Panel	Gene set	Order No.
Diagnostic Panels		
Blood and Immune Disorders	Erythrocytes incl. Dyserythropoetic anemia, Diamond-Blackfan anemia, Megaloblastic anemia, Haemolytic anemia	BID01
	Platelets, Coagulation disorders	BID02
	Thrombocytopenia	BID03
	Bone marrow failure syndromes incl. Fanconi anemia, Dyskeratosis congenita, Aplastic anemia	BID04
	Bone marrow failure syndromes, Leukemia	BID05
	Bone marrow failure syndromes, Metabolism	BID06
	Antibody deficiencies incl. Hyper-IgM-Syndrome, Agammaglobinemia, Common variable immunodeficiency	BID07
	Complement deficiencies incl. Neisseria Infections	BID08
	Autoinflammatory diseases incl. Periodic fever syndromes, Early-onset chronic inflammatory bowel diseases	BID09
	Immune dysregulation incl. hemophagocytic lymphohistiocytosis, Lymphoproliferative syndrome	BID10
	Defects of phagocytosis incl. Neutropenia, Mycobacteriosis, Leukocyte adhesion defect, Chronic granulomatous disease	BID11
	Defects in innate immunity incl. Chronic mucocutaneous candidiasis	BID12
	Combined immunodeficiencies	BID13
	Syndromes with immunodeficiency incl. Hyper-IgE-Syndrome, Interferonopathies	BID14
Ciliopathies	Primary Ciliary Dyskinesia	CIL01
	Joubert Syndrome	CIL02
	Bardet-Biedl Syndrome	CIL03
	Senior Loken Syndrome	CIL04
Connective Tissue and Skeletal Diseases	Stickler syndrome	CTD01
	Connective tissue diseases: Ehlers-Danlos syndrome, Marfan syndrome, Loeys-Dietz syndrome, thoracic aortic aneurysm and related disorders	CTD02
Skin Diseases	Oculocutaneous albinism	DRM01
	Syndromic albinism and related disorders : Hermansky-Pudlak syndrome, Griscelli syndrome, Waardenburg syndrome	DRM02
	Hyperpigmentation: Dowling-Degos and related disorders	DRM03
	Ichthyosis, palmoplantar keratoderma, and related disorders of cornification	DRM04
	Epidermolysis bullosa and related genetic blistering disorders	DRM05

Panel	Gene set	Order No.
Skin Diseases (continued)	Connective tissue diseases: Ehlers-Danlos syndrome, Marfan syndrome, Loeys-Dietz syndrome, thoracic aortic aneurysm and related disorders	DRM07
	Ectodermal dysplasia, selective tooth agenesis, trichothiodystrophy, and hypotrichosis	DRM08
	Dyskeratosis congenita	DRM09
	Photodermatosis: Xeroderma pigmentosum, Cockayne syndrome, COFS syndrome and related disorders	DRM10
	Neurofibromatosis / Schwannomatosis	DRM11
	Vascular disorders: hereditary hemorrhagic telangiectasia, cerebral cavernous malformations, association with MoyaMoya, and related disorders	DRM12
	Progeria syndromes and Lipodystrophy	DRM13
Hearing Loss	Hearing loss, nonsyndromic, autosomal recessive and X-linked	EAR01
	Hearing loss, nonsyndromic, autosomal dominant and X-linked	EAR02
	Syndromic Hearing Loss	EAR03
Epilepsy, Metabolic and Brain Development Disorders Epilepsy	Familial and Idiopathic Epilepsy	EPI01
	Epilepsy and Developmental Delay (incl. Epileptic Encephalopathies)	EPI02
	Progressive Myoclonus Epilepsy and Neuronal Ceroid Lipofuscinosis	EPI05
	GPI anchor deficiency with or without Hyperphosphatasia	EPI12
	Migraine	EPI14
	Hyperekplexia	EPI15
	Metabolic/Mitochondrial Epilepsy	EPI19
Metabolic Diseases	Congenital Disorders of Glycosylation (CDG syndrome)	MET01
	Lysosomal Disorders	MET02
	Peroxisome Biogenesis Disorders: Zellweger spectrum disorder	MET03
	Pyridoxine- and Folic Acid-dependent epilepsy	MET04
	Urea Cycle Disorders	MET05
	Glycine Encephalopathy	MET06
	Hyperphenylalaninemia	MET07
	Maple Syrup Urine Disease and DLD Deficiency	MET08
	Molybdenum Cofactor Deficiency and Sulfite Oxidase Deficiency	MET09
	Methylmalonic Acidemia	MET10
	3-Methylglutaconic aciduria	MET11
	Hyperinsulinemic Hypoglycemia	MET12
	Maturity-onset Diabetes of the Young (MODY)	MET13
	Glycogen Storage Diseases	MET14
	Fatty acid oxidation disorders	MET15

Panel	Gene set	Order No.
Brain Development Disorders	Microcephaly and Pontocerebellar Hypoplasia	BRN01
	Neuronal Migration Disorders	BRN02
	Holoprosencephaly spectrum	BRN03
	Macrocephaly	BRN04
	Leukodystrophy and Leukoencephalopathy	BRN05
	Aicardi-Goutières Syndrome	BRN06
	Cornelia de Lange Syndrome	BRN08
	Cerebral Microangiopathies	BRN09
	Leukodystrophy and Leukoencephalopathy and Differential Diagnoses	BRN10
	Kabuki Syndrome	BRN11
	Coffin-Siris syndrome	BRN12
Eye Diseases	Usher Syndrome	EYE01
	Retinitis pigmentosa, autosomal dominant and X-linked	EYE02
	Retinitis pigmentosa, autosomal recessive and X-linked	EYE03
	Achromatopsia	EYE04
	CNGB3, exon 10 only Achromatopsia (NGS panel)	
	Bardet-Biedl Syndrome	EYE05
	Congenital Stationary Night Blindness	EYE06
	Joubert Syndrome	EYE07
	Leber Congenital Amaurosis	EYE08
	Zellweger syndrome spectrum (Refsum/ Zellweger/ neonatale adrenoleukodystrophiy)	EYE10
	Senior Loken Syndrome	EYE11
	Stargardt Disease and Macular Dystrophies	EYE12
	Cone Rod Dystrophies	EYE13
	Flecked Retina Disorders	EYE14
	Vitreoretinopathies (Wagner syndrome/ Norrie/ Coats)	EYE15
	Stickler Syndrome	EYE16
	Optic atrophy (incl. LHON (Leber Hereditary Optic Neuropathy))	EYE17
	Oculocutaneous albinism	EYE18
	Syndromic albinism (Hermansky-Pudlak/ Waardenburg/ Vici/ Griscelli)	EYE19
	Ocular malformations (microphthalmia/ anophtalmia/ nanophthalmia/ colomba)	EYE20
	Cataract	EYE21
	Septo-optical dysplasia	EYE22
	Glaucoma	EYE23
	Corneal dystrophies	EYE24
	Ectopia lentis	EYE25

Panel	Gene set	Order No.
Cardiac Diseases	Cardiomyopathy, dilated	HRT01
	Cardiomyopathy, hypertrophic	HRT02
	Cardiomyopathy, restrictive	HRT12
	Left Ventricular Noncompaction Cardiomyopathy (LVNC)	HRT03
	Atrial Fibrillation and Short QT Syndrome	HRT04
	Long QT Syndrome	HRT05
	Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy (ARVD/C)	HRT06
	Brugada Syndrome	HRT07
	Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT), Paroxysmal/ Idiopathic Ventricular Fibrillation / Tachycardia	HRT08
	Congenital Heart Defects	HRT09
	RASopathies	HRT10
Ion Channel Diseases	Episodic Ataxia	ION01
	Neuropathic Pain Syndromes	ION06
	Paroxysmal Dyskinesias	ION07
	Malignant Hyperthermia	ION10
Kidney Diseases	Nephronophthisis	KID01
	NPHP1 deletion/duplication analysis (MLPA)	
	Cystic Kidney Disease	KID02
	PKD1/PKD2 sequencing Cystic Kidney Disease (NGS panel)	
	Renal Tubular Dysgenesis	KID03
	Renal Dysplasia, Renal Agenesia, CAKUT	KID04
	Nephrotic Syndrome	KID05
	Focal Segmental Glomerulosclerosis	KID06
	Alport Syndrome and Disorders of Glomerular Basement Membrane (GBM)	KID07
	C1q Deficiency	KID08
	Renal tubular acidosis	KID09
	Bartter Syndrome and Differential Diagnoses	KID10
	Hypophosphatemic rickets	KID11
	Pseudohypoaldosteronism	KID12
	Diabetes insipidus, nephrogenic	KID13
	Hyperoxaluria	KID14
	Atypical Hemolytic Uremic Syndrome and Differential Diagnoses	KID15
	Primary Inherited Aminoacidurias	KID16
	Branchiootorenal Syndrome	KID17
	Bardet-Biedl Syndrome	KID18
	Joubert Syndrome	KID19
	Meckel Syndrome	KID20
	Senior-Loken Syndrome	KID21

Panel	Gene set	Order No.
Liver Diseases	Cholestasis familial	LIV01
	Hypercholanemia and Bile acid synthesis defects	LIV02
	Impairment of the transport in hepatocytes and cholangiocytes	LIV03
	Impairment of the organo morphogensis	LIV04
	Metabolic disorders of hepatocytes, including Tyrosinemia, Glycogen storage diseases, Hyperammonemia, Shwachman-Diamond syndrome, Disorder of fatty acid oxidations and Peroxisomal diseases	LIV05
	Lysosomal storage disorders	LIV06
	Recurrent acute liver failure	LIV07
	Hepatic Mitochondriopathies	LIV08
Mitochondrio-pathies	Mitochondrial DNA (mtDNA)	MIT01
	Progressive external ophthalmoplegia (PEO/CPEO)	MIT16
	Leigh syndrome (nuclear genes)	MIT03
	Mitochondrial encephalopathy / Mitochondrial Hepato(encephalo)pathy (nuclear genes)	MIT04
	Mitochondrial DNA-depletion and deletion syndromes (nuclear genes)	MIT05
	Pyruvate Metabolism Disorders (nuclear genes)	MIT06
	Combined oxidative phosphorylation deficiency (COXPD)	MIT07
	Complex I-Deficiency	MIT08
	Complex II-Deficiency	MIT09
	Complex III-Deficiency	MIT10
	Complex IV-Deficiency	MIT11
	Complex V-Deficiency	MIT12
	CoQ10 Deficiency and Acyl-CoA-Dehydrogenase Deficiency	MIT13
	Methylglutaconic Aciduria (MGA)	MIT14
	MELAS and MERRF syndrome	MIT15
	Nuclear encoded Mitochondriopathies	MIT02
Neurodegenerative Diseases	Parkinson's disease, autosomal dominant	NDD01
	Parkinson's disease, autosomal recessive	NDD02
	Atypical Parkinson's disease	NDD03
	Dystonia-Parkinsonism	NDD04
	Parkinson's disease	NDD05
	Primary torsion dystonia	NDD06
	Dystonia Plus syndrome	NDD07
	Paroxysmal dyskinesia	NDD08
	Hereditary Degenerative Syndromes	NDD09
	Dystonia	NDD10
	Neurodegeneration with brain iron accumulation (NBIA)	NDD11
	Neuroacanthocytosis (full analysis), including:	NDD12
	Neuroacanthocytosis (NGS Panel) JPH3 repeat analysis	

Panel	Gene set	Order No.
Neurodegenerative Diseases (continued)	Choreatic movement disorders, step-by-step analysis: Step 1: HTT, JPH3 repeat analysis Step 2: Choreatic movement disorders (NGS Panel)	NDD13
	Ataxia, autosomal dominant, step-by-step analysis: Step 1: SCA1, SCA2, SCA3, SCA6, SCA7, SCA17 repeat analysis Step 2: Ataxia, autosomal dominant (NGS Panel)	NDD25
	Ataxia, autosomal recessive and X-linked, step-by-step analysis: Step 1: FXN repeat analysis Step 2: Ataxia, autosomal recessive and X-linked (NGS Panel)	NDD26
	Episodic ataxia	NDD30
	Ataxia and differential diagnoses, step-by-step analysis: Step 1A: SCA1, SCA2, SCA3, SCA6, SCA7, SCA17 repeat Step 1B: FXN repeat analysis Step 2: Ataxia and differential diagnoses (NGS Panel)	NDD14
	Frontotemporal dementia (FTD), step-by-step analysis: Step 1: C9orf72 repeat analysis Step 2: Frontotemporal dementia (FTD) (NGS Panel)	NDD15
	Alzheimer's disease (full analysis), including: Step 1: C9orf72 repeat analysis Step 2: Alzheimer's disease (NGS Panel)	NDD16
	Dementia, step-by-step analysis: Step 1: C9orf72 repeat analysis Step 2: Dementia (NGS Panel)	NDD17
	Amyotrophic lateral sclerosis (ALS), step-by-step analysis: Step 1: C9orf72 repeat analysis Step 2: Amyotrophic lateral sclerosis (ALS) (NGS Panel)	NDD18
	Hereditary spastic paraplegia (HSP), autosomal dominant (NGS	NDD27
	Hereditary spastic paraplegia (HSP), autosomal recessive and X-	NDD28
	Hereditary spastic paraplegia (HSP) all	NDD20
	Neuronal ceroid lipofuscinosis (NCL)	NDD21
	Leukodystrophy and Leukoencephalopathy	NDD29
	Leukodystrophy / Leukoencephalopathy and differential diagnoses	NDD22
	Cerebral small vessel disease	NDD23
	Basal ganglia calcification	NDD24

Panel	Gene set	Order No.
Neuromuscular Diseases	Spinal Muscular Atrophy (SMA), step-by-step analysis: Step 1: SMN1 deletion/duplication analysis (MLPA) Step 2: Spinal Muscular Atrophy (NGS Panel)	NMD01
	Hereditary Neuropathies, step-by-step analysis: Step 1: PMP22 deletion/duplication analysis (MLPA) Step 2: Hereditary Neuropathies (NGS Panel)	NMD02
	Congenital and Distal Myopathies	NMD03
	Limb-girdle Muscular Dystrophies	NMD04
	Muscular Dystrophies, step-by-step analysis: Step 1: DMD deletion/duplication analysis (MLPA) Step 2: Muscular Dystrophies (NGS Panel)	NMD05
	Congenital Myasthenic Syndromes and Arthrogryposis	NMD06
	Myotonia	NMD07
	Metabolic Myopathies	NMD08
	Walker-Warburg Syndrome	NMD10
	Periodic Paralysis	NMD12
Rasopathies	Disorders of the RAS/MAP Kinase Pathway	RAS01
Skeletal disorders	Metaphyseal dysplasia	SKT01
	Multiple epiphyseal dysplasia and pseudoachondroplasia	SKT02
	Spondylometaphyseal dysplasia and Spondylo-epi-(meta)-physeal dysplasia	SKT03
	Micromelic dysplasia: acromelic, acromesomelic, mesomelic and rhizo-mesomelic dysplasia	SKT04
	Short-rib dysplasia	SKT05
	Chondrodysplasia punctata	SKT06
	Osteogenesis imperfecta and related skeletal dysplasia with decreased bone density	SKT07
	Osteopetrosis and related skeletal dysplasia with increased bone density	SKT08
	Hypophosphatemic rickets and related skeletal dysplasia with abnormal mineralization	SKT09
	Limb malformations: isolated brachydactyly, synostoses, split-hand/foot, polydactyly, syndactyly, and selected genetic syndromes with limb malformations	SKT10
	Craniosynostosis	SKT11
	Potentially lethal skeletal disorders	SKT12
	Seckel syndrome, 3-M syndrome, Rubinstein–Taybi syndrome, Kabuki syndrome, and further selected genetic syndromes with skeletal involvement	SKT13

Panel	Gene set	Order No.
Focussed Panels	Afibrinogenemia / Dysfibrinogenemia	SSP01
	Common variable immunodeficiency (CVID)	SSP02
	Tuberous sclerosis	SSP03
	Hereditary breast and ovarian cancer (small)	SSP04
	Hereditary breast and ovarian cancer (large)	SSP05
	Lynch-Syndrom	SSP06
	Hereditary hemorrhagic telangiectasia	SSP07
	Neurofibromatosis	SSP08
	Hyperekplexia	SSP09
	Holoprosencephaly	SSP10
	Refsum disease	SSP11
	Episodic ataxia	SSP12
	Dopa-responsive dystonia	SSP13
	Neuropathic pain syndromes	SSP14
	Malignant hyperthermia	SSP15
	Familial intrahepatic cholestasis	SSP16
	Maple syrup urine disease	SSP17
	Maturity onset diabetes of the young (MODY)	SSP18
	Kabuki syndrome	SSP19
	Craniosynostoses	SSP20

Cancer and Tumor Diag.		
Cancer, Germline	Colorectal Cancer	CAN01
	Polyposis syndrome	CAN11
	Gastric Cancer	CAN13
	Cowden Syndrome	CAN14
	Pancreatic cancer	CAN06
	Breast and Ovarian Cancer	CAN02
	Breast and Ovarian Cancer - extended	CAN21
	Prostate Cancer	CAN03
	Pheochromocytoma and Paraganglioma	CAN04
	Other Familial Tumor Syndromes	CAN05
	Tumors of the Central Nervous System	CAN51
	Renal cell carcinoma	CAN07
	Xeroderma pigmentosum	CAN08
	Melanoma	CAN09
	Fanconi anemia	CAN10
Cancer, Somatic	Somatic Cancer - Treatment Decision Panel	TUM01
	Somatic Cancer - Exome-Sequencing	TUM02
Cancer, ovarian	BRCA1/2 analysis in tumor tissue or normal tissue (blood)	BRC01
	BRCA1/2 analysis in tumor tissue and normal tissue (blood)	BRC02

Panel	Gene set	Order No.
Prevention Panel		
Prevention Panel	Prevention Panel (tumor diseases, cardiovascular diseases, thrombosis and coagulation disorders, iron- and copper storage diseases, hypercholesteremia, glaucoma, pharmacogenetics and malignant hyperthermia)	PRV01
Whole Exome Analysis		
	Trio Exome Analysis	EXM03
	Additional Patient for Trio Exome Additional Evaluation ACMG Variants in Index patient Additional Evaluation ACMG Variants in Parents/Additional Patient, each:	
Array CGH		
Array CGH	Array CGH (180k)	CGH01