Oxford University Hospitals
NHS Foundation Trust

Oxford Genetics Laboratories
Price list for Rare Disease Services from April 2022

Please contact the laboratory for the prices of any tests not listed in this document.

Services with an R code are funded within the NHS Genomic Medicine Service for referrals from NHS England. Prices for referrals from elsewhere are given below or indicated as NHS Highly Specialised Services (HSS) funded.

Musculoskeletal services

Clinical indication / Test	NHS (not England)	Private
R24 - Achondroplasia (*FGFR3*) Screen for common mutation	£170	£195.50
R23 - Apert syndrome (*FGFR2*) *HSS funded service for UK* Screen for common mutations	HSS	£195.50
R99 – Common craniosynostosis syndromes *HSS funded service for UK* **Including: Craniofrontonasal syndrome (*EFNB1*)**		
R99.1 Craniosynostosis panel by Sanger sequencing (*FGFR1* exon 7, *FGFR2* exons 3, 5, 8, 10, 11, 14-18, *FGFR3* exons 7 &10, *TWIST1*, *EFNB1*, *TCF12* and *ERF*), including: R99.2 Dosage analysis for *TWIST, ERF, TCF12* and *EFNB* *[+]For private referrals, individual gene analysis may be available on request – contact lab for details)*	HSS	£862.50[+]
R99.2 Dosage analysis only	HSS	£253
R100 – Rare craniosynostosis syndromes *HSS funded service for UK* This panel test is available by trio WGS for English referrals, and by singleton WES for non-English referrals. *[+]For private referrals, individual gene analysis for IL11RA, ALX4, MSX2, POR, ZIC1, GLI3, RAB23 and FLNA may be available on request – contact lab for details).*	HSS	£977.50[+]
R415 - Cleidocranial dysplasia (*RUNX2* sequencing & dosage analysis)	£500	£575
R416 - Non-syndromic metopic/sagittal synostosis - *SMAD6* sequencing	HSS	£517.50
R104 – Skeletal Dysplasia: this panel is available by trio WGS for English referrals. Gene testing is available for the following disorders for non-English referrals:		
Boston-type craniosynostosis (*MSX2* targeted sequencing)	£170	£195.50
Brachydactyly type B1 (*ROR2* targeted sequencing)	£220	£253
Frontonasal dysplasia testing (*ALX1*, *ALX3* & *ALX4* sequencing & dosage	£500	£575
GDF5-associated disorders (*GDF5* sequencing & dosage)	£450	£517.50
GLI3-associated disorders (*GLI3* sequencing & dosage)	£600[#]	£690[#]
LADD syndrome (*FGF10* sequencing & dosage + *FGFR2*, *FGFR3* hotspots)	£450	£517.50
NOG-associated disorders (*NOG* sequencing & dosage)	£450	£517.50
Parietal foramina (*ALX4* & *MSX2*, sequencing & dosage analysis)	£450	£517.50
Robinow syndrome (*DVL1*, *DVL2*, *DVL3*, *ROR2*, *WNT5A* sequencing & *ROR2*, *WNT5A* dosage)	£600[#]	£690[#]
HOXD13-related disorders (*HOXD13* sequencing & dosage)	£450	£517.50
Treacher-Collins syndrome (*TCOF1, POLR1C, POLR1D* sequencing & dosage)	£750[#]	£862.5[#]
X-linked Otopalatodigital spectrum disorders (*FLNA* sequencing)	£750[#]	£862.5[#]
ZRS regulatory region of SHH-associated skeletal disorders (sequencing & dosage)	£220	£253
R382 – Hypochondroplasia. Sequencing of *FGFR3* hotspots	£220	£253
R25 – Thanatophoric dysplasia. Sequencing of *FGFR3* hotspots	£450	£517.50
R52 - Short stature - SHOX deficiency. Diagnostic testing by sequencing and MLPA	£450	£517.50
R390 - multiple exostoses. Diagnostic testing by sequencing and MLPA	£650	£747.50

[#]Targeted sequencing may be available at reduced cost – call to discuss

Non-Malignant Haematology

Clinical indication / Test	NHS (not England)	Private
R90 Bleeding and platelet disorders NGS screen for panel of genes associated with coagulation, bleeding, and platelet disorders.	£850	£977.50
Targeted or full gene screening and dosage analysis (if applicable) is available for these disorders:		
R117 - Factor VIII deficiency (Haemophilia A)		
R117.1 Test for *F8* intron 1 and intron 22 inversions R117.2 & R117.3 *F8* sequencing and MLPA	£170 £650	£195.50 £747.50
R118 - Factor IX deficiency (Haemophilia B) – sequencing and MLPA	£450	£517.50
R121 - Von Willebrand disease – sequencing and MLPA	£750	£862.50
R112 - Factor II deficiency - sequencing	£500	£575
R115 - Factor V deficiency - sequencing	£600	£690
R116 - Factor VII deficiency – sequencing and MLPA	£450	£517.50
R119 - Factor X deficiency – sequencing and MLPA	£450	£517.50
R120 - Factor XI deficiency - sequencing	£550	£632.50
R122 - Factor XIII deficiency - sequencing	£500	£575
R123 - Combined vitamin K-dependent clotting factor deficiency - sequencing	£550	£632.50
R124 - Combined Factor V and VIII deficiency – sequencing	£550	£632.50
These gene screens are also available for non-English referrals:		
PROC	£450	£517.50
PROS1	£500	£575
SERPINC1	£450	£517.50
FGA/FGB/FGG	£500	£575
MYH9/GP1BA/GP1BB	£500	£575
ITGA2B/ITGB3	£500	£575
R97 Thrombophilia with a likely monogenic cause NGS gene panel screen	£750	£862.50
Factor V Leiden and common Prothrombin mutation analysis	£60	£69
R96 Iron metabolism disorders – NOT common *HFE* mutations NGS gene panel screen	£750	£862.50
R95 Iron overload – hereditary haemochromatosis testing (*HFE* genotyping) Screen for the common *HFE* p.His63Asp and p.Cys282Tyr variants	£85	£97.75
R361 Haemoglobinopathy trait or carrier testing (single gene sequencing and MLPA	£375	£431.25
R362 Carrier testing for sickle cell disease	£170	£195.50
R93 Thalassaemia and other haemoglobinopathies (single gene sequencing and MLPA)	£375	£431.25
R94 HbSS variant targeted mutation testing	£170	£195.50
R92 – Rare anaemia - NGS gene panel screen	£850	£977.50
R91 Cytopenia – NOT Fanconi anaemia - NGS gene panel screen	£850	£977.50
R229 Confirmed Fanconi anaemia or Bloom syndrome – NGS gene panel screen	£750	£862.50
R258.2 Cytopenia – Fanconi breakage testing indicated – NGS gene panel screen (R91 & R229 panels combined) [R258.1 Breakage testing NOT undertaken in Oxford]	£850	£977.50
R259 Nijmegen breakage syndrome – *NBN* sequencing	£550	£632.50
R313 Neutropaenia consistent with ELANE mutations – *ELANE* sequencing	£450	£517.50
R338 Monitoring for G(M)CSF escape mutations – *CSF3R* sequencing	£550	£632.50
R405 Hereditary erythrocytosis – NGS gene panel screen	£750	£862.50
R406 Thrombocythaemia – NGS gene panel screen	£750	£862.50

Neurology

Clinical indication / Test	NHS (not England)	Private
R80 - Congenital Myasthenic Syndromes *HSS funded service for Scotland* NGS gene panel screen	£750	£862.50
R59 Epilepsy panel is provided by trio WGS for English referrals. The following test may be available for non-English referrals (contact lab to discuss): R59.4 *SLC2A1* MLPA	£220	£253
R87 Cerebral malformations – this panel is available by WGS for English referrals. The following test is available for non-NHSE referrals: X-linked periventricular nodular heterotopia - *FLNA* screening & dosage	£750	£862.50
R68.1 - Huntington disease Direct mutation test	£220	£253
R383.1 - Huntington disease Exclusion testing by linkage	£450	£517.50
R58 – Adult onset neurodegenerative disorders:		
R58.3 ALS and MND direct mutation test – PCR & TP-PCR R58.4 Adult onset neurodegenerative disorders panel is provided by WGS for English referrals (contact lab to discuss)	£220	£253
R66 – Paroxysmal central nervous system disorders NGS gene panel screen.	£750	£862.50
R73 – Duchenne or Becker muscular dystrophy:		
R73.1 *DMD* gene sequencing	£750	£862.50
R73.2 *DMD* gene dosage by MLPA	£450	£517.50
R378.1 Linkage testing in families	POA	POA
Myotonic dystrophy Type 1 – Direct mutation test – PCR	£220	£253
Myotonic dystrophy Type 2 – Direct mutation test – PCR	£220	£253

Cardiology

Clinical indication / Test	NHS (not England)	Private
R127 Long QT syndrome – NGS gene panel screen.	£750	£862.50
R128 Brugada syndrome and cardiac sodium channel disease – *SCN5A* screen only	£650	£747.50
R129 Catecholaminergic polymorphic VT (CPVT) – NGS gene panel screen.	£750	£862.50
Catecholaminergic polymorphic VT (CPVT) – Dosage analysis by MLPA	£220	£253
R130 Short QT syndrome – NGS gene panel screen.	£750	£862.50
R131 Hypertrophic cardiomyopathy - NGS gene panel screen.	£750	£862.50
R132 Dilated and arrhythmogenic cardiomyopathy - NGS gene panel screen.	£850	£977.50
R133 Arrhythmogenic right ventricular cardiomyopathy – NGS gene panel screen	£850	£977.50
R135 Paediatric or syndromic cardiomyopathy – This gene panel is provided by trio WGS for routine NHSE referrals (contact lab to discuss). NGS gene panel screen available for urgent cases and non NHSE referrals.	£1020	£1173
R138 Sudden unexplained death or survivors of a cardiac event- NGS gene panel screen.	£850	£977.50
R328 Progressive cardiac conduction disease – NGS gene panel screen.	£750	£862.50
Individual gene tests available for non-English referrals:		
Pan-cardiomyopathy (includes genes in R131, R132 and *TAZ*)	£850	£977.50
Andersen-Tawil syndrome – *KCNJ2* screen only	£220	£253
Timothy syndrome – *CACNA1C* sequencing of exons 2-5 only	£220	£253

Mitochondrial

HSS funded service for England and Scotland

Clinical indication / Test	NHS (not England)	Private
The following targeted tests are available and selected as clinically appropriate. Results are reported together on a single report: R64 MELAS or MIDD Diagnostic testing - m.3243A>G R65 Aminoglycoside exposure posing risk to hearing m.1555A>G (NB this is not HSS funded in all cases) R299 Possible mitochondrial disorder – mitochondrial DNA rearrangement testing R350 MERRF syndrome - m.8344A>G R351 NARP syndrome or maternally inherited Leigh syndrome - m.8993T>C/G R397 Maternally inherited cardiomyopathy - m.4300A>G	£220	£253
R300 Possible mitochondrial disorder – whole mitochondrial genome sequencing (High read depth NGS screen)	£750	£862.50
R301 Possible mitochondrial disorder –mitochondrial DNA depletion (muscle or liver tissue/DNA required)	£220	£253
R315 POLG-related disorder:		
R315.1 Targeted sequencing for 4 common pathogenic variants	£220	£253
R315.2 *POLG* sequencing	£550	£632.50
POLG Dosage analysis by MLPA	£220	£253
R316 Pyruvate dehydrogenase (PDH) deficiency - NGS gene panel screen	£750	£862.50
R317 Mitochondrial liver disease, including transient infantile liver failure - screen for *BCS1L, DGUOK, MPV17, POLG, TRMU, TWNK* -	£750	£862.50
R352 Mitochondrial DNA maintenance disorder - NGS gene panel screen	£750	£862.50
R353 Mitochondrial disorder with complex I deficiency - NGS gene panel screen	£850	£977.50
R354 Mitochondrial disorder with complex II deficiency - NGS gene panel screen	£750	£862.50
R355 Mitochondrial disorder with complex III deficiency - NGS gene panel screen	£750	£862.50
R356 Mitochondrial disorder with complex IV deficiency - NGS gene panel screen	£750	£862.50
R357 Mitochondrial disorder with complex V deficiency - NGS gene panel screen	£750	£862.50
R63 Possible mitochondrial disorder – nuclear genes - NGS gene panel screen	£1020	£1173
R394 Mitochondrial neurogastrointestinal encephalopathy – *TYMP* sequencing	£450	£517.50
R395 Thiamine metabolism dysfunction syndrome 2 - *SLC19A3* sequencing	£450	£517.50
R396 Mitochondrial complex V deficiency, *TMEM70* type - *TMEM70* sequencing	£220	£253
R42 Leber hereditary optic neuropathy:		
R42.1 – Targeted mutation testing	£170	£195.50
R42.2 – whole mitochondrial genome sequencing (High read depth NGS screen)	£750	£862.50
Biochemical assay for PDH enzyme activity in fibroblasts	£500	£575

Cancer

Clinical indication / Test	NHS (not England)	Private
R208 Inherited Breast (and Ovarian) Cancer NGS gene panel screen (incl. dosage)	£600	£690
R207 Inherited Ovarian Cancer panel – NGS gene panel screen (including dosage)	£750	£862.50
R209 Colorectal Cancer panel - NGS gene panel screen (including dosage)	£750	£862.50
R211 Inherited Polyposis panel - NGS gene panel screen (including dosage)	£750	£862.50
R210 Lynch syndrome / Inherited MMR deficiency:		
NGS panel, including dosage (*MLH1*, *MSH2*, *MSH6*, *PMS2*)	£750	£862.50
Microsatellite Instability	£220	£253
MS-MMR (promoter methylation analysis)	£220	£253
R226 Inherited parathyroid cancer - Single gene (*CDC73*) analysis (including dosage)	*£450*	£517.50
R414 *APC* associated polyposis/ Familial adenomatous polyposis coli (FAP) Mutation screening (including dosage analysis)	£600	£750
Familial Isolated Pituitary adenoma Mutation screening *AIP*	£375	£431.25
GREM1 associated mixed polyposis Testing for known mutation	£220	£253
Juvenile polyposis syndrome (JPS) Mutation screening and dosage analysis (*SMAD4* & *BMPR1A)*	£600	£690
Polymerase Proofreading-Associated Polyposis (PPAP) Colorectal cancer - Mutation screen - *POLD1*, *POLE*	£600	£690
The following tests are available to private patients:		
Li- Fraumeni (R216) - Mutation screen incl. dosage of *TP53*	N/A	£575
PTEN (R213) - Mutation screen incl. dosage	N/A	£575
Prostate cancer - Mutation screen by gene panel	N/A	£862.50
Inherited Pancreatic Cancer (R367) - Mutation screen by gene panel (including dosage)	N/A	£862.50
Von Hippel Lindau syndrome (R225) - Mutation screen incl. dosage of *VHL*	N/A	£517.50
Familial Malignant Melanoma (R254) - Mutation screening *BAP1*, *CDKN2A* & *CDK4*	N/A	£862.50
Inherited renal carcinoma (R224)	N/A	£862.50

Family and Miscellaneous tests

Clinical indication / Test	NHS (not England)	Private
R346 DNA extraction and storage	£50	£57.50
R242 Predictive test by sequencing	£170	£195.50
R240 Diagnostic test by sequencing	£170	£195.50
R375 Family follow-up testing by sequencing	£170	£195.50
Family test by dosage (MLPA)	£220	£253
Prenatal test including R321 maternal cell contamination test	£450	£517.50
R264 Zygosity test	£220	£253
NGS sequencing panels – reanalysis		
R387 Re-interrogation NGS – Unmasking NGS data plus Sanger	£375	£431.25
R387 Re-interrogation NGS – Unmasking NGS data no Sanger	£220	£253
Variant reclassification letter	£145	£166.75
R370 Variant confirmation and interpretation	£220	£253

Endocrinology

Clinical indication / Test	NHS (not England)	Private
R151 Familial hyperparathyroidism – NGS gene panel screen (including dosage)	£750	£862.50
R152 Hypocalciuric hypercalcaemia - NGS gene panel screen (including dosage)	£600	£690
R153 Familial hypoparathyroidism - NGS gene panel screen (including dosage)	£750	£862.50
R319 Calcium-sensing receptor phenotypes – Single gene sequencing *CASR*	£375	£431.25
R156 Carney complex - Single gene sequencing (*PRKAR1A*)	£450	£517.50
R217 Multiple endocrine neoplasia (Endocrine neoplasms) - NGS gene panel screen (including dosage)	£750	£862.50
R218 Multiple endocrine neoplasia type 2 - Single gene sequencing (*RET*)	£375	£431.25
R226 Inherited parathyroid cancer - Single gene sequencing and dosage (*CDC73*)	£450	£517.50
Other tests that may be available (contact lab)		
R223 Inherited phaeochromocytoma and paraganglioma	£750	£862.50
CDC73 sequencing (HPT-JT syndrome)	£450	£517.50
GCM2 codons 379_395 sequencing	£170	£195.50
AIRE sequencing	£500	£500
GATA3 sequencing	£450	£450
GCM2 sequencing	£450	£450
GNA11 sequencing	£375	£375
PTH sequencing	£375	£375
MEN1 sequencing & dosage	£450	£450
CDKN1B sequencing & dosage	£375	£375
CDKN1B sequencing only	£170	£195.50
Succinate dehydrogenase and familial paraganglioma syndromes SDHB/C/D seq)	£600	£690

Eyes

Clinical indication / Test	NHS (not England)	Private
R32 Retinal disorders – NGS gene panel screen.	£1020	£1173
R38 Aniridia - NGS gene panel screen	£750	£862.50
R39 Albinism or congenital nystagmus – NGS gene panel screen	£750	£862.50
R41 Optic neuropathy (HSS for Wolfram only) – NGS gene panel screen	£750	£862.50
R262 Corneal dystrophy - NGS gene panel screen	£750	£862.50
R42 Leber hereditary optic neuropathy R42.1 – Targeted mutation testing	£170	£195.50