Oxford Molecular Genetics Laboratory

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www.ouh.nhs.uk/geneticslab

INHERITED EYE DISEASE NGS SERVICES

INTRODUCTION

Next generation sequencing (NGS) phenotype panels are available for the following Inherited Eye Diseases:

Panel	No of Genes	Conditions	Cost*
Panel 1 RP and RP-like phenotypes	111	Retinitis pigmentosa (RP), cone rod dystrophy (CORD), Leber's congenital amaurosis (LCA), *ABCA4* retinopathies (recessive), Choroideremia, Goldmann Favre/Enhanced S-cone syndrome, X-linked retinoschisis, late-onset retinal degeneration (LORD), congenital stationary night blindness (CSNB) (including Oguchi), familial exudative vitreoretinopathy, Bietti crystalline corneoretinal dystrophy	£650
Panel 2 Syndromic retinal dystrophies	84	Abetalipoproteinemia, Alagille, Alstrom, Bardet-Biedl (BBS), McKusick-Kaufman, Batten disease, Bietti crystalline corneoretinal dystrophy, Gyrate atrophy, ciliopathy-related retinal degeneration, nephronophthisis, Norrie, Refsum, Senior-Loken, Usher syndrome, Zellweger, X-linked retinoschisis, Blau, Knobloch	£650
Panel 3 Macular phenotypes	17	Stargardt disease (recessive), Best disease, dominant drusen (Doyne honeycomb retinal dystrophy-DHRD/mallatia levantinese), Sorsby fundus dystrophy, Pattern dystrophy (including dominant Stargardt disease), macular degeneration with hypotrichosis	£375
Panel 4 Non-progressive conditions	17	Oculocutaneous albinism (OCA), Achromatopsia, Tritanopia	£500
Panel 5 Congenital stationary night blindness	16	Congenital stationary night blindness (CSNB) - including Oguchi	£500
Panel 6 Optic nerve disease	12	i) Optic atrophy (dominant and recessive), optic atrophy plus syndromic features, Wolfram syndrome, mitochondrial optic nerve disease. ii) Optic atrophy NGS panel plus analysis for the 3 common mtDNA LHON mutations: m.3460G>A, m.11778G>A & m.14484T>C	£375 £500

* N.B. OUH Trust translational research price for NHS referrals only

REFERRALS

- Referrals must be accompanied by the referral proforma (click link)
- Referrals are accepted from Consultant Ophthalmologists, Clinical Geneticists and Genetic Counsellors

TECHNICAL INFORMATION

- Samples are prepared using Agilent's Haloplex^{TM} Targeted Enrichment system followed by next generation sequencing on the Illumina MiSeq Personal Sequencer
- Regions of interest include all coding exons +/- 10bp at intron/exon boundaries.
- Average coverage for the total region of interest is 98% at 30x read depth.
- Multiplex amplification probe ligation analysis (MLPA) is available for *ABCA4*, *CHM* and *USH2A*.
- Sanger sequencing is used to confirm all putative pathogenic variants.

TARGET REPORTING TIMES

Diagnostic screens: 80 days

N.B. Details are correct for the date of printing only – last updated 30/10/2016

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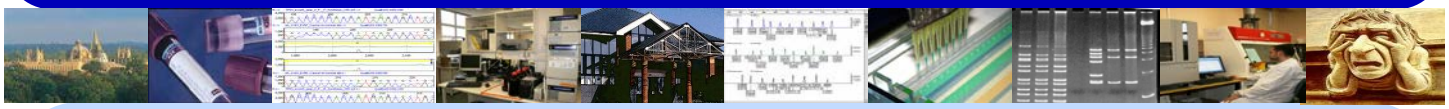

INHERITED EYE DISEASE NGS PHENOTYPE PANELS & GENES

INTRODUCTION

Next generation sequencing (NGS) phenotype panels include the following genes:

Panel 1 – RP and RP-like phenotypes – 111 genes
ABCA4, ABHD12, ADAM9, AIPL1, ARL6, BBS1, BEST1, C1QTNF5, C2ORF71, C8ORF37, CA4, CABP4, CACNA1F, CACNA2D4, CAPN5, CC2D2A, CDHR1, CEP290, CERKL, CHM, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CRB1, CRX, CYP4V2, DHDDS, EYS, FAM161A, FLVCR1, FSCN2, FZD4, GNAT1, GNPTG, GPR179, GRK1, GRM6, GUCA1A, GUCA1B, GUCY2D, IDH3B, IMPDH1, IMPG2, IQCB1, KCNJ13, KCNV2, KLHL7, LCA5, LRAT, LRIT3, LRP5, MAK, MERTK, NDP, NMNAT1, NR2E3, NRL, NYX, OFD1, OTX2, PDE6A, PDE6B, PDE6C, PDE6G, PDE6H, PITPNM3, POC1B, PRCD, PROM1, PRPF3, PRPF31, PRPF6, PRPF8, PRPH2, RAB28, RAX2, RBP3, RBP4, RD3, RDH5, RDH12, RGR, RHO, RIMS1, RLBP1, ROM1, RP1, RP2, RP9, RPE65, RPGR, RPGRIP1, RPGRIP1L, RS1, SAG, SEMA4A, SLC24A1, SNRNP200, SPATA7, TOPORS, TRPM1, TSPAN12, TTC8, TTPA, TULP1, UNC119, USH2A, WDR19, ZNF513

Panel 2 – Syndromic retinal degenerations – 83 genes
ABHD12, AHI1, ALMS1, APOB, ARL6, B9D1, B9D2, BBS1, BBS2, BBS4, BBS5, BBS7, BBS9, BBS10, BBS12, CC2D2A, CCDC28B, CDH23, CEP41, CEP164, CEP290, CIB2, CLRN1, COL18A1, CSPP1, CYP4V2, DFNB31, GLIS2, GPR98, HARS, INPP5E, INVS, IQCB1, JAG1, LZTFL1, MKKS, MKS1, MTTP, MYO7A, NDP, NEK8, NOD2, NOTCH2, NPHP1, NPHP3, NPHP4, OAT, OFD1, PCDH15, PDE6D, PDZD7, PEX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26, PHYH, POC1B, RPGRIP1L, RS1, SDCCAG8, TMEM67, TMEM138, TMEM216, TMEM231, TMEM237, TRIM32, TTC8, USH1C, USH1G, USH2A, WDPCP, WDR19, XPNPEP3, ZNF423

Panel 3 – Macular phenotypes – 17 genes
ABCA4, BEST1, C1QTNF5, CDH3, CNGB3, EFEMP1, ELOVL4, FSCN2, GUCA1A, GUCA1B, IMPG1, IMPG2, PROM1, PRPH2, RP1L1, RS1, TIMP3

N.B. For Stargardt (recessive) referrals:
Sanger sequencing will be undertaken for *ABCA4* gene regions not covered at a depth of 30x.
ABCA4 MLPA analysis will also be undertaken on any samples which do not have 2 pathogenic variants.

Panel 4 – Non-progressive conditions – 17 genes
C10ORF11, CNGA3, CNGB3, GNAT2, GPR143, LYST, MITF, MYO5A, OCA2, OPN1SW, PDE6C, PDE6H, RAB27A, SLC24A5, SLC45A2, TYR, TYRP1

Panel 5 – Congenital Stationary Night Blindness – 16 genes
CABP4, CACNA1F, CACNA2D4, GNAT1, GPR179, GRK1, GRM6, LRIT3, NYX, PDE6B, RBP4, RDH5, RHO, SAG, SLC24A1, TRPM1

Panel 6 – Optic nerve disease – 12 genes
ACO2, AFG3L2, C12ORF65, CISD2, MFN2, OPA1, OPA3, POLG, SLC25A46, SPG7, TMEM126A, WFS1

N.B. Analysis for the 3 common LHON mutations is also available for patients who have not had this pre-screen

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