Next Generation Sequencing Service at SDGS

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Testing Workflow

Library Preparation

- 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.

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).
- 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.
- 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).

Post analysis

- Confirmation of clinically significant sequence variants by Sanger sequencing 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 have uncertain clinical significance.

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]), when detected by SureSelect NGS workflows run on the MiSeq and HiSeq2500 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

Next Generation Sequencing Panels

Inborn Errors of Metabolism

(SureSelect Design ID: 0660881)

Glycogen Storage Diseases (GSDs)			
Condition	**Gene**	**Subpanel(s)**	**Primary transcript**
Glycogen storage disease III	*AGL*	Liver/Muscle/Heart	NM_000642.2
Glycogen storage disease XII	*ALDOA*	Muscle	NM_000034.3
Glycogen storage disease XIII	*ENO3*	Muscle	NM_001976.4
Epilepsy, progressive myoclonic 2A (Lafora)	*EPM2A*	Generalised	NM_005670.3
Fructose-1,6-bisphosphatase deficiency	*FBP1*	Liver	NM_000507.3
N/A - no mutations reported in gene to date	*FBP2*	Muscle	NM_003837.2
Glycogen storage disease Ia	*G6PC*	Liver	NM_000151.2
Glycogen storage disease II	*GAA*	Generalised/Muscle	NM_000152.3
Glycogen storage disease IV / Adult polyglucosan body disease	*GBE1*	Liver/Muscle/Heart	NM_000158.3
Glycogen storage disease XV	*GYG1*	Muscle/Heart	NM_004130.3
N/A - no mutations reported in gene to date	*GYG2*	Liver	NM_003918.2
Glycogen storage disease 0, muscle	*GYS1*	Muscle/Heart	NM_002103.4
Glycogen storage disease, type 0	*GYS2*	Liver	NM_021957.3
Danon disease	*LAMP2*	Generalised	NM_002294.2
Glycogen storage disease XI	*LDHA*	Muscle	NM_005566.3
Epilepsy, progressive myoclonic 2B (Lafora)	*NHLRC1*	Generalised	NM_198586.2
Hemolytic anaemia due to phosphofructokinase deficiency	*PFKL*	Liver	NM_002626.4
Glycogen storage disease VII	*PFKM*	Muscle	NM_00116686.1
Glycogen storage disease X	*PGAM2*	Muscle	NM_000290.3
Phosphoglycerate kinase 1 deficiency	*PGK1*	Muscle	NM_000291.3
GSD XIV, Phosphoglucomutase 1 deficiency	*PGM1*	Muscle	NM_002633.2
GSD Ixd; muscle phosphorylase kinase deficiency	*PHKA1*	Muscle	NM_002637.3
Glycogen storage disease IXa1; GSD9A1	*PHKA2*	Liver	NM_000292.2
Glycogen storage disease IXb; GSD9B	*PHKB*	Liver	NM_000293.2
N/A - no mutatiopns reported in gene to date	*PHKG1*	Muscle	NM_006213.3
Glycogen storage disease IXc	*PHKG2*	Liver	NM_000294.2
Glycogen storage disease of heart, lethal congenital	*PRKAG2*	Heart	NM_016203.3
Glycogen storage disease VI	*PYGL*	Liver	NM_002863.4
Glycogen storage disease, type V; McArdle disease	*PYGM*	Muscle	NM_005609.2
RBCK1 deficiency	*RBCK1*	Muscle/Heart	NM_031229.2
Fanconi-Bickel syndrome	*SLC2A2*	Liver	NM_000340.1
Glycogen storage disease Ib / Ic	*SLC37A4*	Liver	NM_001164277.1

32 genes

Fatty Acid Metabolism Disorders			
Condition	**Gene**	**Subpanel(s)**	**Primary transcript**
ACAD9 deficiency (complex I deficiency)	*ACAD9*	FAO	NM_014049.4
MCAD deficiency	*ACADM*	FAO	NM_000016.4
SCAD deficiency	*ACADS*	SCAD	NM_000017.2
VLCAD deficiency	*ACADVL*	FAO	NM_000018.2
Beta-ketothiolase deficiency	*ACAT1*	Ketolysis	NM_000019.3
CPTI deficiency	*CPT1A*	FAO	NM_001876.3
CPTI deficiency (muscle)	*CPT1B*	FAO	NM_004377.3
CPTII deficiency	*CPT2*	FAO	NM_000098.2
MADD	*ETFA*	FAO	NM_000126.3
MADD	*ETFB*	FAO	NM_001985.2
MADD (riboflavin-responsive)	*ETFDH*	FAO	NM_004453.2
HADH (SCHAD) deficiency	*HADH*	FAO	NM_001184705.2
Mitochondrial TFP deficiency	*HADHA*	FAO	NM_000182.4
Mitochondrial TFP deficiency	*HADHB*	FAO	NM_000183.2
Mitochondrial HMG-CoA lyase deficiency	*HMGCL*	FAO	NM_000191.2
Mitochondrial HMG-CoA synthase deficiency	*HMGCS2*	FAO	NM_005518.3
SCOT deficiency	*OXCT1*	Ketolysis	NM_000436.3
Systemic primary carnitine deficiency (carnitine transporter)	*SLC22A5*	FAO	NM_003060.3
CACT deficiency	*SLC25A20*	FAO	NM_000387.5
Riboflavin deficiency (RFVT1)	*SLC52A1*	FAO	NM_001104577.1
Brown-Vialetto-van Laere syndrome (RFVT2)	*SLC52A2*	FAO	NM_024531.4
Brown-Vialetto-van Laere syndrome (RFVT3)	*SLC52A3*	FAO	NM_033409.3

22 genes

FAO - Fatty acid oxidaton/ketogenesis

Ketolysis - Ketolysis

SCAD - Suspected SCAD (by specific request secondary to/combined with FAO panel)

Peroxisomal Disorders		
Condition	**Gene**	**Primary transcript**
X-linked adrenoleukodystrophy	*ABCD1*	NM_000033.3
Peroxisomal Acyl-CoA oxidase deficiency	*ACOX1*	NM_004035.6
Rhizomelic chondrodysplasia punctata type 3 (RCDP3)	*AGPS*	NM_003659.3
Type I primary hyperoxaluria (HP1)	*AGXT*	NM_000030.2
Alpha-methylacyl-CoA racemase (AMACR) deficiency	*AMACR*	NM_014324.5
Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission	*DNM1L*	NM_012062.4
Rhizomelic chondrodysplasia punctata type 2 (RCDP2) (DHAPAT deficiency)	*GNPAT*	NM_014236.3
D-Bifunctional protein deficiency	*HSD17B4*	NM_001199291.1
Peroxisome Biogenesis Disorders, Zellweger Syndrome Spectrum	*PEX1*	NM_000466.2
Peroxisome Biogenesis Disorders, Zellweger Syndrome Spectrum	*PEX2*	NM_000318.2
Peroxisome Biogenesis Disorders, Zellweger Syndrome Spectrum	*PEX3*	NM_003630.2
Peroxisome Biogenesis Disorders, Zellweger Syndrome Spectrum	*PEX5*	NM_001131023.1
Peroxisome Biogenesis Disorders, Zellweger Syndrome Spectrum	*PEX6*	NM_000287.3
Rhizomelic chondrodysplasia punctata type 1 (RCDP1)	*PEX7*	NM_000288.3
Peroxisome Biogenesis Disorders, Zellweger Syndrome Spectrum	*PEX10*	NM_002617.3
Peroxisome Biogenesis Disorders, Zellweger Syndrome Spectrum	*PEX11B*	NM_003846.2
Peroxisome Biogenesis Disorders, Zellweger Syndrome Spectrum	*PEX12*	NM_000286.2
Peroxisome Biogenesis Disorders, Zellweger Syndrome Spectrum	*PEX13*	NM_002618.3
Peroxisome Biogenesis Disorders, Zellweger Syndrome Spectrum	*PEX14*	NM_004565.2
Peroxisome Biogenesis Disorders, Zellweger Syndrome Spectrum	*PEX16*	NM_004813.2
Peroxisome Biogenesis Disorders, Zellweger Syndrome Spectrum	*PEX19*	NM_002857.3
Peroxisome Biogenesis Disorders, Zellweger Syndrome Spectrum	*PEX26*	NM_017929.5
Adult Refsum disease	*PHYH*	NM_006214.3
Leukoencephalopathy with dystonia and motor neuropathy	*SCP2*	NM_002979.4
	24 genes	

Hyperammonaemia / Urea Cycle Disorders			
Condition	**Gene**	**Subpanel(s)**	**Primary transcript**
Arginase deficiency (hyperargininemia)	*ARG1*	UCD/Hyperamm	NM_000045.2
Argininosuccinic aciduria	*ASL*	UCD/Hyperamm	NM_000048.3
Citrullinemia type I	*ASS1*	UCD/Hyperamm	NM_000050.4
Carbamoylphosphate synthetase I (CPS1) deficiency	*CPS1*	CPS_NAGS/UCD/ Hyperamm	NM_001875.3
Hyperinsulinism-hyperammonemia syndrome	*GLUD1*	Hyperamm	NM_005271.3
MMA (mutase)	*MUT*	Hyperamm	NM_000255.3
N-acetyl glutamate synthetase (NAGS) deficiency	*NAGS*	CPS_NAGS/UCD/ Hyperamm	NM_153006.2
Ornithine aminotransferase (OAT) deficiency	*OAT*	Hyperamm	NM_000274.3
Ornithine transcarbamylase (OTC) deficiency	*OTC*	UCD/Hyperamm	NM_000531.5
PA	*PCCA*	Hyperamm	NM_000282.3
PA	*PCCB*	Hyperamm	NM_001178014.1
Citrullinaemia type 2 (citrin deficiency)	*SLC25A13*	UCD/Hyperamm	NM_014251.2
HHH syndrome	*SLC25A15*	UCD/Hyperamm	NM_014252.3
Lysinuric protein intolerance	*SLC7A7*	Hyperamm	NM_001126105.2

14 genes

CPS_NAGS - CPS1 and NAGS only
UCD - Urea cycle disorders
Hyperamm - Hyperammonaemia

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	*ETFAO*	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

Connective Tissue Disorders

(SureSelect Design ID: 0782441).

Ehlers Danlos Syndrome (EDS)		
Subpanel	**Gene**	**Primary transcript**
Vascular	*COL3A1*	NM_000090.3
Classical	*COL5A1*	NM_000093.3
	COL5A2	NM_000393.3
Kyphoscoliotic	*B4GALNT7*	NM_007255.2
	CHST14	NM_130468.3
	DSE	NM_001080976
	FKBP14	NM_017946.2
	PLOD2	NM_000302.3
	PRDM5	NM_018699.2
	RIN2	NM_001242581.1
	SLC39A13	NM_001128225.2
	ZNF469	NM_00127464.1
	TGFBR1	NM_004612.2
	TGFBR2	NM_001024847.2
Dermatosparaxis	*ADAMTS2*	NM_014244.4
EDS-Arthrochalasic	*COL1A1* (Exons 6-10)	NM_000088.3
	COL1A2 (Exons 6-10)	NM_000089.3

Familial Thoracic Aortic Aneurysm (FTAA) panel		
Condition	**Gene**	**Primary transcript**
FTAA	CBS	NM_000071.2
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

Osteogenesis Imperfecta (OI) panels		
Subpanels	**Gene**	**Primary transcript**
Dominant, Extended	*COL1A1*	NM_001025295.2
Dominant, Extended	*COL1A2*	NM_006129.4
Dominant	*IFITM5 (*c.-14 only)	NM_001199.3
Recessive, Extended	*BMP1*	NM_052854.3
Recessive, Extended	*BMP1*	NM_006371.4
Recessive, Extended	*CREB3L1*	NM_021939.3
Recessive, Extended	*CRTAP*	NM_001025295.2
Recessive, Extended	*FKBP10*	NM_22356.3
Recessive, Extended	*IFITM5* full	NM_152860.1
Recessive, Extended	*LEPRE1* (*P3H1*)	NM_000918.2
Recessive, Extended	*OSX* (*SP7*)	NM_182943.2
Recessive, Extended	*P4HB*	NM_005032.6
Recessive, Extended	*PLOD2*	NM_000942.4
Recessive, Extended	*PLS3*	NM_014822.2
Recessive, Extended	*PPIB*	NM_000942.4
Recessive, Extended	*SEC24D*	NM_014822.2
Recessive, Extended	*SERPINF1*	NM_002615.5
Recessive, Extended	*SERPINH1*	NM_001235.2
Recessive, Extended	*SPARC*	NM_003118.3
Recessive, Extended	*TAPT1*	NM_153365.2
Recessive, Extended	*TMEM38B*	NM_018112.1
Recessive, Extended	*WNT1*	NM_005430.3
Recessive, Extended	*XYLT2*	NM_022167.3

Familial Porencephaly (FP) panel		
Condition	**Gene**	**Primary transcript**
Familial Porencephaly	*COL4A1*	NM_001845.4
Familial Porencephaly	*COL4A2*	NM_001846.2

Alport Syndrome (AP) panel		
Condition	**Gene**	**Primary transcript**
Alport Syndrome	*COL4A3*	NM_000091.4
Alport Syndrome	*COL4A4*	NM_000092.4
Alport Syndrome	*COL4A5*	NM_000495.3

Bethlem Myopathy panel		
Condition	**Gene**	**Primary transcript**
Bethlem Myopathy	*COL6A1*	NM_001848.2
Bethlem Myopathy	*COL6A2*	NM_001849.3
Bethlem Myopathy	*COL6A3*	NM_004369.3

Cutis Laxa (CL) panel		
Condition	**Gene**	**Primary transcript**
Cutis Laxa	*ATP6V0A2*	NM_012463.3
Cutis Laxa	*ATP7A*	NM_000052.4
Cutis Laxa	*EFEMP2(FBLN4)*	NM_016938.4
Cutis Laxa	*ELN*	NM_000501.2
Cutis Laxa	*FBLN5*	NM_006329.3
Cutis Laxa	*LTBP4*	NM_003573.2 (NG_021201.1)
Cutis Laxa	*PYCR1*	NM_006907.2

Stickler Syndrome panel		
Condition	**Gene**	**Primary transcript**
Stickler syndrome	*COL11A1*	NM_001854.3
Stickler syndrome	*COL11A1*	NM_080629.2
Stickler syndrome	*COL11A2*	NM_080680.2
Stickler syndrome	*COL2A1*	NM_001844.4
Stickler syndrome	*COL9A1*	NM_001851.4
Stickler syndrome	*COL9A2*	NM_001852.3
Stickler syndrome	*COL9A3*	NM_001853.3

Hereditary Cancer Panels

(SureSelect Design ID: 3014041).

Breast and Ovarian gene panel (Primary)			
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 (By request)	151623	*TP53*	NM_000546.5

Ovarian gene panel (Extended)			
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, early-onset	114480	*BRIP1*	NM_032043.2
Breast cancer, susceptibility to	114480	*PALB2*	NM_024675.3
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

Breast and Ovarian gene panel (Extended)			
Condition	**OMIM**	**Gene**	**Primary transcript**
Breast cancer susceptibility	114480	*ATM*	NM_000051.3
Tumor predisposition syndrome	614327	*BAP1*	NM_004656.3
Breast-ovarian cancer, familial, 1	604370	*BRCA1*	NM_007294.3
Breast-ovarian cancer, familial, 2	612555	*BRCA2*	NM_000059.3
Breast cancer, early-onset	114480	*BRIP1*	NM_032043.2
Breast cancer, lobular	114480	*CDH1*	NM_004360.4
Breast cancer, susceptibility to	114480	*CHEK2*	NM_007194.3
Breast cancer, susceptibility to	114480	*PALB2*	NM_024675.3
Breast-ovarian cancer, familial, susceptibility to, 3	613399	*RAD51C*	NM_058216.2
Breast-ovarian cancer, familial, susceptibility to, 4	614291	*RAD51D*	NM_002878.3
Li-Fraumeni Syndrome	151623	*TP53*	NM_000546.5
Cowden Syndrome	158350	*PTEN*	NM_000314.4
Peutz-Jeghers Syndrome	175200	*STK11*	NM_000455.4
Breast cancer	114480	*PPM1D*	NM_003620.3
Nijmegen Breakage Syndrome	251260	*NBN*	NM_002485.4

Colorectal Cancer gene panel (Primary – HNPCC/Lynch syndrome)			
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 (By request)	614337	*PMS2*	NM_000535.5

Colorectal Cancer gene panel (Primary - FAP)			
Condition	**OMIM**	**Gene**	**Primary transcript**
Familial Adenamatous Polyposis	175100	*APC*	NM_000038.5
Colorectal cancer, hereditary nonpolyposis, type 1	120435	*MUTYH*	NM_001128425.1

Colorectal Cancer gene Panel (Extended)			
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 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
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			
Condition	**OMIM**	**Gene**	**Primary transcript**
Paragangliolas 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

Renal Cancer gene Panel			
Condition	**OMIM**	**Gene**	**Primary transcript**
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

Fanconi Anaemia			
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

Polycystic Disease Panels

(SureSelect Design ID: 3144591).

Autosomal Dominant Polycystic Kidney Disease – primary panel			
Condition	**OMIM**	**Gene**	**Primary transcript**
Polycystic kidney disease 1	173900	*PKD1*	NM_001009944.2
Polycystic kidney disease 2	613095	*PKD2*	NM_000297.3

Autosomal Recessive Polycystic Kidney Disease			
Condition	**OMIM**	**Gene**	**Primary transcript**
Polycystic kidney disease 4	263200	*PKHD1*	NM_138694.3
Polycystic kidney disease 5	617610	*DZIP1L*	NM_173543.2

Autosomal Dominant 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
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 liver disease	609214	*SEC61B*	NM_006808.2
Polycystic kidney disease 6 with or without polycystic liver disease	618061	*DNAJB11*	NM_016306.5

Autosomal Dominant Tubulointerstitial Kidney Disease (aka Medullary cystic and Glomerulocystic kidney disease)			
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

Polycystic Kidney and Liver Disease full panel			
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
Polycystic 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

Neurogenetic Motor Disorders

(SureSelect Design ID: 0836801).

Hereditary Spastic Paraparesis 88 gene panel			
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
Spastic paraplegia		*FARS2*	NM_006567.3
Amyotrophic lateral sclerosis 11, ALS11	612577	*FIG4*	NM_014845.5
Spastic paraplegia 68, SPG68		*FLRT1*	NM_013280.4

Hereditary Spastic Paraparesis 88 gene panel			
Condition	**OMIM**	**Gene**	**Primary transcript**
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
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

Hereditary Spastic Paraparesis 88 gene panel			
Condition	**OMIM**	**Gene**	**Primary transcript**
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			
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
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 29 gene panel			
Condition	**OMIM**	**Gene**	**Primary transcript**
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			
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
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

Familial Amyotrophic Lateral Sclerosis with or without Frontotemporal Dementia 42 gene panel			
Condition	**OMIM**	**Gene**	**Primary transcript**
Frontotemporal Dementia, FTD	600274	*MAPT*	NM_001123066.3
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 gene 27 gene panel			
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

Dementia gene 27 gene panel			
Condition	**OMIM**	**Gene**	**Primary transcript**
Perry syndrome	168605	*DCTN1*	NM_004082.4
Neuropathy, hereditary sensory, type IE, HSN1E	614116	*DNMT1*	NM_001005360.2
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_001123066.3
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: 0658791)

Dystonia and Parkinsonism gene 27 gene panel			
Condition	**OMIM**	**Gene**	**Primary transcript**
Ataxia, spastic, 5, autosomal recessive; SPAX5	614487	*AFG3L2*	NM_006796.2
Dystonia 24; DYT24	615034	*ANO3*	NM_031418.2
Dystonia-12; DYT12	128235	*ATP1A3*	NM_152296.4
Wilson disease; WD	277900	*ATP7B*	NM_000053.3
Cerebrotendinous xanthomatosis; CTX	213700	*CYP27A1*	NM_000784.3
Spastic paraplegia 35, autosomal recessive; SPG35	612319	*FA2H*	NM_024306.4
Neurodegeneration with brain iron accumulation 3; NBIA3	606159	*FTL*	NM_000146.3
Parkinson disease, late-onset; PD	168600	*GBA*	NM_000157.3
Dystonia, DOPA-responsive, with or without hyper-phenylalaninemia; DRD	128230	*GCH1*	NM_000161.2
Dystonia 25; DYT25	615073	*GNAL*	NM_182978.3
Parkinson disease 8, autosomal dominant; PARK8	607060	*LRRK2*	NM_198578.3
Dementia, frontotemporal, with or without parkinsonism; FTD	600274	*MAPT*	NM_001123066.3
Neurodegeneration with brain iron accumulation 1; NBIA1 (Pantothenate kinase-associated neurodegeneration; PKAN,	234200	*PANK2*	NM_153638.2
Parkinson disease, juvenile, type 2; PARK2	600116	*PARK2*	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
Paroxysmal nonkinesigenic dyskinesia 1; PNKD1	118800	*PNKD*	NM_015488.4
Spinocerebellar ataxia 14; SCA14	605361	*PRKCG*	NM_002739.3
Episodic kinesigenic dyskinesia 1; EKD1 (Dystonia 10; DYT10)	128200	*PRRT2*	NM_145239.2
Dystonia-11, myoclonic; DYT11	159900	*SGCE*	NM_001099401.1
Allan-Herndon-Dudley syndrome; AHDS	300523	*SLC16A2*	NM_006517.4
Dystonia 9; DYT9	601042	*SLC2A1*	NM_006516.2
Parkinson disease 1 autosomal dominant; PARK1	168601	*SNCA*	NM_000345.3
Spastic paraplegia 11, autosomal recessive; SPG11	604360	*SPG11*	NM_025137.3
Dystonia, dopa-responsive, due to sepiapterin reductase deficiency; SPRD	612716	*SPR*	NM_003124.4
Segawa syndrome, autosomal recessive	605407	*TH*	NM_199292.2
Dystonia 6, torsion; DYT6	602629	*THAP1*	NM_018105.2
Neurodegeneration with brain iron accumulation 5; NBIA5	300894	*WDR45*	NM_007075.3

Episodic Ataxia 7 gene panel			
Condition	**OMIM**	**Gene**	**Primary transcript**
Cerebellar ataxia, areflexia, pes cavus, optic atrophy, and sensorineural hearing loss; CAPOS	601338	*ATP1A3*	NM_152296.4
Episodic ataxia, type 2; EA2; Migraine, familial hemiplegic, 1, with or without progressive cerebellar ataxia; FHM1	108500	*CACNA1A*	NM_001127221.1
Episodic ataxia, type 5; EA5	613855	*CACNB4*	NM_000726.3
Episodic ataxia, type1 (including episodic ataxia/myokymia syndrome); EA1	160120	*KCNA1*	NM_000217.2
Episodic kinesigenic dyskinesia 1; EKD1 (Dystonia 10; DYT10)	128200	*PRRT2*	NM_145239.2
Episodic ataxia, type 6; EA6	612656	*SLC1A3*	NM_004172.4
Dystonia 9; DYT9	601042	*SLC2A1*	NM_006516.2

Hemiplegic Migraine 6 gene panel			
Condition	**OMIM**	**Gene**	**Primary transcript**
Migraine, familial hemiplegic, 2; FHM2	602481	*ATP1A2*	NM_000702.3
Episodic ataxia, type 2; EA2; Migraine, familial hemiplegic, 1, with or without progressive cerebellar ataxia; FHM1	108500	*CACNA1A*	NM_001127221.1
Episodic kinesigenic dyskinesia 1; EKD1 (Dystonia 10; DYT10)	128200	*PRRT2*	NM_145239.2
Migraine, familial hemiplegic, 3; FHM3	609634	*SCN1A*	NM_001165963.1
Episodic ataxia, type 6; EA6	612656	*SLC1A3*	NM_004172.4
Dystonia 9; DYT9	601042	*SLC2A1*	NM_006516.2

Hereditary Ataxia Stages 1 and 2

(SureSelect Design ID: 3097261)

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
Polyneuropathy, 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

Hereditary Ataxia (Stage 1) 107 gene panel				
Condition	**OMIM**	**Inheritance**	**Gene**	**Primary Transcript**
Pinocerebellar ataxia, autosomal recessive 17; SCAR17	616127	AR	*CWF19L1*	NM_018294.5
Spastic paraplegia 56, autosomal recessive; SPG56	615030	AR	*CYP2U1*	NM_183075.2
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

Hereditary Ataxia (Stage 1) 107 gene panel				
Condition	**OMIM**	**Inheritance**	**Gene**	**Primary Transcript**
Tay-Sachs disease, GM2-gangliosidosis, several forms; TSD	272800	AR	*HEXA*	NM_000520.5
Sandhoff disease;	268800	AR	*HEXB*	NM_000521.3
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

Hereditary Ataxia (Stage 1) 107 gene panel				
Condition	**OMIM**	**Inheritance**	**Gene**	**Primary Transcript**
Spinocerebellar ataxia 14; SCA14	176980	AD	*PRKCG*	NM_002739.3
Huntington disease-like 1; HDL1	603218	AD	*PRNP*	NM_000311.4
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	*SYNE1*	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
Cerebellar ataxia;		AR	CLN6	NM_017882.2
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
Neurodegeneration with brain iron accumulation 3; NBIA3;	134790	AD	FTL	NM_000146.3

Hereditary Ataxia (Stage 2) 54 gene panel				
Condition	**OMIM**	**Inheritance**	**Gene**	**Primary Transcript**
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
Occult macular dystrophy; OCMD	613587	AR	*RP1L1*	NM_178857.5
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
Epileptic encephalopathy, early infantile, 4; EIEE4	612164	AD	*STXBP1*	NM_003165.3

Hereditary Ataxia (Stage 2) 54 gene panel				
Condition	**OMIM**	**Inheritance**	**Gene**	**Primary Transcript**
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

Bleeding & Thrombotic disorders

(SureSelect Design ID: 0657461)

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