Deutsche Akkreditierungsstelle GmbH

Annex to the Accreditation Certificate D-ML-13133-01-00 according to DIN EN ISO 15189:2014

Valid from: 31.03.2021
Date of issue: 31.03.2021

Holder of certificate:

Zentrum für Nephrologie und Stoffwechsel
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***angestellter Arzt**
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Examinations in the field:

Medical Laboratory Diagnostics

Medical laboratory field:
Human genetics (molecular human genetics)

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

This document is a translation. The definitive version is the original German annex to the accreditation certificate.
The requirements for the management system in DIN EN ISO 15189 are written in a language relevant to medical laboratories and are generally in accordance with the principles of DIN EN ISO 9001.
The certificate together with its annex reflects the status at the time of the date of issue. The current status of the scope of accreditation can be found in the database of accredited bodies of Deutsche Akkreditierungsstelle GmbH.
https://www.dakks.de/en/content/accredited-bodies-dakks

Medical laboratory field: Human genetics (molecular human genetics)

Type of test:

Molekularbiological Tests (Amplifications)**

Analyte (measurement parameter)	Test material (matrix)	Test technique
Abetalipoproteinemia (MTTP)	genomic DNA	sanger sequencing
Aceruloplasminemia/Hypoceruloplasminemia (CP)	genomic DNA	sanger sequencing
ACTH-independent macronodular adrenal hyperplasia 1 (ARMC5, GNAS)	genomic DNA	sanger sequencing
ACTH-independent macronodular adrenal hyperplasia 2 (ARMC5)	genomic DNA	sanger sequencing
Afibrinogenemia (FGA, FGB, FGG)	genomic DNA	sanger sequencing
Agammaglobulinemia, X-linked (BTK)	genomic DNA	sanger sequencing
Albright hereditary osteodystrophy (GNAS)	genomic DNA	sanger sequencing
Alloimmune thrombocytopenia (ITGA2B, ITGB3)	genomic DNA	sanger sequencing
Alport syndrome (COL4A3, COL4A4, COL4A5)	genomic DNA	sanger sequencing
Leiomyomatosis with alport	genomic DNA	sanger sequencing
Alström syndrome (ALMS1)	genomic DNA	sanger sequencing
Alzheimer disease (APOE, APP, CLU, HFE)	genomic DNA	sanger sequencing
Aminoaciduria (EHHADH, HNF4A, SLC2A2, SLC3A1, SLC7A7, SLC7A9, SLC16A1, SLC34A1)	genomic DNA	sanger sequencing
Finnish type Amyloidosis (GSN)	genomic DNA	sanger sequencing

Analyte (measurement parameter)	Test material (matrix)	Test technique
Aniridia-Wilms-tumor syndrome (PAX6, WT1)	genomic DNA	sanger sequencing
Apolipoprotein deficiency (APOA1, APOA2, APOA5, APOB, APOC1, APOC2, APOC3, APOE, APOL1, CLU)	genomic DNA	sanger sequencing
Apparent mineralocorticoid excess (HSD11B2)	genomic DNA	sanger sequencing
Argininosuccinic aciduria (ASL)	genomic DNA	sanger sequencing
Arteriosclerosis (APOB, APOE, HABP2, LDLR, LPA, MTHFR, SLC3A1)	genomic DNA	sanger sequencing
ATTR amyloidosis (TTR)	genomic DNA	sanger sequencing
Autosomal dominant polycystic kidney disease (GANAB, PKD1, PKD2)	genomic DNA	sanger sequencing
Autosomal dominant tubulointerstitial kidney disease (ADTKD) (HNF1B, REN, SEC61A1, UMOD)	genomic DNA	sanger sequencing
Autosomal dominant protein C deficiency (PROC)	genomic DNA	sanger sequencing
Autosomal dominant protein S deficiency (PROS1)	genomic DNA	sanger sequencing
Autosomal recessive polycystic kidney and hepatic disease 1 (PKHD1)	genomic DNA	sanger sequencing
Autosomal recessive protein C deficiency (PROC)	genomic DNA	sanger sequencing
Autosomal recessive protein S deficiency (PROS1)	genomic DNA	sanger sequencing

Analyte (measurement parameter)	Test material (matrix)	Test technique
Bardet-Biedl syndrome (ARL6, BBIP1, BBS1, BBS2, BBS4, BBS5, BBS7, BBS9, BBS10, BBS12, CEP290, IFT27, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP)	genomic DNA	sanger sequencing
Bartter syndrome (BSND, CASR, CLCNKA, CLCNKB, GNA11, KCNJ1, SLC12A1)	genomic DNA	sanger sequencing
Graves disease (GC)	genomic DNA	sanger sequencing
Benign hyperproreninemia (REN)	genomic DNA	sanger sequencing
Bernard-Soulier syndrome (GP1BA, GP1BB, GP9)	genomic DNA	sanger sequencing
Chondrodysplasia of Blomstrand type (PTH1R)	genomic DNA	sanger sequencing
Branchiootorenal dysplasia (EYA1, SIX5)	genomic DNA	sanger sequencing
Branchiootic syndrome (EYA1, SIX1)	genomic DNA	sanger sequencing
Bronchiectasis with or without elevated sweat chloride (SCNN1A, SCNN1B, SCNN1G)	genomic DNA	sanger sequencing
Brunner syndrome (MAOA)	genomic DNA	sanger sequencing
C3 glomerulonephritis (ADAMTS13, C1QA, C1QB, C1QC, C3, CD46, CFB, CFD, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CLU, DGKE, PIGA, THBD)	genomic DNA	sanger sequencing
Caroli disease (PKHD1)	genomic DNA	sanger sequencing
CFHR5 Nephropathy (CFHR5)	genomic DNA	sanger sequencing
Charcot-Marie-Tooth disease (INF2)	genomic DNA	sanger sequencing
Chylomicronemia (APOA5, APOC2, APOE, GPIHBP1, LPL)	genomic DNA	sanger sequencing
CINCA syndrome (NLRP3)	genomic DNA	sanger sequencing
Citrullinemia (ASS1, SLC25A13)	genomic DNA	sanger sequencing

Analyte (measurement parameter)	Test material (matrix)	Test technique
Coenzyme Q10 deficiency (COQ2, COQ6, COQ8A, COQ9, PDSS1, PDSS2)	genomic DNA	sanger sequencing
Conn syndrome (CACNA1H, KCNJ5)	genomic DNA	sanger sequencing
Crigler-Najjar syndrome (UGT1A)	genomic DNA	sanger sequencing
Cystinosis (CTNS)	genomic DNA	sanger sequencing
Cystinuria (SLC3A1, SLC7A9)	genomic DNA	sanger sequencing
Developmental delay, epilepsy, and neonatal diabetes melitus (KCNJ11)	genomic DNA	sanger sequencing
Dense deposit disease (ADAMTS13, C1QA, C1QB, C1QC, C3, CD46, CFB, CFD, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CLU, DGKE, PIGA, THBD)	genomic DNA	sanger sequencing
Denys-Drash syndrome (WT1)	genomic DNA	sanger sequencing
Diabetes mellitus with insulin	genomic DNA	sanger sequencing
Diabetic nephropathy (ACE, AGT)	genomic DNA	sanger sequencing
Dihydroxyadenin urolithiasis (APRT)	genomic DNA	sanger sequencing
Dilated cardiomyopathy 1A (LMNA)	genomic DNA	sanger sequencing
Donnai-Barrow syndrome (LRP2)	genomic DNA	sanger sequencing
Leri-Weill dyschondrosteosis (SHOX)	genomic DNA	sanger sequencing
Dysfibrinogenemia (FGA, FGB, FGG)	genomic DNA	sanger sequencing
EAST syndrome (KCNJ10)	genomic DNA	sanger sequencing
Eiken syndrome (PTH1R)	genomic DNA	sanger sequencing
Epstein syndrome (MYH9)	genomic DNA	sanger sequencing
Hereditary susceptibility to diabetes (IRS1, PDX1)	genomic DNA	sanger sequencing
Hereditary pancreatic disease (NEK8, NPHP3, PRSS1, SPINK1)	genomic DNA	sanger sequencing
Adult type lactose intolerance (LCT, MCM6)	genomic DNA	sanger sequencing
Erythrocyte lactate transporter defect (SLC16A1)	genomic DNA	sanger sequencing

Analyte (measurement parameter)	Test material (matrix)	Test technique
Factor XII deficiency (F12)	genomic DNA	sanger sequencing
Factor XIII A subunit deficiency (F13A1)	genomic DNA	sanger sequencing
Factor XIII B subunit deficiency (F13B)	genomic DNA	sanger sequencing
Familial erythrocytosis 2 (VHL)	genomic DNA	sanger sequencing
Familial cold autoinflammatory syndrome 1 (NLRP3)	genomic DNA	sanger sequencing
Familial mediterranean fever (MEFV, NOD2, SAA1, TNFRSF1A)	genomic DNA	sanger sequencing
Fanconi-Bickel syndrome (SLC2A2)	genomic DNA	sanger sequencing
Fanconi renotubular syndrome (EHHADH, HNF4A, SLC34A1)	genomic DNA	sanger sequencing
FGF23-induced hypophosphatemic rickets (DMP1, ENPP1, FGF23, PHEX)	genomic DNA	sanger sequencing
Fibrodysplasia ossificans progressiva (ACVR1)	genomic DNA	sanger sequencing
Fibronectin glomerulopathy (FN1)	genomic DNA	sanger sequencing
Fish-eye disease (LCAT)	genomic DNA	sanger sequencing
Focal segmental glomerulosclerosis (FSGS) (ACTN4, APOL1, CD2AP, CLU, INF2, MYO1E, PAX2, TRPC6)	genomic DNA	sanger sequencing
Frasier syndrome (WT1)	genomic DNA	sanger sequencing
Fructose intolerance (ALDOB)	genomic DNA	sanger sequencing
Fructose malabsorption (SLC2A5)	genomic DNA	sanger sequencing
Fructosuria (KHK)	genomic DNA	sanger sequencing
Generalized arterial calcification of infancy (ENPP1)	genomic DNA	sanger sequencing
Generalized lipodystrophy (AGPAT2, BSCL2)	genomic DNA	sanger sequencing
Genetic hyperbilirubinemia (UGT1A)	genomic DNA	sanger sequencing

Analyte (measurement parameter)	Test material (matrix)	Test technique
Gestational diabetes mellitus (TRPM6)	genomic DNA	sanger sequencing
Gilbert syndrome (UGT1A)	genomic DNA	sanger sequencing
Gitelman syndrome (SLC12A3)	genomic DNA	sanger sequencing
Thrombasthenia of Glanzmann and Naegeli (ITGA2B, ITGB3)	genomic DNA	sanger sequencing
Glomerulocystic kidney disease with hyperuricemia and isosthenuria (HNF1B, UMOD)	genomic DNA	sanger sequencing
Glucose-Galactose malabsorption (SLC5A1)	genomic DNA	sanger sequencing
Glucocorticoid triggered hypertension (NR3C1)	genomic DNA	sanger sequencing
Glucocorticoid resistance (NR3C1)	genomic DNA	sanger sequencing
Goodpasture syndrome (COL4A3, COL4A5)	genomic DNA	sanger sequencing
HADH deficiency (HADH)	genomic DNA	sanger sequencing
Hemochromatosis (BMP2, FTH1, HAMP, HFE, HFE2, SLC40A1, TFR2)	genomic DNA	sanger sequencing
Hemolytic uremic Syndrome (ADAMTS13, C3, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CLU, DGKE, PIGA, THBD)	genomic DNA	sanger sequencing
HANAC syndrome (COL4A1, COL4A2)	genomic DNA	sanger sequencing
Hypoparathyroidism, sensorineural deafness, and renal dysplasia syndrome (GATA3)	genomic DNA	sanger sequencing

Analyte (measurement parameter)	Test material (matrix)	Test technique
Hereditary distal motor neuron neuropathy type 5A (BSCL2)	genomic DNA	sanger sequencing
Hereditary leiomyomatosis and renal cell cancer (FH)	genomic DNA	sanger sequencing
Hereditary sensory and autonomic neuropathy type 2A (WNK1)	genomic DNA	sanger sequencing
Hereditary angioedema (F12, SERPING1)	genomic DNA	sanger sequencing
Hereditary glaucoma (OPA1)	genomic DNA	sanger sequencing
Hereditary lymphedema (VEGFC)	genomic DNA	sanger sequencing
Histamine intolerance (AOC1, HNMT, MAOA, MAOB)	genomic DNA	sanger sequencing
Hyper-IgM syndrome (AICDA, CD40, CD40LG, UNG)	genomic DNA	sanger sequencing
Hyperaldosteronism (CACNA1H, CYP11B1, CYP11B2, KCNJ5)	genomic DNA	sanger sequencing
Hyperalphalipoproteinemia (APOC3, CETP)	genomic DNA	sanger sequencing
Hypercholesterolemia (APOB, LDLR, LDLRAP1, PCSK9)	genomic DNA	sanger sequencing
Hyperinsulinemic hypoglycemia (ABCC8, GCK, GLUD1, HADH, HNF1A, HNF4A, INSR, KCNJ11, SLC16A1)	genomic DNA	sanger sequencing
Hypercatabolic hypoproteinemia (B2M)	genomic DNA	sanger sequencing
Hyperlipemia (APOA1, APOA5, APOC3, APOE, CETP, LCAT, LDLR, LIPC, LIPE, LPL, PCSK9, PPARG)	genomic DNA	sanger sequencing
Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome (SLC25A15)	genomic DNA	sanger sequencing
Hyperoxaluria (AGXT, GRHPR, HOGA1)	genomic DNA	sanger sequencing

Analyte (measurement parameter)	Test material (matrix)	Test technique
Hyperparathyroidism (AP2S1, CASR, CDC73, GCM2, GNA11)	genomic DNA	sanger sequencing
Hyperphosphatemic familial tumoral calcinosis (FGF23, GALNT3, KL)	genomic DNA	sanger sequencing
Hypertension and brachydactyly syndrome (PDE3A)	genomic DNA	sanger sequencing
Hypertriglyceridemia (APOA5, APOE, GPIHBP1, LIPC, LIPE, LPL)	genomic DNA	sanger sequencing
Hyperuricemia (REN, UMOD)	genomic DNA	sanger sequencing
Hypoaldosteronism (CYP11B2)	genomic DNA	sanger sequencing
Hypoalphalipoproteinemia (ABCA1, APOA1)	genomic DNA	sanger sequencing
Hypobetalipoproteinemia (APOB)	genomic DNA	sanger sequencing
Hypoinsulinemic hypoglycemia (AKT2)	genomic DNA	sanger sequencing
Hypokalemic periodic paralysis 1 (CACNA1S)	genomic DNA	sanger sequencing
Hypomagnesemia (CLDN16, CLDN19, CNNM2, EGF, EGFR, FXYD2, TRPM6)	genomic DNA	sanger sequencing
Hypomethylation syndrome (ZFP57)	genomic DNA	sanger sequencing
Hypoparathyroidism (AP2S1, CASR, GCM2, GNA11, PTH)	genomic DNA	sanger sequencing
Hypophosphatemic rickets with hyperparathyroidism (KL)	genomic DNA	sanger sequencing
Fanconi-type hypophosphatemic rickets (CLCN5, OCRL, SLC34A1)	genomic DNA	sanger sequencing
Hypophosphatasia (ALPL)	genomic DNA	sanger sequencing
IgA nephropathy (CFHR1, CFHR3, CFHR5)	genomic DNA	sanger sequencing

Analyte (measurement parameter)	Test material (matrix)	Test technique
Infantile sialic acid storage disorder (SLC17A5)	genomic DNA	sanger sequencing
Inclusion body myopathy 2 (GNE)	genomic DNA	sanger sequencing
Insulin resistance (CIDEC, ENPP1, IRS1, IRS2, PPARG)	genomic DNA	sanger sequencing
Ivemark syndrome (NEK8, NPHP3)	genomic DNA	sanger sequencing
Parathyroid carcinoma (CDC73)	genomic DNA	sanger sequencing
Kelley-Seegmiller syndrome (HPRT1)	genomic DNA	sanger sequencing
Kenny-Caffey syndrome (TBCE)	genomic DNA	sanger sequencing
Combined pituitary hormone deficiency (HESX1, LHX3, LHX4, POU1F1, PROP1)	genomic DNA	sanger sequencing
Complement component C1q deficiency (C1QA, C1QB, C1QC)	genomic DNA	sanger sequencing
Complement component C1r/C1s deficiency (C1R)	genomic DNA	sanger sequencing
Complement component C1s deficiency (C1S)	genomic DNA	sanger sequencing
Complement C2 deficiency (C2)	genomic DNA	sanger sequencing
Complement C3 deficiency (C3)	genomic DNA	sanger sequencing
Complement component C4 deficiency (C4A, SERPING1)	genomic DNA	sanger sequencing
Complement C5 deficiency (C5)	genomic DNA	sanger sequencing
Complement C8 deficiency (C8A, C8B, C8G)	genomic DNA	sanger sequencing
Complement C9 deficiency (C9)	genomic DNA	sanger sequencing
Complement factor D deficiency (CFD)	genomic DNA	sanger sequencing
Complement factor I deficiency (CFI)	genomic DNA	sanger sequencing
Properdin deficiency, X-linked (CFP)	genomic DNA	sanger sequencing

Analyte (measurement parameter)	Test material (matrix)	Test technique
Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency (CYP17A1)	genomic DNA	sanger sequencing
Congenital anomalies of kidney and urinary tract 1 (DSTYK)	genomic DNA	sanger sequencing
Kowarski syndrome (GH1)	genomic DNA	sanger sequencing
Lactase deficiency (LCT)	genomic DNA	sanger sequencing
Langer mesomelic dysplasia (SHOX)	genomic DNA	sanger sequencing
Laron syndrome (GHR)	genomic DNA	sanger sequencing
Leber congenital amaurosis (CEP290)	genomic DNA	sanger sequencing
Leprechaunism (INSR)	genomic DNA	sanger sequencing
Lesch-Nyhan syndrome (HPRT1)	genomic DNA	sanger sequencing
Liddle syndrome (NEDD4, NR3C2, OXSR1, SCNN1B, SCNN1G, STK39)	genomic DNA	sanger sequencing
Lipoprotein glomerulopathy (APOE)	genomic DNA	sanger sequencing
Long QT syndrome (KCNJ5)	genomic DNA	sanger sequencing
Lowe disease (OCRL)	genomic DNA	sanger sequencing
Lupus erythematosus nephritis (C1QA, C1QB, C1QC, CFHR1, CFHR3)	genomic DNA	sanger sequencing
Lysinuric protein intolerance (SLC7A7)	genomic DNA	sanger sequencing
Macular degeneration (APOE, C2, C3, CFH, CFHR1, CFHR3)	genomic DNA	sanger sequencing
Malouf syndrome (LMNA)	genomic DNA	sanger sequencing
Measles infection susceptibility (CD46)	genomic DNA	sanger sequencing
McCune-Albright syndrom (GNAS)	genomic DNA	sanger sequencing
Medullary cystic kidney disease (UMOD)	genomic DNA	sanger sequencing

Analyte (measurement parameter)	Test material (matrix)	Test technique
Sea-blue histiocyte disease (APOE)	genomic DNA	sanger sequencing
MELAS syndrome (EHHADH, ND1, ND5, ND6, TRNC, TRNH, TRNK, TRNQ, TRNS1, TRNS2)	genomic DNA	sanger sequencing
Membranoproliferative glomerulonephritis (MPGN) (ADAMTS13, C1QA, C1QB, C1QC, C3, CD46, CFB, CFD, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CLU, DGKE, PIGA, THBD)	genomic DNA	sanger sequencing
Membranous nephropathy (PLA2R1)	genomic DNA	sanger sequencing
Meningococcal infection susceptibility (C3, C5, C8A, C8B, C8G, C9, CD46, CFB, CFD, CFH, CFP)	genomic DNA	sanger sequencing
Methionine adenosyltransferase deficiency (MAT1A)	genomic DNA	sanger sequencing
Mevalonic aciduria (MVK)	genomic DNA	sanger sequencing
Mitochondrial diabetes (TRNE, TRNK, TRNL1, TRNS2)	genomic DNA	sanger sequencing
MODY diabetes (ABCC8, APPL1, BLK, CEL, GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1)	genomic DNA	sanger sequencing
Monocarboxylate transporter 1 deficiency (SLC16A1)	genomic DNA	sanger sequencing
Dent disease (CLCN5, OCRL)	genomic DNA	sanger sequencing
Fabry disease (GLA)	genomic DNA	sanger sequencing
Muckle-Wells syndrome (NLRP3)	genomic DNA	sanger sequencing
Metaphyseal chondrodysplasia of Murk Jansen type (PTH1R)	genomic DNA	sanger sequencing
MYH9 related disorders (MYH9)	genomic DNA	sanger sequencing
Nail-patella syndrome (LMX1B)	genomic DNA	sanger sequencing
Susceptibility to obesity (ENPP1)	genomic DNA	sanger sequencing

Analyte (measurement parameter)	Test material (matrix)	Test technique
Susceptibility to thyrotoxic periodic paralysis 1 (CACNA1S)	genomic DNA	sanger sequencing
Neonatal diabetes mellitus (ABCC8, GCK, GLIS3, INS, KCNJ11, ZFP57)	genomic DNA	sanger sequencing
Nephrolithiasis diarrhea syndrome (SLC26A6)	genomic DNA	sanger sequencing
Nephronophthisis (CEP290, INVS, IQCB1, NEK8, NPHP1, NPHP3, NPHP4, SDCCAG8)	genomic DNA	sanger sequencing
Nephropathy with pretibial epidermolysis bullosa and deafness (CD151)	genomic DNA	sanger sequencing
Nephrotic syndrome (ARHGDIA, COQ8B, DGKE, EMP2, FAT1, GPC5, LAMB2, NPHS1, NPHS2, PLCE1, PTPRO, WT1)	genomic DNA	sanger sequencing
Nonpapillary renal cell carcinoma (HNF1A, HNF1B, VHL)	genomic DNA	sanger sequencing
Hereditary renal amyloidosis (APOA1, B2M, CST3, FGA, LYZ)	genomic DNA	sanger sequencing
Renal cysts and diabetes (RCAD) (HNF1B)	genomic DNA	sanger sequencing
Nonaka myopathy (GNE)	genomic DNA	sanger sequencing
Norum disease (LCAT)	genomic DNA	sanger sequencing
Ornithine aminotransferase deficiency (OAT)	genomic DNA	sanger sequencing
Ornithine carbamoyltransferase deficiency (OTC)	genomic DNA	sanger sequencing
Osteoglophonic dysplasia (FGFR1)	genomic DNA	sanger sequencing
Osteopathia striata with cranial sclerosis (AMER1)	genomic DNA	sanger sequencing
Osteopetrosis (CA2, LRP5)	genomic DNA	sanger sequencing

Analyte (measurement parameter)	Test material (matrix)	Test technique
Osteoporosis/renal Osteodystrophy (CASR, LRP5, RXRA, VDR)	genomic DNA	sanger sequencing
Ovalocytosis (SLC4A1)	genomic DNA	sanger sequencing
Pancreatic agenesis 1 (PDX1)	genomic DNA	sanger sequencing
Papillorenal syndrome (PAX2)	genomic DNA	sanger sequencing
Periodontal Ehlers-Danlos syndrome (C1R, C1S)	genomic DNA	sanger sequencing
Paroxysmal nocturnal hemoglobinuria (PIGA)	genomic DNA	sanger sequencing
Partial lipodystrophy (CIDEC, LMNA, PLIN1, PPARG)	genomic DNA	sanger sequencing
Pheochromocytoma (GDNF, KIF1B, MAX, RET, SDHB, SDHD, TMEM127, VHL)	genomic DNA	sanger sequencing
Porencephaly (COL4A1, COL4A2)	genomic DNA	sanger sequencing
Susceptibility to nephrolithiasis (ALPL, CASR)	genomic DNA	sanger sequencing
Progressive osseous heteroplasia (GNAS)	genomic DNA	sanger sequencing
Pseudohypoaldosteronism (CUL3, KLHL3, NR3C2, SCNN1A, SCNN1B, SCNN1G, WNK1, WNK4)	genomic DNA	sanger sequencing
Pseudohypoparathyroidism type IB (GNAS, STX16)	genomic DNA	sanger sequencing
Rabson-Mendenhall syndrome (INSR)	genomic DNA	sanger sequencing
Raine syndrome (FAM20C)	genomic DNA	sanger sequencing
Renal-hepatic-pancreatic dysplasia (NPHP3)	genomic DNA	sanger sequencing

Analyte (measurement parameter)	Test material (matrix)	Test technique
Renal dysplasia with hypopituitarism and diabetes (HNF1A)	genomic DNA	sanger sequencing
Renal glucosuria (SLC5A2)	genomic DNA	sanger sequencing
Renal hypodysplasia/aplasia (DSTYK, FGF20, ITGA8, PAX2, RET, UPK3A)	genomic DNA	sanger sequencing
Renal tubular acidosis (ATP6V0A4, ATP6V1B1, CA2, SLC4A1, SLC4A4, VIPAS39, VPS33B)	genomic DNA	sanger sequencing
Nephrogenic diabetes insipidus (AQP2, AVPR2)	genomic DNA	sanger sequencing
Renal tubular dysgenesis (ACE, AGT, AGTR1, REN)	genomic DNA	sanger sequencing
Resistance to trypanosoma brucei (APOL1)	genomic DNA	sanger sequencing
Salla disease (SLC17A5)	genomic DNA	sanger sequencing
Thyroid hormone resistance (RXRA, THRB)	genomic DNA	sanger sequencing
Thyroid cancer (HABP2, RET)	genomic DNA	sanger sequencing
Schimke Immunoosseous dysplasia (SMARCAL1)	genomic DNA	sanger sequencing
Poor response to Eculizumab (C5)	genomic DNA	sanger sequencing
Pregnancy exacerbated hypertension (NR3C2)	genomic DNA	sanger sequencing
Severe obesity (PPARG)	genomic DNA	sanger sequencing
Senior-Loken syndrome (CEP290, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8)	genomic DNA	sanger sequencing
Sialuria (GNE)	genomic DNA	sanger sequencing
Somatic nephroblastoma (WT1)	genomic DNA	sanger sequencing
Spastic paraplegia 17 with amyotrophy of hands and feet (BSCL2)	genomic DNA	sanger sequencing

Analyte (measurement parameter)	Test material (matrix)	Test technique
Disorders of the renal phosphate transporters (SLC9A3R1, SLC20A2, SLC34A1, SLC34A3)	genomic DNA	sanger sequencing
Thin basement membrane nephropathy (COL4A3, COL4A4, COL4A5)	genomic DNA	sanger sequencing
Syndromic microphthalmia 6 (BMP4)	genomic DNA	sanger sequencing
Tangier Disease (ABCA1)	genomic DNA	sanger sequencing
Thrombotic thrombocytopenic purpura (ADAMTS13)	genomic DNA	sanger sequencing
Vitamin D-dependent rickets (RXRA, VDR)	genomic DNA	sanger sequencing
Vitamin D hydroxylation-deficient rickets (CYP2R1, CYP27B1)	genomic DNA	sanger sequencing
von Hippel-Lindau syndrome (VHL)	genomic DNA	sanger sequencing
Growth hormone deficiency (BTK, GH1, GHRH, GHRHR, GHSR)	genomic DNA	sanger sequencing
Growth hormone hypersensitivity (GHR)	genomic DNA	sanger sequencing
Growth hormone insensitivity (IGF1, IGF1R, IGFALS, STAT5B)	genomic DNA	sanger sequencing
WAGR syndrome (PAX6, WT1)	genomic DNA	sanger sequencing
Open angle glaucoma 1 (ASB10, MYOC, NTF4, OPTN, WDR36)	genomic DNA	sanger sequencing
Open angle glaucoma 3 (CYP1B1, LTBP2)	genomic DNA	sanger sequencing
Wolfram syndrome (CISD2, WFS1)	genomic DNA	sanger sequencing
X-linked familial short stature (SHOX)	genomic DNA	sanger sequencing
Y-linked familial short stature (SHOX)	genomic DNA	sanger sequencing
Failure of tooth eruption (PTH1R)	genomic DNA	sanger sequencing

Analyte (measurement parameter)	Test material (matrix)	Test technique
Central diabetes insipidus (AVP)	genomic DNA	sanger sequencing
Brain small vessel disease with hemorrhage (COL4A1, COL4A2)	genomic DNA	sanger sequencing
Amyloidosis, cerebroarterial (APP, CST3, ITM2B)	genomic DNA	sanger sequencing
Abetalipoproteinemia (MTTP)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Achondroplasia (FGFR3)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Aceruloplasminemia/Hypoceruloplasminemia (CP)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
ACTH-independent macronodular adrenal hyperplasia (ARMC5, GNAS)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Obesity, adrenal insufficiency, and red hair due to POMC deficiency (POMC)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Afibrinogenemia (FGA, FGB, FGG)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Agammaglobulinemia, X-linked (BTK)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Acro-renal-ocular syndrome (SALL4)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Alagille syndrome 2 (NOTCH2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Albright hereditary osteodystrophy (GNAS)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Allan-Herndon-Dudley syndrome (SLC16A2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Alloimmune thrombocytopenia (ITGA2B, ITGB3)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Alport syndrome (COL4A3, COL4A4, COL4A5)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Leiomyomatosis with alport syndrome (COL4A5, COL4A6)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Alström syndrome (ALMS1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Alzheimer disease (APOE, APP, CLU, CYP2D6, HFE)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Aminoaciduria (EHHADH, HNF4A, SLC1A1, SLC2A2, SLC3A1, SLC6A18, SLC6A19, SLC6A20, SLC7A7, SLC7A9, SLC16A1, SLC34A1, SLC36A2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Finnish type Amyloidosis (GSN)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Congenital abnormalities of the kidney and urinary tract (BMP7, CHD1L)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Aniridia-Wilms-tumor syndrome (PAX6, WT1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Antley-Bixler syndrome (FGFR2, POR)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Apert syndrome (FGFR2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Müllerian aplasia and hyperandrogenism (WNT4)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Apolipoprotein deficiency (APOA1, APOA2, APOA5, APOB, APOC1, APOC2, APOC3, APOE, APOH, APOL1, APOM, CLU)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Apparent mineralocorticoid excess (HSD11B2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Argininosuccinic aciduria (ASL)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Arteriosclerosis (APOB, APOE, HABP2, LDLR, LPA, MTHFR, PON1, SLC3A1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Arts syndrome (PRPS1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
ATTR amyloidosis (TTR)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Autoimmune diabetes (FOXP3, HLA-DQA1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Autoimmune polyendocrinopathy syndrome (AIRE, FOXP3)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Autosomal dominant obesity (MC4R)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Autosomal dominant polycystic kidney disease (PKD1, PKD2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Autosomal dominant tubulointerstitial kidney disease (ADTKD) (HNF1B, MUC1, REN, UMOD)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Autosomal dominant cerebellar ataxia, deafness and narcolepsy (DNMT1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Autosomal dominant protein C deficiency (PROC)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Autosomal dominant protein S deficiency (PROS1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Autosomal dominant Robinow syndrome 1 (WNT5A)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Autosomal recessive polycystic kidney and hepatic disease 1 (PKHD1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Autosomal rezessive deafness (ATP2B2, CDH23, PCDH15, SLC26A4)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Autosomal recessive spastic paraplegia type 44 (GJC2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Autosomal recessive protein C deficiency (PROC)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Autosomal recessive protein S deficiency (PROS1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Axenfeld-Rieger anomaly (FOXC1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Bamforth-Lazarus syndrome (FOXE1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Bardet-Biedl syndrome (ARL6, BBS1, BBS2, BBS4, BBS5, BBS7, BBS9, BBS10, BBS12, CCDC28B, CEP290, LZTFL1, MKKS, MKS1, SDCCAG8, TMEM67, TRIM32, TTC8, WDPCP)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Bartter syndrome (BSND, CASR, CLCNKA, CLCNKB, KCNJ1, SLC12A1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Graves disease (GC)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Benign hyperproreninemia (REN)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Bernard-Soulier syndrome (GP1BA, GP1BB, GP9)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Chondrodysplasia of Blomstrand type (PTH1R)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
BNAR syndrome (FREM1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Branchio-oculo-facial syndrome (TFAP2A)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Branchiootorenal dysplasia (EYA1, SIX2, SIX5)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Branchiootic syndrome (EYA1, SIX1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Bronchiectasis with or without elevated sweat chloride (SCNN1A, SCNN1B, SCNN1G)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Brunner syndrome (MAOA)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
C3 glomerulonephritis (ADAMTS13, C1QA, C1QB, C1QC, C3, CD46, CFB, CFD, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CLU, PIGA, THBD)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Caroli disease (PKHD1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
CFHR5 Nephropathy (CFHR5)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Charcot-Marie-Tooth disease (INF2, PRPS1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
CHARGE syndrome (CHD7, SEMA3E)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Choreoathetosis with hypothyroidism and neonatal respiratory distress (NKX2-1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Chylomicronemia (APOA5, APOC2, APOE, GPIHBP1, LPL)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
CINCA syndrome (NLRP3)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Citrullinemia (ASS1, SLC25A13)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
COACH syndrome (CC2D2A, RPGRIP1L, TMEM67)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Coenzyme Q10 deficiency (COQ2, COQ6, COQ8A, COQ9, PDSS1, PDSS2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Conn syndrome (CACNA1D, CACNA1H, CTNNB1, KCNJ5)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Coumarin resistance (CYP2A6, CYP2C9, CYP4F2, VKORC1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
CR1 deficiency (CR1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Crigler-Najjar syndrome (UGT1A)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Crouzon syndrome (FGFR3)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Cystinosis (CTNS)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Cystinuria (SLC3A1, SLC7A9)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Developmental delay, epilepsy, and neonatal diabetes melitus (KCNJ11)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Dense deposit disease (ADAMTS13, C1QA, C1QB, C1QC, C3, CD46, CFB, CFD, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CLU, PIGA, THBD)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Denys-Drash syndrome (WT1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Diabetes mellitus with insulin resistance and acanthosis nigricans (INSR)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Diabetic nephropathy (ACE, AGT, AKR1B1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Diabetic retinopathy (PON1, VEGFA)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Dicarboxylic aminoaciduria (SLC1A1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Dihydroxyadenin urolithiasis (APRT)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Dilated cardiomyopathy 1A (LMNA)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Donnai-Barrow syndrome (LRP2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Dubin-Johnson syndrome (ABCC2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Leri-Weill dyschondrosteosis (SHOX)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Dysfibrinogenemia (FGA, FGB, FGG)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
EAST syndrome (KCNJ10)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Ehlers-Danlos syndrome due to tenascin-X deficiency (TNXB)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Eiken syndrome (PTH1R)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Emberger syndrome (GATA2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Eosinophil peroxidase deficiency (EPX)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Epidermolysis bullosa (ITGB4)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Epigenetic dyslipidemia (ABCG1, CPT1A, SREBF1, TNNT1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Epstein syndrome (MYH9)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Susceptibility to acute myeloid leukemia (GATA2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Susceptibility to myelodysplastic syndrome (GATA2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hereditary susceptibility to diabetes (IRS1, MAPK8IP1, PDX1, SH2B1, TBC1D1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hereditary pancreatic disease (NEK8, NPHP3, PRSS1, SPINK1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Glomerular basement membrane disorders (PXDN)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Adult type lactose intolerance (LCT, MCM6)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Erythrocyte lactate transporter defect (SLC16A1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Factor XII deficiency (F12)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Factor XIII A subunit deficiency (F13A1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Factor XIII B subunit deficiency (F13B)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Familial erythrocytosis 2 (VHL)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Familial gestational hyperthyroidism (TSHR)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Familial and sporadic pituitary adenomas (CDH23)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Familial cold autoinflammatory syndrome 1 (NLRP3)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Familial mediterranean fever (MEFV, NOD2, SAA1, TNFRSF1A)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Fanconi-Bickel syndrome (SLC2A2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Fanconi renotubular syndrome (EHHADH, HNF4A, SLC34A1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
FGF23-induced hypophosphatemic rickets (DMP1, ENPP1, FGF23, PHEX)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Fibrodysplasia ossificans progressiva (ACVR1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Fibronectin glomerulopathy (FN1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Fish-eye disease (LCAT)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Focal segmental glomerulosclerosis (FSGS) (ACTN4, ALG13, APOL1, ARHGAP24, CD2AP, CLU, INF2, ITGA9, LAMA5, MYO1E, NXF5, PAX2, TRPC6)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Fraser syndrome (FRAS1, FREM2, GRIP1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Frasier syndrome (WT1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Early-onset obesity (NR0B2, POMC)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Fructose-1,6-bisphosphatase deficiency (FBP1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Fructose intolerance (ALDOB)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Fructosuria (KHK)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Galactosemia (GALT)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Generalized arterial calcification of infancy (ABCC6, ENPP1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Generalized lipodystrophy (AGPAT2, BSCL2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Generalized thyrotropin-releasing hormone resistance (TRHR)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Genetic hyperbilirubinemia (SLCO1B1, SLCO1B3, UGT1A)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
BMI effecting trait 14 (FTO)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Gestational diabetes mellitus (TRPM6)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Disturbed regulators of lipid and carbohydrate metabolism (GCKR, GPD1, MLXIPL, TRIB1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Disordered steroidogenesis due to POR deficiency (POR)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Gilbert syndrome (UGT1A)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Gillessen-Kaesbach-Nishimura syndrome (ALG9)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Gitelman syndrome (SLC12A3)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Thrombasthenia of Glanzmann and Naegeli (ITGA2B, ITGB3)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Glomerulocystic kidney disease with hyperuricemia and isosthenuria (HNF1B, UMOD)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Glucose-Galactose malabsorption (SLC5A1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Glucocorticoid triggered hypertension (NR3C1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Glucocorticoid resistance (NR3C1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Glycogen storage disease 1 (G6PC, SLC37A4)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Glycoprotein 1a deficiency (ITGA2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Goodpasture syndrome (COL4A3, COL4A5)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
HADH deficiency (HADH)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hajdu-Cheney syndrome (NOTCH2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hemochromatosis (BMP2, FTH1, HAMP, HFE, HFE2, SLC40A1, TFR2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hemolytic uremic Syndrome (ADAMTS13, C3, C4BPA, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CLU, PIGA, THBD)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
HANAC syndrome (COL4A1, COL4A2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hartnup disorder (SLC6A19)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hypoparathyroidism, sensorineural deafness, and renal dysplasia syndrome (GATA3)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Hennekam syndrome (CCBE1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hepatic CPT-deficiency type 1A (CPT1A)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hepatorenal tyrosinemia (FAH)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hereditary benign chorea (NKX2-1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hereditary distal motor neuron neuropathy type 5A (BSCL2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hereditary leiomyomatosis and renal cell cancer (COL4A6, FH)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hereditary myokymia type 1 (KCNA1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hereditary sensory and autonomic neuropathy type 2A (WNK1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hereditary sensory neuropathy type 1E (DNMT1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hereditary angioedema (F12, SERPING1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hereditary glaucoma (OPA1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Hereditary lymphedema (FLT4, FOXC2, GJC2, KIF11)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Brain malformations with urinary tract defects (NFIA)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Histamine intolerance (AOC1, HNMT, MAOA, MAOB)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
HIV resistance (CCR5, CXCR1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Homocystinuria-megaloblastic anemia (MTR, MTRR)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Humoral hypercalcemia of malignancy (PTHLH)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hyper-IgM syndrome (AICDA, CD40, CD40LG, UNG)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hyperaldosteronism (CACNA1D, CACNA1H, CYP11B1, CYP11B2, KCNJ5)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hyperalphalipoproteinemia (APOC3, CETP)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hypercalcemia (CASR, CYP24A1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hypercholesterolemia (APOB, LDLR, LDLRAP1, PCSK9)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Hyperglycinuria (SLC36A2, SLC6A19, SLC6A20)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hyperinsulinemic hypoglycemia (ABCC8, GCK, GLUD1, HADH, HNF1A, HNF4A, INSR, KCNJ11, SLC16A1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hypercatabolic hypoproteinemia (B2M)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hyperlipemia (APOA1, APOA4, APOA5, APOC3, APOE, ATF6, C5AR2, CETP, CREB3L3, GALNT2, GCKR, LCAT, LDLR, LEPR, LIPC, LIPE, LIPG, LPL, PCSK9, PNPLA2, PPARG, RXRG, USF1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome (SLC25A15)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hyperoxaluria (AGXT, GRHPR, HOGA1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hyperparathyroidism (CASR, CDC73, GCM2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hyperphosphatemic familial tumoral calcinosis (FGF23, GALNT3, KL)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hypertriglyceridemia (ANGPTL4, APOA5, APOE, GPD1, GPIHBP1, LIPC, LIPE, LMF1, LPL)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Hyperuricemia (ABCG2, REN, UMOD)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hypoaldosteronism (CYP11B2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hypoalphalipoproteinemia (ABCA1, APOA1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hypobetalipoproteinemia (ANGPTL3, APOB)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hypogonadotropic hypogonadism 6 with or without anosmia (FGF8)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hypoinsulinemic hypoglycemia (AKT2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hypokalemic periodic paralysis 1 (CACNA1S)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hypomagnesemia (CLDN16, CLDN19, CNNM2, EGF, EGFR, FXYD2,TRPM6, TRPM7)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hypomethylation syndrome (DNMT1, DNMT3A, DNMT3B, KHDC3L, MECP2, NLRP2, NLRP7, ZFP57)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hypomyelinating leukodystrophy 2 (GJC2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Hypoparathyroidism (CASR, GCM2, PTH)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hypophosphatemic rickets with hyperparathyroidism (KL)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Fanconi-type hypophosphatemic rickets (CLCN5, OCRL, SLC34A1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hypophosphatasia (ALPL)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
IgA nephropathy (CFHR1, CFHR3, CFHR5, SPRY2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Imerslund-Grasbeck syndrome (AMN, CUBN)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Iminoglycinuria (SLC36A2, SLC6A19, SLC6A20)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Immunodeficiency 21 (GATA2, STAT1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Infantile sialic acid storage disorder (SLC17A5)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Capillary infantile hemangioma (FLT4)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Inclusion body myopathy 2 (GNE)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Insulin resistance (CIDEC, ENPP1, IRS1, IRS2, PPARG)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Ivemark syndrome (NEK8, NPHP3)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
IVIC syndrome (SALL4)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Joubert syndrome (AHI1, CC2D2A, RPGRIP1L, SUFU, TMEM67, TTC21B)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Juvenile myelomonocytic leukemia (NF1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Kabuki syndrome (KDM6A, KMT2D)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Parathyroid carcinoma (CDC73)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Kelley-Seegmiller syndrome (HPRT1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Kenny-Caffey syndrome (TBCE)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Knobloch syndrome 1 (COL18A1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Combined pituitary hormone deficiency (HESX1, LHX3, LHX4, POU1F1, PROP1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Combined deficiency of vitamin K-dependent clotting factors type 1 (GGCX)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Complement component C1q deficiency (C1QA, C1QB, C1QC)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Complement component C1r/C1s deficiency (C1R)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Complement component C1s deficiency (C1S)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Complement C2 deficiency (C2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Complement C3 deficiency (C3)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Complement component C4 deficiency (C4A, C4B, SERPING1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Complement C5 deficiency (C5)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Complement C6 deficiency (C6)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Complement C7 deficiency (C7)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Complement C8 deficiency (C8A, C8B)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Complement C9 deficiency (C9)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Complement factor D deficiency (CFD)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Complement factor I deficiency (CFI)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Properdin deficiency, X-linked (CFP)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency (CYP17A1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Congenital disorder of glycosylation (ALG9, PMM2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Congenital cadiac malformations (NKX2-5)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Congenital nongoitrous hypothyroidism (NKX2-5, PAX8, THRA, TSHB, TSHR)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Congenital contractural arachnodactyly (FBN2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Congenital insensitivity to pain with anhidrosis (NTRK1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Congenital hypogonadotropic hypogonadism without anosmia 5 (CHD7)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Kowarski syndrome (GH1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
L-ferritin deficiency (FTL)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Lactase deficiency (LCT)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Lacrimoauriculodentodigital syndrome (FGF10)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Langer mesomelic dysplasia (SHOX)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Laron syndrome (GHR)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Leber congenital amaurosis (AIPL1, CEP290, CRB1, CRX, GDF6, GUCY2D, IMPDH1, KCNJ13, LCA5, LRAT, NMNAT1, PRPH2, RD3, RDH12, RPE65, RPGRIP1, SPATA7, TULP1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Legius syndrome (SPRED1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Leprechaunism (INSR)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Lesch-Nyhan syndrome (HPRT1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Liddle syndrome (NEDD4L, NR3C2, OXSR1, SCNN1B, SCNN1G, STK39)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Lipoprotein glomerulopathy (APOE)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Orofacial cleft 11 (BMP4)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Long QT syndrome (KCNH2, KCNJ5, KCNQ1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Lowe disease (OCRL)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Lupus erythematosus nephritis (C1QA, C1QB, C1QC, CFHR1, CFHR3)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Lysinuric protein intolerance (SLC7A7)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Macular degeneration (APOE, ARMS2, C2, C3, CFH, CFHR1, CFHR3, FBN2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Left-right axis malformations (LEFTY2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Malouf syndrome (LMNA)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Lysosomal acid lipase deficiency (LIPA)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Mannose-binding protein deficiency (MBL2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Manitoba oculotrichoanal syndrome (FREM1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Measles infection susceptibility (CD46)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
McCune-Albright syndrom (GNAS)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Meckel syndrome (CC2D2A, RPGRIP1L, TMEM67)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Mediterranean macrothrombocytopenia (ABCG5, ABCG8)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Medullary cystic kidney disease (MUC1, UMOD)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Sea-blue histiocyte disease (APOE)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
MELAS syndrome (EHHADH)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Membranoproliferative glomerulonephritis (MPGN) (ADAMTS13, C1QA, C1QB, C1QC, C3, CD46, CFB, CFD, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CLU, PIGA, THBD)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Membranous nephropathy (HLA-DQA1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Meningococcal infection susceptibility (C3, C5, C7, C8A, C8B, C9, CD46, CFB, CFD, CFH, CFP)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Mesangioproliferative glomerulonephritis (CXCR1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Methionine adenosyltransferase deficiency (MAT1A)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Methylmalonic aciduria (MUT, MMACHC, MMADHC)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Mevalonic aciduria (MVK)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Mitchell-Riley syndrome (RFX6)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
MODY diabetes (ABCC8, BLK, CEL, GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Monocarboxylate transporter 1 deficiency (SLC16A1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Dent disease (CLCN5, OCRL)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Fabry disease (GLA)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Wilson disease (ATP7B)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Mowat-Wilson syndrome (ZEB2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Muckle-Wells syndrome (NLRP3)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Muenke syndrome (FGFR3)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Metaphyseal chondrodysplasia of Murk Jansen type (PTH1R)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
MYH9 related disorders (MYH9)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Myoclonus-nephropathy syndrome (SCARB2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Nail-patella syndrome (LMX1B)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Susceptibility to autoimmune thyroid disease 3 (TG, ZFAT)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Susceptibility to obesity (ADRB2, ADRB3, CARTPT, ENPP1, GHRL, PPARGC1B, SDC3, UCP1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Susceptibility to nocturnal asthma (ADRB2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Susceptibility to cystic renal dysplasia (BICC1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Susceptibility to thyrotoxic periodic paralysis 1 (CACNA1S)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Nemaline myopathy 5 (TNNT1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Severe neonatal-onset encephalopathy with microcephaly (MECP2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Neonatal diabetes mellitus (GCK, GLIS3, HYMAI, INS, PLAGL1, ZFP57)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Nephrolithiasis diarrhea syndrome (SLC26A6)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Nephronophthisis (CEP290, DCDC2, GLIS2, INVS, IQCB1, NEK8, NPHP1, NPHP3, NPHP4, RPGRIP1L, SDCCAG8, TMEM67, TTC21B, WDR19)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Nephropathy with pretibial epidermolysis bullosa and deafness (CD151)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Nephrotic syndrome (LAMB2, NPHS1, NPHS2, PLCE1, PTPRO, WT1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Neurofibromatosis (NF1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Non-autoimmune hyperthyroidism (TSHR)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Noninsulin-dependent diabetes mellitus 1 (CAPN10)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Nonpapillary renal cell carcinoma (DIRC2, FLCN, HNF1A, HNF1B, OGG1, RNF139, VHL)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Hereditary renal amyloidosis (APOA1, B2M, CST3, FGA, LYZ)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Renal cysts and diabetes (RCAD) (HNF1B)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Nonaka myopathy (GNE)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Noonan syndrome 6 (TRHR)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Norum disease (LCAT)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Fasting plasma glucose trait loci (G6PC2, GCKR)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Ornithine aminotransferase deficiency (OAT)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Ornithine carbamoyltransferase deficiency (OTC)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Osteoglophonic dysplasia (FGFR1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Osteopathia striata with cranial sclerosis (AMER1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Osteopetrosis (CA2, LRP5)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Osteoporosis/renal Osteodystrophy (CASR, LRP5, VDR)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Ovalocytosis (SLC4A1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
PAI transcription modulator (SERPINE1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Pancreatogenic diabetes (GATA6, PDX1, PTF1A, RFX6)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Papillorenal syndrome (PAX2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Periodontal Ehlers-Danlos syndrome (C1R, C1S)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Paroxysmal nocturnal hemoglobinuria (PIGA)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Partial lipodystrophy (CIDEC, LMNA, PLIN1, PPARG)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Pendred syndrome (SLC26A4)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Pheochromocytoma (GDNF, KIF1B, MAX, RET, SDHB, SDHD, TMEM127, VHL)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Phosphoribosylpyrophosphate synthetase superactivity (PRPS1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Plasminogen activator inhibitor deficiency (SERPINE1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Polycystic liver disease (LRP5, PRKCSH, SEC63)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Polycystic kidney disease with hyperinsulinemic hypoglycemia (PMM2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Porencephaly (COL4A1, COL4A2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Susceptibility to nephrolithiasis (ALPL, CASR, SLC26A1, TRPV5, ZNF365)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Primary ciliary dyskinesia 3 with or without situs inversus (DNAH5)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Progressive osseous heteroplasia (GNAS)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Protein Z deficiency (PROZ, SERPINA10)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Pseudohypoaldosteronism (CUL3, KLHL3, NR3C2, SCNN1A, SCNN1B, SCNN1G, WNK1, WNK4)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Pseudohypoparathyroidism type IB (GNAS, STX16)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency (GGCX)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Pulmonary alveolar microlithiasis (SLC34A2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Rabson-Mendenhall syndrome (INSR)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Raine syndrome (FAM20C)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Recurrent hydatidiform mole (KHDC3L, NLRP7)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Renal-hepatic-pancreatic dysplasia (NPHP3)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Renal dysplasia with hypopituitarism and diabetes (HNF1A)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Renal glucosuria (SLC5A2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Renal hypodysplasia/aplasia (FGF20, PAX2, RET, UPK3A)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Renal Hypouricemia (SLC22A12, SLC2A9)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Renal tubular acidosis (ATP6V0A4, ATP6V1B1, CA2, SLC4A1, SLC4A4, VIPAS39, VPS33B)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Nephrogenic diabetes insipidus (AQP2, AVPR2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Renal tubular dysgenesis (ACE, AGT, AGTR1, REN)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Resistance to trypanosoma brucei (APOL1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Rett syndrome (MECP2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Salla disease (SLC17A5)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Salt-sensitive essential hypertension (CYP3A5)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Thyroid hormone resistance (SECISBP2, THRA, THRB)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Thyroid cancer (FOXE1, HABP2, HRAS, MINPP1, NKX2-1, NRAS, NTRK1, RET, TSHR)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Schimke Immunoosseous dysplasia (SMARCAL1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Poor response to Eculizumab (C5)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Pregnancy exacerbated hypertension (NR3C2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Severe obesity (PPARG, SIM1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Severe obesity and type 2 diabetes (UCP3)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
X-linked non-syndromic sensorineural deafness type DFN (PRPS1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Senior-Loken syndrome (CEP290, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
SERKAL syndrome (WNT4)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Short QT syndrome (KCNH2, KCNJ2, KCNQ1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Sialuria (GNE)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Sitosterolemia (ABCG5, ABCG8)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Smith-Lemli-Opitz syndrome (DHCR7)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Somatic nephroblastoma (GPC3, WT1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Spastic paraplegia 17 with amyotrophy of hands and feet (BSCL2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Late-onset obesity (AGRP)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Statin intolerance (SLCO1B1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Disorders of mRNA editing (APOBEC3B, APOBEC3G, APOBEC3H)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Disorders of the renal phosphate transporters (SLC9A3R1, SLC20A2, SLC34A1, SLC34A3)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Thin basement membrane nephropathy (COL4A3, COL4A4, COL4A5)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Intellectual disability-severe speech delay-mild dysmorphism syndrome (FOXP1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Multiple synostoses syndrome 3 (FGF9)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Immunodeficiency-centromeric instability-facial anomalies syndrome (DNMT3B)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Syndromic microphthalmia 6 (BMP4)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Tangier Disease (ABCA1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Tatton-Brown-Rahman syndrome (DNMT3A)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Thanatophoric dysplasia (FGFR3)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Thrombophilia due to heparin cofactor 2 deficiency (SERPIND1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Thrombotic thrombocytopenic purpura (ADAMTS13)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Bleeding disorder platelet-type 9 (ITGA2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Thyroid dyshormonogenesis (DUOX2, DUOXA2, IYD, SLC5A5, TG, TPO)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Townes-Brocks syndrome (SALL1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Aplasia of lacrimal and salivary glands (FGF10)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Neutral lipid storage disease (ABHD5, PNPLA2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Trigonocephaly 2 (FREM1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Urofacial syndrome (HPSE2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Usher syndrome (CDH23, PCDH15)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Inflammatory bowel disease (ABCB1, SEL1L)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Inherited leanness (AGRP)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Attenuated cholesterol lowering by statins (HMGCR, KIF6)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Reduced response to beta-2-adrenoreceptor agonist (ADRB2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Vesicoureteral reflux (ROBO2, SOX17, TNXB)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Vitamin D-dependent rickets, type 2A (VDR)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Vitamin D-dependent rickets (RXRA, VDR)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Vitamin D hydroxylation-deficient rickets (CYP2R1, CYP27B1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
von Hippel-Lindau syndrome (VHL)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Growth hormone deficiency (BTK, GH1, GHRH, GHRHR, GHSR)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Growth hormone hypersensitivity (GHR)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Growth hormone insensitivity (IGF1, IGF1R, IGFALS, SH2B1, STAT5B)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
WAGR syndrome (PAX6, WT1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Watson syndrome (NF1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Open angle glaucoma 1 (ASB10, MYOC, NTF4, OPTN, WDR36)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Open angle glaucoma 3 (CYP1B1, LTBP2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Williams-Beuren syndrome (ELN)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Wiskott–Aldrich syndrome (WAS)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Wolcott-Rallison syndrome (EIF2AK3)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Wolfram syndrome (CISD2, WFS1)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Wolman disease (LIPA)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
X-linked syndromic mental retardation Lubs type (MECP2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)

Analyte (measurement parameter)	Test material (matrix)	Test technique
X-linked susceptibility to autism (MECP2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
X-linked familial short stature (SHOX)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Y-linked familial short stature (SHOX)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Failure of tooth eruption (PTH1R)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Central diabetes insipidus (AVP)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Brain small vessel disease with hemorrhage (COL4A1, COL4A2)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Amyloidosis, cerebroarterial (APP, CST3, ITM2B)	genomic DNA	next generation sequencing (Illumina-MiSeq-plattform, TruSight One Panel)
Agammaglobulinemia, X-linked (BTK)	genomic DNA	MLPA (MRC-Holland, Kit P210)
Alport syndrome (COL4A3, COL4A4, COL4A5)	genomic DNA	MLPA (MRC-Holland, Kit P191, P192, P439, P444)
Alzheimer disease (APP)	genomic DNA	MLPA (MRC-Holland, Kit P170)
Aniridia-Wilms-tumor syndrome (PAX6, WT1)	genomic DNA	MLPA (MRC-Holland, Kit P118, P219)
Antenatal Bartter syndrome type 2 (KCNJ1)	genomic DNA	MLPA (MRC-Holland, Kit P200 with in-house MLPA probes)
Apolipoprotein deficiency (APOC2)	genomic DNA	MLPA (MRC-Holland, Kit P200 with in-house MLPA probes)
Autosomal dominant hypercholesterolemia 1 (LDLR)	genomic DNA	MLPA (MRC-Holland, Kit P062)
Autosomal dominant polycystic kidney disease (PKD1, PKD2)	genomic DNA	MLPA (MRC-Holland, Kit P351, P352)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Branchiootorenal dysplasia 1 (EYA1)	genomic DNA	MLPA (MRC-Holland, Kit P153)
C3 glomerulonephritis (CD46, CFH, CFHR1, CFHR2, CFHR3, CFI)	genomic DNA	MLPA (MRC-Holland, Kit P236, P296)
Caroli disease (PKHD1)	genomic DNA	MLPA (MRC-Holland, Kit P341, P342)
Chylomicronemia (APOC2, LPL)	genomic DNA	MLPA (MRC-Holland, Kit P200 with in-house MLPA probes, P218)
Cystinosis (CTNS)	genomic DNA	MLPA (MRC-Holland, Kit P200 with in-house MLPA probes)
Cystinuria (SLC3A1, SLC7A9)	genomic DNA	MLPA (MRC-Holland, Kit P426)
Dense deposit disease (CD46, CFH, CFHR1, CFHR2, CFHR3, CFI)	genomic DNA	MLPA (MRC-Holland, Kit P236, P296)
Denys-Drash syndrome (WT1)	genomic DNA	MLPA (MRC-Holland, Kit P118)
Leri-Weill dyschondrosteosis (SHOX)	genomic DNA	MLPA (MRC-Holland, Kit P018)
Epstein syndrome (MYH9)	genomic DNA	MLPA (MRC-Holland, Kit P432)
Hereditary pancreatic disease (PRSS1, SPINK1)	genomic DNA	MLPA (MRC-Holland, Kit P242)
Factor XII deficiency (F12)	genomic DNA	MLPA (MRC-Holland, Kit P243)
Familial hypocalciuric hypercalcemia type 1 (CASR)	genomic DNA	MLPA (MRC-Holland, Kit P177)
Familial hypocalciuric hypercalcemia type 3 (AP2S1)	genomic DNA	MLPA (MRC-Holland, Kit P200 with in-house MLPA probes)
Familial partial lipodystrophy type 3 (PPARG)	genomic DNA	MLPA (MRC-Holland, Kit P224)
Familial mediterranean fever (MEFV)	genomic DNA	MLPA (MRC-Holland, Kit P094)
Frasier syndrome (WT1)	genomic DNA	MLPA (MRC-Holland, Kit P118)
Fructose intolerance (ALDOB)	genomic DNA	MLPA (MRC-Holland, Kit P255)
Gitelman syndrome (SLC12A3)	genomic DNA	MLPA (MRC-Holland, Kit P136)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Hemochromatosis (HAMP, HFE, HFE2, SLC40A1, TFR2)	genomic DNA	MLPA (MRC-Holland, Kit P347)
Hemolytic uremic Syndrome (CD46, CFH, CFHR1, CFHR2, CFHR3, CFI)	genomic DNA	MLPA (MRC-Holland, Kit P236, P296)
Hypoparathyroidism, sensorineural deafness, and renal dysplasia syndrome (GATA3)	genomic DNA	MLPA (MRC-Holland, Kit P234)
Hereditary leiomyomatosis and renal cell cancer (FH)	genomic DNA	MLPA (MRC-Holland, Kit P198)
Hereditary angioedema (F12, SERPING1)	genomic DNA	MLPA (MRC-Holland, Kit P243)
Hyperaldosteronism type 1 (CYP11B1, CYP11B2)	genomic DNA	MLPA (MRC-Holland, Kit P200 with in-house MLPA probes)
Hyperinsulinemic hypoglycemia 1 (ABCC8, HNF1A, HNF4A)	genomic DNA	MLPA (MRC-Holland, Kit P117, P241)
Hyperoxaluria (AGXT, GRHPR)	genomic DNA	MLPA (MRC-Holland, Kit P305)
Hyperparathyroidism (AP2S1, CASR, CDC73)	genomic DNA	MLPA (MRC-Holland, Kit P177,P200 with in-house MLPA probes, P466)
Hypoaldosteronism (CYP11B2)	genomic DNA	MLPA (MRC-Holland, Kit P200 with in-house MLPA probes)
Hypomagnesemia (CLDN16)	genomic DNA	MLPA (MRC-Holland, Kit P200 with in-house MLPA probes)
Hypoparathyroidism (AP2S1, CASR)	genomic DNA	MLPA (MRC-Holland, Kit P177, P200 with in-house MLPA probes)
Hypophosphatasia (ALPL)	genomic DNA	MLPA (MRC-Holland, Kit P200 with in-house MLPA probes)
Parathyroid carcinoma (CDC73)	genomic DNA	MLPA (MRC-Holland, Kit P466)
Kelley-Seegmiller syndrome (HPRT1)	genomic DNA	MLPA (MRC-Holland, Kit P447)
Classic Bartter syndrome (CLCNKB)	genomic DNA	MLPA (MRC-Holland, Kit P266)
Langer mesomelic dysplasia (SHOX)	genomic DNA	MLPA (MRC-Holland, Kit P018)
Lesch-Nyhan syndrome (HPRT1)	genomic DNA	MLPA (MRC-Holland, Kit P447)

Analyte (measurement parameter)	Test material (matrix)	Test technique
Lysinuric protein intolerance (SLC7A7)	genomic DNA	MLPA (MRC-Holland, Kit P426)
Malouf syndrome (LMNA)	genomic DNA	MLPA (MRC-Holland, Kit P048)
Membranoproliferative glomerulonephritis (MPGN) (CD46, CFH, CFHR1, CFHR2, CFHR3, CFI)	genomic DNA	MLPA (MRC-Holland, Kit P236, P296)
MODY diabetes (ABCC8, APPL1, CEL, GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1)	genomic DNA	MLPA (MRC-Holland, Kit ME033, P117, P170, P241, P357)
Fabry disease (GLA)	genomic DNA	MLPA (MRC-Holland, Kit P159)
MYH9 related disorders (MYH9)	genomic DNA	MLPA (MRC-Holland, Kit P432)
Nail-patella syndrome (LMX1B)	genomic DNA	MLPA (MRC-Holland, Kit P289)
Nephronophthisis (NPHP1)	genomic DNA	MLPA (MRC-Holland, Kit P387)
Renal cysts and diabetes (RCAD) (HNF1B)	genomic DNA	MLPA (MRC-Holland, Kit P241, P357)
Ornithine carbamoyltransferase deficiency (OTC)	genomic DNA	MLPA (MRC-Holland, Kit P079)
Osteoglophonic dysplasia (FGFR1)	genomic DNA	MLPA (MRC-Holland, Kit P133)
Pseudohypoparathyroidism type IB (GNAS, STX16)	genomic DNA	MLPA (MRC-Holland, Kit ME031)
Polycystic kidney disease 4 (PKHD1)	genomic DNA	MLPA (MRC-Holland, Kit P341,
Protein C deficiency (PROC)	genomic DNA	MLPA (MRC-Holland, Kit P265)
Protein S deficiency (PROS1)	genomic DNA	MLPA (MRC-Holland, Kit P112)
Pseudohypoparathyroidism (GNAS, STX16)	genomic DNA	MLPA (MRC-Holland, Kit ME031)
Renal dysplasia with hypopituitarism and diabetes (HNF1A)	genomic DNA	MLPA (MRC-Holland, Kit P241)
Renal hypodysplasia/aplasia (RET)	genomic DNA	MLPA (MRC-Holland, Kit P169)
Nephrogenic diabetes insipidus (AVPR2)	genomic DNA	MLPA (MRC-Holland, Kit P200 with in-house MLPA probes)
Thin basement membrane nephropathy (COL4A3, COL4A4, COL4A5)	genomic DNA	MLPA (MRC-Holland, Kit P191, P192, P439, P444)
Transient neonatal diabetes mellitus (KCNJ11, ZFP57)	genomic DNA	MLPA (MRC-Holland, Kit ME033)
Thromboembolic diseases (SERPINC1)	genomic DNA	MLPA (MRC-Holland, Kit P227)

Analyte (measurement parameter)	Test material (matrix)	Test technique
von Hippel-Lindau syndrome (VHL)	genomic DNA	MLPA (MRC-Holland, Kit P016)
Growth hormone deficiency (GH1)	genomic DNA	MLPA (MRC-Holland, Kit P216)
Growth hormone hypersensitivity (GHR)	genomic DNA	MLPA (MRC-Holland, Kit P262)
Growth hormone insensitivity (IGF1, IGF1R, IGFALS, STAT5B)	genomic DNA	MLPA (MRC-Holland, Kit P217, P262)
WAGR syndrome (PAX6, WT1)	genomic DNA	MLPA (MRC-Holland, Kit P118, P219)
Open angle glaucoma 3A (CYP1B1)	genomic DNA	MLPA (MRC-Holland, Kit P200 with in-house MLPA probes)
Williams-Beuren syndrome (ELN)	genomic DNA	MLPA (MRC-Holland, Kit P029)
Wolfram syndrome 1 (WFS1)	genomic DNA	MLPA (MRC-Holland, Kit P163)
X-linked dominant hypophosphatemic rickets (PHEX)	genomic DNA	MLPA (MRC-Holland, Kit P223)
Central diabetes insipidus (AVP)	genomic DNA	MLPA (MRC-Holland, Kit P200 with in-house MLPA probes)
Venous thromboembolic diseases (F2, F5)	genomic DNA	DNA-Strip-technology