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: November 2025

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)	www.eqascheme.org/
Centers for Disease Control and Prevention – Newborn Screening Quality Assurance Program	www.cdc.gov
SKML (Dutch Foundation for Quality Assessment in Medical Laboratories) (ISO/IEC 17043:2010)	www.skml.nl

2 Molecular Analysis

Analyte	Centre
A	
ACE (Angiotensin I Converting Enzyme)	INSTAND, RfB
Aldolase B	INSTAND, RfB
Alpha-1-Antitrypsin	INSTAND, RfB
Amyloidosis, Hereditary	EMQN
Antithrombin III	INSTAND, RfB
Apolipoprotein B100 (ApoB100)	INSTAND, RfB
Apolipoprotein E (ApoE): E2/E3/E4	INSTAND, RfB
Ataxias and Hereditary Spastic Paraplegia Charcot Marie Tooth disease (CMT) and Hereditary Liability to Pressure Palsies (HNPP), including PMP22, GJB1, MPZ, MFN2 and other associated genes	GenQA
Autoinflammatory Disease	EMQN
B	
Beckwith-Wiedemann Syndrome, Silver-Russel Syndromes	EMQN
BRCA in Ovarian Cancer – Germline	EMQN, GenQA
C	
Calcium Disorders Arrythmias: Brugada syndrome, Long QT syndrome, Catecholaminergic polymorphic ventricular tachycardia (CPVT) and Progressive cardiac conduction disease. Cardiomyopathies: Hypertrophic cardiomyopathy, dilated cardiomyopathy, arrhythmogenic ventricular cardiomyopathies and paediatric cardiomyopathy. Syndromic aortopathies: Marfan syndrome, Ehlers Danlos syndrome and Loeys Dietz syndrome. Non syndromic aortopathy.	GenQA
Cardiac Disorders Brugada syndrome, Long QT syndrome, Catecholaminergic polymorphic ventricular tachycardia (CPVT), general arrhythmia, cardiomyopathies, Marfan syndrome, Ehlers Danlos syndrome	GenQA, EMQN
Charcot-Marie-Tooth Disease and Related Sensory and Motor Neuropathies Charcot Marie Tooth disease (CMT) and Hereditary Liability to Pressure Palsies (HNPP), including PMP22, GJB1, MPZ, MFN2 and other associated genes	EMQN, GenQA
Chromosome Instability Syndromes Ataxia telangiectasia, Bloom syndrome, Cornelia de Lange syndrome, Fanconi anaemia, ICR, syndrome, mosaic variegated aneuploidy, Nijmegen syndrome, Roberts syndrome and Seckel syndrome	GenQA
COL1A1	RfB
Congenital Adrenal Hyperplasia (CAH), *see Differences of Sexual Development*	EMQN, GenQA
Connexin 26 (GJB2 Gen, GJB6 Gen)	INSTAND
Cystic Fibrosis	CF network, GenQA

Analyte	Centre
Cytopenias, inherited[new] Dyskeratosis congenita, Diamond Blackfan anemia, Fanconi anemia, Shwachman Diamond syndrome, congenital thrombocytopenia, and inherited causes of neutropenia. Targeted testing for variants in genes associated with inherited cytopenias	GenQA (pilot)
D	
Developmental Delay Interpretation of genetic causes of developmental delay, includes molecular and cytogenomic test results	GenQA
Differences in Sex Development (DSD) Androgen insensitivity syndrome, Congenital adrenal hyperplasia, cytogenomic abnormalities and other disorders associated with a DSD NGS panel	GenQA
E	
Epilepsy Disorders Monogenic epilepsies and genetic epilepsy syndromes including: Tuberous sclerosis, Rett syndrome and Dravet syndrome.	GenQA
Eye Disorders Retinopathies, structural eye disorders, optic atrophy, cataracts and albinism	GenQA
F	
Fabry Disease, *see Inborn Errors of Metabolism*	GenQA
Factor II	INSTAND, RfB
Factor V-Leiden	INSTAND, RfB
Factor XIII	INSTAND, RfB
Familial colorectal cancer and polyposis Lynch syndrome, Familial Adenomatous Polyposis and MUTYH-associated Polyposis (MAP)	EMQN, GenQA
Familial Endocrine Tumour Predisposition Disorders MEN1, MEN2, VHL and FMTC disorders	GenQA
Familial Hypercholesterolemia (FH) LDLR, APOB and PCSK9	GenQA, EMQN
Familial SHOX Gene Related Disorders	EMQN
Fibrinogen, Beta	RfB
Fibrinogen receptor HPA 1a/1b	INSTAND
Fragile X Syndrome (FRAX) Only Full Version	EMQN, GenQA
Friedreich Ataxia (FRDA)	EMQN, GenQA
G	
Gastroenterology and Hepatology Disorders Hereditary Pancreatitis, Gilbert syndrome, Cholestasis, Hirschsprung disease, Polycystic liver disease	GenQA
Gluten Intolerance *see HLA*	INSTAND, UKNEQAS
Globin, Alpha and Beta	INSTAND
GPIIIa	RfB
H	
Haemoglobinopathies	INSTAND, UKNEQAS

Analyte	Centre
Hereditary Breast and Ovarian Cancer (HBOC) Disorders Familial Breast and Ovarian Cancer (BRCA1 and BRCA2), Cowden Syndrome and Li-Fraumeni	EMQN, GenQA
Hereditary Cystic Kidney Disease	EMQN
Hereditary Hearing Loss (DFNB1)	EMQN
Hereditary Endocrine Cancer	EMQN
Hereditary Haemochromatosis (HFE)	EMQN, UKNEQAS
Hereditary Motor and Sensory Neuropathy and Hereditary Neuropathy with Liability to Pressure Palsies (HMSN/HNPP)	EMQN
Hereditary Renal/Kidney Cancer (VHL)	EMQN
Homocystinuria due to MTHFR deficiency	INSTAND, RfB
HLA-B27	INSTAND, RfB, UKNEQAS
HLA-A	INSTAND, UKNEQAS
HLA-DRB1, HLA-DQB1, HLA-DQA1 (Gluten Intolerance)	INSTAND, UKNEQAS
Huntington Disease (HD) and DRPLA	EMQN, GenQA
Hypertrophic Cardiomyopathies (Panel Testing)	EMQN
Hypotonic Infant Spinal Muscular Atrophy type 1 (SMA), Prader Willi Syndrome (PWS) and Myotonic Dystrophy type 1 (DM1)	GenQA
I	
Imprinting Disorders Angelman Syndrome (AS), Beckwith Wiedemann Syndrome (BWS), Silver Russell Syndrome (SRS), Wilms tumour and Temple syndrome	EMQN, GenQA
Inborn Errors of Metabolism MCADD, Fabry disease, galactosaemia, lysosomal storage disease, urea cycle disorders, organic acidaemias	GenQA
Infertility Chromosomal mosaicism, CFTR, FMR1, Y-deletions	GenQA
K	
Kollagenrezeptor GP Ia/IIa	INSTAND
L	
Lactose Intolerance (LCT -13910C>T)	INSTAND, RfB
Lynch Syndrome (HNPCC), *see Familial Colorectal Cancer and Polyposis*	EMQN, GenQA
M	
Maternal Cell Contamination and Sexing (MCC & Sexing)	GenQA
Microdeletion Syndromes Prader-Willi syndrome, Angelman syndrome, Williams syndrome and Di-George syndrome	GenQA
Mitochondrial Disease Mitochondrial and POLG-related disorders	EMQN, GenQA
Monogenic Diabetes (MONODIAB)	EMQN
Morbus Crohn (NOD2)	INSTAND, RfB

Analyte	Centre
Muscular Dystrophies DMD-related and other muscular dystrophies	GenQA
Myotonic Dystrophy (Type 1 & 2) (DM), *see Hypotonic Infant*	EMQN, GenQA
N	
Neurodegenerative Disorders Alzheimer disease, Frontotemporal dementia, Motor neurone disease/Amyotrophic lateral sclerosis (ALS), Parkinson disease and Spinal and bulbar muscular atrophy (SBMA)	GenQA
Neurofibromatosis and Rasopathies Neurofibromatosis (types 1 and 2), Noonan syndrome and schwannomatosis	GenQA
Neurological Disease, Rare	EMQN
Neuromuscular Disorders	EMQN
O	
Osteogenesis Imperfecta	EMQN
P	
Parathyroid and Calcium regulation Disorders Hypercalcaemia and hypocalcaemia including: Familial hypoparathyroidism, Albright hereditary osteodystrophy, Pseudohypoparathyroidism, Pseudopseudohypoparathyroidism, Isolated hyperparathyroidism, Hypocalciuric hypercalcaemia, Calcium sensing receptor phenotypes	GenQA
Phaeochromocytoma and Paraganglioma Disorders	GenQA
Phenylketonuria (PKU)	EMQN
Plasminogen Activator Inhibitor (PAI-1)	INSTAND. RfB
POLG, *see Mitochondrial Disorder*	GenQA
Porphyries	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
R	
Retinal Disorders, Inherited	EMQN
Retinoblastoma	EMQN
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

Analyte	Centre
S	
Severe Combined Immunodeficiency (SCID), *see Primary Immunodeficiency Disorders*	EMQN, GenQA
Silver-Russell Syndrome, *see Beckwith-Wiedemann Syndrome*	EMQN
Skeletal dysplasia FGFR2/FGFR3 related disorders, Osteogenesis Imperfecta and other skeletal dysplasias	GenQA
Spastic paraplegia, *see Ataxia and spastic paraplegia*	GenQA
Spinal Muscular Atrophy (SMA), *see Hypotonic Infant*	EMQN, GenQA
Spinocerebellar Ataxia's (SCA)	EMQN
Stickler Syndrome (Panel testing)	EMQN
T	
TNF Alpha (periodontitis)	RfB
TPMT (Thiopurine S-Methyltransferase)	INSTAND, RfB
Tuberous Sclerosis, *see Epilepsy Disorders*	GenQA
U	
UDP-Glucuronyltransferase 1 (UGT1A1) - Gilbert syndrome	INSTAND, RfB, GenQA (pilot)
V	
Von Hippel-Lindau Syndrome (VHL), *see also Familial Endocrine tumour predisposition disorders*	GenQA
W	
Wilson Disease (WIL)	EMQN
X	
X-Inactivation	GenQA
Y	
Y-Chromosome Microdeletion (AZF)	EMQN

3 Classification and Nomenclature

Analyte	Centre
Classification and Interpretation of germline CNV	GenQA
Classification and Interpretation of germline SNVs and Indels	GenQA
Classification of germline SNVs and Indels	GenQA
BRCA und HRR gene variant classification	GenQA
Germline SNV classification	GenQA
Germline CNV classification	GenQA
HGVS nomenclature	GenQA

Analyte	Centre
Clinical Genetics- Cardiovascular Disorders	GenQA
Clinical Genetics - Dysmorphology	GenQA
Clinical Genetics – Inherited Metabolic Disorders	GenQA
Clinical Genetics – Monogenic Disorders	GenQA

4 Cytogenetic Analysis

Method	Centre
CNV Detection – Prenatal	GenQA, EMQN, BVDH
CNV Detection – Postnatal (Array/NGS)	GenQA, EMQN, BVDH
Constitutional Cytogenetics – Prenatal (Amniotic Fluid, Chorionic Villus)	BVDH, GenQA
Constitutional Cytogenetics – Postnatal (Blood)	BVDH, GenQA
Molecular Rapid Aneuploidy (MRA), FISH or QF-PCR	GenQA, BVDH
FISH – Constitutional Abnormality	BVDH, GenQA
Non-invasive Prenatal Testing (NIPT) for Common Aneuploidies	EMQN, GenQA
Non-invasive Prenatal Testing (NIPT) for Fetal Sex Determination (X-linked disorders)	EMQN, GenQA
Non-invasive Prenatal Testing for Common Microdeletions	GenQA
Pregnancy Loss (G-banding)	GenQA
Strukturanalyse	BVDH

5 Molecular Testing on Blood Spots

Analyte	Centre
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	Centre
Aminoglycoside Ototoxicity	GenQA
Cytochrome P450 2C9 (CYP2C9)	INSTAND, RfB
Cytochrome P450 2C19 (CYP2C19)	GenQA, INSTAND, RfB

Analyte	Centre
Cytochrome P450 2D6 (CYP2D6)	GenQA, INSTAND, RfB,
Dihydropyrimidine dehydrogenase (DPYD)	GenQA, INSTAND
Abacavir Hypersensitivität HLA B*5701	INSTAND, RfB. UKNEQAS
Interleukin IL-28B	INSTAND, RfB
Thiopurine-S-Methyltransferase Deficiency (TPMT)	GenQA, INSTAND, RfB
UDP-Glycosyltransferase 1 (UGT1A1)	GenQA*new*, INSTAND, RfB,
Vitamin K epoxide reductase complex subunit 1 (VKORC1)	INSTAND, RfB
Pharmacogenetics DPYD / UGT1A1 testing only	EMQN
Pharmacogenetics APOE	GenQA
Pharmacogenetic Panel CYP2B6, CYP2C9, CYP2C19, CYP2D6, CYP3A5, DPYD, fVl, NUDT15, SLCO1B1, TPMT, UGT1A1 and VKORC1	EMQN, GenQA

7 Methodological

Method	Centre
cfDNA Extraction from Plasma	GenQA, EMQN
DNA Extraction from Blood	GenQA, RfB,
DNA Extraction from Blood for Long Read Sequencing	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)
Trio sequencing - prenatal	GenQA
Trio sequencing - postnatal	GenQA
NSG Bioinformatic	BVDH
NGS Data Analysis	BVDH
Optical Genome Mapping – Rare Disease	GenQA

8 Preimplantation Genetic Testing

Analyte	Centre
Genetic Testing for Monogenic Disorders (PGT-M)	GenQA
Genetic Testing for Aneuploidies (PGT-A)	GenQA
Genetic Testing for Structural Rearrangement (PGT-SR)	GenQA
Genetic Testing of Blastomeres for Structural Rearrangements (FISH)	GenQA

9 Preproductive Genomics

Analyte	Organisation
Reproductive Carrier Screening Molecular testing for preconception carrier screening	GenQA

10 Genetic counselling

Analyte	Organisation
Genetic Counselling	BVDH, GenQA

Contact

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