Schweizerische Eidgenossenschaft
Confédération suisse
Confederazione Svizzera
Confederaziun svizra

Swiss Confederation

Federal Department of Home Affairs FDHA

Federal Office of Public Health FOPH
Health Protection Directorate

Medical Genetics External Quality Control Assessment Schemes

Date: February 2024

1 Control Centres

Control Centre	Information
Gesellschaft zur Förderung der Qualitätssicherung in medizinischen Laboratorien e.V. (INSTAND) (ISO/IEC 17043:2010) In collaboration with MQ Zürich	www.instandev.de www.mqzh.ch
Referenzinstitut für Bioanalytik (RfB) (ISO/IEC 17043:2010) In collaboration with MQ Zürich	www.rfb.bio www.mqzh.ch
European Molecular Genetics Quality Network (EMQN) (ISO/IEC 17043:2010**)** In collaboration with CSCQ	www.emqn.org www.cscq.ch
Genomics Quality Assessment (GenQA) (ISO/IEC 17043:2010)	www.genqa.org
UK NEQAS (ISO/IEC 17043:2010)	www.ukneqas.org.uk
Berufsverband Deutscher Humangenetiker e. V. (BVDH) (ISO/IEC 17043:2010)	www.bvdh.de
European CF Network (CF Network) (ISO/IEC 17043:2010)	eqascheme.org
Centers for Disease Control and Prevention – Newborn Screening Quality Assurance Program	www.cdc.gov
Hôpitaux Universitaires Genève **Laboratoire National de Référence pour l'Histocompatibilité**	www.hug.ch/batlab/centres-nationaux-reference

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
Amyloidosis, Hereditary	EMQN (pilot)
Antithrombin III	INSTAND, RfB
Apolipoprotein B100 (ApoB100)	INSTAND, RfB
Apolipoprotein E (ApoE): E2/E3/E4	INSTAND, RfB
Ataxias and Hereditary Spastic Paraplegia Friedreich ataxia, spinocerebellar ataxia, hereditary spastic paraplegia, frataxin	GenQA
Autosomal Dominant Polycystic Kidney Disease (ADPKD)	EMQN
B	
Beckwith-Wiedemann Syndrome, Silver-Russel Syndromes	EMQN
BRCA in Ovarian Cancer – Germline	EMQN, GenQA
Breast and Ovarian Cancer Hereditary (BRCA1/2 Testing Only)	EMQN
Breast and Ovarian Cancer Hereditary (Panel Testing)	EMQN
Breast Cancer (AKT pathway testing)	EMQN (pilot)
Breast Cancer (ESR1 testing)	EMQN (pilot)
Breast/Ovarian Cancer **Cowden, Li Fraumeni, Peutz Jeuger Syndrome**	EMQN, GenQA
C	
Calcium Disorders Hypercalcaemia and hypocalcaemia including: Familial hypoparathyroidism, Albright hereditary osteodystrophy, Pseudohypoparathyroidism, Pseudopseudohypoparathyroidism, Isolated hyperparathyroidism, Hypocalciuric hypercalcaemia, Calcium sensing receptor phenotypes	GenQA
Cardiac Genetics (Arrhythmias) **Cardiac Genetics (Hypertrophic Cardiomyopathies)**	EMQN, GenQA
Cardiac Disorders Brugada syndrome, Long QT syndrome, Catecholaminergic polymorphic ventricular tachycardia (CPVT), general arrhythmia, cardiomyopathies, Marfan syndrome, Ehlers Danlos syndrome	GenQA
Charcot-Marie-Tooth Disease and Related Sensory and Motor Neuropathies	EMQN, GenQA

Analyte	Organization
Chromosome Instability Syndromes Fanconi anaemia, Blood syndrome, Ataxia telangiectasia and Niijemen Syndrome, Robert Syndrome, ICR, Cornelia de Lange syndrome, Mosaic variegated aneuploidy and Seckel syndrome	GenQA
Congenital Adrenal Hyperplasia (CAH), *see Disorders of Sexual Development*	EMQN, GenQA
Cystic Fibrosis	CF network, GenQA
E	
Epilepsy Disorders **Tuberous Sclerosis, Rett Syndrome, Dravet Syndrome and focal seizures**	GenQA
Eye Disorders	GenQA
D	
Dementia/ALS	GenQA
Differences in Sex Development Androgen insensitivity syndrome, Congenital adrenal hyperplasia, cytogenomic abnormalities and other disorders associated with a DSD NGS panel	GenQA
Duchenne/Becker Muscular Dystrophy (DMD)	EMQN, GenQA
F	
Fabry Disease	GenQA
Factor II	INSTAND, RfB
Factor V	INSTAND, RfB
Factor XIII	INSTAND, RfB
Familial Adenomatous Polyposis Colon Cancer (FAP), *see Polyposis*	EMQN, GenQA
Familial Colorectal Cancer and Polyposis	GenQA
Familial Endocrine Tumour Predisposition Disorders Von Hippel-Lindau disease (VHL), Multiple Endocrine Neoplasia (MEN) and Familial medullary thyroid carcinoma (FMTC)	GenQA
Familial Hypercholesterolemia (FH)	GenQA, EMQN
Familial Medullary Carcinoma, *see Familial Endocrine Tumour Predisposition Disorders*	GenQA
Familial SHOX-Related Disorders	EMQN
Fibrinogen, Beta	INSTAND, RfB
Fragile X Syndrome (FRAX) Only Full Version	EMQN, GenQA
Friedreich Ataxia (FRDA)	EMQN, GenQA
G	
Gastrohepatology Disorders	GenQA
Gluten Intolerance	see HLA
Globin, Alpha and Beta	INSTAND
GPIa	INSTAND
GPIIIa	RfB

Analyte	Organization
H	
Hereditary Deafness (DFNB1)	EMQN
Hereditary Haemochromatosis (HFE)	EMQN, UKNEQAS
Hereditary Motor and Sensory Neuropathy and Hereditary Neuropathy with Liability to Pressure Palsies (HMSN/HNPP)	EMQN
Systemic Autoinflammatory Disease	EMQN
Huntington Disease (HD)	EMQN, GenQA
HLA B27	INSTAND, RfB, UKNEQAS, HUG
HLA-A	INSTAND, UKNEQAS, HUG
HLA-DRB1, HLA-DQB1, HLA-DQA1 (Gluten Intolerance)	INSTAND, HUG, UKNEQAS
Hypertrophic Cardiomyopathies (Panel Testing)	EMQN
Hypotonic Infant Myotonic Dystrophy Type 1, Prader-Willi Syndrome, Spinal Muscular Atrophy	GenQA
I	
Imprinting disorders Angelman Syndrome (AS), BeckwithWiedemann Syndrome (BWS), Silver Russell Syndrome (SRS), Wilms tumour and Temple syndrome	EMQN, GenQA
Inborn Errors of Metabolism	GenQA
Infertility (online), *see List 3 Clinical Genetics* Chromosomal mosaicism, CFTR, FMR1, Y-deletions	GenQA
L	
Lactose Intolerance (LCT -13910C>T)	INSTAND, RfB
Linkage Analysis (online), *see List 3 Clinical Genetics*	GenQA
Long QT Syndrome, *see Arrythmia*	GenQA
Lynch Syndrome (HNPCC), *see Familial Colorectal Cancer and Polyposis*	EMQN, GenQA
M	
Maternal Cell Contamination and Sexing (MCC & Sexing)	GenQA
Medium Chain Acyl CoA Dehydrogenase Deficiency Molecular Scheme (MCADD) Blood Spot	GenQA
MTHFR	INSTAND, RfB
Microdeletion Syndromes Prader-Willi syndrome, Angelman syndrome, Williams syndrome and Di-George syndrome	GenQA
MSI, Microsatellite Instability Testing	GenQA, EMQN
Mitochondrial Diseases (mtDNA), Metabolic Disorders	EMQN
Mitochondrial Disorder (including POLG)	GenQA
Molecular Rapid Aneuploidy (MRA), *see list 4 Cytogenetic Analysis*	GenQA, BVDH
Monogenic Diabetes (MONODIAB)	EMQN
Morbus Crohn (NOD2)	INSTAND, RfB
Multiple Endocrine Neoplasia Type 2, *see Familial Endocrine Tumour Predisposition Disorders*	EMQA GenQA

Analyte	Organization
Myotonic Dystrophy (Type 1 & 2) (DM)	EMQN, GenQA
N	
Neuroblastoma	GenQA
Neurodegenerative Disorders Alzheimer Disease, Frontotemporal Dementia, Amylotrophic Disease, Parkinson Disease	GenQA
Neurofibromatosis Type 1 and Rasopathies	GenQA
Neurological Disease, Rare	EMQN
Neuromuscular Disease, Rare	EMQN (pilot)
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
Porphyrias	EMQN
Polyposis Syndromes (Familial Adenomatous Polyposis and MUTYH-associated Polyposis)	EMQN; GenQA
Primary Immunodeficiency Disorders Severe Combined Immunodeficiency (SCID), Agammaglobulinaemia, Hereditary angioedema, Chronic granulomatous disease and Hyper IgE syndrome	GenQA
Prothrombin (F2), *see Factor II*	INSTAND, CSCQ, RfB
Prader-Willi and Angelman Syndromes	EMQN, GenQA
Q	
QF-PCR	See MRA
R	
Retinoblastoma	EMQN
Ophthalmological Disease	EMQN (pilot)
Renal Disorders Haematuria, tubulointerstitial kidney disease, cystic renal disease and Alport syndrome	GenQA
Respiratory Disorders Pneumothorax, respiratory insufficiency, bronchiectasis (ciliopathies/PCD and surfactants) and pulmonary arterial disease	GenQA
RYR1 related Myopathies and Malignant Hyperthermia susceptibility	EMQN
S	
Severe Combined Immunodeficiency (SCID)	EMQN
Sickle Cell Anemia (Hemoglobin S)	INSTAND

Analyte	Organization
Silver-Russell Syndrome, *see Beckwith-Wiedemann Syndrome*	EMQN
Skeletal dysplasia FGFR2/FGFR3 related disorders, OI and other skeletal dysplasias	GenQA
Spastic paraplegia, *see Ataxia and spastic paraplegia*	GenQA
Spinal Muscular Atrophy (SMA)	EMQN, GenQA
Spinocerebellar Ataxia's (SCA)	EMQN
Stickler Syndrome (Panel testing)	EMQN
T	
Thalassemia	INSTAND
TNF Alpha (periodontitis)	INSTAND, RfB
TPMT (Thiopurine S-Methyltransferase)	INSTAND, RfB
Tuberous Sclerosis, *see Epilepsy Disorders*	GenQA
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), *see also Familial Endocrine tumour predisposition disorders*	EMQN, GenQA
W	
Wilson Disease (WIL)	EMQN
X	
X-Inactivation	GenQA
Y	
Y-Chromosome Microdeletion (AZF)	EMQN

3 Clinical Genetics

Online case scenario

Analyte	Organization
Cardiovascular Disease	GenQA
Dysmorphology	GenQA
Exome Sequencing Data Interpretation	GenQA
Infertility	GenQA
ISCN Accuracy	GenQA
Linkage Analysis	GenQA
Metabolic disorders	GenQA
Monogenic Disorder	GenQA

Analyte	Organization
Pathogenicity of Copy Number Variant (CNV)	GenQA
Pathogenicity of Sequence Variants	GenQA
Development Delay	GenQA
Severe Intellectual Delay	GenQA
Variant Validation	GenQA

4 Cytogenetic Analysis

Method	Organization
CNV Detection – Prenatal	GenQA, EMQN, BVDH
CNV Detection – Postnatal (Array/NGS)	GenQA, EMQN, BVDH
Constitutional Cytogenetics – Prenatal (Amniotic Fluid, Chorionic Villus)	BVDH, ACLF, GenQA
Constitutional Cytogenetics – Postnatal (Blood)	BVDH, ACLF, GenQA
Molecular Rapid Aneuploidy (MRA), FISH or QF-PCR	GenQA, BVDH
FISH – Constitutional Abnormality	BVDH, GenQA
Microdeletion Syndromes	GenQA
Non-invasive Prenatal Testing (NIPT) for Common Aneuploidies	EMQN, GenQA
Non-invasive Prenatal Testing (NIPT) for Fetal Sex Determination	EMQN, GenQA
Non-invasive Prenatal Testing for Common Microdeletions	GenQA
Pregnancy Loss (G-banding)	GenQA
Pregnancy Loss (molecular methods)	GenQA
Strukturanalyse (analyze of structure)	BVDH

5 Molecular Testing on Blood Spots

Analyte	Organisation
Cystic Fibrosis	GenQA, CDC
Medium Chain Acyl-CoA Dehydrogenase Deficiency	GenQA
Severe Combined Immunodeficiency (SCID), TREC	GenQA, CDC
Spinal Muscular Atrophy	GenQA

6 Pharmacogenetics

Analyte	Organisation
Aminoglycoside Induced Deafness (MT-RNR1)	GenQA
Butyrylcholinesterase (BCHE)	INSTAND, RfB
Cytochrome P450 2C9 (CYP2C9)	INSTAND, RfB
Cytochrome P450 2C19 (CYP2C19)	INSTAND, RfB
Cytochrome P450 2D6 (CYP2D6)	INSTAND, RfB, GenQA (pilot)
5-Fluorouracil Toxicity/DPYD	GenQA, EMQN (pilot)
HLA B*5701	INSTAND, RfB. UKNEQAS
Interleukin IL-28B	INSTAND, RfB
Pharmacogenetics (Drug Intolerance and Effectivity)	EMQN, GenQA (pilot)
Clopidogrel effectiveness (Cyp2C19)	GenQA (pilot)
Thiopurine-S-Methyltransferase Deficiency (TPMT)	GenQA, INSTAND, RfB
UDP-Glycosyltransferase 1 (UGT1A1)	INSTAND, RfB
VKORC1	INSTAND, RfB

7 Methodological

Method	Organisation
DNA Extraction from Blood	RfB, GenQA
DNA Extraction form Saliva	GenQA
DNA Sequencing (Sanger)	EMQN, RfB
DNA Sequencing (NGS Germline SNVs and Indels)	EMQN, GenQA, BVDH
DNA Sequencing (NGS Germline) - CNV Testing	EMQN, GenQA (pilot)
DNA Sequencing (NGS) Somatic	EMQN (pilot), GenQA
Exome trio sequencing - prenatal	GenQA
Exome trio sequencing - postnatal	GenQA
NGS data analysis	BVDH

8 Preimplantation Genetic Testing

Analyte	Organisation
Genetic Testing for Monogenic Disorders	GenQA
Genetic Testing for Aneuploidies (Array/NGS)	GenQA
Genetic Testing for Chromosomal Rearrangement (Array/NGS)	GenQA

Analyte	Organisation
Genetic Testing for Aneuploidies and Structural Rearrangements (FISH)	GenQA
Polar Bodies (Array/NGS)	GenQA
Genetic Testing for Aneuploidy and Structural Rearrangements (Array/NGS)	GenQA

Screening

Analyte	Organisation
Carrier Screening for Preconception Carrier Screening	GenQA

Genetic counseling

Analyte	Organisation
Genetic Counseling	BVDH, GenQA

Contact

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