Deutsche Akkreditierungsstelle GmbH

Annex to the Accreditation Certificate D-ML-13242-01-00 according DIN EN ISO 15189:2014

Period of validity: 22.11.2018 to 16.08.2022 Date of issue: 24.01.2019

Holder of certificate:

MGZ Medizinisch Genetisches Zentrum GbR
Bayerstraße 3 - 5, 80335 München

Examinations in the fields:

Medical Laboratory Diagnostics

Medical laboratory fields:

Human genetics (human molecular genetics, cytogenetics)

Within the given type of examination marked with ** the medical laboratory is permitted, without being required to inform and obtain prior approval from DAkkS GmbH, the modification, development and refinement of examination procedures. The listed examination procedures are exemplary. The medical laboratory maintains a current list of all examination procedures within the flexible scope of accreditation.

Medical laboratory field: Human genetics (cytogenetics)

Type of examination:
Chromosome analysis **

Analyte (measurement parameter)	Test material (matrix)	Test technique
Chromosomal changes	Chromosome preparations	Staining (GTG)
extraction of chromosomes from cell culture	Fibroblasts	Chromosome analysis from fibroblasts
extraction of chromosomes from cell culture	Amniotic fluid	Chromosome analysis from amniotic fluid cells
extraction of chromosomes from cell culture	Chorionic villus tissue	Chromosome analysis from chorionic villus/placental tissue
extraction of chromosomes from cell culture	Miscarriage tissue	Chromosome analysis from miscarriage tissue
Identification of minute chromosomal/numerical changes and structurally changed chromosomes	Chromosome preparations	Chromosome analysis by fluorescence in situ hybridization (FISH)
Long-term culture of chorionic villi/placental cells	Chorionic villi, placental cells	Chromosome analysis from chorionic villus/placental tissue
Long-term culture of miscarriage tissue	Miscarriage tissue	Chromosome analysis from miscarriage tissue
Long-term culture of fibroblasts	Skin biopsy	Chromosome analysis from fibroblasts
Long-term culture of amniotic fluid	Amniotic fluid	Chromosome analysis from amniotic fluid cells
Detection of standard variants of chromosomes	Chromosome preparations	Staining (CBG)
Standard variants of the short arms of the chromosomes (nucleolus-organizing regions of the D and G groups of chromosomes)	Chromosome preparations	Staining (NOR)
Numerical chromosome aberrations	Buccal mucosa swab	Chromosome analysis by fluorescence in situ hybridization (FISH)
Numerical chromosome aberrations	Heparin blood, umbilical cord blood	Chromosome analysis by fluorescence in situ hybridization (FISH)
Numerical changes of chromosomes 13, 18, 21, X and Y	Fixed amniotic cell nuclei	Interphase analysis by FISH
Preparation of chromosomes from lymphocyte culture	Lymphocyte culture	Chromosome analysis from lymphocytes
Somatic chromosome mosaicism	Buccal mucosa swab	Chromosome analysis by fluorescence in situ hybridization (FISH)
T-lymphocyte culture	Sodium heparin blood, lithium heparin blood	Chromosome analysis from lymphocytes

Analyte (measurement parameter)	Test material (matrix)	Test technique
Chromosomal microarray analysis	Genomic DNA	Comparative genome hybridization using chromosomes with oligonucleotide-based array
Single cell diagnostics array CGH	Single cells	Comparative genomic hybridization (array CGH) using BAC clone-based Cytochip

Medical laboratory field: Human genetics (human molecular genetics)

Type of examination:
Molecular biological tests (amplification procedures)**

Analyte (measurement parameter)	Test material (matrix)	Test technique
Eye diseases		
Aniridia: *PAX6*	Extracted DNA	PCR, DNA sequencing, MLPA
Glaucoma: *CYP1B1, LTBP2*	Extracted DNA	PCR, DNA sequencing
Corneal dystrophies: ID172	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Isolated lens luxation: *ADAMTSL4, LTBP2*	Extracted DNA	PCR, DNA sequencing
Isolated lens luxation: *FBN1*	Extracted DNA	PCR, DNA sequencing, MLPA
Juvenile glaucoma basic: ID171	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Cataract - basic diagnostics: ID168	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Cataract: ID120	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Keratoconus: *VSX1*	Extracted DNA	PCR, DNA sequencing
Knobloch syndrome: *COL18A1*	Extracted DNA	PCR, DNA sequencing
Autosomal dominant optic atrophy: *OPA1*	Extracted DNA	PCR, DNA sequencing, MLPA
Autosomal dominant optic atrophy: *OPA3*	Extracted DNA	PCR, DNA sequencing
Autosomal recessive optic atrophy: *TMEM126A*	Extracted DNA	PCR, DNA sequencing

Analyte (measurement parameter)	Test material (matrix)	Test technique
Hereditary optic atrophy: ID125	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
LHON (Leber's hereditary optic neuropathy): MT-ND1, MT-ND2, MT-ND3,MT-ND4L, MT-ND4, MT-ND5, MT-ND6	Extracted DNA	PCR, DNA sequencing
Lens luxation: ID123	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Lens luxation basic: ID170	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Microphthalmia-anophthalmia-coloboma complex (MAC): *OTX2, GDF6, RAX, MAB21L2*	Extracted DNA	PCR, DNA sequencing
Microphthalmia-anophthalmia-coloboma complex (MAC): *SOX2*	Extracted DNA	PCR, DNA sequencing, MLPA
Microphthalmia-anophthalmia-coloboma complex (MAC)_basic: ID167	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Microphthalmia-anophthalmia-coloboma complex (MAC)_complete: ID119	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Ocular/oculocutaneous albinism: ID124	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Optic atrophy with mental retardation, Bosch-Boonstra optic atrophy: *NR2F1*	Extracted DNA	PCR, DNA sequencing
Peters-plus syndrome: *B3GALTL (B3GLCT)*	Extracted DNA	PCR, DNA sequencing
Stickler syndrome/high myopia: ID122	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Stickler syndrome: *COL2A1, COL11A1*	Extracted DNA	PCR, DNA sequencing, MLPA
Stickler syndrome basic: ID169	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Usher syndrome: *HARS*	Extracted DNA	PCR, DNA sequencing
Anterior chamber dysgenesis / Axenfeld-Rieger syndrome: *FOXC1, PITX2*	Extracted DNA	PCR, DNA sequencing, MLPA
Anterior chamber dysgenesis / Axenfeld-Rieger syndrome: ID121	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)

Analyte (measurement parameter)	Test material (matrix)	Test technique
X-chromosomal congenital nystagmus: *FRMD7*	Extracted DNA	PCR, DNA sequencing, MLPA
Vascular and connective tissue diseases		
Adenosine deaminase 2 (ADA2) deficiency: *CECR1*	Extracted DNA	PCR, DNA sequencing
Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL): *NOTCH3*	Extracted DNA	PCR, DNA sequencing
Cerebral autosomal recessive arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL): *HTRA1*	Extracted DNA	PCR, DNA sequencing
Cerebral microangiopathy / porencephaly / apoplexy: *COL4A1, COL4A2*	Extracted DNA	PCR, DNA sequencing
Chilblain lupus erythematosus, retinal vasculopathy with cerebral leukodystrophy: *TREX1*	Extracted DNA	PCR, DNA sequencing
Collagen type IV-associated diseases and phenocopies: ID129	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Ehlers-Danlos syndrome (EDS): ID130	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Ehlers-Danlos syndrome, classic type: ID282	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Ehlers-Danlos syndrome, vascular type: ID173	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Familial thoracic aortic aneurysm: *FBN1, TGFBR1, TGFBR2*	Extracted DNA	PCR, DNA sequencing, MLPA
Vascular and connective tissue diseases total panel ID175	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Marfan syndrome and type 1 fibrillinopathy: ID174	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Fabry disease: *GLA*	Extracted DNA	PCR, DNA sequencing, MLPA
Osler's disease, Osler-Rendu-Weber syndrome: *GDF2*	Extracted DNA	PCR, DNA sequencing
Osler's disease, Osler-Rendu-Weber syndrome: *ACVRL1, ENG*	Extracted DNA	PCR, DNA sequencing, MLPA

Analyte (measurement parameter)	Test material (matrix)	Test technique
Thoracic aortic aneurysms and aortic dissections (TAAD): ID127	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
MELAS: mt-tRNALeu (m.3243A>G), mt-tRNAs	Extracted DNA	PCR, DNA sequencing
Juvenile stroke/stroke-like episodes: ID128	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Thoracic aortic aneurysms and aortic dissections (TAAD): *ACTA2*	Extracted DNA	PCR, DNA sequencing
TREX1-*associated diseases: TREX1*	Extracted DNA	PCR, DNA sequencing
Cerebral hemorrhage with cataract: *JAM3*	Extracted DNA	PCR, DNA sequencing
Neurogenetics		
Movement disorders/ataxia		
Episodic ataxia type 2: *CACNA1A, KCNA1*	Extracted DNA	PCR, DNA sequencing, MLPA
Episodic ataxia - basic diagnostics: ID226	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Episodic ataxia and phenocopies: ID090	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Glut1 deficiency syndrome: *SLC2A1*	Extracted DNA	PCR, DNA sequencing, MLPA
Fragile X-associated tremor ataxia syndrome FAXTAS: *FMR1* - by premutant PCR	Extracted DNA	PCR, size-specific DNA fragment analysis
Ataxia, dominant		
Spinocerebellar ataxia type 1, 2, 3 (SCA1,2,3): ATXN1,2,3	Extracted DNA	PCR, size-specific DNA fragment analysis
Spinocerebellar ataxia type 6 (SCA6): *CACNA1A*	Extracted DNA	PCR, size-specific DNA fragment analysis
Spinocerebellar ataxia 7 *(SCA7): ATXN7*	Extracted DNA	PCR, size-specific DNA fragment analysis
Spinocerebellar ataxia 8 (SCA8): ATXN8	Extracted DNA	PCR, size-specific DNA fragment analysis
Spinocerebellar ataxia 17 (SCA17): *TBP*	Extracted DNA	PCR, size-specific DNA fragment analysis
Dentatorubral-pallidoluysian atrophy (DRPLA): *ATN1*	Extracted DNA	PCR, size-specific DNA fragment analysis
Ataxia - dominant - after repeat-associated SCA: ID569	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Ataxia with oculomotor apraxia (AOA1, AOA2): *APTX, SETX*	Extracted DNA	PCR, DNA sequencing, MLPA
Ataxia - total panel: ID149	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Spinocerebellar ataxia 13 (SCA13): *KCNC3*	Extracted DNA	PCR, DNA sequencing
Spinocerebellar ataxia 18 (SCA18): *IFRD1*	Extracted DNA	PCR, DNA sequencing
Alexander disease: *GFAP*	Extracted DNA	PCR, DNA sequencing
Cerebral microangiopathy / porencephaly / apoplexy: *COL4A1, COL4A2*	Extracted DNA	PCR, DNA sequencing
Leukocytopenia with brainstem and spinal cord involvement and elevated lactate values: *DARS2*	Extracted DNA	PCR, DNA sequencing
Ataxia, recessive		
Friedreich's ataxia: *FRDA*	Extracted DNA	PCR, size-specific DNA fragment analysis
Ataxia - recessive, with sensory neuropathy: ID283	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Ataxia - recessive, with spasticity: ID284	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Ataxia, with oculomotor apraxia: ID223	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Ataxia telangiectasia, ATM gene: ID224	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Ataxia, autosomal recessive - basic diagnostics: ID092	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Ataxia, autosomal recessive: ID225	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Charlevoix-Saguenay ataxia: *SACS*	Extracted DNA	PCR, DNA sequencing, MLPA
Spinocerebellar ataxia type 10, autosomal recessive (SCAR10): *ANO10*	Extracted DNA	PCR, DNA sequencing

Analyte (measurement parameter)	Test material (matrix)	Test technique
Movement disorders / spastic paraparesis		
Hereditary spastic paraparesis basic diagnostics: ID147	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Hereditary spastic paraparesis (HSP): ID148	Extracted DNA	PCR, Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Spastic paraparesis type 53: *VPS37A (p.Lys382Asn)*	Extracted DNA	PCR, DNA sequencing
Spastic paraparesis: *ATL1, SPAST, CYP7B1, SPG7, SPG11*	Extracted DNA	PCR, DNA sequencing, MLPA
Movement disorders/dystonia/dyskinesia		
Dystonia, isolated - basic diagnostics: ID227	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Dystonia with myoclonus: ID228	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Dystonia (DYT1, DYT5A, DYT5B, DYT6): *TOR1A, GCH1, TH, THAP1*	Extracted DNA	PCR, DNA sequencing, MLPA
Myoclonic dystonia: *SGCE*	Extracted DNA	PCR, DNA sequencing, MLPA
Dystonia with parkinsonism and/or DOPA-responsive: ID229	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Dyskinesia with facial myocymia: *ADCY5*	Extracted DNA	PCR, DNA sequencing
Dyskinesia, episodic kinesigenic: *PRRT2*	Extracted DNA	PCR, DNA sequencing
Dyskinesia, paroxysmal/with dystonia: ID230	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Dystonia, dyskinesia - total panel: ID231	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Movement disorders/chorea		
Huntington's disease: *HTT*	Extracted DNA	PCR, size-specific DNA fragment analysis
Spinocerebellar ataxia 17 (SCA17): *TBP*	Extracted DNA	PCR, size-specific DNA fragment analysis
Dentatorubral-pallidoluysian atrophy (DRPLA): *ATN1*	Extracted DNA	PCR, size-specific DNA fragment analysis
Choreiform movement disorder: ID232	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Movement disorders/Parkinson's disease		
Parkinson's disease, autosomal dominant or x-linked: ID234	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Parkinson's disease: ID235	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Dementia/neurodegeneration/leukodystrophy		
Apolipoprotein E4 allele in Alzheimer's disease: *ApoE*	Extracted DNA	PCR, DNA sequencing
Huntington's disease: *HTT*	Extracted DNA	PCR, size-specific DNA fragment analysis
Chorea acanthocytosis: *VPS13A*	Extracted DNA	PCR, DNA sequencing, MLPA
Dementia, familial of Alzheimer type: *APP, PSEN1*	Extracted DNA	PCR, DNA sequencing, MLPA
Dementia, familial of Alzheimer type: *PSEN2*	Extracted DNA	PCR, DNA sequencing
Dementia: ID238	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Frontotemporal dementia, FTD-ALS: *C9orf72* -repeat	Extracted DNA	PCR, size-specific DNA fragment analysis
Leukodystrophy - comprehensive diagnostics: ID241	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Leukodystrophy, with hypomyelination - basic diagnostics: ID239	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Leukodystrophy, with hypomyelination: ID240	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
McLeod syndrome: *XK*	Extracted DNA	PCR, DNA sequencing
Neuroacanthocytosis: ID236	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Neurodegeneration with iron storage in brain (NBIA): ID080	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Prion disease: *PRNP* -repeat	Extracted DNA	PCR, DNA sequencing, size-specific DNA fragment analysis
Riboflavin transporter deficiency (Brown-Vialetto-van Laere syndrome): *SLC52A2, SLC52A3*	Extracted DNA	PCR, DNA sequencing

Analyte (measurement parameter)	Test material (matrix)	Test technique
Epilepsy		
Childhood absence epilepsy (pyknolepsie): *CACNA1H*	Extracted DNA	PCR, DNA sequencing
Neonatal siezure - basic diagnostics: ID242	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Benign familial neonatal + infantile epilepsy: *KCNQ2, KCNQ3*	Extracted DNA	PCR, DNA sequencing, MLPA
Benign familial neonatal + infantile epilepsy: *PRRT2, SCN2A*	Extracted DNA	PCR, DNA sequencing
Dravet syndrome: *SCN1A*	Extracted DNA	PCR, DNA sequencing, MLPA
Fever-related seizures: ID146	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Focal epilepsy with speech disorder: *GRIN2A*	Extracted DNA	PCR, DNA sequencing, MLPA
Glut1 deficiency syndrome: *SLC2A1*	Extracted DNA	PCR, DNA sequencing, MLPA
Benign familial neonatal and infantile epilepsy: ID003	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Familial focal epilepsy: ID004	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Epilepsy and hypomyelination: ID005	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Progressive myoclonus epilepsy: ID008	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Epilepsy - total panel: ID085	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
X-chromosomal West syndrome: *ARX*	Extracted DNA	PCR, DNA sequencing, long-range TP-PCR and size-specific DNA fragment analysis, MLPA
Rett-like syndrome: *CDKL5*	Extracted DNA	PCR, DNA sequencing, MLPA
Unverricht-Lundborg disease: *CSTB*	Extracted DNA	PCR, DNA sequencing, size-specific DNA fragment analysis

Analyte (measurement parameter)	Test material (matrix)	Test technique
Generalized epilepsy with fever siezure plus (GEFS +): *SCN1A*	Extracted DNA	PCR, DNA sequencing, MLPA
Generalized epilepsy with fever siezure plus (GEFS +): *GABRA1, GABRD, GABRG, SCN1B, SCN2A, STX1B*	Extracted DNA	PCR, DNA sequencing
Juvenile myoclonus epilepsy: *EFHC1*	Extracted DNA	PCR, DNA sequencing
Lafora epilepsy: *EPM2A*	Extracted DNA	PCR, DNA sequencing, MLPA
Lafora epilepsy: *NHLRC1*	Extracted DNA	PCR, DNA sequencing
MERRF: mt-tRNALys (m.8344A>G), mt-tRNAs	Extracted DNA	PCR, DNA sequencing
Phophoglycerate dehydrogenase deficiency: *PHGDH*	Extracted DNA	PCR, DNA sequencing
POLG-associated epilepsy: *POLG*	Extracted DNA	PCR, DNA sequencing, MLPA
Progressive myoclonus epilepsy: *GOSR2*	Extracted DNA	PCR, DNA sequencing
Pyridoxine-dependent epilepsy: *ALDH7A1*	Extracted DNA	PCR, DNA sequencing
Pyridoxine phosphate oxidase deficiency: *PNPO*	Extracted DNA	PCR, DNA sequencing
Cerebral folate receptor deficiency: *FOLR1*	Extracted DNA	PCR, DNA sequencing
Epileptic encephalopathies		
Encephalopathies (mitochondrial/epileptic): ID265	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Epileptic encephalopathy -basic diagnostics - ID001	Extracted DNA	PCR, Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Epileptic encephalopathy: *CHD2*	Extracted DNA	PCR, DNA sequencing
Exom analysis by panel ID265: ID165	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Early childhood epilepsy - basic diagnostics: ID243	Extracted DNA	PCR, Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Infantile epileptic encephalopathy (EIEE2): *CDKL5*	Extracted DNA	PCR, DNA sequencing, MLPA
Infantile epileptic encephalopathy (EIEE4): *STXBP1*	Extracted DNA	PCR, DNA sequencing, MLPA

Analyte (measurement parameter)	Test material (matrix)	Test technique
Infantile epileptic encephalopathy: *GABRA1, DOCK7, HCN1, KCNT1, SLC25A22*	Extracted DNA	PCR, DNA sequencing
Infantile epilepsy with mental retardation (girls only): *PCDH19*	Extracted DNA	PCR, DNA sequencing, MLPA
Necrotizing encephalopathy (ANE): *RANBP2*	Extracted DNA	PCR, DNA sequencing
Neuronal ceroid lipofuscinosis: *PPT1*	Extracted DNA	PCR, DNA sequencing
Migraine/paroxysmal neurological disorders		
Familial hemiplegic migraine: ID150	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Familial hemiplegic migraine: *ATP1A2*	Extracted DNA	PCR, DNA sequencing, MLPA
Alternating childhood hemiplegia: *ATP1A3*	Extracted DNA	PCR, DNA sequencing, MLPA
Familial hemiplegic migraine: *CACNA1A, SCN1A*	Extracted DNA	PCR, DNA sequencing, MLPA
Paroxysmal stress-dependent dystonia: *SLC2A1*	Extracted DNA	PCR, DNA sequencing, MLPA
Hyperekplexia: ID151	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Neuromuscular diseases		
Fetal akinesia/arthrogryposis multiplex congenita (AMC)		
Arthrogryposis multiplex congenita basic panel: ID581	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Fetal akinesia/arthrogryposis multiplex congenita (AMC) ID078	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Fetal akinesia: *CHRNA1, CHRNB1, CHRND, DOK7, RAPSN*	Extracted DNA	PCR, DNA sequencing
Fetal akinesia: *CHRNG*	Extracted DNA	PCR, DNA sequencing
Malignant hyperthermia		
Malignant hyperthermia, susceptibility: ID076	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Malignant hyperthermia: *CACNA1S, RYR1*	Extracted DNA	PCR, DNA sequencing, MLPA

Analyte (measurement parameter)	Test material (matrix)	Test technique
Myotonia/myotonic dystrophy		
Myotonic dystrophy type 1 (DM1): *DMPK*- repeat	Extracted DNA	PCR, size-specific DNA fragment analysis
Myotonic dystrophy type 2 (DM2, PROMM): *ZNF9*- repeat	Extracted DNA	PCR, size-specific DNA fragment analysis
Myotonia, chloride channel-associated: *CLCN1*	Extracted DNA	PCR, DNA sequencing, MLPA
Myotonia, paramyotonia, sodium channel-associated: *SCN4A*	Extracted DNA	PCR, DNA sequencing, MLPA
Schwartz-Jampel syndrome, type 1: ID245	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Neuropathy with neuromyotonia: *HINT1*	Extracted DNA	PCR, DNA sequencing
Myotonia - basic diagnostics: ID244	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
CAV3 -associated myotonia: *CAV3*	Extracted DNA	PCR, DNA sequencing
Periodic paralysis		
Andersen-Tawil syndrome: *KCNJ2*	Extracted DNA	PCR, DNA sequencing, MLPA
Periodic paralysis: CACNA1S, RYR1, SCN4A	Extracted DNA	PCR, DNA sequencing, MLPA
Periodic paralysis - ID077	Extracted DNA	PCR, Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Congenital myasthenic syndrome/neonatal apnea		
CMS - congenital myasthenic syndromes - basic diagnostics: ID246	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Congenital myasthenic syndromes: ID33	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Congenital myasthenic syndromes: *CHAT, CHRNE, CHRNA1, CHRNB1, COLQ, DOK7, RAPSN*	Extracted DNA	PCR, DNA sequencing
Congenital myasthenic syndromes: *AGRN, ALG2, ALG14, DPAGT1, GFPT1, MUSK, SYT2*	Extracted DNA	PCR, DNA sequencing
Congenital myasthenic syndromes: *SCN4A*	Extracted DNA	PCR, DNA sequencing, MLPA
Congenital central hypoventilation: *PHOX2B*	Extracted DNA	PCR, DNA sequencing, size-specific DNA fragment analysis, MLPA
Neonatal apneas: ID038	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Myopathies/muscular dystrophies		
Congenital myopathies, core myopathy: *RYR1*	Extracted DNA	PCR, DNA sequencing, MLPA
Congenital myopathies, core myopathy: *CCDC78, MEGF10, SEPN1*	Extracted DNA	PCR, DNA sequencing
Muscle weakness with early manifestation (infancy to childhood): ID037	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Muscle weakness with late manifestation (adulthood): ID045	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Myopathy/muscular dystrophy: ID089	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Oculopharyngeal muscular dystrophy (OPMD): *PABPN1* -repeat	Extracted DNA	PCR, DNA sequencing, size-specific DNA fragment analysis
Tubular aggregate myopathy, Stormorken syndrome: *STIM1*	Extracted DNA	PCR, DNA sequencing
Limb girdle dystrophies/distal myopathies		
Muscular dystrophy Duchenne/Becker-Kiener - ID020 (DMD)	Extracted DNA	PCR, Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Muscular dystrophy Duchenne/Becker-Kiener: *DMD*	Extracted DNA	PCR, DNA sequencing, MLPA
Limb girdle muscular dystrophies / LGMD (non-DMD-associated): ID247	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Muscular dystrophy limb girdle dystrophy: ANO5, CAPN3, DYSF, FKRP, MYOT, PLEC, SGCA, SGCB, SGCD	Extracted DNA	PCR, DNA sequencing, MLPA
Muscular dystrophy limb girdle dystrophy: *CAV3, DES, DNAJB6, TCAP, TRIM32*	Extracted DNA	PCR, DNA sequencing
Muscular dystrophy limb girdle dystrophy: *LMNA*	Extracted DNA	PCR, DNA sequencing, MLPA
Emery-Dreifuss muscular dystrophy: ID022	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Inclusion body myopathy/distal myopathy: ID574	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Myofibrillar myopathy: ID575	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Myofibrillar myopathy: *CRYAB, DES, DNAJB6, FHL1, FLNC, LDB3*	Extracted DNA	PCR, DNA sequencing
Myofibrillar myopathy: *BAG3, MYOT*	Extracted DNA	PCR, DNA sequencing, MLPA
Guiding symptom distal myopathy: ID047	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Congenital muscular dystrophies/structural myopathies		
Congenital muscular dystrophy - collagen6-associated (Bethlem/Ullrich): ID249	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Congenital muscular dystrophy (CMD): *CHKB, COL6A1, COL6A2, COL6A3, COL12A1, SEPN1*	Extracted DNA	PCR, DNA sequencing
Congenital muscular dystrophy (CMD): *LMNA*	Extracted DNA	PCR, DNA sequencing, MLPA
Congenital muscular dystrophy (CMD): *FKRP, LAMA2*	Extracted DNA	PCR, DNA sequencing, MLPA
Nemaline myopathy: *ACTA1, CFL2, KBTBD13, KLHL40, KLHL41, NEB, TNNT1, TPM2, TPM3*	Extracted DNA	PCR, DNA sequencing
Nemaline myopathy - NEB: ID329	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
RYR gene: ID257	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Centronuclear myopathy: ID028	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Structural myopathies - nemaline/core/centronuclear: ID026	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Guiding symptom contractures and/or rigid spine: ID043	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Congenital muscular dystrophy: ID251	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Fiber type disproportions: *ACTA1, MYL2, SEPN1, TPM2, TPM3*	Extracted DNA	PCR, DNA sequencing
Fiber type disproportions: *MYH7, RYR1*	Extracted DNA	PCR, DNA sequencing, MLPA
Centronuclear myopathy: *MTM1, RYR1*	Extracted DNA	PCR, DNA sequencing, MLPA
Centronuclear myopathy: *BIN1, DNM2*	Extracted DNA	PCR, DNA sequencing

Analyte (measurement parameter)	Test material (matrix)	Test technique
Inclusion body myopathy: *DES, GNE, MYH2, VCP*	Extracted DNA	PCR, DNA sequencing
Muscular dystrophy Emery-Dreifuss: *LMNA*	Extracted DNA	PCR, DNA sequencing, MLPA
Muscular dystrophy Emery-Dreifuss: *EMD*	Extracted DNA	PCR, DNA sequencing
Facioscapulohumeral muscular dystrophy		
FSHD permissive haplotype: 4qA161 haplotype	Extracted DNA	PCR, DNA sequencing
Facioscapulohumeral muscular dystrophy (FSHD2): *SMCHD1*	Extracted DNA	PCR, DNA sequencing
FSHD2 and FSHD1 phenocopies: ID252	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Chronic progressive external ophthalmoplegia, CPEO		
Analysis for mtDNA deletions	Extracted DNA	PCR, long-range TP-PCR and size-specific DNA fragment analysis
Chronic progressive external ophthalmoplegia (CPEO), autosomal dominant: ID253	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Chronic progressive external ophthalmoplegia (CPEO), autosomal recessive: ID254	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
CPEO - chronic progressive external ophthalmoplegia (dominant, recessive and phenocopies): ID255	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Metabolic myopathies		
CPT2 deficiency: *CPT2*	Extracted DNA	PCR, DNA sequencing
Glutaric aciduria type 2: *ETFDH*	Extracted DNA	PCR, DNA sequencing
Myoadenylate deaminase (MAD) deficiency: *AMPD1 (p.Gln45*)*	Extracted DNA	PCR, DNA sequencing
Myopathy with lipid storage disease - *CPT2, ETFDH, PNPLA2*	Extracted DNA	PCR, DNA sequencing
McArdle: *PYGM*	Extracted DNA	PCR, DNA sequencing
Pompe disease: *GAA*	Extracted DNA	PCR, DNA sequencing, MLPA
Metabolic myopathy / fatty acid oxidation disorder - basic diagnostics: ID256	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Metabolic myopathy / fatty acid oxidation disorder / myalgia / rhabdomyolysis: ID025	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Rhabdomyolysis, stress-intolerance: *LPIN1, MT-CYB*	Extracted DNA	PCR, DNA sequencing
Visceral myopathy		
Visceral myopathy: *ACTG2*	Extracted DNA	PCR, DNA sequencing
Neuropathies/motor neuron diseases		
Neuropathy, early childhood manifestation: ID016	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Neuropathy - total panel: ID086	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Neuropathies / CMT motor and sensitive		
Neuropathy, demyelinating: *PMP22 - dose*	Extracted DNA	MLPA
Neuropathy, motor-sensitive, demyelinating / CMT1 - basic diagnostics: ID286	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Neuropathy, motor-sensitive, axonal / CMT2 - basic diagnostics: ID287	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Neuropathy, motor / dHMN - basic diagnostics: ID258	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Neuropathy, motor-sensitive / CMT1, CMT2, dHMN comprehensive diagnostics: ID288	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
HMSN1, predominantly demyelinating: *MPZ*	Extracted DNA	PCR, DNA sequencing
HMSN1, predominantly demyelinating: *GJB1 (Cx32)*	Extracted DNA	PCR, DNA sequencing, MLPA
HMSN1, predominantly demyelinating: *FGD4, FIG4, PRPS1*	Extracted DNA	PCR, DNA sequencing
HMSN1, predominantly demyelinating: *SH3TC2*	Extracted DNA	PCR, DNA sequencing, MLPA
HMSN1, predominantly demyelinating: *LITAF (SIMPLE), NDRG1*	Extracted DNA	PCR, DNA sequencing
HMSN1, predominantly demyelinating: *EGR2*	Extracted DNA	PCR, DNA sequencing, MLPA

Analyte (measurement parameter)	Test material (matrix)	Test technique
HMSN1, predominantly demyelinating: *MTMR2,PRX*	Extracted DNA	PCR, DNA sequencing, MLPA
HMSN1, predominantly demyelinating with glaucoma: *SBF2*	Extracted DNA	PCR, DNA sequencing, MLPA
HMSN1, predominantly demyelinating with cataract: *FAM126A*	Extracted DNA	PCR, DNA sequencing
HMSN1, predominantly demyelinating with maculopathy: *FBLN5*	Extracted DNA	PCR, DNA sequencing
HMSN2, predominantly axonal: *MFN2*	Extracted DNA	PCR, DNA sequencing, MLPA
HMSN2, predominantly axonal: *GDAP1 LMNA, NEFL, RAB7A*	Extracted DNA	PCR, DNA sequencing, MLPA
HMSN2, predominantly axonal: *AARS, DYNC1H1, HSPB1, HSPB3, HSPB8, LRSAM1, MARS, MED25 , TRPV4*	Extracted DNA	PCR, DNA sequencing
HMSN2, predominantly axonal: *BSCL2* (Exon3)	Extracted DNA	PCR, DNA sequencing
HMSN, intermediate: *DNM2*	Extracted DNA	PCR, DNA sequencing
HMSN, intermediate: *KARS, YARS*	Extracted DNA	PCR, DNA sequencing
HMSN, dominant intermdiary (CMTDIE): *INF2*	Extracted DNA	PCR, DNA sequencing
HMSN with optic atrophy: *SLC25A46*	Extracted DNA	PCR, DNA sequencing
Hereditary sensory neuropathy with spastic paraplegia: *CCT5*	Extracted DNA	PCR, DNA sequencing
Neuromyotonia and axonal neuropathy: *HINT1*	Extracted DNA	PCR, DNA sequencing
Distal hereditary motoneuropathies (dHMN): *GARS*	Extracted DNA	PCR, DNA sequencing, MLPA
Distal hereditary motoneuropathies (dHMN): *SETX* (AOA2)	Extracted DNA	PCR, DNA sequencing, MLPA
Distal hereditary motoneuropathies (dHMN): *DCTN1*	Extracted DNA	PCR, DNA sequencing
Erythermalgia, pain tolerance: *SCN9A*	Extracted DNA	PCR, DNA sequencing
Peripheral neuropathy with myopathy, hoarseness and hearing loss (PNMHH): *MYH14*	Extracted DNA	PCR, DNA sequencing

Analyte (measurement parameter)	Test material (matrix)	Test technique
Familial episodic pain syndrome: *SCN10A*	Extracted DNA	PCR, DNA sequencing
Slowed nerve conduction velocity, autosomal dominant: *ARHGEF10*	Extracted DNA	PCR, DNA sequencing
Giant axon neuropathy (GAN1): *GAN*	Extracted DNA	PCR, DNA sequencing
Neuropathies / HSAN - sensitive		
Neuropathy, sodium channel-associated: ID289	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Hereditary sensory-autonomic neuropathy (HSAN1A): *SPTLC1*	Extracted DNA	PCR, DNA sequencing, MLPA
Hereditary sensory autonomic neuropathy (HSAN1C, HSN1E, HSAN2, HSAN2B, HSAN2C, HSAN4, HSAN5, HSAN7): *SPTLC2, DNMT1, WNK1, FAM134B, KIF1A, NTRK1, NGFB (NGF), SCN11A*	Extracted DNA	PCR, DNA sequencing
Neuropathy, sensory / HSAN1 and HSAN2 ID259	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Neuropathy, sensory - familial dysautonomy / HSAN3 and HSAN4: ID260	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Neuropathy, sensory (autonomous) - HS(A)N - comprehensive diagnostics: ID015	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Hereditary neuropathy with pressure paralysis (HNPP): *PMP22* (deletion, point mutation)	Extracted DNA	PCR, DNA sequencing, MLPA
Hereditary neuralgic amyotrophy (HNS): *SEPT9*	Extracted DNA	PCR, DNA sequencing, MLPA
Amyloid polyneuropathy: *TTR*	Extracted DNA	PCR, DNA sequencing
Congenital cataract facial dysmorphism-neuropathy syndromes: *CTDP1*	Extracted DNA	PCR, DNA sequencing
Spinal / spinobulbar muscular atrophy		
Spinobulbar muscular atrophy type Kennedy (SBMA, SMAX1): *AR*	Extracted DNA	PCR, size-specific DNA fragment analysis
Spinal muscular atrophy type 0-IV: *SMN1*	Extracted DNA	MLPA
SMN1 gene point mutations: ID578	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Myotonic dystrophy type 1 (DM1): *DMPK*- repeat	Extracted DNA	PCR, size-specific DNA fragment analysis

Analyte (measurement parameter)	Test material (matrix)	Test technique
Prader-Willi syndrome: Methylation status and deletion/duplication test	Extracted DNA	MS-MLPA (methylation-sensitive)
Differential diagnoses of infant SMA: ID261	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Structural myopathy - basic diagnostics infant: ID294	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Differential diagnoses of adult SMA: ID262	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Motoneuropathy proximal / spinal muscular atrophy: ID152	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Spinal muscular atrophy, X-linked, infantile (SMAX2): *UBA1*	Extracted DNA	PCR, DNA sequencing
Spinal muscular atrophy, lower extremity dominant (SMA-LED): *DYNC1H1*	Extracted DNA	PCR, DNA sequencing
Spinal muscular atrophy with respiratory distress (SMARD1): *IGHMBP1*	Extracted DNA	PCR, DNA sequencing, MLPA
Spinal muscular atrophy, dominant: *BICD2, TRPV4, VAPB*	Extracted DNA	PCR, DNA sequencing
Amyotrophic lateral sclerosis		
Amyotrophic lateral sclerosis: *C9orf72*-repeat	Extracted DNA	PCR, size-specific DNA fragment analysis
Amyotrophic lateral sclerosis - basic diagnostics: ID263	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Amyotrophic lateral sclerosis: ID019	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Amyotrophic lateral sclerosis: *ANG, CHCHD10, FIG4, FUS, SOD1, TARDBP, UBQLN2, VAPB*	Extracted DNA	PCR, DNA sequencing
Mitochondrial diseases		
Mitochondrial encephalopathy / hepatomyopathy		
Leigh/Leigh-like syndrome (primary molecular genetic: *BCS1L, C12orf65, COX6B1, FASTKD2, MT-ATP6, SURF1*	Extracted DNA	PCR, DNA sequencing
Leigh syndrome basic diagnostics nucleic gene defects: ID266	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Leigh/Leigh-like syndrome (primary molecular genetic: *PDHA1*	Extracted DNA	PCR, DNA sequencing, MLPA
Leigh/Leigh-like syndrome (primary molecular genetic: *ECHS1, SLC19A3*	Extracted DNA	PCR, DNA sequencing
Mitochondrial hepato (encephalopathy) - basic diagnostics: ID264	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Alpers syndrome: *POLG*	Extracted DNA	PCR, DNA sequencing, MLPA
Early childhood (hepato) encephalomyopathy: *C10orf2 (TWNK, PEO1)*	Extracted DNA	PCR, DNA sequencing
Early childhood (hepato) encephalomyopathy: *DGUOK, SUCLA2, TK2*	Extracted DNA	PCR, DNA sequencing, MLPA
Early childhood (hepato) encephalomyopathy, hepatic form: *MPV17*	Extracted DNA	PCR, DNA sequencing, MLPA
Early childhood (hepato) encephalomyopathy, myopathic form: *RRM2B*	Extracted DNA	PCR, DNA sequencing, MLPA
Early childhood (hepato) encephalomyopathy, encephalomyopathic form: *SUCLG1*	Extracted DNA	PCR, DNA sequencing, MLPA
CoQ10 deficiency: ID054	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Coenzyme Q10 defects: *APTX*	Extracted DNA	PCR, DNA sequencing, MLPA
Coenzyme Q10 defects: *COQ2, COQ8A (ADCK3, CABC1), COQ9, ETFA, ETFB, ETFDH, PDSS1, PDSS2*	Extracted DNA	PCR, DNA sequencing
Coenzyme Q10 defects: *COQ6*	Extracted DNA	PCR, DNA sequencing
Mitochondriopathies - total panel: ID087	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Complete sequencing of mtDNA	Extracted DNA	PCR, DNA sequencing
Encephalopathies (mitochondrial/epileptic): ID265	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Exom analysis by panel ID265: ID165	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Mitochondrial myopathy		
mtDNA deletion/s	Extracted DNA	PCR, long-range TP-PCR and size-specific DNA fragment analysis
Mitochondrial myopathy: ID069	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Mitochondrial syndromes		
Hydroxyacyl-CoA dehydrogenase deficiency: *HADH, HADHA, HADHB*	Extracted DNA	PCR, DNA sequencing
MELAS: mt-tRNALeu (m.3243A>G), mt-tRNAs	Extracted DNA	PCR, DNA sequencing
MERRF: mt-tRNALys (m.8344A>G), mt-tRNAs	Extracted DNA	PCR, DNA sequencing
Leber's hereditary optic neuropathy (LHON)*: MT-ND1, MT-ND2, MT-ND3, MT-ND4L, MT-ND4, MT-ND5, MT-ND6*	Extracted DNA	PCR, DNA sequencing
Neuropathy, ataxia, retinitis pigmentosa (NARP): *MT-ATP6, MT-ATP8*	Extracted DNA	PCR, DNA sequencing
Kearns-Sayre syndrome/CPEO: mtDNA from muscle DNA	Extracted DNA	PCR, DNA sequencing
Kearns-Sayre syndrome/CPEO: *SLC25A4 (ANT1), C10orf2 (TWNK, PEO1)*	Extracted DNA	PCR, DNA sequencing
Kearns-Sayre syndrome/CPEO: *OPA1, POLG, POLG2, RRM2B, TK2*	Extracted DNA	PCR, DNA sequencing, MLPA
Kearns-Sayre syndrome/CPEO: *MGME1*	Extracted DNA	PCR, DNA sequencing
Congenital muscular dystrophy with enlarged mitochondria: *CHKB*	Extracted DNA	PCR, DNA sequencing
Mitochondrial cardiomyopathy: *COX15, MT-ATP6, MT-ATP8,* mt-tRNAs*, SCO2, SLC25A4, TMEM70*	Extracted DNA	PCR, DNA sequencing
Mitochondrial cardiomyopathy: *AGK, CHKB, SLC25A3*	Extracted DNA	PCR, DNA sequencing
Mitochondrial cardiomyopathy: *SDHA*	Extracted DNA	PCR, DNA sequencing, MLPA
Detection of point mutations of mtDNA from muscle DNA	Extracted DNA	PCR, DNA sequencing
MNGIE/mitochondrial neurogastrointestinal encephalomyopathy: *TYMP*	Extracted DNA	PCR, DNA sequencing

Analyte (measurement parameter)	Test material (matrix)	Test technique
Mitochondrial translation defect: *GFM1 (EGF1), MRPS16,* mt-tRNAs, *TRMU, TSFM, TUFM*	Extracted DNA	PCR, DNA sequencing
Mitochondrial translation defect: *AARS, DARS2, EARS2, MTFMT, MTO1, PUS1, YARS*	Extracted DNA	PCR, DNA sequencing
Multiple acyl-CoA dehydrogenase defects: *ETFA, ETFB, ETFDH*	Extracted DNA	PCR, DNA sequencing
Multiple acyl-CoA dehydrogenase defects: *ACADVL*	Extracted DNA	PCR, DNA sequencing, MLPA
Multiple acyl-CoA dehydrogenase defects: *ACADS, ACADM*	Extracted DNA	PCR, DNA sequencing
Rhabdomyolysis, stress-intolerance: *CPT2*	Extracted DNA	PCR, DNA sequencing
Rhabdomyolysis, stress-intolerance: *ACADVL*	Extracted DNA	PCR, DNA sequencing, MLPA
Sensory ataxic neuropathy, dysarthia, ophthalmoplegia (SANDO): *POLG*	Extracted DNA	PCR, DNA sequencing, MLPA
Sensory ataxic neuropathy, dysarthia, ophthalmoplegia (SANDO): *C10orf2 (TWNK, PEO1)*	Extracted DNA	PCR, DNA sequencing
Thiamine-responsive megaloblastic anaemia (TRMA): *SLC19A2*	Extracted DNA	PCR, DNA sequencing
Wolfram syndrome (DIDMOAD): *WFS1*	Extracted DNA	PCR, DNA sequencing, MLPA
Wolfram syndrome (DIDMOAD): *CISD2*	Extracted DNA	PCR, DNA sequencing
Complex I defect: *NDUFA1, NDUFA8, NDUFA11, NDUFS1, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2*	Extracted DNA	PCR, DNA sequencing
Complex I defect: *ACAD9*	Extracted DNA	PCR, DNA sequencing
Complex II defect: *SDHA*	Extracted DNA	PCR, DNA sequencing, MLPA
Complex II defect: *SDHAF1*	Extracted DNA	PCR, DNA sequencing
Complex II defect: *ISCU*	Extracted DNA	PCR, DNA sequencing
Complex III defect: *BCS1L, MT-CYB*	Extracted DNA	PCR, DNA sequencing

Analyte (measurement parameter)	Test material (matrix)	Test technique
Complex III defect: *TTC19*	Extracted DNA	PCR, DNA sequencing
Complex IV defect: *COX6B1, COX10, COX15, FASTKD2, MT-CO1-3, SCO1, SCO2, SURF1, TACO1,*	Extracted DNA	PCR, DNA sequencing
Complex IV defect: *ETHE1*	Extracted DNA	PCR, DNA sequencing
Complex V defect: *MT-ATP6, MT-ATP8, TMEM70*	Extracted DNA	PCR, DNA sequencing
Pyruvate dehydrogenase complex: *PDHA1*	Extracted DNA	PCR, DNA sequencing, MLPA
Pyruvate dehydrogenase complex: *PDHX*	Extracted DNA	PCR, DNA sequencing
mtDNA depletion: *AGK, C12orf65, DGUOK, MPV17, POLG, RRM2B, SUCLA2, SUCLG1, TK2, C10orf2 (TWNK, PEO1)*	Extracted DNA	PCR, real-time PCR
Normal activity: *OPA1, POLG, POLG2, RRM2B*	Extracted DNA	PCR, DNA sequencing, MLPA
Normal activity: SLC25A4 (ANT1), C10orf2 (TWNK, PEO1)	Extracted DNA	PCR, DNA sequencing
Normal activity: *SLC25A3*	Extracted DNA	PCR, DNA sequencing
Targeted diagnostics of mtDNA: MT-ATP6, MT-ATP8, MT-CO1-3, MT-CYB, MT-ND1-6, mt-tRNAs	Extracted DNA	PCR, DNA sequencing
***mt* DNA / deletion, depletion, sequence analysis**		
Analysis for mtDNA deletions	Extracted DNA	PCR, long-range TP-PCR and size-specific DNA fragment analysis
Analysis for mtDNA depletions	Extracted DNA	PCR, real-time PCR
Complete sequencing of mtDNA	Extracted DNA	PCR, DNA sequencing
Coagulation/haematology		
Thrombophilia: *F2 (Factor II-20210G>A), F5 (Factor-V-Leiden mutations)*	Extracted DNA	PCR, DNA sequencing
Methylenetetrahydrofolate reductase mutation: *MTHFR*	Extracted DNA	PCR, DNA sequencing
X-linked agammaglobulinemia (type Bruton): *BTK*	Extracted DNA	PCR, DNA sequencing

Analyte (measurement parameter)	Test material (matrix)	Test technique
Alpha thalassemia: *HBA1, HBA2*	Extracted DNA	PCR, DNA sequencing, MLPA
Beta thalassemia: *HBB*	Extracted DNA	PCR, DNA sequencing
Sickle cell anemia: *HBB (p.Glu7Val)*	Extracted DNA	PCR, DNA sequencing
Heart diseases		
Cardiomyopathy, hypertrophic - basic diagnostics: ID176	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Cardiomyopathy, hypertrophic: ID072	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Cardiomyopathy, dilated: *MYBPC3*	Extracted DNA	PCR, DNA sequencing, MLPA
Cardiomyopathy, hypertrophic left ventricular: *MYL2*	Extracted DNA	PCR, DNA sequencing
Cardiomyopathy, dilated - basic diagnostics: ID177	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Cardiomyopathy, dilated: ID073	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Cardiomyopathy - infancy/early cardiomyopathy: ID110	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Cardiomyopathy, mitochondrial: ID074	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Cardiomyopathy in the context of a neuromuscular disease: ID035	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Cardiomyopathies, comprehensive diagnostics: ID088	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Barth syndrome: *TAZ*	Extracted DNA	PCR, DNA sequencing
Danon disease: *LAMP2*	Extracted DNA	PCR, DNA sequencing
Long QT syndrome (LQTS) / Brugada syndrome - basic diagnostics: ID178	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Long QT syndrome (LQTS): ID132	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Long QT syndrome (LQTS): *KCNQ1*	Extracted DNA	PCR, DNA sequencing, MLPA

Analyte (measurement parameter)	Test material (matrix)	Test technique
Arrhythmogenic right ventricular dysplasia (ARVD) - basic diagnostics: ID179	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Catecholaminergic polymorphic ventricular tachycardia (CPVT) - basic diagnostics: ID180	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Catecholaminergic polymorphic ventricular tachycardia (CPVT) / arrhythmogenic right ventricular dysplasia (ARVD): ID133	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Arrhythmogenic diseases - comprehensive diagnostics: ID181	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Congenital heart defects, syndromal: ID182	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Congenital heart defect, isolated - basic diagnostics: ID183	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Congenital heart defect, isolated: ID184	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Congenital heart defects - total panel: ID185	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Noonan syndrome, PTPN11 gene: ID159	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Noonan syndrome, other genes: ID285	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Rasopathies: ID160	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Marfan syndrome and type 1 fibrillinopathies: ID174	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Marfan syndrome: *FBN1*	Extracted DNA	PCR, DNA sequencing, MLPA
Heart diseases - total panel: ID186	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Brain malformations / neuronal migration disorders		
Gyration disorders (basic diagnostics): ID571	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Gyration disorders (lissencephaly, pachygyria, polymicrogyria): ID136	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Periventricular nodular heterotopias: ID137	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Aicardi-Goutières syndrome and phenocopy: ID138	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Holoprosencephaly: ID139	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Neuronal migration disorder with microcephaly: ID140	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Pontocerebellar hypoplasia: ID141	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
ARX-associated diseases: *ARX*	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Tuberous sclerosis - TSC2 gene: ID539	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Brain malformations / neuronal migration disorders - total panel: ID144	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Exom analysis by panel ID144: ID165	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Liver and pancreatic diseases		
Pancreatitis: *CASR, CFTR, CPA1, CTRC, PRSS1, SPINK1*	Extracted DNA	PCR, DNA sequencing, MLPA
Alagille syndrome: ID187	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Progressive familial intrahepatic cholestasis: ID188	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Cholestasis / parenchymal liver damage: ID189	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Porphyria: ID191	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Congenital bile acid synthesis defect: *CYP7B1*	Extracted DNA	PCR, DNA sequencing
Liver and pancreatic diseases - total panel: ID192	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
x-chromosomal mental retardation		
Pulmonary arterial hypertension (PAH): ID193	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Cystic fibrosis (mucoviscidosis): Screening of the most common mutations	Extracted DNA	PCR, DNA sequencing, MLPA
Cystic fibrosis *(CFTR* gene) by deletions / duplications of all *CFTR* exons	Extracted DNA	MLPA
Cystic fibrosis *(CFTR* gene): Complete analysis of *CFTR* gene	Extracted DNA	PCR, DNA sequencing, MLPA
Primary ciliary dyskinesia: *CCNO*	Extracted DNA	PCR, DNA sequencing
Congenital respiratory diseases: ID197	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Nephrology, endocrinology and electrolytes		
Adrenogenital syndrome (AGS): *CYP21A2*	Extracted DNA	PCR, DNA sequencing, MLPA
Polycystic kidneys: *PKD1, PKD2*	Extracted DNA	PCR, DNA sequencing, MLPA
Polycystic kidneys, autosomal recessive: *PKHD1*	Extracted DNA	PCR, DNA sequencing, MLPA
Autosomal dominant tubulointerstitial kidney disease (ADTKD): ID200	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Polycystic liver disease: ID201	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Polycystic kidneys: ID202	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Alport syndrome / thin basement membrane syndrome: ID203	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Branchio-oto-renal / brachiootic syndrome (BOR and BOS): ID204	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Renal tubular dysgenesis: ID205	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Anomalies of the urinary tract: ID206	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Renal hypoplasia and renal agenesis: ID207	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Fraser syndrome: ID208	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Congenital anomalies of the kidneys and urinary tract (CAKUT): ID209	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Hyperkalemia / pseudohypoaldosteronism: ID211	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Hypokalemia and Gitelmann/Bartter syndrome: ID212	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Maturity-onset diabetes of the young (MODY): ID213	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Kidney stones: ID214	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Nephrotic syndrome - basic diagnostics: ID215	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Nephrotic syndrome: ID216	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Focal segmental glomerulosclerosis (FSGS) - basic diagnostics: ID217	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Focal segmental glomerulosclerosis (FSGS): ID218	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Kidney diseases, syndromal: ID219	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Kidney disease, non-syndromal: ID220	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Endocrinology / electrolytes: ID221	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Kidney diseases and electrolytes - total panel: ID222	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Pharmacogenetics		
5FU toxicity: *DPYD (c.1905+1G>A)*	Extracted DNA	PCR, DNA sequencing
Thiopurine toxicity: *TPMT*	Extracted DNA	PCR, DNA sequencing
Reproductive genetics		
Adrenogenital syndrome (AGS): *CYP21A2*	Extracted DNA	PCR, DNA sequencing, MLPA
Azoospermia factor: *AZF*	Extracted DNA	PCR, size-specific DNA fragment analysis
Congenital bilateral aplasia vas deferens (CBAVD): *CFTR*	Extracted DNA	PCR, DNA sequencing, MLPA
Premature ovarian insufficiency (POI): *FMR1* by premutant PCR	Extracted DNA	PCR, size-specific DNA fragment analysis
Gender determining region: *SRY* (deletion analysis)	Extracted DNA	PCR, size-specific DNA fragment analysis
Single cell diagnostics for chromosome changes	Polar bodies	Whole genome amplification (WGA), chromosome copy number determination
Single cell diagnostics for chromosome changes	Trophectoderm	Preimplantation diagnostics by whole genome amplification (WGA), chromosome copy number determination
Single cell diagnostics for monogenic diseases	Polar bodies	PCR, DNA sequencing, whole genome amplification (WGA), micro/mini satellite analysis
Single cell diagnostics for monogenic diseases	Trophectoderm	Preimplantation diagnostics by PCR, DNA sequencing, whole genome amplification (WGA), micro/mini satellite analysis, SNP microarray analysis
Retardation and dysmorphic syndromes		
X-chromosomal mental retardation		
Fragile X syndrome: *FMR1*	Extracted DNA	PCR, size-specific DNA fragment analysis
Alpha thalassemia with mental retardation: *ATRX*	Extracted DNA	PCR, DNA sequencing, MLPA
Börjeson-Forssman Lehmann syndrome: *PHF6*	Extracted DNA	PCR, DNA sequencing

Analyte (measurement parameter)	Test material (matrix)	Test technique
X-chromosomal mental retardation (type Cabezas): *CUL4B*	Extracted DNA	PCR, DNA sequencing, MLPA
X-chromosomal mental retardation - basic diagnostics ID268	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Creatine transporter deficiency: *SLC6A8*	Extracted DNA	PCR, DNA sequencing, MLPA
X-chromosomal mental retardation: ID154	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Methylation interference		
Angelman syndrome: Methylation status and deletion/duplication test	Extracted DNA	MS-MLPA (methylation-sensitive)
Angelman syndrome in conspicuous methylation test: Uniparental disomy 15 (UPD15)	Extracted DNA	PCR, microsatellite analysis
Angelman syndrome: *UBE3A*	Extracted DNA	PCR, DNA sequencing, MLPA
Angelman/Rett-like phenotypes - basic diagnostics: ID269	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Angelman/Rett-like phenotypes: ID155	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Beckwith-Wiedemann syndrome: Methylation status and deletion/duplication test	Extracted DNA	MS-MLPA (methylation-sensitive)
Beckwith-Wiedemann syndrome: *CDKN1C*	Extracted DNA	PCR, DNA sequencing
Beckwith-Wiedemann syndrome :, uniparental disomy 11 (UPD11)	Extracted DNA	PCR, microsatellite analysis
Asymmetric large growth and phakomatoses: ID270	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Prader-Willi syndrome: Methylation status and deletion/duplication test	Extracted DNA	MS-MLPA (methylation-sensitive)
Prader-Willi syndrome in conspicuous methylation test: Uniparental disomy 15 (UPD15)	Extracted DNA	PCR, microsatellite analysis
Prader-Willi syndrome: *MAGEL2*	Extracted DNA	PCR, DNA sequencing
Silver Russel syndrome 11p15.5: Methylation status and deletion/duplication test	Extracted DNA	MS-MLPA (methylation-sensitive)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Silver-Russel syndrome 7p12.1 and 7q32.2: Methylation status and deletion/duplication test	Extracted DNA	MS-MLPA (methylation-sensitive)
Silver Russel syndrome: Uniparental disomy 7 maternal (UPD7)	Extracted DNA	PCR, microsatellite analysis
Silver-Russel-Sydrom, in non-conspicuous methylation test: IMAGe syndrome: *CDKN1C*	Extracted DNA	PCR, DNA sequencing
Temple syndrome: Methylation test 14q32	Extracted DNA	MS-MLPA (methylation-sensitive)
Other syndromes		
Achondroplasia (ACH), Hypochondroplasia (HCH), Thanatophoric dysplasia: *FGFR3*	Extracted DNA	PCR, DNA sequencing
Anderman syndrome: *SLC12A6*	Extracted DNA	PCR, DNA sequencing
CHARGE syndrome: ID301	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Coffin Lowry syndrome (CLS): *RPS6KA3*	Extracted DNA	PCR, DNA sequencing, MLPA
Coffin-Siris syndrome 4: *SMARCA4*	Extracted DNA	PCR, DNA sequencing
Coffin-Siris and Nicolaides-Baraitser syndrome (CSS, NCBRS): ID158	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Cohesinopathies (Cornelia de Lange syndrome): ID157	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Cornelia de Lange syndrome: ID580	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Epidermolysis bullosa simplex: *PLEC*	Extracted DNA	PCR, DNA sequencing
Kabuki syndrome: ID271	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Kabuki syndrome: *KDM6A, KMT2D (MLL2)*	Extracted DNA	PCR, DNA sequencing, MLPA
Lipodystrophy Berardeinelli-Seip: *BSCL2* (exon3)	Extracted DNA	PCR, DNA sequencing
Macrocephaly - basic diagnostics: ID272	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Macrocephaly: ID156	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Metabolic dysmorphia syndromes: ID273	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Generalized lipodystrophy type 4 (CGL4): *PTRF (CAVIN1)*	Extracted DNA	PCR, DNA sequencing
Legius syndrome: *SPRED1*	Extracted DNA	PCR, DNA sequencing, MLPA
Loeys-Dietz syndrome: *SMAD3, TGFB2*	Extracted DNA	PCR, DNA sequencing
Loeys-Dietz syndrome: *TGFBR1, TGFBR2*	Extracted DNA	PCR, DNA sequencing, MLPA
Marshall-Smith syndrome: *NFIX*	Extracted DNA	PCR, DNA sequencing
Mediterranean fever, familial: *MEFV*	Extracted DNA	PCR, DNA sequencing
MEF2C-associated developmental disorder: *MEF2C*	Extracted DNA	PCR, DNA sequencing, MLPA
Mowat-Wilson syndrome: *ZEB2*	Extracted DNA	PCR, DNA sequencing, MLPA
Neurofibromatosis 1: ID290	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Opitz G/BBB syndrome: *SPECC1L*	Extracted DNA	PCR, DNA sequencing
Opitz G/BBB syndrome: *MID1*	Extracted DNA	PCR, DNA sequencing, MLPA
Occipital horn syndrome: *ATP7A*	Extracted DNA	PCR, DNA sequencing, MLPA
Pitt-Hopkins syndrome: *TCF4*	Extracted DNA	PCR, DNA sequencing, MLPA
PTEN-associated diseases: *PTEN* (mutated in multiple advanced cancers 1)	Extracted DNA	PCR, DNA sequencing, MLPA
MECP2 duplication syndrome	Extracted DNA	MLPA
Rett syndrome: *MECP2*	Extracted DNA	PCR, DNA sequencing, MLPA
Rett-like syndrome: *FOXG1*	Extracted DNA	PCR, DNA sequencing, MLPA
Rett-like syndrome: *CDKL5*	Extracted DNA	PCR, DNA sequencing, MLPA
Sotos and other large growth syndromes: ID291	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Sotos syndrome: *NSD1*	Extracted DNA	PCR, DNA sequencing, MLPA

Analyte (measurement parameter)	Test material (matrix)	Test technique
Sotos-like large-growth syndromes: *DNMT3A, NFIX, SETD2*	Extracted DNA	PCR, DNA sequencing
Syndromic craniosynostoses: ID275	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Smith-Magenis syndrome: *RAI1*	Extracted DNA	PCR, DNA sequencing, MLPA
Stüve-Wiedemann syndrome: *LIFR*	Extracted DNA	PCR, DNA sequencing
Syndromes with extremity malformations: ID276	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Syndromic diseases - total panel: ID161	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
UPD14 (mat)-like phenotype: Methylation test	Extracted DNA	MS-MLPA (methylation-sensitive)
Weill-Marchesani syndrome: *ADAMTS10, ADAMTS17*	Extracted DNA	PCR, DNA sequencing
Weill-Marchesani syndrome: *FBN1*	Extracted DNA	PCR, DNA sequencing, MLPA
X-chromosomal mental retardation, type Christianson: *SLC9A6*	Extracted DNA	PCR, DNA sequencing
Weaver syndrome: *EZH2*	Extracted DNA	PCR, DNA sequencing
Wiedemann Steiner syndrome: *KMT2A*	Extracted DNA	PCR, DNA sequencing
Microdeletion syndrome - targeted diagnostics		
Autism (16p11.2)	Extracted DNA	MLPA
Cri-du-chat syndrome (5p15.2)	Extracted DNA	MLPA
DiGeorge syndrome (22q11.2)	Extracted DNA	MLPA
Ichthyosis (x-chrom): Microdeletion Xp22.31	Extracted DNA	MLPA
Microdeletion syndrome (1p36)	Extracted DNA	MLPA
Miller-Dieker lissencephaly syndrome: Microdeletion 17p13.3	Extracted DNA	MLPA
Phelan-McDermid syndrome, SHANK3 (22q13.3)	Extracted DNA	MLPA
SHOX, microdeletion Xp22.32	Extracted DNA	MLPA
Smith-Magenis syndrome, microdeletion 17p11.2	Extracted DNA	MLPA
Wiliams-Beuren syndrome: Microdeletion 7q11.23	Extracted DNA	MLPA

Analyte (measurement parameter)	Test material (matrix)	Test technique
Wolf-Hirschhorn syndrome: Microdeletion 4p16.3	Extracted DNA	MLPA
Uniparental disomy 7 (UPD7)	Extracted DNA	PCR, microsatellite analysis
Uniparental disomy 11 (UPD11)	Extracted DNA	PCR, microsatellite analysis
Uniparental disomy 15 (UPD15)	Extracted DNA	PCR, microsatellite analysis
Uniparental disomy 2 (UPD2)	Extracted DNA	PCR, microsatellite analysis
Uniparental disomy 6 (UPD6)	Extracted DNA	PCR, microsatellite analysis
Uniparental disomy 9 (UPD9)	Extracted DNA	PCR, microsatellite analysis
Uniparental disomy 13 (UPD13)	Extracted DNA	PCR, microsatellite analysis
Uniparental disomy 14 (UPD14)	Extracted DNA	PCR, microsatellite analysis
Uniparental disomy 16 (UPD16)	Extracted DNA	PCR, microsatellite analysis
Uniparental disomy 20 (UPD20)	Extracted DNA	PCR, microsatellite analysis
Uniparental disomy 22 (UPD22)	Extracted DNA	PCR, microsatellite analysis
Metabolic diseases		
Adrenogenital syndrome (AGS): *CYP21A2*	Extracted DNA	PCR, DNA sequencing, MLPA
Adrenoleukodystrophy, X-chromosomal (X-ALD): *ABCD1*	Extracted DNA	PCR, DNA sequencing, MLPA
Alpha1-antitrypsin: *SERPINA1*	Extracted DNA	PCR, DNA sequencing
Apolipoprotein B 100 (Apo B100) - familial hypercholesterolemia: *APOB*	Extracted DNA	PCR, DNA sequencing
Cystic fibrosis (mucoviscidosis): Screening of the most common mutations	Extracted DNA	PCR, DNA sequencing, MLPA
Cystic fibrosis *(CFTR* gene) by deletions / duplications of all *CFTR* exons	Extracted DNA	MLPA
Cystic fibrosis *(CFTR* gene): Complete analysis of *CFTR* gene	Extracted DNA	PCR, DNA sequencing, MLPA
Danon disease: *LAMP2*	Extracted DNA	PCR, DNA sequencing
Glycine encephalopathy: ID595	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Guanidino-acetate-methyltransferase deficiency: *GAMT*	Extracted DNA	PCR, DNA sequencing
Hereditary hemochromatosis (HFe): *HFE* (p.His63Asp, p.Cys282Tyr)	Extracted DNA	PCR, DNA sequencing
Cogenital glycosylation defect (CDG) syndrome: ID162	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Methylmalonic acidemia: ID278	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Molybdenum cofactor deficiency: ID279	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Gilbert's syndrome *UGT1A1*	Extracted DNA	PCR, size-specific DNA fragment analysis
Neuronal lipofuscinosis (NCL): ID280	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Menkes syndrome: *ATP7A*	Extracted DNA	PCR, DNA sequencing, MLPA
Maturity-onset diabetes of the young (MODY): ID213	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Ethylmalonic encephalopathy: *ETHE1*	Extracted DNA	PCR, DNA sequencing
tumor diseases		
Breast and ovarian cancer		
Breast and ovarian cancer - only BRCA: ID109	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Hereditary breast and ovarian cancer: ID094	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Hereditary breast cancer basic diagnostics: ID359	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Breast cancer: ID095	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Ovarian cancer - basic diagnostics: ID361	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Ovarian cancer - advanced diagnostics: ID096	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Ovarian cancer: ID097	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Breast cancer / ovarian cancer: *ATM, BRCA1, BRCA2, BRIP1, RAD51C*	Extracted DNA	PCR, DNA sequencing, MLPA
Breast cancer / ovarian cancer: *RAD51D*	Extracted DNA	PCR, DNA sequencing
Breast cancer / ovarian cancer: *CHEK2*	Extracted DNA	PCR, DNA sequencing, MLPA
Gynecological tumors - endometrial cancer		
Endometrial cancer: ID707	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Hereditary colon cancer		
HNPCC / Lynch syndrome - MLH1 and PMS2: ID363	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
HNPCC / Lynch syndrome - MSH2, MSH6 and EPCAM: ID364	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
HNPCC / Lynch syndrome: ID099	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
HNPCC, Lynch syndrome: *MLH1*	Extracted DNA	PCR, DNA sequencing, MLPA
HNPCC, Lynch syndrome: *MSH2*	Extracted DNA	PCR, DNA sequencing, MLPA
HNPCC, Lynch syndrome: *MSH6*	Extracted DNA	PCR, DNA sequencing, MLPA
HNPCC, Lynch syndrome: *PMS2*	Extracted DNA	PCR, DNA sequencing, MLPA
MLH1 promoter methylation	Extracted DNA	MS-MLPA (methylation-sensitive)
Colorectal carcinomas - basic diagnostics without HNPCC/Lynch syndrome: ID365	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Colorectal carcinomas: ID358	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Polyposis syndromes		
Adenomatous polyposis: *APC, MUTYH*	Extracted DNA	PCR, DNA sequencing, MLPA
Adenomatous polyposis: *AXIN2, NTHL1, POLD1, POLE*	Extracted DNA	PCR, DNA sequencing
Cowden syndrome: *PTEN* (mutated in multiple advanced cancers 1)	Extracted DNA	PCR, DNA sequencing, MLPA

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik
Juvenile polyposis: *BMPR1A, ENG, SMAD4*	Extracted DNA	PCR, DNA sequencing, MLPA
Juvenile polyposis: ID565	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Peutz-Jeghers syndrome: *STK11*	Extracted DNA	PCR, DNA sequencing, MLPA
Adenomatous polyposis: ID098	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Polyposis coli: ID362	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Mixed polyposis: ID702	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Serratated polyposis syndrome: *RNF43*	Extracted DNA	PCR, DNA sequencing
Sessil serratated polyposis: ID566	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Gastrointestinal tumors, others		
Pancreatic cancer - basic diagnostics: ID366	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Pancreatic cancer: ID101	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Pancreatitis-associated pancreatic cancer: ID703	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Gastric cancer, E-cadherin: *CDH1*	Extracted DNA	PCR, DNA sequencing, MLPA
Gastric cancer: ID102	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Gastric cancer basic diagnostics: ID643	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Gastrointestinal stromal tumor - basic diagnostics: ID103	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Gastrointestinal stromal tumor: ID104	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Gastrointestinal stromal tumors: KIT, PDGFRA	Extracted DNA	PCR, DNA sequencing, MLPA

Analyte (measurement parameter)	Test material (matrix)	Test technique
Renal cancer		
Von Hippel-Lindau syndrome: *VHL*	Extracted DNA	PCR, DNA sequencing, MLPA
Renal cancer- basic diagnostics: ID367	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Renal cancer: ID106	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Nephrological/urological tumors - prostate cancer		
Prostate cancer: ID708	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Endocrinological tumors		
Paraganglioma-pheochromocytoma syndrome: ID105	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Paraganglioma pheochromocytoma syndrome (PGL1, PGL3, PGL4): *SDHD, SDHC, SDHB*	Extracted DNA	PCR, DNA sequencing, MLPA
Pheochromocytoma: *MAX*	Extracted DNA	PCR, DNA sequencing, MLPA
Pheochromocytoma: *TMEM127*	Extracted DNA	PCR, DNA sequencing
Thyroid cancer - basic diagnostics: ID107	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Thyroid cancer: ID108	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Primary hyperparathyroidism: ID706	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Multiple endocrine neoplasia type 2 (MEN2A/2B): *RET*	Extracted DNA	PCR, DNA sequencing, MLPA
Multiple endocrine neoplasia type 1 (MEN1): *MEN1*	Extracted DNA	PCR, DNA sequencing, MLPA
Multiple endocrine neoplasia type 4 (MEN4): *CDKN1B*	Extracted DNA	PCR, DNA sequencing, MLPA
Tumors of the central nervous system		
Medulloblastoma: ID646	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Brain tumors: ID647	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Childhood tumor diseases		
Constitutional MMR deficiency (CMMRD): ID645	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Childhood tumor diseases - comprehensive diagnostics: ID648	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Rhabdoid tumors: ID701	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Other tumor diseases		
Li-Fraumeni syndrome: *TP53*	Extracted DNA	PCR, DNA sequencing, MLPA
Malignant melanoma: *CDKN2A (p16)*	Extracted DNA	PCR, DNA sequencing, MLPA
Malignant melanoma: *POT1*	Extracted DNA	PCR, DNA sequencing
Wilms tumor: WT1	Extracted DNA	PCR, DNA sequencing, MLPA
Schwannomatosis: *LZTR1, SMARCB1*	Extracted DNA	PCR, DNA sequencing, MLPA
DICER1 syndrome: *DICER1*	Extracted DNA	PCR, DNA sequencing
Hereditary tumor diseases, comprehensive diagnostics - ID093	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
B-cell leukemia: *PAX5*	Extracted DNA	PCR, DNA sequencing, MLPA
Birt-Hogg-Dubé syndrome: *FLCN*	Extracted DNA	PCR, DNA sequencing, MLPA
Carney complex: *PRKAR1A*	Extracted DNA	PCR, DNA sequencing
Gorlin-Goltz syndrome, basal-cell nevus syndrome: *PTCH1, SUFU*	Extracted DNA	PCR, DNA sequencing, MLPA
Leiomyomatosis / renal cell carcinoma: *FH*	Extracted DNA	PCR, DNA sequencing, MLPA
Osler's disease, Osler-Rendu-Weber syndrome: *GDF2*	Extracted DNA	PCR, DNA sequencing
Osler's disease, Osler-Rendu-Weber syndrome: *ACVRL1, ENG*	Extracted DNA	PCR, DNA sequencing, MLPA
Nijmegen breakage syndrome (NBS): *NBN*	Extracted DNA	PCR, DNA sequencing
Pancreatic cancer: *PALB2*	Extracted DNA	PCR, DNA sequencing, MLPA
Pancreatic cancer: *p16/CDKN2A*	Extracted DNA	PCR, DNA sequencing, MLPA

Analyte (measurement parameter)	Test material (matrix)	Test technique
Papillary renal cell carcinoma: *MET*	Extracted DNA	PCR, DNA sequencing
Neurofibromatosis / schwannomatosis: ID704	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Malignant melanoma: ID705	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Tumor diseases - molecular pathological analysis of tumor tissue		
Microsatellite analysis from tumor tissue	Extracted DNA	PCR, microsatellite analysis
MLH1 promoter methylation	Extracted DNA	MS-MLPA (methylation-sensitive)
BRAF (Exon15 Kodon600)	Extracted DNA	PCR, DNA sequencing
RAS (*KRAS/NRAS 12/13, 59, 61, 117, 146)*	Extracted DNA	PCR, DNA sequencing
BRCA1	Extracted DNA	PCR, DNA sequencing, MLPA
BRCA2	Extracted DNA	PCR, DNA sequencing, MLPA
KIT	Extracted DNA	PCR, DNA sequencing, MLPA
MSH2	Extracted DNA	PCR, DNA sequencing, MLPA
MSH6	Extracted DNA	PCR, DNA sequencing, MLPA
MLH1	Extracted DNA	PCR, DNA sequencing, MLPA
PMS2	Extracted DNA	PCR, DNA sequencing, MLPA
Further diagnostics		
Narcolepsy - *HLA-DQ B1*0602*	Extracted DNA	PCR, detection with size-specific DNA fragment analysis
X inactivation	Extracted DNA	Methylation-sensitive digest, PCR, fragment analysis
Contamination test	Extracted DNA	PCR, microsatellite analysis
Crohn's disease: *NOD2*	Extracted DNA	PCR, DNA sequencing
Severe recessive diseases: ID164	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)
Clinical exoms: ID112	Extracted DNA	Next-generation sequencing (in-solution hybridization / high-throughput sequencing)