

Service: Molecular Genetics

Service Summary: Tests by site

A current profile of disease and location for testing is provided below.

Key:

AB= Aberdeen	DUN = Dundee	ED = Edinburgh	GLA = Glasgow
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Disease	AB	DUN	ED	GLA	Service
Achondroplasia			X		Common mutations
Acral peeling skin syndrome		X			Sequence analysis of TGM5 gene
ADPKD		X			Microsatellite analysis
ARPKD		X			Sequence analysis of PKHD1
Alpha 1 antitrypsin deficiency			X	X	Z mutation only
Aneuploidy screening by QF PCR	X	X	X	X	QF-PCR, chr. 13,18,21, X & Y
Angelman syndrome				X	Inc. UBE3A mutations
ARVC	X				PKP2 mutations
BCIE (keratin)		X			Sequence analysis of K1 and K10 genes mgenes
Breast Cancer	X			X	BRCA 1 & 2 mutations
Cadasil				X	
Cerebral cavernous malformations		X			Sequence CCM genes; MLPA
CHARGE syndrome				X	CHD7 mutations
Choroideremia			X		Linkage only
Crigler-Najjar syndrome		X			Sequence UGT1 gene

Disease	AB	DUN	ED	GLA	Service
Cystic fibrosis (CF)	X	X	X	X	
Duchenne/ Becker muscular dystrophy (DMD/ BMD)				X	Inc. dystrophin mutations
Deafness (GJB2, GJB6)		X			Sequence GJB2; GJB6 deletions
Developmental delay	X	X	X	X	Common microdeletions
Di George syndrome (22q11 del)			X		microdeletions
Dentatorubral-pallidolusian atrophy (DRPLA)			X	X	
Dystonia - torsion	X	X			DYT1 deletion
Epidermolysis bullosa (keratin)		X			Sequence K5 and K14
EPPK		X			Sequence K1 and K9
Factor V Leiden	X	X		X	Specific mutation
Familial adenomatous polyposis (FAP)	X	X			Sequence APC and MYH; MLPA for deletions
Familial hypercholesterolaemia	X				LDLR sequence & specific mutations
Fibrodysplasia Ossificans Progressiva (FOP)				X	Specific mutation
Fragile X	X		X	X	Mutation test
Friedreichs ataxia			X	X	Mutation test
Gilbert syndrome		X			Common mutation in UGT1 promoter
GRA (hyperaldosteronism)	X				
Haemochromatosis	X	X		X	Common mutations
Hereditary liability to pressure palsy (HLPP)	X				Common mutations
Hereditary motor sensory neuropathy (HMSN, CMT)	X				MLPA, P0,PMP22,CX32 mutations
Hereditary Hemorrhagic Telangiectasia (HHT)			X		ENG, ALK1, SMAD4 mutations
Hereditary non-polyposis colorectal cancer (HNPCC)		X	X		MMR genes; MSI testing; MYH mutations
HOCM	X				TNNT2,MYH7 mutations
Huntington disease			X		Mutation test
Hypochondroplasia			X		Common mutations

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Hypokalemic periodic paralysis 1 (HOKPP1)		X			Common mutations in CACN1AS and SCN4a
IBS		X			Sequence K2 gene
Incontinentia pigmenti				X	Common del. & X inactivation
Ichthyosis; recessive congenital form		X			Sequence TGM1 and ALOX12
Ichthyosis vulgaris		X			Common FLG mutations
Lesch-Nyhan syndrome				X	HPRT mutations
Leber hereditary optic neuropathy (LHON)		X			Common mutations
Long QT syndrome	X				KCNQ1, KCNH2, SCN5A and other gene sequences
Marfan syndrome					Microsatellite markers
MCAD		X			Sequence ACADM gene
Medullary thyroid carcinoma		X			Common mutation in RET
Mitochondrial		X			Selected mutations
Methylenetetrahydrofolate reductase deficiency (MTHFR)				X	C677T mutation
Multiple endocrine neoplasia type 1		X			Sequence MEN1 gene
Multiple endocrine neoplasia type 2a/2b		X			Common mutations in RET
Myotonic dystrophy	X	X	X	X	Mutation test
Myotonic dystrophy type 2	X				Mutation test
Nephronophthisis (familial juvenile)				X	Common deletion NPH1
Neurofibromatosis (NF1)		X			Microsatellite markers
Oculopharyngeal muscular dystrophy (OPMD)		X			Repeat in OPMD gene; sequence 25 codons
Ornithine transcarbamylase deficiency (OTC)			X		OTC gene screen
Pachyonychia congenita		X			Confirm identified mutation
Pelizaeus Merzbacher		X			Sequence PLP1; GJA12; MLPA
Peutz Jeghers syndrome		X			Sequence STK11; MLPA
Phaeochromocytoma/ paraganglioma		X			Sequence SDHB/SDHC/SDHD; MLPA

Disease	AB	DUN	ED	GLA	Service
Prader Willi syndrome				X	
Prothrombin (factor II)	X	X		X	g.20210G->A mutation
Proximal symphalangism		X			Sequence NOG1; GDF5
Retinoblastoma		X			RB1 mutations; MLPA
Rett syndrome			X		MECP2 mutations
Russell-Silver syndrome (UPD7)			X		Linkage
Severe Myoclonic Epilepsy of Infancy (SMEI)				X	SCN1A mutations
Spinal bulbular muscular strophy (SBMA, Kennedy's)			X	X	Mutation test
Spinal muscular atrophy (SMA)			X		Common mutations, linkage
Spinocerebellar ataxia (SCA)			X		SCA1,2,3,6,7 & 8 mutations
Von Hippel-Lindau (VHL)		X			Sequence VHL; MLPA
X-inactivation				X	
X-linked mental retardation			X		Linkage, some mutation testing
Y deletion		X		X	Multiplex PCR to detect deletions; microsatellite markers