Next Generation Sequencing Service at SDGS

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Testing Workflow

Library Preparation

SureSelect/Illumina

- Shearing of genomic DNA using the Covaris E220 sonicator.
- End repair, A tailing and ligation of adaptors using SureSelectXT library system (Agilent Technologies).
- Enrichment by SureSelect target enrichment (Agilent Technologies) using custom in house designed probes. Samples have barcode tags added following target enrichment.
- Sequencing on the Illumina HiSeq using the HiSeq Rapid SBS Kit v2 performing 2 x 108 bp paired end reads.

SureSelect/Illumina lab work performed in Leeds (Central NEY GLH lab)

- Shearing of genomic DNA using the Covaris E220 sonicator.
- End repair, A tailing and ligation of adaptors using SureSelectXT library system (Agilent Technologies).
- Enrichment by SureSelect target enrichment (Agilent Technologies) using custom in house designed probes. Samples have barcode tags added following target enrichment.
- Sequencing on the Illumina NextSeq using the NextSeq 500/550 High Output Kit v2.5 (300 Cycles), performing paired 150bp reads.

AmpliSeq/Ion Torrent

- Generate amplicon libraries using Ion AmpliSeq (Thermo Fisher Scientific) primer pools, and add patient specific barcodes.
- Ion Chef System provides automated, high-throughput preparation of template-positive Ion Sphere Particles (ISPs) and Ion 520, 530 or 540 chip loading.
- Sequencing on the Ion S5 or S5 Prime system.

Data Analysis

- Based on the open source 'Best Practices' workflow by the Broad Institute (for additional information, see http://www.broadinstitute.org/gatk/guide/best-practices).
- BWA alignment of reads to human genome build hg19.
- Generation of depth of coverage reports. Checked using Alamut Visual (Interactive Biosoftware).
- For SureSelect/Illumina: a minimum threshold of 30-fold read depth is set for exonic sequences and intronic sequences up to and including 5 bp from exon. A minimum threshold of 18-fold read depth is set for intronic sequences from 6 bp to 25 bp from exon. For AmpliSeq/Ion Torrent minimum 50x for exonic regions and ±5bp into introns and minimum 30x for intronic regions between 6 and 25bp from exons.
- Identification of variants using HaplotypeCaller. Annotation from dbSNP and COSMIC (currently dbSNP150 and COSMIC v67 but updated with new releases)

- Variants filtered against in-house polymorphism lists and Best Practice Guidelines for the evaluation of pathogenicity and the reporting of sequence variants in clinical molecular genetics (Association for Clinical Genetic Science).
- Assessment of copy number variants using DeCON software for SureSelect/Illumina data generated in Sheffield. This analysis can detect single-exon deletions, with smallest deletion detected 116bp (largest = 200kbp). With thresholds in place, at exon-level resolution, non-mosaic CNVs sensitivity 1.0 and specificity 0.989.
- Assessment of copy number variants using comparative depth read analysis for SureSelect/Illumina data generated in Leeds (Central NEY GLH lab), includes analysis of single exon deletions and duplications (smallest deletion = 43bp) with sensitivity 100%.
- Note copy number variants are not currently analysed using AmpliSeq/Ion Torrent platform

SureSelect/Illumina lab work performed in Leeds (Central NEY GLH lab)

- Samples are analysed using varDB, a tool for filtering variants designed, built, and maintained by Leeds Genetics Laboratory as part of the NEY GLH.
- Genes and transcripts of interest are applied to each Sample. Variants are filtered using the following criteria against annotations derived from Alamut Batch v1.9 (database version 2018-01-18):
    - In a Gene associated with the Sample.
    - In a Transcript associated with the Sample.
    - Does not have an artefact signature (relative to locally sequenced population).
    - Has a HGMD classification of DM or DM? (irrespective of population frequency)
    - Has an ExAC All or gnomAD All frequency less than 0.06.
    - For synonymous variants:
        - distNearestSS between -3 and 3
        - nearestSSChange less than -0.1
        - localSS_varMaxEntScore greater than 4.0
    - For intronic variants:
        - nearestSSChange less than -0.1
        - localSS_varMaxEntScore greater than 4.0
- Individual variant calls which pass filtering are then investigated further to check whether they are genuine variant calls.
- Genuine variant calls are then classified by Clinical Scientists according to the ACMG Variant Classification guidelines.

Post analysis

- Confirmation of clinically significant sequence variants by Sanger sequencing and/or MLPA (or ddPCR) as necessary (see below).
- Filling of gaps with low depth of coverage by Sanger sequencing as necessary. Gaps that fall within genes that have an individual whole gene sequencing service listed on the UKGTN service website as filled. The locations of all gaps that are not filled by Sanger sequencing are clearly indicated on the report.
- Creation of a diagnostic report combining clonal and Sanger sequence data that includes clinical interpretation of all the variants identified that are clearly pathogenic, likely to be pathogenic or variants of uncertain significance with high probability of pathogenicity (see ACGS variant interpretation guidelines 2020).

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Confirmation of variants detected by NGS

A risk assessment was carried out in June 2016 regarding the necessity for confirming variants identified by NGS using orthogonal technology (typically Sanger sequencing). The risk assessment was based on best practice guidelines, analysis of the quality of NGS data produced and the robustness of the tube transfer checking process within the laboratory.

The conclusion was: -

SDGS will no longer performs confirmation of single nucleotide substitution variants rated as class 3, 4 and 5, with a QUAL score of ≥3000 (and depth of coverage of ≥30x [≥18x for -25_-6 and +6_+25]), detected by SureSelect NGS workflows run on the MiSeq and HiSeq2500 platforms, and AmpliSeq/Ion Torrent workflows run of S5 and S5 Prime platforms.

Exclusions to this include: -

- Indel variants - these are confirmed by Sanger sequencing, regardless of their QUAL score.
- Ampliseq/S5/S5Prime – all variants are confirmed by Sanger sequencing.
- Rare genes with highly homologous pseudogenes
- Somatic cancer panels

All copy number variants detected by analysis of NGS data (Sheffield and Leeds) must be confirmed an alternative method (usually MLPA or ddPCR) before clinical reporting.

Next Generation Sequencing Panels

Inborn Errors of Metabolism

(SureSelect Design ID: 0660881) wetwork in Sheffield as interim before moving to Manchester

Rhabdomyolysis / Metabolic Myopathies

Condition	Gene	Original panel	Primary transcript
very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	*ACADVL*	FAO	NM_000018.2
Glycogen storage disease III	*AGL*	GSD	NM_000642.2
Glycogen storage disease XII	*ALDOA*	GSD	NM_000034.2
Muscular dystrophy, limb-girdle, type IC; Myopathy, distal, Tateyama type; Rippling muscle disease	*CAV3*	Rhabdo	NM_001234.3
CARNITINE PALMITOYLTRANSFERASE I, MUSCLE	*CPT1B*	FAO	NM_004377.3
Carnitine palmitoyltransferase II (CPT II) deficiency	*CPT2*	FAO	NM_000098.2
Glycogen storage disease XIII	*ENO3*	GSD	NM_001976.4
MADD	*ETFA*	FAO	NM_000126.3
MADD	*ETFB*	FAO	NM_001985.2
MADD (riboflavin-responsive)	*ETFDH*	FAO	NM_004453.2
N/A - no mutations reported to date	*FBP2*	GSD	NM_003837.2
Glycogen storage disease II	*GAA*	GSD	NM_000152.3
Glycogen storage disease IV / Polyglucosan body disease, adult form	*GBE1*	GSD	NM_000158.3
Glycogen storage disease XV	*GYG1*	GSD	NM_004130.3
Glycogen storage disease 0, muscle	*GYS1*	GSD	NM_002103.4
Mitochondrial TFP deficiency	*HADHA*	FAO	NM_000182.4
Mitochondrial TFP deficiency	*HADHB*	FAO	NM_000183.2
Hereditary myopathy with lactic acidosis	*ISCU*	Rhabdo	NM_213595.2
Glycogen storage disease XI	*LDHA*	GSD	NM_005566.3
Autosomal recessive recurrent acute myoglobinuria	*LPIN1*	Rhabdo	NM_145693.2
Glycogen storage disease VII	*PFKM*	GSD	NM_00116686.1
Glycogen storage disease X	*PGAM2*	GSD	NM_000290.3
Phosphoglycerate kinase 1 deficiency	*PGK1*	GSD	NM_000291.3
Glycogen storage disease XIV	*PGM1*	GSD	NM_002633.2
GSD IXd; muscle phosphorylase kinase deficiency	*PHKA1*	GSD	NM_002637.3
	PHKG1	GSD	NM_006213.3
Glycogen storage disease, type V; McArdle disease	*PYGM*	GSD	NM_005609.2
RBCK1 deficiency	*RBCK1*	GSD	NM_031229.2
Central Core Disease	*RYR1*	Rhabdo	NM_000540.2
Systemic primary carnitine deficiency (carnitine transporter)	*SLC22A5*	FAO	NM_003060.3

30 genes

GSD - Glycogen Storage Diseases

FAO - Fatty Acid oxidaton/metbaolism disorders

Rhabdo - genes unique to rhabdomyolysis/metaboilc myopathies panel

Familial Hypercholesterolemia panel

(wet work in Leeds Central NEY GLH lab – FH panel)

Condition	Gene	Primary transcript
Familial hypercholesterolaemia	LDLR	NM_000527.4
Autosomal dominant type B hypercholesterolaemia	APOB	NM_000384.2
Hyperlipoproteinemia, type III	APOE	NM_000041.3
Familial hypercholesterolaemia 3	PCSK9	NM_174936.3
Autosomal recessive hypercholesterolaemia	LDLRAP1	NM_015627.2

Musculoskeletal Service

(SureSelect Design ID: 3278641) Labwork in NEYGLH central lab

R101 Ehlers Danlos Syndrome (EDS)		
Condition	**Gene**	**Primary transcript**
Dermatosparaxis EDS	ADAMTS2	NM_014244.4
Classical Like EDS II	AEBP1	NM_001129.4
Cutis Laxa	ALDH18A1	NM_002860.3
Cutis Laxa	ATP6V0A2	NM_012463.3
Cutis Laxa	ATP6V1A	NM_001690.3
Cutis Laxa	ATP7A	NM_000052.6
Spondylodysplastic EDS	B3GALT6	NM_080605.3
Spondylodysplastic EDS	B4GALT7	NM_007255.2
Meester-Loeys syndrome	BGN	NM_001711.5
Peridontal EDS	C1R	NM_001733.5
Peridontal EDS	C1S	NM_201442.3
Homocystinuria	CBS	NM_000071.2
Musculocontracural EDS	CHST14	NM_130468.3
Myopathic EDS	COL12A1	NM_004370.5
Arthrochalasia EDS	COL1A1	NM_000088.3
Cardiac-Valvular EDS, Arthrochalasia EDS	COL1A2	NM_000089.3
Vascular EDS	COL3A1	NM_000090.3
Classical EDS	COL5A1	NM_000093.4
	COL5A2	NM_000393.4
Bethlem Myopathy	COL6A1	NM_001848.2
Bethlem Myopathy	COL6A2	NM_001849.3

Bethlem Myopathy	COL6A3	NM_004369.3
Musculocontractural EDS	DSE	NM_013352.3
Cutis Laxa	EFEMP2	NM_016938.4
Cutis Laxa	ELN	NM_001278939.1
Cutis Laxa	FBLN5	NM_006329.3
Marfan's Syndrome	FBN1	NM_000138.4
Beal's Syndrome	FBN2	NM_001999.3
Kyphoscoliotic EDS	FKBP14	NM_017946.3
Geroderma osteodysplasticum	GORAB	NM_152281.2
FTAA	LOX	NM_002317.6
Cutis Laxa	LTBP4	NM_003573.2
Kyphoscoliotic EDS	PLOD1	NM_000302.3
Brittle Cornea syndrome	PRDM5	NM_018699.3
Cutis Laxa	PYCR1	NM_006907.3
Cutis Laxa	RIN2	NM_018993.3
Gaze palsy with progressive scoliosis	ROBO3	NM_022370.3
Shprintzen-Goldberg syndrome	SKI	NM_003036.3
Spondylodysplastic EDS	SLC39A13	NM_152264.4
Loeys-Dietz Syndrome	SMAD2	NM_005901.5
Loeys-Dietz Syndrome	SMAD3	NM_005902.3
Loeys-Dietz Syndrome	TGFB2	NM_003238.4
Loeys-Dietz Syndrome	TGFB3	NM_003239.4
Loeys-Dietz Syndrome	TGFBR1	NM_004612.3
Loeys-Dietz Syndrome	TGFBR2	NM_003242.5
Classical Like EDS	TNXB	NM_019105.6
Brittle Cornea syndrome	ZNF469	NM_001127464.2
R102 Osteogenesis Imperfecta		
Hypophospatasia	ALPL	NM_000478.5
Spondylodysplastic EDS	B3GALT6	NM_080605.3
Spondylodysplastic EDS	B4GALT7	NM_007255.2
OI type XIII	BMP1	NM_006129.4
Hyperparathyroidism	CASR	NM_000388.3
OI type I - IV	COL1A1	NM_000088.3
OI type I - IV	COL1A2	NM_000089.3
OI type XVI	CREB3L1	NM_052854.3
OI type VII	CRTAP	NM_006371.4
Dentinogenesis Imperfecta II	DSPP	NM_014208.3
OI type XI	FKBP10	NM_021939.3
Geroderma osteodysplasticum	GORAB	NM_152281.2
OI type V	IFITM5	NM_001025295.2
Osteoporosis-pseudoglioma syndrome	LRP5	NM_002335.3
Bone fragility	NBAS	NM_015909.3
Hajdu-Cheney syndrome	NOTCH2	NM_024408.3
OI type XIII	P3H1	NM_022356.3

Cole-Carpenter syndrome I	P4HB	NM_000918.3
Bruck syndrome	PLOD2	NM_182943.2
X-linked osteoporosis	PLS3	NM_005032.6
OI type IX	PPIB	NM_000942.4
Cole-Carpenter syndrome II	SEC24D	NM_014822.3
OI type VI	SERPINF1	NM_002615.6
OI type X	SERPINH1	NM_001235.3
OI type XII	SP7	NM_001173467.2
OI type XVII	SPARC	NM_003118.3
Osteochondrodysplasia	TAPT1	NM_153365.2
OI type XVIII	TENT5A (FAM46A)	NM_017633.2
OI type XIV	TMEM38B	NM_018112.2
Hyperparathyroidism	TRPV6	NM_018646.5
OI type XV	WNT1	NM_005430.3

Musculoskeletal special panel
ID: WG_IAD186831_improve.20191114 (wet work in Sheffield)

R390 Multiple exotoses	EXT1	NM_000127.3
	EXT2	NM_207122.2
R25 Thanatophoric dysplasia	FGFR3	NM_000142.4

Familial Thoracic Aortic Aneurysm (FTAA) panel
for non NHSE referrals only SureSelect ID: 0782441 (wetwork in Sheffield)

Condition	Gene	Primary transcript
FTAA	*ACTA2*	NM_001613.2
FTAA	*CBS*	NM_000071.2
FTAA	*COL1A1*	NM_000088.3
EDS-Vascular	*COL3A1*	NM_000090.3
Marfan's Syndrome	*FBN1*	NM_000138.4
Beal's Syndrome	*FBN2*	NM_001999.3
FTAA	*FLNA*	NM_001110556.1
FTAA	*GATA5*	NM_080473.4
FTAA	*MFAP5*	NM_003480.3
FTAA	*MYH11*	NM_002474.2
FTAA	*MYLK*	NM_053025.3
FTAA	*NOTCH1*	NM_017617.3
FTAA	*PRKG1*	NM_006258.2
FTAA	*SKI*	NM_003036.3
FTAA	*SLC2A10*	NM_030777.3
FTAA	*SMAD3*	NM_005902.3
FTAA	*SMAD4*	NM_005359.3

FTAA	*TGFB2*	NM_001135599.2
FTAA	*TGFB3*	NM_003239.2
Loeys-Dietz Syndrome	*TGFBR1*	NM_004612.2
Loey's Dietz Syndrome/Marfan's syndrome	*TGFBR2*	NM_001024847.2
FTAA	*SLC2A10*	NM_000071.2

Hereditary Cancer Panels

Breast and Ovarian cancer genes
(wet work in Leeds Central NEY GLH lab – Breast Ovarian Bowel BOB panel)

R208: Breast & Ovarian cancer

Condition	OMIM	Gene	Primary transcript
Breast-ovarian cancer, familial, 1	604370	*BRCA1*	NM_007294.3
Breast-ovarian cancer, familial, 2	612555	*BRCA2*	NM_000059.3
Breast cancer, susceptibility to	114480	*PALB2*	NM_024675.3
Li-Fraumeni Syndrome (optional add R216)	151623	*TP53*	NM_000546.5

R207: Ovarian cancer

Breast-ovarian cancer, familial, 1	604370	*BRCA1*	NM_007294.3
Breast-ovarian cancer, familial, 2	612555	*BRCA2*	NM_000059.3
Breast cancer, susceptibility to	114480	*PALB2*	NM_024675.3
Breast cancer, early-onset	114480	*BRIP1*	NM_032043.2
Breast-ovarian cancer, familial, susceptibility to, 3	613399	*RAD51C*	NM_058216.2
Breast-ovarian cancer, familial, susceptibility to, 4	614291	*RAD51D*	NM_002878.3
Colorectal cancer, hereditary nonpolyposis, type 2	609310	*MLH1*	NM_000249.3
Colorectal cancer, hereditary nonpolyposis, type 1	120435	*MSH2*	NM_000251.2
Colorectal cancer, hereditary nonpolyposis, type 5	614350	*MSH6*	NM_000179.2
Colorectal cancer, hereditary nonpolyposis, type 4	614337	*PMS2*	NM_000535.5
Colorectal cancer, hereditary nonpolyposis, type 8	613244	*EPCAM*	NM_002354.2

Colorectal Cancer gene panels
(wet work in Leeds Central NEY GLH lab – Breast Ovarian Bowel BOB panel)

R210: Lynch syndrome (HNPCC)

Condition	OMIM	Gene	Primary transcript
Colorectal cancer, hereditary nonpolyposis, type 2	609310	*MLH1*	NM_000249.3
Colorectal cancer, hereditary nonpolyposis, type 1	120435	*MSH2*	NM_000251.2
Colorectal cancer, hereditary nonpolyposis, type 5	614350	*MSH6*	NM_000179.2
Colorectal cancer, hereditary nonpolyposis, type 4	614337	*PMS2*	NM_000535.5
Colorectal cancer, hereditary nonpolyposis, type 8	613244	*EPCAM*	NM_002354.2

R211: Inherited Polyposis

Condition	OMIM	Gene	Primary transcript

Colorectal Cancer gene panels

(wet work in Leeds Central NEY GLH lab – Breast Ovarian Bowel BOB panel)

R210: Lynch syndrome (HNPCC)

Condition	OMIM	Gene	Primary transcript
Familial Adenamatous Polyposis	175100	*APC*	NM_000038.5
Polyposis, juvenile intestinal	174900	*BMPR1A*	NM_004329.2
Colorectal cancer, hereditary nonpolyposis, type 8	613244	*EPCAM*	NM_002354.2
Colorectal cancer, hereditary nonpolyposis, type 2	609310	*MLH1*	NM_000249.3
Colorectal cancer, hereditary nonpolyposis, type 1	120435	*MSH2*	NM_000251.2
Colorectal cancer, hereditary nonpolyposis, type 5	614350	*MSH6*	NM_000179.2
Colorectal cancer, hereditary nonpolyposis, type 1	120435	*MUTYH*	NM_001128425.1
Familial Adenomatous Polyposis, type 3	602656	*NTHL1*	NM_002528.6
Colorectal cancer, hereditary nonpolyposis, type 4	614337	*PMS2*	NM_000535.5
Cowden Syndrome	158350	*PTEN*	NM_000314.4
Polyposis, juvenile intestinal	174900	*SMAD4*	NM_005359.5
Peutz-Jeghers Syndrome	175200	*STK11*	NM_000455.4
Colorectal cancer, susceptibility to, 10	612591	*POLD1*	NM_001308632.1
Colorectal cancer, susceptibility to, 12	615083	*POLE*	NM_006231.3

Renal Cancer gene Panel

non-NHS patients only (SureSelect design ID: 3014041) wetwork in Sheffield

Condition	OMIM	Gene	Primary transcript
Paragangliomas 4	115310	*SDHB*	NM_003000.2
Leiomyomatosis and renal cell cancer	150800	*FH*	NM_000143.3
von Hippel-Lindau syndrome	193300	*VHL*	NM_000551.2
Tumor predisposition syndrome	614327	*BAP1*	NM_004656.3
Melanoma, and renal cell carcinoma, susceptibility to, 8	614456	*MITF*	NM_000248.3
Renal cell carcinoma, papillary, 1, familial and somatic	605074	*MET*	NM_001127500.2
Tuberous sclerosis-1	191100	*TSC1*	NM_000368.4
Tuberous sclerosis-2	191092	*TSC2*	NM_000548.4
Cowden syndrome 1	158350	*PTEN*	NM_000314.4
Wilms tumor, type 1	194070	*WT1*	NM_024426.4
Birt-Hogg-Dube syndrome	135150	*FLCN*	NM_144997.6
Renal cell carcinoma	144700	*HNF1A*	NM_001306179.1

Polycystic Disease Panels

SureSelect Design ID: 3144591 – wetwork in Sheffield

Polycystic Kidney and Liver Disease – non NHS patients and NHS interim to WGS

R193.1 Renal cystic panel – PKD1

Condition	OMIM	Gene	Primary transcript
Polycystic kidney disease 1	173900	*PKD1*	NM_001009944.2
Polycystic kidney disease 2	613095	*PKD2*	NM_000297.3

R193.3 Renal Cystic panel – interim to WGS

Condition	OMIM	Gene	Primary transcript
Polycystic kidney disease 1	173900	*PKD1*	NM_001009944.2
Polycystic kidney disease 2	613095	*PKD2*	NM_000297.3
Polycystic kidney disease 4,with hepatic disease	263200	*PKHD1*	NM_138694.3
Polycystic kidney disease 5	617610	*DZIP1L*	NM_173543.2
Polycstic liver disease 1	174050	*PRKCSH*	NM_002743.3
Polycystic liver disease 2	617004	*SEC63*	NM_007214.4
Polycystic liver disease 4 with or without kidney cysts	617875	*LRP5*	NM_002335.2
Polycystic kidney disease 3	600666	*GANAB*	NM_198335.3
Polycystic liver disease 3 with or without kidney cysts	617874	*ALG8*	NM_024079.4
Polycystic liver disease	609214	*SEC61B*	NM_006808.2
Polycystic kidney disease 6 with or without polycystic liver disease	618061	*DNAJB11*	NM_016306.5
Hyperuricemic nephropathy, familial juvenile 2	613092	*REN*	NM_000537.3
Medullary cystic kidney disease 2	603860	*UMOD*	NM_003361.3
Renal cysts and diabetes syndrome	137920	*HNF1B*	NM_000458.3
Hyperuricemic nephropathy, familial juvenile, 4	617056	*SEC61A1*	NM_006808.2
Tuberous sclerosis-1	191100	*TSC1*	NM_000368.4
Tuberous sclerosis-2	191092	*TSC2*	NM_000548.4

Autosomal Dominant Tubulointerstitial Kidney Disease

(aka Medullary cystic & Glomerulocystic kidney disease) – non-NHS patients only

Condition	OMIM	Gene	Primary transcript
Hyperuricemic nephropathy, familial juvenile 2	613092	*REN*	NM_000537.3
Medullary cystic kidney disease 2	603860	*UMOD*	NM_003361.3
Renal cysts and diabetes syndrome	137920	*HNF1B*	NM_000458.3
Hyperuricemic nephropathy, familial juvenile, 4	617056	*SEC61A1*	NM_006808.2
Tuberous sclerosis-1	191100	*TSC1*	NM_000368.4
Tuberous sclerosis-2	191092	*TSC2*	NM_000548.4

Neurogenetic Disorders

Neurogenetic Motor Disorders

(SureSelect Design ID: 0836801).

Hereditary Spastic Paraparesis
88 gene panel non-NHS patients only

Condition	OMIM	Gene	Primary transcript
Adrenoleukodystrophy, ALD	300100	*ABCD1*	NM_000033.3
Ataxia, spastic, 5, autosomal recessive, SPAX5	614487	*AFG3L2*	NM_006796.2
Spastic paraplegia 9A, autosomal dominant, SPG9A	601162	*ALDH18A1*	NM_002860.3
Spastic paralysis, infantile onset ascending, IAHSP	607225	*ALS2*	NM_020919.3
Spastic paraplegia 63, SPG63	615686	*AMPD2*	NM_001257360.1
Pettigrew syndrome, PGS	304340	*AP1S2*	NM_001272071.1
Spastic paraplegia 47, autosomal recessive, SPG47	614066	*AP4B1*	NM_006594.4
Spastic paraplegia 51, autosomal recessive, SPG51	613744	*AP4E1*	NM_007347.4
Spastic paraplegia 50, autosomal recessive, SPG50	612936	*AP4M1*	NM_004722.3
Spastic paraplegia 52, autosomal recessive, SPG52	614067	*AP4S1*	NM_007077.4
Spastic paraplegia 48, autosomal recessive, SPG48	613647	*AP5Z1*	NM_014855.2
Argininemia	207800	*ARG1*	NM_001244438.1
Spastic paraplegia 61, autosomal recessive, SPG61	615685	*ARL6IP1*	NM_015161.2
Spastic paraplegia 66, SPG66		*ARSI*	NM_001012301.2
Spastic paraplegia 3A, autosomal dominant, SPG3A	182600	*ATL1*	NM_015915.4
Spastic paraplegia		*ATP2B4*	NM_001001396.2
Spastic paraplegia 26, autosomal recessive, SPG26	609195	*B4GALNT1*	NM_001478.4
Spastic paraplegia 17, autosomal dominant, SPG17	270685	*BSCL2*	NM_032667.6
Spastic paraplegia 55, autosomal recessive, SPG55	615035	*C12orf65*	NM_152269.4
Spastic paraplegia 43, autosomal recessive, SPG43	615043	*C19orf12*	NM_001031726.2
Neuropathy, hereditary sensory, with spastic paraplegia,	256840	*CCT5*	NM_012073.4
Spastic paraplegia 73, autosomal dominant, SPG73	616282	*CPT1C*	NM_001199752.2
Cerebrotendinous xanthomatosis, CTX	213700	*CYP27A1*	NM_000784.3
Spastic paraplegia 56, autosomal recessive, SPG56	615030	*CYP2U1*	NM_183075.2
Spastic paraplegia 5A, autosomal recessive, SPG5A	270800	*CYP7B1*	NM_004820.3
Spastic paraplegia 28, autosomal recessive, SPG28	609340	*DDHD1*	NM_001160148.1
Spastic paraplegia 54, autosomal recessive, SPG54	615033	*DDHD2*	NM_015214.2
Spastic paraplegia		*DNM2*	NM_001005360.2
Leukoencephalopathy with vanishing white matter, VWM	603896	*EIF2B5*	NM_003907.2
Spastic paraplegia 64, autosomal recessive, SPG64	615683	*ENTPD1*	NM_001164178.1
Spastic paraplegia 62, autosomal recessive, SPG62		*ERLIN1*	NM_001100626.1
Spastic paraplegia 18, autosomal recessive, SPG18	611225	*ERLIN2*	NM_007175.6
Spastic paraplegia 35, autosomal recessive, SPG35	612319	*FA2H*	NM_024306.4

Hereditary Spastic Paraparesis
88 gene panel non-NHS patients only

Condition	OMIM	Gene	Primary transcript
Spastic paraplegia		*FARS2*	NM_006567.3
Amyotrophic lateral sclerosis 11, ALS11	612577	*FIG4*	NM_014845.5
Spastic paraplegia 68, SPG68		*FLRT1*	NM_013280.4
Cerebral palsy, spastic quadriplegic, 1, CPSQ1	603513	*GAD1*	NM_000817.2
Giant axonal neuropathy-1, GAN1	256850	*GAN*	NM_022041.3
Spastic paraplegia 46, autosomal recessive, SPG46	614409	*GBA2*	NM_020944.2
Spastic paraplegia		*GCH1*	NM_000161.2
Oculodentodigital dysplasia, ODDD	164200	*GJA1*	NM_000165.3
Spastic paraplegia 44, autosomal recessive, SPG44	613206	*GJC2*	NM_020435.3
Tay-Sachs disease, TSD	272800	*HEXA*	NM_000520.4
Spastic paraplegia 13, autosomal dominant, SPG13	605280	*HSPD1*	NM_002156.4
Spastic paraplegia 74, autosomal recessive, SPG74	616451	*IBA57*	NM_001010867.3
Mental retardation, X-linked, syndromic, Claes-Jensen type, MRXSCJ	300534	*KDM5C*	NM_004187.3
Spastic paraplegia 8, autosomal dominant, SPG8	603563	*KIAA0196*	NM_014846.3
Spastic paraplegia 30, autosomal recessive, SPG30	610357	*KIF1A*	NM_001244008.1
Spastic ataxia 2, autosomal recessive, SPAX2	611302	*KIF1C*	NM_006612.5
Spastic paraplegia 10, autosomal dominant, SPG10	604187	*KIF5A*	NM_004984.2
Spastic paraplegia		*KLC4*	NM_201523.2
MASA syndrome, MASA	303350	*L1CAM*	NM_000425.3
Spastic paraplegia		*LYST*	NM_000081.3
Spastic paraplegia		*MAG*	NM_002361.3
Spastic paraplegia 70, SPG70		*MARS*	NM_004990.3
Spastic ataxia 3, autosomal recessive, SPAX3	611390	*MARS2*	NM_138395.3
Spastic paraplegia		*MT-ATP6*	NC_012920.1
Ataxia, spastic, 4, SPAX4	613672	*MTPAP*	NM_018109.3
Spastic paraplegia 6, autosomal dominant, SPG6	600363	*NIPA1*	NM_144599.4
Spastic paraplegia 45, autosomal recessive, SPG45	613162	*NT5C2*	NM_012229.4
Spastic paraplegia 67, SPG67		*PGAP1*	NM_024989.3
Spastic paraplegia 2, X-linked, SPG2	312920	*PLP1*	NM_001128834.2
Spastic paraplegia 39, autosomal recessive, SPG39	612020	*PNPLA6*	NM_001166111.1
Alzheimer disease, type 3, with spastic paraparesis and apraxia, AD3	607822	*PSEN1*	NM_000021.3
Spastic paraplegia 69, SPG69		*RAB3GAP2*	NM_012414.3
Spastic paraplegia 31, autosomal dominant, SPG31	610250	*REEP1*	NM_001164730.1
Spastic paraplegia 72, autosomal recessive, SPG72	615625	*REEP2*	NM_001271803.1
Spastic paraplegia 12, autosomal dominant, SPG12	604805	*RTN2*	NM_005619.4
Spastic ataxia, Charlevoix-Saguenay type, SACS	270550	*SACS*	NM_014363.5
Amyotrophic lateral sclerosis 16, juvenile, ALS16	614373	*SIGMAR1*	NM_005866.3
Allan-Herndon-Dudley syndrome, AHDS	300523	*SLC16A2*	NM_006517.4

Hereditary Spastic Paraparesis
88 gene panel non-NHS patients only

Condition	OMIM	Gene	Primary transcript
Dystonia 9, DYT9	601042	*SLC2A1*	NM_006516.2
Spastic paraplegia 42, autosomal dominant, SPG42	612539	*SLC33A1*	NM_001190992.1
Spastic paraplegia 4, autosomal dominant, SPG4	182601	*SPAST*	NM_014946.3
Spastic paraplegia 11, autosomal recessive, SPG11	604360	*SPG11*	NM_025137.3
Spastic paraplegia 20, autosomal recessive, SPG20	275900	*SPG20*	NM_015087.4
Mast syndrome	248900	*SPG21*	NM_016630.6
Spastic paraplegia 7, autosomal recessive, SPG7	607259	*SPG7*	NM_003119.3
Spastic paraplegia 49, autosomal recessive, SPG49	615031	*TECPR2*	NM_014844.3
Spastic paraplegia 57, autosomal recessive, SPG57	615658	*TFG*	NM_006070.5
Spastic paraplegia 59, autosomal recessive, SPG59		*USP8*	NM_005154.4
Spastic ataxia 1, autosomal dominant, SPAX1	108600	*VAMP1*	NM_014231.4
Spastic paraplegia 53, autosomal recessive, SPG53	614898	*VPS37A*	NM_152415.2
Neurodegeneration with brain iron accululation 5, NBIA5	300894	*WDR45*	NM_007075.3
Spastic paraplegia 60, autosomal recessive, SPG60		*WDR48*	NM_020839.3
Spastic paraplegia 71, SPG71		*ZFR*	NM_016107.3
Spastic paraplegia 15, autosomal recessive, SPG15	270700	*ZFYVE26*	NM_015346.3
Spastic paraplegia 33, autosomal dominant, SPG33	610244	*ZFYVE27*	NM_001002261.3

Spinal Muscular Atrophy 29 gene panel
non-NHS patients only

Condition	OMIM	Gene	Primary transcript
Charcot-Marie-Tooth disease, axonal, type 2N, CMT2N	613287	*AARS*	NM_001605.2
Spinal muscular atrophy with progressive myoclonic epilepsy, SMAPME	159950	*ASAH1*	NM_004315.4
Spinal muscular atrophy, distal, X-linked 3, SMAX3	300489	*ATP7A*	NM_000052.4
Spinal muscular atrophy, lower extremity-predominant, 2, AD, SMALED2	609797	*BICD2*	NM_001003800.1
Spastic paraplegia 17, autosomal dominant, SPG17	270685	*BSCL2*	NM_032667.6
Spinal muscular atrophy, Jokela type, SMAJ	615048	*CHCHD10*	NM_001301339.1
Neuropathy, distal hereditary motor, type VIIB, HMN7B	607641	*DCTN1*	NM_004082.4
Spinal muscular atrophy, distal, autosomal recessive, 5, DSMA5	614881	*DNAJB2*	NM_001039550.1
Spinal muscular atrophy, lower extremity-predominant 1, AD, SMALED1	158600	*DYNC1H1*	NM_001376.4
Pontocerebellar hypoplasia, type 1B, PCH1B	614678	*EXOSC3*	NM_016042.3
Pontocerebellar hypoplasia, type 1C, PCH1C	616081	*EXOSC8*	NM_181503.2
Neuronopathy, distal hereditary motor, type IID, HMN2D	615575	*FBXO38*	NM_030793.4
Neuropathy, distal hereditary motor, type VA, HMN5A	600794	*GARS*	NM_002047.2
Tay-Sachs disease, TSD	272800	*HEXA*	NM_000520.4
Neuropathy, distal hereditary motor, type IIB, HMN2B	608634	*HSPB1*	NM_001540.3

Spinal Muscular Atrophy 29 gene panel
non-NHS patients only

Condition	OMIM	Gene	Primary transcript
Neuronopathy, distal hereditary motor, type IIC, HMN2C	613376	*HSPB3*	NM_006308.2
Neuropathy, distal hereditary motor, type IIA, HMN2A	158590	*HSPB8*	NM_014365.2
Spinal muscular atrophy, distal, autosomal recessive, 1, DSMA1	604320	*IGHMBP2*	NM_002180.2
Spinal muscular atrophy		*LAS1L*	NM_031206.4
Spinal muscular atrophy, distal, autosomal recessive, 4, DSMA4	611067	*PLEKHG5*	NM_198681.3
Neuronopathy, distal hereditary motor, type VB, HMN5B	614751	*REEP1*	NM_001164730.1
Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, CEMCOX1	604377	*SCO2*	NM_005138.2
Spinal muscular atrophy, distal, autosomal recessive, 2, DSMA2	605726	*SIGMAR1*	NM_005866.3
Neuronopathy, distal hereditary motor, type VIIA, HMN7A	158580	*SLC5A7*	NM_021815.4
Spinal muscular atrophy-1, SMA1	253300	*SMN1*	NM_000344.3
Scapuloperoneal spinal muscular atrophy, SPSMA	181405	*TRPV4*	NM_021625.4
Spinal muscular atrophy, X-linked 2, infantile, SMAX2	301830	*UBA1*	NM_003334.3
Spinal muscular atrophy, late-onset, Finkel type, SMAFK	182980	*VAPB*	NM_004738.4
Pontocerebellar hypoplasia type 1A, PCH1A	607596	*VRK1*	NM_003384.2

Neurodegenerative Disorders

(SureSelect Design ID: 0836801).

Familial Amyotrophic Lateral Sclerosis with or without Frontotemporal Dementia
42 gene panel – non-NHS patients only

Condition	OMIM	Gene	Primary transcript
Primary lateral sclerosis, juvenile, PLSJ	606353	*ALS2*	NM_020919.3
Amyotrophic lateral sclerosis 9, ALS9	611895	*ANG*	NM_001145.4
Familial Amyotrophic Lateral Sclerosis		*ANXA11*	NM_145869.1
Familial Amyotrophic Lateral Sclerosis		*ARHGEF28*	NM_001080479.2
Spinocerebellar ataxia 2, SCA2	183090	*ATXN2*	NM_002973.3
Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, FTDALS2	615911	*CHCHD10*	NM_001301339.1
Amyotrophic lateral sclerosis 17, ALS17	614696	*CHMP2B*	NM_014043.3
Cerebrotendinous xanthomatosis, CTX	213700	*CYP27A1*	NM_000784.3
Familial Amyotrophic Lateral Sclerosis		*DAO*	NM_001917.4
Neuropathy, distal hereditary motor, type VIIB, HMN7B	607641	*DCTN1*	NM_004082.4
Amyotrophic lateral sclerosis 19, ALS19	615515	*ERBB4*	NM_005235.2
Familial Amyotrophic Lateral Sclerosis		*EWSR1*	NM_013986.3
Amyotrophic lateral sclerosis 11, ALS11	612577	*FIG4*	NM_014845.5

Familial Amyotrophic Lateral Sclerosis with or without Frontotemporal Dementia

42 gene panel – non-NHS patients only

Condition	OMIM	Gene	Primary transcript
Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, ALS6	608030	*FUS*	NM_004960.3
Spastic paraplegia 46, autosomal recessive, SPG46	614409	*GBA2*	NM_020944.2
Frontotemporal lobar degeneration with TDP43 inclusions, GRN related,	607485	*GRN*	NM_002087.3
Amyotrophic lateral sclerosis 20, ALS20	615426	*HNRNPA1*	NM_031157.2
Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 2, IBMPFD2	615422	*HNRNPA2B1*	NM_031243.2
Frontotemporal Dementia, FTD	600274	*MAPT*	NM_005910.5
Amyotrophic lateral sclerosis 21, ALS21	606070	*MATR3*	NM_199189.2
Amyotrophic lateral sclerosis, susceptibility to, ALS1	105400	*NEFH*	NM_021076.3
Familial Amyotrophic Lateral Sclerosis		*NEK1*	NM_001199397.1
Amyotrophic lateral sclerosis 12, ALS12	613435	*OPTN*	NM_001008211.1
Amyotrophic lateral sclerosis 18, ALS18	614808	*PFN1*	NM_005022.3
Amyotrophic lateral sclerosis, susceptibility to, ALS1	105400	*PRPH*	NM_006262.3
Amyotrophic lateral sclerosis 4, juvenile, ALS4	602433	*SETX*	NM_015046.5
Amyotrophic lateral sclerosis 16, juvenile, ALS16	614373	*SIGMAR1*	NM_005866.3
Amyotrophic lateral sclerosis 1, ALS1	105400	*SOD1*	NM_000454.4
Spastic paraplegia 4, autosomal dominant, SPG4	182601	*SPAST*	NM_014946.3
Amyotrophic lateral sclerosis 5, juvenile, ALS5	602099	*SPG11*	NM_025137.3
Spastic paraplegia 20, autosomal recessive, SPG20	275900	*SPG20*	NM_015087.4
Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, FTDALS3	616437	*SQSTM1*	NM_003900.4
Familial Amyotrophic Lateral Sclerosis		*SS18L1*	NM_198935.2
Familial Amyotrophic Lateral Sclerosis		*TAF15*	NM_139215.2
Amyotrophic lateral sclerosis 10, with or without FTD, ALS10	612069	*TARDBP*	NM_007375.3
Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, FTDALS4	616439	*TBK1*	NM_013254.3
Amyotrophic lateral sclerosis 22 with or without frontotemoral dementia, ALS22	616208	*TUBA4A*	NM_006000.2
Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, ALS15	300857	*UBQLN2*	NM_013444.3
Amyotrophic lateral sclerosis 8, ALS8	608627	*VAPB*	NM_004738.4
Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, ALS14	613954	*VCP*	NM_007126.3
Familial Amyotrophic Lateral Sclerosis		*VPS54*	NM_016516.2
Pontocerebellar hypoplasia type 1A, PCH1A	607596	*VRK1*	NM_003384.2

Dementia 27 gene panel

non-NHS patients only

Condition	OMIM	Gene	Primary transcript
Cerebral amyloid angiopathy, APP Related	605714	*APP*	NM_000484.3
Alzheimer disease 1, familial, AD	104300	*APP*	NM_000484.3
Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, FTDALS2	615911	*CHCHD10*	NM_001301339.1
Frontotemporal Dementia, Chromosome 3-Linked, FTD3	600795	*CHMP2B*	NM_014043.3
Leukoencephalopathy, diffuse hereditary, with spheroids, HDLS	221820	*CSF1R*	NM_005211.3
Cerebrotendinous xanthomatosis, CTX	213700	*CYP27A1*	NM_000784.3
Perry syndrome	168605	*DCTN1*	NM_004082.4
Neuropathy, hereditary sensory, type IE, HSN1E	614116	*DNMT1*	NM_001130823.2
Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, ALS6	608030	*FUS*	NM_004960.3
Frontotemporal lobar degeneration with TDP43 inclusions, GRN related	607485	*GRN*	NM_002087.3
Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 2, IBMPFD2	615422	*HNRNPA2B1*	NM_031243.2
Cerebral arteriopathy, autosomal recessive, with subcortical infarcts and leukoencephalopathy, CARASIL	600142	*HTRA1*	NM_002775.4
Cerebral amyloid angiopathy, ITM2B-related, 1	176500	*ITM2B*	NM_021999.4
Frontotemporal Dementia, FTD	600274	*MAPT*	NM_005910.5
Amyotrophic lateral sclerosis 21, ALS21	606070	*MATR3*	NM_199189.2
Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy, CADASIL	125310	*NOTCH3*	NM_000435.2
Gerstmann-Straussler disease, GSD	137440	*PRNP*	NM_000311.3
Alzheimer disease, type 3, AD3	607822	*PSEN1*	NM_000021.3
Alzheimer disease-4, AD4	606889	*PSEN2*	NM_000447.2
Mast syndrome	248900	*SPG21*	NM_016630.6
Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, FTDALS3	616437	*SQSTM1*	NM_003900.4
Amyotrophic lateral sclerosis 10, with or without FTD, ALS10	612069	*TARDBP*	NM_007375.3
Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, FTDALS4	616439	*TBK1*	NM_013254.3
Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy, PLOSL	221770	*TREM2*	NM_018965.3
Amyotrophic lateral sclerosis 22 with or without frontotemoral dementia, ALS22	616208	*TUBA4A*	NM_006000.2
Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy, PLOSL	221770	*TYROBP*	NM_006000.2
Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, ALS15	300857	*UBQLN2*	NM_013444.3
Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, ALS14	613954	*VCP*	NM_007126.3

Neurogenetic Movement Disorders

(SureSelect Design ID: 3156571) wet work in Sheffield

Hereditary Dystonia

73 gene panel non-NHS patients only

Condition	OMIM	Gene	Primary Transcript
Aicardi-Goutieres, AGS6	615010	*ADAR*	NM_001111.4
Dyschromatosis Symmetrica Hereditaria, DSH	127400	*ADAR*	NM_001111.4
Dyskinesia, Familial, With Facial Myokymia, FDFM	606703	*ADCY5*	NM_183357.2
Ataxia, Spastic, 5, Autosomal Recessive, SPAX5;	604581	*AFG3L2*	NM_006796.2
Spinocerebellar Ataxia 28, SCA28	610246	*AFG3L2*	NM_006796.2
Dystonia 24, DYT24	615034	*ANO3*	NM_031418.2
Ataxia, Early Onset, With Occulomotor Apraxia And Hypoalbuminemia, EAOH	606350	*APTX*	NM_175073.4
Spinocerebellar Ataxia, Autosomal Recessive 25, SCAR25	617584	*ATG5*	NM_004849.1
Ataxia-Telangiectasia, AT	208900	*ATM*	NM_000051.3
Cerebellar Ataxia, Areflexia, Pes Cavus, Optic Atrophy, And Sensorineural Hearing Loss, CAPOS	601338	*ATP1A3*	NM_152296.4
Dystonia 12, DYT12	128235	*ATP1A3*	NM_152296.4
Alternating Hemiplegia Of Childhood 2, AHC2	614820	*ATP1A3*	NM_152296.4
Kufor-Rakeb Syndrome, KRS	606693	*ATP13A2*	NM_022089.3
Wilson Disease, WD	606882	*ATP7B*	NM_000053.3
Neurodegeneration With Brain Iron Accumulation 4, SPG43 / NBIA4	614298	*C19orf12*	NM_001031726.3
Dystonia 23, DYT23	614860	*CACNA1B*	NM_000718.3
Mitochondrial Dna Depletion Syndrome 7 (Hepatocerebral Type), MTDPS7	271245	*CCNY*	NM_145012.5
Dystonia 23, DYT23	614860	*CIZ1*	NM_001257975.1
Frontotemporal Dementia, Chromosome 3-Linked; , FTD3	600795	*CHMP2B*	NM_014043.3
Coenzyme Q10 Deficiency, Primary, 5, COQ10D5	614654	*COQ9*	NM_020312.3
Neurodegeneration With Brain Iron Accumulation 6, NBIA6	615643	*COASY*	NM_025233.6
Aceruloplasminemia,	604290	*CP*	NM_000096.3
Myoclonic Epilepsy Of Unverricht And Lundborg	254800	*CSTB*	NM_000100.3
Spastic Paraplegia 56, Autosomal Recessive, SPG56	615030	*CYP2U1*	NM_183075.2
Cerebrotendinous Xanthomatosis; , CTX	606530	*CYP27A1*	NM_000784.3
Perry Syndrome,	168605	*DCTN1*	NM_004082.4
Woodhouse-Sakati Syndrome,	241080	*DCAF17*	NM_025000.3
Spastic Paraplegia 35, Autosomal Recessive, SPG35	612319	*FA2H*	NM_024306.4
Parkinson Disease 15, Autosomal Recessive Early-Onset, PARK15	260300	*FBXO7*	NM_012179.3
Rett Syndrome, Congenital Variant,	613454	*FOXG1*	NM_005249.4
Neurodegeneration With Brain Iron Accumulation 3, NBIA3	606159	*FTL*	NM_000146.3
Parkinson Disease, Late-Onset, PD	168600	*GBA*	NM_001005741.2
Dystonia, Dopa-Responsive, DRD	128230	*GCH1*	NM_000161.2

Hereditary Dystonia

73 gene panel non-NHS patients only

Condition	OMIM	Gene	Primary Transcript
Dystonia 25, DYT25	615073	*GNAL*	NM_001142339.2
Neurodevelopmental Disorder With Involuntary Movements, NEDIM	617493	*GNAO1*	NM_020988.2
3-Methylglutaconic Aciduria, Type Viii, MGCA8	617248	*HTRA2*	NM_013247.4
Dystonia 28, Childhood-Onset, DYT28	617284	*KMT2B*	NM_014727.2
Dystonia, Childhood-Onset, With Optic Atrophy And Basal Ganglia Abnormalities, DYTOABG	617282	*MECR*	NM_016011.4
Niemann-Pick Disease, Type C1 Niemann-Pick Disease, Type D, Included, NPC1	257220	*NPC1*	NM_000271.4
Niemann-Pick Disease, Type C2, NPC2	607625	*NPC2*	NM_006432.3
Neurodegeneration With Brain Iron Accumulation 1, NBIA1	234200	*PANK2*	NM_153638.3
Hypoprebetalipoproteinemia, Acanthocytosis, Retinitis Pigmentosa, And Pallidal Degeneration	607236	*PANK2*	NM_153638.3
Parkinson Disease 2, Autosomal Recessive Juvenile, PARK2	600116	*PRKN*	NM_004562.2
Parkinson Disease 7, Autosomal Recessive Early-Onset, PARK7	606324	*PARK7*	NM_007262.4
Parkinson Disease 6, Autosomal Recessive Early-Onset, PARK6	605909	*PINK1*	NM_032409.2
Neurodegeneration With Brain Iron Accumulation 2b, NBIA2A/2B	610217	*PLA2G6*	NM_003560.3
Paroxysmal Nonkinesigenic Dyskinesia 1, PNKD1	118800	*PNKD*	NM_015488.4
Ataxia-Oculomotor Apraxia 4, AOA4	616267	*PNKP*	NM_007254.3
Spinocerebellar Ataxia 14, SCA14	605361	*PRKCG*	NM_002739.3
Dystonia 16, DYT16	612067	*PRKRA*	NM_003690.4
Episodic Kinesigenic Dyskinesia 1; EKD1 (Dystonia 10; DYT10)	614386	*PRRT2*	NM_145239.2
Microphthalmia, Syndromic 12, MCOPS12	615524	*RARB*	NM_000965.4
Lissencephaly 2; LIS2 And Myoclonus Dystonia, LIS2	257320	*RELN*	NM_005045.3
Epileptic Encephalopathy, Early Infantile, 6, EIEE6	607208	*SCN1A*	NM_001165963.1
Neonatal Epilepsy With Late-Onset Episodic Ataxia.,		*SCN2A*	NM_021007.2
Dystonia 11, Myoclonic, DYT11	159900	*SGCE*	NM_003919.2
Mental Retardation, X-Linked, Syndromic, Christianson Type, MRXSCH	300243	*SLC9A6*	NM_006359.2
Allan-Herndon-Dudley Syndrome, AHDS	300095	*SLC16A2*	NM_006517.4
Dystonia 9, DYT9	138140	*SLC2A1*	NM_006516.2
Hypermanganesemia With Dystonia, Polycythemia, And Cirrhosis, HMDPC	613280	*SLC30A10*	NM_018713.2
Hypermanganesemia With Dystonia 2, HMNDYT2	608736	*SLC39A14*	NM_015359.5
Parkinsonism-Dystonia, Infantile, PKDYS	613135	*SLC6A3*	NM_001044.4
Parkinson Disease 1, Autosomal Dominant, PARK1	168601	*SNCA*	NM_000345.3
Parkinson Disease 4, Autosomal Dominant, PARK4	605543	*SNCA*	NM_000345.3

Hereditary Dystonia

73 gene panel non-NHS patients only

Condition	OMIM	Gene	Primary Transcript
Spastic Paraplegia 11, Autosomal Recessive, SPG11	604360	*SPG11*	NM_025137.1
Dystonia, Dopa-Responsive, Due To Sepiapterin Reductase Deficiency	612716	*SPR*	NM_003124.4
Mitochondrial Depletion Disorder		*SUCLA2*	NM_003850.2
Epileptic Encephalopathy, Early Infantile, 53, EIEE53	617389	*SYNJ1*	NM_003895.3
Dystonia 3, Torsion, X-Linked, DYT3	314250	*TAF1*	NM_004606.3
Segawa Syndrome, Autosomal Recessive	605407	*TH*	NM_199292.2
Dystonia 6, Torsion, DYT6	602629	*THAP1*	NM_018105.2
Dystonia 1, Torsion, Autosomal Dominant, DYT1	128100	*TOR1A*	NM_000113.2
Dystonia 4, Torsion, Autosomal Dominant, DYT4	128101	*TUBB4A*	NM_006087.3
Striatonigral Degeneration, Childhood-Onset, SNDC	617054	*VAC14*	NM_018052.4
Choreoacanthocytosis, CHAC	200150	*VPS13A*	NM_033305.2
Neurodegeneration With Brain Iron Accumulation 5, NBIA5	300894	*WDR45*	NM_007075.3
Galloway-Mowat Syndrome 1, GAMOS1	251300	*WDR73*	NM_032856.3
Basal Ganglia Calcification, Idiopathic, 6, IBGC6	616413	*XPR1*	NM_004736.3
Gabriele-De Vries Syndrome, GADEVS	617557	*YY1*	NM_003403.4

Hereditary Parkinsonism

53 gene panel – non-NHS patients only

Condition	OMIM	Gene	Primary Transcript
Dystonia 24, DYT24	615034	*ANO3*	NM_031418.2
Kufor-Rakeb Syndrome, KRS	606693	*ATP13A2*	NM_022089.3
Cerebellar Ataxia, Areflexia, Pes Cavus, Optic Atrophy, And Sensorineural Hearing Loss;, CAPOS	601338	*ATP1A3*	NM_152296.4
Neurodegeneration With Brain Iron Accumulation 4, SPG43 / NBIA4	614298	*C19orf12*	NM_001031726.3
Parkinson Disease 22, PARK22	616710	*CHCHD2*	NM_016139.3
Msa1, Susceptibility To, MSA1	146500	*COQ2*	NM_015697.7
Aceruloplasminemia	604290	*CP*	NM_000096.3
Leukoencephalopathy, Hereditary Diffuse, With Spheroids, HDLS	221820	*CSF1R*	NM_005211.3
Perry Syndrome	168605	*DCTN1*	NM_004082.4
Parkinson Disease 19, Juvenile-Onset, PARK19	615528	*DNAJC6*	NM_001256864.1
Parkinson Disease 21, PARK21	616361	*DNAJC13*	NM_015268.3
Parkinson Disease 15, Autosomal Recessive Early-Onset, PARK15	260300	*FBXO7*	NM_012179.3
Neurodegeneration With Brain Iron Accumulation 3, NBIA3	606159	*FTL*	NM_000146.3

Hereditary Parkinsonism

53 gene panel – non-NHS patients only

Condition	OMIM	Gene	Primary Transcript
Parkinson Disease, Late-Onset, PD	168600	*GBA*	NM_001005741.2
Dystonia, Dopa-Responsive, DRD	128230	*GCH1*	NM_000161.2
Frontotemporal Lobar Degeneration With TDP43 Inclusions, GRN-Related	607485	*GRN*	NM_002087.3
Tay-Sachs Disease, GM2-Gangliosidosis, Several Forms, TSD	272800	*HEXA*	NM_000520.5
Sandhoff Disease	268800	*HEXB*	NM_000521.3
Parkinson Disease 13, Autosomal Dominant, Susceptibility To, PARK13	610297	*HTRA2*	NM_013247.4
Parkinson Disease 8, Autosomal Dominant, PARK8	607060	*LRRK2*	NM_198578.3
Parkinson-Dementia Syndrome	260540	*MAPT*	NM_005910.5
Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1		*NOTCH3*	NM_000435.2
3-Methylglutaconic Aciduria, Type III, MGCA3	258501	*OPA3*	NM_025136.3
Neurodegeneration With Brain Iron Accumulation 1, NBIA1	234200	*PANK2*	NM_153638.3
Hypoprebetalipoproteinemia, Acanthocytosis, Retinitis Pigmentosa, And Pallidal Degeneration,	607236	*PANK2*	NM_153638.3
Parkinson Disease 7, Autosomal Recessive Early-Onset, PARK7	606324	*PARK7*	NM_007262.4
Parkinson Disease 6, Autosomal Recessive Early-Onset, PARK6	605909	*PINK1*	NM_032409.2
Neurodegeneration With Brain Iron Accumulation 2b, NBIA2A/2B	610217	*PLA2G6*	NM_003560.3
Parkinson Disease 2, Autosomal Recessive Juvenile, PARK2	600116	*PRKN*	NM_004562.2
Dystonia 16	612067	*PRKRA*	NM_003690.4
Alzheimer Disease 3	607822	*PSEN1*	NM_000021.3
Alzheimer Disease 4	606889	*PSEN2*	NM_000447.2
Waisman Syndrome, WSMN	311510	*RAB39B*	NM_171998.3
Parkinsonism-Dystonia, Infantile, PKDYS	613135	*SLC6A3*	NM_001044.4
Basal Ganglia Calcification, Idiopathic, 1, IBGC1	213600	*SLC20A2*	NM_006749.4
Hypermanganesemia With Dystonia 1, HMNDYT1	613280	*SLC30A10*	NM_018713.2
Hypermanganesemia With Dystonia 2, HMNDYT2	608736	*SLC39A14*	NM_015359.5
Niemann-Pick Disease, Type B	607616	*SMPD1*	NM_000543.4
Niemann-Pick Disease, Type A	257200	*SMPD1*	NM_000543.4
Parkinson Disease 1, Autosomal Dominant, PARK1	168601	*SNCA*	NM_000345.3
Parkinson Disease 4, Autosomal Dominant, PARK4	605543	*SNCA*	NM_000345.3
Dementia, Lewy Body, DLB	127750	*SNCB*	NM_001001502.2
Juvenille Parkinsonism		*SPG11*	NM_025137.1
Dystonia, Dopa-Responsive, Due To Sepiapterin Reductase Deficiency	612716	*SPR*	NM_003124.4

Hereditary Parkinsonism

53 gene panel – non-NHS patients only

Condition	OMIM	Gene	Primary Transcript
Parkinson Disease 20, Early-Onset, PARK20	615530	*SYNJ1*	NM_003895.3
Dystonia 3, Torsion, X-Linked, DYT3	314250	*TAF1*	NM_004606.3
Segawa Syndrome, Autosomal Recessive	605407	*TH*	NM_199292.2
Dystonia 6, Torsion, DYT6	602629	*THAP1*	NM_018105.2
Dystonia 1, Torsion, Autosomal Dominant, DYT1	128100	*TOR1A*	NM_000113.2
Dystonia 4, Torsion, Autosomal Dominant, DYT4	128101	*TUBB4A*	NM_006087.3
Parkinson Disease Type 5, PARK5	613643	*UCHL1*	NM_004181.4
Parkinson Disease 23, Autosomal Recessive Early-Onset; PARK23, PARK23	616840	*VPS13C*	NM_020821.2
Chorea-Acanthocytosis, CHAC	200150	*VPS13A*	NM_033305.2
Parkinson Disease 17, PARK17	614203	*VPS35*	NM_018206.5
Neurodegeneration With Brain Iron Accumulation 5, NBIA5	300894	*WDR45*	NM_007075.3
Basal Ganglia Calcification, Idiopathic, 6, IBGC6	616413	*XPR1*	NM_004736.3

Hereditary Chorea 30 gene panel

non NHS patients only

Condition	OMIM	Gene	Primary Transcript
Dyskinesia, Familial, With Facial Myokymia, FDFM	606703	*ADCY5*	NM_183357.2
Ataxia Telangiectasia, ATM	208900	*ATM*	NM_000051.3
Wilson Disease, WD	277900	*ATP7B*	NM_000053.3
Cerebellar Ataxia, Mental Retardation, And Dysequilibrium Syndrome 4, CAMRQ4	615268	*ATP8A2*	NM_016529.5
Aceruloplasminemia	604290	*CP*	NM_000096.3
Rett Syndrome, Congenital Variant	613454	*FOXG1*	NM_005249.4
Friedreich Ataxia 1, FRDA	229300	*FXN*	NM_000144.4
Gm2-Gangliosidosis, Ab Variant,	272750	*GM2A*	NM_000405.4
Neurodevelopmental Disorder With Involuntary Movements, NEDIM	617493	*GNAO1*	NM_020988.2
Chorea, Benign Hereditary, BHC	118700	*NKX2-1*	NM_001079668.2
3-Methylglutaconic Aciduria, Type III, MGCA3	258501	*OPA3*	NM_025136.3
Dyskinesia, Limb And Orofacial, Infantile-Onset, IOLOD	616921	*PDE10A*	NM_001130690.2
Basal Ganglia Calcification, Idiopathic, 4, IBGC4	615007	*PDGFRB*	NM_002609.3
Paroxysmal Nonkinesigenic Dyskinesia 1, PNKD1, DYT8	118800	*PNKD*	NM_015488.4
Huntington Disease-Like 1, HDL1	603218	*PRNP*	NM_000311.4
Microphthalmia, Syndromic 12, MCOPS12	615524	*RARB*	NM_000965.4
Gordon Holmes Syndrome, GDHS	212840	*RNF216*	NM_207111.3

Hereditary Chorea 30 gene panel
non NHS patients only

Condition	OMIM	Gene	Primary Transcript
Epileptic Encephalopathy, Early Infantile, 6, EIEE6	607208	*SCN1A*	NM_001165963.1
Epileptic Encephalopathy, Early Infantile, 13, EIEE13	614558	*SCN8A*	NM_014191.3
Pontocerebellar Hypoplasia, Type 2d, PCH2D	613811	*SEPSECS*	NM_016955.3
Parkinsonism-Dystonia, Infantile, PKDYS	613135	*SLC6A3*	NM_001044.4
Basal Ganglia Calcification, Idiopathic, 1, IBGC1	213600	*SLC20A2*	NM_006749.4
Severe Motor Delay And Intellectual Disability		*SYT1*	NM_005639.2
Pontocerebellar Hypoplasia, Type 2b, PCH2B	612389	*TSEN2*	NM_025265.3
Pontocerebellar Hypoplasia, Type 2a, PCH2A	277470	*TSEN34*	NM_024075.4
Pontocerebellar Hypoplasia, Type 2a, PCH2A	277470	*TSEN54*	NM_207346.2
Pontocerebellar Hypoplasia, Type 2e, Parkinson Disease Type 17, PCH2E	615851	*VPS53*	NM_001128159.2
Chorea-Acanthocytosis, CHAC	200150	*VPS13A*	NM_033305.2
Mcleod Syndrome, MCLDS	300842	*XK*	NM_021083.3
Basal Ganglia Calcification, Idiopathic, 6, IBGC6	616413	*XPR1*	NM_004736.3

Abnormal Metal Transport
9 gene panel non-NHS patients only

Condition	OMIM	Gene	Primary Transcript
Wilson Disease, WD	277900	*ATP7B*	NM_000053.3
Neurodegeneration With Brain Iron Accumulation 4, SPG43 / NBIA4	614298	*C19orf12*	NM_001031726.3
Aceruloplasminemia	604290	*CP*	NM_000096.3
Neurodegeneration With Brain Iron Accumulation 3, NBIA3	606159	*FTL*	NM_000146.3
Neurodegeneration With Brain Iron Accumulation 1, NBIA1	234200	*PANK2*	NM_153638.3
Neurodegeneration With Brain Iron Accumulation 2b, NBIA2A/2B	610217	*PLA2G6*	NM_003560.3
Hypermanganesemia With Dystonia 1, HMNDYT1	613280	*SLC30A10*	NM_018713.2
Hypermanganesemia With Dystonia 2, HMNDYT2	608736	*SLC39A14*	NM_015359.5
Neurodegeneration With Brain Iron Accumulation 5, NBIA5	300894	*WDR45*	NM_007075.3

Paroxysmal neurological disorders

(SureSelect Design ID: 3097261)

Hemiplegic Migraine
10 gene panel – non-NHS patients only

Condition	OMIM Gene Ref	Gene	Primary transcript
Familial hemiplegic migraine type 2; Alternating hemiplegia of childhood type 1	182340	*ATP1A2*	NM_000702.3
Alternating hemiplegia of childhood type 2	182350	*ATP1A3*	NM_152296.4
Familial hemiplegic migraine, type 1 (with progressive cerebellar ataxia)	601011	*CACNA1A*	NM_001127221.1
Susceptibility to migraine type 6, with or without aura	601013	*CACNA1E*	NM_001205293.1
Susceptibility to migraine type 13, with or without aura	613655	*KCNK18*	NM_181840.1
Seizures, benign familial infantile, 2	614386	*PRRT2*	NM_145239.2
Familial hemiplegic migraine, type 3	182389	*SCN1A*	NM_001165963.1
Episodic ataxia, type 6	600111	*SLC1A3*	NM_004172.4
Dystonia 9 GLUT1 deficiency syndrome	138140	*SLC2A1*	NM_006516.2
Retinal vasculopathy with cerebral leukodystrophy	606609	*TREX1*	NM_033629.4

Episodic Ataxia 11 gene panel

non-NHS patients only

Condition	OMIM Gene Ref	Gene	Primary transcript
Episodic ataxia type 2 Spinocerebellar ataxia type 6 Early infantile epileptic encephalopathy type 42	601011	CACNA1A	NM_001127221.1
Episodic ataxia type 5	601949	CACNB4	NM_000726.3
Episodic ataxia type1 (including episodic ataxia / myokymia syndrome)	176260	KCNA1	NM_000217.2
Episodic kinesigenic dyskinesia 1	614386	PRRT2	NM_145239.2
Episodic ataxia type 6	600111	SLC1A3	NM_004172.4
Dystonia 9 GLUT1 deficiency syndrome	138140	SLC2A1	NM_006516.2
Spinocerebellar ataxia type 27	601515	FGF14	NM_175929.2
Intellectual disability, ataxia and arthrogryposis	611549	NALCN	NM_052867.2
Early infantile epileptic encephalopathy type 11	182390	SCN2A	NM_021007.2
Early infantile epileptic encephalopathy type 4	602926	STXBP1	NM_003165.3
Episodic ataxia type 8	609890	UBR4	NM_020765.2

Hereditary Ataxia Stages 1 and 2

(SureSelect Design ID: 3097261)

Hereditary Ataxia (Stage 1)

107 gene panel

Condition	OMIM	Inheritance	Gene	Primary Transcript

Hereditary Ataxia (Stage 1)

107 gene panel

Condition	OMIM	Inheritance	Gene	Primary Transcript
Achalasia-Addisonianism-Alacrima syndrome; AAAS; AAAS	231550	AR	*AAAS*	NM_015665.5
Anemia, sideroblastic and spinocerebellar ataxia; ASAT	301310	XL	*ABCB7*	NM_004299.4
PolNEYuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract; PHARC	612674	AR	*ABHD12*	NM_001042472.2
Coenzyme Q10 deficiency, primary, 4; CoQ10D4	612016	AR	*ADCK3*	NM_020247.4
Spinocerebellar ataxia 28 ; SCA28	610246	AD	*AFG3L2*	NM_006796.2
Spastic ataxia 5, autosomal recessive; SPAX5	614487	AR	*AFG3L2*	NM_006796.2
Spastic paraplegia 63; SPG63	615686	AR	*AMPD2*	NM_001257360.1
Spinocerebellar ataxia, autosomal recessive 10; SCAR10	613728	AR	*ANO10*	NM_018075.3
Pettigrew syndrome; PGS	304340	XL	*AP1S2*	NM_001272071.1
Ataxia, early onset, with occulomotor apraxia and hypoalbumine mia; EAOH	606350	AR	*APTX*	NM_175073.4
Metachromatic leukodystrophy; MLD	250100	AR	*ARSA*	NM_000487.5
Cerebellar ataxia, cayman type; ATCAY; ATCAY	601238	AR	*ATCAY*	NM_033064.4
Ataxia-telangiectasia; AT	208900	AR	*ATM*	NM_000051.3
Cerebellar ataxia, areflexia, pes cavus, optic atrophy, and sensorineural hearing loss; CAPOS	601338	AD	*ATP1A3*	NM_152296.4
Dystonia 12; DYT12	128235	AD	*ATP1A3*	NM_152296.4
Alternating hemiplegia of childhood 2; AHC2	614820	AD	*ATP1A3*	NM_152296.4
Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 3; CAMRQ3	613227	AR	*CA8*	NM_004056.4
Episodic ataxia, type 2; EA2	601011	AD	*CACNA1A*	NM_001127221.1
Spinocerebellar ataxia type 42; SCA42	604065	AD	*CACNA1G*	NM_018896.4
Cerebellar ataxia, nonprogressive, with mental retardation; CANPMR	614756	AD	*CAMTA1*	NM_015215.3
Mental retardation and microcephaly with pontine and cerebellar hypoplasia; micpch; MICPCH	300749	XL	*CASK*	NM_003688.3
Pontocerebellar hypoplasia, type 8; PCH8	614961	AR	*CHMP1A*	NM_002768.4
Cerebellar ataxia		AR	*CLN6*	NM_017882.2
Mitochondrial complex iv deficiency	220110	AR	*COX20*	NM_198076.4
Aceruloplasminemia	604290	AR	*CP*	NM_000096.3
Pinocerebellar ataxia, autosomal recessive 17; SCAR17	616127	AR	*CWF19L1*	NM_018294.5
Spastic paraplegia 56, autosomal recessive; SPG56	615030	AR	*CYP2U1*	NM_183075.2

Hereditary Ataxia (Stage 1)

107 gene panel

Condition	OMIM	Inheritance	Gene	Primary Transcript
Cerebrotendinous xanthomatosis; CTX	606530	AR	*CYP27A1*	NM_000784.3
Leukoencephalopathy with brainstem and spinal cord involvement and lactate elevation; lbsl; LBSL	611105	AR	*DARS2*	NM_018122.4
Spastic paraplegia 54, autosomal recessive; SPG54	615033	AR	*DDHD2*	NM_015214.2
Ceroid lipofuscinosis, neuronal, 4B, Parry type	162350	AD	*DNAJC5*	NM_025219.2
Cerebellar ataxia, deafness, and narcolepsy; ADCADN	604121	AD	*DNMT1*	NM_001005360.2
Leukoencephalopathy with vanishing white matter; VWN	603896	AR	*EIF2B1*	NM_001414.3
Leukoencephalopathy with vanishing white matter; VWN	603896	AR	*EIF2B2*	NM_014239.3
Leukoencephalopathy with vanishing white matter; VWN	603896	AR	*EIF2B3*	NM_020365.4
Leukoencephalopathy with vanishing white matter; VWN	603896	AR	*EIF2B4*	NM_015636.3
Leukoencephalopathy with vanishing white matter; VWN	603896	AR	*EIF2B5*	NM_003907.2
Spinocerebellar ataxia 34; SCA34	133190	AD	*ELOVL4*	NM_022726.3
Epilepsy, progressive myoclonic 2A (Lafora)	254780	AR	*EPM2A*	NM_005670.3
Pontocerebellar hypoplasia, type 1b; PCH1B	614678	AR	*EXOSC3*	NM_016042.3
Spinocerebellar ataxia 27; SCA27	609307	AD	*FGF14*	NM_175929.2
Posterior column ataxia with retinitis pigmentosa; AXPC1; AXPC1	609033	AR	*FLVCR1*	NM_014053.3
Neurodegeneration due to cerebral folate transport deficiency	613068	AR	*FOLR1*	NM_016725.2
Friedreich ataxia (including Friedreich ataxia with retained reflexes); FRDA (FARR)	606829	AR	*FXN*	NM_000144.4
Spastic paraplegia 46, autosomal recessive; SPG46	609471	AR	*GBA2*	NM_020944.2
Spastic paraplegia 44, autosomal recessive; SPG44	613206	AR	*GJC2*	NM_020435.3
Epilepsy, progressive myoclonic, 6; EPM6; EPM6	614018	AR	*GOSR2*	NM_004287.3
Spinocerebellar ataxia, autosomal recessive 18; SCAR18	616204	AR	*GRID2*	NM_001510.3
Spinocerebellar ataxia, autosomal recessive 13; SCAR13	614831	AR	*GRM1*	NM_001278064.1
Tay-Sachs disease, GM2-gangliosidosis, several forms; TSD	272800	AR	*HEXA*	NM_000520.5
Sandhoff disease;	268800	AR	*HEXB*	NM_000521.3

Hereditary Ataxia (Stage 1)

107 gene panel

Condition	OMIM	Inheritance	Gene	Primary Transcript
Spinocerebellar ataxia 15; SCA15	147265	AD	*ITPR1*	NM_001168272.1
Spinocerebellar ataxia 29, congenital nonprogressive; SCA29		?	*ITPR1*	NM_001168272.1
Episodic ataxia, type1 (including episodic ataxia/myokymia syndrome); EA1	176260	AD	*KCNA1*	NM_000217.2
Spinocerebellar ataxia 13; SCA13	176264	AD	*KCNC3*	NM_004977.2
Spinocerebellar ataxia 19; SCA19	605411	AD	*KCND3*	NM_004980.4
Spinocerebellar ataxia 22; SCA22		AD	*KCND3*	NM_004980.4
Seizures, sensorineural deafness, ataxia, mental retardation, and electrolyte imbalance; sesames; SESAMES	612780	AR	*KCNJ10*	NM_002241.4
Spastic ataxia 2, autosomal recessive; SPAX2	611302	AR	*KIF1C*	NM_006612.5
Spastic ataxia 3, autosomal recessive; SPAX3	611390	AR	*MARS2*	NM_138395.3
Methylmalonic aciduria and homocystinuria, CBLC type;	277400	AR	*MMACHC*	NM_015506.2
Ataxia-telangiectasia-like disorder 1; ATLD1	604391	AR	*MRE11A*	NM_005591.3
Abetalipoproteinemia; ABL	200100	AR	*MTTP*	NM_000253.3
Myoclonic Epilepsy Of Lafora;	254780	AR	*NHLRC1*	NM_198586.2
Niemann-pick disease, type C1; NPC1	257220	AR	*NPC1*	NM_000271.4
Niemann-pick disease, type C2; NPC2	607625	AR	*NPC2*	NM_006432.3
Mental retardation, x-linked, with cerebellar hypoplasia and distinctive facial appearance; MRX60	300486	XL	*OPHN1*	NM_002547.2
Gillespie syndrome; GLSP	206700	AD	*PAX6*	NM_000280.4
Spinocerebellar ataxia 23; SCA23	131340	AD	*PDYN*	NM_024411.4
Peroxisome biogenesis disorder 8a (Zellweger); PBD8A	614876	AR	*PEX16*	NM_004813.2
Peroxisome biogenesis disorder 8b; PBD8B	614877	AR	*PEX16*	NM_004813.2
Neurodegeneration with brain iron accumulation 2b; NBIA2A/2B	610217	AR	*PLA2G6*	NM_003560.3
Spinocerebellar ataxia, autosomal recessive 2; SCAR2	213200	AR	*PMPCA*	NM_015160.2
Ataxia-oculomotor apraxia 4; AOA4	616267	AR	*PNKP*	NM_007254.3
Spastic paraplegia 39; SPG39	612020	AR	*PNPLA6*	NM_006702.4
Sensory ataxic neuropathy, dysarthria, and ophthalmoparesis; SANDO	607459	AR	*POLG*	NM_002693.2
Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism; HLD7	607694	AR	*POLR3A*	NM_007055.3
Spinocerebellar ataxia 14; SCA14	176980	AD	*PRKCG*	NM_002739.3
Huntington disease-like 1; HDL1	603218	AD	*PRNP*	NM_000311.4

Hereditary Ataxia (Stage 1)

107 gene panel

Condition	OMIM	Inheritance	Gene	Primary Transcript
Episodic kinesigenic dyskinesia 1 (Dystonia 10); EKD1 /DYT10	614386	AD	*PRRT2*	NM_145239.2
Gordon Holmes syndrome; GDHS	212840	AR	*RNF216*	NM_207111.3
Autosomal dominant sensory ataxia-1 (SNAX1); SNAX1	608984	AD	*RNF170*	NM_001160223.1
Spastic ataxia, Charlevoix-Saguenay type; SACS; SACS	604490	AR	*SACS*	NM_014363.5
Chylomicron retention disease; CMRD	246700	AR	*SAR1B*	NM_001033503.2
Pontocerebellar hypoplasia, type 2D; PCH2D	613811	AR	*SEPSECS*	NM_016955.3
Spinocerebellar ataxia, autosomal recessive 1; SCAR1	608465	AR	*SETX*	NM_015046.5
(Ataxia-oculomotor apraxia-2; AOA2); AOA2		AR	*SETX*	NM_015046.5
Marinesco-Sjogren syndrome; MSS; MSS	608005	AR	*SIL1*	NM_001037633.1
Episodic ataxia, type 6; EA6	600111	AD	*SLC1A3*	NM_004172.4
Dystonia 9; DYT9	138140	AD	*SLC2A1*	NM_006516.2
Mental retardation, x-linked, syndromic, christianson type; MRXSCH; MRXSCH	300243	XLD	*SLC9A6*	NM_006359.2
Spinocerebellar ataxia, autosomal recessive 20; SCAR20	616354	AR	*SNX14*	NM_153816.5
Spastic paraplegia 7, autosomal recessive; SPG7	602783	AR	*SPG7*	NM_003119.3
Spinocerebellar ataxia 5; SCA5	604985	AR	*SPTBN2*	NM_006946.2
Spinocerebellar ataxia, autosomal recessive 14; SCAR14		AD	*SPTBN2*	NM_006946.2
Congenital disorder of glycosylation, type 1Q; CDG1Q	611715	AR	*SRD5A3*	NM_024592.4
Spinocerebellar ataxia, autosomal recessive 16; SCAR16	615768	AR	*STUB1*	NM_005861.3
Spinocerebellar ataxia, autosomal recessive 8; SCAR8	610743	AR	*SNEY1*	NM_033071.3
Spinocerebellar ataxia 35; SCA35	613900	AD	*TGM6*	NM_198994.2
Spinocerebellar ataxia 21; SCA21	607454	AD	*TMEM240*	NM_001114748.1
Spinocerebellar ataxia, autosomal recessive 7; SCAR7	609270	AR	*TPP1*	NM_000391.3
Pontocerebellar hypoplasia, type 2b; PCH2B	612389	AR	*TSEN2*	NM_025265.3
Pontocerebellar hypoplasia, type 2a; PCH2A	277470	AR	*TSEN54*	NM_207346.2
Spinocerebellar ataxia 11; SCA11	611695	AD	*TTBK2*	NM_173500.3
Mitochondrial complex III deficiency, nuclear type 2; MC3DN2	615157	AR	*TTC19*	NM_017775.3
Vitamin E, familial isolated deficiency of; VED	600415	AR	*TTPA*	NM_000370.3

Hereditary Ataxia (Stage 1)

107 gene panel

Condition	OMIM	Inheritance	Gene	Primary Transcript
Dystonia 4, torsion, autosomal dominant; DYT4	128101	AD	*TUBB4A*	NM_006087.3
Spinocerebellar ataxia, infantile-onset; IOSCA	606075	AR	*TWNK*	NM_006087.3
Spastic ataxia 1, autosomal dominant; SPAX1	185880	AD	*VAMP1*	NM_014231.4
Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 1; CAMRQ1	224050	AR	*VLDLR*	NM_003383.3
Pontocerebellar hypoplasia, type 1a; PCH1A	607596	AR	*VRK1*	NM_003384.2
Galloway-mowat syndrome; GAMOS	251300	AR	*WDR73*	NM_032856.3
Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2; CAMRQ2	610185	AR	*WDR81*	NM_001163809.1
Wolfram syndrome 1; WFS1	222300	AR	*WFS1*	NM_006005.3
Spinocerebellar ataxia, autosomal recessive 12; SCAR12	605131	AR	*WWOX*	NM_016373.3

Hereditary Ataxia (Stage 2)

54 gene panel

Condition	OMIM	Inheritance	Gene	Primary Transcript
Joubert syndrome 3; JBTS3	608629	AR	AHI1	NM_017651.4
Wilson disease; WD	606882	AR	ATP7B	NM_000053.3
Joubert syndrome 17; JBTS17	614615	AR	C5orf42	NM_023073.3
Coach syndrome;	216360	AR	CC2D2A	NM_001080522.2
Joubert syndrome 15; JBTS15	614464	AR	CEP41	NM_018718.2
Joubert syndrome 5; JBTS5	610188	AR	CEP290	NM_025114.3
Spinocerebellar ataxia, autosomal recessive 7; SCAR7	609270	AR	COPRS	NM_018405.3
Mitochondrial complex iv deficiency;	220110	AR	COX14	NM_032901.3
Mitochondrial complex iv deficiency or Leigh syndrome;	220110	AR	COX10	NM_001303.3
Mitochondrial complex iv deficiency;	220110	AR	COX6B1	NM_001863.4
Msa1, susceptibility to; MSA1	146500	AR/AD	COQ2	NM_015697.7
Coenzyme Q10 deficiency, primary, 5; COQ10D5	614654	AR	COQ9	NM_020312.3
Mental retardation, autosomal dominant 19; MRD19	615075	AD	CTNNB1	NM_001904.3
Joubert syndrome 21; JBTS21	615636	AR	CSPP1	NM_024790.6
Pyruvate dehydrogenase e2 deficiency; PDHDD	245348	AR	DLAT	NM_001931.4
Spinocerebellar ataxia 26; SCA26	130610	AD	EEF2	NM_001961.3
Mitochondrial complex iv deficiency;	220110	AR	FASTKD2	NM_014929.3

Hereditary Ataxia (Stage 2)

54 gene panel

Condition	OMIM	Inheritance	Gene	Primary Transcript
Neurodegeneration with brain iron accumulation 3; NBIA3;	134790	AD	FTL	NM_000146.3
Krabbe disease	245200	AR	*GALC*	NM_000153.3
Gaucher disease, type I	230800	AR	*GBA*	NM_001005741.2
Frontotemporal lobar degeneration with TDP43 inclusions	607485	AR	*GRN*	NM_002087.3
Spinocerebellar ataxia 18; SCA18	603502	AD	*IFRD1*	NM_001550.3
Sensorimotor neuropathy with ataxia; SMNA	603502	AD	*IFRD1*	NM_001550.3
Joubert syndrome 1; JBTS1	213300	AR	*INPP5E*	NM_019892.4
Early-onset epileptic encephalopathy		AD	*KCNA2*	NM_004974.3
Poretti-Boltshauser syndrome; PTBHS	615960	AR	*LAMA1*	NM_005559.3
No known		?	*LIG3*	NM_013975.3
Epilepsy, progressive myoclonic, 9; EPM9	616540	AR	*LMNB2*	NM_032737.3
Congenital contractures of the limbs and face, hypotonia, and developmental delay; CLIFAHDD	616266	AD	*NALCN*	NM_052867.2
Joubert syndrome 4; JBTS4	609583	AR	*NPHP1*	NM_000272.3
Coenzyme q10 deficiency, primary, 2; COQ10D2	614651	AR	*PDSS1*	NM_014317.3
Coenzyme q10 deficiency, primary, 3; COQ10D3	614652	AR	*PDSS2*	NM_020381.3
Mitochondrial complex iv deficiency	220110	AR	*PET100*	NM_001171155.1
Peroxisome biogenesis disorder 5b; PBD5B	614867	AR	*PEX2*	NM_000318.2
Peroxisome biogenesis disorder 4a (Zellweger); PBD4A	614862	AR	*PEX6*	NM_000287.3
Not known		AR	*PITRM1*	NM_001242309.1
Coach syndrome	216360	AR	*RPGRIP1L*	NM_015272.2
Epileptic encephalopathy, early infantile, 11; EIEE11	613721	AD	*SCN2A*	NM_021007.2
Mitochondrial complex iv deficiency	220110	AR	*SCO1*	NM_004589.3
Spinocerebellar ataxia, autosomal recessive 21; SCAR21; SCAR21	616719	AR	*SCYL1*	NM_020680.3
Allan-Herndon-Dudley syndrome; AHDS; AHDS	300095	XL	*SLC16A2*	NM_006517.4
Basal ganglia calcification, idiopathic, 1; IBGC1	213600	AD	*SLC20A2*	NM_006749.4
Spinocerebellar ataxia with blindness and deafness		AR	*SLC52A2*	NM_024531.4
Niemann-pick disease, type b	607616	AR	*SMPD1*	NM_000543.4
Niemann-pick disease, type a	257200	AR	*SMPD1*	NM_000543.4

Hereditary Ataxia (Stage 2)
54 gene panel

Condition	OMIM	Inheritance	Gene	Primary Transcript
Epileptic encephalopathy, early infantile, 4; EIEE4	612164	AD	*STXBP1*	NM_003165.3
Mitochondrial complex iv deficiency	220110	AR	*TACO1*	NM_016360.3
Spinocerebellar ataxia, autosomal recessive 23; SCAR23	616949	AR	*TDP2*	NM_016614.2
Spastic paraplegia 49, autosomal recessive; SPG49	615031	AR	*TECPR2*	NM_014844.3
Joubert syndrome 6; JBTS6	610688	AR	*TMEM67*	NM_153704.5
Coach syndrome	216360	AR	*TMEM67*	NM_153704.5
Joubert syndrome 16; JBTS16	614465	AR	*TMEM138*	NM_016464.4
Joubert syndrome 2; JBTS2	608091	AR	*TMEM216*	NM_001173990.2
Joubert syndrome 20; JBTS20	614970	AR	*TMEM231*	NM_001077416.2
Wieacker-Wolff syndrome; WRWF	341580	XLR	*ZC4H2*	NM_018684.3

R56.1 :: Adult onset dystonia, chorea or related movement disorder
Panel: Adult onset movement disorder (Version 1.14)
https://panelapp.genomicsengland.co.uk/panels/540/
Agilent SureSelect Reagent Design ID: 3297121 – Laboratory work in YNEGLH Central Lab

Rating	Gene	HGNC	OMIM	Primary Transcript	Additional Transcripts
Green	*ACTB*	132	102630	NM_001101.4	
Green	*AFG3L2*	315	604581	NM_006796.2	
Green	*ANO3*	14004	610110	NM_031418.3	
Green	*APTX*	15984	606350	NM_175073.2	NM_001195248.1
Green	*ATM*	795	607585	NM_000051.3	
Green	*ATP13A2*	30213	610513	NM_022089.3	
Green	*ATP1A2*	800	182340	NM_000702.3	
Green	*ATP1A3*	801	182350	NM_152296.4	
Green	*ATP7B*	870	606882	NM_000053.3	
Green	*C19orf12*	25443	614297	NM_001031726.3	
Green	*CACNA1A*	1388	601011	NM_001127221.1	
Green	*CHMP2B*	24537	609512	NM_014043.3	
Green	*CP*	2295	117700	NM_000096.3	
Green	*CSF1R*	2433	164770	NM_005211.3	
Green	*CYP27A1*	2605	606530	NM_000784.3	
Green	*DCAF17*	25784	612515	NM_025000.3	
Green	*DCTN1*	2711	601143	NM_004082.4	
Green	*DNAJC6*	15469	608375	NM_001256864.1	
Green	*FBXO7*	13586	605648	NM_012179.3	

R56.1 :: Adult onset dystonia, chorea or related movement disorder
Panel: Adult onset movement disorder (Version 1.14)
https://panelapp.genomicsengland.co.uk/panels/540/
Agilent SureSelect Reagent Design ID: 3297121 – Laboratory work in YNEGLH Central Lab

Rating	Gene	HGNC	OMIM	Primary Transcript	Additional Transcripts
Green	*FTL*	3999	134790	NM_000146.3	
Green	*GBA*	4177	606463	NM_001005741.2	
Green	*GCH1*	4193	600225	NM_000161.2	
Green	*GFAP*	4235	137780	NM_002055.4	
Green	*GNAL*	4388	139312	NM_001142339.2	NM_182978.3
Green	*GRN*	4601	138945	NM_002087.3	
Green	*GTPBP2*	4670	607434	NM_019096.4	
Green	*HPCA*	5144	142622	NM_002143.2	
Green	*KMT2B*	15840	606834	NM_014727.2	
Green	*LRRK2*	18618	609007	NM_198578.3	
Green	*LYST*	1968	606897	NM_000081.3	
Green	*MAPT*	6893	157140	NM_005910.5	NM_016835.4
Green	*MYORG*	19918	618255	NM_020702.4	
Green	*NKX2-1*	11825	600635	NM_001079668.2	
Green	*PANK2*	15894	606157	NM_153638.3	
Green	*PARK7*	16369	602533	NM_007262.4	
Green	*PDE10A*	8772	610652	NM_001130690.2	NM_006661.3
Green	*PDGFB*	8800	190040	NM_002608.3	
Green	*PDGFRB*	8804	173410	NM_002609.3	
Green	*PINK1*	14581	608309	NM_032409.2	
Green	*PLA2G6*	9039	603604	NM_003560.3	
Green	*PNKD*	9153	609023	NM_015488.4	
Green	*PRKN*	8607	602544	NM_004562.2	
Green	*PRKRA*	9438	603424	NM_003690.4	
Green	*PRNP*	9449	176640	NM_000311.4	
Green	*PRRT2*	30500	614386	NM_145239.2	
Green	*RAB39B*	16499	300774	NM_171998.3	
Green	*RNF216*	21698	609948	NM_207111.3	
Green	*SGCE*	10808	604149	NM_003919.2	
Green	*SLC19A3*	16266	606152	NM_025243.3	
Green	*SLC20A2*	10947	158378	NM_006749.4	
Green	*SLC2A1*	11005	138140	NM_006516.2	
Green	*SLC30A10*	25355	611146	NM_018713.2	
Green	*SNCA*	11138	163890	NM_000345.3	
Green	*SPG11*	11226	610844	NM_025137.3	
Green	*SPR*	11257	182125	NM_003124.4	
Green	*SYNJ1*	11503	604297	NM_003895.3	

R56.1 :: Adult onset dystonia, chorea or related movement disorder
Panel: Adult onset movement disorder (Version 1.14)
https://panelapp.genomicsengland.co.uk/panels/540/
Agilent SureSelect Reagent Design ID: 3297121 – Laboratory work in YNEGLH Central Lab

Rating	Gene	HGNC	OMIM	Primary Transcript	Additional Transcripts
Green	*TBK1*	11584	604834	NM_013254.3	
Green	*THAP1*	20856	609520	NM_018105.2	
Green	*TIMM8A*	11817	300356	NM_004085.3	
Green	*TOR1A*	3098	605204	NM_000113.2	
Green	*TUBB4A*	20774	602662	NM_006087.3	
Green	*VPS13A*	1908	605978	NM_033305.2	
Green	*VPS35*	13487	601501	NM_018206.5	
Green	*WDR45*	28912	300526	NM_007075.3	
Green	*XPR1*	12827	605237	NM_004736.3	

R57.1 :: Childhood onset dystonia, chorea or related movement disorder
Panel: Childhood onset dystonia or chorea or related movement disorder (Version 1.58)
https://panelapp.genomicsengland.co.uk/panels/847/
Agilent SureSelect Reagent Design ID: 3297121 – Laboratory work in YNEGLH Central Lab

Rating	Gene	HGNC	OMIM	Primary Transcript	Additional Transcripts
Green	*ABAT*	23	137150	NM_020686.5	
Green	*ACOX1*	119	609751	NM_004035.6	NM_007292.5
Green	*ACTB*	132	102630	NM_001101.4	
Green	*ADAR*	225	146920	NM_001111.4	
Green	*ADCY5*	236	600293	NM_183357.2	
Green	*AFG3L2*	315	604581	NM_006796.2	
Green	*ALDH18A1*	9722	138250	NM_002860.3	
Green	*ANO3*	14004	610110	NM_031418.3	
Green	*AP1S2*	560	300629	NM_001272071.1	
Green	*APTX*	15984	606350	NM_175073.2	NM_001195248.1
Green	*ARSA*	713	607574	NM_000487.5	
Green	*ATM*	795	607585	NM_000051.3	
Green	*ATP13A2*	30213	610513	NM_022089.3	
Green	*ATP1A2*	800	182340	NM_000702.3	
Green	*ATP1A3*	801	182350	NM_152296.4	
Green	*ATP7B*	870	606882	NM_000053.3	
Green	*BCAP31*	16695	300398	NM_001139441.1	
Green	*BCS1L*	1020	603647	NM_004328.4	
Green	*C19orf12*	25443	614297	NM_001031726.3	
Green	*C9orf72*	28337	614260	NM_018325.4	
Green	*CACNA1A*	1388	601011	NM_001127221.1	
Green	*CACNA1G*	1394	604065	NM_018896.4	

R57.1 :: Childhood onset dystonia, chorea or related movement disorder
Panel: Childhood onset dystonia or chorea or related movement disorder (Version 1.58)
https://panelapp.genomicsengland.co.uk/panels/847/
Agilent SureSelect Reagent Design ID: 3297121 – Laboratory work in YNEGLH Central Lab

Rating	Gene	HGNC	OMIM	Primary Transcript	Additional Transcripts
Green	*CACNB4*	1404	601949	NM_000726.4	
Green	*CLN3*	2074	607042	NM_001042432.1	
Green	*CLN5*	2076	608102	NM_006493.2	
Green	*CLPB*	30664	616254	NM_030813.5	
Green	*COASY*	29932	609855	NM_025233.6	
Green	*COL6A3*	2213	120250	NM_004369.3	
Green	*COX10*	2260	602125	NM_001303.3	
Green	*COX15*	2263	603646	NM_078470.5	NM_004376.6
Green	*CSTB*	2482	601145	NM_000100.3	
Green	*DCAF17*	25784	612515	NM_025000.3	
Green	*DDC*	2719	107930	NM_000790.3	
Green	*DLAT*	2896	608770	NM_001931.4	
Green	*DLD*	2898	238331	NM_000108.4	
Green	*DNAJC12*	28908	606060	NM_021800.2	
Green	*ECHS1*	3151	602292	NM_004092.3	
Green	*FA2H*	21197	611026	NM_024306.4	
Green	*FBXO7*	13586	605648	NM_012179.3	
Green	*FOLR1*	3791	136430	NM_016725.2	
Green	*FOXRED1*	26927	613622	NM_017547.3	
Green	*FTL*	3999	134790	NM_000146.3	
Green	*FXN*	3951	606829	NM_000144.4	
Green	*GBA*	4177	606463	NM_001005741.2	
Green	*GCDH*	4189	608801	NM_000159.3	
Green	*GCH1*	4193	600225	NM_000161.2	
Green	*GJC2*	17494	608803	NM_020435.3	
Green	*GLB1*	4298	611458	NM_000404.3	
Green	*GLRA1*	4326	138491	NM_000171.3	
Green	*GM2A*	4367	613109	NM_000405.4	
Green	*GNAL*	4388	139312	NM_001142339.2	NM_182978.3
Green	*GNAO1*	4389	139311	NM_020988.2	
Green	*GTPBP2*	4670	607434	NM_019096.4	
Green	*HCFC1*	4839	300019	NM_005334.2	
Green	*HEXA*	4878	606869	NM_000520.5	
Green	*HIBCH*	4908	610690	NM_014362.3	
Green	*HPCA*	5144	142622	NM_002143.2	
Green	*HSPD1*	5261	118190	NM_002156.4	
Green	*HTRA2*	14348	606441	NM_013247.4	
Green	*IFIH1*	18873	606951	NM_022168.3	

R57.1 :: Childhood onset dystonia, chorea or related movement disorder
Panel: Childhood onset dystonia or chorea or related movement disorder (Version 1.58)
https://panelapp.genomicsengland.co.uk/panels/847/
Agilent SureSelect Reagent Design ID: 3297121 – Laboratory work in YNEGLH Central Lab

Rating	Gene	HGNC	OMIM	Primary Transcript	Additional Transcripts
Green	*KCNA1*	6218	176260	NM_000217.2	
Green	*KCNMA1*	6284	600150	NM_002247.3	
Green	*KCNQ2*	6296	602235	NM_172107.3	
Green	*KCTD17*	25705	616386	NM_001282684.1	
Green	*KIF1C*	6317	603060	NM_006612.5	
Green	*KMT2B*	15840	606834	NM_014727.2	
Green	*LRPPRC*	15714	607544	NM_133259.3	
Green	*MARS2*	25133	609728	NM_138395.3	
Green	*MECR*	19691	608205	NM_016011.4	
Green	*MRE11*	7230	600814	NM_005591.3	
Green	*MTFMT*	29666	611766	NM_139242.3	
Green	*NDUFA1*	7683	300078	NM_004541.3	
Green	*NDUFA10*	7684	603835	NM_004544.3	
Green	*NDUFAF5*	15899	612360	NM_024120.4	
Green	*NDUFAF6*	28625	612392	NM_152416.3	
Green	*NDUFS1*	7707	157655	NM_005006.6	
Green	*NDUFS4*	7711	602694	NM_002495.3	
Green	*NDUFS7*	7714	601825	NM_024407.4	
Green	*NDUFS8*	7715	602141	NM_002496.3	
Green	*NDUFV1*	7716	161015	NM_007103.3	
Green	*NGLY1*	17646	610661	NM_018297.3	
Green	*NKX2-1*	11825	600635	NM_001079668.2	
Green	*NKX6-2*	19321	605955	NM_177400.2	
Green	*NPC1*	7897	607623	NM_000271.4	
Green	*NPC2*	14537	601015	NM_006432.3	
Green	*OCLN*	8104	602876	NM_002538.3	
Green	*OPA3*	8142	606580	NM_025136.3	
Green	*PANK2*	15894	606157	NM_153638.3	
Green	*PCCA*	8653	232000	NM_000282.3	
Green	*PCCB*	8654	232050	NM_000532.4	NM_001178014.1
Green	*PDE10A*	8772	610652	NM_001130690.2	NM_006661.3
Green	*PDE2A*	8777	602658	NM_002599.4	
Green	*PDGFB*	8800	190040	NM_002608.3	
Green	*PDHA1*	8806	300502	NM_000284.3	NM_001173454.1
Green	*PDHX*	21350	608769	NM_003477.2	
Green	*PET100*	40038	614770	NM_001171155.1	
Green	*PINK1*	14581	608309	NM_032409.2	
Green	*PLA2G6*	9039	603604	NM_003560.3	

R57.1 :: Childhood onset dystonia, chorea or related movement disorder
Panel: Childhood onset dystonia or chorea or related movement disorder (Version 1.58)
https://panelapp.genomicsengland.co.uk/panels/847/
Agilent SureSelect Reagent Design ID: 3297121 – Laboratory work in YNEGLH Central Lab

Rating	Gene	HGNC	OMIM	Primary Transcript	Additional Transcripts
Green	*PNKD*	9153	609023	NM_015488.4	
Green	*PNKP*	9154	605610	NM_007254.3	
Green	*POLR3A*	30074	614258	NM_007055.3	
Green	*PRKN*	8607	602544	NM_004562.2	
Green	*PRKRA*	9438	603424	NM_003690.4	
Green	*PRNP*	9449	176640	NM_000311.4	
Green	*PRRT2*	30500	614386	NM_145239.2	
Green	*PTS*	9689	612719	NM_000317.2	
Green	*QDPR*	9752	612676	NM_000320.2	
Green	*RAB39B*	16499	300774	NM_171998.3	
Green	*RNASEH2B*	25671	610326	NM_024570.3	
Green	*RNASEH2C*	24116	610330	NM_032193.3	
Green	*RNASET2*	21686	612944	NM_003730.4	
Green	*SAMHD1*	15925	606754	NM_015474.3	
Green	*SCN8A*	10596	600702	NM_014191.3	NM_001330260.1
Green	*SERAC1*	21061	614725	NM_032861.3	
Green	*SETX*	445	608465	NM_015046.6	
Green	*SGCE*	10808	604149	NM_003919.2	
Green	*SLC19A3*	16266	606152	NM_025243.3	
Green	*SLC20A2*	10947	158378	NM_006749.4	
Green	*SLC2A1*	11005	138140	NM_006516.2	
Green	*SLC30A10*	25355	611146	NM_018713.2	
Green	*SLC39A14*	20858	608736	NM_015359.5	
Green	*SLC6A3*	11049	126455	NM_001044.4	
Green	*SLC6A8*	11055	300036	NM_005629.3	
Green	*SPR*	11257	182125	NM_003124.4	
Green	*SUCLA2*	11448	603921	NM_003850.2	
Green	*SUOX*	11460	606887	NM_000456.2	
Green	*SURF1*	11474	185620	NM_003172.3	
Green	*SYNJ1*	11503	604297	NM_003895.3	
Green	*TAF1*	11535	313650	NM_004606.4	
Green	*TH*	11782	191290	NM_199292.2	
Green	*THAP1*	20856	609520	NM_018105.2	
Green	*TIMM8A*	11817	300356	NM_004085.3	
Green	*TOR1A*	3098	605204	NM_000113.2	
Green	*TPK1*	17358	606370	NM_022445.3	
Green	*TREX1*	12269	606609	NM_033629.4	
Green	*TUBB4A*	20774	602662	NM_006087.3	

R57.1 :: Childhood onset dystonia, chorea or related movement disorder
Panel: Childhood onset dystonia or chorea or related movement disorder (Version 1.58)
https://panelapp.genomicsengland.co.uk/panels/847/
Agilent SureSelect Reagent Design ID: 3297121 – Laboratory work in YNEGLH Central Lab

Rating	Gene	HGNC	OMIM	Primary Transcript	Additional Transcripts
Green	*VAC14*	25507	604632	NM_018052.4	
Green	*VAMP1*	12642	185880	NM_014231.4	
Green	*VAMP2*	12643	185881	NM_014232.2	
Green	*VPS13A*	1908	605978	NM_033305.2	
Green	*VPS13D*	23595	608877	NM_015378.3	
Green	*WDR45*	28912	300526	NM_007075.3	
Green	*WDR73*	25928	616144	NM_032856.3	
Green	*YY1*	12856	600013	NM_003403.4	
Green	*ZSWIM6*	29316	615951	NM_020928.1	

R66.1 :: Paroxysmal central nervous system disorders
Panel: Paroxysmal central nervous system disorders (Version 1.2)
https://panelapp.genomicsengland.co.uk/panels/541/
Agilent SureSelect Reagent Design ID: 3297121 – Laboratory work in NEYGLH Central Lab

Rating	Gene	HGNC	OMIM	Primary Transcript	Additional Transcripts
Green	*ADCY5*	236	600293	NM_183357.2	
Green	*ATAD1*	25903	614452	NM_032810.3	
Green	*ATP1A2*	800	182340	NM_000702.3	
Green	*ATP1A3*	801	182350	NM_152296.4	
Green	*CACNA1A*	1388	601011	NM_001127221.1	
Green	*CSNK1D*	2452	600864	NM_001893.4	
Green	*DNMT1*	2976	126375	NM_001130823.2	
Green	*GLRA1*	4326	138491	NM_000171.3	
Green	*GLRB*	4329	138492	NM_000824.4	
Green	*KCNA1*	6218	176260	NM_000217.2	
Green	*KCNMA1*	6284	600150	NM_002247.3	
Green	*KCNQ2*	6296	602235	NM_172107.3	
Green	*PDE10A*	8772	610652	NM_001130690.2	NM_006661.3
Green	*PNKD*	9153	609023	NM_015488.4	
Green	*PRRT2*	30500	614386	NM_145239.2	
Green	*SCN1A*	10585	182389	NM_001165963.2	
Green	*SLC1A3*	10941	600111	NM_004172.4	
Green	*SLC2A1*	11005	138140	NM_006516.2	
Green	*SLC6A5*	11051	604159	NM_004211.4	
Green	*VAMP2*	12643	185881	NM_014232.2	

R58.1 :: Adult onset neurodegenerative disorder
Panel: Neurodegenerative disorders - adult onset (Version 2.31)
https://panelapp.genomicsengland.co.uk/panels/474/
Agilent SureSelect Reagent Design ID: 3297111 – Laboratory work in NEYGLH Central Lab

Rating	Gene	HGNC	OMIM	Primary Transcript	Additional Transcripts
Green	*ABCD1*	61	300371	NM_000033.3	
Green	*AFG3L2*	315	604581	NM_006796.2	
Green	*ALS2*	443	606352	NM_020919.3	
Green	*ANG*	483	105850	NM_001145.4	
Green	*ANXA11*	535	602572	NM_001157.2	
Green	*APP*	620	104760	NM_000484.3	
Green	*ARSA*	713	607574	NM_000487.5	
Green	*ATP13A2*	30213	610513	NM_022089.3	
Green	*ATP1A3*	801	182350	NM_152296.4	
Green	*ATP7B*	870	606882	NM_000053.3	
Green	*AUH*	890	600529	NM_001698.2	
Green	*C19orf12*	25443	614297	NM_001031726.3	
Green	*CACNA1G*	1394	604065	NM_018896.4	
Green	*CCNF*	1591	600227	NM_001761.2	
Green	*CHCHD10*	15559	615903	NM_213720.2	
Green	*CHCHD2*	21645	616244	NM_016139.3	
Green	*CHMP2B*	24537	609512	NM_014043.3	
Green	*CLCN2*	2020	600570	NM_004366.5	
Green	*CLN6*	2077	606725	NM_017882.2	
Green	*COASY*	29932	609855	NM_025233.6	
Green	*CP*	2295	117700	NM_000096.3	
Green	*CSF1R*	2433	164770	NM_005211.3	
Green	*CTSF*	2531	603539	NM_003793.3	
Green	*CYP27A1*	2605	606530	NM_000784.3	
Green	*CYP7B1*	2652	603711	NM_004820.4	
Green	*DARS2*	25538	610956	NM_018122.4	
Green	*DCTN1*	2711	601143	NM_004082.4	
Green	*DNAJC5*	16235	611203	NM_025219.2	
Green	*DNAJC6*	15469	608375	NM_001256864.1	
Green	*DNMT1*	2976	126375	NM_001130823.2	
Green	*EIF2B1*	3257	606686	NM_001414.3	
Green	*EIF2B2*	3258	606454	NM_014239.3	
Green	*EIF2B3*	3259	606273	NM_020365.4	
Green	*EIF2B4*	3260	606687	NM_015636.3	
Green	*EIF2B5*	3261	603945	NM_003907.2	
Green	*ELOVL4*	14415	605512	NM_022726.3	
Green	*EPM2A*	3413	607566	NM_005670.3	
Green	*FBXO7*	13586	605648	NM_012179.3	
Green	*FIG4*	16873	609390	NM_014845.5	
Green	*FTL*	3999	134790	NM_000146.3	

R58.1 :: Adult onset neurodegenerative disorder
Panel: Neurodegenerative disorders - adult onset (Version 2.31)
https://panelapp.genomicsengland.co.uk/panels/474/
Agilent SureSelect Reagent Design ID: 3297111 – Laboratory work in NEYGLH Central Lab

Rating	Gene	HGNC	OMIM	Primary Transcript	Additional Transcripts
Green	*FUS*	4010	137070	NM_004960.3	
Green	*GCH1*	4193	600225	NM_000161.2	
Green	*GFAP*	4235	137780	NM_002055.4	
Green	*GRN*	4601	138945	NM_002087.3	
Green	*HEXA*	4878	606869	NM_000520.5	
Green	*HEXB*	4879	606873	NM_000521.3	
Green	*HNRNPA1*	5031	164017	NM_031157.3	
Green	*HTRA1*	9476	602194	NM_002775.4	
Green	*ITM2B*	6174	603904	NM_021999.4	
Green	*KCNC3*	6235	176264	NM_004977.2	
Green	*KCND3*	6239	605411	NM_004980.4	
Green	*KIF5A*	6323	602821	NM_004984.3	
Green	*LRRK2*	18618	609007	NM_198578.3	
Green	*LYST*	1968	606897	NM_000081.3	
Green	*MAPT*	6893	157140	NM_005910.5	NM_016835.4
Green	*MYORG*	19918	618255	NM_020702.4	
Green	*NHLRC1*	21576	608072	NM_198586.2	
Green	*NOTCH3*	7883	600276	NM_000435.2	
Green	*NPC1*	7897	607623	NM_000271.4	
Green	*NPC2*	14537	601015	NM_006432.3	
Green	*OPTN*	17142	602432	NM_001008211.1	
Green	*PANK2*	15894	606157	NM_153638.3	
Green	*PARK7*	16369	602533	NM_007262.4	
Green	*PDGFB*	8800	190040	NM_002608.3	
Green	*PDGFRB*	8804	173410	NM_002609.3	
Green	*PFN1*	8881	176610	NM_005022.3	
Green	*PINK1*	14581	608309	NM_032409.2	
Green	*PLA2G6*	9039	603604	NM_003560.3	
Green	*PRKN*	8607	602544	NM_004562.2	
Green	*PRNP*	9449	176640	NM_000311.4	
Green	*PSEN1*	9508	104311	NM_000021.3	
Green	*PSEN2*	9509	600759	NM_000447.2	
Green	*RNF216*	21698	609948	NM_207111.3	
Green	*SETX*	445	608465	NM_015046.6	
Green	*SLC20A2*	10947	158378	NM_006749.4	
Green	*SNCA*	11138	163890	NM_000345.3	
Green	*SOD1*	11179	147450	NM_000454.4	
Green	*SPAST*	11233	604277	NM_014946.3	
Green	*SPG11*	11226	610844	NM_025137.3	
Green	*SQSTM1*	11280	601530	NM_003900.4	

R58.1 :: Adult onset neurodegenerative disorder
Panel: Neurodegenerative disorders - adult onset (Version 2.31)
https://panelapp.genomicsengland.co.uk/panels/474/
Agilent SureSelect Reagent Design ID: 3297111 – Laboratory work in NEYGLH Central Lab

Rating	Gene	HGNC	OMIM	Primary Transcript	Additional Transcripts
Green	*SYNJ1*	11503	604297	NM_003895.3	
Green	*TARDBP*	11571	605078	NM_007375.3	
Green	*TBK1*	11584	604834	NM_013254.3	
Green	*TMEM240*	25186	616101	NM_001114748.1	
Green	*TREM2*	17761	605086	NM_018965.3	
Green	*TTC19*	26006	613814	NM_017775.3	
Green	*TYROBP*	12449	604142	NM_003332.3	
Green	*UBQLN2*	12509	300264	NM_013444.3	
Green	*VAPB*	12649	605704	NM_004738.4	
Green	*VCP*	12666	601023	NM_007126.4	
Green	*VPS13A*	1908	605978	NM_033305.2	
Green	*VPS35*	13487	601501	NM_018206.5	
Green	*WDR45*	28912	300526	NM_007075.3	
Green	*XPR1*	12827	605237	NM_004736.3	

R58.1 :: Adult onset neurodegenerative disorder
Panel: Neurodegenerative disorders - adult onset (Version 2.31)
https://panelapp.genomicsengland.co.uk/panels/474/
Agilent SureSelect Reagent Design ID: 3297111 – Laboratory work in NEYGLH Central Lab

R58.1.1 Slice 1		R58.1.2 Slice 2		R58.1.3 Slice 3	
Dementia		Parkinson Disease		ALS	
ABCD1	NPC1	ATP13A2	PARK7	ALS2	PFN1
APP	NPC2	ATP1A3	PINK1	ANG	SETX
ARSA	PDGFB	CHCHD2	PLA2G6	ANXA11	SOD1
CCNF	PDGFRB	CSF1R	PRKN	CCNF	SPG11
CHCHD10	PRNP	DCTN1	SNCA	CHCHD10	SQSTM1
CHMP2B	PSEN1	DNAJC6	SPG11	CHMP2B	TARDBP
CLN6	PSEN2	FBXO7	SYNJ1	FIG4	TBK1
CSF1R	SLC20A2	GCH1	VPS13A	FUS	UBQLN2
CTSF	SQSTM1	GRN	VPS35	HNRNPA1	VAPB
FUS	TARDBP	LRRK2	WDR45	OPTN	VCP
GRN	TBK1	MAPT	XPR1		
HTRA1	TREM2	PANK2			
ITM2B	TYROBP				
MAPT	VCP				
NOTCH3	XPR1				

R60.1 :: Adult onset hereditary spastic paraplegia
Panel: Hereditary spastic paraplegia - adult onset (Version 1.12)
https://panelapp.genomicsengland.co.uk/panels/567/
Agilent SureSelect Reagent Design ID: 3297111 – Laboratory work in NEYGLH Central Lab

Rating	Gene	HGNC	OMIM	Primary Transcript	Additional Transcripts

R60.1 :: Adult onset hereditary spastic paraplegia
Panel: Hereditary spastic paraplegia - adult onset (Version 1.12)
https://panelapp.genomicsengland.co.uk/panels/567/
Agilent SureSelect Reagent Design ID: 3297111 – Laboratory work in NEYGLH Central Lab

Rating	Gene	HGNC	OMIM	Primary Transcript	Additional Transcripts
Green	*ABCD1*	61	300371	NM_000033.3	
Green	*ADAR*	225	146920	NM_001111.4	
Green	*AFG3L2*	315	604581	NM_006796.2	
Green	*AIMP1*	10648	603605	NM_004757.3	NM_001142416.1
Green	*ALDH18A1*	9722	138250	NM_002860.3	
Green	*ALS2*	443	606352	NM_020919.3	
Green	*AP4B1*	572	607245	NM_006594.4	
Green	*AP4E1*	573	607244	NM_007347.4	
Green	*AP4M1*	574	602296	NM_004722.3	
Green	*AP4S1*	575	607243	NM_007077.4	
Green	*AP5Z1*	22197	613653	NM_014855.2	
Green	*ARG1*	663	608313	NM_000045.3	
Green	*ATL1*	11231	606439	NM_015915.4	
Green	*ATP13A2*	30213	610513	NM_022089.3	
Green	*B4GALNT1*	4117	601873	NM_001478.4	
Green	*BSCL2*	15832	606158	NM_032667.6	NM_001122955.3
Green	*C12orf65*	26784	613541	NM_152269.4	
Green	*C19orf12*	25443	614297	NM_001031726.3	
Green	*CAPN1*	1476	114220	NM_001198868.1	
Green	*CYP27A1*	2605	606530	NM_000784.3	
Green	*CYP2U1*	20582	610670	NM_183075.2	
Green	*CYP7B1*	2652	603711	NM_004820.4	
Green	*DARS1*	2678	603084	NM_001349.3	
Green	*DDHD1*	19714	614603	NM_001160148.1	NM_001160147.1
Green	*DDHD2*	29106	615003	NM_015214.2	
Green	*ENTPD1*	3363	601752	NM_001098175.1	NM_001776.5
Green	*ERLIN1*	16947	611604	NM_006459.3	
Green	*ERLIN2*	1356	611605	NM_007175.6	
Green	*FA2H*	21197	611026	NM_024306.4	
Green	*FARS2*	21062	611592	NM_006567.4	
Green	*FXN*	3951	606829	NM_000144.4	
Green	*GBA2*	18986	609471	NM_020944.2	
Green	*GCH1*	4193	600225	NM_000161.2	
Green	*HACE1*	21033	610876	NM_020771.3	
Green	*KCNA2*	6220	176262	NM_004974.3	
Green	*KDM5C*	11114	314690	NM_004187.3	
Green	*KIDINS220*	29508	615759	NM_020738.3	
Green	*KIF1A*	888	601255	NM_001244008.1	
Green	*KIF1C*	6317	603060	NM_006612.5	

R60.1 :: Adult onset hereditary spastic paraplegia
Panel: Hereditary spastic paraplegia - adult onset (Version 1.12)
https://panelapp.genomicsengland.co.uk/panels/567/
Agilent SureSelect Reagent Design ID: 3297111 – Laboratory work in NEYGLH Central Lab

Rating	Gene	HGNC	OMIM	Primary Transcript	Additional Transcripts
Green	*KIF5A*	6323	602821	NM_004984.3	
Green	*L1CAM*	6470	308840	NM_000425.4	
Green	*NIPA1*	17043	608145	NM_144599.4	
Green	*NKX6-2*	19321	605955	NM_177400.2	
Green	*NT5C2*	8022	600417	NM_012229.4	
Green	*OPA3*	8142	606580	NM_025136.3	
Green	*PLP1*	9086	300401	NM_001128834.2	
Green	*PNPLA6*	16268	603197	NM_006702.4	
Green	*POLR3A*	30074	614258	NM_007055.3	
Green	*PSEN1*	9508	104311	NM_000021.3	
Green	*REEP1*	25786	609139	NM_001164730.1	NM_022912.2
Green	*REEP2*	17975	609347	NM_001271803.1	
Green	*RTN2*	10468	603183	NM_005619.4	
Green	*SACS*	10519	604490	NM_014363.5	
Green	*SERAC1*	21061	614725	NM_032861.3	
Green	*SLC16A2*	10923	300095	NM_006517.4	
Green	*SLC1A4*	10942	600229	NM_003038.4	
Green	*SPART*	18514	607111	NM_015087.4	
Green	*SPAST*	11233	604277	NM_014946.3	
Green	*SPG11*	11226	610844	NM_025137.3	
Green	*SPG21*	20373	608181	NM_016630.6	
Green	*SPG7*	11237	602783	NM_003119.3	
Green	*TFG*	11758	602498	NM_006070.5	
Green	*TUBB4A*	20774	602662	NM_006087.3	
Green	*UBAP1*	12461	609787	NM_001171201.1	
Green	*UCHL1*	12513	191342	NM_004181.4	
Green	*WASHC5*	28984	610657	NM_014846.3	
Green	*WDR45B*	25072	609226	NM_019613.3	
Green	*ZFYVE26*	20761	612012	NM_015346.3	

R61.1 :: Childhood onset hereditary spastic paraplegia
Panel: Hereditary spastic paraplegia - childhood onset (Version 2.18)
https://panelapp.genomicsengland.co.uk/panels/568/
Agilent SureSelect Reagent Design ID: 3297111 – Laboratory work in NEYGLH Central Lab

Rating	Gene	HGNC	OMIM	Primary Transcript	Additional Transcripts
Green	*ABCD1*	61	300371	NM_000033.3	
Green	*ADAR*	225	146920	NM_001111.4	
Green	*AFG3L2*	315	604581	NM_006796.2	

R61.1 :: Childhood onset hereditary spastic paraplegia
Panel: Hereditary spastic paraplegia - childhood onset (Version 2.18)
https://panelapp.genomicsengland.co.uk/panels/568/
Agilent SureSelect Reagent Design ID: 3297111 – Laboratory work in NEYGLH Central Lab

Rating	Gene	HGNC	OMIM	Primary Transcript	Additional Transcripts
Green	*AIMP1*	10648	603605	NM_004757.3	NM_001142416.1
Green	*ALDH18A1*	9722	138250	NM_002860.3	
Green	*ALS2*	443	606352	NM_020919.3	
Green	*AP4B1*	572	607245	NM_006594.4	
Green	*AP4E1*	573	607244	NM_007347.4	
Green	*AP4M1*	574	602296	NM_004722.3	
Green	*AP4S1*	575	607243	NM_007077.4	
Green	*AP5Z1*	22197	613653	NM_014855.2	
Green	*ARG1*	663	608313	NM_000045.3	
Green	*ATL1*	11231	606439	NM_015915.4	
Green	*ATP13A2*	30213	610513	NM_022089.3	
Green	*B4GALNT1*	4117	601873	NM_001478.4	
Green	*BSCL2*	15832	606158	NM_032667.6	NM_001122955.3
Green	*C12orf65*	26784	613541	NM_152269.4	
Green	*C19orf12*	25443	614297	NM_001031726.3	
Green	*CAPN1*	1476	114220	NM_001198868.1	
Green	*CPT1C*	18540	608846	NM_001199752.2	
Green	*CYP27A1*	2605	606530	NM_000784.3	
Green	*CYP2U1*	20582	610670	NM_183075.2	
Green	*CYP7B1*	2652	603711	NM_004820.4	
Green	*DARS1*	2678	603084	NM_001349.3	
Green	*DDHD1*	19714	614603	NM_001160148.1	NM_001160147.1
Green	*DDHD2*	29106	615003	NM_015214.2	
Green	*ENTPD1*	3363	601752	NM_001098175.1	NM_001776.5
Green	*ERLIN1*	16947	611604	NM_006459.3	
Green	*ERLIN2*	1356	611605	NM_007175.6	
Green	*FA2H*	21197	611026	NM_024306.4	
Green	*FARS2*	21062	611592	NM_006567.4	
Green	*FXN*	3951	606829	NM_000144.4	
Green	*GBA2*	18986	609471	NM_020944.2	
Green	*GCH1*	4193	600225	NM_000161.2	
Green	*HACE1*	21033	610876	NM_020771.3	
Green	*KCNA2*	6220	176262	NM_004974.3	
Green	*KDM5C*	11114	314690	NM_004187.3	
Green	*KIDINS220*	29508	615759	NM_020738.3	
Green	*KIF1A*	888	601255	NM_001244008.1	
Green	*KIF1C*	6317	603060	NM_006612.5	

R61.1 :: Childhood onset hereditary spastic paraplegia
Panel: Hereditary spastic paraplegia - childhood onset (Version 2.18)
https://panelapp.genomicsengland.co.uk/panels/568/
Agilent SureSelect Reagent Design ID: 3297111 – Laboratory work in NEYGLH Central Lab

Rating	Gene	HGNC	OMIM	Primary Transcript	Additional Transcripts
Green	*KIF5A*	6323	602821	NM_004984.3	
Green	*L1CAM*	6470	308840	NM_000425.4	
Green	*NIPA1*	17043	608145	NM_144599.4	
Green	*NKX6-2*	19321	605955	NM_177400.2	
Green	*NT5C2*	8022	600417	NM_012229.4	
Green	*OPA3*	8142	606580	NM_025136.3	
Green	*PLP1*	9086	300401	NM_001128834.2	
Green	*PNPLA6*	16268	603197	NM_006702.4	
Green	*POLR3A*	30074	614258	NM_007055.3	
Green	*REEP1*	25786	609139	NM_001164730.1	NM_022912.2
Green	*REEP2*	17975	609347	NM_001271803.1	
Green	*RTN2*	10468	603183	NM_005619.4	
Green	*SACS*	10519	604490	NM_014363.5	
Green	*SERAC1*	21061	614725	NM_032861.3	
Green	*SLC16A2*	10923	300095	NM_006517.4	
Green	*SLC1A4*	10942	600229	NM_003038.4	
Green	*SLC2A1*	11005	138140	NM_006516.2	
Green	*SPART*	18514	607111	NM_015087.4	
Green	*SPAST*	11233	604277	NM_014946.3	
Green	*SPG11*	11226	610844	NM_025137.3	
Green	*SPG21*	20373	608181	NM_016630.6	
Green	*SPG7*	11237	602783	NM_003119.3	
Green	*TFG*	11758	602498	NM_006070.5	
Green	*TUBB4A*	20774	602662	NM_006087.3	
Green	*UBAP1*	12461	609787	NM_001171201.1	
Green	*UCHL1*	12513	191342	NM_004181.4	
Green	*WASHC5*	28984	610657	NM_014846.3	
Green	*WDR45B*	25072	609226	NM_019613.3	
Green	*ZFYVE26*	20761	612012	NM_015346.3	

R78.1 :: Hereditary neuropathy or pain disorder – NOT PMP22 copy number
Panel: Hereditary neuropathy NOT PMP22 copy number (Version 1.2)
https://panelapp.genomicsengland.co.uk/panels/846/
Agilent SureSelect Reagent Design ID: 3297101 – Laboratory work in NEYGLH Central Lab

Rating	Gene	HGNC	OMIM	Primary Transcript	Additional Transcripts

R78.1 :: Hereditary neuropathy or pain disorder – NOT PMP22 copy number
Panel: Hereditary neuropathy NOT PMP22 copy number (Version 1.2)
https://panelapp.genomicsengland.co.uk/panels/846/
Agilent SureSelect Reagent Design ID: 3297101 – Laboratory work in NEYGLH Central Lab

Rating	Gene	HGNC	OMIM	Primary Transcript	Additional Transcripts
Green	*AARS*	20	601065	NM_001605.2	
Green	*ABCA1*	29	600046	NM_005502.3	
Green	*AIFM1*	8768	300169	NM_004208.3	NM_145812.2
Green	*ATL1*	11231	606439	NM_015915.4	
Green	*ATP1A1*	799	182310	NM_000701.7	
Green	*ATP7A*	869	300011	NM_000052.6	
Green	*BICD2*	17208	609797	NM_001003800.1	
Green	*BSCL2*	15832	606158	NM_032667.6	NM_001122955.3
Green	*CHCHD10*	15559	615903	NM_213720.2	
Green	*COX6A1*	2277	602072	NM_004373.3	
Green	*CPOX*	2321	612732	NM_000097.5	
Green	*CYP27A1*	2605	606530	NM_000784.3	
Green	*DCTN1*	2711	601143	NM_004082.4	
Green	*DNAJB2*	5228	604139	NM_001039550.1	
Green	*DNM2*	2974	602378	NM_001005361.2	
Green	*DNMT1*	2976	126375	NM_001130823.2	
Green	*DST*	1090	113810	NM_001144769.2	NM_001723.5, NM_183380.3
Green	*DYNC1H1*	2961	600112	NM_001376.4	
Green	*EGR2*	3239	129010	NM_000399.4	
Green	*ELP1*	5959	603722	NM_003640.4	
Green	*FBLN5*	3602	604580	NM_006329.3	
Green	*FGD4*	19125	611104	NM_139241.3	
Green	*FIG4*	16873	609390	NM_014845.5	
Green	*GARS*	4162	600287	NM_002047.2	
Green	*GDAP1*	15968	606598	NM_018972.2	
Green	*GJB1*	4283	304040	NM_000166.5	
Green	*GNB4*	20731	610863	NM_021629.3	
Green	*HARS*	4816	142810	NM_002109.5	
Green	*HINT1*	4912	601314	NM_005340.6	
Green	*HK1*	4922	142600	NM_000188.2	NM_033500.2
Green	*HMBS*	4982	609806	NM_000190.3	
Green	*HSPB1*	5246	602195	NM_001540.4	
Green	*HSPB8*	30171	608014	NM_014365.2	
Green	*IGHMBP2*	5542	600502	NM_002180.2	
Green	*INF2*	23791	610982	NM_022489.3	
Green	*KIF1A*	888	601255	NM_001244008.1	
Green	*KIF5A*	6323	602821	NM_004984.3	
Green	*LITAF*	16841	603795	NM_004862.3	
Green	*LMNA*	6636	150330	NM_170707.3	

R78.1 :: Hereditary neuropathy or pain disorder – NOT PMP22 copy number
Panel: Hereditary neuropathy NOT PMP22 copy number (Version 1.2)
https://panelapp.genomicsengland.co.uk/panels/846/
Agilent SureSelect Reagent Design ID: 3297101 – Laboratory work in NEYGLH Central Lab

Rating	Gene	HGNC	OMIM	Primary Transcript	Additional Transcripts
Green	*LRSAM1*	25135	610933	NM_138361.5	
Green	*MCM3AP*	6946	603294	NM_003906.4	
Green	*MFN2*	16877	608507	NM_014874.3	
Green	*MME*	7154	120520	NM_007289.2	
Green	*MORC2*	23573	616661	NM_001303256.2	
Green	*MPV17*	7224	137960	NM_002437.4	
Green	*MPZ*	7225	159440	NM_000530.7	
Green	*MT-ATP6*	7414	516060	Analysis available via mitochondrial service	
Green	*MTMR2*	7450	603557	NM_016156.5	
Green	*NDRG1*	7679	605262	NM_006096.3	
Green	*NEFH*	7737	162230	NM_021076.3	
Green	*NEFL*	7739	162280	NM_006158.4	
Green	*NGF*	7808	162030	NM_002506.2	
Green	*NTRK1*	8031	191315	NM_002529.3	
Green	*PLEKHG5*	29105	611101	NM_020631.4	
Green	*PMP2*	9117	170715	NM_002677.4	
Green	*PMP22*	9118	601097	NM_000304.3	
Green	*PPOX*	9280	600923	NM_000309.4	
Green	*PRDM12*	13997	616458	NM_021619.2	
Green	*PRPS1*	9462	311850	NM_002764.3	
Green	*PRX*	13797	605725	NM_181882.2	
Green	*RAB7A*	9788	602298	NM_004637.5	
Green	*REEP1*	25786	609139	NM_001164730.1	NM_022912.2
Green	*RETREG1*	25964	613114	NM_001034850.2	
Green	*SBF1*	10542	603560	NM_002972.3	
Green	*SBF2*	2135	607697	NM_030962.3	
Green	*SCN10A*	10582	604427	NM_006514.3	
Green	*SCN11A*	10583	604385	NM_014139.2	
Green	*SCN9A*	10597	603415	NM_002977.3	
Green	*SEPT9*	7323	604061	NM_001113491.1	NM_001113493.1, NM_001293696.1, NM_001113495.1
Green	*SETX*	445	608465	NM_015046.6	
Green	*SH3TC2*	29427	608206	NM_024577.3	
Green	*SIGMAR1*	8157	601978	NM_005866.3	
Green	*SLC52A2*	30224	607882	NM_024531.4	
Green	*SLC52A3*	16187	613350	NM_033409.3	
Green	*SLC5A7*	14025	608761	NM_021815.4	
Green	*SMN1*	11117	600354	NM_000344.3	
Green	*SPG11*	11226	610844	NM_025137.3	

R78.1 :: Hereditary neuropathy or pain disorder – NOT PMP22 copy number
Panel: Hereditary neuropathy NOT PMP22 copy number (Version 1.2)
https://panelapp.genomicsengland.co.uk/panels/846/
Agilent SureSelect Reagent Design ID: 3297101 – Laboratory work in NEYGLH Central Lab

Rating	Gene	HGNC	OMIM	Primary Transcript	Additional Transcripts
Green	*SPTLC1*	11277	605712	NM_006415.3	
Green	*SPTLC2*	11278	605713	NM_004863.3	
Green	*TFG*	11758	602498	NM_006070.5	
Green	*TRIM2*	15974	614141	NM_001130067.1	
Green	*TRPV4*	18083	605427	NM_021625.4	
Green	*TTR*	12405	176300	NM_000371.3	
Green	*VRK1*	12718	602168	NM_003384.2	
Green	*WARS*	12729	191050	NM_004184.3	
Green	*WNK1*	14540	605232	NM_018979.3	NM_213655.4
Green	*YARS*	12840	603623	NM_003680.3	

Haematology disorders

Bleeding & Thrombotic disorders

(SureSelect Design ID: 0657461) wetwork in Sheffield for non-NHS patients only

Sub-panels			
Disease Names	**Gene(s)**	**OMIM**	**Primary Transcript**
ADAMTS13 deficiency	*ADAMTS13*	274150	NM_139025.4
Factor V deficiency	*F5*	227400	NM_000130.4
Haemophilia A	*F8*	306700	NM_000132.3
Haemophilia A and B (where the type of haemophilia is unknown)	*F8* *F9*	306700 306900	NM_000132.3 NM_000133.3
Haemophilia A and 2N VWD (where the cause of reduced FVIII:C is unknown)	*F8* *VWF*	306700 277480	NM_000132.3 NM_000552.3
Factor XIII deficiency	*F13A1* *F13B*	613225 613235	NM_000129.3 NM_001994.2
Fibrinogen disorders	*FGA* *FGB* *FGG*	134820 134830 134850	NM_000508.3 NM_005141.4 NM_021870.2
Glanzmann thrombasthenia	*ITGAT2B* *ITGB3*	273800 273800	NM_000419.3 NM_000212.2
MYH9 related disorders	*MYH9*	160775	NM_002473.5
von Willebrand disease	*VWF*	277480	NM_000552.3

Bone Marrow Failure Disorders

(SureSelect Design ID: 0724921) wetwork in Sheffield for non-NHS patients only

Sub-panels			
Condition	**OMIM**	**Gene**	**Primary transcript**
Fanconi anaemia, complementation group A	227650	*FANCA*	NM_000135.2
Fanconi anaemia, complementation group B	300514	*FANCB*	NM_001018113.1
Fanconi anaemia, complementation group C	227645	*FANCC*	NM_000136.2
Fanconi anaemia, complementation group D1	605724	*BRCA2*	NM_000059.3
Fanconi anaemia, complementation group D2	227646	*FANCD2*	NM_033084.3
Fanconi anaemia, complementation group E	600901	*FANCE*	NM_021922.2
Fanconi anaemia, complementation group F	603467	*FANCF*	NM_022725.3
Fanconi anaemia, complementation group G	614082	*FANCG*	NM_004629.1
Fanconi anaemia, complementation group I	609053	*FANCI*	NM_032043.2
Fanconi anaemia, complementation group J	609054	*BRIP1*	NM_001113378.1
Fanconi anaemia, complementation group L	614083	*FANCL*	NM_001114636.1

Fanconi anaemia, complementation group M	614087	*FANCM*	NM_020937.3
Fanconi anaemia, complementation group N	610832	*PALB2*	NM_024675.3
Fanconi anaemia, complementation group O	613390	*RAD51C*	NM_058216.2
Fanconi anaemia, complementation group P	613951	*SLX4*	NM_032444.2
Fanconi anaemia, complementation group Q	615272	*ERCC4*	NM_005236.2
Diamond-Blackfan Anaemia	305371	*GATA1*	NM_002049.3
	603634	*RPL5*	NM_000969.3
	604175	*RPL11*	NM_000975.3
	180468	*RPL35A*	NM_000996.2
	603658	*RPS7*	NM_001011.3
	603632	*RPS10*	NM_001014.4
	130620	*RPS14*	NM_00102507.1
	180472	*RPS17*	NM_001021.5
	603474	*RPS19*	NM_001022.3
	602412	*RPS24*	NM_001142285.1
	603701	*RPS26*	NM_001029.3
Myelodysplastic syndrome	137295	*GATA2*	NM_001145661.1
	159530	*MPL*	NM_005373.2
	607444	*SBDS*	NM_016038.2
	151385	*RUNX1*	NM_001754.4
	602122	*SRP72*	NM_006947.3
Severe Congenital Neutropenia	130130	*ELANE*	NM_001972.3
	611045	*G6PC3*	NM_138387.3
	600871	*GFI1*	NM_005263.4
	605998	*HAX1*	NM_006118.3
	300392	*WAS*	NM_000377.2
Thrombocytopenia with absent radii	605313	*RBM8A* (& microarray)	NM_005105.4

Haems mini-panel

(Ampliseq Design ID: IAD162775_197) Wetwork in Sheffield

Gene	Primary transcript
F2	NM_000506.4
F8	NM_000132.3
F9	NM_000133.3
F11	NM_000128.3
GP1BA	NM_000173.6
GP1BB	NM_000407.4
GP9	NM_000174.4
MPL	NM_005373.2
PROC	NM_000312.3
PROS1	NM_000313.3

SERPINC1	NM_000488.3
VKORC1	NM_024006.5
F7	NM_000131.4
F10	NM_000504.3
GGCX	NM_000821.6
F5	NM_000130.4
ADAMTS13	NM_139025.4
VWF	NM_000552.4
F13A1	NM_000129.3
F13B	NM_001994.2
NBN	NM_002485.4
ELANE	NM_001972.3
PAX5	NM_016734.2
ETV6	NM_001987.4
CSF3R	NM_156039.3
GATA2	NM_001145661.1
RUNX1	NM_001754.4
CEBPA	NM_001287424.1
ANKRD26	NM_014915.2
DDX41	NM_016222.3
SRP72	NM_006947.3
HFE	NM_000410.3
HFE2 (HJV)	NM_213653.3
HAMP	NM_021175.3
TFR2	NM_003227.3
SLC40A1	NM_014585.5
LMAN1	NM_005570.3
MCFD2	NM_139279.5
ITGA2B	NM_000419.4
ITGB3	NM_000212.2
FGA	NM_000508.4
FGB	NM_005141.4
FGG	NM_021870.2

R90.1 :: Bleeding and Platelet Disorders (Version 1.2)
https://panelapp.genomicsengland.co.uk/panels/545/
Agilent SureSelect Reagent Design ID: 3278641 – Laboratory work in YNEGLH Central Lab

Rating	Gene	HGNC	Primary Transcript	Slicing panels
Green	*ABCG5*	HGNC:13886	NM_022436.2	
Green	*ABCG8*	HGNC:13887	NM_022437.2	
Green	ACTB	HGNC:132	NM_001101.1	
Green	ACTN1	HGNC:163	NM_001130004.1	R90 Gene Slice 5 Macrothrombocytopenia / R91 Gene Slice 1 Thrombocytopenia
Green	ACVRL1	HGNC:175	NM_000020.2	
Green	ADAMTS13	HGNC:1366	NM_139025.4	
Green	*ANKRD26*	HGNC:29186	NM_014915.2	
Green	ANO6	HGNC:25240	NM_001025356.2	

Green	AP3B1	HGNC:566	NM_003664.4	R90 Gene Slice 4 Hermansy-Pudlak Syndrome
Green	AP3D1	HGNC:568	NM_001261826.2	R90 Gene Slice 4 Hermansy-Pudlak Syndrome
Green	ARPC1B	HGNC:704	NM_005720.3	
Green	BLOC1S3	HGNC:20914	NM_212550.4	R90 Gene Slice 4 Hermansy-Pudlak Syndrome
Green	BLOC1S6	HGNC:8549	NM_012388.3	R90 Gene Slice 4 Hermansy-Pudlak Syndrome
Green	CDC42	HGNC:1736	NM_001791.3	
Green	*CYCS*	HGNC:19986	NM_018947.5	
Green	DIAPH1	HGNC:2876	NM_005219.4	R90 Gene Slice 5 Macrothrombocytopenia
Green	DTNBP1	HGNC:17328	NM_032122.4	R90 Gene Slice 4 Hermansy-Pudlak Syndrome
Green	ENG	HGNC:3349	NM_000118.3	
Green	*ETV6*	HGNC:3495	NM_001987.4	
Green	F10	HGNC:3528	NM_000504.3	
Green	F11	HGNC:3529	NM_000128.3	
Green	F12	HGNC:3530	NM_000505.3	
Green	F13A1	HGNC:3531	NM_000129.3	
Green	F13B	HGNC:3534	NM_001994.2	
Green	F2	HGNC:3535	NM_000506.4	
Green	F5	HGNC:3542	NM_000130.4	R90 Gene Slice 8 haemophilia like bleeding/low factor VIII
Green	F7	HGNC:3544	NM_000131.4	
Green	F8	HGNC:3546	NM_000132.3	R90 Gene Slice 8 haemophilia like bleeding/low factor VIII
Green	F9	HGNC:3551	NM_000133.3	
Green	FERMT3	HGNC:23151	NM_031471.5	
Green	FGA	HGNC:3661	NM_021871.3	R90 Gene Slice 1 Fibrinogen Deficiency
Green	FGB	HGNC:3662	NM_005141.4	R90 Gene Slice 1 Fibrinogen Deficiency
Green	FGG	HGNC:3694	NM_000509.5	R90 Gene Slice 1 Fibrinogen Deficiency
Green	FLI1	HGNC:3749	NM_002017.4	R91 Gene Slice 1 Thrombocytopenia
Green	*FYB1*	HGNC:4036	NM_001465.5	
Green	*GATA1*	HGNC:4170	NM_002049.3	
Green	GBA	HGNC:4177	NM_001005741.2	
Green	GFI1B	HGNC:4238	NM_004188.6	
Green	GGCX	HGNC:4247	NM_000821.6	
Green	GNE	HGNC:23657	NM_001128227.2	
Green	GP1BA	HGNC:4439	NM_000173.6	R90 Gene Slice 2 Bernard Soulier syndrome/ R90 Gene Slice 5 Macrothrombocytopenia
Green	GP1BB	HGNC:4440	NM_000407.4	R90 Gene Slice 2 Bernard Soulier syndrome
Green	GP6	HGNC:14388	NM_001083899.2	
Green	GP9	HGNC:4444	NM_000174.4	R90 Gene Slice 2 Bernard Soulier syndrome
Green	HOXA11	HGNC:5101	NM_005523.5	
Green	HPS1	HGNC:5163	NM_000195.4	R90 Gene Slice 4 Hermansy-Pudlak Syndrome
Green	HPS3	HGNC:15597	NM_032383.4	R90 Gene Slice 4 Hermansy-Pudlak Syndrome

Green	HPS4	HGNC:15844	NM_022081.5	R90 Gene Slice 4 Hermansy-Pudlak Syndrome
Green	HPS5	HGNC:17022	NM_181507.1	R90 Gene Slice 4 Hermansy-Pudlak Syndrome
Green	HPS6	HGNC:18817	NM_024747.5	R90 Gene Slice 4 Hermansy-Pudlak Syndrome
Green	ITGA2B	HGNC:6138	NM_000419.4	R90 Gene Slice 3 Glanzman Thrombasthenia
Green	ITGB3	HGNC:6156	NM_000212.2	R90 Gene Slice 3 Glanzman Thrombasthenia
Green	KDSR	HGNC:4021	NM_002035.2	
Green	KLKB1	HGNC:6371	NM_000892.4	
Green	KNG1	HGNC:6383	NM_001102416.2	
Green	LMAN1	HGNC:6631	NM_005570.3	R90 Gene Slice 8 haemophilia like bleeding/low factor VIII
Green	LYST	HGNC:1968	NM_000081.3	
Green	MCFD2	HGNC:18451	NM_139279.5	R90 Gene Slice 8 haemophilia like bleeding/low factor VIII
Green	*MECOM*	HGNC:3498	NM_001105078.3	
Green	MPIG6B	HGNC:13937	NM_025260.3	
Green	*MPL*	HGNC:7217	NM_005373.2	
Green	MYH9	HGNC:7579	NM_002473.5	R90 Gene Slice 5 Macrothrombocytopenia
Green	NBEA	HGNC:7648	NM_015678.4	
Green	NBEAL2	HGNC:31928	NM_015175.2	
Green	P2RY12	HGNC:18124	NM_022788.4	
Green	PLA2G4A	HGNC:9035	NM_024420.2	
Green	PLAU	HGNC:9052	NM_002658.4	
Green	PRKACG	HGNC:9382	NM_002732.3	
Green	RASGRP2	HGNC:9879	NM_153819.1	
Green	RBM8A	HGNC:9905	NM_005105.4	
Green	RUNX1	HGNC:10471	NM_001754.4	
Green	SERPINE1	HGNC:8583	NM_000602.4	
Green	SERPINF2	HGNC:9075	NM_000934.3	
Green	SLFN14	HGNC:32689	NM_001129820.1	
Green	*SRC*	HGNC:11283	NM_005417.4	
Green	STIM1	HGNC:11386	NM_003156.3	
Green	STXBP2	HGNC:11445	NM_006949.3	
Green	TBXA2R	HGNC:11608	NM_001060.5	
Green	TBXAS1	HGNC:11609	NM_001061.4	
Green	THBD	HGNC:11784	NM_000361.2	
Green	*THPO*	HGNC:11795	NM_000460.3	
Green	TPM4	HGNC:12013	NM_001145160.1	
Green	TUBB1	HGNC:16257	NM_030773.3	R90 Gene Slice 5 Macrothrombocytopenia
Green	VIPAS39	HGNC:20347	NM_022067.3	
Green	VKORC1	HGNC:23663	NM_024006.5	

Green	VPS33B	HGNC:12712	NM_018668.4	
Green	VWF	HGNC:12726	NM_000552.4	R90 Gene Slice 8 haemophilia like bleeding/low factor VIII
Green	*WAS*	HGNC:12731	NM_000377.2	

R91.1 :: Cytopenia NOT Fanconi Anaemia (Version 1.29)
https://panelapp.genomicsengland.co.uk/panels/519/
Agilent SureSelect Reagent Design ID: 3297121 – Laboratory work in YNEGLH Central Lab

Rating	Gene	HGNC	Primary Transcript	Slicing Panels
Green	*ACD*	HGNC:25070	NM_001082486.1	R91 Gene Slice 3 Inherited Bone marrow failure
Green	*ADA2*	HGNC:1839	NM_001282225.1	R91 Gene Slice 1 Thrombocytopenia / R91 Gene Slice 2 Neutropenia / R91 Gene Slice 3 Inherited Bone marrow failure
Green	*ANKRD26*	HGNC:29186	NM_014915.2	R91 Gene Slice 1 Thrombocytopenia / R91 Gene Slice 3 Inherited Bone marrow failure
Green	*CSF3R*	HGNC:2439	NM_000760.3	R91 Gene Slice 2 Neutropenia
Green	*CTC1*	HGNC:26169	NM_025099.5	R91 Gene Slice 3 Inherited Bone marrow failure
Green	*CXCR4*	HGNC:2561	NM_003467.2	R91 Gene Slice 2 Neutropenia / R91 Gene Slice 3 Inherited Bone marrow failure
Green	*CYCS*	HGNC:19986	NM_018947.5	R91 Gene Slice 1 Thrombocytopenia
Green	*DKC1*	HGNC:2890	NM_001363.4	R91 Gene Slice 3 Inherited Bone marrow failure
Green	*DNAJC21*	HGNC:27030	NM_001012339.2	R91 Gene Slice 3 Inherited Bone marrow failure
Green	*ELANE*	HGNC:3309	NM_001972.3	R91 Gene Slice 2 Neutropenia
Green	*ERCC6L2*	HGNC:26922	NM_020207.4	R91 Gene Slice 3 Inherited Bone marrow failure
Green	*ETV6*	HGNC:3495	NM_001987.4	R91 Gene Slice 1 Thrombocytopenia
Green	*FYB1*	HGNC:4036	NM_001465.5	R91 Gene Slice 1 Thrombocytopenia
Green	*G6PC3*	HGNC:24861	NM_138387.3	R91 Gene Slice 2 Neutropenia / R91 Gene Slice 3 Inherited Bone marrow failure
Green	*GATA1*	HGNC:4170	NM_002049.3	R91 Gene Slice 1 Thrombocytopenia
Green	*GATA2*	HGNC:4171	NM_032638.4	R91 Gene Slice 3 Inherited Bone marrow failure
Green	*GFI1*	HGNC:4237	NM_005263.4	R91 Gene Slice 2 Neutropenia / R91 Gene Slice 3 Inherited Bone marrow failure
Green	*HAX1*	HGNC:16915	NM_006118.3	R91 Gene Slice 2 Neutropenia / R91 Gene Slice 3 Inherited Bone marrow failure
Green	*JAGN1*	HGNC:26926	NM_032492.3	R91 Gene Slice 2 Neutropenia
Green	*KIF23*	HGNC:6392	NM_138555.3	R91 Gene Slice 3 Inherited Bone marrow failure
Green	*KLF1*	HGNC:6345	NM_006563.4	R91 Gene Slice 3 Inherited Bone marrow failure
Green	*MECOM*	HGNC:3498	NM_001105078.3	R91 Gene Slice 1 Thrombocytopenia
Green	*MPL*	HGNC:7217	NM_005373.2	R91 Gene Slice 1 Thrombocytopenia / R91 Gene Slice 3 Inherited Bone marrow failure
Green	*PARN*	HGNC:8609	NM_002582.3	R91 Gene Slice 3 Inherited Bone marrow failure
Green	*RMRP*	HGNC:10031	NR_003051.3	R91 Gene Slice 2 Neutropenia
Green	*RPL11*	HGNC:10301	NM_000975.4	

R91.1 :: Cytopenia NOT Fanconi Anaemia (Version 1.29)
https://panelapp.genomicsengland.co.uk/panels/519/
Agilent SureSelect Reagent Design ID: 3297121 – Laboratory work in YNEGLH Central Lab

Rating	Gene	HGNC	Primary Transcript	Slicing Panels
Green	*RPL15*	HGNC:10306	NM_002948.4	
Green	*RPL26*	HGNC:10327	NM_000987.4	
Green	*RPL27*	HGNC:10328	NM_000988.4	
Green	*RPL31*	HGNC:10334	NM_001098577.2	
Green	*RPL35A*	HGNC:10345	NM_000996.3	
Green	*RPL5*	HGNC:10360	NM_000969.4	
Green	*RPL9*	HGNC:10369	NM_000661.4	
Green	*RPS10*	HGNC:10383	NM_001014.4	
Green	*RPS17*	HGNC:10397	NM_001021.5	
Green	*RPS19*	HGNC:10402	NM_001022.3	
Green	*RPS24*	HGNC:10411	NM_033022.3	
Green	*RPS26*	HGNC:10414	NM_001029.4	
Green	*RPS29*	HGNC:10419	NM_001032.4	
Green	*RPS7*	HGNC:10440	NM_001011.3	
Green	*RTEL1*	HGNC:15888	NM_032957.4	R91 Gene Slice 3 Inherited Bone marrow failure
Green	*SAMD9*	HGNC:1348	NM_017654.3	R91 Gene Slice 3 Inherited Bone marrow failure
Green	*SAMD9L*	HGNC:1349	NM_152703.4	R91 Gene Slice 1 Thrombocytopenia / R91 Gene Slice 3 Inherited Bone marrow failure
Green	*SBDS*	HGNC:19440	NM_016038.3	R91 Gene Slice 3 Inherited Bone marrow failure
Green	*SRC*	HGNC:11283	NM_005417.4	R91 Gene Slice 1 Thrombocytopenia
Green	*TAZ*	HGNC:11577	NM_000116.4	R91 Gene Slice 2 Neutropenia
Green	*TERC*	HGNC:11727	NR_001566.1	R91 Gene Slice 3 Inherited Bone marrow failure
Green	*TERT*	HGNC:11730	NM_198253.2	R91 Gene Slice 3 Inherited Bone marrow failure
Green	*THPO*	HGNC:11795	NM_000460.3	R91 Gene Slice 1 Thrombocytopenia
Green	*TINF2*	HGNC:11824	NM_001099274.1	R91 Gene Slice 3 Inherited Bone marrow failure
Green	*USB1*	HGNC:25792	NM_024598.3	R91 Gene Slice 2 Neutropenia / R91 Gene Slice 3 Inherited Bone marrow failure
Green	*VPS45*	HGNC:14579	NM_007259.4	R91 Gene Slice 2 Neutropenia / R91 Gene Slice 3 Inherited Bone marrow failure
Green	*WAS*	HGNC:12731	NM_000377.2	R91 Gene Slice 1 Thrombocytopenia / R91 Gene Slice 2 Neutropenia
Green	*WIPF1*	HGNC:12736	NM_001077269.1	R91 Gene Slice 1 Thrombocytopenia / R91 Gene Slice 2 Neutropenia
Green	*WRAP53*	HGNC:25522	NM_018081.2	R91 Gene Slice 3 Inherited Bone marrow failure

R92.1 :: Rare Anaemia (Version 1.2)
https://panelapp.genomicsengland.co.uk/panels/518/
Agilent SureSelect Reagent Design ID: 3297121 – Laboratory work in YNEGLH Central Lab

Rating	Gene	HGNC	Primary Transcript	Slicing panels
Green	*ABCB7*	HGNC:48	NM_004299.5	R92 Gene Slice 7 Sideroblastic Anaemia
Green	*ABCG5*	HGNC:13886	NM_022436.2	R92 Gene Slice 9 Sitosterolaemia
Green	*ABCG8*	HGNC:13887	NM_022437.2	R92 Gene Slice 9 Sitosterolaemia
Green	*ADA2*	HGNC:1839	NM_001282225.1	R92 Gene Slice 6 Diamond-Blackfan Anaemia
Green	*AK1*	HGNC:361	NM_000476.2	R92 Gene Slice 4 Enzymopathy / R92 Gene Slice 8 Haemolytic Anaemia
Green	*ALAS2*	HGNC:397	NM_000032.4	R92 Gene Slice 7 Sideroblastic Anaemia
Green	*ALDOA*	HGNC:414	NM_000034.3	R92 Gene Slice 8 Haemolytic Anaemia
Green	*AMN*	HGNC:14604	NM_030943.3	R92 Gene Slice 10 Megaloblastic Anaemia
Green	*ANK1*	HGNC:492	NM_000037.3	R92 Gene Slice 3 Membranopathy / R92 Gene Slice 8 Haemolytic Anaemia
Green	*C15orf41*	HGNC:26929	NM_001130010.2	R92 Gene Slice 5 Congenital Dyserythropoietic Anaemia
Green	*CBLIF (GIF)*	HGNC:4268	NM_005142.2	R92 Gene Slice 10 Megaloblastic Anaemia
Green	*CD59*	HGNC:1689	NM_203330.2	
Green	*CDAN1*	HGNC:1713	NM_138477.3	R92 Gene Slice 5 Congenital Dyserythropoietic Anaemia
Green	*COX4I2*	HGNC:16232	NM_032609.2	R92 Gene Slice 5 Congenital Dyserythropoietic Anaemia
Green	*CUBN*	HGNC:2548	NM_001081.3	R92 Gene Slice 10 Megaloblastic Anaemia
Green	*CYB5R3*	HGNC:2873	NM_000398.6	R92 Gene Slice 1 Methaemoglobinaemia / R92 Gene Slice 4 Enzymopathy
Green	*DHFR*	HGNC:2861	NM_000791.3	R92 Gene Slice 10 Megaloblastic Anaemia
Green	*EPB41*	HGNC:3377	NM_004437.3	R92 Gene Slice 3 Membranopathy
Green	*EPB42*	HGNC:3381	NM_000119.2	R92 Gene Slice 3 Membranopathy
Green	*G6PD*	HGNC:4057	NM_001042351.2	R92 Gene Slice 8 Haemolytic Anaemia
Green	*GATA1*	HGNC:4170	NM_002049.3	R92 Gene Slice 5 Congenital Dyserythropoietic Anaemia / R92 Gene Slice 6 Diamond-Blackfan Anaemia
Green	*GCLC*	HGNC:4311	NM_001498.3	R92 Gene Slice 8 Haemolytic Anaemia
Green	*GLRX5*	HGNC:20134	NM_016417.2	R92 Gene Slice 7 Sideroblastic Anaemia
Green	*GPI*	HGNC:4458	NM_000175.4	R92 Gene Slice 4 Enzymopathy / R92 Gene Slice 8 Haemolytic Anaemia
Green	*GSR*	HGNC:4623	NM_000637.4	R92 Gene Slice 4 Enzymopathy / R92 Gene Slice 8 Haemolytic Anaemia
Green	*GSS*	HGNC:4624	NM_000178.3	R92 Gene Slice 4 Enzymopathy / R92 Gene Slice 8 Haemolytic Anaemia
Green	*HBA1*	HGNC:4823	NM_000558.4	R92 Gene Slice 1 Methaemoglobinaemia / R92 Gene Slice 8 Haemolytic Anaemia
Green	*HBA2*	HGNC:4824	NM_000517.4	R92 Gene Slice 1 Methaemoglobinaemia /

R92.1 :: Rare Anaemia (Version 1.2)
https://panelapp.genomicsengland.co.uk/panels/518/
Agilent SureSelect Reagent Design ID: 3297121 – Laboratory work in YNEGLH Central Lab

Rating	Gene	HGNC	Primary Transcript	Slicing panels
				R92 Gene Slice 8 Haemolytic Anaemia
Green	*HBB*	HGNC:4827	NM_000518.4	R92 Gene Slice 1 Methaemoglobinaemia / R92 Gene Slice 8 Haemolytic Anaemia
Green	*HBD*	HGNC:4829	NM_000519.3	R92 Gene Slice 1 Methaemoglobinaemia / R92 Gene Slice 8 Haemolytic Anaemia
Green	*HBG1*	HGNC:4831	NM_000559.2	R92 Gene Slice 1 Methaemoglobinaemia / R92 Gene Slice 8 Haemolytic Anaemia
Green	*HBG2*	HGNC:4832	NM_000184.2	R92 Gene Slice 1 Methaemoglobinaemia / R92 Gene Slice 8 Haemolytic Anaemia
Green	*HK1*	HGNC:4922	NM_000188.2	R92 Gene Slice 4 Enzymopathy / R92 Gene Slice 8 Haemolytic Anaemia
Green	*HSPA9*	HGNC:5244	NM_004134.6	R92 Gene Slice 7 Sideroblastic Anaemia
Green	*KCNN4*	HGNC:6293	NM_002250.2	R92 Gene Slice 3 Membranopathy
Green	*KIF23*	HGNC:6392	NM_138555.3	R92 Gene Slice 5 Congenital Dyserythropoietic Anaemia
Green	*KLF1*	HGNC:6345	NM_006563.4	R92 Gene Slice 5 Congenital Dyserythropoietic Anaemia
Green	*LPIN2*	HGNC:14450	NM_014646.2	R92 Gene Slice 5 Congenital Dyserythropoietic Anaemia
Green	*MTR*	HGNC:7468	NM_000254.2	R92 Gene Slice 10 Megaloblastic Anaemia
Green	*MTRR*	HGNC:7473	NM_002454.2	R92 Gene Slice 10 Megaloblastic Anaemia
Green	*NT5C3A*	HGNC:17820	NM_016489.12	R92 Gene Slice 4 Enzymopathy / R92 Gene Slice 8 Haemolytic Anaemia
Green	*PFKM*	HGNC:8877	NM_000289.5	R92 Gene Slice 4 Enzymopathy / R92 Gene Slice 8 Haemolytic Anaemia
Green	*PIEZO1*	HGNC:28993	NM_001142864.3	R92 Gene Slice 3 Membranopathy / R92 Gene Slice 8 Haemolytic Anaemia
Green	*PKLR*	HGNC:9020	NM_000298.5	R92 Gene Slice 4 Enzymopathy / R92 Gene Slice 8 Haemolytic Anaemia
Green	*PUS1*	HGNC:15508	NM_025215.5	R92 Gene Slice 7 Sideroblastic Anaemia
Green	*RHAG*	HGNC:10006	NM_000324.2	R92 Gene Slice 3 Membranopathy / R92 Gene Slice 8 Haemolytic Anaemia
Green	*RPL11*	HGNC:10301	NM_000975.4	R92 Gene Slice 6 Diamond-Blackfan Anaemia
Green	*RPL15*	HGNC:10306	NM_002948.4	R92 Gene Slice 6 Diamond-Blackfan Anaemia
Green	*RPL26*	HGNC:10327	NM_000987.4	R92 Gene Slice 6 Diamond-Blackfan Anaemia
Green	*RPL27*	HGNC:10328	NM_000988.4	R92 Gene Slice 6 Diamond-Blackfan Anaemia
Green	*RPL31*	HGNC:10334	NM_001098577.2	R92 Gene Slice 6 Diamond-Blackfan Anaemia
Green	*RPL35A*	HGNC:10345	NM_000996.3	R92 Gene Slice 6 Diamond-Blackfan Anaemia

R92.1 :: Rare Anaemia (Version 1.2)
https://panelapp.genomicsengland.co.uk/panels/518/
Agilent SureSelect Reagent Design ID: 3297121 – Laboratory work in YNEGLH Central Lab

Rating	Gene	HGNC	Primary Transcript	Slicing panels
Green	*RPL5*	HGNC:10360	NM_000969.4	R92 Gene Slice 6 Diamond-Blackfan Anaemia
Green	*RPL9*	HGNC:10369	NM_000661.4	R92 Gene Slice 6 Diamond-Blackfan Anaemia
Green	*RPS10*	HGNC:10383	NM_001014.4	R92 Gene Slice 6 Diamond-Blackfan Anaemia
Green	*RPS17*	HGNC:10397	NM_001021.5	R92 Gene Slice 6 Diamond-Blackfan Anaemia
Green	*RPS19*	HGNC:10402	NM_001022.3	R92 Gene Slice 6 Diamond-Blackfan Anaemia
Green	*RPS24*	HGNC:10411	NM_033022.3	R92 Gene Slice 6 Diamond-Blackfan Anaemia
Green	*RPS26*	HGNC:10414	NM_001029.4	R92 Gene Slice 6 Diamond-Blackfan Anaemia
Green	*RPS27*	HGNC:10416	NM_001030.5	R92 Gene Slice 6 Diamond-Blackfan Anaemia
Green	*RPS29*	HGNC:10419	NM_001032.4	R92 Gene Slice 6 Diamond-Blackfan Anaemia
Green	*RPS7*	HGNC:10440	NM_001011.3	R92 Gene Slice 6 Diamond-Blackfan Anaemia
Green	*SBDS*	HGNC:19440	NM_016038.3	
Green	*SEC23B*	HGNC:10702	NM_006363.5	R92 Gene Slice 5 Congenital Dyserythropoietic Anaemia
Green	*SLC11A2*	HGNC:10908	NM_000617.2	
Green	*SLC19A2*	HGNC:10938	NM_006996.2	R92 Gene Slice 10 Megaloblastic Anaemia
Green	*SLC25A38*	HGNC:26054	NM_017875.3	R92 Gene Slice 7 Sideroblastic Anaemia
Green	*SLC2A1*	HGNC:11005	NM_006516.2	R92 Gene Slice 3 Membranopathy / R92 Gene Slice 8 Haemolytic Anaemia
Green	*SLC4A1*	HGNC:11027	NM_000342.3	R92 Gene Slice 3 Membranopathy / R92 Gene Slice 8 Haemolytic Anaemia
Green	*SPTA1*	HGNC:11272	NM_003126.3	R92 Gene Slice 3 Membranopathy / R92 Gene Slice 8 Haemolytic Anaemia
Green	*SPTB*	HGNC:11274	NM_001024858.3	R92 Gene Slice 3 Membranopathy / R92 Gene Slice 8 Haemolytic Anaemia
Green	*TCN2*	HGNC:11653	NM_000355.3	R92 Gene Slice 10 Megaloblastic Anaemia
Green	*TF*	HGNC:11740	NM_001063.3	
Green	*TMPRSS6*	HGNC:16517	NM_153609.3	
Green	*TPI1*	HGNC:12009	NM_000365.5	R92 Gene Slice 4 Enzymopathy / R92 Gene Slice 8 Haemolytic Anaemia
Green	*TRNT1*	HGNC:17341	NM_182916.2	R92 Gene Slice 7 Sideroblastic Anaemia
Green	*UMPS*	HGNC:12563	NM_000373.3	R92 Gene Slice 10 Megaloblastic Anaemia
Green	*XK*	HGNC:12811	NM_021083.3	R92 Gene Slice 3 Membranopathy / R92 Gene Slice 8 Haemolytic Anaemia

R92.1 :: Rare Anaemia (Version 1.2)
https://panelapp.genomicsengland.co.uk/panels/518/
Agilent SureSelect Reagent Design ID: 3297121 – Laboratory work in YNEGLH Central Lab

Rating	Gene	HGNC	Primary Transcript	Slicing panels
Green	*YARS2*	HGNC:24249	NM_001040436.2	R92 Gene Slice 9 Sitosterolaemia

R96.1 :: Iron Metabolism Disorders NOT common HFE mutations (Version 1.2)
https://panelapp.genomicsengland.co.uk/panels/515/
Agilent SureSelect Reagent Design ID: 3297111 – Laboratory work in YNEGLH Central Lab

Rating	Gene	HGNC	Primary Transcript	Slicing panels
Green	*ABCB7*	HGNC:48	NM_004299.5	
Green	*ALAS2*	HGNC:397	NM_000032.2	
Green	ATP7B	HGNC:870	NM_000053.3	
Green	BMP6	HGNC:1073	NM_001718.5	
Green	CP	HGNC:2295	NM_000096.3	
Green	CYBRD1	HGNC:20797	NM_024843.3	
Green	FTL	HGNC:3999	NM_000146.3	
Green	GBA	HGNC:4177	NM_001005741.2	
Green	*GLRX5*	HGNC:20134	NM_016417.2	
Green	HAMP	HGNC:15598	NM_021175.3	
Green	HFE	HGNC:4886	NM_000410.3	
Green	HJV (HFE2)	HGNC:4887	NM_213653.3	
Green	*SLC11A2*	HGNC:10908	NM_000617.2	
Green	*SLC25A38*	HGNC:26054	NM_017875.3	
Green	SLC40A1	HGNC:10909	NM_014585.5	
Green	*TF*	HGNC:11740	NM_001063.3	
Green	TFR2	HGNC:11762	NM_003227.3	
Green	*TMPRSS6*	HGNC:16517	NM_153609.3	

R97.1 :: Thrombophilia with a likely monogenic cause (Version 1.4)
https://panelapp.genomicsengland.co.uk/panels/516/
Agilent SureSelect Reagent Design ID: 3297111 – Laboratory work in YNEGLH Central Lab

Rating	Gene	HGNC	Primary Transcript	Slicing panels
Green	ADAMTS13	HGNC:1366	NM_139025.4	
Green	F2	HGNC:3535	NM_000506.4	
Green	F5	HGNC:3542	NM_000130.4	
Green	FGA	HGNC:3661	NM_021871.3	
Green	FGB	HGNC:3662	NM_005141.4	
Green	FGG	HGNC:3694	NM_000509.5	

R97.1 :: Thrombophilia with a likely monogenic cause (Version 1.4)
https://panelapp.genomicsengland.co.uk/panels/516/
Agilent SureSelect Reagent Design ID: 3297111 – Laboratory work in YNEGLH Central Lab

Rating	Gene	HGNC	Primary Transcript	Slicing panels
Green	HRG	HGNC:5181	NM_000412.4	
Green	PIGA	HGNC:8957	NM_002641.3	
Green	PLG	HGNC:9071	NM_000301.3	
Green	PROC	HGNC:9451	NM_000312.3	
Green	PROS1	HGNC:9456	NM_000313.3	
Green	SERPINC1	HGNC:775	NM_000488.3	
Green	SERPIND1	HGNC:4838	NM_000185.3	
Green	SERPINE1	HGNC:8583	NM_000602.4	
Green	THBD	HGNC:11784	NM_000361.2	

R124.1:: Combined factor V and VIII deficiency (Version 1.2)
https://panelapp.genomicsengland.co.uk/panels/517/
Agilent SureSelect Reagent Design ID: 3297111 – Laboratory work in YNEGLH Central Lab

Rating	Gene	HGNC	Primary Transcript	
Green	LMAN1	HGNC:6631	NM_005570.3	
Green	MCFD2	HGNC:18451	NM_139279.5	

R229 :: Confirmed Fanconi or Bloom Syndrome (Version 1.7)
https://panelapp.genomicsengland.co.uk/panels/508/
Agilent SureSelect Reagent Design ID: 3297111 – Laboratory work in YNEGLH Central Lab

Rating	Gene	HGNC	Primary Transcript	Slicing Panels
Green	BLM	HGNC:1058	NM_000057.4	
Green	BRCA1	HGNC:1100	NM_007294.4	
Green	BRCA2	HGNC:1101	NM_000059.4	
Green	BRIP1	HGNC:20473	NM_032043.3	
Green	ERCC4	HGNC:3464	NM_005236.3	
Green	FANCA	HGNC:3582	NM_000135.4	
Green	FANCB	HGNC:3583	NM_001018113.3	
Green	FANCC	HGNC:3584	NM_000136.3	
Green	FANCD2	HGNC:3585	NM_033084.6	
Green	FANCE	HGNC:3586	NM_021922.3	
Green	FANCF	HGNC:3587	NM_022725.4	
Green	FANCG	HGNC:3588	NM_004629.2	

R229 :: Confirmed Fanconi or Bloom Syndrome (Version 1.7)
https://panelapp.genomicsengland.co.uk/panels/508/
Agilent SureSelect Reagent Design ID: 3297111 – Laboratory work in YNEGLH Central Lab

Rating	Gene	HGNC	Primary Transcript	Slicing Panels
Green	FANCI	HGNC:25568	NM_001113378.2	
Green	FANCL	HGNC:20748	NM_018062.4	
Green	PALB2	HGNC:26144	NM_024675.4	
Green	SLX4	HGNC:23845	NM_032444.4	
Green	TOP3A	HGNC:11992	NM_004618.5	
Green	UBE2T	HGNC:25009	NM_014176.4	

R405.1 :: Hereditary Erythrocytosis (Version 1.19)
https://panelapp.genomicsengland.co.uk/panels/157/
Agilent SureSelect Reagent Design ID: 3297101 – Laboratory work in YNEGLH Central Lab

Rating	Gene	HGNC	Primary Transcript	Slicing Panels
Green	*EGLN1*	HGNC:1232	NM_022051.3	
Green	*EPAS1*	HGNC:3374	NM_001430.5	
Green	*EPO*	HGNC:3415	NM_000799.4	
Green	*EPOR*	HGNC:3416	NM_000121.4	
Green	*HBA1*	HGNC:4823	NM_000558.4	
Green	*HBA2*	HGNC:4824	NM_000517.4	
Green	*HBB*	HGNC:4827	NM_000518.4	
Green	*VHL*	HGNC:12687	NM_000551.4	

R406.1 :: Thrombocythemia (Version 1.1)
https://panelapp.genomicsengland.co.uk/panels/945/
Agilent SureSelect Reagent Design ID: 3297101 – Laboratory work in YNEGLH Central Lab

Rating	Gene	HGNC	Primary Transcript	Slicing panels
Green	*JAK2*	HGNC:6192	NM_004972.4	
Green	*MPL*	HGNC:7217	NM_005373.2	
Green	*SH2B3*	HGNC:29605	NM_005475.3	
Green	*THPO*	HGNC:11795	NM_000460.3	

Gastrohepatology

SureSelect Gastrohepatology panel ID: Gastrohep LIVE v3 Design ID: 3268241 (wet work in Leeds Central NEY GLH lab

R171: Cholestasis

Condition	OMIM	Gene	Primary transcript

R171: Cholestasis

Condition	OMIM	Gene	Primary transcript
Progressive familial intrahepatic cholestasis 2	603201	ABCB11	NM_003742.2
Progressive familial intrahepatic cholestasis 3	602347	ABCB4	NM_000443.3
Dubin-Johnson syndrome	601107	ABCC2	NM_000392.4
Congenital bile acid synthesis defect 2	604741	AKR1D1	NM_005989.3
Hereditary fructose intolerance	612724	ALDOB	NM_000035.3
Congenital bile acid synthesis defect 4	604489	AMACR	NM_014324.5
Progressive familial intrahepatic Cholestasis1	602397	ATP8B1	NM_005603.5
Familial Hypercholanemia	602938	BAAT	NM_001701.3
Mitochondrial complex III deficiency, nuclear type 1	603647	BCS1L	NM_004328.4
Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis	603718	CLDN1	NM_021101.4
Cerebrotendinous xanthomatosis	606530	CYP27A1	NM_000784.3
Bile acid synthesis	118455	CYP7A1	NM_000780.3
Congenital bile acid synthesis defect 3	603711	CYP7B1	NM_004820.4
Sclerosing cholangitis, neonatal	605755	DCDC2	NM_016356.4
Tyrosinemia, type I	613871	FAH	NM_000137.2
Congenital bile acid synthesis defect 1	607764	HSD3B7	NM_025193.3
Alagille syndrome 1	601920	JAG1	NM_000214.2
Microvillus inclusion disease	606540	MYO5B	NM_001080467.2
Alagille syndrome 2	600275	NOTCH2	NM_024408.3
Niemann-Pick disease type C1	607623	NPC1	NM_000271.4
Niemann-pick disease type C2	601015	NPC2	NM_006432.3
Progressive familial intrahepatic cholestasis-5	603826	NR1H4	NM_005123.3
Peroxisome biogenesis disorder 1A (or 1B)	602136	PEX1	NM_000466.2
Peroxisome biogenesis disorder 3A (or 3B)	601758	PEX12	NM_000286.2
Peroxisome biogenesis disorder 7A (or 7B)	608666	PEX26	NM_017929.5
Peroxisome biogenesis disorder 4A (or 4B)	601498	PEX6	NM_000287.3
Alpha-1-Antitrypsin Deficiency	613490	SERPINA1	NM_000295.4
Citrullinemia, type II or neonatal intrahepatic cholestasis caused by citrin deficiency	603859	SLC25A13	NM_014251.2
Transaldolase deficiency	602063	TALDO1	NM_006755.1
Familial Hypercholanemia	607709	TJP2	NM_004817.3
Hyperbilirubinemia Crigler-Najjar syndrome, type I	191740	UGT1A1	NM_000463.2
Arthrogryposis, renal dysfunction, and cholestasis 2	613401	VIPAS39	NM_022067.3
Arthrogryposis, renal dysfunction, and cholestasis 1	608552	VPS33B	NM_018668.4

R173: Polycystic Liver disease

Condition	OMIM	Gene	Primary transcript
Polycystic liver disease 1	174050	*PRKCSH*	NM_002743.3
Polycystic liver disease 2	617004	*SEC63*	NM_007214.4

R171: Cholestasis			
Condition	**OMIM**	**Gene**	**Primary transcript**
Polycystic liver disease 4 with or without kidney cysts	617875	*LRP5*	NM_002335.2
Polycystic kidney disease 3	600666	*GANAB*	NM_198335.3
Polycystic kidney disease 4,with hepatic disease	263200	*PKHD1*	NM_138694.3
Polycystic liver disease 3 with or without kidney cysts	617874	*ALG8*	NM_024079.4
Polycystic kidney disease 6 with or without polycystic liver disease	618061	*DNAJB11*	NM_016306.5
Polycystic kidney disease 1	173900	*PKD1*	NM_001009944.2
Polycystic kidney disease 2	613095	*PKD2*	NM_000297.3

R331: Intestinal failure			
Condition	**OMIM**	**Gene**	**Primary transcript**
Congenital secretory sodium diarrhea 3	270420	*SPINT2*	NM_021102.3
Trichohepatoenteric syndrome 1	614589	*TTC37*	NM_014639.3
Diarrhea 7, protein-losing enteropathy type	604900	*DGAT1*	NM_012079.5
Familial hemophagocytic lymphohistiocytosis-5	613101	*STXBP2*	NM_006949.3
Trichohepatoenteric syndrome 2	600478	*SKIV2L*	NM_006929.4
Diarrhea 6	601330	*GUCY2C*	NM_004963.3
Microvillus inclusion disease	600876	*STX3*	NM_004177.4
Congenital secretory sodium diarrhea 8	616868	*SLC9A3*	NM_004174.3
Diarrhea 5, with congenital tufting enteropathy	613217	*EPCAM*	NM_002354.2
Microvillus inclusion disease	606540	*MYO5B*	NM_001080467.2
Congenital secretory chloride Diarrhea 1	214700	*SLC26A3*	NM_000111.2

S5 Gastrohepatology special panel ID: WG_IAD186831_improve.20191114 (wet work in Sheffield)

R172 Wilson Disease			
Condition	**OMIM**	**Gene**	**Primary transcript**
Wilson Disease	606882	*ATP7B*	NM_000053.4

R175: Pancreatitis			
Condition	**OMIM**	**Gene**	**Primary transcript**
Hereditary Pancreatitis	167790	*SPINK1*	NM_003122.5
Hereditary Pancreatitis	602421	*CFTR*	NM_000492.4
Hereditary Pancreatitis	276000	*PRSS1*	NM_002769.5

R177: Hirschsprung disease			
Condition	**OMIM**	**Gene**	**Primary transcript**
Hirschsprung disease	164761	*RET*	NM_020975.6

Respiratory

SureSelect Design ID: 3188541. Laboratory wetwork in NEYGLH Central lab

R189 Respiratory ciliopathies including non-CF bronchiectasis		
Condition	Gene	Primary transcript
Primary ciliary dyskinesia 23	ARMC4	**NM_018076.4**
Primary ciliary dyskinesia 26	C21orf59	NM_021254.3
Primary ciliary dyskinesia 17	CCDC103	NM_213607.2
Primary ciliary dyskinesia 20	CCDC114	NM_144577.3
Primary ciliary dyskinesia 30	CCDC151	NM_145045.4
Primary ciliary dyskinesia 14	CCDC39	NM_181426.1
Primary ciliary dyskinesia 15	CCDC40	NM_017950.3
Primary ciliary dyskinesia 27	CCDC65	NM_033124.4
Primary ciliary dyskinesia 29	CCNO	NM_021147.4
Primary ciliary dyskinesia 38	CFAP300/ C11orf70	NM_032930.2
Cystic fibrosis	CFTR	NM_000492.3
Primary ciliary dyskinesia 13	DNAAF1	NM_178452.5
Primary ciliary dyskinesia 10	DNAAF2	NM_018139.2
Primary ciliary dyskinesia 2	DNAAF3	NM_178837.4
Primary ciliary dyskinesia 25	DNAAF4	NM_130810.3
Primary ciliary dyskinesia 18	DNAAF5	NM_017802.3
Primary ciliary dyskinesia 7	DNAH11	NM_001277115.1
Primary ciliary dyskinesia 3	DNAH5	NM_001369.2
Primary ciliary dyskinesia 40	DNAH9	NM_001372.4
Primary ciliary dyskinesia 1	DNAI1	NM_012144.3
Primary ciliary dyskinesia 9	DNAI2	NM_023036.5
Primary ciliary dyskinesia 34	DNAJB13	NM_153614.3
Primary ciliary dyskinesia 16	DNAL1	NM_031427.3
Primary ciliary dyskinesia 21	DRC1	NM_145038.4
Primary ciliary dyskinesia 33	GAS8	NM_001481.2
Primary ciliary dyskinesia 5	HYDIN	NM_001270974.2
Primary ciliary dyskinesia 21	LRRC56	NM_198075.3
Primary ciliary dyskinesia 19	LRRC6	NM_012472.5
Primary ciliary dyskinesia 42	MCIDAS	NM_001190787.2
Immunodeficiency	NFKB1	NM_003998.3
Immunodeficiency	NFKB2	NM_001288724.1
Primary ciliary dyskinesia 36	PIH1D3	NM_173494.2
Immunodeficiency	PIK3CD	NM_005026.3
Immunodeficiency	PIK3R1	NM_181523.2
Combined immunodeficiency	RAG1	NM_000448.2
Combined immunodeficiency	RAG2	NM_000536.2
Primary ciliary dyskinesia 24	RSPH1	NM_080860.3
Primary ciliary dyskinesia 32	RSPH3	NM_031924.4
Primary ciliary dyskinesia 11	RSPH4A	NM_001010892.2
Primary ciliary dyskinesia 12	RSPH9	NM_152732.4
Pseudohypoaldosteronism type I	SCNN1A	NM_001038.6

Bronchiectasis	SCNN1B	NM_000336.3
Bronchiectasis	SCNN1G	NM_001039.4
Primary ciliary dyskinesia 28	SPAG1	NM_003114.4
Primary ciliary dyskinesia 35	TTC25	NM_031421.5
Primary ciliary dyskinesia 22	ZMYND10	NM_015896.3
R188 Pulmonary Arterial Hypertension		
HHT	ACVRL1	NM_000020.2
Pulmonary arterial hypertension	ATP13A3	NM_024524.3
Pulmonary arterial hypertension	BMPR2	NM_001204.6
Pulmonary arterial hypertension	EIF2AK4	NM_001013703.4
HHT	ENG	NM_000118.3
Pulmonary arterial hypertension	GDF2	NM_016204.3
Pulmonary arterial hypertension	KCNK3	NM_002246.2
Pulmonary arterial hypertension	SMAD9	NM_001127217.2
Pulmonary arterial hypertension	SOX17	NM_022454.3
R186 Hereditary Haemorrhagic Telangiectasia		
HHT	ACVRL1	NM_000020.2
	ENG	NM_000118.3
	EPHB4	NM_004444.4
	SMAD4	NM_005359.5
R139 Laterality disorders and isomerism		
Primary ciliary dyskinesia 23	ARMC4	NM_018076.4
Primary ciliary dyskinesia 26	C21orf59	NM_021254.3
Primary ciliary dyskinesia 17	CCDC103	NM_213607.2
Primary ciliary dyskinesia 20	CCDC114	NM_144577.3
Primary ciliary dyskinesia 30	CCDC151	NM_145045.4
Primary ciliary dyskinesia 14	CCDC39	NM_181426.1
Primary ciliary dyskinesia 15	CCDC40	NM_017950.3
Primary ciliary dyskinesia 38	CFAP300/ C11orf70	NM_032930.2
Primary ciliary dyskinesia 13	DNAAF1	NM_178452.5
Primary ciliary dyskinesia 2	DNAAF3	NM_178837.4
Primary ciliary dyskinesia 25	DNAAF4	NM_130810.3
Primary ciliary dyskinesia 18	DNAAF5	NM_017802.3
Primary ciliary dyskinesia 7	DNAH11	NM_001277115.1
Primary ciliary dyskinesia 3	DNAH5	NM_001369.2
Primary ciliary dyskinesia 1	DNAI1	NM_012144.3
Primary ciliary dyskinesia 9	DNAI2	NM_023036.5
Primary ciliary dyskinesia 21	LRRC56	NM_198075.3
Primary ciliary dyskinesia 19	LRRC6	NM_012472.5
Primary ciliary dyskinesia 36	PIH1D3	NM_173494.2
Primary ciliary dyskinesia 28	SPAG1	NM_003114.4
Primary ciliary dyskinesia 22	ZMYND10	NM_015896.3
Visceral heterotaxy 6	CFAP53	NM_145020.4
Right atrial Isomerism	GDF1	NM_001492.6
Visceral heterotaxy 7	MMP21	NM_147191.1
Visceral heterotaxy 5	NODAL	NM_018055.5
Visceral heterotaxy 1	ZIC3	NM_003413.4

Primary ciliary dyskinesia 40	DNAH9	NM_001372.4
R190 Pneumothorax		
Vascular EDS	COL3A1	NM_000090.3
Marfan syndrome	FBN1	NM_000138.4
Primary spontaneous pneumothorax, Birt-Hogg-Dube syndrome	FLCN	NM_144997.5
Emphysema due to AA1 deficiency	SERPINA1	NM_000295.5
Loeys-Dietz syndrome	TGFB2	NM_001135599.2
Loeys-Dietz syndrome	TGFB3	NM_003239.2
Loeys-Dietz syndrome	TGFBR1	NM_004612.2
Loeys-Dietz syndrome	TGFBR2	NM_003242.5
Tuberous sclerosis 1	TSC1	NM_000368.4
Tuberous sclerosis 2	TSC2	NM_000548.3
R192 Surfactant deficiency		
Surfactant deficiency 3	ABCA3	NM_001089.2
Neuroendocrine cell hyperplasia of infancy	NKX2-1	NM_001079668.2
Surfactant deficiency 1	SFTPB	NM_198843.3
Surfactant deficiency 2	SFTPC	NM_003018.3

Oncology

Pharmacogenetics solid tumour panels:

Oncomine Focus NGS Assay (DNA and Fusions)	https://www.thermofisher.com/uk/en/home/clinical/preclinical-companion-diagnostic-development/oncomine-oncology/oncomine-focus-assay.html

Pan Cancer panels:

Illumina TruSight RNA fusion panel	https://www.illumina.com/content/dam/illumina-marketing/documents/products/gene_lists/gene_list_trusight_rna_fusion_panel.xlsx

Haemato Oncology panels:

Fluidigm custom 48x48 access array	Gene	Primary transcript
Targeted Analysis	ASXL1	NM_015338.6
	BCOR	NM_001123385.1
	CALR	NM_004343.3
	CBL	NM_005188.4
	CEBPA (sanger sequencing)	NM_001287424.1
	CSF3R	NM_156039.3

	FLT3	NM_004119.2
	IDH1	NM_005896.4
	IDH2	NM_001289910.1
	JAK2	NM_001322194.2
	KIT	NM_000222.3
	KRAS	NM_033360.4
	MPL	NM_005373.3
	NPM1	NM_002520.7
	NRAS	NM_002524.5
	RHOA	NM_001664.4
	RUNX1	NM_001001890.3
	SETBP1	NM_001130110.2
	SF3B1	NM_001005526.2
	SRSF2	NM_003016.4
	STAT3	NM_139276.3
	TP53	NM_000546.6
	U2AF1	NM_006758.3
	WT1	NM_000378.6
	ZRSR2	NM_005089.4
Whole Gene analysis	DNMT3A	NM_022552.5
	EZH2	NM_004456.5
	STAG2	NM_006603.5
	TET2	NM_017628.4