Oxford University Hospitals
NHS Foundation Trust

Oxford Genetics Laboratories
Price list for Rare Disease Services from April 2024

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.

General Core

Clinical indication ID	Test ID	Clinical Indication	Target/Genes	Test Method	Standard Price	Private Price
R240 / R242	R240.1	Diagnostic testing for known mutation(s) & Predictive testing for known familial mutation(s)	Specific Target	Targeted variant testing - Sanger sequencing	£185.00	£212.75
R240 / R242	R242.1	Diagnostic testing for known mutation(s) & Predictive testing for known familial mutation(s)	Specific Target	Targeted variant testing - MLPA	£240.00	£276.00
R244	R244.1	Carrier testing for known familial mutation(s)	Specific Target	Targeted variant testing - Sanger sequencing	£185.00	£212.75
R321	R321.1	Maternal cell contamination testing	Genomewide	Identity testing	£485.00	£557.75
R370	R370.1	Validation test	Specific Target	Targeted variant testing	£240.00	£276.00
R375	R375.1	Family follow-up testing to aid variant interpretation	Specific Target	Targeted variant testing	£185.00	£212.75
R387	R387.1	Reanalysis of existing data - NGS / WGS screen	As per updated indication	Other	£405.00	£465.75
R442	R442.1	Variant re-interpretation	Specific Target	Targeted variant testing	£155.00	£178.25
R443	R443.1	Confirmation test	Specific Target	Targeted variant testing	£240.00	£276.00
R322	R322.1	Skin fibroblasts to be cultured and stored	No target identified at this stage	Other	£135.00	£155.25
R346	R346.1	DNA to be stored	No target identified at this stage	DNA extraction and storage	£55.00	£63.25
		Blood culture			£80.00	£92.00
		Admin supplementary charge			£45.00	£51.75
		Supplementary work up (e.g. Cytogenetic segregation analysis)			£55.00	£63.25

Cardiology

Clinical indication ID	Test ID	Clinical Indication	Target/Genes	Test Method	Standard Price	Private Price
R127	R127.1	Long QT syndrome	Long QT syndrome	Small panel	£805.00	£925.75
R128	R128.1	Brugada syndrome and cardiac sodium channel disease	Brugada syndrome	Small panel	£695.00	£799.25
R129	R129.1	Catecholaminergic polymorphic VT	Catecholaminergic polymorphic VT	Small panel	£805.00	£925.75
R130	R130.1	Short QT syndrome	Short QT syndrome	Small panel	£805.00	£925.75
R131	R131.1	Hypertrophic cardiomyopathy	Hypertrophic cardiomyopathy - teen and adult	WES or Medium Panel	£805.00	£925.75
R132	R132.1	Dilated and Arrhythmogenic cardiomyopathy	Dilated cardiomyopathy - teen and adult	WES or Medium Panel	£910.00	£1,046.50
R133	R133.1	Arrhythmogenic right ventricular cardiomyopathy	Arrhythmogenic cardiomyopathy	Small panel	£910.00	£1,046.50

R135	R135.2	Paediatric or syndromic cardiomyopathy	Cardiomyopathies - including childhood onset	WES	£1,095.00	£1,259.25
R138	R138.1	Sudden unexplained death or survivors of a cardiac event	Sudden cardiac death	WES or Medium Panel	£910.00	£1,046.50
R328	R328.1	Progressive cardiac conduction disease	Progressive cardiac conduction disease	WES or Small Panel	£805.00	£925.75

Developmental Disorders

Clinical indication ID	Test ID	Clinical Indication	Target/Genes	Test Method	Standard Price	Private Price
R137	R137.1	Congenital heart disease - microarray	Genomewide	Microarray	£320.00	£368.00
R184	R184.1	Cystic fibrosis diagnostic test	CFTR	Targeted variant testing	£240.00	£276.00
R184	R184.3	Cystic fibrosis diagnostic test	CFTR	MLPA or equivalent	£240.00	£276.00
R185	R185.1	Cystic fibrosis carrier testing	CFTR	Targeted variant testing		
R199	R199.1	Congenital anomalies of the kidney and urinary tract - familial	Genomewide	Microarray	£320.00	£368.00
R21	R21.1	Fetal anomalies with a likely genetic cause	Genomewide	Common aneuploidy testing	£145.00	£166.75
R22	R22.1	Fetus with a likely chromosomal abnormality	Genomewide	Common aneuploidy testing	£145.00	£166.75
R22	R22.2	Fetus with a likely chromosomal abnormality	Genomewide	Microarray	£320.00	£368.00
R26	R26.1	Likely common aneuploidy	Genomewide	Common aneuploidy testing	£145.00	£166.75
R265	R265.1	Chromosomal mosaicism - karyotype	Genomewide	Karyotype	£205.00	£235.75
R27	R27.2	Congenital malformation and dysmorphism syndromes - microarray and sequencing	Genomewide	Microarray	£320.00	£368.00
R27	R27.3	Congenital malformation and dysmorphism syndromes	Paediatric disorders (486)	WGS	N/A	N/A
R28	R28.1	Congenital malformation and dysmorphism syndromes – microarray only	Genomewide	Microarray	£320.00	£368.00
R29	R29.2	Intellectual disability – microarray and sequencing	Genomewide	Microarray	£320.00	£368.00
R29	R29.4	Intellectual disability – microarray and sequencing	Intellectual disability (285)	WGS	N/A	N/A
R297	R297.1	Possible structural chromosomal rearrangement - karyotype	As determined by indication	Karyotype	£205.00	£235.75
R298	R298.1	Possible structural or mosaic chromosomal abnormality - FISH	As determined by indication	FISH	£120.00	£138.00
R314	R314.1	Ambiguous genitalia presenting neonatally	Sex chromosomes	Common aneuploidy testing	£145.00	£166.75
R314	R314.2	Ambiguous genitalia presenting neonatally	Sex chromosomes	Karyotype	£205.00	£235.75
R318	R318.1	Recurrent miscarriage with products of conception available for testing	Genomewide	Common aneuploidy testing	£145.00	£166.75
R318	R318.2	Recurrent miscarriage with products of conception available for testing	Genomewide	Microarray	£320.00	£368.00
R343	R343.1	Chromosomal mosaicism - microarray	Genomewide	Microarray	£320.00	£368.00
R377	R377.1	Intellectual disability – microarray only	Genomewide	Microarray	£320.00	£368.00
R402	R402	Premature ovarian failure	Genomewide	Karyotype	£205.00	£235.75
R69	R69.3	Hypotonic infant	Genomewide	Microarray	£320.00	£368.00

R69	R69.5	Hypotonic infant	Hypotonic infant (490)	WGS	N/A	N/A
R69	R69.6	Hypotonic infant	Hypotonic infant (490)	Confirmatory STR testing	£240.00	£276.00
R89	R89.2	Ultra-rare and atypical monogenic disorders	Genomewide	Microarray	£320.00	£368.00
R89	R89.3	Ultra-rare and atypical monogenic disorders	Relevant panel(s) in PanelApp	WGS	N/A	N/A

Endocrinology

Clinical indication ID	Test ID	Clinical Indication	Target/Genes	Test Method	Standard Price	Private Price
R151	R151.1	Familial hyperparathyroidism or Hypocalciuric hypercalcaemia	Familial hyperparathyroidism (480) and Hypocalciuric hypercalcaemia (481)	Small panel	£805.00	£925.75
R153	R153.1	Familial hypoparathyroidism	Familial hypoparathyroidism (312)	Small panel	£805.00	£925.75
R156	R156.1	Carney complex	PRKAR1A	Single gene sequencing >=10 amplicons	£485.00	£557.75
R217	R217.1	Endocrine neoplasia	Endocrine neoplasms (648)	Small panel	£805.00	£925.75
R218	R218.1	Multiple endocrine neoplasia type 2	RET	Single gene sequencing >=10 amplicons	£405.00	£465.75
R223	R223.1	Inherited phaeochromocytoma and paraganglioma excluding NF1	Inherited phaeochromocytoma and paraganglioma excluding NF1 (649)	Small panel	£805.00	£925.75
R226	R226.1	Inherited parathyroid cancer	CDC73	Single gene sequencing >=10 amplicons	£485.00	£557.75
R319	R319.1	Calcium-sensing receptor phenotypes	CASR	Single gene sequencing >=10 amplicons	£405.00	£465.75

Familial Hypercholesterolaemia

Clinical indication ID	Test ID	Clinical Indication	Target/Genes	Test Method	Standard Price	Private Price
R134	R134.1	Familial hypercholesterolaemia	Familial hypercholesterolaemia – targeted panel (772)	Small panel	£385.00	£485.00
R134	R134.2	Familial hypercholesterolaemia	LDLR	MLPA or equivalent	£275.00	£316.25

Haematology (Non-Malignant)

Clinical indication ID	Test ID	Clinical Indication	Target/Genes	Test Method	Standard Price	Private Price
R112	R112.1	Factor II deficiency	F2	Single gene sequencing >=10 amplicons	£535.00	£615.25
R115	R115.1 / R115.2	Factor V deficiency	F5	Single gene sequencing & Dosage	£645.00	£741.75
R116	R116.1	Factor VII deficiency	F7	Single gene sequencing >=10 amplicons	£485.00	£557.75
R116	R116.2	Factor VII deficiency	F7	MLPA or equivalent	£240.00	£276.00
R117	R117.1	Factor VIII deficiency	F8	Targeted variant testing	£185.00	£212.75

R117	R117.2	Factor VIII deficiency	F8	Single gene sequencing >=10 amplicons	£695.00	£799.25
R117	R117.3	Factor VIII deficiency	F8	MLPA or equivalent	£240.00	£276.00
R118	R118.1	Factor IX deficiency	F9	Single gene sequencing >=10 amplicons	£485.00	£557.75
R118	R118.2	Factor IX deficiency	F9	MLPA or equivalent	£240.00	£276.00
R119	R119.1	Factor X deficiency	F10	Single gene sequencing <10 amplicons	£485.00	£557.75
R119	R119.2	Factor X deficiency	F10	MLPA or equivalent	£240.00	£276.00
R120	R120.1	Factor XI deficiency	F11	Single gene sequencing >=10 amplicons	£590.00	£678.50
R120	R120.2	Factor XI deficiency	F11	MLPA or equivalent	£240.00	£276.00
R121	R121.1	von Willebrand disease	VWF	Single gene sequencing >=10 amplicons	£805.00	£925.75
R121	R121.2	von Willebrand disease	VWF	MLPA or equivalent	£485.00	£557.75
R122	R122.1	Factor XIII deficiency	F13A1;F13B	Small panel	£535.00	£615.25
R123	R123.1	Combined vitamin K-dependent clotting factor deficiency	VKORC1;GGCX	Small panel	£590.00	£678.50
R124	R124.1	Combined factor V and VIII deficiency	Combined factor V and VIII deficiency (517)	Small panel	£590.00	£678.50
R229	R229.1	Confirmed Fanconi anaemia or Bloom syndrome - mutation testing	FANCA;FANCB;FANCD2;PALB2	Small panel & MLPA or equivalent	£805.00	£925.75
R258	R258.2	Cytopenia - Fanconi breakage testing indicated	Confirmed Fanconi anaemia or Bloom syndrome (508)	WES or Medium Panel & Exon level CNV detection by MLPA or equivalent	£910.00	£1,046.50
R259	R259.2	Nijmegen breakage syndrome	NBN	Single gene sequencing >=10 amplicons	£590.00	£678.50
R313	R313.1	Neutropaenia consistent with ELANE mutations	ELANE	Single gene sequencing <10 amplicons	£485.00	£557.75
R338	R338.1	Monitoring for G(M)CSF escape mutations	CSF3R	Single gene sequencing >=10 amplicons	£590.00	£678.50
R361	R361.1 / R361.2	Haemoglobinopathy trait or carrier testing	HBA1;HBB	Small panel & MLPA	£375.00	£431.25
R362	R362.1	Carrier testing for sickle cell disease	HbS variant	Targeted variant testing	£185.00	£212.75
R405	R405.1	Hereditary Erythrocytosis	Hereditary Erythrocytosis (157)	Small panel	£805.00	£925.75
R406	R406.1	Thrombocythaemia	Thrombocythaemia (945)	Small panel	£805.00	£925.75
R90	R90.1	Bleeding and platelet disorders	Bleeding and platelet disorders (545)	WES or Medium Panel	£910.00	£1,046.50
R91	R91.1	Cytopenia - NOT Fanconi anaemia	Cytopenia - NOT Fanconi anaemia (519)	WES or Medium Panel	£910.00	£1,046.50

R92	R92.1	Rare anaemia	HBA1; HBA2; HBG1; HBG2; HBB; RPL11;RPL35A;RPS17;RPS19;RPS26;RPL5;PKLR	Panel & MLPA or equivalent	£910.00	£1,046.50
R93	R93.1 / R93.2	Thalassaemia and other haemoglobinopathies	HBA1;HBB	Small panel & MLPA or equivalent	£405.00	£465.75
R94	R94.1	HbSS sickle cell anaemia	HbS variant	Targeted variant testing	£185.00	£212.75
R95	R95.1	Iron overload - hereditary haemochromatosis testing	HFE common variants	Targeted variant testing	£95.00	£109.25
R96	R96.1	Iron metabolism disorders - NOT common HFE mutations	Iron metabolism disorders (515)	Small panel	£805.00	£925.75
R97	R97.1	Thrombophilia with a likely monogenic cause	Thrombophilia (516)	WES or Small Panel	£805.00	£925.75
		Factor 5 Leiden and prothrombin			£75.00	£86.25

Inherited Cancer

Clinical indication ID	Test ID	Clinical Indication	Target/Genes	Test Method	Standard Price	Private Price
R207	R207.1	Inherited ovarian cancer (without breast cancer)	Inherited ovarian cancer (without breast cancer) (143)	Small panel	£805.00	£925.75
R208	R208.1	Inherited breast cancer and ovarian cancer	BRCA1;BRCA2;PALB2; ATM; CHEK2; RAD51C; RAD51D	Small panel	£805.00	£925.75
R210	R210.2	Inherited MMR deficiency (Lynch syndrome)	Inherited MMR deficiency (Lynch syndrome) (503)	Small panel	£805.00	£925.75
R210	R210.4	Inherited MMR deficiency (Lynch syndrome)	MLH1 hypermethylation	Methylation testing	£240.00	£276.00
R211	R211.1	Inherited polyposis and early onset colorectal cancer - germline testing	Inherited polyposis (504)	Small panel	£805.00	£925.75
R213	R213.1	PTEN Hamartoma Tumor Syndrome	PTEN	Single gene sequencing >=10 amplicons	N/A	£614.62
R216	R216.1	Li Fraumeni Syndrome	TP53; POT1	Small panel	N/A	£614.62
R224	R224.1	Inherited renal cancer	Inherited renal cancer (521)	Small panel	N/A	N/A
R225	R225.1	Von Hippel Lindau syndrome	VHL	Single gene sequencing >=10 amplicons	N/A	£553.16
R367	R367.1	Inherited pancreatic cancer	Inherited pancreatic cancer (524)	Small panel	N/A	£921.93
R404	R404.1	Testing of unaffected individuals for inherited cancer predisposition syndromes	As dictated by clinical indication	Single gene sequencing >=10 amplicons	POA	POA
R404	R404.3	Testing of unaffected individuals for inherited cancer predisposition syndromes	Relevant inherited cancer panel	Small panel	POA	POA
R414	R414.1	APC associated Polyposis	APC	Single gene sequencing >=10 amplicons	£645.00	£741.75
R430	R430.1	Inherited prostate cancer	BRCA1, BRCA2, MLH1, MSH2, MSH6, ATM, PALB2, CHEK2	Small panel	£805.00	£925.75
R444	R444.1	NICE approved PARP inhibitor treatment	Breast cancer: BRCA1; BRCA2; PALB2; RAD51C; RAD51D; ATM; CHEK2	Small panel	£645.00	£741.75

R444	R444.2	NICE approved PARP inhibitor treatment	Prostate cancer: BRCA1, BRCA2	Small panel	N/A	N/A

Mitochondrial

Clinical indication ID	Test ID	Clinical Indication	Target/Genes	Test Method	Standard Price	Private Price
R299	R299.1	Possible mitochondrial disorder - mitochondrial DNA rearrangement testing	Mitochondrial genome	Other	£240.00	£276.00
R300	R300.1	Possible mitochondrial disorder - whole mitochondrial genome sequencing	Mitochondrial genome	Other	£805.00	£925.75
R301	R301.1	Possible mitochondrial disorder - mitochondrial DNA depletion testing	Mitochondrial genome	Other	£240.00	£276.00
R315	R315.1	POLG-related disorder	Common POLG mutations	Targeted variant testing	£240.00	£276.00
R315	R315.2	POLG-related disorder	POLG	Single gene sequencing >=10 amplicons	£590.00	£678.50
R316	R316.1	Pyruvate dehydrogenase (PDH) deficiency	Pyruvate dehydrogenase (PDH) deficiency (531)	WES or Medium panel	£805.00	£925.75
R317	R317.1	Mitochondrial liver disease, including transient infantile liver failure	Mitochondrial liver disease (532)	Small panel	£805.00	£925.75
R350	R350.1	MERRF syndrome	Common MERRF mutations	Targeted variant testing	£240.00	£276.00
R351	R351.1	NARP syndrome or maternally inherited Leigh syndrome	MT-ATP6;MT-ND6	Small panel	£240.00	£276.00
R351	R351.2	NARP syndrome or maternally inherited Leigh syndrome	m.8993T>C/G	Targeted variant testing	£240.00	£276.00
R352	R352.1	Mitochondrial DNA maintenance disorder	Mitochondrial DNA maintenance disorder (533)	WES or Medium Panel	£805.00	£925.75
R353	R353.1	Mitochondrial disorder with complex I deficiency	Mitochondrial disorder with complex I deficiency (534)	WES or Medium Panel	£910.00	£1,046.50
R354	R354.1	Mitochondrial disorder with complex II deficiency	Mitochondrial disorder with complex II deficiency (535)	WES or Small Panel	£805.00	£925.75
R355	R355.1	Mitochondrial disorder with complex III deficiency	Mitochondrial disorder with complex III deficiency (536)	WES or Small Panel	£805.00	£925.75
R356	R356.1	Mitochondrial disorder with complex IV deficiency	Mitochondrial disorder with complex IV deficiency (537)	WES or Small Panel	£805.00	£925.75
R357	R357.1	Mitochondrial disorder with complex V deficiency	Mitochondrial disorder with complex V deficiency (538)	WES or Small Panel	£805.00	£925.75
R394	R394.1	Mitochondrial neurogastrointestinal encephalopathy	TYMP	Single gene sequencing >=10 amplicons	£485.00	£557.75
R395	R395.1	Thiamine metabolism dysfunction syndrome 2	SLC19A3	Single gene sequencing <10 amplicons	£485.00	£557.75
R396	R396.1	Mitochondrial Complex V deficiency, TMEM70 type	TMEM70	Single gene sequencing <10 amplicons	£240.00	£276.00
R397	R397.1	Maternally inherited cardiomyopathy	m.4300A>G	Targeted variant testing	£240.00	£276.00
R42	R42.1	Leber hereditary optic neuropathy	Three common LHON variants	Targeted variant testing	£185.00	£212.75
R42	R42.2	Leber hereditary optic neuropathy	Mitochondrial genome	Other	£805.00	£925.75

R63	R63.1	Possible mitochondrial disorder - nuclear genes	Possible mitochondrial disorder - nuclear genes (539)	WES or Large Panel	£1,095.00	£1,259.25
R64	R64.1	MELAS or MIDD	MTTL1 3243A>G	Targeted variant testing	£240.00	£276.00
R65	R65.1	Aminoglycoside exposure posing risk to hearing	MT-RNR1 1555A>G	Targeted variant testing	£240.00	£276.00
N/A	N/A	Biochemical assay for PDH enzyme activity in fibroblasts	Biochemical test		£590.00	£678.50

Musculoskeletal

Clinical indication ID	Test ID	Clinical Indication	Target/Genes	Test Method	Standard Price	Private Price
R100	R100.2	Rare syndromic craniosynostosis or isolated multisuture synostosis	Genomewide	Microarray		
R100	R100.3	Rare syndromic craniosynostosis or isolated multisuture synostosis	Craniosynostosis (168)	WGS	HSS	£1,044.85
R104	R104.3	Skeletal dysplasia	Skeletal dysplasia (309)	WGS	N/A	N/A
R104	R104.4	Skeletal dysplasia	Osteopetrosis (943)	WES or Large Panel	N/A	N/A
R23	R23.1	Apert syndrome	FGFR2 c.755 and c.758	Targeted variant testing	HSS	£212.75
R24	R24.1	Achondroplasia	FGFR3 c.1138	Targeted variant testing	£185.00	£212.75
R25	R25.1	Thanatophoric dysplasia	FGFR3	Sequencing of FGFR3 hotspots	£485.00	£557.75
R382	R382.1	Hypochondroplasia	FGFR3 c.1620	Targeted variant testing	£240.00	£276.00
R390	R390.1 / R390.2	Multiple exostoses	EXT1;EXT2	Small panel & MLPA	£645.00	£741.75
R415	R415.1 / R415.2	Cleidocranial Dysplasia	RUNX2	Single gene & MLPA	£590.00	£678.50
R416	R416.1	Syndromic and non syndromic craniosynostosis involving midline sutures	SMAD6	Single gene sequencing <10 amplicons	HSS	£553.16
R52	R52.1 / R52.2	Short stature - SHOX deficiency	SHOX	Single gene sequencing & MLPA	£485.00	£557.75
R99	R99.1	Common craniosynostosis syndromes	Common craniosynostosis syndromes (507)	Small panel	HSS	POA
R99	R99.2	Common craniosynostosis syndromes	Common craniosynostosis syndromes (507)	Exon level CNV detection by MLPA or equivalent	HSS	POA

Skeletal Dysplasia: Gene testing is available for the following disorders for non-English referrals:

R104	Boston-type craniosynostosis ()	MSX2	targeted sequencing	£185.00	£212.75
R104	Brachydactyly type B1	ROR2	targeted sequencing	£240.00	£276.00
R104	Frontonasal dysplasia testing	ALX1, ALX3 & ALX4	sequencing & dosage	£535.00	£615.25
R104	*GDF5*-associated disorders	GDF5	sequencing & dosage	£485.00	£557.75
R104 #	*GLI3*-associated disorders	GLI3	sequencing & dosage	£645.00	£741.75
R104	*NOG*-associated disorders	NOG	sequencing & dosage	£485.00	£557.75
R104	Parietal foramina	ALX4 & MSX2	sequencing & dosage analysis	£485.00	£557.75

R104 #	Robinow syndrome	DVL1, DVL2, DVL3, ROR2, WNT5A sequencing & ROR2, WNT5A dosage	DVL1, DVL2, DVL3, ROR2, WNT5A sequencing & ROR2, WNT5A dosage	£645.00	£741.75
R104	*HOXD13*-related disorders	HOXD13	sequencing & dosage	£485.00	£557.75
R104 #	Treacher-Collins syndrome	TCOF1, POLR1C, POLR1D	sequencing & dosage	£805.00	£925.75
R104 #	X-linked Otopalatodigital spectrum disorders	FLNA	sequencing	£805.00	£925.75
R104	ZRS regulatory region of SHH-associated skeletal disorders		sequencing & dosage	£240.00	£276.00

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

Neurology

Clinical indication ID	Test ID	Clinical Indication	Target/Genes	Test Method	Standard Price	Private Price	
R383	R383.1	Linkage testing for Huntington disease	HTT	Other	£485.00	£557.75	
R410	R410.1	Myotonic dystrophy type 2 (DM2)	CNBP (ZNF9) STR	STR testing	£240.00	£276.00	
R54	R54.3	Hereditary ataxia with onset in adulthood	Hereditary ataxia - adult onset (466)	WGS	N/A	N/A	
R55	R55.4	Hereditary ataxia with onset in childhood	Hereditary ataxia and cerebellar anomalies - childhood onset (488)	WGS	N/A	N/A	
R58	R58.4	Adult onset neurodegenerative disorder	Neurodegenerative disorders - adult onset (474)	WGS	N/A	N/A	
R58	R58.5	Adult onset neurodegenerative disorder	Neurodegenerative disorders - adult onset (474) STR	Confirmatory STR testing	£0.00	£0.00	
R59	R59.2	Early onset or syndromic epilepsy	Genomewide	Microarray	£320.00	£368.00	
R59	R59.3	Early onset or syndromic epilepsy	Epilepsy - early onset or syndromic (402)	WGS	N/A	N/A	
R59	R59.4	SLC2A1 MLPA			£240.00	£276.00	
R66	R66.1	Paroxysmal central nervous system disorders	Paroxysmal central nervous system disorders (541)	WES or Medium Panel	£805.00	£925.75	
R68	R68.1	Huntington disease	HTT STR	STR testing	£240.00	£276.00	
R72	R72.1	Myotonic dystrophy type 1	DMPK STR	STR testing	£240.00	£276.00	
R73	R73.1	Duchenne or Becker muscular dystrophy	DMD	Single gene sequencing >=10 amplicons	£805.00	£925.75	
R73	R73.2	Duchenne or Becker muscular dystrophy	DMD	MLPA or equivalent	£485.00	£557.75	
R80	R80.1	Congenital myaesthenic syndrome	Congenital myaesthenic syndrome (232)	WES or Medium Panel	£805.00	£925.75	Scotland is HSS
R80	R80.2	Congenital myaesthenic syndrome	Congenital myaesthenic syndrome (232)	Exon level CNV detection by MLPA or equivalent	£240.00	£276.00	Scotland is HSS
R83	R83.2	Arthrogryposis	Genomewide	Microarray	£320.00	£368.00	
R84	R84.2	Cerebellar anomalies	Genomewide	Microarray	£320.00	£368.00	
R86	R86.2	Hydrocephalus	Genomewide	Microarray	£320.00	£368.00	
R87	R87.2	Cerebral malformation	Genomewide	Microarray	£320.00	£368.00	
R87	R87.3	Cerebral malformation	Cerebral malformations (491)	WGS	N/A	N/A	
R88	R88.2	Severe microcephaly	Genomewide	Microarray	£320.00	£368.00	

Ophthalmology

Clinical indication ID	Test ID	Clinical Indication	Target/Genes	Test Method	Standard Price	Private Price
R262	R262.1	Corneal dystrophy	Corneal dystrophies (658)	WES or Medium panel	£805.00	£925.75
R31	R31.3	Bilateral congenital or childhood onset cataracts	Cataracts (230)	WGS	N/A	N/A
R32	R32.2	Retinal disorders	Retinal disorders (307)	WGS	£1,095.00	£1,259.25
R33	R33.1	Possible X-linked retinitis pigmentosa	RPGR exon ORF15	Targeted variant testing	N/A	N/A
R36	R36.2	Structural eye disease	Structural eye disease (509)	WGS	N/A	N/A
R38	R38.2	Sporadic aniridia	Aniridia (510)	Small panel	£805.00	£925.75
R39	R39.1	Albinism or congenital nystagmus	Albinism or congenital nystagmus (511)	WES or Medium panel	£805.00	£925.75
R39	R39.2	Albinism or congenital nystagmus	Albinism or congenital nystagmus (511)	Exon level CNV detection by MLPA or equivalent		
R41	R41.1	Optic neuropathy	Optic neuropathy (186)	WES or Medium panel	£805.00	£925.75
R41	R41.3	Optic neuropathy	Three common LHON variants	Targeted variant testing	£185.00	£212.75

Note: these prices are not applicable to work covered under the NHSE contract
N/A = not available
HSS = funded for all NHS patients by the higher specialist services, commissioned via NHSE.
POA = price on application