KWALITEIT BEWAKING IN DE BELGISCHE CENTRA VAN MENSELIJKE ERFELIJKHEID:

KWALITEITSCONTROLE VAN ZELDZAME ZIEKTEN GENETISCHE TESTS

Lijst van de financierbare externe kwaliteitsevaluaties (EKE)-2024

EKE	Leverancier(s)
Abnormal ultrasound	GenQA
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
Carrier screening	GenQA
Charcot-Marie-Tooth disease/HNPP	EMQN
Charcot Marie Tooth disease and related sensory and motor neuropathies	GenQA
Chromosome instability syndromes	GenQA
Classification of germline SNVs and indels	GenQA
Classification and Interpretation of germline SNVs and indels	GenQA
Congenital Adrenal Hyperplasia	EMQN
Cystic fibrosis	CF NETWORK
Cystic fibrosis and CFTR related disorders	GenQA
Developmental delay	GenQA

Differences in sex development	GenQA
DNA isolation & testing for FV other than leiden + HFE + FII	RfB
DNA Sequencing - NGS (v Germline CNV testing)	EMQN/GenQA
DNA Sequencing - NGS (v Germline SNVs and indels)	EMQN/GenQA
DNA Sequencing Sanger	EMQN
DPYD	RfB/CAP/GenQA/INSTAND/EMQN
Duchenne / Becker Muscular Dystrophy	EMQN
Eye disorders	GenQA
Epilepsy disorder	GenQA
Exome trio sequencing – prenatal	GenQA
Exome trio sequencing - postnatal	GenQA
Familial autosomal dominant hypercholesterolemia	EMQN
Familial Colorectal Cancer and Polyposis	GenQA
Familial endocrine tumour predisposition disorders	GenQA
Familial SHOX-related disorders	EMQN
Fragile X Syndrome	EMQN/GenQA
Friedreich Ataxia	EMQN
Gastroenterology and hepatology disorders	GenQA
Haemophilie (Genetics of Heritable Bleeding & Thrombotic Disorders)	UKNEQAS Blood Coagulation
Hereditary amyloidosis	EMQN
Hereditary Breast and Ovarian Cancer (HBOC)	EMQN/ GenQA
Hereditary Haemochromatosis	EMQN/INSTAND/ RfB
Hereditary Hearing loss	EMQN
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	GenQA
Infertility	GenQA
Inherited cancer sequencing panel (BRCA2, CDKN2A, MLH1, MSH2,MSH6, and PMS2)	Cap

Interpretation of RNA splicing variants	GenQA
Ophthalmological Disease (Inherited Retinal Disorders; IRD)	EMQN
ISCN	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
Microdeletion syndromes	GenQA
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
Newborn screening- Molecular testing for Cystic fibrosis on blood spots	GenQA
Newborn screening- Molecular testing for MCADD c.985A>G on blood spot	GenQA
Newborn screening- Molecular testing for SCID on blood spot	GenQA
Newborn screening- Molecular testing for Spinal Muscular Atrophy on blood spot	GenQA
Neurodegenerative disorders	GenQA

Neurofibromatosis and rasopathies	GenQA
NIPT for common aneuploidies	EMQN/GenQA
NIPT for common microdeletions	GenQA
Osteogenesis imperfecta	EMQN/GenQA
Pathogenicity of germline postnatal CNVs	GenQA
Pathogenicity of germline postnatal CNV (interpretation only)	GenQA
Pathogenicity of prenatal CNVs	GenQA
Pathogenicity of prenatal CNVs (interpretation only)	GenQA
Pathogenicity of RNA splicing variants	GenQA
Pharmacogenetic panel	EMQN/GenQA
Pharmacogenetic DPYD/UGT1A only	EMQN
Phenylketonuria	EMQN
Polyposis syndromes (Familial Adenomatous Polyposis Colon Cancer; MUYTH associated polyposis)	EMQN
Porphyrias	EMQN
Postnatal constitutional CNV detection	EMQN/GenQA
Postnatal karyotyping	GenQA
Prader-Willi and Angelman Syndromes	EMQN
Prediction of Clopidogrel effectiveness	GenQA
Pregnancy loss	GenQA
Preimplantation genetic testing for aneuploidies (PGT-A)	GenQA
Preimplantation genetic testing for structural rearrangements (PGT-SR)	GenQA
Preimplantation genetic testing (PGT) of blastomere (FISH)	GenQA
Preimplantation genetic testing for monogenic disorders (PGT-M)	GenQA
Prenatal constitutional CNV detection	GenQA
Prenatal karyotyping	GenQA
Primary Immunodeficiency disorders	GenQA
Rapid prenatal testing for common aneuploidies	GenQA
Rare neurological disorders	EMQN
Rare neuromuscular Disease	EMQN
Renal disorders	GenQA
Respiratory disorders	GenQA

Retinoblastoma	EMQN
RETT syndrome	CAP
RYR1-malignant hyperthermia	EMQN
Severe combined immuno deficiencies	EMQN
Skeletal dysplasis	GenQA
Spinal Muscular Atrophy	EMQN
Spinocerebellar Ataxia's (SCA 1,2,3,6,7)	EMQN
Stickler syndrome	EMQN
Systemic Autoinflammatory Diseases	EMQN
TPMT	INSTAND/GenQA
Variant Validation	GenQA
Von Hippel Lindau Syndrome	EMQN
Wilson disease	EMQN
Y-Microdeletions syndromes	EMQN/GenQA

Laatste wijziging: 15/02/2024