Oxford University Hospitals
NHS Foundation Trust

OXFORD MEDICAL GENETICS LABORATORIES

CURRENT DISEASE SERVICES – APRIL 2018

Please note that there is a 15% surcharge for Non-NHS patients.
Highly specialised services (HSS) are indicated.
Note: This test list is due to be replaced by a Nationally agreed genomic test directory in Autumn 2018.

DISEASE and Comments	Cost
Achondroplasia (*FGFR3*)	
Screen for common mutation	**£145**
Amyotrophic lateral sclerosis (ALS) / Motor neurone disease (MND)	
Direct mutation test – PCR & TP-PCR	**£220**
Direct mutation test – Southern blot	**£220**
Andersen-Tawil syndrome	
Testing for known mutations	**£170**
KCNJ2 Mutation screening	**£220**
Angelman syndrome (AS)	
Deletion and Methylation analysis	**£220**
Family studies for Uniparental disomy (*Parental samples are also required)*	**£450**
UBE3A mutation screen	**£500**
Testing for known mutations	**£170**
Apert syndrome	
Screen for common mutations	**£145**
Arrhythmogenic right ventricular cardiomyopathy (ARVC)	
Testing for known mutations	**£170**
NGS screen of candidate genes (9 gene panel) inc MLPA of *PKP2*	**£1,020**
Ataxia	
Testing for known mutations	**£145**
NGS screen of candidate genes (98 gene panel)	**£1,020**
Autosomal Dominant Hypocalcaemia (ADH)	
Testing for known mutations	**£170**
Mutation screening *CASR* (FHH1)	**£375**
Mutation Screening *GNA11* (FHH2)	**£375**
Beckwith-Wiedemann syndrome	
Deletion and methylation analysis	**£220**
Breast/Ovarian cancer (BRCA1/ BRCA2)	
Testing for known mutations	**£170**
BRCA1/BRCA2 Mutation screen (incl dosage)	**£600**
NGS screen of panel of candidate genes	**£1020**
Targetted analysis (e.g Ashkenazi screen)	**£220**
Brugada syndrome	
Testing for known mutations	**£170**
NGS screen of candidate genes (17 gene panel)	**£650**
Calcium Sensing Receptor (FHH1, ADH1, FIHP)	
Testing for known mutations	**£170**
Mutation screening	**£375**

Cancer (Bowel/breast/ovarian)	
Testing for known mutations	**£170**
BRCA1/BRCA2 Mutation screen	**£600**
NGS screen of candidate genes	**£1020**
See individual listings for other cancer syndromes	
Cardiomyopathy (see Familial dilated/hypertrophic cardiomyopathy)	
Carpenter syndrome *HSS funded service*	
Testing for known mutations	**£145**
Mutation screening of *RAB23*	**£450**
Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT)	
Testing for known mutations	**£170**
NGS screen of candidate genes (7 gene panel)	**£650**
Cleidocranial dysplasia *HSS funded service*	**£145**
Testing for known mutations	**£500**
Mutation screening (including dosage analysis)	**£220**
Dosage analysis only	
Congenital Myasthenic Syndromes *HSS funded service*	
Testing for known mutations	**£145**
Mutation screening of candidate genes:	
▪ *CHAT / COLQ / GFPT1*	**£550 per gene**
▪ *CHRNE / CHRNB1 / CHRND / CHRNG*	**£500 per gene**
▪ *CHRNA1 / RAPSN / DOK7 / DPAGT1*	**£450 per gene**
Craniofacial disorders *HSS funded service*	
Testing for known mutations	**£145**
Apert syndrome targeted testing	**£145**
Level 1 mutation screening of candidate genes & dosage analysis	**£450**
Level 2 mutation screening	**£450**
ERF mutation screen	**£450**
TCF12 mutation screen	**£550**
IL11RA mutation screen	**£500**
ZIC1 mutation screen	**£450**
SMAD6 mutation screen	**£**
Dosage analysis only	**£220**
Craniofrontonasal syndrome *HSS funded service*	
Testing for known mutations	**£145**
Mutation screening (including dosage analysis)	**£500**
Dosage analysis only	**£220**
Cystic Fibrosis (CF)	
Screen for common mutations	**£145**
Duchenne & Becker Muscular Dystrophy (DMD/BMD)	
Testing for known mutations	**£170**
Duplication and deletion screening	**£220**
Family studies by linkage	**£450**
Mutation screening of *DMD* gene by NGS	**£650**
Epilepsy	
WES screen of candidate genes (209 gene panel)	**£1300**
Testing for known mutations	**£170**

Familial Dilated Cardiomyopathy (DCM)	
Testing for known mutations	**£170**
NGS screen of candidate genes (36 gene panel)	**£1,020**
Familial adenomatous polyposis coli (FAP)	
Testing for known mutations	**£170**
Mutation screening (including dosage analysis)	**£650**
Familial Hypertrophic Cardiomyopathy (HCM)	
Testing for known mutations	**£170**
NGS screen of candidate genes (19 gene panel)	**POA**
Familial Hypocalciuric Hypercalcaemia (FHH)	
Testing for known mutations	**£170**
Mutation screening *CASR* (FHH1)	**£375**
Mutation screening *GNA11* (FHH2)	**£375**
Mutation Screening *AP2S1* (FHH3)	**£145**
Simultaneous Mutation Screening *CASR, AP2S1 & GNA11*	**£600**
Familial Isolated Pituitary adenoma	
Testing for known mutations	**£145**
Mutation screening *AIP*	**£220**
Familial Malignant Melanoma	
Testing for known mutations	**£170**
Mutation screening *CDKN2A & CDK4*	**£450**
***FLNA*-associated disorders – please see 'X-linked periventricular nodular heterotopia…'**	
Fragile X syndrome (FRAXA)	
Direct mutation test – PCR	**£145**
Direct mutation test – Asuragen	**£220**
Direct mutation test - Southern blot	**£220**
Frontonasal dysplasia testing - *ALX3*	
Mutation screening (including dosage analysis)	**£450**
***GDF5*-associated skeletal disorders**	
Testing for known mutations	**£145**
Mutation screening and dosage analysis	**£450**
***GLI3*-associated skeletal disorders Pallister-Hall syndrome, Grieg Cephalopolysyndactyly syndrome, Preaxial polydactyly type IV, Preaxial polydactyly type A1**	
Testing for known mutations	**£145**
Targeted *GLI3* screen (PHS)	**£450**
Mutation screening of *GLI3* and dosage analysis	**£600**
Dosage analysis only	**£220**
GLUT1 deficiency syndrome – *SLC2A1*	
Testing for known mutations	**£145**
Mutation screening (including dosage analysis)	**£500**
***GREM1* associated mixed polyposis**	
Testing for known mutation	**£220**

Hereditary Motor and Sensory neuropathies (X-linked CMT)

Testing for known mutations	**£170**
Mutation screening of *GJB1 (Cx32)*	**£220**

Hereditary papillary renal carcinoma

Testing for known mutations	**£170**
Mutation screening *MET*	**£220**

Huntington disease (HD)

Direct mutation test	**£220**
Exclusion testing by linkage (*family members are also required*)	**£450**

Hyperparathyroidism Jaw Tumour syndrome (&FIHP)
Proforma required

Testing for known mutations	**£170**
Mutation screening *CDC73* (incl dosage)	**£375**
Dosage analysis only	**£220**

Hyperparathyroidism

Testing for known mutations	**£170**
Mutation screening of candidate genes:-	
▪ 9 gene mutation screen (*MEN1, CASR, CDC73, RET, CDKN1B, CDKN2B, CDKN2C, CDKN1A, AP2S1* codon 15)	**£750**

Or individual genes:

▪ *MEN1* screen	**£375**
▪ *CDC73* screen	**£375**
▪ *CASR* screen	**£375**

Hypoparathyroidism

Testing for known mutations	**£170**
Mutation screening of candidate genes:-	
▪ 6 gene mutation screen (*CASR,PTH,GCM2,GATA3,AIRE,GNA11*)	**£850**

Or individual genes:

▪ *AIRE* screen	**£500**
▪ *GATA3* screen	**£450**
▪ *CASR* screen	**£375**
▪ *GCM2* screen	**£450**
▪ *PTH* screen	**£220**
▪ *GNA11* screen	**£375**

Hypochondroplasia (*FGFR3*)

Testing for known mutations	**£145**
Screen for common mutations	**£220**

Joubert syndrome and related disorders

Testing for known mutations	**£170**
NGS screen of candidate genes (29 gene panel) includes MLPA of *NPHP1*	**£900**

Juvenile polyposis syndrome (JPS)

Testing for known mutations	**£170**
Mutation screening and dosage analysis (*SMAD4* & *BMPR1A)*	**£600**

LADD syndrome/ ALSG

Testing for known mutations	**£145**
Mutation screening *(FGFR3 exon 13, FGFR2 exon 16 and FGF10 exons 1-3)* and dosage analysis	**£450**

Lebers Hereditary Optic Neuropathy (LHON)	
LHON screen	**£170**
Li-Fraumeni syndrome	
Testing for known mutations	**£170**
Mutation screening and dosage analysis *TP53*	**£450**
Long QT syndrome (LQT)	
Testing for known mutations	**£170**
NGS screen of candidate genes (16 gene panel)	**£650**
Lynch syndrome (previously HNPCC)	
Testing for known mutations	**£170**
Mutation screening and dosage (*MLH1*, *MSH2, MSH6* genes)	**£800**
Microsatellite Instability	**£220**
Dosage only	**£220**
MS-MMR (promoter methylation analysis)	**£220**
Mitochondrial diseases	
HSS funded service (except for LHON - see above)	
Testing for known mutations	**£145**
Testing for m.1555A>G only (HSS funded only if patient has hearing loss)	**£145**
Screen for common mitochondrial DNA mutations	**£220**
mtDNA whole sequencing (by NGS)	**£550**
mtDNA depletion analysis (muscle or liver tissue/DNA required)	**£170**
Mutation screening of nuclear genes associated with mtDNA maintenance disorders:-	
▪ *POLG* target screen (5 common mutations)	**£220**
▪ NGS screen of candidate genes (21-22 genes including those listed below)	**£1020**
▪ *DGUOK, MPV17, POLG & TRMU* sequencing	**£800**
▪ *POLG & TWNK (C10orf2)* sequencing	**£600**
▪ *SUCLA2 & SUCLG1* sequencing	**£600**
▪ *POLG* sequencing	**£550**
▪ *TK2 / TRMU* single gene sequencing	**£500 per gene**
▪ *DGUOK / MPV17 / POLG2 / RRM2B* single gene sequencing	**£450 per gene**
▪ *SLC25A4 (ANT1) / TWNK (C10orf2)* single gene sequencing	**£220 per gene**
▪ *OPA1* sequencing & dosage analysis (not all cases fulfil HSS funding requirements – call lab to discuss)	**£700**
Mutation screening of nuclear genes associated with Leigh syndrome and/or PDH deficiency:-	
▪ *SURF1* sequencing	**£450**
▪ *PDHA1* sequencing	**£500**
▪ PDH subunit sequencing (5 gene Sanger panel incl. *PDHA1*)	**£900**
▪ PDH regulatory genes & cofactors sequencing (14 gene Sanger panel)	**£1020**
▪ NGS screen of PDH subunits, regulatory genes & cofactors (19 genes)	**£1020**
Biochemical assay for PDH enzyme activity in fibroblasts	**£250**
MEN1 & FIHP & MEN4	
Testing for known mutations	**£170**
Mutation screening *MEN1* gene (incl dosage)	**£375**
Dosage analysis only, *MEN1* and/or CDKN1B	**£220**
Screen *CDKN1B* (MEN4) (excl dosage as same kit as MEN1)	**£170**
MEN2 & FMTC	
Testing for known mutations	**£170**
Mutation screening in *RET* exons 10 & 11	**£220**
Mutation screening in *RET* exons 5, 8, 10, 11, 13, 14, 15 & 16	**£450**

MYH polyposis

Test for 2 common mutations	**£220**
Testing for known mutations	**£170**

MHYRE syndrome

Testing for known/common mutation	**£145**

Myotonic dystrophy Type 1

Direct mutation test – PCR	**£220**
Direct mutation test – Southern blot	**£220**

NGS sequencing panels – see individual diseases for panel costs

Re-interrogation NGS – Unmasking NGS data plus Sanger	**£375**
Re-interrogation NGS – Unmasking NGS data no Sanger	**£220**
Variant reclassification letter	**£145**
Variant confirmation and interpretation	**£220**

***NOG*-associated skeletal disorders**

Testing for known mutations	**£145**
Mutation screening and dosage analysis	**£450**

Painful channelopathies/neuropathies

Testing for known mutations	**£170**
Mutation screening of candidate genes –	
▪ 5 gene screen *SCN9A, SCN10A, SCN11A, TRPA1, NAGLU*	**£650**

Parietal foramina & Boston-type craniosynostosis

HSS funded service

Testing for known mutations	**£145**
Mutation screening (*ALX4* and *MSX2)* and dosage analysis	**£450**
Dosage analysis only	**£220**

Pheochromocytoma (isolated)

Testing for known mutations	**£170**
Dosage analysis (familial Mutation)	**£220**
10 gene mutation screen (*VHL, SDHB, SDHC, SDHD, SDHA, TMEM127, MAX, SDHAF2, RET* ex 10&11, *FH*)	**£600**
Isolated pheo >45yrs – *TMEM127 &SDHB*	**£500**

Individual gene prices on request.

Polymerase Proofreading-Associated Polyposis (PPAP)
Colorectal cancer (*POLD1*/*POLE*)

Testing for known mutations	**£170**
Mutation screen	**£375**

Prader-Willi syndrome (PWS)

Deletion and methylation analysis	**£220**
Family studies for Uniparental disomy (*Parental samples are also required)*	**£450**

PTEN

PTEN mutation screening and dosage analysis	**£500**

Inherited Retinal Dystrophy (IRD)
N.B. All IRD prices are currently subsidised by an OUH Trust translational research grant

NGS screen of candidate genes:_	
RP and RP-like phenotypes (111 gene panel)	**£650**
Syndromic retinal dystrophies (84 gene panel)	**£650**
Macular phenotypes (17 gene panel), includes Stargardt disease	**£375**
Non-progressive "Stationary" conditions (17 gene panel)	**£500**
Congenital Stationary Night Blindness (16 gene panel)	**£500**
Optic nerve disease (11 gene panel)	**£375**
Optic nerve panel plus 3 LHON common mtDNA mutations	**£500**
Testing for known mutations	**£170**

Robinow syndrome (autosomal dominant)

Testing for known mutations	**£145**
WNT5A, DVL1 and DVL3 mutation screen and *WNT5A* dosage analysis	**£450**
DVL1 or *DVL3* targeted screen	**£220**

Robinow syndrome (recessive) & Brachydactyly type B1

Testing for known mutations	**£145**
Targeted *ROR2* mutation screen (BDB1)	**£220**
ROR2 mutation screening and dosage analysis	**£500**
Dosage analysis only	**£220**

Russell Silver syndrome

Deletion and methylation analysis	**£220**
Family studies for Uniparental disomy (*Parental samples are also required)*	**£450**

Succinate dehydrogenase (SDH) / Familial Paraganglioma Syndromes

Testing for known mutations	**£170**
SDHB/C/D Mutation screening	**£600**

Spinal and bulbar muscular atrophy

Direct mutation test	**£220**
Linkage analysis (*Family members are also required)*	**£450**

Spinal muscular atrophy

SMN1 dosage analysis	**£220**
Linkage analysis (*Parental samples are also required)*	**£450**

Synpolydactyly type II, Brachydactyly type D & type E

Sequencing of known mutations	**£145**
Dosage analysis of known mutations	**£220**
HOXD13 mutation screening and dosage analysis	**£450**

TP53

TP53 mutation screening and dosage analysis	**£450**

Treacher Collins syndrome types 1-3

Testing for known mutations	**£145**
TCOF1 mutation screening and dosage analysis	**£600**
POLR1C mutation screening	**£500**
POLR1D mutation screening	**£220**
POLR1C & D mutation screening and dosage analysis	**£500**
Dosage analysis only	**£220**

Timothy Syndrome	
Testing for known mutations	**£170**
Screen for known mutations in *CACNA1C*	**£220**
Uromodulin (*UMOD*) & *REN*	
Mutation screening	**£450**
UPD14 & 16	
Family studies for Uniparental disomy (*Parental samples are also required)*	**£450**
Von Hippel Lindau syndrome (VHL)	
Testing for known mutations	**£145**
Mutation screening and dosage analysis	**£450**
X-linked periventricular nodular heterotopia, Otopalatodigital syndromes types 1 & 2, Melnick-Needles syndrome, Frontometaphyseal dysplasia	
Testing for known mutations	
FLNA mutation screening (full gene)	**£145**
[Targeted screening may be available at reduced cost – call to discuss]	**£800**
ZRS regulatory region of *SHH*-associated skeletal disorders	
Testing for known mutations	**£145**
Mutation screening and dosage analysis	**£220**
Zygosity testing	
To aid in the diagnosis of medical conditions only	
Ideally, parental samples are also required	**£220**
Microsatellite analysis	