Schweizerische Eidgenossenschaft
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Swiss Confederation

Federal Department of Home Affairs FDHA
Federal Office of Public Health FOPH
Public Health Directorate

Medical Genetics External Quality Control Assessment Schemes

Date: September, 2019

1 Control Centres

Control Centre	Information
Gesellschaft zur Förderung der Qualitätssicherung in medizinischen Laboratorien e.V. (INSTAND)	www.instandev.de
In collaboration with MQ Zürich	www.mqzh.ch
Referenzinstitut für Bioanalytik (RfB)	www.rfb.bio
In collaboration with MQ Zürich	www.mqzh.ch
European Molecular Genetics Quality Network (EMQN)	www.emqn.org
In collaboration with CSCQ	www.cscq.ch
Genomics Quality Assessment (GenQA) Formerly United Kingdom National External (UK NEQAS) for Molecular Genetics and Cytogenetic European Quality Assessment Service (CEQAS)	www.genqa.org
Berufsverband Deutscher Humangenetiker e. V. (BVDH)	www.bvdh.de
European CF Network (CF Network)	cf.eqascheme.org
Association des Cytogénéticiens de Langue Française (ACLF)	www.eaclf.org
Centers for Disease Control and Prevention – Newborn Screening Quality Assurance Program	www.cdc.gov

2 Molecular Analysis

Analyte	Organization
A	
ACE (Angiotensin I Converting Enzyme)	INSTAND, RfB
Achondroplasia (ACH), *see skeletal dysplasia*	GenQA
Adenomatous Polyposis of the Colon (APC), *see Familial Adenomatous Polyposis Colon Cancer (FAP)*	GenQA
Aldolase B	INSTAND, RfB
Alpha-1-Antitrypsin	INSTAND, RfB
Antithrombin III	INSTAND, RfB
Apolipoprotein B100 (ApoB100)	INSTAND, RfB
Apolipoprotein E (ApoE): E2/E3/E4	INSTAND, RfB
Arrhythmias	EMQN
Arrhythmia and Cardiomyopathies (Brugada Syndrome, Long QT Syndrome, Catecholaminergic polymorphic ventricular tachycardia)	GenQA
Ataxia and spastic paraplegia (Friedreich's ataxia, spinobellar ataxia, hereditary spastic paraplegia)	GenQA
Autosomal Dominant Polycystic Kidney Disease (ADPKD)	EMQN
B	
Beckwith-Wiedemann Syndrome (11p-imprinting disorders)	EMQN
BRCA in ovarian cancer – germline **Breast and Ovarian Cancer Hereditary (BRCA1/2 testing only)** **Breast and Ovarian Cancer Hereditary (Panel testing)**	EMQN, GenQA (Pilot)
Breast/Ovarian Cancer, Familial (Cowden, Li Fraumeni, Peutz Jeuger Syndrome)	EMQN, GenQA
C	
Cardiomyopathies, *see also Arrythmia and Cardiomyopathies*	GenQA
C9orf72 related Frontotemperal Dementia and/or Amytrophic Lateral Sclerosis (C9orf72)	GenQA (Pilot)
Charcot-Marie-Tooth Disease	EMQN
Charcot-Marie-Tooth Disease, Hereditary liability to pressure palsies	GenQA
Congenital Adrenal Hyperplasia (CAH)	EMQN
Cystic Fibrosis	CF network, GenQA
D	
Dementia/ALS	GenQA
Duchenne/Becker Muscular Dystrophies	EMQN, GenQA
F	
Fabry disease	GenQA
Factor II	INSTAND, RfB

Analyte	Organization
Factor V	INSTAND, RfB
Factor XIII	INSTAND, RfB
Familial Adenomatous Polyposis Colon Cancer (FAP), *see polyposis*	EMQN, GenQA
Familial Hypercholesterolemia (FH)	GenQA, EMQN
Familial SHOX-related Disorders	EMQN
Fibrinogen, Beta	INSTAND, RfB
Fragile X Syndrome (FRAX)	EMQN, GenQA
Friedreich Ataxia	EMQN
G	
Gluten intolerance	see HLA
Globin, alpha and beta	INSTAND
GPIa	INSTAND
GPIIIa	RfB
H	
Hereditary Deafness	EMQN
Hereditary Hemochromatosis	EMQN
Hereditary Motor and Sensory Neuropathy and Hereditary Neuropathy with Liability to Pressure Palsies (HMSN/HNPP)	EMQN
Hereditary Recurrent Fevers (HRF)	EMQN
Huntington Disease (HD)	EMQN, GenQA
HLA B27	INSTAND, RfB, UKNEQAS Immunology
HLA-A	INSTAND, UKNEQAS Immunology
HLA-DRB1, HLA-DQB1 (gluten intolerance)	INSTAND
Hypertrophic Cardiomyopathies	EMQN
Hypotonic infant **(Myotonic Dystrophy Type 1, Prader-Willi Syndrome, Spinal Muscular Atrophy)**	GenQA
I	
11p-Imprinting Disorders (BWS/SRS)	EMQN
Imprinting and uniparental disomy for AS/BWS	GenQA
Interpretation (and reporting)	EMQN
L	
Lactose Intolerance (LCT -13910C>T)	INSTAND, RfB
Long QT Syndrome, *see Arrythmia*	GenQA
Lynch Syndrome (HNPCC)	EMQN
M	
Maternal Cell Contamination with Sexing (MCC & sexing)	GenQA, UKNEQAS
Medium Chain Acyl CoA Dehydrogenase Deficiency Molecular Scheme (MCADD) Blood Spot	GenQA

Analyte	Organization
MTHFR	INSTAND, RfB
MSI, Microsatellite Instability	GenQA
Mitochondrial Diseases	EMQN
Mitochondrial disorder (including POLG)	GenQA
Molecular Rapid Aneuploidy (MRA)	GenQA, BVDH
Monogenic Diabetes (MODY)	EMQN
Morbus Crohn (NOD2)	INSTAND, RfB
Multiple Endocrine Neoplasia Type 2 (MEN2)	EMQN
Myotonic Dystrophy (DM)	EMQN
N	
Neuroblastoma (Array and FISH)	GenQA (Pilot)
Neurofibromatosis Type 1 and 2 and Schwannomatosis	GenQA
O	
Osteogenesis Imperfecta	EMQN, GenQA
Osteoporosis, COL1A1	INSTAND, RfB
P	
Phaechromocytoma and Paraganglioma Disorders	GenQA
Phenylketonuria (PKU)	EMQN
Plasminogen Activator Inhibitor (PAI 1)	INSTAND. RfB
POLG, *see mitochondrial disorder*	GenQA
Porphyria	EMQN
Polyposis **(Familial adenomatous polyposis and MUTYH-associated Polyposis)**	GenQA
Prothrombin (F2), *see Factor II*	INSTAND, CSCQ, RfB
Prader-Willi and Angelman Syndromes	EMQN
Q	
QF-PCR	See MRA
R	
Retinoblastoma	EMQN
Retinal disorders	GenQA
RYR1 related myopathies and malignant hyperthermia	EMQN (Pilot)
S	
Sickle Cell Anemia (Hemoglobin S)	INSTAND
Silver-Russell Syndrome	EMQN
Skeletal dysplasia **(Achondroplasia, Hypochondroplasia and Thanatophoric dysplasia)**	GenQA
Spastic paraplegia, *see Ataxia and spastic paraplegia*	GenQA
Spinal Muscular Atrophy (SMA)	EMQN
Spinocerebellar Ataxias (SCA)	EMQN
Stickler Syndrome	EMQN (Pilot)

Analyte	Organization
T	
Thalassemia	INSTAND
TNF Alpha (periodontitis)	INSTAND, RfB
TPMT (Thiopurine S-Methyltransferase)	INSTAND, RfB
U	
UDP-Glucuronyltransferase 1 (UGT1A1) - Gilbert syndrome	INSTAND, RfB
V	
Vitamin K Epoxide Reductase Complex, Subunit 1 (VKORC1)	INSTAND, RfB
Von Hippel-Lindau Syndrome (VHL)	EMQN
W	
Wilson Disease	EMQN
X	
X-inactivation	GENQA
Y	
Y-Chromosome Microdeletion (AZF)	EMQN

3 Clinical Genetics

Online case scenario

Analyte	Organization
Cardiovascular disease	GenQA
Dysmorphology	GenQA
Monogenic Disorder	GenQA
Pathogenicity of sequence variants	GenQA
Severe Intellectual Disability Interpretation	GenQA (Pilot)

4 Cytogenetic Analysis

Method	Organization
Chromosome Breakage Syndromes	GenQA
Constitutional CNV detection (array)	GenQA
Constitutional Cytogenetics – Prenatal	BVDH, ACLF, GenQA
Constitutional Cytogenetics – Postnatal (Blood)	BVDH, ACLF, GenQA
Constitutional Cytogenetics – Molecular Rapid Aneuploidy (MRA)	GenQA, BVDH
FISH – Constitutional Abnormality	BVDH, GenQA
FISH – Rapid Prenatal Aneuploidy	BVDH, GenQA

Method	Organization
Microarray/Array CGH	EMQN, GenQA, BVDH
Microarray/Array CGH, Prenatal	GenQA
Strukturanalyse (analyze of structure)	BVDH
Non-invasive Prenatal Testing for common aneuploidies	EMQN, GenQA (Pilot)
Non-invasive Prenatal Testing for fetal sexing	EMQN, GenQA (Pilot)
Non-invasive Prenatal Testing for common microdeletions	GenQA (Pilot)

5 Molecular Testing on Blood Spots

Analyte	Organisation
Cystic Fibrosis	GenQA, CDC
Medium Chain Acyl-CoA Dehydrogenase Deficiency	GenQA

6 Pharmacogenetics

Analyte	Organisation
Butyrylcholinesterase (BCHE)	INSTAND, RfB
Cytochrome P450 2C9 (CYP2C9)	INSTAND, RfB
Cytochrome P450 2C19 (CYP2C19)	INSTAND, RfB
Cytochrome P450 2D6 (CYP2D6)	INSTAND, RfB
FSH Receptor	INSTAND
Growth Hormone Receptor (GH-R)	INSTAND
HLA B*5701	INSTAND, RfB
Interleukin IL-28B	INSTAND, RfB
Pharmacogenetics panel (drug intolerance and effectivity)	EMQN (Pilot)
Thiopurine-S-Methyltransferase deficiency (TPMT)	INSTAND, RfB
UDP-Glycosyltransferase 1 (UGT1A1)	INSTAND, RfB
VKORC1	INSTAND, RfB

7 Methodological

Method	Organisation
DNA extraction from blood	RfB, GenQA, UKNEQAS
DNA extraction form saliva	GenQA, UKNEQAS

Method	Organisation
DNA quantification	GenQA
DNA Sequencing (Sanger)	EMQN, RfB
DNA Sequencing (NGS) Germline	EMQN, GenQA (Pilot)
DNA Sequencing (NGS) Somatic	EMQN, GenQA (Pilot)
Variant Validation	GenQA

8 Preimplantation Genetic Testing

Analyte	Organisation
Preimplantation Genetic Testing for monogenic disorders	GenQA, UKNEQAS
Preimplantation Genetic Testing for Blastomere FISH	GenQA
Preimplantation Genetic Testing for Blastomere/Trophectoderm Chromosomal Rearrangement	GenQA, UKNEQAS
Preimplantation Genetic Testing Array/NGS for Polar Bodies	GenQA
Preimplantation Genetic Testing of Trophectoderm/ Blastomere Array/NGS	GenQA