Oxford University Hospitals
NHS Foundation Trust

Oxford Genetics Laboratories
Price List For Rare Disease Services from April 2021

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. Contracts for highly specialised services (HSS) will continue for relevant devolved nations until at least April 2021. Prices for other services are given below.

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 Seven gene 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* NGS screen for 55 gene panel. This test will be moving to WGS in phase 1 for English referrals, and is available 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[+]
Non-syndromic metopic/sagittal synostosis - *SMAD6* sequencing	HSS	£517.50
R104 – Skeletal Dysplasia: this panel will be available by WGS in phase 1 for English referrals. Gene testing is available for the following disorders in the interim and for non-English referrals. Panel testing via singleton WES may be available for urgent cases (contact laboratory to discuss):		
Boston-type craniosynostosis (*MSX2* targeted sequencing)	£170	£195.50
Brachydactyly type B1 (*ROR2* targeted sequencing)	£220	£253
Cleidocranial dysplasia (*RUNX2* sequencing & dosage analysis)	£500	£575
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 108 genes associated with coagulation, bleeding, and platelet disorders. This test is moving to WGS in phase 3 for English referrals.	£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 of small panel	£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 analysis of 20 genes associated with thrombotic disorders	£750	£862.50
Factor V Leiden and common Prothrombin mutation analysis	£60	£69
R96 Iron metabolism disorders – NOT common *HFE* mutations NGS analysis of 26 genes	£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
R229 (also R285.2) Confirmed Fanconi anaemia or Bloom syndrome – NGS screen for 18 gene panel.	£750	£862.50
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 screen for 79 gene panel. This test will be moving to WGS in phase 3 for English referrals.	£850	£977.50
R91 Cytopenia – NOT Fanconi anaemia - NGS screen for 56 gene panel. This test will be moving to WGS in phase 3 for English referrals	£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 analysis of 8 genes	£750	£862.50
R406 Thrombocythaemia – NGS analysis of 3 genes	£750	£862.50

Neurology

Clinical indication / Test	NHS (not England)	Private
R80 - Congenital Myasthenic Syndromes *HSS funded service for Scotland* NGS screen for 26 gene panel. This test is moving to WGS in phase 2 for English referrals.	£750	£862.50
R59 – Early onset or syndromic Epilepsy – In England this test will be moving to WGS in the first phase with analysis of 430 green genes on the panel. An interim singleton WES NGS screen for 337 genes is available for urgent referrals only.	£1020	£1173
R59.4 *SLC2A1* MLPA	£220	£253
R87 Cerebral malformations – this panel will be available by WGS in phase 1 for English referrals. The following test is available in the interim and 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:		
R58.1 NGS screen for 94 green genes on the panel. In England this test will be moving to WGS in phase 2.	£1020	£1173
Dementia slice (NGS sequencing and analysis – 30 genes)	£850	£977.50
Parkinson Disease and complex Parkinsonism slice (NGS sequencing and analysis – 23 genes)	£850	£977.50
Amyotrophic lateral sclerosis (ALS)/ Motor neuron disease (MND) slice (NGS sequencing and analysis – 20 genes)	£850	£977.50
R58.2 MLPA dosage analysis	£220	£253
R58.3 ALS and MND direct mutation test – PCR & TP-PCR	£220	£253
R58.3 ALS and MND direct mutation test – Southern blot	£220	£253
R66 – Paroxysmal central nervous system disorders NGS screen for 20 green genes on the panel. In England this test will be moving to WGS in phase 3	£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
R54 – Hereditary ataxia with onset in adulthood R54.1 Interim NGS screen for 98 green genes on the panel. In England this test will be moving to WGS in phase 1and will include 186 green genes	£850	£977.50
R55 Hereditary ataxia with onset in childhood & R84 Cerebellar anomalies: R55.1 Interim NGS screen for 98 genes. The full panel will be available by WGS in phase 1 for English referrals	£850	£977.50
Joubert syndrome slice (NGS sequencing and analysis – 28 genes)	£850	£977.50
Myotonic dystrophy Type 1 – Direct mutation test – PCR	£220	£253
Myotonic dystrophy Type 2 – Direct mutation test – PCR	£220	£253

Eyes

Clinical indication / Test	NHS (not England)	Private
R31 Bilateral congenital or childhood onset cataracts – NGS screen for 99 green genes on the panel. In England this test will be moving to WGS in phase 2.	£1020	£1173
COFS & Cockayne syndrome slice (NGS sequencing and analysis – 4 genes)	£750	£862.50
Lowe syndrome slice (NGS sequencing and analysis – 1 gene)	£650	£747.50
Nance-Horan syndrome slice (NGS sequencing and analysis – 1 gene)	£650	£747.50
Warburg Micro syndrome slice (NGS sequencing and analysis – 3 genes)	£750	£862.50
R32 Retinal disorders – NGS screen for 201 green genes on the panel. In England this test will be moving to WGS in phase 3.	£1020	£1173
Best disease slice (NGS sequencing and analysis – 1 gene)	£650	£747.50
Choroideremia slice (NGS sequencing and analysis – 1 gene)	£650	£747.50
FEVR slice (NGS sequencing and analysis – 5 genes)	£750	£862.50
Macular dystrophy slice (NGS sequencing and analysis – 7 genes)	£750	£862.50
Retinoschisis slice (NGS sequencing and analysis – 3 genes)	£750	£862.50
Usher syndrome slice (NGS sequencing and analysis – 13 genes)	£750	£862.50
CSNB slice (NGS sequencing and analysis – 16 genes)	£850	£977.50
Achromatopsia slice (NGS sequencing and analysis – 28 genes)	£850	£977.50
R36 Structural eye disease – NGS screen for 112 green genes on the panel. In England this test will be moving to WGS in phase 3.	£1020	£1173
CHARGE syndrome slice (NGS sequencing and analysis – 1 gene)	£650	£747.50
Glaucoma slice (NGS sequencing and analysis – 10 genes)	£750	£862.50
Isolated ectopia lentis slice (NGS sequencing and analysis – 2 genes)	£650	£747.50
Anophthalmia/microphthalmia slice (NGS sequencing and analysis – 34 genes)	£850	£977.50
Ocular coloboma slice (NGS sequencing and analysis – 21 genes)	£850	£977.50
R38 Aniridia - NGS screen for 4 green genes on the panel	£750	£862.50
R39 Albinism or congenital nystagmus – NGS screen for 23 green genes on the panel	£750	£862.50
R41 Optic neuropathy (HSS for Wolfram only) – NGS screen for 21 green genes on the panel	£750	£862.50
R262 Corneal dystrophy - NGS screen for 22 green genes on the panel	£750	£862.50
R42 Leber hereditary optic neuropathy R42.1 – Targeted mutation testing	£170	£195.50

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 mutations	£220	£253
R315.2 *POLG* sequencing	£550	£632.50
POLG Dosage analysis by MLPA	£220	£253
R316 Pyruvate dehydrogenase (PDH) deficiency - NGS screen for 24 genes on the panel	£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 screen for 23 genes on the panel. In England this test will be moving to WGS in phase 2.	£750	£862.50
R353 Mitochondrial disorder with complex I deficiency - NGS screen for 50 genes on the panel. In England this test will be moving to WGS in phase 2.	£850	£977.50
R354 Mitochondrial disorder with complex II deficiency - NGS screen for 8 genes on the panel. In England this test will be moving to WGS in phase 2	£750	£862.50
R355 Mitochondrial disorder with complex III deficiency - NGS screen for 15 genes on the panel. In England this test will be moving to WGS in phase 2.	£750	£862.50
R356 Mitochondrial disorder with complex IV deficiency - NGS screen for 40 genes on the panel. In England this test will be moving to WGS in phase 2.	£750	£862.50
R357 Mitochondrial disorder with complex V deficiency - NGS screen for 19 genes on the panel. In England this test will be moving to WGS in phase 2.	£750	£862.50
R63 Possible mitochondrial disorder – nuclear genes - NGS screen for 311 genes on the panel. In England this test will be moving to WGS in phase 2.	£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
Biochemical assay for PDH enzyme activity in fibroblasts	£500	£575

Cancer

Clinical indication / Test	NHS (not England)	Private
R208 Inherited Breast (and Ovarian) Cancer *BRCA1/BRCA2/PALB2* NGS mutation screen (incl. dosage)	£600	£690
R207 Inherited Ovarian Cancer panel – NGS screen (including dosage) for 8 green genes on the panel.	£750	£862.50
R209 Colorectal Cancer panel - NGS screen (including dosage) for 14 green genes on the panel.	£750	£862.50
R211 Inherited Polyposis panel - NGS screen (including dosage) for 14 green genes on the panel.	£750	£862.50
R210 Lynch syndrome / Inherited MMR deficiency:		
NGS panel, including dosage (*MLH1*, *MSH2*, *MSH6*, *PMS2*)	£750	£862.50
Microsatellite Instability (R210.1)	£220	£253
MS-MMR (promoter methylation analysis) (R210.4)	£220	£253
R226 Inherited parathyroid cancer - Single gene sequencing (*CDC73*)	*£450*	£517.50
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 small panel	N/A	£862.50
Inherited Pancreatic Cancer (R367) - Mutation screen by small panel (including dosage) for 3 green genes on the panel	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 – Screen for 6 green genes on the panel (*AP2S1, CASR, CDC73, CDKN1B, MEN1, RET*	£750	£862.50
R152 Hypocalciuric hypercalcaemia - Screen for 3 green genes on the panel (*AP2S1, CASR, GNA11*)	£600	£690
R153 Familial hypoparathyroidism - Screen for 7 green genes on the panel (*AIRE, CASR, GATA3, GCM2, GNA11, PTH, TBCE)*	£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) - Screen for 5 green genes on the panel *(AIP, CDC73, CDKN1B,MEN1,RET)*	£750	£862.50
R218 Multiple endocrine neoplasia type 2 - Single gene sequencing (*RET*)	£375	£431.25
R226 Inherited parathyroid cancer - Single gene sequencing (*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

Cardiology

Clinical indication / Test	NHS (not England)	Private
R127 Long QT syndrome – NGS screen for 10 green genes on the panel.	£750	£862.50
R128 Brugada syndrome – *SCN5A* screen only	£650	£747.50
R129 Catecholaminergic polymorphic VT (CPVT) – NGS screen for 6 green genes on the panel.	£750	£862.50
Catecholaminergic polymorphic VT (CPVT) – Dosage analysis by MLPA	£220	£253
R130 Short QT syndrome – NGS screen for 4 green genes on the panel.	£750	£862.50
R131 Hypertrophic cardiomyopathy – teen and adult - NGS screen for 21 green genes on the panel.	£750	£862.50
R132 Dilated cardiomyopathy – teen and adult - NGS screen for 32 green genes on the panel.	£850	£977.50
R133 Arrythmogenic cardiomyopathy – R133.1 NGS screen for 11 green genes on the panel. Current screen 9 genes, including dosage analysis of *PKP2* by MLPA	£850	£977.50
R135 Paediatric or syndromic cardiomyopathy - NGS screen for 143 green genes on the panel. In England this test will be moving to WGS in phase 3.	£1020	£1173
R138 Molecular autopsy (sudden cardiac death) - NGS screen for 53 green genes on the panel.	£850	£977.50
R328 Progressive cardiac conduction disease – NGS screen for 11 green genes on the panel.	£750	£862.50
Individual gene tests available in for non-English referrals:		
Andersen-Tawil syndrome – *KCNJ2* screen only	£220	£253
Timothy syndrome – *CACNA1C* sequencing of exons 2-5 only	£220	£253