndrome
565471-565482	Angelman syndrome
565390-565401	Angioedema type III (hot spot mutations - p.Thr328Lys; p. Thr328Arg), Hereditary
565471-565482	Angioedema, Hereditary (gene panel)
565471-565482	Aniridia
565471-565482	Anterior segment dysgenesis
565456-565460	Antithrombine III deficiency (thrombophilia) (SERPINC1 gene)
565471-565482	Arterial Calcification of Infancy,generalized
565456-565460	Arterial Tortuosity Syndrome
565493-565504	Arteriovenous malformation (gene panel)
565493-565504	Ataxia (gene panel)
565530-565541	Ataxia telangiectasia
565390-565401	Atopic dermatitis (hot spot mutations - p.Arg501*; c.2282del4)
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-tumor predisposition syndrome
565456-565460	Becker muscular dystrophy / Duchenne muscular dystrophy (deletion/duplication DMD gene)
565471-565482	Becker muscular dystrophy / Duchenne muscular dystrophy (Full sequencing DMD gene)
565456-565460	Beckwith-Wiedemann syndrome
565456-565460	Beta-globin hemoglobinopathies
565471-565482	Beta-globin hemoglobinopathies (full sequencing)
565390-565401	Beta-globin hemoglobinopathies, phenotype modifiers (hot spot mutations - rs7482144 (Xmn1) at promoter 158 bp 5' upstream of HBG2 / 32C-T in the 5' UTR of the HBS1L)
565390-565401	Beta-globin hemoglobinopathies, Sickle cell anemia, Sickle cell disorder (hot spot mutation - p.Glu6Val, p.Glu6Lys)
565471-565482	Bethlem myopathy / Ullrich congenital muscular dystrophy / Myosclerosis Myopathy
565471-565482	Bicuspid aortic valve
565471-565482	Bile Acid Primary Malabsorption
565493-565504	Bile Acid Synthesis Congenital Defect (gene panel)
565530-565541	Birt-Hogg-Dubé syndrome
565471-565482	Blepharophimosis type I /II
565530-565541	Bloom syndrome
565493-565504	Brain malformations

565552-565563	Breast and Ovarian Cancer, HBOC, Familial (gene panel)
565530-565541	Breast cancer, hereditary (5 genes)
565515-565526	Breast cancer, hereditary (CHEK2) (Hot spot mutation - c.1100delC)
565530-565541	Breast cancer, hereditary (PALB2 gene)
565515-565526	Breast cancer, hereditary / Li-Fraumeni syndrome (Hot spot mutation - 1100delC)
565515-565526	Breast or Pancreatic Cancer, metastatic
565493-565504	Bronchiectasis (gene panel)
565456-565460	Brugada syndrome
565493-565504	Brugada syndrome (gene panel)
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 mutations - p.Glu285Ala, p.Tyr231*)
565552-565563	Cancer (Breast, ovary, colon), Hereditary (gene panel)
565552-565563	Cancer, Hereditary (gene panel)
565471-565482	Candidiasis, familial 7 / Immunodeficiency 31A (AD) / Immunodefyciency 31B (AR)
565471-565482	Capillary malformation – microcephaly
565493-565504	Cardiac arrhytmia, inherited (gene panel)
565456-565460	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency
565471-565482	Cardiofaciocutaneous syndrome (5 genes)
565471-565482	Cardiomyopathy, hypertrophic
565493-565504	Cardiomyopathy, Hypertrophic (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 Hypoventilation Syndrome, congenital / Ondine syndrome
565471-565482	Central Precocious Puberty (5 genes)
565471-565482	Cerebral cavernous malformation (3 genes)
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
565456-565460	Charcot-Marie-Tooth, X-linked
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
565471-565482	Choroideremia
565471-565482	Chronic granulomatous disease, X-linked
565493-565504	Chronic progressive external ophthalmoplegia (CPEO) (Full sequencing of mtDNA genome)
565493-565504	Ciliary dyskinesia, primary (PCD) Heterotaxyies (gene panel)
565493-565504	Ciliopathy (gene panel)
565493-565504	Cleft lip and palate
565456-565460	Clouston syndrome
565471-565482	Coagulopathies (2 genes)
565390-565401	Coeliac disease (HLA-DQ2, HLA-DQ8) - Pharmacogenetics
565530-565541	Colon and endometrial cancer susceptibility (POLD1 gene)
565390-565401	Colon cancer susceptibility (POLE and POLD1 gene) (hot spot mutations - p.Leu424Val; p.Ser489Asn)
565552-565563	Colorectal cancer, familial (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	Complement factor H deficiency / Hemolytic uremic syndrome, atypical, susceptibility to, 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)
565456-565460	Contractural arachnodactyly, congenital (Arthrogryposis Distal Type 9 / Beals-Hecht syndrome)
565493-565504	Corneal dystrophy
565456-565460	Costello Syndrome- Schimmelpenning syndrome
565530-565541	Cowden disease / PTEN hamartoma tumor syndrome
565456-565460	COX10 homolog, cytochrome c oxidase assembly protein
565390-565401	Craniosynostosis / Apert syndrome (hot spot mutations - exon 7)
565390-565401	Craniosynostosis / Crouzon syndrome (hot spot mutation - exon 9)
565456-565460	Craniosynostosis Boston type
565456-565460	Craniosynostosis syndromes (Apert, Crouzon)
565390-565401	Craniosynostosis, Muenke syndrome (hot spot mutation - p.Pro250 in FGFR3 gene)
565471-565482	Creatine deficiency by Guanidinoacetate methyltransferase deficiency (2 genes)
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)
565471-565482	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*2,*3,*4,*5,*6,*7,*8,*9,*10,*11,*15,*17,*29,*35,*41 genotyping - drug metabolism - Pharmacogenetics
565390-565401	CYP3A4*22 genotyping - drug metabolism - Pharmacogenetics
565390-565401	CYP3A5*3,(*6) genotyping - drug metabolism - Pharmacogenetics
565353-565364	Cystic Fibrosis / Congenital absence of the vas deferens / CFTR-related disorders (hot spot mutations)
565471-565482	Cystic Fibrosis / Congenital absence of the vas deferens / CFTR-related disorders (Sequencing CFTR gene)
565353-565364	Cystic Fibrosis and related disorders (TG repeat intron 8)
565471-565482	Deficiency of Vitamin K-Dependent Clotting Factors
565493-565504	Dementia, early onset (gene panel)
565456-565460	Dentatorubral pallidoluysian atrophy (DRPLA) - CAG repeat expansion
565493-565504	Dermatogenetic panel, severe, rare and hereditary genodermatoses (394 genes))
565493-565504	Diabetes neonatal / Maturity onset Diabete of the Young (MODY) (gene panel)
565471-565482	Diabetes, Maturity onset Diabete of the Young (MODY) (gene panel)
565390-565401	Diabetes, mitochondrial (hot spot mutation - m.3243A>G, MTTL1 (tRNA-Leu))
565530-565541	Dicer1 tumor predisposition syndrome
565390-565401	Dihydropyrimidine dehydrogenase deficiency/5-fluorouracil toxicity - Pharmacogenetics (4 variants: DPYD*2A, DPYD*13, c.2846A>T, HapB3)
565456-565460	Dihydropyrimidine dehydrogenase deficiency/5-fluorouracil toxicity - Pharmacogenetics (exons 2-6-10-11-13-14-18-19-22 - IVS14+1)
565471-565482	Disorder of glycosylation, congenital (1 tier)
565493-565504	Disorders of glycosylation, congenital (gene panel)
565493-565504	Disorders of sex development - Primary Ovarian insufficiency - Hypogonadotropic Hypogonadism (gene panel)
565471-565482	Dravet syndrome / Severe myoclonic epilepsy of infancy / Generalized epilepsy with febrile seizures plus
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)
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 (2 genes)
565471-565482	Emberger syndrome / Immunodeficiency 21
565493-565504	Endocrine Disorders - Hyper(Hypo)parathyroidism (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
565471-565482	Epidermal nevus syndrome (gene panel)
565493-565504	Epidermolysis bullosa (gene panel)
565493-565504	Epilepsy (gene panel)
565493-565504	Epileptic encephalopathy, early onset (845 genes)
565471-565482	Episodic ataxia 2
565471-565482	Exudative Vitreoretinopathy, familial, autosomaal dominant
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)
565390-565401	Fanconi anemia (FANCC) (hot spot mutation - c.345+4A>T)
565552-565563	Fanconi anemia (gene panel)
565456-565460	Feingold syndrome
565471-565482	Fever, Mediterranean,Familial
565493-565504	Fever, periodic (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
565456-565460	Fructosemia (ALDOB gene)
565456-565460	FSHR - Ovarian Hyperstimulation Syndrome

565471-565482	Fukuyama congenital muscular dystrophy
565552-565563	Gastric Cancer (10 genes)
565530-565541	Gastric cancer, diffuse
565530-565541	Gastric cancer, lobular breast cancer
565530-565541	Gastrointestinal stromal tumor (2 genes)
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	Giant Axonal Neuropathy 1
565390-565401	Gilbert syndrome / Irinotecan sensitivity (homozygous A(TA)7TAA allele)
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
565390-565401	Glycogen storage disease type III (exon 21)
565552-565563	Gorlin syndrome (gene panel)
565530-565541	Gorlin syndrome (PTCH1; SUFU genes)
565471-565482	Hallervorden-Spatz disease (Neurodegeneration with brain iron accumulation type 1) / HARP syndrome (Hypoprebetalipoproteinemia, Acanthocytosis, Retinitis pigmentosa, and Pallidal degeneration)
565493-565504	Hearing loss (deafness) (gene panel)
565471-565482	Hearing loss (Deafness), autosomal dominant 6/14 / Wolfram syndrome
565456-565460	Hearing loss (deafness), autosomal dominant 9 (COCH partial sequencing)
565456-565460	Hearing loss (Deafness), autosomal recessive 1A
565471-565482	Hearing loss (Deafness), X-linked
565456-565460	Hearing loss, Frequent hearing deficiency (1 tier)
565471-565482	Hearing loss, STRC gene
565493-565504	Heart / Cardio disorders (gene panel)
565493-565504	Heart defect,congenital
565456-565460	Hemangioma, congenital
565552-565563	Hematologic Familiar Forms
565493-565504	Hemiplegic Migraine, Familial (gene panel)
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
565471-565482	Hemochromatosis hereditary type 2 to type 5 (gene panel)
565456-565460	Hemochromatosis, juvenile
565493-565504	Hemolytic Anemias due to unknown or doubtful origin, hereditary (gene panel)
565471-565482	Hemophilia A
565456-565460	Hemophilia A (inversions)
565471-565482	Hemophilia B
565456-565460	Hepatic failure, early onset, and neurologic disorder (cytochrome C oxidase deficiency)
565493-565504	Hepatorenal disorders (gene panel)
565471-565482	Hirschsprung disease
565331-565342	Homocystinuria (hot spot mutation)
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)
565493-565504	Hypercholanemia, Familial (genepanel)
565471-565482	Hypercholesterolemia, Familial (gene panel)
565471-565482	Hyperekplexia (gene panel)
565493-565504	Hyperinsulinism (gene panel)
565515-565526	Hypermethylation promoter MLH1/BRAF1
565471-565482	Hyperoxaluria
565471-565482	Hyperparathyroidism (gene panel)
565456-565460	Hyperthyroidism (familial gestational or familial nonautoimmune, hypothyroidism, thyrotropin)
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)
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)
565456-565460	Hypomagnesemia with Hypercalciuria and Nephrocalcinosis, Familial
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

565456-565460	Immune deficiency, X-linked, with hyperIgM
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)
565493-565504	Intellectual disability (gene panel)
565456-565460	Iron sulphur cluster assembly protein IscU (myopathy with SDH def)
565456-565460	Isolated mitochondrial respiratory chain complex V deficiency (Complex V assembly deficiency)
565456-565460	Jewish mutation panel (Tay Sachs, Fanconi, Dysautonomia, Canavan) (4 genes; 7 hot spot mutations)
565530-565541	Juvenile polyposis (2 genes)
565471-565482	Kabuki syndrome (gene panel)
565471-565482	Kallmann syndrome / Hypogonadotropic Hypogonadism
565456-565460	Kearns Sayre Syndrome
565456-565460	Kennedy disease / Spinal and bulbar muscular atrophy (SBMA) - AR gene 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 and transitional cell carcinoma renal pelvis) (gene panel)
565456-565460	Klippel-Feil syndrome
565471-565482	Krabbe disease
565493-565504	Leber Congenital Amaurosis - Retinal dystrophy, early onset (gene panel)
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)
565471-565482	Leigh / Narp Syndrome (1st tier-3 genes)
565493-565504	Leigh / NARP Syndrome (gene panel)
565456-565460	Leigh syndrome / Hypertrophic cardiomyopathy
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)
565456-565460	Lipodystrophy type 1, conggential generalized
565471-565482	Lipoid proteinosis
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
565493-565504	Long QT syndrome (gene panel)
565471-565482	Lymphoproliferative syndrome, X-linked (SH2D1A gene) / Duncan's disease
565471-565482	Lymphoproliferative syndrome, X-linked (XIAP gene)
565515-565526	Lynch syndrome MLH1 hypermethylation and BRAF mutation
565530-565541	Lynch-like syndrome
565493-565504	Lysosomal Storage Disease (64 genes)
565456-565460	Macrozoospermia (AURKC gene)
565456-565460	Macular dystrophy
565456-565460	Male infertility
565493-565504	Malformation, congenital (gene panel)
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 diabetes and deafness / Mitochondrial myopathy with reversible cytochrome C oxidase deficiency / mitochondrial tRNA glutamic acid
565471-565482	Mc Ardle disease, glycogene storage disease type V
565390-565401	Mc Cune Albright syndrome
565390-565401	Medium chain acyl-CoA dehydrogenase deficiency (MCAD hot spot mutation - p.Lys329Glu)
565471-565482	Medium chain acyl-CoA dehydrogenase deficiency (MCAD-ACADM)
565456-565460	Medullary Cystic Kidney Disease 1 (MUC1 gene)
565530-565541	Medullary thyroid carcinoma (3 genes)
565552-565563	Medulloblastoma (3 genes)
565530-565541	Medulloblastoma (gene panel)
565552-565563	Melanoma / Familial Atypical Multiple Mole Melanoma Syndrome (gene panel)
565530-565541	Melanoma, familial / Familial Atypical Multiple Mole Melanoma Syndrome, FAMMM (gene panel)
565530-565541	Meningioma (gene panel)
565456-565460	Mental retardation, X-linked, syndromic, Borck type
565493-565504	Metabolic diseases with hepatic disorders (20 genes)
565493-565504	Metabolic disorders (gene panel)
565471-565482	Metachromatic leukodystrophy
565456-565460	Methylmalonic aciduria type B
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
565552-565563	Mismatch Repair Deficiency Syndrome, constitutional (gene panel)
565456-565460	Mitochondial disease/paraganglioma
565456-565460	Mitochondrial complex III deficiency / Bjornstad syndrome / Gracile syndrome / Leigh syndrome
565471-565482	Mitochondrial complex IV deficiency
565456-565460	Mitochondrial complex V (ATP Synthase) deficiency (TMEM70 gene)
565456-565460	Mitochondrial complex V (ATP Synthase) deficiency, nuclear type 1 (ATPAF2 gene)
565493-565504	Mitochondrial disorders (gene panel)
565493-565504	Mitochondrial disorders, mitchondrial DNA based (Full sequencing of mtDNA genome)
565456-565460	Mitochondrial DNA depletion syndrome (DGUOK gene)
565456-565460	Mitochondrial DNA depletion syndrome (MPV17 gene)
565456-565460	Mitochondrial DNA depletion syndrome (RRM2B gene)
565471-565482	Mitochondrial DNA depletion syndrome (SUCLA2 gene)
565456-565460	Mitochondrial DNA depletion syndrome (SUCLG1 gene)
565456-565460	Mitochondrial DNA depletion syndrome (TK2 gene) / Mitochondrial DNA depletion syndrome, myopathic form
565456-565460	Mitochondrial DNA depletion syndrome (TYMP gene) / Mitochondrial neurogastrointestinal encephalomyopathy
565456-565460	Mitochondrial DNA mutation load
565456-565460	Mitochondrial DNA-associated Leigh syndrome / mitochondrial tRNA valine
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 (gene panel)
565390-565401	Mitochondrial non-syndromic sensorineural deafness with susceptibility to aminoglycoside exposure (hot spot mutation - 1555A-G in MT-RNR1)
565456-565460	Mitochondrial respiratory chain complex II deficiency
565456-565460	Mitochondropathy with primary renal involvement/coenzyme 10 deficiency / Leigh syndrome with nephrotic syndrome
565493-565504	Movement Disorders (gene panel)
565493-565504	mtDNA depletion syndrome (gene panel)
565456-565460	mtDNA depletion syndrome (encephalomyopathic)
565456-565460	mtDNA depletion syndrome (hepatocerebral)
565456-565460	mtDNA depletion syndrome (myopathic)
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 type 1 and 4 (MEN1; CDKN1B genes)
565530-565541	Multiple endocrine neoplasia (gene panel)
565515-565526	Multiple Endocrine Neoplasia type 2A and 2B / familial medullary thyroid carcinoma / Hirschsprung (1st tier screeening)
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
565456-565460	Multiple mitochondrial dysfunctions syndrome 1
565456-565460	Multiple mitochondrial dysfunctions syndrome 2 - BOLA3 deficiency
565471-565482	Multiple osteochondromas (2 genes)
565530 565541	Myeloid neoplasms with germline predisposition (Hereditary MDS/Acute Leukemia) (gene panel)
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
565390-565401	Myoglobinuria (hot spot mutation - p.Glu769Gly)
565493-565504	Myopathy (gene panel)
565471-565482	Myotonia, congenital, (Becker-Thomsen disease) (CLCN1 gene)
565456-565460	Myotonic dystrophy type 2 - CCTG repeat expansion
565456-565460	Myotonic dystrophy type 1 Steinert disease
565456-565460	NADH dehydrogenase 1 Alpha Subcomplex 12 / Leigh syndrome with leukodystrophy
565471-565482	Nanophthalmos
565390-565401	Narcolepsy (HLA-DQB1*0602 Genotyping) - Pharmacogenetics
565493-565504	Nephropathies, hereditary (218 genes)
565471-565482	Nephrotic syndrome - steroid resistant

565493-565504	Nephrotic syndrome, Focal Segmental Glomerulosclerosis (FSGS) , Alport syndrome and podocytopathy (33 genes)
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: developmental delay, intellectual disability, autistic 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
565493-565504	Neuromuscular disorders (gene panel)
565493-565504	Neuropathy (gene panel)
565456-565460	Neuropathy with Liability to Pressure Palsies, hereditary (HNPP)
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
565493-565504	Non-cholestatic jaundice with direct bilirubin (3 genes)
565552-565563	Nonpolyposis colorectal cancer, hereditary / Lynch syndrome (gene panel)
565456-565460	Noonan syndrome (Screening PTPN11)
565456-565460	Norrie disease (NDP gene)
565456-565460	Obesity, Monogenic early onset
565493-565504	Obesity (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 (gene panel)
565456-565460	Oculopharyngeal Muscular Dystrophy - GCN repeats expansion
565471-565482	Oligodontia-colorectal cancer syndrome
565493-565504	Onco-endocrine pathologies (50 genes)
565493-565504	Optic atrophy (OPA1 + gene panel)
565493-565504	Osteogenesis imperfecta / Osteoporose (gene panel)
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 idiopathic (PRSS1 gene)
565456-565460	Pancreatitis, hereditary
565471-565482	Pancreatitis, hereditary (gene panel)
565552-565563	Paraganglioma and pheochromocytoma (extended gene panel)
565530-565541	Paraganglioma-pheochromocytoma (gene panel) - ULG
565471-565482	Paralysis (myotonia), periodic / Paramyotonia congenita (SCN4A gene)
565530-565541	Parkinson (gene panel)
565493-565504	Paroxysmal Episodic Disorders (gene panel)
565456-565460	Pearson Syndrome
565493-565504	Pediatric oncopredisposition
565471-565482	Pendred syndrome
565493-565504	Perrault syndrome (gene panel)
565471-565482	Persistent Müllerian duct syndrome
565530-565541	Peutz-Jeghers Syndrome
565471-565482	Phenylketonuria
565471-565482	Pitt-Hopkins syndrome
565530-565541	Pituitary adenoma (4 genes)
565471-565482	Polyarteritis nodosa, childhood-onset / Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome
565493-565504	Polycystic kidney disease type 1 and 2
565471-565482	Polycythaemia vera, familial (TET2 gene)
565456-565460	Polycythaemia vera, familial / Familial thrombocythemia (congenital thrombocytosis, THCYT3) (JAK2 gene)
565456-565460	Polycythaemia vera, familial / Secondary familial erythrocytosis (EGLN1 gene)
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 Failure/Primary Ovarian Insufficiency (POF/POI) (32 genes)
565493-565504	Primary Arterial Hypertension (18 genes)
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) (113 genes)

565493-565504	Primary Electrical disorders / Brugada syndrome / Long QT syndrome (LQT) / Short QT syndrome (SQT) / Arrhythmogenic right ventricular dysplasia (ARVD) / Catecholaminergic polymorphic ventricular tachycardia (CPVT) (gene panel)
565456-565460	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
565493-565504	Progressive Myoclonic Epilepsy (gene panel)
565530-565541	Prostate cancer
565552-565563	Prostate cancer (gene panel)
565390-565401	Prostate cancer susceptibility (hot spot mutation - rs138213197 (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
565530-565541	Rare non-epithelial ovarian neoplasms (2 genes)
565493-565504	RASopathy (gene panel)
565456-565460	Recurrent metabolic encephalomyopathic crises-rhabdomyolysis-cardiac arrhythmia-intellectual disability syndrome (TANGO2 gene)
565530-565541	Renal carcinoma (4 genes)
565552-565563	Renal cell carcinoma (kidney cancer) (gene panel)
565471-565482	Renal cysts and diabetes syndrome / Maturity onset Diabete of the Young (MODY), type 5
565493-565504	Renal or urinary tract malformation (CAKUT) (gene panel)
565471-565482	Rendu-Osler-Weber disease (4 genes)
565390-565401	Resistance to clopidrogel - PON1 genotyping (Q192R) - Pharmacogenetics
565390-565401	Resistance to vitamin K antagonists - VKORC1 genotyping (3 exons sequencing and -1639G>A + 1173C>T) - Pharmacogenetics
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 / MECP2 Duplication Syndrome
565530-565541	Rhabdoid Tumor Predisposition Syndrome (SMARCA4; SMARCB1 genes)
565530-565541	Rhabdomyosarcoma
565456-565460	Rokitansky syndrome
565493-565504	Saddan dysplasia
565456-565460	Saethre-Chotzen syndrome
565471-565482	Sandhoff disease
565530 565541	Sarcoma (gene panel)
565390-565401	Schinzel-Giedion midface retraction syndrome
565530-565541	Schwannoma Predisposition Syndrome (gene panel)
565456-565460	Secondary familial erythrocytosis (EPAS1 gene)
565456-565460	Secondary familial erythrocytosis (VHL gene)
565456-565460	Segawa syndrome (GCH1 gene)
565471-565482	Segawa syndrome (TH gene)
565456-565460	Septo-optic dysplasia
565530-565541	Sessile serrated polyposis syndrome
565530-565541	Neutropenia, Severe Congenital
565390-565401	Sex determining region Y (presence/absence of the gene)
565456-565460	Sex determining region Y
565493-565504	Short Stature (gene panel)
565456-565460	Silver-Russell syndrome
565493-565504	Skeletal dysplasia (gene panel)
565493-565504	Skin disorders (gene panel)
565390-565401	SLCO1B1*1b,*5,*15 genotyping (transport protein) - Pharmacogenetics
565456-565460	Smith Lemli Opitz
565471-565482	Smith Lemli Opitz
565515-565526	Somatic analysis of the BRCA genes (2 genes)
565456-565460	Sorsby fundus dystrophy (TIMP3)
565493-565504	Spastic Paraplegia (gene panel)
565493-565504	Spastic Paraplegia, hereditary (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
565456-565460	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	Spondyloepiphyseal dysplasia congenita / Achondrogenesis / Kniest dysplasia / Hypochondrogenesis
565471-565482	Stargardt disease
565471-565482	Stickler syndrome (gene panel)
565493-565504	Stroke (gene panel)
565471-565482	Subcortical band heterotopia, lissencephaly
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
565456-565460	Tay-Sachs syndrome / GM2-gangliosidosis diagnostic (HEXA gene hot spot mutations - c.1274_1277dupTATC, c.1421+1G>C and c.805G>A (p.Gly269Ser))
565471-565482	Tay-Sachs syndrome / GM2-gangliosidosis diagnostic (Full sequencing)
565471-565482	Telangiectasia,hereditary hemorrhagic of Rendu Osler and Weber (gene panel)
test	test
565471-565482	Thalassemia Alpha (2 genes)
565456-565460	Thanatophoric dysplasia (hot spot mutations in FGFR3 gene)
565456-565460	Thiamine-responsive encephalopathy
565456-565460	Thiopurine S-Methyltransferase deficiency - TPMT and NUDT15 genotyping - Pharmacogenetics
565390-565401	Thiopurine S-Methyltransferase deficiency - TPMT genotyping of limited set of polymorphisms - Pharmacogenetics
565493-565504	Thoracic Aortic Aneurysm, familial (gene panel)
565456-565460	Thrombocythemia (congenital thrombocytosis, THCYT1) (THPO gene), familial
565456-565460	Thrombocythemia (congenital thrombocytosis, THCYT2) (MPL gene), familial
565456-565460	Thrombophilia due to protein C deficiency (PROC gene)
565493-565504	Thyroid disgenesis (38 genes)
565456-565460	Thyroid hormone receptor resistance (THRB gene)
565390-565401	Torsion dystonia 1 (hot spot mutation - c.907_909delGAG)
565471-565482	Treacher Collins (3 genes)
565493-565504	Trombosis - Hemostasis (gene panel)
565471-565482	Tuberous sclerosis (2 genes)
565493-565504	Tubulopathy (gene panel)
565530-565541	Tumor predisposition syndrome
565390-565401	Uniparental Disomy
565493-565504	Usher syndrome (gene panel)
565471-565482	Usher syndrome, type IIA
565471-565482	van der Woude syndrome / Popliteal pterygium syndrome
565471-565482	Vas deferens, congenital bilateral aplasia of, X-linked (ADGRG2 gene)
565471-565482	Vascular malformations (somatic)
565471-565482	Vascular mineralisation
565493-565504	Venous malformation (3 genes)
565456-565460	Vitamin D resistent rickets
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
565471-565482	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
565471-565482	X-linked Opitz G/BBB syndrome
565390-565401	Zygosity (medical)

criteria voor codes 565493-565504 en 565552-565563 werden verstengd. Minimale panel size werd opgetrokken en volgende panels werden verplaatst van 565493-565504/565552-565563 naar 565471-565482/565530-565541
Alport syndroom
Diabetes, Maturity onset Diabete of the Young (MODY) (genen panel)
Ectrodactyly / cleft lip/palate syndrome type 3 / Ectodermal dysplasia
Porencephaly / Hemorrhagic stroke / Cerebral small vessel disease / Idiopathic cerebral white matter lesions / HANAC / Isolated retinal arteriolar tortuosity
Pseudoxanthoma Elasticum
Pancreatitis, hereditary (gene panel)
Becker muscular dystrophy / Duchenne muscular dystrophy (Full sequencing DMD gene)
Von Willebrand disease
Caroli disease
Tuberous sclerosis
Rhabdomyosarcoma

criteria voor tier1/tier2 testen werd verstrengd, Alle tier 1 testen werden geëvalueerd door de mixed WG en een algoritme gebaseerd op diagnostische opbrengst versus kostprijs werd opgesteld om als criterium te gebruiken
Voor volgende TIER 1 testen werd bepaald dat zij niet meer aangerekend kunnen worden:
MC4R in combinatie met obesitas genen panel
DYT1 in combinatie met distonie genenpanel
Optische atrofie (OPA1)
Oculair albinisme type I
Spastic Paraplegia (SPG4)

criteria voor codes 565493-565504 en 565552-565563 werden verstengd. Minimale panel size werd opgetrokken
criteria voor tier1/tier 1 testen werd verstrengd, Alle tier 1 testen werden geëvalueerd door de mixed WG en een algoritme gebaseerd op diagnostische opbrengst versus kostprijs werd opgesteld om als criterium te gebruiken