DIAGENOM GmbH
Molecular and cytogenetic diagnostics

DIAGENOM GmbH
Robert-Koch-Str. 10
18059 Rostock, Germany
Tel.: +49-381-440 22 410
Fax: +49-381-440 22 419
mail@diagenom.de
www.diagenom.de

Catalogue of services

Bone- /Connective tissue disease

- Achondroplasia/Hypochondroplasia (FGFR3)
- Amelogenesis imperfecta (ENAM)
- Apert-/Crouzon-/Pfeiffer syndrome (FGFR2)
- Arthrogryposis, distal
  - type 1 (TPM2)
  - type 2A Freeman-Sheldon syndrome (MYH3)
  - type 2B Sheldon-Hall syndrome (TNNT3, TNNI2, MYH3)
- Beals syndrome (FBN2)
- Chondrodysplasia punctata 1 (PEX7)
- Chondrodysplasia punctata 2 (EBP)
- Cleidocranial dysplasia (RUNX2)
- Cleidocranial dysplasia [CCD] (RUNX2)
- DTDST-associated dysplasia (SLC26A2)
- Duchenne/Becker muscular dystrophy (DMD)
- Dyggve-Melchior-Clausen dysplasia [DMC] (DYM)
- Dysplasia, campomelic (SOX9)
- Ehlers-Danlos syndrome
  - type 1+2 (COL5A1, COL5A2)*
  - type 4 (COL3A1)*
  - type 6 (PLOD1)*
- Hypocalciuric hypercalcemia (CASR)*
- Hypophosphatemic rickets, autosomal dominant (FGF23)*
- Hypophosphatemic rickets, X-linked (PHEX)*
- Hypophosphatasia (ALPL)
- Ichthyosis, lamellar (TGM1)*
- Larsen syndrome (FLNB)*
- Loeys-Dietz syndrome (TGFBR1, TGFBR2)*
- Marfan syndrome (FBN1)
- Osler-Rendu-Weber disease (ENG, ACVRL1)*
- Osteochondrodysplasia, recessive (DYM)*
- Osteogenesis imperfecta (COL1A1, COL1A2)
- Osteopetrosis (TCIRG1)*
- Smith-McCort dysplasia [SMC] (DYM)*
- Stickler syndrome
  - type 1 (COL2A1)*
  - type 2 (COL11A1)*

Cardiology

- Dilatative cardiomyopathy, screen
  1. LMNA*
  2. MYH7*
  3. TNNT2*
  4. SCN5A*
- Hypertrophic cardiomyopathy, screen
  1. MYH7*
  2. MYEPC3*
  3. TNNT2*
  4. TPM1*
  5. MYL3*
- Long QT syndrome (Romano Ward, autosomal dominant)
  - KCNQ1*
  - KCNH2*
  - SCN5A*
  - KCNE1*
  - KCNE2*
- Long QT syndrome (Jervell-Lange-Nielsen form, autosomal recessive)*
  - KCNQ1*
  - KCNE1*
- Non-syndromic congenital heart defects [HTX1] (ZIC3)*
- Wolf-Parkinson-White syndrome (PRKAG2)*

Endocrinology

- Apparent mineralocorticoid excess (HSD11B2)*
- Azoospermia (Y-chromosome microdeletions; AZFa-c)
- Congenital adrenal hyperplasia
  - 21-hydroxylase deficiency (CYP21A2)
  - 11-β-hydroxylase deficiency (CYP11B1)*
  - 3-β-hydroxysteroid-dehydrogenase deficiency (HSD3B2)*
  - 17-α-hydroxylase deficiency (CYP17A1)*
- Hyperinsulinemic hypoglycemia
  - type 1 (ABCC8)*
  - type 2 (KCNJ11)*
  - type 6 (GLUD1)*
- Pituitary hormone deficiency (PROP1)*
- Kallmann syndrome
  - type 1 (KAL1)*
  - type 2 (FGFR1)*
  - type 3 (PROKR2)*
  - type 4 (PROK2)*
  - type 5 (CDH7)*
  - type 6 (FGF8)*
- MODY diabetes
  - type 1 (HNF4A)*
  - type 2 (GCK)
  - type 3 (HNF1A)
  - type 4 (PDX1)*
  - type 5 (HNF1B)
  - type 6 (NEUROD1)*
- Thyroid hormone resistance (THRB)*

Hematology / Hemostaseology

Hematology

- Agammaglobulinemia, type Bruton (BTK)*
- Acute myeloid leukemia (CEBPA, NPM1)*
- Chronic myeloid leukemia
  - BCR/ABL fusion gene, *quantitative* or *qualitative*† (BCR, ABL1)*
- Mastocytosis [D816V Mutation] (KIT)*
- Polycythaemia vera/essential Thrombocythemia (JAK2)†

DIAGENOM GmbH
Robert-Koch-Str. 10
18059 Rostock, Germany
Tel.: +49-381-440 22 410
Fax: +49-381-440 22 419
mail@diagenom.de
www.diagenom.de

Hemoglobinopathy

- α-Thalassemia (HBA1, HBA2)
- β-Hemoglobinopathy (HBB)
  - β -Thalassemia
  - Sickle cell anemia
  - other β-hemoglobinopathy (HbC, HbD etc.)*
- δβ-Thalassemia incl. Hb Lepore and HPFH (HBB, HBD*)
- Glucose-6-phosphate dehydrogenase deficiency (G6PD)*
- Pyruvate kinase deficiency (PKLR)*

Hemostaseology

- Antithrombin deficiency (SERPINC1)
- Factor II deficiency (F2)*
- Factor V deficiency (F5)*
- Factor VII deficiency (F7)
- Factor VIII deficiency [hemophilia A] (F8)*
- Factor IX deficiency [hemophilia B] (F9)
- Factor X deficiency (F10)*
- Factor XI deficiency (F11)*
- Factor XII deficiency (F12)*
- Factor XIII deficiency (F13A1, F13B)*
- Fibrinogen; A-, Dys-, hypofibrinogenemia (FGA, FGB, FGG)*
- Hemolytic uremic syndrome (CFH)*
- Neutropenia, congenital
  - type 1 (ELANE)*
  - type 3 (HAX1)*
- Protein C deficiency (PROC)
- Protein S deficiency (PROS1)
- Thrombotic thrombocytopenic purpura (ADAMTS13)*
- Von Willebrand syndrome (VWF)

Thrombocytopenia / Thrombocytopathy

- Bernard-Soulier syndrome (GP1BA, GP1BB, GP9)*
- Gray-Platelet syndrome (NBEAL2)*
- MYH9-related thrombocytopenia (MYH9)*
- Glanzmann thrombasthenia (ITGA2B, ITGB3)*
- Wiskott-Aldrich syndrome (WAS)*

Metabolic diseases

- Adipositas (LEP)* (LEPR)* (MC3R)* (MC4R)* (POMC)*
- Alpha-1-Antitrypsin deficiency (SERPINA1)
- Alpha-Mannosidose (MAN2B1)*
- Angioedema, hereditary
  - type 1/2 C1-INH-deficiency (SERPING1)*
  - type 3 (F12)*
- Angiotensin I-Converting Enzyme IVS16 Polymorphism (ACE)*
- Apolipoprotein C2 deficiency (APOC2)
- Apolipoprotein E genotype (APOE)
- Berardinelli-Seip lipodystrophy
  - type 1 (AGPAT2)*
  - type 2 (BSCL2)*
- Beta-Mannosidosis (MANBA)*
- Bisalbuminemia (ALB)*
- CANDLE syndrome (PSMB8)*
- Carnitin Palmitoyltransferase II-deficiency (CPT2)*
- CDG syndrome type 1a (PMM2)*
- Ceroid lipofuscinosis, neuronal, II (TPP1)*
- Creatin-transporter defect (SLC6A8)*
- Crigler-Najjar syndrome 1/2 (UGT1A1)
- Cystathionine β-synthase deficiency (CBS)*
- Cystinosis (CTNS)*
- Cystic fibrosis [CF, mucoviscidosis] (CFTR)
- Dubin-Johnson syndrome (ABCC2)
- Fabry disease (GLA)
- Farber disease (ASAH1)*
- Fever syndromes, hereditary
  - Familial Mediterranean fever (MEFV)
  - Hyper-IgD syndrome (MVK)*
  - Periodic fever, familial (TNFRSF1A)*
  - Cryopyrinopathies (NLRP3)*
- Fructose intolerance, hereditary (ALDOB)*
- Fucosidosis (FUCA1)*
- Galactokinase deficiency (GALK1)*
- Galaktosemia (GALT)*
- Gaucher disease (GBA)*
- Gilbert syndrome (UGT1A1-Promotor)
- Intrinsic factor deficiency (GIF)*
- Krabbe disease (GALC)*
- Lecithin-Cholesterol-Acyltransferase deficiency (LCAT)*
- Lipoprotein lipase deficiency (LPL)
- McArdle disease, Glycogen storage disease type V (PYGM)*
- Niemann-Pick disease
  - type A/B (SMPD1)
  - type C1 (NPC1)*
  - type C2 (NPC2)*
- Pompe disease, Glycogen storage disease type II (GAA)*
- Sandhoff disease, GM2 Gangliosidosis 2 (HEXB)*
- Schindler disease (NAGA)*
- Tay-Sachs disease, GM2-Gangliosidosis (HEXA)*
- Wilson disease (ATP7B)*
- Metachromatic leukodystrophy (ARSA)*
- Mucolipidosis type II, I-Cell disease (GNPTAB)*
- Mucopolysaccharidosis
  - type 1 Hurler/Scheie (IDUA)*
  - type 2 Hunter (IDS)*
  - type 3a Sanfilippo syndrome A (SGSH)*
  - type 3b Sanfilippo syndrome B (NAGLU)*
  - type 3c Sanfilippo syndrome C (HGSNAT)*
  - type 3d Sanfilippo syndrome D (GNS)*
  - type 4a Morquio A (GALNS)*
  - type 4b Morquio B (GLB1)*
  - type 6 Maroteaux-Lamy syndrome (ARSB)*
  - type 7 Sly syndrome (GUSB)*
  - type 9 (HYAL1)*
- Multiple sulphatase deficiency (SUMF1)*
- Ornithine transcarbamylase deficiency (OTC)*
- Pancreatitis, hereditary (SPINK1, PRSS1, CFTR)
- Phenylketonuria/Hyperphenylalaninemia (PAH)*
- Hyperphenylalaninemia, BH4-deficient
  - type A (PTS)*
  - type B (GCH1)*
  - type C (QDPR)*
  - type D (PCBD1)*

DIAGENOM GmbH
Robert-Koch-Str. 10
18059 Rostock, Germany
Tel.: +49-381-440 22 410
Fax: +49-381-440 22 419
mail@diagenom.de
www.diagenom.de

- Glutaric acidemia (ETFDH)*
- Glycine encephalopathy (GLDC, AMT)*
- Glycogen storage disease type 1a (G6PC)*
- GM1-Gangliosidosis (GLB1)*
- Hemochromatosis [HFE]
  - classic hemochromatosis [C282Y/H63D/S65C] (HFE)
  - classic hemochromatosis [sequencing] (HFE)
  - type 2 juvenile (HJV, HAMP)
  - type 3 (TFR2)
  - type 4 (SLC40A1)
- Homocystinuria, B12-responsive, cbl G (MTR)*
- Hypercholesterinemia type B (APOB)
- Hypercholesterinemia, familial, LDL-receptordeficiency (LDLR)
- Hypercholesterinemia, type3 [AD] (PCSK9)
- Hypercholesterinemia [AR] (LDLRAP1)*
- Porphyria
  - Acute intermittent porphyria (HMBS)*
  - Chronic hepatic porphyria (UROD)*
  - Erythrohepatic protoporphyria (FECH, ALAS2)*
  - Hereditary coproporphyria (CPOX)*
  - Congenital erythropoietic porphyria (UROS)*
  - Porphyria variegata (PPOX)*
- Prosaposin deficiency (PSAP)*
- Pyruvate decarboxylase deficiency (PDHA1)*
- Schwachman-Diamond syndrome (SBDS)*
- Superoxide dismutase 2 polymorphism (SOD2)*
- Tyrosinemia
  - type 1 (FAH)*
  - type 2 (TAT)*

Molecular pathology

- BRAF-associated tumor (BRAF V600E)*
- EGFR-associated tumor (EGFR)
- Gastrointestinal stromal tumor [GIST] (KIT, PDGFRA)*
- HNPCC: Microsatellite instability (MMR-Gene)*
- KRAS-associated tumor (KRAS)

Muscle diseases

- Bethlem myopathy (COL6A1, COL6A2, COL6A3)*
- Bloch-Sulzberger syndrome (IKBKG)*
- Brachydactyly, type B2 [BDB2] (NOG)*
- Central Core disease (RYR1)*
- Emery-Dreifuss muscular dystrophy (EMD)*
- Limb-girdle muscular dystrophy
  - type 1C (CAV3)*
  - type 2A (CAPN3)*
  - type 2B (DYSF)*
  - type 2D (SGCA)*
- Kennedy disease [SBMA] (AR)*
- Muscular dystrophy, congenital merosin-deficient (LAMA2)*
- Muscular dystrophy-Dystroglycanopathy A5 [MDDGA5] (FKRP)*
- Myasthenic syndrome, congenital, postsynaptic (CHRNE)*
- Myotonia congenita, type Becker [AR] (CLCN1)*
- Oculopharyngeal muscular dystrophy (PABPN1)*
- Spinal muscular atrophy 1/2/3/4 [MLPA] (SMN1)*
- Spinal muscular atrophy 1/2/3/4 [Seq.] (SMN1)*

Nephrology

- Alport syndrome (COL4A3, COL4A4, COL4A5)*
- Bartter syndrome
  - type 2 (KCNJ1)*
  - type 3 (CLCNKB)*
- Cystinuria (SLC3A1, SLC7A9)*
- Epstein syndrome (MYH9)*
- Gitelman syndrome (SLC12A3)*
- Lowe syndrome (OCRL)*
- Nephrotic syndrome
  - Finnish type (NPHS1)*
  - Steroid-resistant (NPHS2)*
- Polycystic kidney disease [AD] (PKD1, PKD2)*
- Polycystic kidney disease [AR] (PKHD1)*

Neurogenetics

Dystonia

- DYT1 [Torsion dystonia 1] (TOR1A)*
- DYT5A [DOPA-reponsive dystonia] (GCH1)
- DYT5B [DOPA-reponsive dystonia] (TH)*
- DYT6 [Torsion dystonia 6] (THAP1)
- DYT8 [Paroxysmal nonkinesigenic dyskinesia 1] (PNKD1)*
- DYT11 [Myoclonus dystonia] (SGCE)*
- DYT12 [Dystonia 12] (ATP1A3)*
- DYT16 [Dystonia 16] (PRKRA)*
- DYT18 [Dystonia 18] (SLC2A1)*
- DYT20 [Paroxysmal nonkinesigenic dyskinesia 2] (PNKD2)*

Epilepsy

- Infantile Epilepsy
  - Dravet syndrome (SCN1A)*
  - with mental retardation, female-restricted (PCDH19)
  - (CDKL5)*
  - (FOXG1)*
  - X-linked West syndrome (ARX)*
  - Ohtahara syndrome (STXBP1)*
  - GLUT1-deficiency syndrome 2 (SLC2A1)
- Early childhood epilepsy syndrome
  - X-linked mental retardation type Christianson [Angelman syndrome-like] (SLC9A6)*
  - Rett syndrome (MECP2)
  - Angelman syndrome [methylation test] (UBE3A)*
- Absence epilepsy of childhood (CACNA1H)*
- Benign familial neonatal convulsions [BFNC]
  - (KCNQ2)* potassium channel Q2
  - (KCNQ3)* potassium channel Q3
- GEFS+ (“Generalized epilepsy with febrile seizures plus”)
  - type 2 (SCN1A)*
  - type 1 [GABRD(Juvenile myoclonic epilepsy)*] (SCN1B)*
  - type 3 [Absence epilepsy of school age] (GABRG2)*
- Myoclonic epilepsy, juvenile (GJD2)*
- Unverricht-Lundborg disease (CSTB)*

Leukodystrophies and vascular diseases

- Adrenoleukodystrophy (ABCD1)
- Aicardi-Goutieres syndrome 1 (TREX1)*
- Alexander disease (GFAP)*
- Canavan disease (ASPA)
- CADASIL (NOTCH3)
- Megalencephalic leukoencephalopathy with subcortical cysts (MLC1)*
- Pelizaeus-Merzbacher disease [PMD] (PLP1)*
- Pelizaeus-Merzbacher like disease (GJC2)*
- Peroxisomal acyl-CoA-oxidase deficiency (ACOX1)*
- Tuberous sclerosis

DIAGENOM GmbH
Robert-Koch-Str. 10
18059 Rostock, Germany
Tel.: +49-381-440 22 410
Fax: +49-381-440 22 419
mail@diagenom.de
www.diagenom.de

- Cerebrotendinous xanthomatosis (CYP27A1)*
- Leukoencephalopathy with brainstem and spinal cord involvement and elevated lactate (DARS2)*
- Leukoencephalopathy with vanishing white matter
  - type 1 (EIF2B1)*
  - type 2 (EIF2B2)*
  - type 3 (EIF2B3)*
  - type 4 (EIF2B4)*
  - type 5 (EIF2B5)*
  - type 1 (TSC1)
  - type 2 (TSC2)
- Wolman disease (LIPA)*

Mitochondriopathy and related diseases

- Leber optic neuropathy (frequent mutations, 90%)* (MTND1, MTND4, MTND6)*
- Leber optic neuropathy (MTND1, MTND2, MTND4, MTND5, MTND6)*
- Leigh syndrome (SURF1)*
- MELAS syndrome (MTTL1, MTND5)*
- MERRF syndrome (MTTK, MTTF, MTTP)*
- PEO with mtDNA deletions
  - type 1 (POLG)*
  - type 2 (SLC25A4)*
  - type 3 (C10ORF2)*
  - type 4 (POLG2)*

Motor and Sensory Neuropathies [HMSN/CMT]

- CMT (most common forms: PMP22, MPZ, GJB1 (~90% CMT1))

Demyelinating

- CMT1A/HNPP [AD] Del./Dup. [MLPA] (PMP22)
- CMT1A/HNPP [AD] sequencing (PMP22)
- CMT1B [AD] (MPZ)
- CMT1C [AD] (LITAF)*
- CMT1D [AD] (EGR2)*
- CMT1F [AD] (NEFL)*
- CMT4A [AR] (GDAP1)*
- CMT4B1 [AR] (MTMR2)*
- CMT4B2 [AR] (SBF2)*
- CMT4C [AR] (SH3TC2)*
- CMT4D [AR] (NDRG1)*
- CMT4F [AR] (PRX)*
- CMT4H [AR] (FGD4)*
- CMT4J [AR] (FIG4)*
- CMTX1 [X-linked] (GJB1)
- Dejerine-Sottas syndrome [DSS] (MPZ, PMP22, PRX, EGR2)

Axonal

- CMT2A1 [AD] (KIF1B)*
- CMT2A2 [AD] (MFN2)*
- CMT2B [AD] (RAB7A)*
- CMT2B1 [AR] (LMNA)*
- CMT2B2 [AR] (MED25)*
- CMT2D [AD] (GARS)*
- CMT2E [AD] (NEFL)*
- CMT2F [AD] (HSPB1)*
- CMT2I/2J [AD] (MPZ)
- CMT2K [AR] (GDAP1)*
- CMT2L [AD] (HSPB8)*
- CMTX1 [X-linked] (GJB1)
- CMTX5 [X-linked] (PRPS1)*

Intermediate

- CMTX1 [X-linked] (GJB1)
- DI-CMT B [AD] (DNM2)*
- DI-CMT C [AD] (YARS)*
- DI-CMT D [AD] (MPZ)

Other neuropathies

Motor neuropathy

- dHMN2A (HSPB8)*
- dHMN2B (HSPB1)*
- dHMN5 (GARS)*
- dHMN6 / DSMA1 (IGHMBP2)*
- dHMN7B (DCTN1)*

Autonomic neuropathy

- HSAN1 (SPTLC1)*
- HSAN2 (HSN2)*
- HSAN3 (IKBKAP)*
- HSAN4 (NTRK1)*
- HSAN5 (NGFB)*

Other

- Familial amyloid polyneuropathy (TTR)*
- Hereditary neuralgic amyotrophy (SEPT9)

Neurodegenerative disease

- Alzheimer, familial
  - FAD1 (APP)*
  - FAD3 (PSEN1)*
  - FAD4 (PSEN2)*
- Amyotrophic lateral sclerosis
  - type 1 (SOD1)*
  - type 2 (ALS2)*
- Ataxia telangiectasia (ATM)*
- Ataxie-oculomotor apraxia
  - type 1 (APTX)*
  - type 2 (SETX)*
- Huntington disease [HD] (HTT)
- Huntington disease-like
  - type 1 (PRNP)*
  - type 2 (JPH3)*
- Episodic ataxia
  - type 2 (CACNA1A)*
  - type 5 (CACNB4)*
- Familial hemiplegic migraine 2 (ATP1A2)*
- Fragile X syndrome (FMR1)*
- Friedreich ataxia (FXN)*
- Friedreich ataxia-like with vitamin E deficiency (TTPA)*
- Frontotemporal dementia (MAPT, GRN)*
- Parkinson, familial
  - type 1/4 (SNCA)*
  - type 2 (PARK2)*
  - type 6 (PINK1)*
  - type 7 (DJ1)*
  - type 9 (ATP13A2)*
  - type 15 (FBXO7)*
- Prion-disease [Creutzfeldt-Jakob, Gerstmann-Straussler, Fatal familial insomnia] (PRNP)*
- Spinal muscular atrophy 1/2/3/4 [MLPA] (SMN1)*
- Spinal muscular atrophy 1/2/3/4 [Seq.] (SMN1)*
- Spinocerebellar ataxia
  - type 1 (ATXN1)*
  - type 2 (ATXN2)*
  - type 3 (ATXN3)*
  - type 5 (SPTBN2)*
  - type 6 (CACNA1A)*
  - type 7 (ATXN7)*
  - type 8 (SCA8)*
  - type 10 (ATXN10)*
  - type 11 (TTBK2)*
  - type 12 (PPP2R2B)*
  - type 13 (KCNC3)*

DIAGENOM GmbH
Robert-Koch-Str. 10
18059 Rostock, Germany
Tel.: +49-381-440 22 410
Fax: +49-381-440 22 419
mail@diagenom.de
www.diagenom.de

- Infantile neuroaxonal dystrophy (PLA2G6)*
- Myotonic dystrophy 1 [DM1] (DMPK)*
- Panthotenate-kinase associated neurodegeneration (PANK2)*
  - type 14 (PRKCG)*
  - type 17 (TBP)*
  - type 27 (FGF14)*

Spastic paraplegia [SPG]

Pure/Complicated	Pure	Complicated
SPG2 [X-linked] (PLP1)*	SPG5A [AR] (CYP7B1)*	SPG1 [X-linked] (L1CAM)*
SPG3A [AD] (ATL1)	SPG6 [AD] (NIPA1)*	SPG15 [AR] (ZFYVE26)*
SPG4 [AD] (SPAST)	SPG8 [AD] (KIAA0196)*	SPG17 [AD] (BSCL2)*
SPG7 [AR] (SPG7)	SPG13 [AD] (HSPD1)*	SPG20 [AR] (SPG20)*
SPG10 [AD] (KIF5A)*	SPG31 [AD] (REEP1)*	SPG21 [AR] (SPG21)*
SPG11 [AR] (SPG11)*	SPG42 [AD] (SLC33A1)*	

Pharmacogenetics

- Postanesthetic apnea (BCHE)*
- Fluorouracil (5-FU)* toxicity (DPYD IVS14+1 G>A)
- Irinotecan toxicity (UGT1A1)*
- IL28B SNP with HCV association (IL28B)*
- Malignant hyperthermia 1 (RYR1)*
- Malignant hyperthermia 5 (CACNA1S)*
- Phenprocoumon sensitivity (VKORC1, CYP2C9)*
- Thiopurine S-Methyltransferase deficiency (TPMT)*

Postnatale Cytogenetics

- classical chromosome analysis
- FISH chromosome analysis
  - specification
    - Chri-du-Chat syndrome (5p15.2)*
    - DiGeorge syndrome (DGS2)* (10p13-14)*
    - DiGeorge syndrome/ CATCH22 (22q11.2)
    - Kallmann syndrome (Xp22.3)*
    - Microdeletion syndrome (1p36)
    - Prader-Willi-/Angelman syndrome (15q11-13)
    - Smith-Magenis syndrome (17p13.3)
    - Subtelomer screening
    - Williams-Beuren syndrome (7q11.23)
    - Wolf-Hirschhorn syndrome (4p16.3)

Prenatal Cytogenetics

- ACHE- determination of amniotic fluid†
- AFP- determination of amniotic fluid†
- cell cultivation
- classical chromosome analysis
- FISH chromosome analysis (quick test)
  - specification
    - number differences of chromosomes 13; 18; 21; X and Y

Sensory disturbances (visual and audiotary)

- Macular degeneration, juvenile (CNGB3)*
- Norrie syndrome (NDP)*
- Retinitis pigmentosa type 49 (CNGA1)*
- Deafness, hereditary 1A [AR] (GJB2)*
- Stargardt disease
  - type 1 (ABCA4)*
  - type 3 (ELOVL4)*
  - type 4 (PROM1)*

Syndromes (developmental)

- Angelman syndrome (MS-MLPA+UBE3A)*
- Beckwith-Wiedemann syndrome (MS-MLPA+CDKN1C)*
- Birt-Hogg-Dube syndrome (FLCN)*
- CHARGE syndrome (CHD7)*
- Cohen syndrome (VPS13B)*
- Septooptic dysplasia (HESX1)*
- Mowat-Wilson syndrome (ZEB2)*
- Noonan syndrome
  - type 1 (PTPN11)*
- Prader-Willi syndrome (MS-MLPA)*
- Silver-Russell syndrome
  - (uniparental disomy [UPD] of chromosome 7)*
- Smith-Lemli-Opitz syndrome (DHCR7)*
- Sotos syndrome (NSD1)*
- Townes-Brocks syndrome (SALL1)*
- Waardenburg syndrome
  - type 1 (PAX3)*
  - type 2A (MITF)*

Tumor Genetics

Molecular Genetics

- Alagille syndrome
  - type 1 (JAG1)*
  - type 2 (NOTCH2)*
- APC-associated polyposis [FAP] (APC)*
- BCR-ABL fusion gene (BCR, ABL1)†
- BRAF-associated tumor conditions [V600E] (BRAF)*
- Breast-Ovarian cancer, familial (BRCA1, BRCA2)
- EGFR-associated tumor conditions (EGFR)
- Gastrointestinal stromal tumor, [GIST] (KIT, PDGFRA)*
- Hereditary diffuse gastric cancer (CDH1)*
- KRAS-associated tumor conditions (KRAS)
- Lymphoproliferative syndrome (SH2D1A)*
- Multiple endocrine neoplasia
  - type 1 (MEN1)*
  - type 2 (RET)*
- MYH-associated polyposis (MUTYH)*
- Neurofibromatosis
  - type 1 (NF1)
  - type 2 (NF2)*
- Paraganglioma-Pheochromocytoma syndrome
  - type 1 (SDHD)*

DIAGENOM GmbH
Robert-Koch-Str. 10
18059 Rostock, Germany
Tel.: +49-381-440 22 410
Fax: +49-381-440 22 419
mail@diagenom.de
www.diagenom.de

- Hereditary non-polyposis colorectal cancer [HNPCC]
  - Microsatellite instability (MMR Gens)*
  - Methylation status (MLH1)
  - type 1 (MSH2)
  - type 2 (MLH1)
  - type 4 (PMS2)
  - type 5 (MSH6)
  - type 4 (SDHB)*
- Peutz-Jeghers syndrome (STK11)*
- Prostate cancer (ELAC2, RNASEL, BRCA2, SRD5A2)*
- Renal cell carcinoma, papillary (MET)*
-
- Von Hippel-Lindau syndrome (VHL)*

Cytogenetics

In addition to chromosome analysis the following testing is possible:

- Acute lymphoblastic leukemia [ALL]
  - optional FISH
    - BCR/ABL t(9;22)
    - c-myc t(8q24)*
    - MLL t(11q23)*
    - ETV6 t(12p13)*
    - P16 (9p21)*
- Acute myeloid leukemia [AML]
  - optional FISH
    - EVI1 3q26*
    - EGR1 5q31*
    - D7S522 7q31*
    - CEP8 centromere 8*
    - AML1/ETO t (8;21)*
    - MLL 11q23*
    - PML/RARA t (15;17)*
    - CBFB inv/t (16q22)*
- Chronic lymphocytic leukemia [CLL]
  - FISH
    - MYB 6q23
    - ATM 11q22.3
    - CEP12 centromere 12
    - D13S319 13q14
    - LAMP1 13q34
    - IGH 14q32
    - TP53 17p13
- Chronic myelogenous leukemia [CML]
  - optional FISH
    - BCR/ABL t(9;22), *qualitative*
  - Molecular genetics
    - BCR/ABL, *quantitative*†
- Myelodysplastic syndrome [MDS]
  - optional FISH
    - TET2 4q24*
    - EGR1 5q31*
    - D7S522 7q31*
    - CEP8 centromere 8*
    - TP53 17p13
    - PTPRT,MYBL 20q12*
    - X/Y
- Myeloproliferative neoplasm [MPN]
  - optional FISH
    - BCL/ABL t(9;22), *qualitative*
    - PDGFRA 4q12*
    - PDGFRB 5q33*
    - FGFR1 8p11*
  - Molecular genetics
    - BCR/ABL, *quantitative*†
    - JAK2-V617F-mutation†
    - JAK2-Exon 12-mutation (PV)†
    - MPL-W515L/K-mutation (ET/PMF)†
- Lymphomas
  - optional FISH
    - ALK 2p23*
    - BCL6 3q27*
    - MYC 8q24*
    - CCND1/IGH t(11;14)
    - BIRC3,MALT1 t(11;18)*
    - IGH 14q32
    - IGH/BCL2 t(14;18)*
    - TP53 17p13
- NSCLC (Non-small-cell lung carcinoma)
  - ALK 2p23*
- Plasma cell neoplasm [MM, PCL, MGUS]
  - FISH
    - IGH 14q32
    - RB1 13q14
    - CCND1/IGH t(11;14)
    - TP53 17p13
  - optional
    - FGFR3/IGH t(4;14)
    - IGH/MAF t(14;16)

If you require the analysis of a sample material not included in our service directory, please do not hesitate to contact us at +49 (0)-381/440 22 410.

Our interdisciplinary team of physicians, scientists and medical technical assistants will be pleased to assist you.

* analysis currently not accredited

† analysed by contract laboratory