ZENTRALE LISTE RINGVERSUCHE

(Ringversuchsregister gem. § 79 Abs. 1 Zi 3 GTG)

Ringversuche zur externen Qualitätssicherung in der molekularen Gendiagnostik

Stand: Juli 2018

Inhalt

I. Ringversuche Molekulargenetik

Diverse

Achondroplasia and Thanatophoric dysplasia

GenQA[12]: Achondroplasia and Thanatophoric dysplasia EQA - DNA samples are distributed for three clinical case scenarios. Testing Achondroplasia and Thanatophoric dysplasia genes. Testing can be performed using any methodology.

Arrythmia

GenQA[12]: EQA to test molecularly for Brugada syndrome, Long QT syndrome, Catecholaminergic polymorphic ventricular tachycardia (CPVT) and general arrhythmia - DNA samples are distributed for three clinical case scenarios including one case requiring gene panel testing, a single gene test and a predictive/family test. Testing can be performed using any methodology.

Adult Autosomal Dominant Polycystic Kidney disease (PKD1)

(OMIM #173900)

- Polycystin 1 (PKD1) Gen (OMIM *601313)

- Polycystin 2 (PKD2 Gen (OMIM *173910)

EMQN[8]: Scheme APKD: Mutations in the PKD1 and PKD2 genes; genotyping and biological/clinical interpretation; for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number variation (e.g. MLPA, NGS based CNV analysis).

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Genetics, Germline Mutation testing
Provider: EMQN; sample: lyophilized DNA

Beckwith-Wiedemann Syndrome (BWS)

(OMIM #130650)

- H19/IGF2-Imprinting Control Region (ICR1) Gen
(OMIM *616186)

- H19, Imprinted Maternally Expresses Noncoding Transcript (H19) Gen
(OMIM *103280)

- KCNQ1-Overlapping Transcript 1 (KCNQ1OT1 / LIT1) Gen
(OMIM *604115)

- Cyclin-Dependent Kinase Inhibitor 1C (CDKN1C) Gen
(OMIM *600856)

EMQN[8]: Scheme BWS/SRS:Maternal hypomethylation at ICR2 (KCNQ1OT1), hypermethylation at maternal ICR1 (H19), copy number variants, segmental mosaic UPD11pat, and maternally- inherited mutations of CDKN1C.

genotyping and Biochemical/clinical interpretation; for methylation and copy number analysis

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Genetics, Germline Mutation testing
Provider: EMQN; sample: lyophilized DNA

Blooms Syndrome (BLM) - RecQ Protein-like 3 (RECQL3) Gen
(OMIM #210900 - OMIM *604610)

CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
"Molecular Genetics" (MGL4)

Canavan Disease - Aspartoacylase (ASPA) Gen
(OMIM #271900 - OMIM *608034)

CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
"Molecular Genetics" (MGL4)

Charcot-Marie-Tooth Disease, type 1A (CMT1A) - Peripheral Myelin Protein 22 (PMP22) Gen (OMIM #118220 - OMIM *601097)
Charcot-Marie-Tooth Disease, X-linked dominant 1 (CMTX1) – Gap Junction Protein beta-1 (GJB1) Gen (OMIM #302800 - OMIM *304040)

EMQN[8]: Scheme CMT/HMSN: Mutation testing in the PMP22 (deletion /duplication), MPZ and GJB1 genes;
genotyping, and biological and clinical interpretation; for copy number analysis of the PMP22 gene (e.g. MLPA) and/or sequence analysis (Sanger/NGS) of PMP22/MPZ/GJB1 genes

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Genetics, Germline Mutation testing
Provider: EMQN; sample: lyophilized DNA

Cystische Fibrose (CF) / Mukoviszidose - Cystic Fibrosis transmembrane conductance regulator (CFTR) Gen
(OMIM #219700 - OMIM *602421)

CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
"Molecular Genetics" (MGL2, MGL5)

CF Network[4]: genotyping, written report, interpretation; DNA samples

INSTAND[13]: Molekulargenetik III, Nr.778

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Genetics – Cystic Fibrosis (Provider: CF Network); sample: purified DNA

RfB (DGKL)[19]: in Kooperation mit EMQN

GenQA[12]: *CTFR* gene
Cystic fibrosis EQA - DNA samples are distributed for three clinical case scenarios. Testing can be performed using any methodology.

Duchenne/Becker-Muskeldystrophie (DMD/BMD) - Dystrophin (DMD) Gen
(OMIM #310200 / OMIM #300376 - OMIM *300377)

CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
"Molecular Genetics" (MGL2)

EMQN[8]: Scheme DMD: mutations in the Dystrophin gene;
genotyping & interpretation; for copy number analysis of the dystrophin gene

INSTAND[13]: Molekulargenetik III, Nr.780

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Genetics, Germline Mutation testing
Provider: EMQN; sample: lyophilised DNA

RfB (DGKL)[19]: in Kooperation mit EMQN

GenQA[12]: Duchenne and Becker muscular dystrophies: *DMD* gene
DNA samples are distributed for three clinical case scenarios.
Testing can be performed using any methodology.

Fabry Disease
(OMIM #301500)

GenQA[12]: Fabry disease EQA - DNA samples are distributed for three clinical case scenarios: *GAL* gene
Testing can be performed using any methodology.

Fragiles X-Syndrom - FMR1 Gen
(OMIM #300624 - OMIM *309550)

CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
"Molecular Genetics" (MGL1)

EMQN[8]: - Full version:
Scheme FRAX-Full: target: Triplet repeat expansions and methylation of mutations the FMR1 gene
full diagnosis of entire range of expansion mutations;
genotyping & interpretation;
for PCR-based and Southern blotting techniques;
CCG repeat analysis only
- Prescreen-only version:
Scheme FRAX-Pre screen: Triplet repeat expansion mutations in the FMR1 gene; no full diagnosis, pre-screening only;
for PCR-based and Southern blotting techniques;
CCG repeat analysis only

INSTAND[13]: Molekulargenetik III, Nr.781

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
- Molecular Genetics, Germline Mutation testing
sample: DNA in TE (full version), lyophilised DNA (pre-screening only version); Provider: EMQN
- Molecular Genetics, Disease Program
FMR1-Related Disorders; sample: purified genomic DNA

RfB (DGKL)[19]: in Kooperation mit EMQN

GenQA[12]: *FRAX* tandem repeat testing
Fragile X syndrome EQA - DNA samples are distributed for three clinical case scenarios. Testing can be performed using any methodology.

Friedreich´sche Ataxie 1 (FRDA) – Frataxin (FXN) Gen
(OMIM #229300 – OMIM *606829)

CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
"Molecular Genetics" (MGL2)

EMQN[8]: Scheme FRDA: mutations in the FXN gene;
genotyping & interpretation; for PCR-based and Southern blotting techniques

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Genetics, Germline Mutation testing
Provider: EMQN; sample: lyophilised DNA

GenQA[12]: Ataxia EQA. The diseases included are Friedreich ataxia and spinocerebellar ataxias - DNA samples are distributed for three clinical case scenarios. Friedreich ataxia and spinocerebellar ataxias tandem repeats.
Testing can be performed using any methodology.

Frontotemporal Dementia and/or Amyotrophic Lateral Sclerosis 1 (FTDALS1) – Chromosome 9 Open Reading Frame 72 (C9orf72)
(OMIM #105550 – OMIM *614260)

GenQA[12]: *C9orf72* and dementia genes. Dementia/Amyotrophic lateral sclerosis EQA. DNA samples are distributed for three clinical case scenarios. Testing can be performed using any methodology.

Hereditary Deafness (DFNB1):

Autosomal recessive deafness 1A (DFNB1A) – Gap Junction Protein beta 2 (GJB2) / Connexin 26 (CNX26) Gen
(OMIM #220290 - OMIM *121011)

Autosomal recessive deafness 1B (DFNB1B) - Gap Junction Protein beta 6 (GJB6) Gen
(OMIM #612645 - OMIM *604418):

CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
Connexin-26
"Molecular Genetics" (MGL3)

EMQN[8]: Scheme DFNB1: mutations in the GJB2 and GJB6 genes;
genotyping & interpretation; for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis)

INSTAND[13]: Molekulargenetik III, Nr.782; GJB2, Connexin 26

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Genetics, Germline Mutation testing
Provider: EMQN; sample: lyophilised DNA
RfB (DGKL)[19]: in Kooperation mit EMQN

Hereditary Recurrent Fevers (HRF):

Familial mediterranean fever (MEFV), AD / AR - MEFV Gen: M680I, M694V
(OMIM #134610 / OMIM #249100 - OMIM *608107)
Periodic Fever, familial - Tumor Necrosis Factor Receptor 1A (TNFRSF1A) Gen: P75L
(OMIM #142680 – OMIM *191190)
Mevalonat Kinase (MVK) Gen: E296Gfs, V377I
(OMIM *251170**)**
NLR Family, Pyrin Domain Containing 3 (NLRP3) Gen: D303N
(OMIM *606416)

EMQN[8]: Scheme HRF: mutations in the MEFV, MVK, TNFRSF1A and NLRP3 genes;
genotyping & interpretation; for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis)
RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Genetics, Germline Mutation testing
Provider: EMQN; sample: lyophilised DNA

Huntington Disease (HD) / Chorea Huntington – Huntingtin (HTT) Gen
(OMIM #143100 - OMIM *613004)
CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
"Molecular Genetics" (MGL2)
EMQN[8]: Scheme HD: mutations in the HTT gene;
genotyping & interpretation; CAG repeat analysis only; PCR-based analysis techniques only
INSTAND[13]: Molekulargenetik III, Nr.784
RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Genetics, Germline Mutation testing
Provider: EMQN; sample: lyophilised DNA
RfB (DGKL)[19]: in Kooperation mit EMQN
GenQA[12]: HD tandem repeat testing
Huntington disease EQA - DNA samples are distributed for three clinical case scenarios. Testing can be performed using any methodology.

Mitochondrial Diseases – Polymerase, DNA, Gamma (POLG) Gen
(OMIM *174763)

GenQA[12]: - POLG testing
POLG EQA - DNA samples are distributed for three clinical case scenarios. Testing can be performed using any methodology.
- Mitochondrial DNA diseases
Mitochondrial diseases EQA - DNA samples are distributed for three clinical case scenarios. Testing can be performed using any methodology.

Mitochondrial Myopathy
(OMIM 251900)

EMQN[8]: Scheme mtDNA: Mutations in mtDNA (mitochondrial genome). Metabolic disorders which MAY be covered by the EQA scheme include MELAS, NARP, LHON, MERRF, Leigh syndrome, and Pearson syndrome. Levels of Homo/heteroplasmy will be assessed; genotyping & interpretation; for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis); Mutation variant screening for any mtDNA gene including Lebers hereditary optic neuropathy (in cooperation with RCPA QAP[18])

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Genetics, Germline Mutation testing, Mitochondrial DNA (mtDNA) metabolic disorders
Provider: EMQN, sample: lyophilised DNA

Myotone Dystrophie Typ 1 (DM1) – Dystrophia Myotonica Protein Kinase (DMPK) Gen
(OMIM #160900 – OMIM *605377)

CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
"Molecular Genetics" (MGL2)

EMQN[8]: Scheme DM: mutations in the DMPK gene;
genotyping & interpretation; CTG repeat analysis only; for PCR-based and Southern blotting techniques

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Genetics, Germline Mutation testing
Provider: EMQN; sample: lyophilised DNA

Neuropathy, Hereditary Motor Sensory and Autonomic, Type III (HSAN3) / Familial Dysautonomia - IKBKAP Gen
(OMIM #223900 - OMIM *603722)

CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
"Molecular Genetics" (MGL4)

Niemann-Pick Disease, Type A/B -
Sphingomyelin Phosphodiesterase 1, Acid Lysosomal (SMPD1)
(OMIM #257200 – OMIM *607608)

CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
"Molecular Genetics" (MGL4)

Prader-Willi-Syndrom / Angelman-Syndrom (PWS/AS) - Chromosome region 15q11-q13

(OMIM #176270 / OMIM #105830)

CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
"Molecular Genetics" (MGL1)

EMQN[8]: Scheme PWAS: 15q11-q13 studies;
genotyping & interpretation; for MS-MLPA, methylation-specific PCR and Southern blotting techniques. Methylation analysis of PWS / AS critical region; uniparental disomy / deletion analysis

INSTAND[13]: Molekulargenetik III, Nr.786; ANCR

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Genetics, Germline Mutation testing;
Provider: EMQN; sample: lyophilised DNA

RfB (DGKL)[19]: in Kooperation mit EMQN

GenQA[12]: Hypotonic infant
EQA to test molecularly for the presence of diseases which cause hyoptonic infants. The diseases included are Angelman syndrome, myotonic dystrophy type 1, Prader-Willi syndrome EQA and spinal muscular atrophy. DNA samples are distributed for three clinical case scenarios. Testing can be performed using any methodology.

POLG-related Disorders – Polymerase, DNA, Gamma (POLG) Gen

(OMIM *174763)

GenQA[12]: - POLG testing
POLG EQA - DNA samples are distributed for three clinical case scenarios. Testing can be performed using any methodology.
- Mitochondrial DNA diseases
Mitochondrial diseases EQA - DNA samples are distributed for three clinical case scenarios. Testing can be performed using any methodology.

Rett Syndrome (RTT) - Methyl-CpG Binding Protein 2 (MECP2) Gen

(OMIM #312750 - OMIM *300005)

CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
- Rett Syndrome (RETT): MECP2 genotyping
- Rett Syndrome (RETT): MECP2 duplication/deletion analysis

GenQA[12]: *MECP2* gene
Rett syndrome EQA - DNA samples are distributed for three clinical case scenarios. Testing can be performed using any methodology.

Short stature gene testing / familial SHOX-related disorders –
Short stature homeobox (SHOX) Gen
(OMIM *312865)

EMQN[8]: Scheme SHOX: mutations in the SHOX gene;
genotyping and interpretation; for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis); Testing of the short stature homeobox for disorders such as Langer mesomelic dysplasia Leri-Weill dyschondrosteosis (LWD).

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Genetics, Germline Mutation testing
Provider: EMQN; sample: lyophilised DNA

Silver-Russel Syndrome (SRS) - H19/IGF2-Imprinting Control Region (ICR1) Gen
(OMIM #180860 - OMIM **616186)

EMQN[8]: Scheme BWS/SRS: paternal methylation at ICR1, CNVs simulating maternalisation of ICR1, UPD7mat, other rare imprinting anomalies, and diverse CNVs;
genotyping and interpretation; for methylation and copy number analysis (e.g. MS-MLPA)

Spinal and Bulbar Muscular Atrophy, X-linked 1 (SMAX1) / Kennedy Disease (KD) - Androgen Receptor (AR)
(OMIM #313200 - OMIM *313700)

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Genetics, Sample Exchange Program;
sample: DNA

Spinal muscular atrophy (SMA), Type 1, 2, 3, 4 – Survival of Motor Neuron 1 (SMN1) Gen
(OMIM #253300, OMIM #253500, OMIM #253400, OMIM #271150 - OMIM *600354)

CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
"Molecular Genetics" (MGL2)

EMQN[8]: Scheme SMA: mutations in the SMN1 gene;
genotyping & interpretation; for copy number analysis of the SMN1/(SMN2) gene(s)

INSTAND[13]: Molekulargenetik III, Nr.787

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Genetics, Germline Mutation testing;
Provider: EMQN ; sample: lyophilised DNA

RfB (DGKL)[19]: in Kooperation mit EMQN

GenQA[12]: Hypotonic infant
EQA to test molecularly for the presence of diseases which cause hyoptonic infants. The diseases included are Angelman syndrome,

myotonic dystrophy type 1, Prader-Willi syndrome EQA and spinal muscular atrophy. DNA samples are distributed for three clinical case scenarios. Testing can be performed using any methodology.

Spinocerebelläre Ataxien (SCA) – SCA 1-7 Gene:

Spinocerebelläre Ataxie 1 (SCA1) - Ataxin 1 (ATXN1) Gen
(OMIM #164400 - OMIM *601556)
Spinocerebelläre Ataxie 2 (SCA2) - Ataxin 2 (ATXN2) Gen
(OMIM #183090 - OMIM *601517)
Spinocerebelläre Ataxie 3 (SCA3) / Machado-Joseph Disease - Ataxin 3 (ATXN3) Gen
(OMIM #109150 - OMIM *607047)
Spinocerebelläre Ataxie 4 (SCA4)
(OMIM %600223)
Spinocerebelläre Ataxie 5 (SCA5) - SPTBN2 Gen
(OMIM #600224 - OMIM *604985)
Spinocerebelläre Ataxie 6 (SCA6) - CACNA1A Gen
(OMIM #183086 - OMIM *601011)
Spinocerebelläre Ataxie 7 (SCA7) - Ataxin 7 (ATXN7) Gen
(OMIM #164500 - OMIM *607640)

CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
"Molecular Genetics" (MGL2)

EMQN[8]: Scheme SCA: mutations in the ATXN 1-7 genes;
genotyping & interpretation; CAG repeat analysis only; for PCR-based analysis techniques only

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Genetics, Germline Mutation testing;
Provider: EMQN ; sample: lyophilised DNA

GenQA[12]: Ataxia EQA
The diseases included are Friedreich ataxia and spinocerebellar ataxias - DNA samples are distributed for three clinical case scenarios. Friedreich ataxia and spinocerebellar ataxias tandem repeats. Testing can be performed using any methodology.

Tay Sachs Disease (TSD) - Hexosaminidase A (HEXA) Gen
(OMIM #272800 - OMIM *606869)

CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
"Molecular Genetics" (MGL4)

Stoffwechselerkrankungen

Acyl CoA Dehydrogenase, Medium chain, Deficiency of (ACADMD, MCADD) – Acyl CoA Dehydrogenase, Medium chain (ACADM) Gen:
(OMIM #201450 - OMIM *607008)

GenQA[12]: ACADM c. 985A>G p.(Lys329Glu) variant
Medium chain acyl-CoA dehydrogenase deficiency molecular testing for the ACADM c.985A>G p.(Lys329Glu) variant EQA - DNA samples are distributed for three clinical case scenarios. Testing can be performed using any methodology.

alpha-1-Antitrypsin (AAT) Mangel – AAT / SERPINA1 / a1-Proteinase Inhibitor Gen
(OMIM #613490 - OMIM *107400)

CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
- Alpha-1 Antitrypsin (SERPINA1) Genotyping (AAT)
The survey will test for the M, S and Z alleles; DNA specimens.

ECAT[6]: MG1, Set C (in Kooperation mit DGKL)

INSTAND[13]: Molekulargenetik II, Set 02-B, Nr.743; PiM, PiS, PiZ / a1-AT S-Locus, a1-AT Z-Locus

ÖQUASTA[15]: Analyte Gruppe 1, Set C (in Kooperation mit DGKL)

RfB (DGKL)[19]: - Molekulargenetik Gruppe 1 (MG1), Set C

UKNEQAS[21]: UKNEQAS Immunology, Immunochemistry & Allergy
- Alpha 1 Antitrypsin Phenotyping

Apolipoprotein B100 (ApoB100) Gen: R3500Q (rs 5742904)
(OMIM +107730)

ECAT[6]: MG1, Set C (in Kooperation mit DGKL)

INSTAND[13]: Molekulargenetik III, Nr.771

ÖQUASTA[15]: Analyte Gruppe 1, Set C (in Kooperation mit DGKL)

RfB (DGKL)[19]: - Molekulargenetik Gruppe 1 (MG1), Set C

Apolipoprotein E (ApoE) Gen: ApoE2, E3, E4 Isoform (Arg158Cys, Cys112Arg)
(OMIM +107741)

CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
- Apolipoprotein E Genotyping (APOE)
APOE testing for hyperlipoproteinemia type III and Alzheimer diseases; test for variants APOe2, APOe3 and APOe4; DNA specimens.

ECAT[6]: MG1, Set C (in Kooperation mit DGKL)

EQUALIS[9]: Product code 032, DNA

INSTAND[13]: - Molekulargenetik I; Nr.734, Blut
- Molekulargenetik II, Set 02-C, Nr.744, DNA

Labquality[14]: in Kooperation mit EQUALIS

ÖQUASTA[15]: Analyte Gruppe 1, Set C (in Kooperation mit DGKL)

RfB (DGKL)[19]: - Molekulargenetik Gruppe 1 (MG1), Set C

Cholesterinester Transfer Protein (CETP) Mangel / Hyperalphalipoproteinämie 1 (HALP1) - CETP-Gen
(OMIM #143470 - OMIM *118470)
ECAT[6]: MG1, Set C (in Kooperation mit DGKL)
ÖQUASTA[15]: Analyte Gruppe 1, Set C (in Kooperation mit DGKL)
RfB (DGKL)[19]: - Molekulargenetik Gruppe 1 (MG1), Set C

Fruktose Intoleranz – Aldolase B (ALDOB) Gen; A149P, A174D, N334K
(OMIM #229600 - OMIM *612724)
ECAT[6]: MG1, Set D (in Kooperation mit DGKL)
INSTAND[13]: Molekulargenetik II, Set 2, Nr.793
ÖQUASTA[15]: Analyte Gruppe 1, Set D (in Kooperation mit DGKL)
RfB (DGKL)[19]: - Molekulargenetik Gruppe 1 (MG1), Set D

Gaucher Disease
CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
"Molecular Genetics" (MGL4)

Gilbert (Meulengracht) Syndrom / Hyperbilirubinämie - UDP-Glycosyltransferase 1 Polypeptid A1 (UGT-1A1) Gen: UGT1A1*28 (2bp Insertion (TA) im TATAA Element)
(OMIM #143500 - OMIM *191740)
CAP[3]: CAP Headquarters, Northfield
- Pharmacogenetics: UGT1A1 (PGX3)
ECAT[6]: MG2, Set A (in Kooperation mit DGKL)
INSTAND[13]: Molekulargenetik II, Set 02-F, Nr.742;
UDP-Glucuronyltransferase 1, TATA-Box
ÖQUASTA[15]: Analyte Gruppe 2, Set A (in Kooperation mit DGKL)
RfB (DGKL)[19]: - Molekulargenetik Gruppe 2 (MG2), Set A

Glycogen Storage Disease, Type 1A (GSD1A) - Glucose 6 phosphate, catalytic (G6PC) Gen
(OMIM #232200 - OMIM *613742)
CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
"Molecular Genetics" (MGL4)

Hämochromatose, hereditäre – HFE-Gen
(OMIM #235200..-..OMIM *613609)
CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
"Molecular Genetics" (MGL1)
ECAT[6]: H63D, C282Y, S65C
MG1, Set D (in Kooperation mit DGKL)
EMQN[8]: Scheme HFE: mutations in the HFE gene;
genotyping & interpretation; for targeted mutations testing as well

as sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis)

EQUALIS[9]: Product code 032, DNA: c.187C>G, c.845G>A

INSTAND[13]:
- Molekulargenetik I; Nr.733
  H63D, C282Y, optional S65C; Blut
- Molekulargenetik II, Set 02-E, Nr.741
  H63D, C282Y, optional S65C; DNA

Labquality[14]: Kooperation mit EQUALIS
c.187C>G, c.845G>A

ÖQUASTA[15]: Analyte Gruppe 1, Set D: H63D, C282Y, S65C
(in Kooperation mit DGKL)

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
- Molecular Diagnostics (Molecular Haematology), Module 2;
  His63Asp, Cys282Tyr, Ser65Cys
- Molecular Genetics, Germline Mutation testing
  Provider: EMQN; sample: lyophilised DNA

RfB (DGKL)[19]:
- Molekulargenetik Gruppe 1 (MG1), Set D (H63D, C282Y, S65C)
- DNA-Isolierung (DI) und Genotypisierung (H63D, C282Y, S65C)
- Genotyping and Interpretation (in Kooperation mit EMQN)

UKNEQAS[21]: Histocompatibility & Immunogenetics
- Testing for HFE Gene Mutations (5A)
  Identification of mutations of the HFE gene; blood
- Interpretative: HFE genotype and Hereditary Haemochromatosis (5B)
  Reporting on clinical scenarios related to HFE typing and hereditary haemochromatosis; Clinical scenarios supplied

Hyperbilirubinämie, Rotor Type (HBLRR) -
Solute Carrier Organic Anion Transporter Family, Member 1B1 (SLCO1B1) Gen
(OMIN #237450 – OMIM *604843)

CAP[3]: CAP Headquarters, Northfield
Pharmacogenetics: Genotype detection and interpretation
SLCO1B1 (PGX) (rs4149056)

Hypercholesterolemia, familial autosomal dominant – LDLR, APOB, PCSK9 Gene
(OMIM #143890, #144010, #603776 – OMIM *606945, +107730, *607786)

EMQN[8]: Scheme FH: mutations in the LDLR, APOB and PCSK9 genes; genotyping & biological/clinical interpretation; for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis); Mutation screening in any LDLR, APOB and PCSK9 exons required

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Genetics, Germline Mutation testing
Provider: EMQN; sample: lyophilised DNA

Lactose-Intoleranz – MCM6 Gen: MCM6 IVS13 C/T (= LCT C-13910T; rs4988235)
(OMIM #223100 - OMIM *601806)

ECAT[6]: MG1, Set D (in Kooperation mit DGKL)
EQUALIS[9]: Product code 032, DNA: g.-13910C>T
INSTAND[13]: Molekulargenetik III, Nr.770
Labquality[14]: Kooperation mit EQUALIS
g.-13910C>T
ÖQUASTA[15]: Analyte Gruppe 1, Set D (in Kooperation mit DGKL)
RfB (DGKL)[19]: - Molekulargenetik Gruppe 1 (MG1), Set D

Maturity onset diabetes of the young (MODY) / Monogenic Diabetes (MonoDiab):
(OMIM #606391)

MODY1 – Hepatocyte nuclear factor 4 alpha (HNF4A) Gen
(OMIM #125850 – OMIM *600281)
MODY2 – Glukokinase (GCK) Gen
(OMIM #125851 – OMIM *138079)
MODY3 – HNF1 Homeobox A (HNF1A) / TCF1 Gen
(OMIM #600496 - OMIM *142410)
MODY5 – HNF1 Homeobox B (HNF1B) / TCF2 Gen
(OMIM #137920 - OMIM *189907)

EMQN[8]: Scheme MONODIAB: mutations in the GCK, HNF1A, HNF1B and HNF4A genes;
genotyping & interpretation; for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis).
RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Genetics, Germline Mutation testing
Provider: EMQN; sample: lyophilised DNA

Mitochondrielle Stoffwechselerkrankungen

CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
"CAP/ACMG Inherited Metabolic Diseases":
- Mitochondrial DNA deletion syndromes (IMD1)
includes disorders like Leigh syndrome, Pearson syndrome, Kearns-Sayre syndrome
- MCAD (IMD2)
- Mitochondrial Cytopathies (IMD3)
includes disorders like Leber hereditary optic neuropathy and Myoclonus epilepsy with ragged red fibers (MERRF)

GenQA[12]: Mitochondrial diseases EQA - DNA samples are distributed for three clinical case scenarios. Testing can be performed using any methodology.

Morbus Wilson / Wilson Disease - ATPase Cu(2+)-Transporting beta Polypeptide (ATP7B) Gen

(OMIM #277900 – OMIM *606882)

ECAT[6]:	C3207A MG1, Set E (in Kooperation mit DGKL)
EMQN[8]:	Scheme WIL: mutations in the ATP7B gene; genotyping & interpretation; for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis)
INSTAND[13]:	Molekulargenetik II, Set 02-D, Nr.745; H1069Q
ÖQUASTA[15]:	Analyte Gruppe 1, Set E: C3207A (in Kooperation mit DGKL)
RCPA QAP[18]:	RCPAQAP Enrolment Office, St.Leonards Molecular Genetics, Germline Mutation testing Provider: EMQN; sample: lyophilised DNA
RfB (DGKL)[19]:	- Molekulargenetik Gruppe 1 (MG1), Set E (C3207A) - Genotyping and Interpretation (in Kooperation mit EMQN)

Mukolipidose, Typ IV - Mucolipin 1 (MCOLN1) Gen

(OMIM #252650 - OMIM *605248)

CAP[3]:	CAP Headquarters, Northfield (in Kooperation mit ACMG[1]) "Molecular Genetics" (MGL4)

Multiple Acyl-CoA Dehydrogenase Deficiency (MCADD)

(OMIM #231680)

GenQA[12]:	*ACADM* c. 985A>G p.(Lys329Glu) variant Molecular testing for MCADD c.985A>G on blood spots (4 distributions per year)

Osteogenesis imperfecta

EMQN[8]:	Scheme OI: mutations in the COL1A1 and COL1A2 genes; genotyping & biological/clinical interpretation; for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis); Mutation screening in any COL1A1 and COL1A2 exons required
RCPA QAP[18]:	RCPAQAP Enrolment Office, St.Leonards Molecular Genetics, Germline Mutation testing Provider: EMQN; sample: lyophilised DNA

Osteoporose - Typ1 Kollagen alpha 1 (COL1A1) Gen: SP1 G/T

(OMIM #166710 - OMIM +120150)

ECAT[6]:	MG1, Set E (in Kooperation mit DGKL)
ÖQUASTA[15]:	Analyte Gruppe 1, Set E (in Kooperation mit DGKL)
RfB (DGKL)[19]:	- Molekulargenetik Gruppe 1 (MG1), Set E

Osteoporose – Vitamin D Rezeptor (VDR) Gen: BsmI B/b
(OMIM #166710 – OMIM *601769)

ECAT[6]:	MG1, Set E; BsmI/ApaI/TaqI (in Kooperation mit DGKL)
ÖQUASTA[15]:	Analyte Gruppe 1, Set E: BsmI-rs1544410/ApaI-rs7975232/TaqI-rs731236 (in Kooperation mit DGKL)
RfB (DGKL)[19]:	- Molekulargenetik Gruppe 1 (MG1), Set E: BsmI/ApaI/TaqI

Phenylketonurie (PKU) – Phenylalanin Hydroxylase (PAH) Gen
(OMIM #261600 – OMIM *612349)

EMQN[8]:	Scheme PKU: mutations in the PAH gene; genotyping & interpretation; for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis)
RCPA QAP[18]:	RCPAQAP Enrolment Office, St.Leonards Molecular Genetics, Germline Mutation testing Provider: EMQN; sample: lyophilised DNA

Porphyria variegata (POR) – Protoporphyrinogen Oxidase (PPOX) Gen, andere Porphyria Gene
(OMIM #176200 - OMIM *600923)

EMQN[8]:	Scheme POR: mutations in the most frequently analysed porphyria genes; genotyping & interpretation; for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis).
RCPA QAP[18]:	RCPAQAP Enrolment Office, St.Leonards Molecular Genetics, Germline Mutation testing Provider: EMQN; sample: lyophilised DNA

Hämatologie, Hämostaseologie, Kardiovaskuläre Erkrankungen

Angiotensin I Converting Enzyme (ACE) Gen: Insertion/Deletion Polymorphism (rs4340)
(OMIM +106180)
ECAT[6]: MG1, Set C (in Kooperation mit DGKL)
ÖQUASTA[15]: Analyte Gruppe 1, Set C (in Kooperation mit DGKL)
RfB (DGKL)[19]: - Molekulargenetik Gruppe 1 (MG1), Set C

Antithrombin III Defizienz (AT3D) - Antithrombin III (AT3) / SERPINC1 Gen
(OMIM #613118 - OMIM *107300)
ECAT[6]: MG1, Set F: AT3 Cambridge TypI/II (in Kooperation mit DGKL)
INSTAND[13]: Molekulargenetik II, Set 03-A, Nr.792;
3 Exons vorgegeben, DNA
ÖQUASTA[15]: Analyte Gruppe 1, Set F: AT3 Cambridge TypI/II
(in Kooperation mit DGKL)
RfB (DGKL)[19]: - Molekulargenetik Gruppe 1 (MG1), Set F: (AT3 Cambridge TypI/II)

Arrhythmias - Pilot
EMQN[8]: Scheme CARDIO(ARR): As testing approaches are still not standardized and vary between laboratories, the exact list of genes to be tested is not specified. This pilot EQA will enable the collection of information regarding the different panels in current use; genotyping & interpretation; pilot
RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Genetics, Germline Mutation testing
Arrhytmias (Provider: EMQN)

beta-Fibrinogen (FGB) Gen: Fibrinogen beta Polymorphismus G-455A
(OMIM *134830)
ECAT[6]: MG1, Set B (in Kooperation mit DGKL)
ÖQUASTA[15]: Analyte Gruppe 1, Set B (in Kooperation mit DGKL)
RfB (DGKL)[19]: - Molekulargenetik Gruppe 1 (MG1), Set B

Fanconi Anemia Complementation Group C
CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
"Molecular Genetics" (MGL4)

Faktor II (Prothrombin) Gen: g.20210G>A Polymorphismus
(OMIM +176930)
CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
- "Molecular Genetics" (MGL1)
- Thrombophilia Mutations (TPM): Factor, II, Factor V
ECAT[6]: MG1, Set A (in Kooperation mit DGKL)
EQUALIS[9]: Product code 032, DNA: g.20210G>A

INSTAND[13]:	- Molekulargenetik I; Nr.731; Blut
	- Molekulargenetik II, Set 01-F, Nr.746; DNA
Labquality[14]:	(in Kooperation mit EQUALIS
ÖQUASTA[15]:	Analyte Gruppe 1, Set A (in Kooperation mit DGKL)
RCPA QAP[18]:	RCPAQAP Enrolment Office, St.Leonards
	Molecular Diagnostics (Molecular Haematology), Module 1
RfB (DGKL)[19]:	- Molekulargenetik Gruppe 1 (MG1), Set A
	- DNA-Isolierung (DI) und Genotypisierung
UKNEQAS[21]:	UKNEQAS Blood Coagulation
	- Factor V Leiden / Molecular Genetics of Thrombophilia Testing: Factor V Leiden, P20210A; blood, DNA

Faktor V Gen: DNA Isolierung und FV Genotypisierung

(OMIM *612309)

ECAT[6]:	DNA isolation and FV genotyping (in Kooperation mit DGKL)
RfB (DGKL)[19]:	DNA-Isolierung (DI) und Genotypisierung: FV Leiden Arg506Gln, FV His1299Arg, FV Cambridge Arg306Thr, FV Hong-Kong Arg306Gly, FII G20210A, MTHFR C677T, HFE His63Asp/Cys282Tyr/Ser65Cys)

Faktor V Gen: Leiden Mutation c.1691G>A (Arg506Gln) (rs6025)

(OMIM *612309)

CAP[3]:	CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
	- "Molecular Genetics" (MGL1)
	- Thrombophilia Mutations (TPM): Factor, II, Factor V
ECAT[6]:	MG1, Set A (in Kooperation mit DGKL)
EQUALIS[9]:	Product code 032, DNA: c.1691G>A
INSTAND[13]:	- Molekulargenetik I; Nr.730; Blut
	- Molekulargenetik II, Set 01-A, Nr.740; DNA
Labquality[14]:	in Kooperation mit EQUALIS
ÖQUASTA[15]:	Analyte Gruppe 1, Set A (in Kooperation mit DGKL)
RCPA QAP[18]:	RCPAQAP Enrolment Office, St.Leonards
	Molecular Diagnostics (Molecular Haematology), Module 1
RfB (DGKL)[19]:	- Molekulargenetik Gruppe 1 (MG1), Set A
	- DNA-Isolierung (DI) und Genotypisierung
UKNEQAS[21]:	UKNEQAS Blood Coagulation
	- Factor V Leiden / Molecular Genetics of Thrombophilia Testing: Factor V Leiden, P20210A; blood, DNA

Faktor V Gen: His1299Arg

(OMIM *612309)

ECAT[6]:	MG1, Set B (in Kooperation mit DGKL)
ÖQUASTA[15]:	Analyte Gruppe 1, Set B (in Kooperation mit DGKL)
RfB (DGKL)[19]:	- Molekulargenetik Gruppe 1 (MG1), Set B
	- DNA-Isolierung (DI) und Genotypisierung

Faktor V Gen: Cambridge Arg306Tyr
(OMIM *612309)
RfB (DGKL)[19]: - DNA-Isolierung (DI) und Genotypisierung

Faktor V Gen: Hong-Kong Arg306Gly
(OMIM *612309)
RfB (DGKL)[19]: - DNA-Isolierung (DI) und Genotypisierung

Faktor VII Gen: R353Q
(OMIM +227500)
ECAT[6]: MG1, Set F (in Kooperation mit DGKL)
ÖQUASTA[15]: Analyte Gruppe 1, Set F (in Kooperation mit DGKL)
RfB (DGKL)[19]: - Molekulargenetik Gruppe 1 (MG1), Set F

Faktor VII Activating Protease (FSAP) / Hyaluronan-binding protein 2 (HABP2) Gen: 1601G>A/Gly534Glu (FSAP-Marburg 1; rs7080536)
(OMIM *603924)
ECAT[6]: MG1, Set E (in Kooperation mit DGKL)
ÖQUASTA[15]: Analyte Gruppe 1, Set E (in Kooperation mit DGKL)
RfB (DGKL)[19]: - Molekulargenetik Gruppe 1 (MG1), Set E

Faktor XII Gen: C46T Polymorphismus (rs1801020)
(OMIM *610619)
ECAT[6]: MG1, Set B (in Kooperation mit DGKL)
ÖQUASTA[15]: Analyte Gruppe 1, Set B (in Kooperation mit DGKL)
RfB (DGKL)[19]: - Molekulargenetik Gruppe 1 (MG1), Set B

Faktor XIII, Subunit A (F13A1) Gen: Val34Leu Polymorphismus (rs5985)
(OMIM +134570)
ECAT[6]: MG1, Set B (in Kooperation mit DGKL)
ÖQUASTA[15]: Analyte Gruppe 1, Set B (in Kooperation mit DGKL)
RfB (DGKL)[19]: - Molekulargenetik Gruppe 1 (MG1), Set B

Glykoprotein Ia/IIa (GPIa/IIa) Defizienz/Kollagenrezeptor Defizienz – Integrin alpha 2 (ITGA2) Gen: C807T
(OMIM +192974)
ECAT[6]: MG1, Set E (in Kooperation mit DGKL)
INSTAND[13]: Molekulargenetik II, Set 01-C, Nr.739
ÖQUASTA[15]: Analyte Gruppe 1, Set E (in Kooperation mit DGKL)
RfB (DGKL)[19]: - Molekulargenetik Gruppe 1 (MG1), Set E

Glycoprotein IIb/IIIa (GPIIb/IIIa) Defizienz/Fibrinogenrezeptor Defizienz – Integrin beta 3, ITGB3 (GPIIIa) Gen
(OMIM +173470)
ECAT[6]: MG1, Set B (in Kooperation mit DGKL)

INSTAND[13]: Molekulargenetik II, Set 01-B, Nr.749; HPA 1a/1b
ÖQUASTA[15]: Analyte Gruppe 1, Set B (in Kooperation mit DGKL)
RfB (DGKL)[19]: - Molekulargenetik Gruppe 1 (MG1), Set B (GPIIIa L33P)

Hämophilie

Hämophilie A – Faktor VIII Defizienz
(OMIM #306700 – OMIM *300841)
UKNEQAS[21]: UKNEQAS Blood Coagulation
- Haemophilia Molecular Genetics
blood or DNA & documentation distributed

Hämophilie B – Faktor IX Defizienz,
(OMIM #306900 – OMIM *300746)
UKNEQAS[21]: UKNEQAS Blood Coagulation
- Haemophilia Molecular Genetics
blood or DNA & documentation distributed

Von Willebrand Disease (VWD), Type 1, 2, 3 - Von Willebrand Faktor (VWF)
(OMIM #193400, OMIM #613554, OMIM #277480 - OMIM *613160)
UKNEQAS[21]: UKNEQAS Blood Coagulation
- Haemophilia Molecular Genetics
blood or DNA & documentation distributed

Hypertrophic Cardiomyopathies
EMQN[8]: Scheme CARDIO(HCM): As testing approaches are still not standardized and vary between laboratories, the exact list of genes to be tested is not specified. This pilot EQA will enable the collection of information regarding the different panels in current use; genotyping & interpretation; pilot
RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Genetics, Germline Mutation testing
Hypertrophic Cardiomyopathies (Provider: EMQN)
GenQA[12]: EQA to test molecularly for cardiomyopathies - DNA samples are distributed for three clinical case scenarios. Cardiomyopathy genes. Testing can be performed using any methodology.

Long QT Syndrome (LQT1, LQT2, LQT3, LQT5, LQT6) –
KCNQ1, KCNH2, SCN5A, KCNE1, KCNE2
(OMIM #192500, #613688, #603830, #613695, #613693 –
OMIM *607542, *152427, *600163, *176261, *603796)
GenQA[12]: EQA to test molecularly for Brugada syndrome, Long QT syndrome, Catecholaminergic polymorphic ventricular tachycardia (CPVT) and general arrhythmia - DNA samples are distributed for three clinical case scenarios including one case requiring gene panel testing, a

single gene test and a predictive/family test.
Testing can be performed using any methodology.

Methylentetrahydrofolat Reduktase (MTHFR) Defizienz / Hyperhomocysteinämie - MTHFR Gen

(OMIM #236250 - OMIM *607093)

CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
C677T, A1298C
"Molecular Genetics" (MGL1)

ECAT[6]: MG1, Set A: C677T, A1298C (in Kooperation mit DGKL)

EQUALIS[9]: Product code 032, DNA: c.677C>T, c.1298A>C

INSTAND[13]: - Molekulargenetik I; Nr.732: C677T, optional A1298C; Blut
- Molekulargenetik II, Set 10,-D Nr.747: C677T, optional A1298C; DNA

Labquality[14]: c.677C>T, c.1298A>C (Kooperation mit EQUALIS)

ÖQUASTA[15]: Analyte Gruppe 1, Set A: C677T, A1298C
(in Kooperation mit DGKL)

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Diagnostics (Molecular Haematology), Module 1: Ala677Val

RfB (DGKL)[19]: - Molekulargenetik Gruppe 1 (MG1), Set A: C677T, A1298C
- DNA-Isolierung (DI) und Genotypisierung: C677T

Plasminogen-Aktivator Inhibitor 1 (PAI 1) / SERPINE1 Gen: 4G/5G

(OMIM *173360)

CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
"Molecular Genetics" (MGL1)

ECAT[6]: MG1, Set A (in Kooperation mit DGKL)

INSTAND[13]: Molekulargenetik I; Nr.735; Blut
Molekulargenetik II, Set 01-E, Nr.748; DNA

ÖQUASTA[15]: Analyte Gruppe 1, Set A (in Kooperation mit DGKL)

RfB (DGKL)[19]: - Molekulargenetik Gruppe 1 (MG1), Set A

Protein C Defizienz - Protein C (PROC) Gen

(OMIM #176860 - OMIM *612283)

INSTAND[13]: Molekulargenetik II, Set 03-C, Nr.790; 3 Exons vorgegeben

Protein S Defizienz - Protein S (PROS1) Gen

(OMIM #612336 – OMIM *176880)

INSTAND[13]: Molekulargenetik II, Set 03-D, Nr.791; 3 Exons vorgegeben

Sichelzell Anämie – Hämoglobin beta Lokus (HBB): S-Variante („Hämoglobin S")

(OMIM #603903 - OMIM +141900)

CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
- Molecular Genetics (MGL2):

Hämoglobin S/C
- Hemoglobinopathies Genotyping (HGM):
Alpha-thalassemia, Beta-thalassemia, Hemoglobin S/C
DNA specimens

UKNEQAS[21]: UKNEQAS Haematology and Transfusion
- DNA Diagnostics for Haemoglobinopathies
Genetic mutations of the alpha and beta globin genes causing Thalassaemia or Haemoglobin variants.

Thalassaemia alpha - Hämoglobin-alpha Lokus 1 (HBA1), Hämoglobin-alpha Lokus 2 (HBA2)

(OMIM #301040 - OMIM *300032, OMIM +141800, OMIM *141850)

CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
- Hemoglobinopathies Genotyping (HGM):
Alpha-thalassemia, Beta-thalassemia, Hemoglobin S/C
DNA specimens

INSTAND[13]: Molekulargenetik III, Nr.794:
Hämoglobinopathien; alpha-Globin, beta-Globin

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Diagnostics (Molecular Haematology), Module 6

UKNEQAS[21]: UKNEQAS Haematology and Transfusion
- DNA Diagnostics for Haemoglobinopathies
Genetic mutations of the alpha and beta globin genes causing Thalassaemia or Haemoglobin variants.

Thalassaemia beta - Hämoglobin beta Lokus (HBB)

(OMIM #603902 - OMIM +141900)

CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
- Hemoglobinopathies Genotyping (HGM):
Alpha-thalassemia, Beta-thalassemia, Hemoglobin S/C
DNA specimens

INSTAND[13]: Molekulargenetik III, Nr.794:
Hämoglobinopathien; alpha-Globin, beta-Globin

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Diagnostics (Molecular Haematology), Module 6

UKNEQAS[21]: UKNEQAS Haematology and Transfusion
- DNA Diagnostics for Haemoglobinopathies
Genetic mutations of the alpha and beta globin genes causing Thalassaemia or Haemoglobin variants.

Hereditäre Tumorerkrankungen

Adenomatous Polyposis of the Colon, familial (APC) - APC Gen
(OMIM #175100 - OMIM *611731)

EMQN[8]: Scheme FAP: mutations in the APC gene;
genotyping & interpretation; for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis)

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Genetics, Germline Mutation testing
Provider: EMQN; sample: lyophilised DNA

GenQA[12]: Familial adenomatous polyposis (FAP) and MUTYH associated polyposis (MAP) pathogenic variants.
EQA for the molecular testing for the presence of familial adenomatous polyposis (FAP) and MUTYH associated polyposis (MAP) - DNA samples are distributed for three clinical case scenarios. Testing can be performed using any methodology.

BRAF/MLH1 promoter methylation analysis for mismatch repair analysis

GenQA[12]: *MLH1* promoter, methylation and *BRAF* pathogenic variants.
Any molecular techniques.

Breast/ovarian cancer, familial - BRCA1 Gen, BRCA2 Gen
(OMIM #604370 - OMIM +113705, OMIM +600185)

CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
- "Molecular Genetics" (MGL3)
- BRCA1/2 Sequencing:
BRCA1/2 DNA sequencing and variant interpretation
BRCA1/2 duplication/deletion analysis

EMQN[8]:
- Full version:
Scheme BRCA-Full: mutations in the BRCA1 and BRCA2 genes; genotyping & biological/clinical interpretation; for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis);
Mutation screening in any BRCA exons required
- Genotyping-only version
Scheme BRCA-Geno: mutations in the BRCA1 + BRCA2 genes; genotyping & biological interpretation; for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis)
Mutation screening in any BRCA exons required
- Molecular testing of BRCA genes in Ovarian Cancer (vSomatic)
Scheme OVARIAN (S): Mutations in the BRCA1 and BRCA2 genes; Artificial paraffin embedded (FFPE) materials; Assessment of genotyping, and biological and clinical interpretation (BRCA1 and

BRCA2 mutation testing within the context of targeted PARP inhibitor treatment) (in cooperation with UKNEQAS[21])

- Molecular testing of BRCA genes in Ovarian Cancer (vGermline) Scheme OVARIAN (G): Mutations in the BRCA1 and BRCA2 genes; DNA (in TE buffer); Assessment of genotyping, and biological and clinical interpretation (BRCA1 and BRCA2 mutation testing within the context of targeted PARP inhibitor treatment) (in cooperation with UKNEQAS[21])

INSTAND[13]: Molekulargenetik III, Nr.779

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards

Molecular Genetics, Germline Mutation testing

- Full version; sample: lyophilised DNA
  Provider: EMQN
- Genotyping-only version; sample: lyophilised DNA
  Provider: EMQN

Molecular Genetics, Somatic Mutation testing;

- Molecular testing (Germline) of BRCA genes in Ovarian Cancer
  Provider: EMQN; genomic DNA
- Molecular testing (Somatic) of BRCA genes in Ovarian Cancer
  Provider: EMQN; sample: rolled sections of paraffin embedded materials

RfB (DGKL)[19]: in Kooperation mit EMQN

GenQA[12]: - *BRCA1* and *BRCA2* genes

Familial breast and ovarian cancer EQA - DNA samples are distributed for three clinical case scenarios. Testing can be performed using any methodology.

- BRCA germline testing in ovarian cancer pilot

BRCA1 and *BRCA2* genes

- BRCA somatic testing in ovarian cancer pilot

BRCA1 and *BRCA2* genes

Hereditary Nonpolyposis Colorectal Cancer (HNPCC)

HNPCC Typ 1 (HNPCC1) / Lynch Syndrome 1 - MSH2 Gen
(OMIM #120435 - OMIM *609309)
HNPCC Typ 2 (HNPCC2) - MLH1 Gen
(OMIM #609310 - OMIM *120436)
HNPCC Typ 5 (HNPCC5) - MSH6 Gen
(OMIM +600678)
HNPCC Typ 4 (HNPCC4) - PMS2 Gen
(OMIM +600259)

CAP[3]: CAP Headquarters, Northfield

- Defective DNA Mismatch Repair/Hereditary Nonpolyposis Colorectal Cancer (HNPCC):
  Microsatellite Instability (MSI) testing (DNA amplification)

MLH1 promoter methylation analysis
- DNA Mismatch Repair by immunohistochemistry (MMR)

EMQN[8]: Scheme HNPCC (Lynch Syndrom): mutations in the MSH2, MLH1, MSH6 and PMS2 genes;
genotyping & interpretation; for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis)

INSTAND[13]: Molekulargenetik III, Nr.783; MSH2, MLH1

QuIP[16]: - Mikrosatelliten-Instabilitätsnachweis (MSI)
10 Fälle (je 3 Objektträger) Normal- & Tumorgewebeproben
Mikrosatelliten-Instabilitätsnachweis bei kolorektalen und Endometrium-Karzinomen durch PCR etc., keine Immunhistochemie
- Mismatch Repair Defizienz (MMRD):
10 Fälle (je 5 Objektträger) Normal- und Tumorgewebe.
Immunhistochemische Expressionsanalyse von MLH1, MSH2, MSH6, PMS2
(in Kooperation mit RfB (DGKL))

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Genetics, Germline Mutation testing, Lynch Syndrome
Provider: EMQN; sample: lyophilised DNA

RfB (DGKL)[19]: in Kooperation mit EMQN)#

GenQA[12]: - *MLH1*, *MSH2*, *MSH6* and *PMS2* genes
Lynch syndrome EQA - DNA samples are distributed for three clinical case scenarios. Testing can be performed using any methodology.
- Microsatellite instability testing
MSI, *MLH1* promoter methylation and *BRAF* pathogenic variants. Any molecular techniques.

Morbus Waldenström / Macroglobulinemia Waldenstrom, Susceptibility to, 1 (WM1) – Myeloid Differentiation Primary Response Gene 88 (MYD88)

(OMIM %153600 – OMIM *602170)

INSTAND[13]: Molekulargenetik IV, Nr.766; MYD88 p.L265P

Multiple endokrine Neoplasie Typ 2 (MEN2) – RET Proto-Onkogen

(OMIM #171400 (MEN2A), OMIM #162300 (MEN2B) – OMIM +164761)

CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
"Molecular Genetics" (MGL3)

EMQN[8]: in the RET proto-oncogene;
genotyping & interpretation; for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis)

INSTAND[13]: - Molekulargenetik II, Set 03-B, Nr.738:
MEN2A, Ret Proto-Onkogen, Exon 10-16

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Genetics, Germline Mutation testing
Provider: EMQN; sample: lyophilised DNA

Retinoblastoma (RB1) – RB1 Gen
(OMIM #180200 - OMIM *614041)
EMQN[8]: Scheme RB: mutations in the RB1 gene;
genotyping & interpretation; for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis)
RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Genetics, Germline Mutation testing
Provider: EMQN; sample: lyophilised DNA

Von Hippel-Lindau-Syndrom (VHL) – VHL Gen
(OMIM #193300 – OMIM *608537)
EMQN[8]: Scheme VHL: mutations in the VHL gene;
genotyping & interpretation; for sequence based analysis (e.g. NGS / Sanger Sequencing) and copy number analysis (e.g. MLPA, NGS based CNV analysis)
RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Genetics, Germline Mutation testing
Provider: EMQN; sample: lyophilised DNA

Reproduktionsmedizinische Genetik

Adrenogenitales Syndrom (AGS) / 21-Hydroxylase-Mangel / Congenital Adrenal Hyperplasia (CAH) - CYP21A2 Gen / 21-Hydroxylase B (Cyp21B) Gen
(OMIM +201910 - OMIM *613815)

EMQN[8]:	Scheme CAH: mutations in the CYP21A2 gene; genotyping & interpretation; for targeted mutation testing as well as sequence based analysis (e.g. NGS / Sanger Sequencing), copy number analysis (e.g. MLPA, NGS based CNV analysis), and Southern blotting techniques
INSTAND[13]:	Molekulargenetik III, Nr.773
RCPA QAP[18]:	RCPAQAP Enrolment Office, St.Leonards Molecular Genetics, Germline Mutation testing Provider: EMQN; sample: lyophilised DNA
RfB (DGKL)[19]:	in Kooperation mit EMQN

Y-Chromosome Microdeletions / Azoospermia Factor (AZF) Regions
(OMIM #415000)

EMQN[8]/ EAA[5]:	Scheme AZF: microdeletions of the Y-chromosome; genotyping & interpretation; for copy number analysis (e.g. STS analysis)
INSTAND[13]:	Molekulargenetik III, Nr.788
RCPA QAP[18]:	RCPAQAP Enrolment Office, St.Leonards Molecular Genetics, Germline Mutation testing Provider: EMQN; sample: lyophilised DNA
RfB (DGKL)[19]:	in Kooperation mit EMQN

Immungenetik, HLA-Typisierung

(Einrichtungen, die nur derartige Analysen durchführen benötigen keine Zulassung gem. § 68 GTG)

KIR Genotyping

UKNEQAS[21]: KIR Genotyping
Participants can register for assessment for the presence/absence of: KIR2DL1, KIR2DL2, KIR2DL3, KIR2DL4, KIR2DL5, KIR3DL1, KIR3DL2, KIR3DL3, KIR3DS1, KIR2DS1, KIR2DS2, KIR2DS3, KIR2DS4, KIR2DS5, KIR2DP1, KIR3DP1

Major Histocompatibility Complex (MHC) Class I - HLA A, HLA B, HLA C

(OMIM +142800, OMIM +142830, OMIM *142840)

CAP[3]: CAP Headquarters, Northfield
HLA Molecular Typing: HLA-A, B, C (DML)

ETRL[11]: HLA typing

INSTAND[13]/EFI[7]: Immungenetik/HLA; Nr.444; Molekulargenetische HLA-Klasse I Typisierung:
444-1: HLA-A niedrig auflösend
444-2: HLA-B niedrig auflösend
444-3: HLA-C niedrig auflösend
444-4: HLA-A hoch auflösend
444-5: HLA-B hoch auflösend
444-6: HLA-C hoch auflösend
444-7: HLA-B*57:01 Bestimmung -> siehe Abacavir-Hypersensitivität, Kapitel „Pharmakogenetik“)

UKNEQAS[21]: UKNEQAS Histocompatibility & Immunogenetics
- HLA Phenotyping (Scheme 1A)
  Phenotyping for HLA-A, B, Cw, DR and DQ by serological methods
- HLA-B27 Testing (Scheme 1B)
  presence or absence of the HLA-B27 antigen; blood
  (-> siehe Spondyloarthropatie/Mb.Bechterew, Kapitel “Autoimmunerkrankungen”)
- HLA DNA Typing (Scheme 4A1)
  Genotyping for HLA-A, B, C, DRB1, DRB3/4/5, DQA1, DQB1, DPA1 by DNA techniques at 1st field resolution; blood
- HLA DNA Typing (Scheme 4A2)
  Genotyping for HLA-A , B, C, DRB1, DRB3/4/5, DQA1, DQB1, DPA1, DPB1 by DNA techniques to 2nd field resolution; blood
- HLA B*57:01 (Scheme 7) Typing for Drug Hypersensitivity
  Typing for presence/absence of HLA-B*57:01; blood
  (-> siehe Abacavir-Hypersensitivität, Kapitel „Pharmakogenetik“)
  DNA techniques; blood
- Interpretative HLA Genotype (Scheme 4A1i)
- Education Scheme

HLA typing of rare and unusual samples by serological and DNA-based techniques; blood or DNA

Major Histocompatibility Complex (MHC) Class II – HLA-DR, HLA-DQ, HLA-DP, insbesondere HLA-DRB1, HLA-DQB1

(OMIM *142857, OMIM *604305)

CAP[3]: CAP Headquarters, Northfield:
HLA Molecular Typing: HLA-DR, DQ, DP (DML)

ETRL[11]: HLA typing

INSTAND[13]/EFI[7]: Immungenetik/HLA, Nr.442: Molekulargenetische HLA-Klasse II Typisierung:
442-1: HLA-DRB niedrig auflösend
444-2: HLA-DQB1 niedrig auflösend
444-3: HLA-DRB1 hoch auflösend
444-4: HLA-DQB1 hoch auflösend
444-5: HLA-DRB1-DNA-Typisierung des sog. „Rheumatoiden Epitops“ -> siehe auch Rheumatoide Arthritis, Kapitel „Autoimmunerkrankungen“)

UHKT[20] Detection of HLA alleles linked with diseases
- Alleles of DQ loci alleles associated with coeliac disease (DQA1*02, *03, *051, DQB1*02, *03:02)

UKNEQAS[21]: UKNEQAS Histocompatibility & Immunogenetics
- HLA Phenotyping (1A)
  Phenotyping for HLA-A, B, Cw, DR and DQ by serological methods DNA techniques; blood
- HLA DNA Typing (4A1)
  Genotyping for HLA-A, B, C, DRB1, DRB3/4/5, DQA1, DQB1, DPA1 by DNA techniques at 1st field resolution; blood
- HLA DNA Typing (4A2)
  Genotyping for HLA-A , B, C, DRB1, DRB3/4/5, DQA1, DQB1, DPA1, DPB1 by DNA techniques to 2nd field resolution; blood
- HLA Genotyping for Coeliac and Other HLA Associated Diseases (scheme 8)
  - Education Scheme
  HLA typing of rare and unusual samples by serological and DNA-based techniques; blood or DNA

Autoimmunerkrankungen, HLA-assoziierte Erkrankungen

Autoimmunität – HLA-DQ

INSTAND[13]: Molekulargenetik III, Nr.772:
HLA DQA1 05, HLA DQB1 02, HLA DQB1 03:02

Autoimmunität - Tumor Nekrosis Faktor alpha (TNFa) Gen: G-238A, G-308A

(OMIM *191160)

ECAT[6]: MG2, Set C (in Kooperation mit DGKL)

ÖQUASTA[15]: Analyte Gruppe 2, Set C (in Kooperation mit DGKL)

RfB (DGKL)[19]: Molekulargenetik Gruppe 2 (MG2), Set C

Birdshot Chorioretinopathy (BR)

(OMIM 605808)

CAP[3]: CAP Headquarters, Northfield
- HLA Disease Association, Drug Risk (DADR2):
HLA-A*29:01, HLA-A*29:02, HLA-DQA1*04:01, HLA-DQA1*05:01, HLA-DQB1*03:02, HLA-DQB1*06:02, HLA-DRB1*03:01, HLA-DRB1*03:02, HLA-DRB1*04:02, HLA-DRB1*04:03, HLA-DRB1*04:06, HLA-DRB1*08:02, HLA-DRB1*08:04, HLA-DRB1*14:04, HLA-DRB1*14:05, HLA-DRB1*14:08, HLA-DRB1*15:01, HLA-DRB1*15:02, DQA1*02, DQA1*03, DQA1*05, DQB1*02:01, DQB1*02:02

Coeliac Disease (CD) / Zöliakie

(OMIM #212750)

CAP[3]: CAP Headquarters, Northfield
- HLA Disease Association, Drug Risk (DADR2):
HLA-A*29:01, HLA-A*29:02, HLA-DQA1*04:01, HLA-DQA1*05:01, HLA-DQB1*03:02, HLA-DQB1*06:02, HLA-DRB1*03:01, HLA-DRB1*03:02, HLA-DRB1*04:02, HLA-DRB1*04:03, HLA-DRB1*04:06, HLA-DRB1*08:02, HLA-DRB1*08:04, HLA-DRB1*14:04, HLA-DRB1*14:05, HLA-DRB1*14:08, HLA-DRB1*15:01, HLA-DRB1*15:02, DQA1*02, DQA1*03, DQA1*05, DQB1*02:01, DQB1*02:02

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Genetics, HLA Genotyping Program:
Coeliac Disease HLA genotyping – Pilot

UHKT[20] Detection of HLA alleles linked with diseases
- Alleles of DQ loci alleles associated with coeliac disease (DQA1*02, *03, *051, DQB1*02, *03:02)

UKNEQAS[21]: UKNEQAS Histocompatibility and Immunogenetics
- HLA Genotyping for Coeliac and Other HLA Associated Diseases (Scheme 8)

Goodpasture Syndrome / Antiglomerular basement membrane disease (ABM)
(OMIM 233450)

CAP[3]: CAP Headquarters, Northfield
- HLA Disease Association, Drug Risk (DADR2):
HLA-A*29:01, HLA-A*29:02, HLA-DQA1*04:01, HLA-DQA1*05:01, HLA-DQB1*03:02, HLA-DQB1*06:02, HLA-DRB1*03:01, HLA-DRB1*03:02, HLA-DRB1*04:02, HLA-DRB1*04:03, HLA-DRB1*04:06, HLA-DRB1*08:02, HLA-DRB1*08:04, HLA-DRB1*14:04, HLA-DRB1*14:05, HLA-DRB1*14:08, HLA-DRB1*15:01, HLA-DRB1*15:02, DQA1*02, DQA1*03, DQA1*05, DQB1*02:01, DQB1*02:02

HLA-assoziierte Erkrankungen

UHKT[20] Detection of HLA alleles linked with diseases
- B*27 (association with Morbus Bechterev and other rheumatoid autoimmune diseases)
- Alleles of DQ loci alleles associated with coeliac disease (DQA1*02, *03, *051, DQB1*02, *03:02)
- DQB1*06:02 (association with narcolepsy)

UKNEQAS[21]: UKNEQAS Histocompatibility and Immunogenetics
- HLA Genotyping for Coeliac and Other HLA Associated Diseases (scheme 8)

Morbus Crohn / Inflammatory Bowel Disease 1 (IBD1) –
Nucleotide-binding oligomerisation domain Protein 2 (NOD2) Gen: R702W, G908R, L1007fsinsC
ATP-Binding Cassette, Subfamily B, Member 1 (ABCB1) / Multidrug Resistance 1 (MDR1) Gen: c.3435C>T
(OMIM #266600 – OMIM *605956, OMIM *171050)

ECAT[6]: in Kooperation mit DGKL
- MG1, Set D: NOD2 (R702W, G908R, L1007fins C)
- MG2, Set E: ABCB1 (MDR1) c.3435>T

INSTAND[13]: Molekulargenetik III, Nr.774

ÖQUASTA[15]:
- Analyte Gruppe 1, Set D (in Kooperation mit DGKL): NOD2
- Analyte Gruppe 2, Set E (in Kooperation mit DGKL): ABCB1 (MDR1) c.3435C>T

RfB (DGKL)[19]:
- Molekulargenetik Gruppe 1 (MG1), Set D: NOD2
- Molekulargenetik Gruppe 2 (MG2), Set E: ABCB1 (MDR1) c.3435C>T

Myositis / Idiopathic inflammatory myopathy (IM)
(OMIM %160750)

CAP[3]: CAP Headquarters, Northfield
- HLA Disease Association, Drug Risk (DADR2):
HLA-A*29:01, HLA-A*29:02, HLA-DQA1*04:01, HLA-DQA1*05:01,

HLA-DQB1*03:02, HLA-DQB1*06:02, HLA-DRB1*03:01, HLA-DRB1*03:02, HLA-DRB1*04:02, HLA-DRB1*04:03, HLA-DRB1*04:06, HLA-DRB1*08:02, HLA-DRB1*08:04, HLA-DRB1*14:04, HLA-DRB1*14:05, HLA-DRB1*14:08, HLA-DRB1*15:01, HLA-DRB1*15:02, DQA1*02, DQA1*03, DQA1*05, DQB1*02:01, DQB1*02:02

Narcolepsy 1 (NRCLP1) / Narkolepsie (N)
(OMIM #161400)

CAP[3]: CAP Headquarters, Northfield
- HLA Disease Association, Drug Risk (DADR2):
HLA-A*29:01, HLA-A*29:02, HLA-DQA1*04:01, HLA-DQA1*05:01, HLA-DQB1*03:02, HLA-DQB1*06:02, HLA-DRB1*03:01, HLA-DRB1*03:02, HLA-DRB1*04:02, HLA-DRB1*04:03, HLA-DRB1*04:06, HLA-DRB1*08:02, HLA-DRB1*08:04, HLA-DRB1*14:04, HLA-DRB1*14:05, HLA-DRB1*14:08, HLA-DRB1*15:01, HLA-DRB1*15:02, DQA1*02, DQA1*03, DQA1*05, DQB1*02:01, DQB1*02:02

UHKT[20] Detection of HLA alleles linked with diseases
DQB1*06:02 (association with narcolepsy)

Pemphigus vulgaris, familial (PV)
(OMIM 169610)

CAP[3]: CAP Headquarters, Northfield
- HLA Disease Association, Drug Risk (DADR2):
HLA-A*29:01, HLA-A*29:02, HLA-DQA1*04:01, HLA-DQA1*05:01, HLA-DQB1*03:02, HLA-DQB1*06:02, HLA-DRB1*03:01, HLA-DRB1*03:02, HLA-DRB1*04:02, HLA-DRB1*04:03, HLA-DRB1*04:06, HLA-DRB1*08:02, HLA-DRB1*08:04, HLA-DRB1*14:04, HLA-DRB1*14:05, HLA-DRB1*14:08, HLA-DRB1*15:01, HLA-DRB1*15:02, DQA1*02, DQA1*03, DQA1*05, DQB1*02:01, DQB1*02:02

Psoriasis 1, susceptibility to (PSORS1) / Psoriasis vulgaris (P)
(OMIM #177900)

CAP[3]: CAP Headquarters, Northfield
- HLA Disease Association, Drug Risk (DADR2):
HLA-A*29:01, HLA-A*29:02, HLA-DQA1*04:01, HLA-DQA1*05:01, HLA-DQB1*03:02, HLA-DQB1*06:02, HLA-DRB1*03:01, HLA-DRB1*03:02, HLA-DRB1*04:02, HLA-DRB1*04:03, HLA-DRB1*04:06, HLA-DRB1*08:02, HLA-DRB1*08:04, HLA-DRB1*14:04, HLA-DRB1*14:05, HLA-DRB1*14:08, HLA-DRB1*15:01, HLA-DRB1*15:02, DQA1*02, DQA1*03, DQA1*05, DQB1*02:01, DQB1*02:02

Rheumatoide Arthritis (RA) - HLA-DRB1
(OMIM #180300 - OMIM *142857
INSTAND[13]/EFI[7]: Immungenetik/HLA, Nr.442: Molekulargenetische HLA-Klasse II Typisierung:
444-5: HLA-DRB1-DNA-Typisierung des sog. „Rheumatoiden Epitops“

Rheumatoide Arthritis, systemic juvenile, susceptibility to - Interleukin 6 (IL-6) Gen: G-174C
(OMIM #604302 - OMIM *147620)
ECAT[6]: MG2, Set F (in Kooperation mit DGKL)
ÖQUASTA[15]: Analyte Gruppe 2, Set F (in Kooperation mit DGKL)
RfB (DGKL)[19]: Molekulargenetik Gruppe 2 (MG2), Set F

Spondyloarthropathy 1 (SPDA1) / Spondylitis ankylosans / Morbus Bechterew – MHC Class I B (HLA-B): HLA B27 Bestimmung
(OMIM #106300 - OMIM *142830)
CAP[3]: CAP Headquarters, Northfield:
HLA-B27 Typing
ECAT[6]: MG2, Set C (in Kooperation mit DGKL)
INSTAND[13]/EFI[7]: Immungenetik/HLA; Nr.440
wahlweise molekulargenetisch und/oder serologisch)
ÖQUASTA[15]: Analyte Gruppe 2, Set C (in Kooperation mit DGKL)
RfB (DGKL)[19]: Molekulargenetik Gruppe 2 (MG2), Set C
UHKT[20] Detection of HLA alleles linked with diseases
B*27 (association with Morbus Bechterev and other rheumatoid autoimmune diseases)
UKNEQAS[21]: UKNEQAS Histocompatibility and Immunogenetics
- HLA-B27 Testing (1B)
presence or absence of the HLA-B27 antigen; blood

Pharmakogenetik, individualisierte Therapie

(Einrichtungen, die nur derartige Analysen durchführen benötigen keine Zulassung gem. § 68 GTG)

Abacavir Hypersensitivität - MHC Class I B (HLA-B): HLA-B*5701 Bestimmung
(OMIM +142830)

CAP[3]:	CAP Headquarters, Northfield - Pharmacogenetics: Genotype detection and interpretation HLA-B*5701, HLA-B*1502 (PGX2) - HLA Disease Association, Drug Risk: HLA-A*31:01, HLA-B*13:01, HLA-B*15:02, HLA-B*57:01, HLA-B*58:01 (DADR1)
ECAT[6]:	MG2, Set E (in Kooperation mit DGKL)
INSTAND[13]/EFI[7]:	Immungenetik/HLA, Nr.444; Molekulargenetische HLA-Klasse I Typisierung: 444-7: HLA-B*57:01 Bestimmung)
ÖQUASTA[15]:	Analyte Gruppe 2, Set E (in Kooperation mit DGKL)
RfB (DGKL)[19]:	Molekulargenetik Gruppe 2 (MG2), Set E
UKNEQAS[21]:	UKNEQAS Histocompatibility and Immunogenetics - HLA B*57:01 (scheme 7) Typing for Drug Hypersensitivity Typing for presence/absence of HLA-B*57:01; blood
RCPA QAP[18]:	RCPAQAP Enrolment Office, St.Leonards Molecular Genetics, HLA Genotyping Program: Human Leukocyte Antigen B*57

Butyrylcholinesterase Defizienz /Postanästhetische Apnoe - Butyrylcholinesterase (BCHE) Gen: Atypische (A)-Variante (N70G), K-Variante (A539T)
(OMIM +177400)

ECAT[6]:	MG2, Set A (in Kooperation mit DGKL)
ÖQUASTA[15]:	Analyte Gruppe 2, Set A (in Kooperation mit DGKL)
RfB (DGKL)[19]:	Molekulargenetik Gruppe 2 (MG2), Set A

Coumarin/Warfarin Resistenz/Sensitivität - Cyp2C9 Gen, VKORC1 Gen:
(OMIM #122700)

Cytochrom p450 2C9 (CYP2C9) Gen: Cyp2C9*2 (Arg144Cys; rs1799853), Cyp2C9*3 (Ile359Leu; rs1047910)
(OMIM *601130)

CAP[3]:	CAP Headquarters, Northfield - Pharmacogenetics: Genotype detection and interpretation CYP2C9 (PGX)
ECAT[6]:	MG2, Set A (in Kooperation mit DGKL)
EQUALIS[9]:	Product code 272
INSTAND[13]:	Molekulargenetik III, Nr.775
ÖQUASTA[15]:	Analyte Gruppe 2, Set A (in Kooperation mit DGKL)
RfB (DGKL)[19]:	Molekulargenetik Gruppe 2 (MG2), Set A

Vitamin K-Epoxidreductase Complex, Subunit1 (VKORC1) Gen
(OMIM *608547)

CAP[3]:	CAP Headquarters, Northfield - Pharmacogenetics: Genotype detection and interpretation VKORC1 (PGX)
ECAT[6]:	MG1, Set B: G-1693A/C1173T (in Kooperation mit DGKL)
INSTAND[13]:	Molekulargenetik III, Nr.775
ÖQUASTA[15]:	Analyte Gruppe 1, Set B: G-1693A/C1173T (in Kooperation mit DGKL)
RfB (DGKL)[19]:	Molekulargenetik Gruppe 1 (MG1), Set B (G-1693A/C1173T)

Dapsone Hypersensitivity Syndrome (DHS) - MHC Class I, B (HLA-B): HLA-B*13:01 Bestimmung (OMIM +142830)

CAP[3]:	CAP Headquarters, Northfield - HLA Disease Association, Drug Risk: HLA-A*31:01, HLA-B*13:01, HLA-B*15:02, HLA-B*57:01, HLA-B*58:01 (DADR1)

Dihydropyrimidin-Dehydrogenase Defizienz / 5-Fluoro-Uracil Toxizität - Dihydropyrimidin-Dehydrogenase (DPD) Gen: Exon 14 skipping Mutation IVS14 G→A +1, D949V (c.2846A>T; rs67376798), DPD*13 (c.1679T>G; rs55886062)
(OMIM #274270 – OMIM *612779)

CAP[3]:	CAP Headquarters, Northfield Pharmacogenetics (PGX): Genotype detection and interpretation DPYD (PGX3)
ECAT[6]:	MG2, Set A (in Kooperation mit DGKL): DPYD*2A (Ex 14 skipping), DPYD*13, DPYD D949V
INSTAND[13]:	Molekulargenetik III, Nr.777
ÖQUASTA[15]:	- Analyte Gruppe 2, Set A (in Kooperation mit DGKL): DPD Ex 14 skipping - Analyte Gruppe 2, Set A (in Kooperation mit DGKL): DPD D949V, c.2846A>T (rs67376798) - Analyte Gruppe 2, Set A (in Kooperation mit DGKL): DPD*13, c.1679T>G (rs55886062)
RfB (DGKL)[19]:	Molekulargenetik Gruppe 2 (MG2), Set A: DPYD *2A (Ex 14 skipp), DPYD*13, DPYD D949V (rs67376798)

Hepatitis C Virus Treatment Response - Interleukin 28B (IL-28B) Gen: C/T (rs12979860)
(OMIM *607402)

CAP[3]:	CAP Headquarters, Northfield - Pharmacogenetics (PGX): Genotype detection and interpretation IL28B (rs12979860) (PGX1)

ECAT[6]: BsmI/ApaI/TaqI
MG2, Set F (in Kooperation mit DGKL)
INSTAND[13]: Molekulargenetik III, Nr.777
ÖQUASTA[15]: Analyte Gruppe 2, Set F (in Kooperation mit DGKL)
RfB (DGKL)[19]: Molekulargenetik Gruppe 2 (MG2), Set F

Poor drug metabolism, Cyp2B6-related (Efavirenz)
- Cytochrom p450 2B6 (CYP2B6) Gen: Cyp2B6*6
(OMIM *123930)
ECAT[6]: BsmI/ApaI/TaqI
MG2, Set E (in Kooperation mit DGKL)
ÖQUASTA[15]: Analyte Gruppe 2, Set E (in Kooperation mit DGKL)
RfB (DGKL)[19]: Molekulargenetik Gruppe 2 (MG2), Set E

Poor drug metabolism, Cyp2C8-related
- Cytochrom p450 2C8 (CYP2C8) Gen: K399R
(OMIM +601129)
ECAT[6]: MG2, Set A (in Kooperation mit DGKL)
ÖQUASTA[15]: Analyte Gruppe 2, Set A (in Kooperation mit DGKL)
RfB (DGKL)[19]: Molekulargenetik Gruppe 2 (MG2), Set A

Poor drug metabolism, Cyp2C19-related (Opremazol, Proguanil, Clopidogrel, Mephenytoin)
- Cytochrom p450 2C19 (CYP2C19) Gen
(OMIM #609535 - OMIM *124020)
CAP[3]: CAP Headquarters, Northfield
- Pharmacogenetics: Genotype detection and interpretation CYP2C19 (PGX)
ECAT[6]: MG2, Set D: Cyp2C19 *2/*17, Cyp2C19*3 (in Kooperation mit DGKL)
EQUALIS[9]: Product code 272
INSTAND[13]: Molekulargenetik III, Nr.775
ÖQUASTA[15]: Analyte Gruppe 2, Set D: Cyp2C19 *2/*17, CYP2C19*3 (in Kooperation mit DGKL)
RfB (DGKL)[19]: Molekulargenetik Gruppe 2 (MG2), Set D (Cyp2C19 *2/*17, Cyp2C19*3)

Poor drug metabolism, Cyp2D6-related (Debrisoquin, Spartein, Nortriptylin, Codein)
- Cytochrom p450 2D6 (CYP2D6) Gen
(OMIM #608902 - OMIM *124030)
CAP[3]: CAP Headquarters, Northfield
- Pharmacogenetics: Genotype detection and interpretation CYP2D6 (PGX)
ECAT[6]: MG2, Set D (in Kooperation mit DGKL)
EQUALIS[9]: Product code 272

INSTAND[13]: Molekulargenetik III, Nr.777
ÖQUASTA[15]: Analyte Gruppe 2, Set D (in Kooperation mit DGKL)
RfB (DGKL)[19]: Molekulargenetik Gruppe 2 (MG2), Set D

Poor drug metabolism, Cyp3A4-related (Tacrolimus, Cyclosporin A)
- Cytochrom p450 3A4 (CYP3A4) Gen: Cyp3A4*22
(OMIM *124010)
CAP[3]: CAP Headquarters, Northfield
- Pharmacogenetics: Genotype detection and interpretation CYP3A4 (PGX)
ECAT[6]: MG2, Set F (in Kooperation mit DGKL)
ÖQUASTA[15]: Analyte Gruppe 2, Set F (in Kooperation mit DGKL)
RfB (DGKL)[19]: Molekulargenetik Gruppe 2 (MG2), Set F

Poor drug metabolism, Cyp3A5-related (Tacrolimus, Cyclosporin A)
- Cytochrom p450 3A5 (CYP3A5) Gen: Cyp3A5*3
(OMIM *605325)
CAP[3]: CAP Headquarters, Northfield
- Pharmacogenetics: Genotype detection and interpretation CYP3A5 (PGX)
ECAT[6]: MG1, Set F (in Kooperation mit DGKL)
ÖQUASTA[15]: Analyte Gruppe 1, Set F (in Kooperation mit DGKL)
RfB (DGKL)[19]: Molekulargenetik Gruppe 1 (MG1), Set F

Stevens-Johnson Syndrome, Allopurinol induced (ASJ) - MHC Class I, B (HLA-B): HLA-B*58:01 Bestimmung (OMIM +142830)
CAP[3]: CAP Headquarters, Northfield
- HLA Disease Association, Drug Risk: HLA-A*31:01, HLA-B*13:01, HLA-B*15:02, HLA-B*57:01, HLA-B*58:01 (DADR1)

Stevens-Johnson Syndrome, Carbamezepine induced (CSJ) - MHC Class I, A (HLA-A), MHC Class I, B (HLA-B): HLA-A*31:01, HLA-B*15:02 Bestimmung
(OMIM +142800, OMIM +142830)
CAP[3]: CAP Headquarters, Northfield
- Pharmacogenetics: Genotype detection and interpretation HLA-B*5701, HLA-B*1502 (PGX2)
- HLA Disease Association, Drug Risk: HLA-A*31:01, HLA-B*13:01, HLA-B*15:02, HLA-B*57:01, HLA-B*58:01 (DADR1)

Thiopurin-S-Methyltransferase (TPMT) Defizienz - TPMT Gen
(OMIM #610460, OMIM *187680)

CAP[3]:	CAP Headquarters, Northfield Pharmacogenetics: Genotype detection and interpretation TPMT (PGX3)
ECAT[6]:	MG2, Set A (in Kooperation mit DGKL)
INSTAND[13]:	Molekulargenetik III, Nr.775
ÖQUASTA[15]:	Analyte Gruppe 2, Set A (in Kooperation mit DGKL)
RfB (DGKL)[19]:	Molekulargenetik Gruppe 2 (MG2), Set A

Somatische Mutationen – Molekulare Hämatoonkologie, Molekularpathologie
(Einrichtungen, die nur derartige Analysen durchführen benötigen keine Zulassung gem. § 68 GTG)

Cell Free DNA

CAP[3]: CAP Headquarters, Northfield
CFDNA: Mix of fragmented cell line gDNA and biosynthetic DNA; not intended for laboratories that perform circulating tumor cell analysis; potential targets included in this Survey are BRAF V600E, EGFR T790M, IDH1, R132C, KRAS G12D, and NRAS Q61R, all within a range of 0.1 to 1.0%

Chimärismus

INSTAND[13]: Engraftment- und Chimärismus-Monitoring, Nr.618
Chimärismus-Diagnostik nach Stammzell-Transplantation

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Diagnostics (Molecular Haematology), Module 5

UHKT[20] Quantitative Analysis of Cell Chimerism after Allogeneic HSCT

UKNEQAS[21]: UKNEQAS Leucocyte Immunophenotyping
- Post-SCT Chimerism Monitoring
  Chimerism testing in the monitoring of patients post stem cell transplant.

DNA Mismatch Repair

CAP[3]: CAP Headquarters, Northfield
- DNA Mismatch Repair (MMR) by immunohistochemistry

Gastrointestinal stromal tumor (GIST)
(OMIM #606764)
- KIT-Onkogen (OMIM *164920)
- Platelet derived growth factor receptor alpha (PGDFRA) Gen (OMIM *173490)

CAP[3]: CAP Headquarters, Northfield
- "Solid Tumors-Other": KIT/PDGRFA (KIT)
- Multigene Tumor Panel (MTP): BRAF, EGFR, HER2 (ERBB2), KIT, KRAS, NRAS, PDGFRA, PIK3CA
  Designed for laboratories performing both non-NGS multiplex testing as well as non-multiplexed platforms (eg, Sanger sequencing)

EMQN[8]: "Oncogene Panel Testing" (ONCOPANEL): mutations in the EGFR, PIK3CA, KRAS, HRAS, NRAS, KIT, TP53 and BRAF genes;
This scheme is being offered to help labs using high thorough put technologies (e.g., NGS, MassArray etc) accurately validate assay sensitivity and specificity; for specific tumour types, please register for the relevant Lung, Melanoma or Colorectal scheme;

	Rolled sections of formalin fixed paraffin embedded (FFPE) materials designed to simulate a real patient sample
QuIP[16]:	- GIST 10 Fälle (je 3 Objekträger) für die Mutationsanalyse des relevanten Exons (Exon 9, Exon 11 von Kit und Exon 18 von PDGFRA) - CD117 2 TMA-Schnitte mit jeweils ca. 10 Stanzen
GenQA[12]:	Molecular analysis of Gastro-Intestinal Stromal Tumours: *KIT* and *PDGFRA* genes Any molecular techniques including FISH and IHC.

Glioma Susceptibility 1 (GLM1)

(OMIM #137800)

CAP[3]:	CAP Headquarters, Northfield Glioma: MGMT, IDH1, IDH2, 10q (PTEN) deletion (GLI)
RCPA QAP[18]:	RCPAQAP Enrolment Office, St.Leonards Molecular Genetics, Sample Exchange Program: - IDH1/IDH2 Sequencing; sample: FFPE - MGMT Hypermethylation; sample: FFPE - PTEN; sample: DNA

Hairy Cell Leukaemia – BRAF V600E

(OMIM *164757)

CAP[3]:	CAP Headquarters, Northfield - Multigene Tumor Panel (MTP): BRAF, EGFR, HER2 (ERBB2), KIT, KRAS, NRAS, PDGFRA, PIK3CA Designed for laboratories performing both non-NGS multiplex testing as well as non-multiplexed platforms (eg, Sanger sequencing)
ECAT[6]:	MG2, Set B: V600E (in Kooperation mit DGKL)
EMQN[8]:	"Oncogene Panel Testing" (ONCOPANEL): mutations in the EGFR, PIK3CA, KRAS, HRAS, NRAS, KIT, TP53 and BRAF genes; This scheme is being offered to help labs using high thorough put technologies (e.g., NGS, MassArray etc) accurately validate assay sensitivity and specificity; for specific tumour types, please register for the relevant Lung, Melanoma or Colorectal scheme; Rolled sections of formalin fixed paraffin embedded (FFPE) materials designed to simulate a real patient sample
ÖQUASTA[15]:	Analyte Gruppe 2, Set B: BRAF V600E, BRAF V600K (in Kooperation mit DGKL)
RfB (DGKL)[19]:	- Molekulargenetik Gruppe 2 (MG2), Set B: BRAF V600E, BRAF V600K - circulating tumor DNA (ctDNA), Set B: BRAF V600E (rs113488022)
UKNEQAS[21]:	UKNEQAS Leucocyte Immunophenotyping - BRAF V600E Hairy Cell Leukaemia

Hematologic Malignancies

CAP[3]: CAP Headquarters, Northfield

- Next-Generation Sequencing - Hematologic Malignancies (NGSHM)
  Methods-based proficiency challenge for laboratories performing targeted next-generation sequencing of genes or mutation hotspots in hematologic malignancies.
  Laboratories will be asked to identify somatic single nucleotide variants and small insertions or deletions in the following genes: ASXL1, ATM, BRAF, CALR, CEBPA, CREBBP, CSF3R, DNMT3A, EZH2, FLT3, IDH1, IDH2, JAK2, KIT, KMT2D, MPL, MYD88, NOTCH1, NPM1, SF3B1, SRSF2, TET2, TP53, U2AF1

Kolorektales Karzinom / Wirksamkeit von Panitumumab oder Cetuximab

- V-Raf Murine Sarcoma Viral Oncogene Homolog B1 (BRAF) (OMIM *164757)
- V-Ki-Ras2 Kirsten Rat Sarcoma Viral Oncogene Homolog (KRAS) (OMIM *190070)
- Neuroblastoma Ras Viral Oncogene Homolog (NRAS) (OMIM *164790)

CAP[3]: CAP Headquarters, Northfield

- "Solid Tumors-Other", KRAS
- "Solid Tumors-Other", BRAF (therapy and HNPCC)
- Multigene Tumor Panel (MTP): BRAF, EGFR, HER2 (ERBB2), KIT, KRAS, NRAS, PDGFRA, PIK3CA
  Designed for laboratories performing both non-NGS multiplex testing as well as non-multiplexed platforms (eg, Sanger sequencing)

ECAT[6]: MG2, Set B: BRAF V600E, BRAF V600K, K-Ras Codon 12, 13, 61 (in Kooperation mit DGKL)

EMQN[8]:

- "Molecular testing in sporadic Colorectal Cancer" (COLOREC): mutations in the KRAS, BRAF, NRAS and PIK3CA genes; genotyping & biological/clinical interpretation; paraffin embedded material
- "Oncogene Panel Testing" (ONCOPANEL): mutations in the EGFR, PIK3CA, KRAS, HRAS, NRAS, KIT, TP53 and BRAF genes;
  This scheme is being offered to help labs using high thorough put technologies (e.g., NGS, MassArray etc) accurately validate assay sensitivity and specificity; for specific tumour types, please register for the relevant Lung, Melanoma or Colorectal scheme;
  Rolled sections of formalin fixed paraffin embedded (FFPE) materials designed to simulate a real patient sample

ESP-EQA[10] wild-type RAS status: KRAS Exon 2,3,4; NRAS Exon 2,3,4;
BRAF testing; genotype and interpretation; full RAS testing required, BRAF testing optional

INSTAND[13]: Molekulargenetik III, Nr.776; Codon 12/13/61 Mutationen

ÖQUASTA[15]: Analyte Gruppe 2, Set B: BRAF V600E, BRAF V600K, K-Ras Codon 12/13/61
(in Kooperation mit DGKL)

QuIP[16]: - BRAF-Mutationsanalyse
(10 Fälle maligner Tumore auf jeweils 3 Objektträgern)
- RAS-Mutationsanalyse:
10 Fälle (je 4 Objektträger) von metastasierten kolorektalen Karzinomen zu untersuchen auf Mutationen in den Exonen 2 (p.12,13), 3 (p.59,61) und 4 (p.117,146);
NRAS Exone 2 (p.12,13), 3 (p.59,61) und 4 (p.117,146)

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
- Molecular Genetics, Oncology Program;
Mutation Detection in Colorectal Cancer
sample: FFPE Tissue (patient-derived tissue & synthetic samples)
- Molecular Genetics, Somatic Mutation Testing;
Molecular testing in sporadic colorectal cancer
Provider: EMQN; sample: real tissue and artificial paraffin embedded materials
Molecular Genetics, Generic Report;
- Mutation Detection in Colorectal Cancer (KRAS, NRAS)

RfB (DGKL)[19]: - Molekulargenetik Gruppe 2 (MG2), Set B (BRAF V600E, BRAF V600K)
- Molekulargenetik Gruppe 2 (MG2), Set B (K-Ras Codon 12, 13, 61)
- circulating tumor DNA (ctDNA), Set A: KRAS C12, C13 (rs121913530, rs121913529, rs121913535, rs112445441, rs121913238, rs121913240, rs17851045)
- circulating tumor DNA (ctDNA), Set B: BRAF V600E (rs113488022)

GenQA[12]: Molecular analysis in colorectal cancer (2 distributions):
KRAS, *NRAS* and *BRAF* genes
Any molecular techniques including FISH and IHC.

Leukämie:

- Leukämie, akut myeloisch (AML) (OMIM #601626)
- Leukämie, akut lymphatisch (ALL) (OMIM %613065)
- Leukämie, chronisch myeloisch (CML) (OMIM #608232)
- Leukämie, chronisch lymphatisch (CLL) (OMIM #151400)
- Myeloproliferative Erkrankungen

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
- Molecular Diagnostics (Molecular Haematology), Module 3: BCR-ABL Qualitative Testing, Quantitative Monitoring
- Molecular Diagnostics (Molecular Haematology), Module 4: T-cell receptor gene, Immunoglobulin heavy chain gene:
- Molecular Diagnostics (Molecular Haematology), Module 7: Jak 2
- Molecular Diagnostics (Molecular Haematology), Module 9: FLT-3 ITD

- Molecular Diagnostics (Molecular Haematology), Module 10: NPM1 – Pilot
- Molecular Diagnostics (Molecular Haematology), Module 11: IDH Mutation Analysis in AML (ID1, IDH2) – Pilot
- Molecular Diagnostics (Molecular Haematology), Module 12: 17p deletion in CLL – Pilot

UHKT[20] Analysis of BCR-ABL Fusion Gene

UKNEQAS[21]: UKNEQAS Leucocyte Immunophenotyping

- BCR-ABL1 and AML translocation Identification: including t(9;22), t(8;21) and t(15;17)
- BCR-ABL Kinase Domain Mutation Status (Pilot): BCR-ABL1 Kinase Mutation analysis for identification of mutation that result in resistance to Tyrosine kinase (TKI) therapy
- BCR-ABL1 Minor Quantification (Pilot): Quantification of minor BCR-ABL1 transcript levels for the monitoring of Ph-positive Acute Lymphocytic Leukaemia (ALL) and a small subset of Chronic Myeloid Leukaemia (CML) patients
- BCR-ABL1 Major Quantification: BCR-ABL Quantitation for the monitoring of BCR-ABL1 positive leukaemia patients receiving Tyrosine Kinase Inhibitor (TKI) therapy
- FLT3 Mutation Status: FLT-3 is member of the platelet derived growth factor receptor subfamily, with 2 types of mutations having prognostic significance in AML; Internal Tandem Duplications (ITD), D835V
- JAK2 V617F Mutation Status: JAK2 V617F Mutation identification for the diagnosis of myeloproliferative neoplasms
- KIT D816V Mutation Status: KIT D816V mutation analysis for the diagnosis and prognostic work up of systemic mastocytosis and other haematological malignancies
- Leukemia Immunophenotyping & Diagnostic Interpretation (Part 1&2)
- Minimal Residual Disease in Acute Lymphatic Leukaemia (Pilot): Stabilised leukaemias issued to assess the level of minimal residual disease
- NPM1 Mutation Status: Nucleophosmin is a chaperone protein. In approximately 35% of adult AML cases mutations in exon 12 of NPM1 are detected. Mutation A, a tandem duplication of TCTG is the most prevalent
- Paediatric Acute Leukaemia Translocation: t(12;21) (ETV6-RUNX1, TEL-AML1), t(4;11) (MLL-AFF1, MLL-AF4) and t(1;19) (E2A-PBX1, TCR3-PBX1) analysis by molecular

techniques in the diagnosis and prognostic work up of Acute Leukaemia

Lung Cancer, Nonsmall Cell Lung Cancer (NSCLC) / Bronchialkarzinom
(OMIM #211980)

- Anaplastic Lymphoma Kinase (ALK) (OMIM *105590)
- V-Raf Murine Sarcoma Viral Oncogene Homolog B1 (BRAF) (OMIM *164757)
- Epidermal Growth Factor Receptor (EGFR) (OMIM *131550)
- V-Ki-Ras2 Kirsten Rat Sarcoma Viral Oncogene Homolog (KRAS) (OMIM *190070)
- Phosphatidylinositol 3-Kinase, Catalytic, Alpha (PIK3CA) (OMIM *171834)

CAP[3]: CAP Headquarters, Northfield
- "Solid Tumors-Other", EGFR
- "Solid Tumors-Other", BRAF
- Multigene Tumor Panel (MTP): BRAF, EGFR, HER2 (ERBB2), KIT, KRAS, NRAS, PDGFRA, PIK3CA
  Designed for laboratories performing both non-NGS multiplex testing as well as non-multiplexed platforms (eg, Sanger sequencing)

ECAT[6]: MG2, Set B (BRAF V600E, BRAF V600K, K-Ras Codon 12, 13, 61 (in Kooperation mit DGKL)

EMQN[8]:
- "Molecular Testing in Lung Cancer, NSCLC" (NSCLC): mutations in the EGFR, PIK3CA, KRAS and BRAF genes; genotyping & biological/clinical interpretation; paraffin embedded material
- "Oncogene Panel Testing" (ONCOPANEL): mutations in the EGFR, PIK3CA, KRAS, HRAS, NRAS, KIT, TP53 and BRAF genes;
  This scheme is being offered to help labs using high thorough put technologies (e.g., NGS, MassArray etc) accurately validate assay sensitivity and specificity; for specific tumour types, please register for the relevant Lung, Melanoma or Colorectal scheme; Rolled sections of formalin fixed paraffin embedded (FFPE) materials designed to simulate a real patient sample

ESP-EQA[10]
- ALK testing (FISH, IHC); resections, digital cases
- ROS1 testing (FISH, IHC); resections, cell line, digital cases
- Molecular NSCLC scheme: EGFR (mandatory), KRAS (optional) and BRAF (optional) variant analysis; resections

QuIP[16]: PD-L1 NSCLC
10 Fälle von Normal- und Tumorgewebe, 2 Objektträger pro Fall (kein HE-Schnitt)

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Genetics, Oncology Program;
- ALK translocation; Oncology Program, sample: FFPE Tissue
- Mutation Detection in Lung Cancer (NSCLC)
  sample: FFPE Tissue (patient-derived tissue & synthetic samples)

Molecular Genetics, Somatic Mutation Testing;

- Molecular testing in Lung Cancer (Provider: EMQN)
  sample: real tissue and artificial paraffin embedded materials
  Molecular Genetics, Generic Report;
- Mutation Detection in Lung Cancer (NSCLC) (EGFR)

RfB (DGKL)[19]:
- Molekulargenetik Gruppe 2 (MG2), Set B (BRAF V600E, BRAF V600K)
- Molekulargenetik Gruppe 2 (MG2), Set B (K-Ras Codon 12, 13, 61)
- circulating tumor DNA (ctDNA), Set A: KRAS C12, C13 (rs121913530, rs121913529, rs121913535, rs112445441, rs121913238, rs121913240, rs17851045)
- circulating tumor DNA (ctDNA), Set B: BRAF V600E (rs113488022)

UKNEQAS[21]: UKNEQAS Immunocytochemistry & In-Situ Hybridization
- NSCLC ALK IHC (Module 10)
- NSCLC PD-L1 IHC (Pilot) (Module 11)

GenQA[12]:
- Molecular analysis in lung cancer (2 distributions):
  EGFR, *ALK*, *KRAS* and *BRAF* genes
  Any molecular techniques including FISH and IHC.
- Additional lung biomarkers
  ROS1, *RET*, *MET* (amplification genes)
  Any molecular techniques including FISH and IHC.
- Testing of circulating free DNA in plasma for lung cancer
  EGFR gene

Magenkarzinom
- HER2 / ERBB2 Onkogen
(OMIM *164870)

CAP[3]: CAP Headquarters, Northfield
- HER2 Immunohistochemistry (HER2)
- Gastric HER2 (GHER2): immunohistochemistry on gastroesophageal adenocarcinomas
- In Situ Hybridisation:
  HER2 (ERBB2) gene amplification (brightfield) (ISH2)
- Multigene Tumor Panel (MTP): BRAF, EGFR, HER2 (ERBB2), KIT, KRAS, NRAS, PDGFRA, PIK3CA
  Designed for laboratories performing both non-NGS multiplex testing as well as non-multiplexed platforms (eg, Sanger sequencing)

QuIP[16]:
- Her2 ISH
  2 TMA-Schnitte mit jeweils ca. 10 Stanzen
- Her2 ICH
  2 TMA-Schnitte mit jeweils ca. 20 Stanzen

Maligne Lymphome:

B-Zell-Neoplasien – Gensegmente der schweren Ketten der Immunglobulingene (IgH): Immunglobulin Heavy Chain – Rearrangement und Mutationen
(OMIM *147100)

ESP-EQA[10]	IG/TR clonality testing in suspected lymphoproliferations/Lymphoma (Euroclonality Network): Immunglobuline (IG) gene rearrangements multiple rearrangements patterns and cross-lineage rearrangements; peripheral blood or FFPE tissue
QuIP[16]:	Klonalität bei malignen Lymphomen (KlonML): 10 Fälle - PCR zum Nachweis von umgelagerten Immunglobulin-Genen (IgH und ggf. IgL) und umgelagerten T-Zellrezeptorgenen (TCRG und ggf. TCRB) - Lymphommarker Low grade-NHL-Panel: CD5, CyclinD1, CD10, BCL2 (4 Marker) Hodgkin-Panel: CD30, PAX5 (2 Marker) Hans-Klassifikatoren: MUM1, BCL6, CD10 (3 Marker)
RCPA QAP[18]:	RCPAQAP Enrolment Office, St.Leonards Molecular Diagnostics (Molecular Haematology), Module 4
CAP[3]:	CAP Headquarters, Northfield Sequence analysis of the clonal immunoglobulin heavy chain V gene (IGHV) to determine somatic hypermutation (SHM) status

Mammakarzinom
- HER2 / ERBB2 Onkogen
(OMIM *164870)

CAP[3]:	CAP Headquarters, Northfield - HER2 Immunochemistry (HER2) - In Situ Hybridisation: HER2 (ERBB2) gene amplification (brightfield) (ISH2) - CAP/ACMG Fluorescence In Situ Hybridization for Paraffin-Embedded Tissue (CYH): Breast Cancer: HER2 gene amplification - Multigene Tumor Panel (MTP): BRAF, EGFR, HER2 (ERBB2), KIT, KRAS, NRAS, PDGFRA, PIK3CA Designed for laboratories performing both non-NGS multiplex testing as well as non-multiplexed platforms (eg, Sanger sequencing)
QuIP[16]:	Mamma-Ringversuche 2018 - Ki67 (Mamma): 2 TMA-Schnitte mit jeweils ca. 24 Stanzen - Progesteronrezeptor (PR-Mamma): 2 TMA-Schnitte mit jeweils ca. 20 Stanzen - Östrogenrezeptor (ER-Mamma): 2 TMA-Schnitte mit jeweils ca. 20 Stanzen

- HER2-ICH (Mamma): 2 TMA-Schnitte mit jeweils ca. 20 Stanzen
- HER2-ISH (Mamma): 2 TMA-Schnitte imt jeweils ca. 20 Stanzen

UKNEQAS[21]: UKNEQAS Immunocytochemistry & In-Situ Hybridization
HER2 ISH (interpretative and technical)

Mammakarzinom, neuroendokrine Tumoren - Ki-67
(OMIM *176741)

QuIP[16]: Mamma-Ringversuch
- Ki67 (Mamma): 2 TMA-Schnitte mit jeweils ca. 24 Stanzen

Mastocytose / Urticaria pigmentosa – KIT Onkogen: D816V
(OMIM 154800 – OMIM *164920)

ECAT[6]: MG2, Set B: cKIT D816V (in Kooperation mit DGKL)

EMQN[8]: "Oncogene Panel Testing" (ONCOPANEL): mutations in the EGFR, PIK3CA, KRAS, HRAS, NRAS, KIT, TP53 and BRAF genes;
This scheme is being offered to help labs using high thorough put technologies (e.g., NGS, MassArray etc) accurately validate assay sensitivity and specificity; for specific tumour types, please register for the relevant Lung, Melanoma or Colorectal scheme;
Rolled sections of formalin fixed paraffin embedded (FFPE) materials designed to simulate a real patient sample

INSTAND[13]: Molekulargenetik IV, Nr.767; KIT p.D816V

ÖQUASTA[15]: Analyte Gruppe 2, Set B: cKIT D816V (in Kooperation mit DGKL)

RfB (DGKL)[19]: - Molekulargenetik Gruppe 2 (MG2), Set B: cKit D816V

UKNEQAS[21]: UKNEQAS Leucocyte Immunophenotyping
- KIT p.Asp816Val (D816V) Mutation Status for Mast Cell Disease

Melanoma – Molecular Testing:

CAP[3]: CAP Headquarters, Northfield
- "Solid Tumors-Other", BRAF
- Multigene Tumor Panel (MTP): BRAF, EGFR, HER2 (ERBB2), KIT, KRAS, NRAS, PDGFRA, PIK3CA
Designed for laboratories performing both non-NGS multiplex testing as well as non-multiplexed platforms (eg, Sanger sequencing)

EMQN[8]: - "Molecular testing for melanoma (MELANOMA): mutations in the BRAF, NRAS, KIT genes;
genotyping & biological/clinical interpretation; paraffin embedded material
- "Oncogene Panel Testing" (ONCOPANEL): mutations in the EGFR, PIK3CA, KRAS, HRAS, NRAS, KIT, TP53 and BRAF genes;
This scheme is being offered to help labs using high thorough put technologies (e.g., NGS, MassArray etc) accurately validate assay sensitivity and specificity; for specific tumour types, please register for the relevant Lung, Melanoma or Colorectal scheme;

Rolled sections of formalin fixed paraffin embedded (FFPE) materials designed to simulate a real patient sample

QuIP[16]: BRAF-Mutationsanalyse:
10 Fälle maligner Tumore auf jeweils 3 Objektträgern

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Genetics, Oncology Program;
- Mutation Detection in Melanoma
sample: FFPE Tissue (patient-derived tissue & synthetic samples)
Molecular Genetics, Somatic Mutation Testing;
- Molecular testing in Melanoma (Provider: EMQN)
sample: real tissue and artificial paraffin embedded materials
Molecular Genetics, Generic Report;
- Mutation Detection in Melanoma (BRAF, NRAS)

GenQA[12]: Molecular analysis in melanoma (2 distributions):
BRAF, *KIT* and *NRAS* genes
Any molecular techniques including FISH and IHC.

Minimal Residual Disease

CAP[3]: CAP Headquarters, Northfield
- BCR/ABL1 p190 (MRD1)
- BCR/ABL1 p210 (MRD)
- PML/RARA (MRD2)

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Diagnostics (Molecular Haematology), Module 8
PML/RARα

Molecular Hematologic Oncology / Molecular Diagnosis of Haematological Malignancies

CAP[3]: CAP Headquarters, Northfield
- Lymphoid malignancy genotyping (MHO, MHO1): IGH, IGH/BCL2 major, IGH/BCL2 minor, IGH/CCND1, IGK, TRB, TRG
- Myeloid malignancy genotyping (MHO2, MHO3): BCR/ABL1 p190, BCR/ABL1 p210, CBFB/MYH11, FLT3 ITD, FLT3 TKD, JAK2 c.1849G>T (p.V617F), NPM1, PML/RARA, RUNX1/RUNX1T1
- DNA extraction and amplification from formalin-fixed, paraffin-embedded (FFPE) tissue (MHO5)

Molecular testing for EGFR gene mutations in ctDNA - pilot

EMQN[8]: Scheme LIQUIDBIOPSY(EGFR): Mutations in the EGFR gene; genotyping & interpretation; pilot

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Genetics, Somatic Mutation testing
Molecular testing for EGFR gene mutations in ctDNA (Provider: EMQN) – Pilot

RfB (DGKL)[19]: circulating tumor DNA (ctDNA), Set C: EGFR T790M (rs121434569))

QuIP: - EGFR-Mutationsanalyse
10 Fälle (je 4 Objetträger für die Mutationen in den Exonen 18, 19, 21)
- T790M (Split 1 & 2)
Blut: 10 Blutproben mit unterschiedlichen Zelllinien (T790M Mutation in EGFR Exon 20 und Primärmutationen in EGFR Exon 19/21 – Allelfrequenz nach EBM-Vorgabe 1%)
Gewebe: 10 Fälle von Normal- und Tumorgewebe, 3 Objektträger pro Fall – T790M Mutation in EGFR Exon 20
- T790M (Split 3&4)
Blut: 10 Blutproben mit unterschiedlichen Zelllinien (T790M Mutation in EGFR Exon 20 und Primärmutationen in EGFR Exon 19/21 – Allelfrequenz nach EBM-Vorgabe 1%)
Gewebe: 10 Fälle von Normal- und Tumorgewebe, 3 Objektträger pro Fall – T790M Mutation in EGFR Exon 20

Molecular testing for RAS gene mutations in ctDNA – pilot

EMQN[8]: Scheme LIQUIDBIOPSY (RAS): Mutations in the KRAS, NRAS, HRAS genes; genotyping & interpretation; pilot

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Genetics, Somatic Mutation testing
Molecular testing for RAS gene mutations in ctDNA (Provider: EMQN) – Pilot

RfB (DGKL)[19]: circulating tumor DNA (ctDNA), Set A: KRAS C12, C13 (rs121913530, rs121913529, rs121913535, rs112445441, rs121913238, rs121913240, rs17851045)

Myeloproliferative Neoplasien – Calreticulin (CALR)-Mutationen

(OMIM *109091)

INSTAND[13]: Molekulargenetik IV, Nr.768; CALR Exon 9 Mutation(en), CALR Spezifikation

Neoplastic Cellularity

CAP[3]: CAP Headquarters, Northfield
- Neoplastic Cellularity (NEO)
online assessment of percent neoplastic cellularity using whole slide images

Neuroblastoma pilot (array and FISH)

GenQA[12]: 1, 11 and 17 abnormalities plus, MYCN amplification
Techniques: FISH, array

Oncogene panel testing

CAP[3]: CAP Headquarters, Northfield
- Multigene Tumor Panel (MTP): BRAF, EGFR, HER2 (ERBB2), KIT,

KRAS, NRAS, PDGFRA, PIK3CA
Designed for laboratories performing both non-NGS multiplex testing as well as non-multiplexed platforms (eg, Sanger sequencing)

EMQN[8]: "Oncogene Panel Testing" (ONCOPANEL): mutations in the EGFR, PIK3CA, KRAS, HRAS, NRAS, KIT, TP53 and BRAF genes;
This scheme is being offered to help labs using high thorough put technologies (e.g., NGS, MassArray etc) accurately validate assay sensitivity and specificity; for specific tumour types, please register for the relevant Lung, Melanoma or Colorectal scheme;
Rolled sections of formalin fixed paraffin embedded (FFPE) materials designed to simulate a real patient sample

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Genetics, Somatic Mutation Testing;
- Oncogen panel testing (Provider: EMQN)
sample: rolled section of paraffin embedded materials

RNA Sequencing to detect gene fusion transcripts

CAP[3]: CAP Headquarters, Northfield
RNA: Total RNA from a cell line engineered to contain desired fusion RNA; for laboratories using RNAseq to detect gene fusion transcripts; not intended to replace the current Survey (SARC) for reverse transcription (RT)-PCR based detection; potential fusion variants include: CD74-ROS1, EML4-ALK, ETV6-NTRK3, FGFR3-TACC3, PAX8-PPARG, SLC45A3-BRAF

Sarcoma Translocation (SARC)

CAP[3]: CAP Headquarters, Northfield
RT-PCR
Sarcoma Translocation Listing:
COL1A1/PDGFB, t(17;22); ETV6-NTRK3, t(12;15); EWSR1/ATF1, t(12;22); EWSR1/ERG, t(21;22); EWSR1/FLI1, t(11;22); EWSR1/FLI1 oder EWSR1/ERG; EWSR1/WT1, t(11;22); FUS/DDIT3, t(12;16); PAX3/FOXO1, t(2;13); PAX7/FOXO1, t(1;13); PAX3/FOXO1 oder PAX7/FOXO1; SS18/SSX1, t(X;18); SS18/SSX2, t(X;18); SS18/SSX1 oder SS18/SSX2

Sarcoma testing by RT-PCR and FISH

GenQA[12]: Sarcoma - Three formalin-fixed paraffin-embedded (FFPE) tissue samples are distributed with clinical case scenarios.
Techniques: RT-PCR, FISH and NGS

Thyroid Cancer

CAP[3]:	CAP Headquarters, Northfield "Solid Tumors-Other", BRAF (1799T>A bzw. Val600Glu)
ECAT[6]:	MG2, Set B: BRAF V600E, BRAF V600K (in Kooperation mit DGKL)
ÖQUASTA[15]:	Analyte Gruppe 2, Set B: BRAF V600E, BRAF V600K (in Kooperation mit DGKL)
RfB (DGKL)[19]:	- Molekulargenetik Gruppe 2 (MG2), Set B (BRAF V600E, BRAF V600K) - circulating tumor DNA (ctDNA), Set B: BRAF V600E (rs113488022)
UKNEQAS[21]:	UKNEQAS Leucocyte and Immunophenotyping BRAF p.Val600Glu (V600E) Mutation Status for Hairy Cell Leukaemia

Tumor Protein p53

(OMIM *191170)

EMQN[8]:	"Oncogene Panel Testing" (ONCOPANEL): mutations in the EGFR, PIK3CA, KRAS, HRAS, NRAS, KIT, TP53 and BRAF genes; This scheme is being offered to help labs using high thorough put technologies (e.g., NGS, MassArray etc) accurately validate assay sensitivity and specificity; for specific tumour types, please register for the relevant Lung, Melanoma or Colorectal scheme; Rolled sections of formalin fixed paraffin embedded (FFPE) materials designed to simulate a real patient sample
INSTAND[13]:	Molekulargenetik IV, Nr.765; TP53 Mutation(en), TP53 Nomenklatur

T-Zell-Lymphome – T-Zell-Rezeptor Gamma (ZCRG) Gen Segmente, T-Zell-Rezeptor Beta (TCRB) Gen Segmente (T-Zell Rezeptor Gen – Rearrangements)

(OMIM *186960)

ESP-EQA[10]	IG/TR clonality testing in suspected lymphoproliferations/Lymphoma (Euroclonality Network): T-cell receptor (TR) gene rearrangements multiple rearrangements patterns and cross-lineage rearrangements; peripheral blood or FFPE tissue
QuIP[16]:	- Klonalität bei malignen Lymphomen (KlonML): 10 Fälle - PCR zum Nachweis von umgelagerten Immunglobulin-Genen (IgH und ggf. IgL) und umgelagerten T-Zellrezeptorgenen (TCRG und ggf. TCRB) - Lymphommarker Low grade-NHL-Panel: CD5, CyclinD1, CD10, BCL2 (4 Marker) Hodgkin-Panel: CD30, PAX5 (2 Marker) Hans-Klassifikatoren: MUM1, BCL6, CD10 (3 Marker)
RCPA QAP[18]:	RCPAQAP Enrolment Office, St.Leonards Molecular Diagnostics (Molecular Haematology), Module 4

UKNEQAS[21]: UKNEQAS Leucocyte Immunophenotyping
- IgH/TCR Clonality Status:
IgH and TCR Clonality testing for the diagnosis of lymphoid malignancy

Methodisch-technische Ringversuche Molekularbiologie

DNA Isolierung

CAP[3]:	CAP Headquarters, Northfield DNA Purification from FFPET (MHO5): DNA purification and amplification of control targets
ECAT[6]:	DNA-Isolierung und FV Genotypisierung (in Kooperation mit DGKL)
ÖQUASTA[15]:	DNA-Isolierung und FV-Genotypisierung (in Kooperation mit DGKL)
RCPA QAP[18]:	RCPAQAP Enrolment Office, St.Leonards Molecular Genetics, Technical Program: DNA Extraction (Pilot)
RfB (DGKL)[19]:	DNA-Isolierung (DI) und FV-Genotypisierung: FV Leiden Arg506Gln, FV His1299Arg, FV Cambridge Arg306Thr, FV Hong-Kong Arg306Gly, FII G20210A, MTHFR C677T, HFE His63Asp/Cys282Tyr/Ser65Cys
GenQA[12]:	- DNA extraction from blood samples Assessment of DNA quality and quantity from blood samples - Three whole-peripheral blood samples in EDTA supplied for DNA extraction. - DNA extraction from FFPE tissue samples Assessment of DNA quality and quantity from formalin-fixed paraffin-embedded (FFPE) tissue. - DNA extraction from FF tissue samples Assessment of DNA quality and quantity from fresh frozen (FF) tissue. - DNA extraction from saliva samples Assessment of DNA quality and quantity from saliva samples.

DNA quantification

GenQA[12]:	DNA Quantification technical EQA for DNA concentration measurement - Six DNA samples with matching blank samples supplied for DNA quantification.

DNA Sequenzierung (Sanger)

CAP[3]:	CAP Headquarters, Northfield (in Kooperation mit ACMG[1]) "CAP/ACMG Molecular Genetics Sequencing": - DNA sequencing interpretation (SEC) - DNA sequencing (SEC1)
ECAT[6]:	inkl. diagnostischer Interpretation (in Kooperation mit DGKL)
EMQN[8]	- Full version: Scheme SEQ-FULL: DNA-sequencing assessment of genotyping, diagnostic interpretation, mutation nomenclature and quality of raw data; suitable for Sanger sequencing technologies only - Data quality assessment only version:

Scheme SEQ-QUAL: DNA-sequencing (gene independent); data quality assessment only; suitable for Sanger sequencing technologies only

EQUALIS[9]: Product code 246: sequencing human genomic DNA

ÖQUASTA[15]: DNA-Sequenzierung (in Kooperation mit DGKL)

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Genetics, Technical; Provider: EMQN; sample: wet DNA in TE
- Data quality assessment only version
- Full version
Molecular Genetics, RCPAQAP Technical Program
- Sanger DNA Sequencing; sample: PCR products

RfB (DGKL)[19]: DNA-Sequenzierung (SQ) und diagnostische Interpretation

Immunohistochemistry

CAP[3]: CAP Headquarters, Northfield
- Immunohistochemistry (MK)

Maternal Cell Contamination (and sexing)

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Genetics, RCPAQAP Technical Program; sample: purified genomic DNA

GenQA[12]: Maternal cell contamination and fetal sexing EQA - DNA samples are distributed for three clinical case scenarios. Fetal sexing and identity testing (contamination). Testing can be performed using any methodology.

Next Generation Sequencing

BVDH (HGQN)[2]: Next Generation Sequencing
NGS Analysepipeline, NGS-Rohdaten online abrufbar

CAP[3]: CAP Headquarters, Northfield
- Next Generation Sequencing (NGS)
Testing of up to 200 variants. For the full list of genes in this program go to www.cap.org, Laboratory Improvement Programs.
- Next-Generation Sequencing - Solid Tumors (NGSST)
Methods-based proficiency challenge for laboratories performing targeted next generation sequencing of cancer genes or mutation hotspots in solid tumors. Laboratories will be asked to identify somatic single nucleotide variants and small insertions or deletions in the following genes: AKT1, ALK, APC, ATM, BRAF, CDH1, CTNNB1, EGFR, ERBB2, FBXW7, FGFR2, GNAQ, GNAS, HRAS, IDH1, KIT, KRAS, MET, NRAS, PDGFRA, PIK3CA, PTEN, SMAD4, SMARCB1, SMO, SRC, STK11, TP53
- Next-Generation Sequencing - Hematologic Malignancies (NGSHM)

Methods-based proficiency challenge for laboratories performing targeted next-generation sequencing of genes or mutation hotspots in hematologic malignancies.
Laboratories will be asked to identify somatic single nucleotide variants and small insertions or deletions in the following genes: ASXL1, ATM, BRAF, CALR, CEBPA, CREBBP, CSF3R, DNMT3A, EZH2, FLT3, IDH1, IDH2, JAK2, KIT, KMT2D, MPL, MYD88, NOTCH1, NPM1, SF3B1, SRSF2, TET2, TP53, U2AF1

- Next-Generation Sequencing Bioinformatics (NGSB1, NGSB2) in silico program augments wet bench NGS proficency testing programs by testing a greater number of variants, at a greater range of variant frequencies: Illumina TruSeq Amplicon Cancer Panel (NGSB1) and Ion Torrent AmpliSeq Cancer Hotspot v2 (NGSB2)
- Next-Generation Sequencing Undiagnosed Disorders – Exome (NGSE): Exome analysis for germline undiagnosed disorders in silico based survey to identify germline variants responsible for a provided clinic phenotype as is encountered in an undiagnosed disease scenario
- Next-Generation Sequencing Bioinformatics Somatic Validated Materials (NGSBV): Somatic in silico mutagenized sequencing file in silico program is designed to optimize bioinformatics pipelines, augment validations, and assist with pipeline verification after changes to NGS/bioinformatics processes. This is not traditional proficiency testing and no results will be returned to the CAP; information regarding the variants introduced will be sent along with the mutagenized file.

EMQN[8]:

- DNA Sequencing NGS (vSomatic) (NEXTGEN (S)): laboratory can choose target; Pilot scheme; Assessment of genotyping and quality of raw data; g.DNA sample derived from FFPE material; for labs doing NGS based SOMATIC testing ONLY
- DNA Sequencing NGS (vGermline) (NEXTGEN (G)): laboratory can choose target; Pilot scheme; Assessment of genotyping and quality of raw data; g.DNA sample derived from FFPE material; for labs doing NGS based GERMLINE testing ONLY
- “Oncogene Panel Testing” (ONCOPANEL): mutations in the EGFR, PIK3CA, KRAS, HRAS, NRAS, KIT, TP53 and BRAF genes;
This scheme is being offered to help labs using high thorough put technologies (e.g., NGS, MassArray etc) accurately validate assay sensitivity and specificity; for specific tumour types, please register for the relevant Lung, Melanoma or Colorectal scheme;
Rolled sections of formalin fixed paraffin embedded (FFPE) materials designed to simulate a real patient sample

RCPA QAP[18]:

RCPAQAP Enrolment Office, St.Leonards
Molecular Genetics, Technical; Provider: EMQN:

- DNA Sequencing – NextGen (vGermline)
  sample: genomic DNA from cell lines
- DNA Sequencing – NextGen (vSomatic)
  sample: rolled section of paraffin embedded material

Molecular Genetics, RCPAQAP Technical Program
- Massively Parallel Sequencing (NGS) – Pilot
  sample: purified genomic DNA

GenQA[12]: - Next Generation Sequencing for inherited disorders (pilot)
Single genes, gene panels, Whole exome sequencing and whole genome sequencing. Variant calling & data assessment.
- Next Generation Sequencing for somatic testing (pilot)
Single genes, gene panels, Whole exome sequencing and whole genome sequencing. DNA and formalin-fixed paraffin embedded tissue. Variant calling & data assessment.

Pathogenicity of sequence variants, Germline Variant Interpretation

CAP[3]: CAP Headquarters, Northfield
Variant interpretation online case review (VIP/VIP1)

GenQA[12]: Pathogenicity of sequence variants
Classification of pathogenicity of sequence variants EQA (interpretation only) - Three clinical cases and variants detected supplied for interpretation, classification and reporting.
No testing required. Interpretation only of the results supplied.

II. Ringversuche Zytogenetik

Konstitutionelle Zytogenetik

Array-Diagnostik / Comparative Genomic Hybridisation (CGH)
BVDH (HGQN)[2]: München

Chromosome breakage Syndromes - Pilot
GenQA[12]: Chromosome breakage and mutations for Fanconi, anaemia, Blooms, Ataxia, Telangiectasia, Nijmegan syndrome.
Chromosome Breakage Syndrome pilot - One clinical postnatal case scenario with solid stained metaphase images.
Techniques: Metaphase analysis of solid stained chromosomes
Analysis & Interpretation

Constitutional Cytogenetics – Postnatal: Blood
GenQA[12]: WGAmicro
Blood - Two clinical postnatal case scenarios with G-banded and FISH images. Two cases - online analysis for the detection of aneuploidy and rearrangements. G-banded metaphase images plus other tests as applicable to analyse the case.
Techniques: G-banding, FISH

Constitutional Cytogenetics – Prenatal: Amniotic Fluid
GenQA[12]: WGAmicro
Amniotic Fluid - Two clinical prenatal case scenarios with G-banded (and FISH) images.
Technique: G-banding
Karyotyping & Interpretation

Constitutional Cytogenetics – Prenatal: Chorionic Villi Sampling (CVS)
GenQA[12]: WGAmicro
Chorionic Villus - Two clinical prenatal case scenarios with G-banded (and FISH) images.
Techniques: G-banding
Karyotyping & Interpretation

Constitutional Cytogenetics – Prenatal: Products of Conception (G-banded only)
GenQA[12]: WGAmicro
Products of conception/Fetal tissue (G-banded) -Two clinical case scenarios with G-banded images.
Techniques: G-banding karyotype and FISH
Karyotyping & Interpretation

Constitutional Cytogenetics – Prenatal: Products of Conception (Array/MLPA/PCR/NGS)

GenQA[12]: WGAmicro & CNV
Products of conception/Fetal tissue (Molecular methods) -Two DNA samples are distributed with clinical case scenarios.
Techniques: Array/Shallow NGS/QF-PCR/MLPA
Analysis & Interpretation

IPS stem cells exploratory pilot

GenQA[12]: WGAmicro and CNV
IPS stem cells exploratory pilot - one online case for metaphase analysis.
Analysis and Interpretation

Molecular testing for cystic fibrosis on blood spots

GenQA[12]: *CFTR* gene
Blood spot cards (4 distributions per year)
Genotyping only

Molecular testing for MCADD c.985A>G on blood spots

GenQA[12]: ACADM c.985A>G p. (Lys329Glu) variant
Blood spot cards (4 distributions per year)
Genotyping only

Postnatal constitutional CNV detection

GenQA[12]: CNVs
Postnatal constitutional CNV detection (array) - DNA samples are distributed for two clinical postnatal case scenarios.
Technique: Array/NGS
Analysis & Interpretation

Prenatal constitutional CNV detection

GenQA[12]: CNV
Prenatal constitutional CNV detection (previously Prenatal microarray) - Two DNA samples are distributed for two clinical prenatal case scenarios plus a CNV survey.
Techniques: Array/NGS
Analysis & Interpretation

Severe developmental delay case scenario pilot

GenQA[12]: WGAmicro & CNV interpretation
Multiple assays to detect genomic causes of severe developmental delay.
Interpretation only

Constitutional Cytogenetics – Postnatal: Microarray (aCGH) - postnatal
(distribution of DNA - technical, analytical and interpretive proficiency assessed)

EMQN[8]: "Constitutional Microarray analysis (Microarrays/arrayCGH)" (aCGH): genomic deletions and duplications; genotyping and interpretation; platform independent – participants use their normal methodology

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Genetics, Technical; Provider: EMQN:
Constitutional Microarray Analysis
sample: genomic DNA

RfB (DGKL)[19]: in Kooperation mit EMQN

Cytogenetics

CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
- Chromosome abnormality (CY, CYBK)
- Karyotype (CY, CYBK)
- Educational challenge, ungraded (CY, CYBK)

CY: online images of metaphase cells
CYBK: prints of metaphase cells
Each challenge includes a case history and images of metaphase cells that are representative of each case; each mailing includes three constitutional and three neoplastic challenges.

Fluorescence In Situ Hybridisation (FISH) – constitutional abnormality

CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
Constitutional and Hematologic Disorders:
- FISH for constitutional abnormality (CYF):

CYF 2018-A:
- Constitutional disorder - Williams syndrome critical region (two slides)
- Constitutional disorder - (two paper/photograph challenges)
- Hematologic disorder - BCR/ABL1 (two slides)
- Hematologic disorder - (two paper/photograph challenges)

CYF 2018-B:
- Constitutional disorder - SRY (two slides)
- Constitutional disorder - (two paper/photograph challenges)
- Hematologic disorder - CLL panel (four slides)
- Hematologic disorder - (two paper/photograph challenges)

CYF is prepared from cell suspension samples.
Four slides and four paper challenges

Laborientierte QS – Postnatale Zytogenetik

BVDH (HGQN)[2]: Berlin

Laborientierte QS – Pränatale Zytogenetik (Chorionzottenanalyse Langzeitkultur)
BVDH (HGQN)[2]: Berlin

Laborientierte QS – Pränatale Zytogenetik (Fruchtwasseranalyse)
BVDH (HGQN)[2]: Berlin

Microarray-Analyse
CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
- Cytogenomic Microarray-Analysis (CYCGH): cytogenomic microarray-analysis for constitutional abnormality (two DNA specimens) and educational paper challenge for constitutional or neoplastic abnormality - Identification of gains or losses and the cytogenetic location of any abnormalities
- Oncology Microarray-Analysis (CYCMY): cytogenomic microarray analysis for oncologic abnormality (one DNA specimen) and educational paper challenge for oncologic abnormality - Identification of gains or losses and the cytogenetic location of any abnormalities

Molecular Rapid Aneuploidy (MRA): QF-PCR, MLPA; BoBs
GenQA[12]: Targeted 13/18/21/X/Y
Three prenatal DNA samples for rapid aneuploidy testing for the chromosomes 13, 18, 21 plus X and Y using either QF-PCR or MLPA or BoBs.
Analysis and Interpretation

Non Invasive Prenatal Testing (NIPT) - Pilot
CAP[3]: CAP Headquarters, Northfield
Noninvasive Prenatal Testing (NIPT) - Cell-free DNA screening for fetal aneuploidy

Non-invasive prenatal testing for common aneuploidies - Pilot
EMQN[8]: Non-invasive prenatal testing (NIPT) for common aneuploidies (NIPT(ANEUPLOIDY)): NIPT for the 3 most common aneuploidies (Chr 13, 18 and 21); pilot (in collaboration with UKNEQAS[21] and CEQAS[4])
GenQA[12]: Non-invasive prenatal testing for aneuploidies - Plasma samples are distributed for 2 or 3 clinical case scenarios.
Chromosomal aneuploidies for 13, 18, 21 and sex chromosomes.
Technique: NGS, Analysis & Interpretation

Non-invasive prenatal testing for sex determination - Pilot
EMQN[8]: Non-invasive prenatal testing (NIPT) for fetal sexing (NIPT(SEXING)): NIPT for the fetal sexing; pilot (in collaboration with UKNEQAS[21])

RCPA QAP[18]: RCPAQAP Enrolment Office, St.Leonards
Molecular Genetics, Technical;
Non-invasive prenatal diagnosis using cell free fetal DNA for sex determination – Pilot (cffDNA-15)
Provider: EMQN; sample: plasma samples

GenQA[12]: Non-invasive prenatal testing for sex determination pilot EQA - Plasma samples are distributed for 2 or 3 clinical case scenarios. Testing can be performed using any methodology.
Genotyping for sex determination. Feedback will be provided for the interpretation of the result and clerical accuracy.

Non-invasive prenatal testing for common microdeletions pilot

GenQA[12]: Non-invasive prenatal diagnosis for common microdeletions pilot - Plasma samples are distributed for 2 or 3 clinical case scenarios. Testing can be performed using any methodology.
Technical, Analysis and Interpretation

Rapid Prenatal Aneuploidy – FISH

GenQA[12]: Rapid Prenatal Aneuploidy FISH - Two fixed cell suspensions from prenatal samples for the detection of aneuploidy (13,18,21,X,Y).
Technique: FISH
Analysis & Interpretation

Molekulare Zytogenetik

BVDH (HGQN)[2]: Jena

Pränataler Schnelltest

BVDH (HGQN)[2]: Schnelltest FISH/PCR, Probenmaterial: Kultivierte, fixierte Fruchtwasserzellen / DNA

Strukturanalyse

BVDH (HGQN)[2]: Marburg

Hämatoonkologie /Tumorzytogenetik

(Einrichtungen, die nur derartige Analysen durchführen benötigen keine Zulassung gem. § 68 GTG)

Cytogenetics

CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
- Chromosome abnormality (CY, CYBK)
- Karyotype (CY, CYBK)
- Educational challenge, ungraded (CY, CYBK)

CY: online images of metaphase cells
CYBK: prints of metaphase cells
Each challenge includes a case history and images of metaphase cells that are representative of each case; each mailing includes three constitutional and three neoplastic challenges.

FISH – Hematologic/Neoplastic Disorder

CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
Constitutional and Hematologic Disorders:
- FISH for constitutional abnormality (CYF):

CYF 2018-A:
Constitutional disorder - Williams syndrome critical region (two slides)
Constitutional disorder - (two paper/photograph challenges)
Hematologic disorder - BCR/ABL1 (two slides)
Hematologic disorder - (two paper/photograph challenges)

CYF 2018-B:
Constitutional disorder - SRY (two slides)
Constitutional disorder - (two paper/photograph challenges)
Hematologic disorder - CLL panel (four slides)
Hematologic disorder - (two paper/photograph challenges)

CYF is prepared from cell suspension samples.
Four slides and four paper

FISH - Paraffin Embedded Tissue

CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
- Breast Cancer – HER2 gene amplification (CYH)
- Brain/Glioma tissue – 1p19q (CYJ)
- Solid Tumor – FOXO1 and DDIT3 gene rearrangement (CYK)
- Lymphoma tissue – CCND1 (CyclinD1) and BCL2 gene rearrangement (CYL)

FISH – Urothelial Carcinoma

CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
Constitutional and Hematologic Disorders:
Urothelial Carcinoma:

- FISH for urothelial carcinoma (CYI)
  CYI: two cell samples from different specimens; FISH to detect chromosome abnormalities

Haematology Cytogenetics - Acute Lymphoblastic Leukaemia/Lymphoma (ALL)

GenQA[12]: ALL
Acute Lymphoblastic Leukaemia (ALL) - Two clinical ALL case scenarios with G-banded plus FISH images.
Karyotyping & Interpretation

Haematology Cytogenetics - Mature B- and T-cell Neoplasms: G-banded

GenQA[12]: Two lymphoma cases - online analysis for the detection of recurrent and non-recurrent abnormalities associated with this disease type. G-banded analysis plus other tests (FISH) provided as applicable to interpret the results.
Techniques: G-banded, FISH
Karyotyping & interpretation

Haematology Cytogenetics - Mature B- and T-cell Neoplasms: FISH for CLL and Lymphoma

GenQA[12]: Mature B & T cell Neoplasms (FISH for CLL and Lymphoma)
A CLL and a lymphoma case for interphase FISH analysis (full EQA) for the detection of recurrent and non-recurrent abnormalities associated with these disorders.
Technique: Fish & array
Analysis & Interpretation

Haematology Cytogenetics - Myeloid Leukaemia (AML, CML, MDS)

UKNEQAS[21]: UKNEQAS Leucocyte Immunophenotyping
Acute Myeloid Leukaemia Gene Panels (Not Accredited)

GenQA[12]: Two myeloid cases - online analysis for the detection of recurrent and non-recurrent abnormalities associated with myeloid disorders. Interphase FISH and G-banded analysis. Other tests results provided as applicable to interpret the results.
Techniques: G-banding, FISH
Karyotyping & Interpretation

Haematology Cytogenetics - Acquired Array

GenQA[12]: CLL/MDS
DNA sample distribution of two hematological cases for analysis and interpretation (CLL plus myeloid) for the detection of recurrent and non-recurrent abnormalities. Array and NGS can be undertaken for copy number loss and gain, absence of heterogeneity only.

Techniques: Array, NGS
Analysis & Interpretation

Haematology Cytogenetics - Myeloma

GenQA[12]: Two myeloma cases (sample plus online analysis) for the detection of recurrent abnormalities associated this disease. Array/NGS can be undertaken on the sample.
Techniques: FISH, array, NGS
Analysis & Interpretation

Haemotology Cytogenetics – Lymphoma (FFPE)

GenQA[12]: A FFPE lymphoma case for interphase FISH analysis for the detection of recurrent abnormalities.
Techniques: FISH
Analysis & Interpretation

Haematology Cytogenetics - Acute Leukaemia (FISH) pilot

GenQA[12]: CML/ALL/AML
Fixed cell suspensions distributed for analysis. FISH will include breakapart probes, fusion probes and copy number probes for recurrent aberrations seen in AML, ALL and CML.
Analytical only

Interphase FISH

BVDH (HGQN)[2]: Kooperation mit QUITZ[17]

Microarray-Analysis

CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
Cytogenomic Microarray-Analysis (CYCGH):
- cytogenomic microarray-analysis for constitutional abnormality
- educational paper challenge for constitutional or neoplastic abnormality

Identification of gains or losses and the cytogenetic location of any abnormalities.

Oncology Cytogenetics - CNS Tumours (formerly Adult Molecular Neuropathology): Oligodendroglioma 1p/19q FISH,
Oligodendroglioma MGMT promoter methylation, IDH

QuIP[16]: - IDH-1
10 Fälle (je 2 Objektträger)

GenQA[12]: CNS tumours (Central nerval system) – Molecular analysis for 1p/19q co-deletion, *MGMT* promoter methylation and *IDH1* & *IDH2* pathogenic variants, *BRAF* pathogenic variants.
FISH/array/different mutation analysis techniques

Oncology Cytogenetics - Sarcoma FISH/FFPE

GenQA[12]: Sarcoma - Three formalin-fixed paraffin-embedded (FFPE) tissue samples are distributed with clinical case scenarios. RT-PCR and FISH testing for the presence of translocations in sarcomas.

Tumorzytogenetik

BVDH (HGQN)[2]: Düsseldorf

III. Ringversuche Präimplantationsdiagnostik

Embryology - Embryotransfer

CAP[3]: CAP Headquarters, Northfield
Embryology (EMB):
- Embryo transfer and quality assessment (three- and five-day-old-embryos); CD-ROM with video-clips

Preimplantation Genetic Testing by NGS and/or arrays for polar body testing

GenQA[12]: Aneuploidy & Structural rearrangements
Preimplantation Genetic Testing of Polar Bodies by NGS and/or array for aneuploidy and chromosomal rearrangements - DNA distributed for one clinical PGT-A and one PGT-ST case scenario.
Techniques: Array, NGS
Analysis & Interpretation

Preimplantation Genetic Testing for Blastomere FISH (Stage 1 & 2)

GenQA[12]: Aneuploidy & Structural rearrangements
Preimplantation Genetic Testing for Blastomere FISH - This is a two part online EQA comprising of two PGT-ST case scenarios and FISH images.
Techniques: FISH
Analysis & Interpretation

Preimplantation Genetic Diagnosis (PGD-M) of trophectoderm and/or blastomere testing for monogenic disorders

GenQA[12]: Monogenic disorders
Technical, analytical and interpretation

Preimplantation Genetic Testing by Sperm FISH for aneuploidy testing

GenQA[12]: Targeted 13/18/21/X/Y
Preimplantation Genetic Testing of Sperm for aneuploidy by FISH (PGT-A) - fixed sperm suspension distributed for two clinical PGT-A case scenarios.
Techniques: FISH
Analysis & Interpretation

Preimplantation Genetic Testing of trophectoderm and/or blastomere for aneuploidies

GenQA[12]: Preimplantation Genetic Testing of Trophectoderm and/or Blastomere for aneuploidies by NGS and/or arrays - DNA distributed for three clinical PGT-A case scenarios.
Techniques: Array, NGS
Analysis & Interpretation

Preimplantation Genetic Testing of trophectoderm and/or blastomere for chromosomal rearrangements

GenQA[12]: Preimplantation Genetic Testing of Blastomere/Trophectoderm for chromosomal rearrangements by NGS and/or arrays - DNA distributed for three clinical PGT-ST case scenarios.
Techniques: Array, NGS
Analysis & Interpretation

IV. Ringversuche „Biochemical Genetics“

(Einrichtungen, die nur derartige Analysen durchführen benötigen keine Zulassung gem. § 68 GTG)

Biochemical Genetics

CAP[3]: CAP Headquarters, Northfield (in Kooperation mit ACMG[1])
- Acylcarnitines, qualitative and quantitatice (BGL)
- Amino Acids, qualitative and quantitatice (BGL)
- Carnitins, qualitative and quantitatice (BGL1)
- Glycosaminoglycans (mucopolysaccharides), qualitative and quantitatice (BGL)
- Organic Acids, qualitative and quantitatice (BGL)
- Educational challenge (BGL)

Down’s Syndrome Screening, 1st trimester

UKNEQAS[21]: UKNEQAS Clinical Chemistry (Peptide Hormones)
- Downs Screening First Trimester (serum)
- Downs Screening First Trimester using dried blood spots

Down’s Syndrome Screening, 2nd trimester

UKNEQAS[21]: UKNEQAS Clinical Chemistry (Peptide Hormones)
- NTD & Downs Screening Second Trimester

Down’s Syndrome Screening, Quality Assurance

Labquality[14]: Helsinki

Neugeborenen-Screening – Angeborene Stoffwechseldefekte

UKNEQAS[21]: UKNEQAS Clinical Chemistry (Birmingham Quality)
- Newborn Screening:
  Phenylketonuria (PKU): phenylalanine, tyrosine and interpretation; Congenital hypothyroidism: TSH and interpretation; MCADD: C8, C10, C8/C10 ratio and interpretation

Thrombophilie-Screening

Thrombophilie Modul: Antithrombin (Aktivität u. Antigen), Protein C (Aktivität u. Antigen), Protein S (Aktivität u. Antigen), APC-Resistenz
Lupus Antikoagulanz/Antiphospholipid Antikörper
D-Dimer
Gerinnungs-Faktoren Modul I: Faktor VIII, IX, XI, XII
Gerinnungs-Faktoren Modul II: Faktor II, V, VII, X
Von Willebrand Faktor Modul: Antigen, Aktivität, Kollagen Bindung, Multimere, Faktor VIII
ADAMTS-13: Aktivität u. Antigen, Inhibitor, Antikörper
Faktor XIII

Fibrinolyse I: Plasminogen, Antiplasmin
Fibrinolyse II: t-PA, PAI-1
Unfraktioniertes Heparin Monitoring (anti-Xa)
Niedermolekulargewicht Heparin Monitoring (anti-Xa)
Homocystein
Faktor IX Inhibitor
Faktor VIII Inhibitor
Thrombin Generation Test
HIT (Heparin-Induzierte Thrombozytopenie) Immunologischer und funktionaler Test
Orale Anti Koagulantien: Orgaran, Fondaparinux, Rivaroxaban, Apixaban, Argatroban, Dabigatran
Thrombozyten Funktion-Bewegung: Postanalytik, Interpretation (elektonische Abfrage)
Thromboelastography (TEG) und Thromboelastometry (ROTEM)
Platelet Dense Granule exercise
Fallstudien bei Blutungsstörungen

CAP[3]:	CAP Headquarters, Northfield
	- Coagulation Special Testing Series (CGS1-6)
	- Coagulation, Calibration, Verification/Linearity (LN35, 36, 37)
	- D-Dimer Calibration, Verification/Linearity (LN 42)
	- Whole Blood Coagulation (WP3,4,6,9,10)
	- Platelet Function (PF, PF1)
	- Thromboelastrogram (TEG)
ECAT[6]:	Leiden
INSTAND[13]:	in Kooperation mit ECAT und eigene RVs (z.B. D-Dimer, Nr.226))
ÖQUASTA[15]:	Gerinnung
RCPA QAP[18]:	RCPAQAP Enrolment Office, St.Leonards
	Haematology Discipline
	- Specialised Haemostasis: Module A-H
	- D-Dimer: Module 1 + 2
	- Haemostasis
RfB (DGKL)[19]:	- Thrombophilie-Screening (TH)
	(in Kooperation mit ECAT; eigener RV zu D-Dimer)
UKNEQAS[21]:	UKNEQAS Blood Coagulation

V. Ringversuche Genetische Beratung

Genetics Training and Competency

GenQA[12]: G-TACT (Genetics Training and Competency Tool)
Online competency assessment modules for individuals working in Specific areas of the genetics laboratory (sample reception, duty scientist, data analysis and report authorization). The annual fee provides access to all available modules.

Clinical Genetics – Cardiovascular genetics

GenQA[12]: Clinical Genetics educational case scenario with multiple stages. Clinical diagnosis, analysis of test results and genetic counselling for cardiovascular disorders.

Clinical Genetics – Dysmorphology

GenQA[12]: Clinical Genetics educational case scenario with multiple stages. Clinical diagnosis, analysis of test results and genetic counselling for dysmorphic patients.

Clinical Genetics – Monogenic

GenQA[12]: Clinical Genetics educational case scenario with multiple stages. Clinical diagnosis, analysis of test results and genetic counselling for monogenic disorders.

Clinical Genetics – Oncogenetics

GenQA[12]: Clinical Genetics educational case scenario with multiple stages. Clinical diagnosis, analysis of test results and genetic counselling for oncology disorders.

Abkürzungen

[1]: ACMG: American College of Medical Genetics and Genomics
[2]: BVDH (HGQN): Berufsverband Deutscher Humangenetiker e. V, Deutschland (BVDH); als QM Datenbank und Plattform für externe QS des BVDH fungiert das Human Genetics Quality Network (HGQN)
[3]: CAP: College of American Pathologists
[4]: CF Network: Cystic Fibrosis European Network;
Kooperation mit EuroGentest Network of Excellence und EMQN
[5]: EAA: European Academy of Andrology
[6]: ECAT: External Quality Control of Diagnostic Assays and Tests (ECAT) Foundation
[7]: EFI: European Federation of Immunogenetics External Proficiency Testing (EPT)
[8]: EMQN: European Molecular Genetics Quality Network
[9]: EQUALIS: External quality assurance in laboratory medicine in Sweden
[10]: ESP-EQA: European Quality Assurance Program, etabliert von der European Society of Pathology (ESP)
[11]: ETRL: Eurotransplant Reference Laboratory
[12] GenQA: Genomics quality assessment: Collaboration between UK NEQAS Molecular Genetics and CEQAS from 1st January 2018.
[13]: INSTAND: Institut für Standardisierung und Dokumentation im medizinischen Laboratorium e. V.
[14]: Labquality: Helsinki, Finland
[15]: ÖQUASTA: Österreichische Gesellschaft für Qualitätssicherung und Standardisierung medizinisch-diagnostischer Untersuchungen
[16]: QuIP: Qualitätssicherungs-Initiative in der Pathologie (QuIP) der Deutsche Gesellschaft für Pathologie e.V. (DGP) und des Bundesverbandes Deutscher Pathologen (BDP) zur diagnostischen Immunhistochemie und Molekularpathologie
[17]: QUITZ: Qualitätssicherung in der Tumorzytogenetik des Kompetenznetz Leukämie
[18]: RCPA QAP: Royal College of Pathologists of Australasia Quality Assurance Programs Pty. Limited, Australien
[19]: RfB (DGKL): Referenzinstitut für Bioanalytik;
Träger des Referenzinstituts für Bioanalytik (RfB) ist die Stiftung für Pathobiochemie und Molekulare Diagnostik der DGKL (Deutsche Vereinte Gesellschaft für Klinische Chemie und Laboratoriumsmedizin e. V., Deutschland)
[20]: UHKT: Ústav hematologie a krevní transfuze (Institute of Hematology and Blood Transfusion), Prag, National Reference Laboratory for DNA Diagnostics
[21]: UKNEQAS: United Kingdom National External Quality Assessment Service

Adressen

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GenQA
From 1st January 2018 Cytogenomics External Quality Assessment Service (CEQAS) and UK National External Quality Assessment Service (UK NEQAS) for Molecular Genetics are partnering to become Genomics Quality Assessment (GenQA).
Homepage: https://www.genqa.org/

- **CEQAS**

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- **UK NEQAS for Molecular Genetics**

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EC4 – European Register of Specialists in Clinical Chemistry and Laboratory Medicine
homepage: www.ec-4.org
Under the direction of the EFLM (European Federation of Clinical Chemistry and Laboratory Medicine).

EuroGentest
homepage: www.eurogentest.org
EuroGentest is a project funded by the European Commission to harmonize the process of genetic testing, from sampling to counseling, across Europe. The ultimate goal is to ensure that all aspects of genetic testing are of high quality thereby providing accurate and reliable results for the benefit of the patients.

European Information System on Proficiency Testing Schemes (eptis)
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The European LeukemiaNet is funded by the http://www.leukemia-net.org/content/home/the_project/funding/. Goal is cure of leukemia by integration of European leukemia research and spread of excellence.

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