Parameter	EQA Provider
ABCB1 (MDR1) c.3435C>T	RfB
Acyl CoA DehydroGenease (ACADM) Gene	UKNEQAS
Adenomatous Polyposis of the Colon, familial (APC) - APC Gene	EMQN
AdrenoGenital syndrome (AGS)	EMQN Instand
Aicardi-Goutieres Syndrome 1 (AGS1)	EMQN
Aldolase B	INSTAND
alpha-1-Antitrypsin (AAT) Deficiency – AAT / SERPINA1 / a1-Proteinase Inhibitor Gene	RfB ECAT INSTAND CAP
Angiotensin I Converting Enzyme (ACE) Gene: Insertion/Deletion Polymorphism (rs4340)	RfB ECAT Instand
Angiotensinogen (AGT) Gene: M235T	INSTAND
Antithrombin III Deficiency (AT3D) - Antithrombin (AT3) / SERPINC1 Gene	INSTAND
Apolipoprotein B100 (ApoB100) Gene: R3500Q (rs5742904)	RfB ECAT INSTAND
Apolipoprotein E (ApoE) Gene: ApoE2, E3, E4; (variants Arg158Cys, Cys112Arg)	RfB CAP ECAT INSTAND EQUALIS Labquality Qualicont
AT3 Cambridge Typ I/II	RfB
beta-Fibrinogen (FGB) Gene: polymorphism G-455A	RfB ECAT Instand
Breast/ovarian cancer, familial - BRCA1 Gene, BRCA2 Gene	CAP EMQN Equalis INSTAND
Butyrylcholinesterase (BCHE) Gene: (A)-Variant (N70G), K-variant (A539T)	Instand RfB
Canavan Disease - Aspartoacylase (ASPA) Gene	CAP
CCR-del-32bp	RfB
Charcot-Marie-Tooth Disease, type 1A (CMT1A)	EMQN
Cholesteryl ester transfer protein / CETP-Gene	RfB ECAT
Connexin 26	CAP INSTAND
Coumarin/Warfarin Sensitivity - Cyp2C9 Gene, VKORC1 Gene	INSTAND RfB CAP ECAT EQUALIS Labquality

Cystic Fibrosis transmembrane conductance regulator (CFTR) Gene	CAP CF Network INSTAND UKNEQAS EMQN SKLM
Cytochrom p450 2B6*6	RfB
Cytochrom p450 2C19	RfB CAP INSTAND SKLM
Cytochrom p450 2C8 (CYP2C8) Gene: K399R	RfB ECAT
Cytochrom p450 2C9	RfB CAP INSTAND
Cytochrom p450 2D6	RfB CAP INSTAND SKLM
Cytochrom p450 3A4*22	RfB CAP
Cytochrom p450 3A5*3	RfB CAP
CYP21A2 21-Hydroxylase	INSTAND
Dihydropyrimidin-Dehydrogenase (DPD) Gene: Exon-skipping mutation IVS14 G>A +1, DPD*13, DPD D949V (rs67376798)	CAP RfB ECAT SKLM
DNA Isolation	RfB ECAT
DNA Sequencing	RfB CAP ECAT EMQN EQUALIS
Duchenne/Becker- Muscular Dystrophy (DMD/BMD) - Dystrophin (DMD) Gene	CAP EMQN INSTAND
Factor II (Prothrombin) Gene: G20210A polymorphism	CAP Controllab RfB ECAT EQUALIS INSTAND Labquality RCPA SKML QAP Qualicont
Factor IX Gene	Instand
Factor V Gene: DNA Isolation and Genotyping	RfB ECAT
Factor V Gene: Hongkong FV-Hong-Kong (ARG306GLY)	RfB ECAT

Factor V Gene: FV Cambridge (ARG306THR)	RfB ECAT
Factor V Gene: H1299R	RfB ECAT
Factor V Gene: Leiden Mutation G1691A (Arg506Gln) (rs6025)	CAP Controllab ECAT EQUALIS INSTAND Labquality SKLM RCPA RfB Qualicont QAP
Factor VII activating Protease (FSAP)- Marburg I polymorphism	RfB ECAT
Factor VII Gene: R353Q	Instand RfB
Factor XII Gene: C46T polymorphism (rs1801020)	RfB ECAT
Factor XIII, Subunit A (F13A1) Gene: Val34Leu polymorphism (rs5985)	RfB ECAT Instand
Familial dysautonomia	CAP
Fanconi Anaemia	CAP
Fibrinogen receptor HPA 1a/1b	Instand
Follicle-Stimulating Hormone Receptor (FSHR) Gene	Instand
Fragile X-Syndrome - FMR1 Gene	CAP EMQN INSTAND
Friedreich ataxia	CAP
Frataxin (FXN) Gene	CAP EMQN
Fructose intolerance – Aldolase B (ALDOB) Gene; A149P, A174D, N334K	RfB ECAT
Gaucher	CAP
Glycogen storage disease type IA	CAP
Gastrointestinal stromal tumor (GIST) – KIT-OnkoGene, Platelet derived growth factor receptor alpha (PGDFRA) Gene	CAP UKNEQAS
Gilbert (Meulengracht) Syndrome/UDP-Glycosyltransferase 1 Polypeptid A1 (UGT-1A1) Gene: UGT1A1*28	RfB ECAT INSTAND CAP
Glycoprotein IIbIIIa (GPIIbIIIa) Deficiency/Fibrinogenrezeptor Deficiency	RfB ECAT Instand
Glykoprotein IaIIa (GPIaIIa) Deficiency /Kollagenrezeptor Deficiency	RfB ECAT Instand
Growth Hormone Receptor (GH-R) Gene: del Exon3	Instand

Haemoglobinopathies	UKNEQAS CAP Equalis
Haemophilia	UKNEQAS
Hämochromatosis, hereditary – HFE-Gene	CAP RfB ECAT EQUALIS INSTAND Labquality EMQN RCPA SKML QAP Qualicont UKNEQAS
Hereditary Deafness (DFNB1)	CAP EMQN
Hereditary Nonpolyposis Colorectal Cancer (HNPCC)	CAP EMQN UKNEQAS INSTAND
Hereditary Recurrent Fevers (HRF)	EMQN ISSAID
HLA-Typification Class I and II	CAP INSTAND
HLA B*1502	CAP
HLA B27	CAP INSTAND SKML
HLA B*5701	RfB CAP
HLA DQ	INSTAND SKML
Huntington Disease (HD) / Chorea Huntington	CAP EMQN INSTAND
Interleukin 28B (IL-28B) Gene: C/T polymorphism	Instand RfB CAP
Interleukin 6	RfB
KRAS Oncogene	Instand CAP KRAS-EQA QUIP RfB UNKEQAS
Lactose-Intolerance – MCM6 Gene: MCM6 IVS13 C/T (=LCT C-13910T; rs4988235)	RfB ECAT EQUALIS INSTAND Labquality SKML

Major Histocompatibility Complex (MHC) Class I & II	CAP Instand UKNQAS
Maturity onset diabetes of the young (MODY)	EMQN
Methylentetrahydrofolat Reduktase (MTHFR) Deficiency	CAP Controllab RfB ECAT Instand Labquality RCPA QAP Qualicont
MHC Class I B (HLA-B): HBLA-B*5701	Instand UKNEQAS
MHC Class I B (HLA-B): HBLA-B*5701 determination	Instand UKNEQAS RfB
Minimal Residual Disease	CAP
Mitochondrial Diseases – Polymerase, DNA, Gamma (POLG) Gene	UKNEQAS CAP
Molecular Hematologic Oncology	CAP RCPA QAP UKNEQAS
Morbus Wilson / Wilson Disease - ATPase Cu(2+)- Transporting beta Polypeptide (ATP7B) Gene	RfB ECAT INSTAND EQUALIS EMQN
Multiple endokrine Neoplasia Typ 2 (MEN2) – RET Proto-Oncogene	CAP EMQN INSTAND
Myotonic dystrophy type 1 (DM1)	CAP EMQN
Neuropathy, Hereditary Sensory and Autonomic, Type III (HSAN3)	CAP
Niemann-Pick type A/B	CAP
Nonsmall Cell Lung Cancer (NSCLC) - Epidermal Growth Factor Receptor (EGFR)	CAP EMQN QuIP UKNEQAS
Nucleotide-binding oligomerisation domain Protein 2 (NOD2) Gene: R702W, G908R, L1007fsinsC	RfB ECAT Instand
Osteoporosis - Typ1 Collagen alpha 1 (COL1A1) Gene: SP1 G/T	RfB ECAT Instand
Osteoporosis – Vitamin D Receptor (VDR) Gene: BsmI B/b	RfB ECAT Instand
Parentage Testing	CAP

Phenylketonuria – Phenylalanin Hydroxylase (PAH) Gene Poor drug metabolism, Cyp2C19	EMQN RfB ECAT EQUALIS Labquality
Plasminogen activator inhibitor-1 (PAI1)	RfB ECAT INSTAND CAP
Poor drug metabolism, Cyp2D6-related	RfB ECAT EQUALIS CAP Labquality
Poor drug metabolism, UGT-1A6-linked	Instand CAP
Porphyria variegata (POR) – Protoporphyrinogen Oxidase (PPOX) Gene	EMQN
Prader-Willi-Syndrom / Angelman-Syndrome (PWS/AS)	CAP EMQN INSTAND
Prion Protein (PRNP) Gene: M129V	INSTAND
Protein C deficiency	INSTAND
Protein S deficiency	INSTAND
Ret Proto-Onkogen, Exon 10-16	INSTAND
Retinoblastoma (RB1) – RB1 Gene	EMQN
RETT (*MECP2*) genotyping	CAP
Rheumatoid arthritis, systemic juvenile, susceptibility to - Interleukin 6 (IL-6) Gene: G-174C	INSTAND
Short stature Gene testing – Short stature homeobox (SHOX) Gene	EMQN
Sickle cell anemia	CAP Instand
SLCO1B1 (rs12979860)	CAP
Spinal muscular atrophy (SMA), Type 1, 2, 3, 4 – Survival of Motor Neuron 1 (SMN1) Gene	CAP EMQN INSTAND
Spinocerebellar ataxias (SCA) – SCA 1-7 Gene	CAP EMQN
Spondyloarthropathy 1 (SPDA1) / Spondylitis ankylosans / Morbus Bechterew – MHC Class I B (HLA-B): HLA B27 Typing	Instand UKNEQAS CAP RfB
Tay Sachs Disease (TSD) - Hexosaminidase A (HEXA) Gene	CAP
Thalassaemia alpha / Mental Retardation Syndrom	CAP RCPA SKML QAP
Thalassaemia beta	CAP RCPA

	SKML QAP
Thiopurin-S-Methyltransferase (TPMT) Deficiency	RfB ECAT INSTAND CAP
Thrombophilia, Molecular Genetics of	UKNQAS
Tumor Nekrosis Factor alpha (TNFa) Gene: G-238A, G-308A	Instand RfB
Von Hippel-Lindau-Syndrom (VHL) – VHL Gene	EMQN
Von Willebrand Disease (VWD), Type 1, 2, 3	Instand
Y-Chromosome microdeletions / Azoospermia factor (AZF) regions	EMQN INSTAND
DNA Sequencing	EMQN RfB CAP
Cell-free DNA screening for fetal aneuploidy	GenQA CAP
Next-Generation Sequencing—Hematologic Malignancies	CAP
Next-Generation Sequencing Undiagnosed Disorders—Exome	CAP
Next-Generation Sequencing Bioinformatics	CAP
Cell-free Tumor DNA CFDNA	CAP RfB