

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 / Platypondylic 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 Trypsinemia, Hereditary
565390-565401	Alpha-1-antitrypsin deficiency (2 hot spot 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	Amyloidosis, 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	Antithrombin 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) / Immunodeficiency 31B (AR)

565471-565482 Capillary malformation – microcephaly

565493-565504 Cardiac arrhythmia, 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 **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 (Arthrogyrosis 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)
 Hallervorden-Spatz disease (Neurodegeneration with brain iron accumulation type 1) / HARP syndrome
 (Hypoprebetalipoproteinemia, Acanthocytosis, Retinitis pigmentosa, and Pallidal degeneration)

565471-565482 Hearing loss (deafness) (gene panel)
 565493-565504 Hearing loss (deafness), autosomal dominant 6/14 / Wolfram syndrome
 565471-565482 Hearing loss (Deafness), autosomal dominant 9 (COCH partial sequencing)
 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
 Hydrocephalia, X-linked/ CRASH (corpus callosum hypoplasia, retardation, adducted thumbs, spastic paraplegia, and
 hydrocephalus) syndrome (L1CAM gene)

565471-565482 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 ichthyosis, 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, congenital 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 hyperthermia
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 Mitochondrial 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, mitochondrial 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)
 565456-565460 Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes (MELAS) (hot spot mutation - m.3243A>G)

565390-565401 Mitochondrial NeuroGastroIntestinal Encephalomyopathy (MNGIE) syndrome (gene panel)
 565493-565504 Mitochondrial non-syndromic sensorineural deafness with susceptibility to aminoglycoside exposure (hot spot mutation - 1555A-G in MT-RNR1)

565390-565401 Mitochondrial respiratory chain complex II deficiency

565456-565460 Mitochondriopathy 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 Mucopolipidosis 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 Mucopolysaccharidosis 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 screening)
 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 syndrome / Catech. polymorphic ventricular tachycardia / Early repolarisation syndrome / Idiopathic 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
 Tay-Sachs syndrome / GM2-gangliosidosis diagnostic (HEXA gene hot spot mutations - c.1274_1277dupTATC, c.1421+1G>C and c.805G>A (p.Gly269Ser))
 565456-565460 Tay-Sachs syndrome / GM2-gangliosidosis diagnostic (Full sequencing)
 565471-565482 Telangiectasia,hereditary hemorrhagic of Rendu Osler and Weber (gene panel)
 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 Thrombocytopenia (congenital thrombocytopenia, THCYT1) (THPO gene), familial
 565456-565460 Thrombocytopenia (congenital thrombocytopenia, THCYT2) (MPL gene), familial
 565456-565460 Thrombophilia due to protein C deficiency (PROC gene)
 565493-565504 Thyroid dysgenesis (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 resistant 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 Zygoty (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 verstengd, 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 verstrengd. 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