UKAS MEDICAL
7883

# Pricing 2018/9

Great Ormond Street Hospital for Children NHS
NHS Foundation Trust

North East Thames
Regional Genetics Service

Please note for any test that requires DNA Extraction there will be an additional £50 charge
* except FH & Free-Fetal/NIPD

*** NCG: Please contact the laboratory for information on patients eligible for NCG funding*

| For detailed information on testing provided please see our service pack: http://www.labs.gosh.nhs.uk/laboratory-services/genetics | For private patients, please contact the lab for prices on 0207 813 8186<br>For Next Generation Sequence Panel gene lists & clinical proformas please see our website http://www.labs.gosh.nhs.uk/laboratory-services/genetics/molecular-genetics-service | Reporting Time (calendar days) | NHS Price | NCG** | MIM No. |
|---|---|---|---|---|---|
| **Molecular Genetic Testing** | | | | | |
| **Sample Preparation** | | | | | |
| DNA Extraction, quantitation & storage | | | £50 | | |
| **General Tests** | | | | | |
| Targeted / familial test, up to 4 pathogenic variants | | 14 | £195 | | |
| Couple report, 1 or 2 known familial pathogenic variant(s) | | 14 | £390 | | |
| Exclusion of Maternal Cell Contamination (maternal & fetal sample required) | | 3 | £195 | | |
| Prenatal Diagnosis including exclusion of maternal cell contamination (by prior arrangement only) | | 3 | £390 | | |
| Predictive Test, single mutation | | 14 | £195 | | |
| X-Inactivation (2 samples) | | 28 | £195 | | |
| **Non Invasive Prenatal Diagnosis (NIPD)** | **Test** | | | | |
| Free fetal DNA gender analysis; price inc. sample extraction | Real Time PCR - SRY | 3 | £290 | | |
| Achondroplasia (ACH) NIPD | NIPD Targeted test: FGFR3 NGS Panel | 5 | £630 | | #100800 |
| Bespoke pre pregnancy NIPD design | Please contact Laboratory | 56 | £790 | | |
| Bespoke Non-Invasive Prenatal Diagnosis (NIPD) | Bespoke NIPD Prenatal Test (once design validated) | 5 | £630 | | |
| Craniosynostosis - Apert syndrome NIPD | NIPD Targeted test: FGFR2 NGS Panel | 5 | £630 | | #101200 |
| Craniosynostosis - Muenke syndrome NIPD | NIPD Targeted test: FGFR3 NGS Panel | 5 | £630 | | #602849 |
| Craniosynostosis - FGFR2 related | NIPD Targeted test: FGFR2 NGS Panel | 5 | £630 | | |
| Cystic fibrosis | NIPD Paternal CFTR mutation exclusion | 5 | £630 | | #219700 |
| Cystic fibrosis - Haplotype analysis | NIPD diagnosis by NGS relative haplotype dosage analysis | 5 | £1,260 | | #219700 |
| Hypochondroplasia (HCH) NIPD | NIPD Targeted test: FGFR3 NGS Panel | 5 | £630 | | #146000 |
| Thanatophoric Dysplasia (TD) NIPD | NIPD Targeted test: FGFR3 NGS Panel | 5 | £630 | | #187600 |
| Skeletal Dysplasia NIPD | NIPD Targeted test: FGFR3 NGS Panel | 5 | £630 | | |
| **Next Generation Sequencing Panels (NGS)** | **Test** | | | | |
| Bardet-Biedl Syndrome (BBS) | 21 Gene panel from Clinical Exome Analysis (GOSHome) | 112 | £935 | ✓ | #209900 |
| Batten Disease (CLN genes) | 14 Gene NGS Panel | 56 | £710 | | |
| Branchio-oto-renal syndrome (BOR) | 3 Gene NGS panel (EYA1, SIX1, SIX5) | 56 | £485 | | |
| Breast / Ovarian cancer BRCA1 & BRCA2 | 2 Gene NGS Panel | 56 | £485 | | |
| BRCA1/2 + additional Breast/Ovarian Cancer Susceptibility Genes | 2-8 Gene NGS Panel (Contact Lab) | 56 | £710 | | |
| Ciliopathies | Subpanel from Clinical Exome Analysed (Contact Lab) | 112 | £935 | | |
| Clinical Exome (GOSHome) | Phenotype specific bespoke virtual panel (up to 250 genes) Contact Lab | 112 | £935 | | |
| Craniosynostosis | Gene NGS Panel (FGFR1, FGFR2, FGFR3, TWIST, ERF, IL11RA, TCF12, EFNB1) | 56 | £710 | ✓ | |
| Dermatology | Subpanel from Clinical Exome Analysed (Contact Lab) | 112 | £935 | | |
| Epilepsy & Severe Delay (EIEE) panel | 82 Gene NGS panel | 112 | £825 | | |
| Familial Hypercholesterolaemia (FH) | 4 gene NGS panel (LDLR, APOB, PCSK9, LDLRAP) | 56 | £485 | | |
| Glass Syndrome | SATB2 gene sequencing by NGS | 56 | £485 | | |
| Hearing Loss Non Syndromic & Syndromic (Including Usher) | 108 Gene NGS panel | 112 | £880 | | |
| Hearing Loss Non Syndromic & Syndromic (Excluding Usher) | 101 Gene NGS panel | 112 | £880 | | |
| Mental retardation syndromes (Coffin Siris, Nicolaides Baraitser) | 5 Gene NGS Panel | 56 | £710 | | |
| Non Syndromic Hearing Loss | 81 Gene NGS panel | 112 | £825 | | |
| Oculome | Subpanel from Clinical Exome Analysed (Contact Lab) | 112 | £935 | | |
| Waardenburg syndrome | 6 Gene NGS Panel | 112 | £825 | | #193500 |
| Usher syndrome | 13 Gene NGS Panel | 112 | £825 | | #276900, #276901 |
| Primary Immunodeficiency (PID) NGS sequencing panel (TIGER) | 71 Gene NGS Panel | 112 | £825 | | |
| Renal Tubulopathy | 37 Gene Subpanel from Clinical Exome Analysed | 112 | £935 | | |
| Very early onset inflamatory bowel disease (VEOIBD) (TIGER) | 41 Gene NGS Panel | 112 | £825 | | |
| PID and VEOIBD (TIGER) | 80 Gene NGS Panel | 112 | £825 | | |
| **NGS Reanalysis / Single gene requests** | **Test** | | | | |
| Next Generation Sequencing -Data Reanalysis | NGS panel reanalysis to include additional gene(s) | 112 | £265 | | |
| NGS Panel, Single Gene Only (excluding clinical exome) | NGS panel. Analysis of a single gene. | 56 | £485 | | |

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| Testing by Disease | Test | Reporting Time (calendar days) | NHS Price | NCG** | MIM No. |
|---|---|---|---|---|---|
| Achondroplasia (ACH) | Targeted test: FGFR3 skeletal dysplasia 5 amplicons | 14 | £195 | | #100800 |
| Albrights Hereditary Osteodystrophy | 3 Gene NGS panel (GNAS, PRKAR1A, PDE4D) | 56 | £485 | | #103580 |
| Aminoglycoside induced deafness | Targeted test: m.1555A>G | 14 | £140 | | #580000 |
| Angelman Syndrome (AS) | Targeted test: MSPCR | 28 | £195 | | #105830 |
| | Linked Markers | 56 | £485 | | |
| Athebaskan-type SCID (radiation sensitive SCID) | Sanger sequence analysis: DCLRE1C (Artemis) | 56 | £485 | | #602450 |
| Autoimmune lymphoproliferative syndrome (ALPs) | Sanger sequence analysis: FAS (TNFRSF6) | 56 | £485 | | #601859 |
| Bardet-Biedl Syndrome (BBS) | 21 Gene panel from Clinical Exome Analysis (GOSHome) | 112 | £935 | ✓ | #209900 |
| Batten Disease (Infantile): NCL1 | Targeted common mutations: PPT1 | 28 | £195 | | #256730 |
| | Sanger sequence analysis: PPT1 | 56 | £485 | | |
| Batten Disease (Late-Infantile): NCL2 | Targeted common mutations: TPP1 | 14 | £140 | | #204500 |
| | Sanger sequence analysis: TPP1 | 56 | £485 | | |
| Batten Disease (Juvenile): NCL3 | Targeted common deletion: CLN3 | 14 | £140 | | #204200 |
| | Sanger sequence analysis: CLN3 | 56 | £485 | | |
| Batten disease (Variant late infantile) | Sanger sequence analysis: CLN5, CLN6, CLN7 & CLN8 | 56 | £710 | | #256731, #601780, #600143 |
| Batten Disease NGS Panel | 14 Gene NGS Panel | 56 | £710 | | |
| Breast Ovarian cancer BRCA1/2 | NGS panel sequencing BRCA1/2 | 56 | £485 | | #604370, #612555 |
| Branchio-oto-renal syndrome (BOR) | 3 Gene NGS panel (EYA1, SIX1, SIX5) | 56 | £485 | | |
| Carbamoyl phosphate synthetase deficiency (CPSI) | Linked Markers | 56 | £485 | | #237200 |
| Cartilage Hair Hypoplasia (CHH) | Sanger sequence analysis:RMRP | 56 | £195 | | #250250 |
| CFHR5 Nephropathy | Targeted test: Common Cypriot Duplication | 14 | £140 | | *608593 |
| Coffin Siris Syndrome | Gene NGS Panel (ARID1A, ARID1B, SMARCB1, SMARCA4) | 56 | £485 | | #601358 |
| Congenital Tufting Enteropathy | Sanger sequence analysis: EPCAM | 56 | £485 | | #613217 |
| Connexin 26 (GJB2) Hearing Loss | Sanger sequence analysis: GJB2 & Targeted test: GJB6 deletion | 56 | £195 | | #220290 |
| Craniosynostosis - Apert syndrome | FGFR2 Targeted test: p.Ser252Trp, p.Pro253Arg | 28 | £195 | ✓ | #101200 |
| Craniosynostosis | 8 Gene NGS Panel (FGFR1, FGFR2, FGFR3, TWIST, ERF, IL11RA, TCF12, EFNB1) | 56 | £710 | ✓ | |
| Cystic fibrosis | Targeted test: CF-EU2 Single Patient Report | 14 | £195 | | #219700 |
| | Targeted test: CF-EU2 Couple Report | 14 | £390 | | |
| Cystinosis | Targeted test: Common 57Kb Deletion | 14 | £140 | ✓ | #219800 |
| | Sanger sequence analysis: CTNS | 56 | £485 | | |
| EAST (SeSAME) syndrome | Sanger sequence analysis: KCNJ10 | 56 | £195 | | #612780 |
| EBV-associated autosomal lymphoproliferative syndrome | Sanger sequence analysis: ITK | 56 | £485 | | #613011 |
| Epilepsy & Severe Delay (EIEE) panel | 82 Gene NGS panel | 112 | £825 | | |
| Fabry disease NGS | NGS analysis: GLA | 56 | £485 | ✓ | #301500 |
| Familial Haemophagocytic Lymphohistiocytosis (FHL2) | Sanger sequence analysis: Perforin (PRF1) | 56 | £485 | ✓ | #603553 |
| Familial Hypercholesterolaemia (FH) | 4 gene NGS panel (LDLR, APOB, PCSK9, LDLRAP) | 56 | £485 | | #143890 #144010 #603776 |
| Fragile X (FRAXA) | FMR1 - Triplet Repeat PCR | 14 | £140 | | #309550 |
| | TP-PCR (Amplidex) | 28 | £195 | | |
| Gastrointestinal Defects & Immunodeficiency (GIDID) | Sanger sequence analysis: TTC7A | 56 | £710 | | #243150 |
| Gaucher Disease | Targeted Test: Common mutations | 28 | £485 | ✓ | #230800 #231000 #230900 |
| Glycogen Storage Disease type 1a (GSD1a) | Targeted test: 2 Common mutations | 28 | £195 | | #232200 |
| | Sanger sequence analysis: GSD1A | 56 | £485 | | |
| Glycogen Storage Disease type 2 - GSD2 (Pompe) | Targeted test: GAA c.-32-13T>G | 14 | £140 | ✓ | #232300 |
| | NGS analysis: GAA | 56 | £485 | ✓ | |
| Hyper IGM syndrome (X-Linked) | Sanger sequence analysis: CD40L | 56 | £485 | ✓ | #308230 |
| Hypochondroplasia (HCH) | Targeted test: FGFR3 skeletal dysplasia 5 amplicons | 28 | £195 | | #146000 |
| IL10-related infantile inflammatory bowel disease | Sanger sequence analysis: IL10, IL10RA, IL10RB | 56 | £485 | | #266600 |

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|---|---|---|---|---|---|
| Immunodeficiency 14 | Targeted Test: PIK3CD 3 amplicons | 56 | £485 | | #615513 |
| Interleukin 7 Receptor Alpha SCID | Sanger sequence analysis: IL7Ra | 56 | £485 | ✓ | #146661 |
| JAK3 deficient severe combined immunodeficiency | Sanger sequence analysis: JAK3 | 56 | £710 | ✓ | #600802 |
| Krabbe disease | Targeted test: GALC Common deletion | 14 | £140 | ✓ | #245200 |
| Long Chain Acyl-CoA deydrogenase Deficiency (LCHADD) | Targeted test: c.1528G>C common mutation | 14 | £140 | | #609016 |
| | Sanger sequence analysis HADHA, HADHB | 56 | £710 | | |
| Loeys Dietz Syndrome | 2 Gene NGS Panel (TGFBR1, TGFBR2) | 56 | £485 | | #610380 |
| Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD) | Targeted test: c.985A>G common mutation | 14 | £140 | | #201450 |
| | Sanger sequence analysis: ACADM | 56 | £485 | | |
| McCune Albright syndrome | Targeted NGS to detect mosaic GNAS pathogenic variants | 28 | £195 | | #174800 |
| Metachromatic Leukodystrophy | Targeted test: ARSA common mutations | 28 | £195 | ✓ | #250100 |
| | Sanger sequence analysis: ARSA | 56 | £485 | | |
| Mucopolysaccharidosis 1 - MPS1 (Hurler / Scheie syndrome) | Targeted Test: IDUA common mutations | 28 | £195 | ✓ | #607014 |
| | Sanger sequence analysis: IDUA | 56 | £485 | | |
| Mucopolysaccharidosis 2 - MPS2 (Hunter syndrome) | Targeted test: IDS common inversion | 14 | £195 | ✓ | #309900 |
| | Sanger sequence analysis and MLPA: IDS | 56 | £485 | | |
| Mucopolysaccharidosis 3A - MPS3A (Sanfilippo syndrome type A) | Sanger sequence analysis: SGSH | 56 | £485 | ✓ | #252900 |
| Mucopolysaccharidosis 3B - MPS3B (Sanfilippo syndrome type B) | Sanger sequence analysis: NAGLU | 56 | £485 | ✓ | #252920 |
| Nephronophthisis (Juvenile) - NPH1 | Targeted Test: Common deletion | 14 | £195 | | #256100 |
| Netherton syndrome | Sanger sequence analysis: SPINK5 | 56 | £710 | | #256500 |
| Nicolaides Baraitser Syndrome | 5 Gene NGS Panel including SMARCA2 | 56 | £485 | | #601358 |
| Non Syndromic Hearing Loss | NGS panel | 112 | £825 | | |
| Syndromic Hearing Loss (USHER or Waardenburg etc) | NGS panel | 112 | £825 | | |
| Hearing Loss Syndromic & Non Syndromic including Usher | NGS panel | 112 | £880 | | |
| Ornithine transcarbamylase deficiency (OTC) | Sanger sequence analysis and MLPA: OTC | 56 | £485 | | #311250 |
| Osteopetrosis (Autosomal Recessive) | Sanger sequence analysis: TCIRG1 | 56 | £485 | | #259700 |
| Pendred Syndrome | NGS Analysis: SLC26A4 | 56 | £485 | | #274600 |
| Popliteal Pterygium syndrome (PPS) | NGS Analysis: IRF6 | 56 | £485 | | #119500 |
| Prader Willi syndrome (PWS) | MSPCR | 28 | £195 | | #176270 |
| | Linked Markers | 56 | £485 | | |
| Primary Congenital Glaucoma | Sanger sequence analysis: CYP1B1 | 56 | £485 | | #231300 |
| Pyridoxine Dependent Epilepsy | Targeted Test: 2 common mutations | 28 | £195 | | #266100 |
| | Sanger sequence analysis: ALDH7A1 (Antiquitin) | 56 | £485 | | |
| RAG- SCID | Sanger sequence analysis: RAG1 & RAG2 | 56 | £485 | ✓ | #601457 |
| Rett syndrome | NGS Analysis: MECP2 | 56 | £485 | | #312750 |
| Rhabdoid Tumour Predisposition Syndrome | NGS analysis: SMARCB1 | 56 | £485 | | #609322 |
| Schindler Disease | Sanger sequence analysis: NAGA | 56 | £485 | | #609241 |
| Primary Immunodeficiency NGS sequencing panel (TIGER) | NGS Panel for severe combined immuno deficiencies | 112 | £825 | | |
| Very early onset inflamatory bowel disease (TIGER) | NGS Panel for VEO-IBD | 112 | £825 | | |
| Skeletal Dysplasia (FGFR3 Related) | Targeted test: FGFR3 skeletal dysplasia 5 amplicons | 28 | £195 | | |
| Steroid Resistant Nephrotic syndrome (NPHS2) | Sanger sequence analysis: Podocin | 56 | £485 | | #600995 |
| Pulmonary Surfactant Metabolism Dysfunction (Surfactant Deficiency) | 8 Gene NGS Panel (inc. SFTPB, SFTPC, ABCA3) | 56 | £710 | | |
| Thanatophoric Dysplasia (TD) | Targeted test: FGFR3 skeletal dysplasia 5 amplicons | 28 | £195 | | #187600 |
| Van der Woude syndrome | NGS Analysis: IRF6 | 56 | £485 | | #119300 |
| Waardenburg syndrome | 7 Gene NGS Panel | 56 | £825 | | #193500 |
| Wiskott Aldrich syndrome | Sanger sequence analysis: WAS | 56 | £485 | ✓ | #301000 |
| | Linked Markers | 56 | £485 | | |
| X-linked Agammaglobulinaemia (XLA) | Sanger sequence analysis: BTK | 56 | £710 | | #307200 |
| X-linked Deafness | NGS Analysis: POU3F4 | 56 | £485 | | #304400 |
| X-linked lymphoproliferative disease type 1 (XLP1) | Sanger sequence analysis: SH2D1A | 56 | £195 | ✓ | #308240 |
| X-linked lymphoproliferative disease type 2 (XLP2) | Sanger sequence analysis: XIAP | 56 | £485 | ✓ | #602450 |
| X-linked SCID (XSCID) | Sanger sequence analysis: IL2Rgamma chain | 56 | £485 | ✓ | #300400 |

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| **Cytogenetic Testing (we can provide shorter reporting times for particularly urgent samples for all of our cytogenetic tests when technically possible) Sample processing where stated includes DNA extraction.** | | | | | |
| **Prenatal** | | | | | |
| **Tissue type** | | | | | |
| Chorionic villi | Rapid QF-PCR testing (trisomy: 13,18,21 + sexing if appropriate) (including processing) | 3 | £160 | | |
| Chorionic villi | CVS SNP Microarray (including processing, QF- PCR) | 14 | £470 | | |
| Chorionic villi | CVS Karyotyping (including processing, QF- PCR and cell culture) | 14 | £470 | | |
| Amniotic fluid | Rapid QF-PCR testing (trisomy: 13,18,21 + sexing if appropriate) (including processing) | 3 | £105 | | |
| Amniotic fluid | AF Microarray (including processing, QF- PCR) | 14 | £415 | | |
| Amniotic fluid | AF Karyotyping (including processing, QF- PCR and cell culture) | 14 | £415 | | |
| **Postnatal Cytogenetic Testing** | | | | | |
| Blood | FISH (trisomy 13,18,21 screen or SRY screen) per screen | 3 | £160 | | |
| Blood | Targeted screen (30 cells) | 28 | £135 | | |
| Blood | Targeted screen (50 cells) | 28 | £160 | | |
| Blood | Targeted screen (100 cells) | 28 | £200 | | |
| Blood | Karyotype (including mosaicism screen if appropriate) | 28 | £210 | | |
| Blood/DNA | Microarray (including processing) | 28 | £315 | | |
| Blood/DNA | Trio microarray (for investigation of UPD) price per parent | 28 | £160 | | |
| **Miscarriage/Fetal Loss** | | | | | |
| POC (products of conception) | Microarray (including processing) | 28 | £470 | | |
| POC (products of conception) | Karyotype | 28 | £345 | | |
| POC (products of conception) | QF-PCR/MLPA aneuploidy combined test | 28 | £210 | | |
| **Follow up (other family members)** | | | | | |
| Blood/DNA | analysis by: FISH/qPCR per parental or sib sample (>one rec'd) | 28 | £210 | | |
| Blood/DNA | analysis by: FISH/qPCR per *singleton* parental or sib sample | 28 | £240 | | |
| Blood/DNA | targeted analysis by: microarray (per parental or sib sample) | - | £210 | | |
| *Proband sample for validation (no charge)* | | - | N/A | | |
| **FISH (including trisomy 13,1,21 screen/SRY screen)** | | | | | |
| Blood | Price per probe or probe kit (karyotyping not included) | 28 | £160 | | |
| Blood | Telomeric FISH screen (karyotyping not included) | 28 | £475 | | |
| **Miscellaneous** | | | | | |
| DNA extraction, quantitation and storage (any tissue) | | - | £50 | | |
| Cell culture and fixed cell storage only (blood samples) | | - | £50 | | |
| Culture of tissue samples (skin biopsy) - not including DNA extraction | | | £85 | | |
| Dissection and culture of tissue samples (other tissues) - not including DNA extraction | | | £105 | | |
| **Sendaways** | | | | | |
| Specified Breakage syndromes: Fanconi, Blooms, AT, Nijmegen | | - | POA | | |


| | | |
|---|---|---|
| *For queries relating to Molecular genetic tests please contact* | Sam Loughlin DipRCPath | 020 7762 6880 |
| *For queries relating to Cytogenetic tests please contact* | Deborah Morrogh FRCPath | 020 7813 8014 |
| *For queries relating to Pricing please contact* | | 020 7762 6869 |