KWALITEIT BEWAKING IN DE BELGISCHE CENTRA VAN MENSELIJKE ERFELIJKHEID:

KWALITEITSCONTROLE VAN ZELDZAME ZIEKTEN GENETISCHE TESTS

Lijst van de financierbare externe kwaliteitsevaluaties (EKE)-2023

EKE	Leverancier(s)
AldoB (149,174,334)	RfB
Alpha and Beta Mutations (DNA for hemoglobinopathies)	UKNEQAS Heamatology and Transfusion
Alpha1-Antitrypsin (a1-PI)	INSTAND
Aminoglycoside Induced deafness	GenQA
ApoB	INSTAND
Alipoprotein E_genotype-APOE E2/E3/E4	INSTAND
Ataxia and spastic paraplegias	GenQA
Autosomal Dominant Polycystic Kidney disease	EMQN
Beckwith - Wiedemann & Silver-Russell syndromes	EMQN
BRCA testing for ovarian, breast, prostate and pancreatic cancer -germline	GenQA
Calcium disorders	GenQA
Cardiac Disorders	GenQA
Cardiac genetics-Arrythmias	EMQN
Cardiac genetics-Hypertrophic cardiomyopathies	EMQN
Charcot-Marie-Tooth disease/HNPP	EMQN/GenQA
Charcot Marie Tooth disease and related sensory and motor neuropathies	GenQA
Chromosome breakage syndrome	GenQA
Congenital Adrenal Hyperplasia	EMQN
Cystic fibrosis	CF NETWORK
Cystic fibrosis and CFTR related disorders	GenQA
Disorders of sex development	GenQA
DNA Sequencing NGS (vGermline)	EMQN/GenQA
DNA Sequencing Sanger	EMQN
DPYD	RfB/CAP/GenQA/INSTAND
Duchenne / Becker Muscular Dystrophy	EMQN

Eye disorders	GenQA
Epilepsy disorder	GenQA
Exome Sequencing Data Interpretation	GenQA
Familial autosomal dominant hypercholesterolemia	EMQN
Familial Colorectal Cancer and Polyposis (Lynch syndrome, Familial adenomatous polyposis (FAP) and MUTYH-associated polyposis)	GenQA
Familial endocrine tumour predisposition disorders	GenQA
Familial SHOX-related disorders	EMQN
Fragile X Syndrome	EMQN
Friedreich Ataxia	EMQN
Gastrohepatology disorders	GenQA
Haemophilie (Genetics of Heritable Bleeding & Thrombotic Disorders)	UKNEQAS Blood Coagulation
Hereditary Breast and Ovarian Cancer (HBOC) Panel testing version	EMQN/ GenQA
Hereditary Deafness	EMQN
Hereditary Haemochromatosis	EMQN/INSTAND/ RfB
HLA-B*57:01 Typing for Drug Hypersensitivity	UKNEQAS for H&I
Huntington Disease	EMQN
Huntington and DRPLA	GenQA
Hypotonic infant	GenQA
Imprinting disorders	GenQA
Inborn errors of metabolism (Sample based or interpretation)	GenQA
Infertility include Sex chromosome disorders karyotyping	GenQA
Inherited cancer sequencing panel (BRCA2, CDKN2A, MLH1, MSH2,MSH6, and PMS2)	Cap
Inherited retinal disorders	EMQN
ISCN Accuracy	GenQA
Linkage analysis	GenQA
Lynch Syndrome	EMQN
Maternal Cell Contamination (MCC) and sexing	GenQA
MGL4 Diseases/disorders related to Ashkenazi Jewish ancestry (Fanconi, Bloom, Canavan, Gaucher, Tay-Sachs, Niemann-Pick	CAP
Mitochondrial DNA (mtDNA) Metabolic Disorders	EMQN

Mitochondrial disease (including POLG)	GenQA
Molecular genetics group 1 : SET A : PAI-4G5G	RfB
Molecular genetics group 1 : SET B: FXIII V34L, GPIIIa, βFib (g455a), VKORC1 (G-1639A/C1173T), FXII c46t, FV-H1299R	RfB
Molecular genetics group 1 : SET D: HFE, AldoB	RfB
Molecular genetics group 1 : SET F: Faktor VII (R353Q), AT3 Cambridge Typ I/II, CYP3A5*3	RfB
Molecular genetics group 2 SETA: TPMT, CYP2C8 (K399R), CYP2C9*2/*3, UGT1A1 (*28), DPYD *2A (Ex 14 skipp), BCHE A/K, DPYD*13, DPYD D949V (rs67376798)	RfB
Molecular genetics group 2 SET D : CYP2D6, CYP2C19 (*2/*17), CYP2C19*3	RfB
Molecular genetics group 2 SET E : HLA B*5701, CYP2B6*6, ABCB1 (MDR1) c.3435C>T, CCR5-del-32bp	RfB
Molecular genetics group 2 SET F : IL28B (C/T Polymorphismus), IL6 (G174C), CYP3A4*22	RfB
Molecular testing for cystic fibrosis (CF) (blood spot)	GenQA
Monogenic Diabetes	EMQN
Multiple Endocrine Neoplasia Type 2	EMQN
Muscular Dystrophies	GenQA
Myotonic Dystrophy	EMQN
Neurodegenerative disorders	GenQA
Neurofibromatosis and rasopathies	GenQA
NIPT for common aneuploidies	EMQN/GenQA
NIPT for common microdeletions pilot	GenQA
Osteogenesis imperfecta	EMQN/GenQA
Pathogenecity of germline sequence variant (classification)	GenQA
Pathogenecity of germline sequence variant (interpretation)	GenQA
Pathogenecity of germline sequence variant (interpretation and classification)	GenQA
Pathogenicity of germline postnatal copy number variants (CNV) (Classification)	GenQA
Pathogenicity of prenatal copy number variants (CNV) (Classification)	GenQA
Pharmacogenetic panel	EMQN/GenQA
Phenylketonuria	EMQN
Polyposis syndromes (Familial Adenomatous Polyposis Colon Cancer; MUYTH associated polyposis)	EMQN
Postnatal constitutional CNV detection	EMQN/GenQA
Postnatal karyotyping	GenQA

Prader-Willi and Angelman Syndromes	EMQN
Pregnancy loss (Molecular methods)	GenQA
Preimplantation Genetic Diagnosis by NGS and/or Arrays for Trophectoderm and/or Blastomere Testing for Aneuploidies	GenQA
Preimplantation Genetic Testing of Blastomere/Trophectoderm for chromosomal rearrangement by NGS and/or arrays	GenQA
Preimplantation Genetic Testing for Blastomere FISH (Stage 1 & 2)	GenQA
Preimplantation Genetic Diagnosis (PGD-M) of trophectoderm and/or blastomere testing for monogenic disorders	GenQA
Prenatal constitutional CNV detection	GenQA
Prenatal karyotyping	GenQA
Primary Immunodeficiency disorders	GenQA
Rapid prenatal testing for common aneuploidies	GenQA
Rare neurological disorders	EMQN
Renal disorders	GenQA
Respiratory disorders	GenQA
Retinoblastoma	EMQN
RETT syndrome	CAP
RYR1-malignant hyperthermia	EMQN
Severe combined immuno deficiencies	EMQN
Severe developmental delay	GenQA
Skeletal dysplasis (including FGFR2 and FGFR3- related disorders)	GenQA
Spinal Muscular Atrophy	EMQN
SMA on bloodspots for newborn screening	GenQA
Spinocerebellar Ataxia's (SCA 1,2,3,6,7)	EMQN
Systemic Autoinflammatory Diseases former Hereditary Recurrent Fevers	EMQN
TPMT	INSTAND
Variant Validation	GenQA
Von Hippel Lindau Syndrome	EMQN
Wilson disease	EMQN
Y-Microdeletions syndromes	EMQN/GenQA