Disorder	Gene	Alias	RIZIV/INAMI nomenclature
46, XY Sex Reversal	NR5A1	SF1	565471-565482
Aarskog-Scott syndrome	FGD1		565471-565482
Achondroplasia	FGFR3	ACH	565390-565401
Achromatopsia	CNGB3, CNGA3		565471-565482
Acrocapitofemoral dysplasia; Brachydactyly, type A1	IHH		565456-546460
Acrodysostose	PRKAR1A		565471-565482
acyl-CoA dehydrogenase family, member 9	ACAD 9	ACAD9	565471-565482
Adams-Oliver syndrome	ARHGAP31, DOCK6, RBPJ, NOTCH1, DLL4, EOGT		565493-565504
Adenomatous Polyposis, familial	APC + MUTYH+(POLD1+POLE)		565552-565563
Adenomatous Polyposis, familial (FAP)	APC		565530-565541
Adenomatous Polyposis, familial, Autosomal Recessive	NTHL1		565515-565526
Adrenogenital syndrome	CYP21A2		565471-565482
Adrenoleukodystrophy	ABCD1		565471-565482
Agnathia-otocephaly complex	PRRX1		565456-565460
Alagille syndrome	JAG1		565471-565482
Alanyl-tRNA synthase 2 (mt)	AARS2		565471-565482
Albright hereditary osteodystrophy	GNAS	AHO	565471-565482
Algrove syndrome (AAAS)	AAAS		565390-565401
Alpha-1-antitrypsin deficiency	SERPINA1	PI	565390-565401
Alport syndrome	COL4A3, COL4A4, COL4A5		565493-565504
ALPS	FASLG	TNFSF6	565456-565460
ALPS type 1A	FAS	TNFRSF6	565456-565460
Alzheimer disease (gene package)	APP, PSEN1, PSEN2		565493-565504
Alzheimer Disease, late onset (AD2)	ApoE		565390-565401
Amyloidosis hereditary / Dystransthyretinemic hyperthyroxinemia	TTR	TBPA	565456-565460
Amyotrophic lateral sclerosis	C9orf72	ALS10	565456-565460
Amyotrophic lateral sclerosis (gene package)	SOD1, TARDBP, FUS,		565456-565460
Androgen insensitivity	AR	DHTR, NR3C4	565456-565460
Angelman syndrome	UBE3A		565471-565482
Angelman/Prader Willi Syndrome	15q11-q13		565456-565460
Aniridia	PAX6		565471-565482
Anterior segment dysgenesis	FOXC1, PITX2, PITX3, FOXE3		565493-565504

Anti-Müllerian hormone receptor type 2	AMHR2		565471-565482
Antithrombine III deficiency (thrombophilia)	SERPINC1	AT3	565456-565460
APECED	AIRE		565471-565482
Apparent mineralocorticoid excess	HSD11B2		565456-565460
ARPKD, POLYCYSTIC KIDNEY DISEASE, AUTOSOMAL RECESSIVE	PKHD1	FCYT	565493-565504
Arterial Tortuosity Syndrome (ATS)	SLC2A10, FBLN4(=EFEMP2)		565471-565482
Ataxia (autosomic dominant and recessive) (gene package)	gene panel		565493-565504
Ataxia telangiectasia	ATM	Breast cancer, susceptibility to	565552-565563
Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia (AOA1)	APTX		565471-565482
Ataxia-ocular apraxia-2 (AOA2) / Amyotrophic lateral sclerosis 4 (ALS4) / Spinocerebellar Ataxia Autosomal Recessive 1 (SCAR1)	SETX		565471-565482
Atopic dermatitis	FLG	-	565390-565401
Atypical Hemolytic Uremic Syndrome (aHUS) panel	gene panel		565493-565504
Autism (gene panel)	gene panel		565493-565504
Autoimmune disease, multisystem, infantile-onset (ADMIO) / Hyper-IgE recurrent infection syndrome	STAT3		565471-565482
Autoimmune lymphoproliferative syndrome, type V	CTLA4		565456-565460
Azoo-/oligozoospermia	Yq11	AZF deletion	565390-565401
Bardet-Biedl syndrome 1	BBS1 + BBS10		565390-565401
BARTTER Syndrome type 1	SLC12A1		565471-565482
BARTTER syndrome type 2	KCNJ1		565456-565460
Bartter syndrome type 3	CLCNKB		565471-565482
Beals-Hecht syndrome; Congenital contractural arachnodactyly; Arthrogryposis Distal Type 9	FBN2		565471-565482
Becker muscular dystrophy / Duchenne muscular dystrophy	DMD		565456-565460
Becker muscular dystrophy / Duchenne muscular dystrophy	DMD		565493-565504
Beckwith-Wiedemann syndrome	11p15.5 (LIT1 - H19)	BWS	565456-565460

Best macular dystrophy	BEST1	VMD2	565471-565482
Beta-globin hemoglobinopathies	HBB	HPFH/deltabeta thalassaemia	565471-565482
Beta-globin hemoglobinopathies, phenotype modifiers	HBG2_XMN1, HBS1L-MYB, BCL11A		565390-565401
Beta-globin hemoglobinopathies, Sickle Cell Anemia+ hemoglobinose C	HBB		565390-565401
Bethlem myopathy gene package; Ullrich congenital muscular dystrophy gene package; Myosclerosis Myopathy gene package	COL6A1, COL6A2, COL6A3, COL12A1		565471-565482
Bicuspid aortic valve (BAV) disease gene package	SMAD6, NOTCH1, NKX2.5, GATA5		565493-565504
Birt Hogg Dube syndrome	FLCN	BHD	565471-565482
Blepharophimosis (BPES) type I /II	FOXL2	BPES	565471-565482
BOLA3-BOLA homolog 3 (Multiple mitochondrial dysfunctions syndrome 2)	BOLA3	-	565456-565460
Brachiootic syndrome 3	SIX1		565456-565460
Branchiootorenal syndrome (BOR) syndrome	EYA1		565471-565482
Branchiootorenal syndrome 2 (BOR)	SIX5		565471-565482
breast cancer susceptibility	BARD1		565515-565526
breast cancer susceptibility	BRIP1		565515-565526
breast cancer susceptibility	RAD51D		565515-565526
Breast cancer, hereditary	BRCA1		565530-565541
Breast cancer, hereditary	BRCA1, BRCA2		565530-565541
Breast cancer, hereditary	BRCA1,BRCA2, TP53, PALB2,CHEK2		565552-565563
Breast cancer, hereditary	BRCA2		565530-565541
Breast cancer, hereditary	CHEK2		565390-565401
Breast cancer, hereditary	PALB2		565530-565541
Breast cancer, hereditary	PALB2		565515-565526
Breast cancer, hereditary / Li-Fraumeni syndrome	CHEK2		565390-565401
Brittle Cornea Syndrome gene package	ZNF469, PRDM5		565471-565482
Bruck syndrome gene package	FKBP10, PLOD2		565471-565482
Brugada syndrome	SCN5A		565471-565482

Brugada syndrome (gene panel)	SCN5A,GPD1L,CACNA1C,CACNAB2,SCN1B,KCNE3,SCN3B,HCN4,CACNA2D1,KCND3,KCNE1L,KCNJ8,MOG1,SLMAP,TRPM4, SCN2B		565493-565504
Buschke-Ollendorff; Melorheostosis; Osteopoikilosis	LEMD3		565471-565482
BUTYRYLCHOLINESTERASE DEFICIENCY, INCLUDED	BCHE	pseudocholinesterase deficiency, butyrylcholinesterase deficiency	565471-565482
C8 deficiency, type I	C8A	C8 ALPHA	565471-565482
C8 deficiency, type II	C8B	C8 BETA	565471-565482
Cadasil	Notch3 (exons 2-24)		565471-565482
Canavan disease	ASPA		565390-565401
Cancer panel (Breast, ovary, colon,…)	gene panel		565552-565563
Candidiasis, familial 7 / Immunodeficiency 31A (AD) / Immunodefyciency 31B (AR)	STAT1		565471-565482
Capillary malformation - arteriovenous malformation	RASA1 - EPHB4 - STAMBP - PTEN		565493-565504
Cardioencephalomyopathy, fatal	SCO2	-	565456-565460
Cardiofaciocutaneous syndrome (CFC)	HRAS /KRAS /BRAF /MEK1/ MEK2		565471-565482
Cardiomyopathy (DCM, HCM, RCM, ARVC, LVNC)	gene panel		565493-565504
Cardiomyopathy, hypertrophic gene package	MYBPC3, MYH7, TNNT2		565471-565482
Cardiopathies, hereditary (gene panel)	gene panel		565493-565504
Carney complex	PRKAR1A		565530-565541
Carnitine Palmitoyl transferase type II	CPT2		565471-565482
CATARACT, JUVENILE, WITH MICROCORNEA AND GLUCOSURIA	SLC16A12	MCT12	565456-565460
CBAVD (gene package)	CFTR+Yq11	CFTR screening +AZF deletion	565456-565460

CDG IA, IB, IC	PMM2, MPI, ALG6	CDG Ia 212065 (PMM2:601785); CDG Ib 602579 (MPI : 154550); CDG Ic 603147 (ALG6 604566)	565471-565482
Central Precocious Puberty	MKRN3, KISS1, KISS1R (GPR54), PROKR2		565471-565482
Cerebral cavernous malformation	KRIT1 - CCM2 - PDCD10	CCM1 - CCM2 - CCM3	565493-565504
Cerebral folate transport deficiency	FOLR1/ FOLR2		565456-565460
Ceroid Lipofuscinosis	CLN6		565456-565460
Charcot-Marie-Tooth (gene panel)	gene panel		565493-565504
Charcot-Marie-Tooth disease, axonal, type 2T	MME		565471-565482
Charcot-Marie-Tooth disease, dominant intermediate D/ Dejerine-Sottas disease /Roussy-Levy syndrome	MPZ		565471-565482
Charcot-Marie-Tooth disease, type 2A2	MFN2		565471-565482
Charcot-Marie-Tooth disease, type 4A (CMT4A)	GDAP1		565471-565482
Charcot-Marie-Tooth type 1 / Hereditary neuropathy with liability to pressure palsies / Dejerine-Sottas syndrome	PMP22		565471-565482
Charcot-Marie-Tooth type 1A (CMT1A) / Hereditary Neuropathy with Liability to Pressure Palsies (HNPP)	PMP22	CMT1A	565456-565460
Charcot-Marie-Tooth, X-linked	GJB1	CX32	565456-565460
Charcot-Marie-Tooth, X-linked	GJB1	CX32	565471-565482
CHARGE syndrome	CHD7, (SEMA3E)		565493-565504
CHED2	SLC4A11		565471-565482
Choroideremia	CHM		565471-565482
Chronic granulomatous disease, X-linked	CYBB		565471-565482
Ciliopathy, polycystic kidney and liver	gene panel		565493-565504
Clouston syndrome	GJB6		565456-565460
Coffin-Lowry syndrome	RPS6KA3	RSK2	565471-565482
COFFIN-SIRIS SYNDROME	ARID1B		565471-565482
colon and endometrial cancer susceptibility (POLD1 gene)	POLD1		565530-565541
colon cancer susceptibility (POLE and POLD1 gene)	POLE and POLD1		565390-565401

Combined immunodeficiency (severe), X-linked	IL2RG	CD132	565456-565460
Combined pituitary hormone deficiency 1 (CPHD)	POU1F1	PIT1, GHF1	565456-565460
Combined pituitary hormone deficiency 2 (CPHD)	PROP1	-	565456-565460
Complement factor H deficiency / Hemolytic uremic syndrome, atypical, susceptibility to, 1	CFH	HF1, HF	565471-565482
Complex IV deficiency	MTCO1-3	COI,II,III	565471-565482
Complex V assembly deficiency	ATPAF1	ATP11	565456-565460
Cone-rod dystrophy (gene package)	ABCA4, CERKL, CNNM4, RPGRIP1, GUCY2D, AIPL1, CRX, PROM1, PRPH2		565493-565504
Congenital Central Hypoventilation Syndrome	PHOX2B		565471-565482
Congenital disorders of glycosylation (gene panel)	gene panel		565493-565504
Congenital heart disease (gene panel)	gene panel		565493-565504
Congenital heart disease nonsyndromic 2	TAB2		565471-565482
congenital hemangioma	GNAQ, GNA11		565456-565460
Congenital malformation syndromes (gene panel)	gene panel		565493-565504
Congenital Myasthenic Syndrome	CHRNE		565456-565460
Congenital Myasthenic Syndrome	CHRNE+RAPSN		565471-565482
Congenital Myasthenic Syndrome	RAPSN		565456-565460
Congenital myotonia (Becker-Thomsen disease)	CLCN1	CLC1	565471-565482
Congenital structural heart defects (gene panel)	gene panel		565493-565504
Costello Syndrome- Schimmelpenning syndrome	HRAS		565456-565460
Cowden disease	PTEN	Bannayan-Riley-Ruvalcaba syndrome (BRRS). Proteus syndrome	565530-565541
COX10 homolog, cytochrome c oxidase	COX10	-	565456-565460
CPEO	mtDNA genome	-	565493-565504
Craniosynostosis	FGFR1		565390-565401
Craniosynostosis	FGFR2		565456-565460
Craniosynostosis /Apert Syndrome	FGFR2		565390-565401
Craniosynostosis /Crouzon	FGFR3		565390-565401

Craniosynostosis Boston type	MSX2		565456-546460
Craniosynostosis, Muenke syndrome (hotspot analysis - p.Pro250)	FGFR3		565390-565401
Craniosynostosis/Apert Syndrome (hotspot analysis - p.Ser252; p.Pro253)	FGFR2		565390-565401
Creatine deficiency by Guanidinoacetate methyltransferase deficiency	GAMT + GATM	AGAT	565471-565482
Crigler Najjar	UGT1A1		565456-565460
Crisponi syndrome	CRLF1		565456-565460
Currarino syndrome	MNX1	HLXB9	565471-565482
Cutis Laxa gene package; Geroderma osteodysplasticum gene package	ELN, FBLN4(=EFEMP2), FBLN5, LTBP4, ATP6V0A2, PYCR1, ALDH18A1, ATP7A, COG7, TALDO1, SCYL1BP1(=GORAB), NAA10, RIN2		565493-565504
Cystic fibrosis (and related disorders)	CFTR		565353-565364
Cystic fibrosis (and related disorders)	CFTR		565471-565482
Cystic Fibrosis and related disorders	SCNN1A	ENAC	565493-565504
Czech dysplasie	COL2A1		565390-565401
Deafness non syndromal gene panel	gene panel		565493-565504
Deafness syndromal gene panel	gene panel		565493-565504
DEAFNESS, AMINOGLYCOSIDE-INDUCED (Mitochondrial Nonsyndromic Hearing Loss and Deafness)	mtDNA (MT-RNR1)	12SrRNA	565456-565460
DEAFNESS, AMINOGLYCOSIDE-INDUCED (Mitochondrial Nonsyndromic Hearing Loss and Deafness)	mtDNA (MT-RNR1)		565390-565401
Deafness, X-linked 2	POU3F4		565471-565482
Deficiency of Vitamin K-Dependent Clotting Factors	VKORC1, GGCX		565471-565482
Dentatorubral pallidoluysian atrophy	ATN1	DRPLA	565456-565460
DFNA6/14	WFS1		565471-565482
DFNA9	COCH		565456-565460
DFNB1A, DFNB1B	GJB2+GJB6 (deletion)	CX26	565456-565460
Diabetes neonatal	KCNJ11+ABCC8+GCK+ HNF1B		565493-565504
Diffuse gastric cancer	CDH1		565530-565541

Di-George	VCF		565456-565460
Dilated cardiomyopthy with conduction defect / Atypical Werner syndrome / Charcot-Marie-Tooth type 2B1 (autosomal recessive axonal neuropathy) CMT2B1 / 'Dunningam-type familial partial lipodystrophy (FPLD) / Emery Dreifuss Muscular Dystrophy (EDMD) / 'Hutchinson-Gilford progeria syndrome (HGPS) / 'Limb girdle muscular dystrophy type 1B (LGMD1B) / 'Mandibulo acral dysplasia (MAD)	LMNA		565471-565482
Disorders of sex development (DSD) panel (gene package)	NR5A1, SOX9, WT1, SRY, AR, DMRT1	SF1 (NR5A1)	565493-565504
Distal Hereditary motor neuropathy (TYPE IIA, IIB, IIC)	HSPB1 + HSPB3 + HSPB8		565471-565482
Dravet syndrome / Severe myoclonic epilepsy of infancy / Generalized epilepsy with febrile seizures plus	SCN1A		565471-565482
Duane-radial ray syndrome (DRSS)	SALL4		565471-565482
Dysautonomia	IKBAP		565390-565401
Early onset epileptic encephalopathy (gene package)	gene panel		565493-565504
Ectodermal dysplasia	GJB6		565456-565460
Ectopia lentis gene package	LTBP2, ADAMTSL4, FBN1		565493-565504
Ectrodactyly; cleft lip/palate syndrome type 3; Ectodermal dysplasia gene package	TP63, AXIN2, WNT10A, PAX9, MSX1, EDA, EDAR, EDARADD	603273	565493-565504
EDS (recessive) panel	B3GALT6, XYLT1, B3GAT3, B4GALT7, PLOD1, FKBP14, SLC39A13, CHST14, DSE, ADAMTS2, RIN2		565493-565504
EDS, arthrochalasis type (type VIIA – type VIIB)	COL1A1, COL1A2 (exons 5-8)		565456-546460
EDS, classic type (type I and II)	COL5A1, COL5A2		565471-565482
EDS, periodontal type	C1R, C1S		565471-565482
EDS, vascular type (type IV)	COL3A1		565471-565482
Ellis-van Creveld syndrome	EVC + EVC2		565493-565504
Emberger syndrome / Immunodeficiency 21	GATA2		565471-565482
Enhanced S-Cone Syndrome	NR2E3		565456-565460
Enzymatic dosage Chitotriosidase		CHIT1	565574-565585

Enzymatic dosage Fabry disease	alpha-galactosidase	GLA	565574-565585
Enzymatic dosage Gaucher disease	beta-glucosidase	GBA	565574-565585
Enzymatic dosage MPS1/Hurler syndrome	alpha-L-iduronidase	IDUA	565574-565585
Enzymatic dosage Pompe disease	alpha-glucosidase	GAA	565574-565585
epidermal nevus syndrome	HRAS, KRAS, NRAS, AKT1, PTEN, FGFR2, FGFR3		56547-565482
Epidermolysis bullosa	gene panel		565493-565504
Epilepsy (including Early Infantile Epileptic Encephalopathy) (gene package)	gene panel		565493-565504
Epilepsy, progressive myoclonic	KCTD7		565456-565460
Epileptic encephalopathy, early infantile, 1 or Infantile spasms without brain malformations (EIEE1), WEST syndrome	ARX		565471-565482
Epileptic encephalopathy, early infantile, 2 (EIEE2) /INFANTILE SPASM SYNDROME, X-LINKED 2 (ISSX2) / RETT	CDKL5	SERINE/THREONINE PROTEIN KINASE 9 (STK9)	565471-565482
Fabry disease	GLA		565471-565482
FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY 1A (FSHMD1A, FSHD1, DMFSH)	D4Z4 MACROSATELLITE REPEAT		565471-565482
FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY 1A (FSHMD1A, FSHD1, DMFSH)	D4Z4 MACROSATELLITE REPEAT		565390-565401
Factor V- cambridge and hong kong variant	F5		565390-565401
Familial Exudative Vitreoretinopathy, autosomaal dominant	FZD4, TSPAN12, LRP5		565471-565482
Familial hemiplegic Migraine FHM1; Episodic ataxia 2	CACNA1A		565493-565504
Familial hemiplegic Migraine FHM2	ATP1A2		565471-565482

Familial Isolated Pituitary Adenoma	AIP		565515-565526
Familial Mediterranean Fever (FMF)	MEFV		565471-565482
Familial melanoma	CDKN2A		565515-565526
Familial melanoma	CDKN2A, CDK4, POT1, BAP1	p16+p14(ARF)	565530-565541
Familial MSD/AML	GATA2, RUNX1, CEBPA		565530-565541
Familial MSD/AML	gene panel		565552-565563
Familial polycythaemia vera (OMIM 263300)	EGLN1/PHD2	modified	565456-546460
Familial polycythaemia vera (OMIM 263300)	JAK2	modified	565456-565460
Familial polycythaemia vera (OMIM 263300)	TET2	modified	565471-565482
Familial Thoracic Aortic Aneurysm (FTAA) gene package	gene panel		565493-565504
Familial thrombocythemia (congenital thrombocytosis, THCYT1, OMIM #187950)	THPO	modified	565456-565460
Familial thrombocythemia (congenital thrombocytosis, THCYT3, OMIM #614521)	JAK2	modified	565456-565460
Familial thrombocythemia(congenital thrombocytosis, THCYT2, OMIM #601977)	MPL	modified	565456-565460
Fanconi anemia	FANCA, FANCB, FANCC, FANCD1 (BRCA2), FANCD2, FANCE, FANCF, FANCG, FANCI, FANCJ (BRIP1), FANCL, FANCM, FANCN (PALB2), FANCO (RAD51C), FANCP (SLX4), FANCQ (ERCC4), FANCS (BRCA1), FANCT (UBE2T)		565552-565563
Fanconi anemia	FANCC		565390-565401
Fanconi Anemia / Breast cancer	RAD51C		565515-565526
Fascioscapulohumeral muscular dystrophy 2	SMCHD1		565493-565504

Feingold syndrome	MYCN		565456-546460
Fetal akinesia deformation sequence / Pena-Shokeir	DOK7		565456-565460
Floating Harbor	SRCAP		565456-565460
Focal SEGMENTAL GLOMERULOSCLEROSIS	ACTN4		565471-565482
FOCAL SEGMENTAL GLOMERULOSCLEROSIS 5; FSGS5	INF2		565456-565460
Fragile X syndrome/ PXPOI / FXTAS	FMR1		565375-565386
Fragile X syndrome/POF/FXTAS	FMR1		565375-565386
Friedreich ataxia (FRDA)	FXN		565456-565460
FRMD7-related infantile nystagmus (FIN), Nystagmus, infantile periodic alternating, X-linked	FRMD7		565471-565482
Frontotemporal lobar degeneration/ ALS (gene package)	GRN, MAPT, FUS , TARDBP		565471-565482
Fructosemia	ALDOB	ALDB, ALDO2	565456-565460
Fukuyama congenital muscular dystrophy (FCMD)	FKTN	FCMD	565471-565482
Gaucher disease diagnostic	GBA	-	565471-565482
Gaucher disease screening	GBA	-	565456-565460
Generalized Arterial Calcification of Infancy	ENPP1, ABCC6		565493-565504
Giant Axonal Neuropathy 1	GAN	GAN1	565471-565482
Gilbert syndrome / Irinotecan sensitivity	UGT1A1		565390-565401
Gitelman Syndrome	SLC12A3		565471-565482
globozoospermia	DPY19L2		565471-565482
Glomerulosclerosis, focal segmental, 2 (FSGS2)	TRPC6		565471-565482
Glycogen storage disease type 9	PHKA2		565471-565482

Glycogen storage disease type 0	GYS2		565471-565482
Glycogen storage disease type 1a	G6PC		565456-565460
GM2-gangliosidosis (Tay-Sachs syndrome) diagnostic	HEXA	-	565471-565482
GM2-gangliosidosis (Tay-Sachs syndrome) screening	HEXA	-	565456-565460
Growth impairment / hypotonia	PREPL		565471-565482
Growth retardation/short stature (gene package)	gene panel		565493-565504
GSD type III	AGL	-	565390-565401
(NEURODEGENERATION WITH BRAIN IRON ACCUMULATION 1; NBIA1) /	PANK2		565471-565482
Heart gene package	gene panel		565493-565504
Hemochromatosis hereditary type 1	HFE		565316-565320
Hemochromatosis hereditary type 4	SLC40A1	Ferroportine	565456-565460
Hemochromatosis, juvenile	HAMP/HJUV	HEPC/HFE2B	565456-565460
HEMOLYTIC UREMIC SYNDROME	CFH+CFI+MCP		565493-565504
Hemolytic uremic syndrome, atypical, susceptibility to, 2	MCP	CD46	565471-565482
Hemolytic uremic syndrome, atypical, susceptibility to, 6	THBD		565456-565460
Hemolytic uremic syndrome, atypical, susceptibility to, type 3; Complement factor I deficiency	CFI		565471-565482
Hemophilia A	FVIII		565471-565482
Hemophilia A	FVIII		565456-565460
Hemophilia B	FIX		565456-565460
Hepatic failure, early onset, and neurologic disorder	SCO1	-	565456-565460
Hereditary angioedema type III	F12		565390-565401
Hereditary angioneurotic edema	SERPING1	C1inh / HAE1	565471-565482
Hereditary Hemolytic Anemias (gene panel) due to unknown or doubtful origin	gene panel		565493-565504
Hereditary Myelodysplastic Syndrome/Acute Leukemia Predisposition Syndromes	gene panel	Myeloid neoplasms with germline mutation	565552-565563
Hereditary neuromyotonia and axonal neuropathy	HINT1		565456-565460
Hereditary nonpolyposis colorectal cancer (HNPCC)	MLH1 - MSH2 - MSH6 - (PMS2) - (EPCAM)		565552-565563

Hereditary nonpolyposis colorectal cancer (HNPCC)	PMS2		565530 565541
Hereditary nonpolyposis colorectal cancer (HNPCC) / Lynch MLH1 NEG (gene package)	BRAF + Méthylation promoteur MLH1		565515-565526
Hereditary spastic paraplegia (gene package)	gene panel		565493-565504
HNFJ4 HYPERURICEMIC NEPHROPATHY, FAMILIAL JUVENILE, 4	SEC61A1		565456-565460
Homocystinuria	MTHFR A1298C		565331-565342
Homocystinuria	MTHFR C677T		565331-565342
Huntington disease	HTT		565456-565460
Hypercalcemia with hypocalciuria / Hypoparathyroidism (Familial)	CASR	PCAR1	565471-565482
HYPERCALCEMIA, INFANTILE	CYP24A1		565471-565482
Hypercholesterolemia familial type B	APOB		565390-565401
Hypercholesterolemia panel	LDLR + APOB E26+ PCSK9 + APOE		565493-565504
Hypercholesterolemia, Familial	LDLR		565471-565482
Hyperekplexia	GLRA1		565390-565401
Hyperekplexia	GLRA1	STHE	565456-565460
Hyperekplexia	SLC6A5		565471-565482
Hyperekplexia	SLC6A5, GLRA1, GLRB		565493-565504
Hyperekplexia and spastic paraparesis / Startle disease, autosomal recessive / Startle disease/hyperekplexia, autosomal dominant	GLRA1		565471-565482
Hyperekplexia, autosomal recessive	GLRB		565471-565482
Hyperinsulinism gene package	GCK+ABCC8+KCNJ11+ HNF4A+INS		565493-565504
Hypermethylation MLH1 + BRAF V600 E			565515-565526
Hyperoxaluria, primary, type 1 (HP1) / OXALOSIS I / GLYCOLIC ACIDURIA	AGXT	SPAT	565471-565482
Hyperoxaluria, primary, type 2	GRHPR	HP2	565456-565460
Hyperoxaluria, primary, type 3	HOGA1	HP3	565456-565460
Hyperparathyroidism, neonatal severe primary	CASR	NSHPT / NSPH / NHPT	565471-565482
Hyperthyroidism, familial gestational	TSHR		565471-565482
Hyperthyroidism, familial nonautoimmune	TSHR		565456-565460

Hyperuricemic nephropathy, familial juvenile 1 (HNFJ1) / medullary cystic kidney disease type 2 (MCKD2)	UMOD		565471-565482
Hyperuricemic nephropathy, familial juvenile 2	REN		565456-565460
Hypocalcemia, autosomal dominant/Hypocalciuric hypercalcemia, familial type I	CASR		565471-565482
Hypocalciuric hypercalcemia, familial type II	GNA11		565456-565460
Hypocalciuric hypercalcemia, familial type III	AP2S1		565456-565460
Hypochondroplasia	FGFR3	HCH	565456-565460
Hypochondroplasia	FGFR3		565390-565401
Hypochondroplasia	FGFR3		565471-565482
Hypogonadism. Female Infertility.	LHB	LSH-beta; LSH-B; CGB4; HH23	565456-565460
Hypogonadotropic hypogonadism (gene panel)	gene panel		565493-565504
Hypokalemic periodic paralysis, type 1	CACNA1S		565390-565401
HYPOPARATHYROIDISM SENSORINEURAL DEAFNESS AND RENAL DISEASE (HDR syndrome)	GATA3		565471-565482
Hypoparathyroidism, familial isolated	CASR	FIH	565471-565482
Hypoparathyroidism, familial isolated	GCMB	FIH	565456-565460
Hypoparathyroidism, familial isolated	PTH	FIH	565456-565460
Hypophosphatasia	ALPL		565471-565482
Hypophosphatemic rickets	PHEX		565471-565482
Hypothyroidism	TITF1		565456-565460
Hypothyroidism	TTF2	FOXE1	565456-565460
Hypothyroidism, congenital (thyroid dysgenesis)	PAX8		565471-565482
Hypothyroidism, familial	TSHR		565471-565482
IBM2	GNE		565471-565482
Ichthyosis	gene panel		565493-565504
Immune deficiency with hyperIgM, type 3	CD40	TNFRSF5	565456-565460
Immune deficiency, X-linked, with hyperIgM	CD40LG	TNFSF5	565456-565460
Immunodeficiency	PIK3CD		565471-565482
Immunodeficiency	PIK3R1		565471-565482
Immunodeficiency 30	IL12Rb1	CD212	565471-565482
Incontinentia pigmenti	IKBKG	NEMO, IKKgamma, FIP3	565456-565460

Inherited bone marrow failure syndromes (IBMFS)	gene panel	IBMFS	565552-565563
Intellectual disability	gene panel		565493-565504
IPEX	FOXP3		565471-565482
ISCU - Iron sulphur cluster assembly protein IscU (myopathy with SDH def)	ISCU	-	565456-565460
Jewish mutation panel (Tay Sachs, Fanconi, Dysautonomia, Canavan)	ASPA+IKBAP+HEXA+FANCC		565456-565460
Juvenile polyposis syndrome	SMAD4/BMPR1A	JPS	565530-565541
KABUKI panel	7 genes (KMT2D, KDM6A, KMT2A, HNRNPK, RAP1A, RAP1B, PACS1-ex4)		565493-565504
Kallmann syndrome	ANOS1	KAL1	565471-565482
Kallmann syndrome, Hypogonadotropic Hypogonadism	FGFR1	KAL2	565471-565482
Kearns Sayre Syndrome	mtDNA del	-	565456-565460
Kennedy disease (SPINAL AND BULBAR MUSCULAR ATROPHY, SBMA)	AR		565456-565460
keratinopathic ichthyosis (epidermolytic ichtyosis, superficial epidermolytic ichthyosis, congenital reticular ichthyosiform erythroderma)	KRT1, KRT2, KRT10		56547-565482
Klippel-Feil syndrome	GDF6	CDMP2	565456-565460
Krabbe disease	GALC	-	565471-565482
Leber Congenital Amaurosis (LCA) (gene package)	AIPL1,CEP290, CRB1, CRX, GUCY2D, IQCB1, LCA5, LRAT, NMNAT1, RDH12, RPE65		565493-565504
Leigh Syndrome	mtDNA	-	565493-565504
Leigh syndrome with complex IV deficiency	SURF1	-	565456-565460
Leigh syndrome/Hypertrophic cardiomyopathy	COX15	-	565456-565460
Leigh/Narp Syndrome	MTATP6/8; MTND5		565471-565482
Leri-Weill dyschondrosteosis (LWD)/ISS	SHOX		565471-565482
Leri-Weill dyschondrosteosis (LWD)/ISS	SHOX		565456-565460
Leydig cell hypoplasia	LHCGR	LCH	565471-565482
LHON	mtDNA		565456-565460
LHON	MTND1, MTND4, MTCO3		565456-565460

Liddle Syndrome	SCNN1A +SCNN1B+SCNN1G	ENAC	565493-565504
Li-Fraumeni syndrome	TP53		565530-565541
Limb Girdle dystrophy type 2C	SGCG		565456-565460
Limb girdle muscular dystrophy type 1C / Rippling muscle disease 2 / sporadic hyperCKemia	CAV3		565456-565460
Lipodystrophy	AGPAT2 + BSCL2		565471-565482
Lipodystrophy, congenital generalized, type 1 (CGL1)	AGPAT2	BSCL1	565456-565460
Lipodystrophy, congenital generalized, type 2 / Silver spastic paraplegia syndrome / Neuropathy, distal hereditary motor, type V / SPG17	BSCL2	seipin	565390-565401
Lipoid proteinosis	ECM1	-	565471-565482
Lissencephaly (Lis) / subcortical band heterotopia (SBH)	PAFAH1B1	LIS1	565471-565482
LISSENCEPHALY 3	TUBA1A	TUBA3	565456-565460
Lissencephaly, X-linked / Subcortical band heterotopia / LIS / SBH	DCX		565471-565482
Long QT syndrome	SCN5A, KCNH2, KCNQ1		565471-565482
Long QT syndrome (gene panel)	KCNQ1,KCNH2,SCN5A ,ANK2,KCNE1,KCNE2, KCNJ2,CACNA1C,CAV 3,SCN4B,AKAP9,SNTA 1,KCNJ5		565493-565504
LONG-CHAIN 3-HYDROXYACYL-CoA DEHYDROGENASE DEFICIENCY (LCHAD deficiency)	HADHA	LCHAD	565390-565401
Low gamma-GT familial intra-hepatic cholestasis (gene package)	ATP8B1+ ABCB11	PFIC1+PFIC2	565493-565504
Low gamma-GT familial intra-hepatic	ABCB11	PFIC2	565471-565482
Low gamma-GT familial intra-hepatic	ATP8B1	PFIC1	565471-565482
LUJAN-FRYNS SYNDROME	MED12		565390-565401
Lutropin-Choriogonadotropin receptor	LHCGR		565471-565482
Lymfoproliferative syndrome, X-linked	SH2D1A		565471-565482
Lymphoproliferative syndrome, X-linked, 2	XIAP		565471-565482
Lysomal Storage Disease package	gene panel		565493-565504
Macrozoospermia	AURKc		565456-565460

Macular dystrophy	PRPH2		565456-565460
Malformations of cortical development (gene panel)	gene panel		565493-565504
Marfan Syndrome	FBN1		565471-565482
Maternally inherited deafness	mtDNA MTTS1	mtDNA MT-TS	565456-565460
Maturity onset Diabete of the Young (MODY) gene package	GCK+HNF1A+HNF4A+ HNF1B, INS, ABCC8		565493-565504
Maturity onset Diabete of the Young (MODY), type 5	HNF1B	TCF2	565471-565482
Mc Ardle disease,glycogene storage disease type V	PYGM		565471-565482
Mc Cune Albright	GNAS1		565331-565342
Mc Cune Albright	GNAS1		565390-565401
MED, SEDC, Achondrogenesis	COL2A1		565493-565504
Medium-Chain Acyl-CoA Dehydrogenase deficiency	ACADM		565390-565401
Medium-Chain Acyl-CoA Dehydrogenase deficiency	ACADM	MEDIUM-CHAIN ACYL-CoA	565471-565482
Medullary Cystic Kidney Disease 1	Mucin-1	MCKD1	565456-565460
MELANOMA, CUTANEOUS	CDK4		565515-565526
Mental retardation, autosomal dominant 17	PACS1		565390-565401
Mental retardation, X-linked,	EIF2S3		565456-565460
Metabolic disorders (gene panel)	gene panel		565493-565504
Metachromatic leukodystrophy	ARSA	-	565471-565482
Methylmalonic aciduria type B	MMAB	-	565456-565460
Microcephaly (gene package)	gene panel	Microcephaly, primary	565493-565504
Microphthalmia, syndromic 5; Retinal dystrophy, early-onset, and pituitary dysfunction	OTX2		565456-565460
Microspherophakia; Megalocornea; primary congenital Glaucoma; primary congenital Glaucoma; Weill-Marchesani syndrome 3 recessive type	LTBP2		565471-565482

Microtia, hearing impairment, and cleft palate	HOXA2		565456-565460
mitochondial disease	SDHAF1		565456-565460
mitochondial disease/paraganglioma	SDHAF2		565456-565460
Mitochondrial complex III deficiency, Bjornstad syndrome, Gracile syndrome, Leigh syndrome	BCS1L	-	565456-565460
Mitochondrial complex IV deficiency	FASTKD2	KIAA09071	565471-565482
Mitochondrial complex V (ATP synthase) deficiency	TMEM70	-	565456-565460
Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1	ATPAF2	ATP12	565456-565460
Mitochondrial Deletion	mtDNA		565456-565460
mitochondrial disease, nuclear based : Combined complex def., CPEO/multiple deletion, Leigh syndrome/isolated C I def., isolated CII def., isolated CIV	gene panel		565493-565504
Mitochondrial disorders (nuclear) (gene panel)	gene panel		565493-565504
Mitochondrial DNA depletion syndrome	DGUOK	DGK	565456-565460
MITOCHONDRIAL MYOPATHY, ENCEPHALOPATHY, LACTIC ACIDOSIS, AND STROKE-LIKE EPISODES (MELAS)	mtDNA		565456-565460
MITOCHONDRIAL MYOPATHY, ENCEPHALOPATHY, LACTIC ACIDOSIS, AND STROKE-LIKE EPISODES (MELAS)	mtDNA MT-TL1	tRNALeucine	565456-565460
MITOCHONDRIAL MYOPATHY, ENCEPHALOPATHY, LACTIC ACIDOSIS, AND STROKE-LIKE EPISODES (MELAS)	mtDNA MT-TL1	tRNALeucine	565390-565401
mitochondrial tRNA glutamic acid	mtDNA MT-TE	tRNAGlu	565456-565460
mitochondrial tRNA valine	mtDNA MT-TV	tRNAVal	565456-565460
Mitochondropathy with primary renal	COQ2	-	565456-565460
MNGIE syndrome	TYMP, POLG, POLG2,	-	565493-565504
MPS type I (Hurler/Scheie syndrome)	IDUA	-	565471-565482
MPS type II (Hunter syndrome)	IDS	-	565456-565460

MPS type IIIA (Sanfilippo A)	SGSH	HSS	565471-565482
MPS type IIIB (Sanfilippo B)	NAGLU	-	565471-565482
MPS type IVA (Morquio A syndrome)	GALNS	-	565471-565482
MPS type IVB (Morquio B syndrome) + GM1-gangliosidosis	GLB1	-	565471-565482
MPS type VII (Sly disease)	GUSB	-	565471-565482
MSI-analysis			565515-565526
mtDNA depletion syndrome	DGUOK, TK2,MPV17,RRM2B,TYMP,Twinkle		565493-565504
mtDNA depletion syndrome	MPV17	-	565456-565460
mtDNA depletion syndrome	RRM2B	P53R2	565456-565460
mtDNA depletion syndrome	SUCLA2	-	565471-565482
mtDNA depletion syndrome	SUCLG1	SUCLA1	565456-565460
mtDNA depletion syndrome	TK2	-	565456-565460
mtDNA depletion syndrome	TYMP	TP, ECGF1, PDECGF	565471-565482
mtDNA depletion syndrome	mtDNA	-	565456-565460
MtDNA mutation load	-	-	565456-565460
mtDNA resequencing	mtDNA	-	565493-565504
MTND6	mtDNA MTND6		565456-565460
Mucolipidosis II and III	GNPTAB	-	565471-565482
Mucopolysacharidose type IIIa (N-sulfoglucosamine sulfohydrolase)	SGSH/MPS3A	MPS3A	565456-565460
Multiple congenital anomalies-	PIGN		565471-565482
Multiple endocrine neoplasia type 1	MEN1		565530-565541

Multiple endocrine neoplasia type 2	RET		565530-565541
Multiple Endocrine Neoplasia type 2A and 2B (MEN2A and MEN2B) /	RET	FMTC	565515-565526
Multiple endocrine neoplasia type 4	CDKN1B		565515-565526
Multiple epiphysaire dysplasie recessief type	SLC26A2		565456-546460
Multiple mitochondrial dysfunctions syndrome 1	NFU1	-	565456-565460
Multiple osteochondromas	EXT1+EXT2		565471-565482
Muscular dystrophy, limb-girdle, type 2A / calpaïnopathy	CAPN3		565471-565482
MYHRE SYNDROME	SMAD4		565390-565401
Myoadenylate deaminase deficiency	AMPD1	-	565390-565401
MYOCLONIC EPILEPSY ASSOCIATED WITH RAGGED-RED FIBERS (MERFF)	mtDNA		565456-565460
MYOCLONIC EPILEPSY ASSOCIATED WITH RAGGED-RED FIBERS (MERFF)	mtDNA MTTK	tRNALysine	565456-565460
MYOCLONIC EPILEPSY ASSOCIATED	mtDNA MTTK	tRNALysine	565390-565401
MYOCLONIC EPILEPSY OF UNVERRICHT AND LUNDBORG / Progressive	CSTB	EPM1	565471-565482
Myoglobinuria	LPIN1	PAP1	565390-565401
Myotonic dystrophy 1 (Steinert disease)	DMPK		565456-565460
Myotonic dystrophy 1 (Steinert disease)	DMPK		565471-565482

Myotonic dystrophy type 2	ZNF9		565456-565460
Myotubular myopathy, X-linked (XLMTM, MTM,MTMX)	MTM1		565471-565482
NADH dehydrogenase 1 Alpha Subcomplex 12	NDUFA12L		565456-565460
Narp/Leigh Syndrome	MTATP6/8; MTND5	ATPase6&8;	565471-565482
Nephrogenic Insipidus Diabetes X-linked	AVPR2	NDI 1/NSIAD	565456-565460
Nephronophthisis 1, juvenile	NPHP1		565471-565482
Nephronophthisis 1, juvenile	NPHP1		565456-565460
NEPHRONOPHTHISIS 2 (NPHP2)	INVS		565471-565482
NEPHRONOPHTHISIS 5 (NPHP5),	IQCB1		565471-565482
Nephropathies, hereditary (gene panel)	gene panel		565493-565504
Nephrotic syndrome -steroid resistant	ADCK4		565471-565482
Nephrotic syndrome, FSGS, Alport	gene panel		565493-565504
Nephrotic syndrome, type 1 (Finnish)	NPHS1		565471-565482
Nephrotic syndrome, type 2 (Steroid-resistant)	NPHS2		565456-565460
Nephrotic syndrome, type 4 (including Frasier, denys-drash) NPHS4	WT1		565456-565460
Nephrotic syndrome, type 7	DGKE		565471-565482
Netherton syndrome	SPINK5	-	565471-565482
Neuraminidase deficiency	NEU1	SIAL1	565456-565460
Neurodegeneration with iron accumulation in brain 2A & 2B	PLA2G6	IPLA2, PNPLA9, IPLA2-VIA	565471-565482

Neurodevelopmental disorders: developmental delay, intellectual disability, autistic disorders (gene panel)	gene panel		565493-565504
Neurofibromatosis type 1	NF1	VON RECKLINGHAUSE	565552-565563
Neurofibromatosis type 1-like	SPRED1		565471-565482
Neurofibromatosis type 2	NF2		565530-565541
Neuromuscular disorders (Muscular dystrophy (Congenital), Myopathy	gene panel		565493-565504
Neuromuscular disorders : motor neuron disease (gene panel)	gene panel		565493-565504
NEUROPATHY, ATAXIA, AND RETINITIS	mtDNA		565390-565401
Neuropathy, hereditary sensory and autonomic, type IA	SPTLC1		565390-565401
Niemann-Pick disease	SMPD1	ASM	565471-565482
Night blindness, congenital stationary (complete), 1A, X-linked	NYX		565456-565460
Noonan syndrome (gene package)	KRAS, MAP2K1, MAP2K2, NRAS, PTN11, RAF1, SHOC2, SOS1		565493-565504

Noonan syndrome 1	PTPN11		565456-565460
Norrie disease	NDP	NORRIN	565456-565460
Obesitas, early onset (gene panel)	8 genes (MC4R,		565493-565504
Obesitas, Monogenic early onset	MC4R		565456-565460
Occipital horn syndrome; Distale	ATP7A		565471-565482
Occult macular dystrophy	RP1L1		565471-565482
Ocular albinism type 1	GPR143	OA1	565471-565482
Oculo Dento Digital Dysplasia (ODDD)	GJA1	Connexin 43 (CX43)	565456-565460
Oculocutaneous albinism type 1, 2 (gene package 1)	TYR, OCA2	OCA1, OCA2	565471-565482
Oculocutaneous albinism type 3, 4, 6, 7 (gene package 2)	TYRP1, SLC45A2, SLC24A5, C10orf11	OCA3, OCA4, OCA6, OCA7	565493-565504
Oculopharyngeal Muscular Dystrophy	PABPN1		565456-565460
Opitz GBBB syndrome, type I	MID1		565471-565482
Opitz-Kaveggia syndrome	MED12		565390-565401
Optic atrophy type 1	OPA1		565471-565482
Oral-facial-digital syndrome 1 / Joubert syndrome 10 / Simpson-Golabi-	OFD1		565471-565482
Osteogenesis imperfecta gene package 1	COL1A1, COL1A2, IFITM5		565493-565504
Osteogenesis imperfecta gene package	B3GALT6, BMP1,		565493-565504
Osteoporosis panel	LRP5, WNT1, PLS3		565493-565504
Ovarian cancer (somatic)	ACADM	-	565471-565482
Ovarian cancer (somatic)	BRCA1+BRCA2		565530-565541
Ovarian cancer (somatic)	BRCA1+BRCA2		565530-565541
Overgrowth & vascular anomalies / CLOVES syndrome	PIK3CA		565456-565460
Overgrowth & vascular anomalies / Proteus syndrome	AKT1		565331-565342
PANCREATIC HYPOPLASIA, INTESTINAL	RFX6	RFXDC1	565471-565482
Pancreatitis idiopathic (complementary analysis)	CTRC		565456-565460

Pancreatitis idiopathic panel	SPINK1 + CTRC + PRSS1		565471-565482
Pancreatitis, hereditary	PRSS1		565456-565460
Pancreatitis, hereditary	PRSS1		565471-565482
Pancreatitis, hereditary	SPINK1		565390-565401
Pancreatitis, hereditary	SPINK1 extended		565456-565460
Papillo renal syndrome/ RENAL-COLOBOMA SYNDROME	PAX2		565471-565482
Paraganglioma-pheochromocytoma	SDHA, SDHB, SDHC, SDHD, MAX,		565552-565563
Paraganglioma-pheochromocytoma	SDHB		565515-565526
Paraganglioma-pheochromocytoma	SDHB, SDHD		565530-565541
Paraganglioma-pheochromocytoma	SDHC		565515-565526
Paraganglioma-pheochromocytoma	SDHD		565515-565526
Paraganglioma-pheochromocytoma (gene package)	SDHB+SDHC+SDHD	paraganglioma	565530-565541
Parietal foramina	ALX4		565471-565482
Parietal foramina	MSX2		565471-565482
Parkinson	PARK2+ LRRK2 hotspot mutaties (ex		565471-565482
Parkinson disease, juvenile, type 2	Park2		565471-565482
Pearson Syndrome	mtDNA del	-	565456-565460
Pelizaeus Merzbacher (PMD) / Spastic paraplegia X-linked (SPG2)	PLP1		565471-565482
Pendred Syndrome	SLC26A4	PDS	565471-565482
Periodic Fever (gene package)	gene panel	FMF/Familial Mediterranean	565493-565504
Periodic paralysis (myotonia)	SCN4A	-	565471-565482
Perrault syndrome	PEO1, CLPP, HARS2,		565493-565504
Peutz Jeghers syndroom	STK11	PJS	565530-565541
Pharmacogenetics	ABCB1		565390-565401

Pharmacogenetics	ABCC2		565390-565401
Pharmacogenetics	CYP2B6		565390-565401
Pharmacogenetics	CYP2C19		565390-565401
Pharmacogenetics	CYP2C9		565390-565401
Pharmacogenetics	SLCO1B1 (protèine OATP1B1)		565390-565401
Pharmacogenetics	UGT1A1	Irinotecan	565390-565401
Pharmacogenetics Abacavir hypersensitivity	HLA5701		565390-565401
Pharmacogenetics AVK resistance	VKORC1		565390-565401
Pharmacogenetics clopidrogel resistance	PON1		565390-565401
Pharmacogenetics Dihydropyrimidine dehydrogenase deficiency/5-	DPYD		565456-565460
Pharmacogenetics Dihydropyrimidine dehydrogenase deficiency/5-	DPYD		565390-565401
Pharmacogenetics HEPATITIS C VIRUS,	IL28B	IL28B	565390-565401
Pharmacogenetics Metabolism	CYP2D6		565456-565460
Pharmacogenetics Metabolism	CYP3A4		565390-565401
Pharmacogenetics Narcolepsy	HLA		565390-565401
Pharmacogenetics Thiopurine S-Methyltransferase deficiency	TPMT		565390-565401
Pharmacogenetics Thiopurine S-Methyltransferase deficiency	TPMT		565456-565460
Pharmacogenetics Transplantation - tacrolimus	CYP3A5		565390-565401
Pharmacogenetics/Gilbert	UGT1A1		565390-565401
Phenylketonuria	PAH	PKU	565471-565482
Pheochromocytoma panel	RET +VHL + SDHA/B/C/D		565552-565563
Pitt Hopkins syndrome	TCF4		565471-565482
Pituitary panel	MEN1 + GNAS + AIP + CDKN1B + PRKAR1A		565552-565563
Polyarteritis nodosa, childhood-onset	CECR1	ADA2	565471-565482
POLYCYSTIC KIDNEY DISEASE 1 and 2	PKD1, PKD2		565493-565504
Polymicrogyria	GPR56	TM7XN1	565471-565482
POLYMICROGYRIA, ASYMMETRIC	TUBB2B	-	565456-565460

Pompe disease	GAA	-	565471-565482
Porencephaly; Hemorrhagic stroke;	COL4A1, COL4A2		565493-565504
Porphyria (gene package)	HMBS, PPOX,CPOX,FECH,ALAS2,UROD,UROS,ALAD		565493-565504
Precocious puberty, male-limited	LHCGR	FMPP	565471-565482
Premature Ovarian failure, autosomal recessive / Ovarian Hyperstimulation	FSHR		565471-565482
Premature Ovarian Failure/Insufficiency (POF/POI) (gene panel)	gene panel		565493-565504
Primary cardiac arrhythmias (atrial fibrillation, Brugada syndome, early	gene panel		565493-565504
Primary congenital glaucoma	CYP1B1		565471-565482
Primary Electrical disorders/Brugada	gene panel		565493-565504
Primary familial erythrocytosis or	EPOR	modified	565456-565460
Primary Hydrocephaly (gene package)	CCDC88C, MPDZ, L1CAM		565493-565504
Primary Hypomagnesemia	CLDN16		565456-565461
Primary immune deficiencies (gene panel)	gene panel		565493-565504
Primary immune deficiencies, APECED	AIRE		565471-565482
Primary immune deficiencies, subpanel: antibody deficiencies	ICOS, TNFRSF13B, TNFRSF13C, CD19,		565493-565504

Primary immune deficiencies, subpanel: chronic mucocutaneous candidiasis (CMC)	STAT3, DOCK8, TYK2, PGM3, CARD9, IL17RA, IL17RC, IL17F, TRAF3IP2, CLEC7A, STAT1, RORC		565493-565504
Primary immune deficiencies, subpanel: severe combined immunodeficiency (SCID)	IL2RG, JAK3, IL7R, IL15RA, PTPRC, CD3D, CD3E, CD3G, CD247, CD8A, ZAP70, RAG1, RAG2, DCLRE1C, ADA, AK2, LIG4, NHEJ1, PNP, PRKDC, LCK		565493-565504
Primary lymphoedema / fetal hydrops	FLT4 - VEGFC - FOXC2 - SOX18 - CCBE1 - KIF11 - PTPN14 - ITGA9 - GATA2 - GJC2 - GJA1 - HGF - FAT4 - PIEZO1 - IKBKG - HRAS - PTPN11 - KRAS - NRAS - RAF1 - SOS1 - RASA1 - EPHB4	VEGFR3 (FLT4)	565493-565504
Primary Pulmonary Hypertension	BMPR2		565471-565482
Progressive external ophthalmoplegia	PEO1	C10orf2, Twinkle	565471-565482
Progressive external ophthalmoplegia/ Alpers syndrome/ SANDO / MNGIE	POLG		565471-565482
Progressive Familial Intrahepatic Cholestasis Type 3 (PFIC3)	ABCB4	MDR3	565471-565482
Progressive Familial Intrahepatic Cholestasis, 1 gene	ATP8B1 and/or ABCB11 and/or ABCB4	ATP8B1/BSEP/MDR3	565471-565482
Progressive Familial Intrahepatic Cholestasis, 2 genes	ATP8B1 and/or ABCB11 and/or ABCB4	ATP8B1/BSEP/MDR3	565493-565504
prostate cancer susceptibility	HOXB13		565390-565401
Protein S deficiency	PROS1	Thrombophilia due to protein S deficiency	565471-565482
Proteus syndroom	AKT1		565471-565482

Pseudohypoaldosteronism type 1	SCNN1A +SCNN1B+SCNN1G	ENAC	565493-565504
Pseudoxanthoma Elasticum (PXE) gene package	ABCC6, ENPP1, GGCX, VEGFA(hotspots)		565493-565504
PTEN Hamartoma Tumor Syndrome	PTEN		565530-565541
Pulmonary Arterial Hypertension (gene package)	gene panel	PPH1 / PHT	565493-565504
PXE-like syndrome with clotting deficiency	GGCX		565471-565482
Pyruvate dehydrogenase deficiency/X-linked Leigh syndrome	PDHA1	PDHCE1A, PDHA, PHE1A	565471-565482
Renal cysts and diabetes syndrome/ Maturity onset Diabete of the Young (MODY), type 5	HNF1B	TCF2	565471-565482
Renal glucosuria (GLYS1)	SLC5A2	SGLT2	565471-565482
Rendu-Osler-Weber disease	ACVRL1 - ENG - SMAD4 - GDF2	ACVRL1 - ENG - SMAD4 - BMP9	565493-565504
Retinal dystrophy panel	gene panel		565493-565504
Retinal dystrophy, early onset	RDH12		565456-565460
Retinitis pigmentosa, AD (gene package)	CRX, NR2E3, PRPF6, PRPF8, PRPF31, PRPH2, RDH12, RHO, RP1, RPE65, SNRNP200		565493-565504
Retinitis pigmentosa, X-linked (gene package)	RPGR ORF15, RPGR exon 1-14, RP2, OFD1		565493-565504
Rett syndrome	MECP2		565471-565482
Rett syndrome, congenital variant	FOXG1		565471-565482
Rhabdoid Tumor Predisposition Syndrome 1	SMARCB1	SWNTS1	565530-565541
Rhabdoid Tumor Predisposition	SMARCA4	BRG1, SNF2-	565552-565563
RIN2 syndrome	RIN2		565471-565482
Rokitansky syndrome	WNT4		565456-565460
Saethre-Chotzen syndrome	TWIST	ACSIII	565456-565460
Sandhoff disease (Hexosaminidase B)	HEXB		565471-565482
Schinzel-Giedion midface retraction syndrome	SETBP1		565390-565401
Schuss-Hoeijmakers sydnrome	PACS1		565390-565401
Schwannomatosis-1	SMARCB1	SWNTS1	565530-565541
Schwannomatosis-2	LZTR1	SWTNS2	565530-565541
Secondary familial erythrocytosis (ECYT2; OMIM 263400)	VHL	modified	565456-565460
Secondary familial erythrocytosis (ECYT3; OMIM 609820)	EGLN1 (PHD2)	modified	565456-565460

Secondary familial erythrocytosis (ECYT4; OMIM 611783)	EPAS1 (HIF2A)	modified	565456-565460
Segawa syndrome	GCH1	Dopa responsive dystonia	565456-565460
Segawa syndrome	TH	-	565471-565482
Septo-optic dysplasia	HESX1	RPX	565456-565460
Severe Congenital Neutropenia	CSF3R	modified	565515-565526
Sex determining region Y	SRY		565390-565401
Sex determining region Y	SRY		565456-565460
Sialidase 1	NEU1	NEU1	565456-565460
Sickle Cell Anemia (HbS)+ hemoglobin C + hemoglobin E	HBB	beta-globin	565390-565401
Silver spastic paraplegia syndrome / Neuropathy, distal hereditary motor, type V / SPG17	BSCL2	seipin	565390-565401
Silver-Russel syndrome	KCNQ1 - H19 (11p15)	SRS	565456-565460
Simpson-Golabi-Behmel syndrome	GPC3		565471-565482
Skeletal dysplasia (gene panel)	gene panel		565493-565504
Skeletal dysplasia gene panel	gene panel		565493-565504
Skin disorders (gene panel)	gene panel		565493-565504
Smith Lemli Opitz	DHCR7		565456-565460
Sorsby fundus dystrophy	TIMP3		565456-565460
Sotos syndrome	NSD1		565471-565482
Sotos syndrome 2; Marshall-Smith syndrome	NFIX		565471-565482
Spastic paraplegia-3A	ATL1	SPG3A	565471-565482
Spastic paraplegia-4	SPAST		565471-565482
Spastic paraplegia-4	SPAST	SPG4	565471-565482
Spinal muscular atrophy type 1 (Werdnig-Hoffmann), type 2, type 3 (Kugelberg-Welander) and type 4	SMN1		565456-565460
Spinal muscular atrophy with respiratory distress (SMARD1, HMN6, DSMA1)	IGHMBP2		565471-565482
SPINAL MUSCULAR ATROPHY, DISTAL, AUTOSOMAL RECESSIVE (DSMA4)	PLEKHG5		565471-565482
Spinocerebellar ataxia (type 13)	KCNC3	SCA13	565390-565401
Spinocerebellar ataxia (type 17)	TBP	SCA17	565456-565460
Spinocerebellar ataxia (type 8, 10, 12, 17) + DRPLA	ATXN8, ATXN10, PPP2R2B, TBP, ATN1		565456-565460

Spinocerebellar ataxia (types 1,2,3,6,7)	ATXN1/ ATXN2/ ATXN3/ CACNA1A/ ATXN7	SCA1-SCA2-SCA3-SCA6-SCA7	565471-565482
Spondyloepiphyseal dysplasia congenita; Achondrogenesis; Hypochondrogenesis; Kniest dysplasia; Multiple epiphysaire dysplasie dominant type	COL2A1		565493-565504
Spondylo-megaepiphyseal-metaphyseal dysplasia	NKX3-2		565456-546460
Spondyloperipheral dysplasia	COL2A1		565456-565460
Stargardt disease	ABCA4		565493-565504
Stickler syndrome	COL11A1, COL2A1, COL9A1, COL9A2,		565493-565504
Stickler syndrome gene package	COL2A1, COL11A1, COL11A2		565493-565504
Subcortical band heterotopia, lissencephaly	DCX		565471-565482
Swachman-Bodian-Diamond syndrome	SBDS		565456-565460
Synpolydactyly / Brachydactyly	HOXD13		565471-565482
Tay Sachs	HEXA		565390-565401
TELANGIECTASIA, HEREDITARY HEMORRHAGIC, OF RENDU, OSLER, AND WEBER	ALK1, ENG	HHT	565471-565482
Thalassemia Alpha	HBA1, HBA2		565471-565482
Thanatophoric dysplasia	FGFR3		565456-565460
Thiamine Transporter	SLC19A3		565456-565460
Thrombophilia due to protein C deficiency	PROC	PC	565456-565460
Thrombophilia Factor 5 variant Hong-kong, cambridge, Liverpool	F5		565390-565401
Thyroid disgenesis (gene panel)	gene panel		565493-565504
Thyroid hormone receptor resistance	THRB	ERBA2	565456-565460
THYROTROPIN DEFICIENCY, ISOLATED	TSHB		565456-565460
Thyrotropin, unresponsiveness to	TSHR		565471-565482
Torsion dystonia 1	TOR1A	DYT1	565390-565401
Townes-Brocks branchiootorenal-like syndrome (TBS)	SALL1		565471-565482
Transport and golgi organization homolog 2	TANGO2	TANGO2	565456-565460

Treacher Collins syndrome	POLR1C, POLR1D, TCOF1		565493-565504
Tuberous sclerosis	TSC1 - TSC2 gene panel		565493-565504
Tubulopathies panel	gene panel		565493-565504
TUMOR PREDISPOSITION SYNDROME	BAP1		565530-565541
Uniparental Disomy		UPD	565390-565401
Uniparental disomy 14	UDP14		565456-565460
Uniparental disomy 7	UDP7		565456-565460
Usher syndrome	CDH23, CLRN1, DFNB31, GPR98, MYO7A, PCDH15, PDZD7, USH1C,		565493-565504
Usher syndrome, type IIA	USH2A		565493-565504
Van der Woude syndrome, Popliteal pterygium syndrome	IRF6		565471-565482
Vas deferens, congenital bilateral aplasia of, X-linked	ADGRG2	-	565471-565482
Vas deferens, congenital bilateral aplasia of, X-linked	ADGRG2	ADGRG2	565471-565482
Vascular mineralisation gene package	ANKH, NT5E(=CD73) en ENPP1		565493-565504
Venous malformation	TEK - GLMN	TIE2 - GLMN	565493-565504
Vitamin D resistent rickets	VDR		565456-565460
Vitelliform Macular Dystrophy	BEST1, PRPH2, IMPG1, IMPG2		565493-565504
V-maf Musculoaponeurotic fibrosarcoma oncogen homolog B	MAFB		565456-565460
Von Hippel Lindau	VHL		565530-565541
Von Willebrand disease	vWF		565493-565504

Waardenburg syndrome	EDN3, EDNRB, MITF, PAX3, SNAI2, SOX10		565493-565504
Waardenburg Syndrome type I / type III	PAX3		565471-565482
Weaver syndrome 2 (614421)	EZH2		565471-565482
Weill-Marchesani syndrome gene package	ADAMTS10, ADAMTS17, FBN1, LTBP2		565493-565504
WHIM syndrome	CXCR4		565456-565460
Wiskott-Aldrich syndrome	WAS		565471-565482
Witkop syndrome	MSX1		565456-565460
Wolfram syndrome	WFS1		565471-565482
X-chromosome inactivation			565456-565460
X-linked agammaglobulinemia	BTK		565471-565482
X-linked creatine deficiency	SLC6A8		565471-565482
X-linked hydrocephalia/CRASH syndrome	L1CAM	CAML1	565471-565482
Zygosity (medical)			565390-565401