# Scottish Genetics Consortium Clinical Exome Panels

*All referrals must have a Clinical Exome proforma completed or testing will not be performed*

## CARDIOLOGY - ABERDEEN

**A clinical exome proforma is not required for Sanger sequencing referrals**

| Sanger sequencing | Genes |
|---|---|
| Barth syndrome | *TAZ* |

## CHROMOSOME BREAKAGE - ABERDEEN

| Subpanel | Genes |
|---|---|
| Ataxia Telangiectasia (& AT-like) | *ATM, MRE11A* |
| Autosomal Recessive Primary Microcephaly | *MCPH1, CDK5RAP2, ASPM, STIL, WDR62, KNL1 (CASC5), CENPJ, CEP135* |
| Bloom Syndrome | *BLM* |
| Cerebro Oculo Facio Skeletal syndrome | *ERC1, ERCC2, ERCC6* |
| Cockayne syndrome | *ERCC6, ERCC8* |
| Fanconi Anaemia | *BRCA2, BRIP, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, PALB2, RAD51C, SLX4* |
| Immunodeficiency-centromeric instability-facial anomalies syndrome | *DNMT3B* |
| Meier-Gorlin Syndrome | *ORC1, ORC4, ORC6, CDT1, CDC6* |
| Natural killer cell and glucocorticoid deficiency with DNA repair defect | *MCM4* |
| Nijmegan breakage syndrome (& NBS-like) | *NBN, RAD50* |
| Rothmund Thomson / RAPADILINO / Baller-Gerold | *RECQL4* |
| Seckel Syndrome | *ATR, RBBP8, CEP152, CENPJ* |
| Tricothiodystrophy | *ERCC2, ERCC3, MPLKIP, GTF2H5* |
| Warsaw breakage syndrome | *DDX11* |
| Werner syndrome | *WRN* |
| Xeroderma Pigmentosum | *XPA, XPC, ERCC1, ERCC3, ERCC4, ERCC5, DDB2, POLH* |

**A clinical exome proforma is not required for Sanger sequencing referrals**

| Sanger sequencing | Genes |
|---|---|
| Ataxia with oculomotor apraxia and hypoalbuminemia | *APTX* |
| Roberts / SC phocomelia syndrome | *ESCO2* |
| Duane-Radial Ray & IVIC Syndrome | *SALL4* |
| Townes-Brocks Syndrome | *SALL1* |
| Holt-Oram Syndrome | *TBX5* |
| Ulnar-mammary syndrome | *TBX3* |
| TAR Syndrome | *RBM8A* |
| LIG4 Syndrome | *LIG4* |

# CONNECTIVE TISSUE DISORDERS – EDINBURGH GENETICS

| Subpanel | Genes |
|---|---|
| **Connective tissue**<br>(please contact to discuss indications not included below) | *ABCC6, ACTA2, ACVR1, ADAMTS2, ALPL, ATP6V0A2, B3GALT6, B4GALT7, BMP1, CBS, CHST14, COL11A1, COL1A1, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, CRTAP, ELN, FBLN5, FBN1, FBN2, FKBP10, FKBP14, IFITM5, LEPRE1 (P3H1), LRP5, MYLK, NOTCH1, NOTCH2, PKD2, PLOD1, PLOD2, PPIB, PRDM5, RIN2, SERPINF1, SERPINH1, SLC2A10, SLC39A13, SMAD3, SP7, TGFB2, TGFBR1, TGFBR2, TNXB, ZNF469* |
| **Ehlers-Danlos Syndrome**<br>(Referral criteria as per Malfait et al (2017) Am J Med Genetics 175C:8-26) Samples for Hypermobile EDS will not be accepted as the genetic basis is unknown) | *ADAMTS2, B3GALT6, B4GALT7, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, CHST14, PLOD1, FKBP14, RIN2, PRDM5, ZNF469, SLC39A13, TNXB, C1R, C1S* |
| **Stickler Syndrome / Cleft Palate**<br>Two or more of the following:<br>• Retinal detachment or: High myopia with onset before 6 years<br>• Cleft palate<br>• Vitreous abnormality<br>• Joint hypermobility or premature joint degeneration<br>• Sensorineural hearing loss<br>• Facial features (flat midface with depressed nasal bridge, reduced nasal protrusion, anteverted nares and micrognathia) | *ANKRD11, COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, FLNA, FLNB, FOXE1, FOXC2, IRF6, IRF7, PLOD3, SATB2, SLC26A2, SOX9, TP63, TBX1, TBX22* |

## DEVELOPMENTAL DISORDERS – EDINBURGH GENETICS

| Subpanel | Genes |
|---|---|
| Enquire to lab regarding coverage / panel (Please provide parental samples if possible) | DDG2P (limited gene panel) |

## ENDOCRINOLOGY - DUNDEE

| Subpanel | Genes |
|---|---|
| Congenital Hypothyroidism<br><br>Stage 2 criteria TBC (discussion with Scottish Paediatric Endocrine Group Sept. 2020) | Stage 1: *DUOX2, FOXE1, NKX2-1, PAX8, TG, THRA, TPO, TSHR* |
| | Stage 2: *DUOXA2, GLIS3, GNAS, HESX1, IGSF1, IRS4, IYD, LHX3, LHX4, OTX2, POU1F1, PRKAR1A, PROP1, SECISBP2, SLC16A2, SLC26A4, SLC5A5, TBL1X, THRB, TRHR, TSHB* |
| Nephrogenic Diabetes Insipidus<br>*Testing now offered by Sanger sequencing* | *AQP2, AVPR2* |
| Hyperinsulinism/Hypersinsulinemic Hypoglycemias | *ABCC8, AKT2, CACNA1D, GCK, GLUD1, GPC3, HADH, HNF1A, HNF4A, INSR, KCNJ11, KDM6A, KMT2D, PMM2, SLC16A1, TRMT10A* |
| Hyperthyroidism | *ALB, SECISBP2, SLC16A2, THRA, THRB, TSHR, TTR* |
| Hypophosphatemic Rickets | *DMP1, ENPP1, FGF23, PHEX, SLC34A3* |
| Pigmented Nodular Adrenocortical Disease | *PDE11A, PDE8B, PRKAR1A* |

# EYES - ABERDEEN

| Subpanel | Genes |
|---|---|
| Anterior Segment Dysgenesis (ASD) | *ADAMTS18, ALDH18A1, ATOH7, B3GLCT, BEST1, BMP7, CHRDL1, CHST6, COL4A1, COL8A2, CRYGC, CYP1B1, DCN, EYA1, FBN1, FOXC1, FOXE3, FOXL2, GJA1, GNPTG, GSN, KERA, KRT12, KRT3, LAMB2, LCAT, LMX1B, LTBP2, MYOC, NOTCH2, OPTN, PAX3, PEX2, PIKFYVE, PITX2, PITX3, PRDM5, PXDN, RAB18, RAB3GAP1, RAB3GAP2, SEC23A, SH3PXD2B, SIX3, SLC16A12, SLC4A11, SLC4A4, TACSTD2, TGFBI, UBIAD1, VSX1, WDR36, ZEB1* |
| Bardet-Biedl Syndrome | *ARL6, BBS1, BBS2, BBS4, BBS5, BBS7, BBS9, BBS10, BBS12, LZTFL1, MKKS, MKS1, SDCCAG8, TTC8, WDPCP* |
| Corneal abnormalities (including corneal dystrophy and BCS) | *ADAMTS18, ALDH18A1, B3GLCT, CHRDL1, CHST6, COL17A1, COL8A2, DCN, GJA1, GRHL2, GSN, HMX1, KERA, KRT12, KRT3, LCAT, MIR184, OVOL2, PIK3R1, PIKFYVE, PRDM5, PXDN, RAB18, RAB3GAP1, RAB3GAP2, SLC16A12, SLC4A11, STS, TACSTD2, TCF4, TGFBI, UBIAD1, ZEB1, ZNF469* |
| Corneal dystrophy | *CHST6, COL17A1, COL8A2, DCN, GRHL2, GSN, KERA, KRT12, KRT3, LCAT, MIR184, OVOL2, PIKFYVE, PRDM5, SLC4A11, STS, TACSTD2, TCF4, TGFBI, UBIAD1, ZEB1 , ZNF469* |
| Brittle cornea syndrome | *PRDM5, ZNF469* |
| Eye Movement Disorder | *CHN1, COL25A1, DCC, FRMD7, HOXA1, HOXB1, KIF21A, MAFB, PHOX2A, ROBO3, SALL1, SALL4, TUBB2B, TUBB3* |
| Glaucoma | *ADAMTS10, ADAMTS17, CYP1B1, FOXC1, FOXD3, FOXE3, JAG1, LTBP2, MYOC, PITX2* |
| Optic Nerve disorders (Including Ocular/Oculocutaneous albinism, foveal hypolasia & nystagmus, and optic atrophy) | *ACO2, ATOH7, C12orf65, CACNA1F, CISD2, FRMD7, GJA1, GPR143, HMX1, HPS3, HPS4, HPS5 , HPS6, LRMDA (C10orf11), LYST, OCA2, OPA1, OPA3, PAX6, RAB18, RAB3GAP1, RAB3GAP2, SLC24A5, SLC45A2, SLC4A11, SOX2, TMEM126A, TYR, TYRP1, WFS1* |
| Oculocutaneous albinism | *GPR143, HPS3, HPS4, HPS5, LRMDA, LYST, OCA2, SLC24A5, SLC45A2, TYR, TYRP1* |
| Familial exudative vitroretinopathy (FEVR) | *ATOH7, FZD4, LRP5, NDP, TSPAN12* |
| Usher syndrome | *MYO7A, USH1C, CDH23, PCDH15, USH1G, ADGRV1, DFNB31 (WHRN), USH2A* |

**A clinical exome proforma is not required for Sanger sequencing referrals**

| Sanger sequencing | Genes |
|---|---|
| Best disease, vitelliform macular dystrophy (VMD), AR bestrophinopathy (ARB) | *BEST1* |
| Choroideraemia | *CHM* including MLPA |
| Macular Dystrophy | *PRPH2* |
| X-linked Congenital Nystagmus | *FRMD7* |
| X-linked Juvenile Retinoschisis | *RS1* – can be performed prior to Retinal Degeneration panel if required |

# GASTROHEPATOLOGY - ABERDEEN

| Subpanel | Genes |
|---|---|
| Cholestasis | *AKR1D1, ALDOB, ABCB11, ABCB4, ATP8B1, BAAT, CLDN1, CYP7A1, HSD3B7, JAG1, NOTCH2, NPC1, NPC2, NR1H4, PEX1, PEX2, SERPINA1, SLC25A13, TJP2, VIPAS39, VPS33B* |
| Porphyrias | *ALAD, ALAS2, CPOX, FECH, HMBS, PPOX, UROD, UROS* |
| Wilsons | *ATP7B* |

**A clinical exome proforma is not required for Sanger sequencing referrals**

| Sanger sequencing | Genes |
|---|---|
| Pancreatitis | *SPINK1, PRSS1* |

# HAEMATOLOGY

## HAEMATOLOGY – ABERDEEN

| Subpanel | Genes |
|---|---|
| Bone Marrow Failure | *BRCA2, BRIP1, CTC1, DKC1, ELANE, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, G6PC3, GATA1, GATA2, GFI1, HAX1, MPL, NHP2, NOP10, PALB2, RAD51C, RPL11, RPL35A, RPL5, RPS10, RPS17, RPS19, RPS24, RPS26, RPS7, RUNX1, SBDS, SLX4, SRP72, TERT, TINF2, WAS, WRAP53* |
| Diamond Blackfan Anaemia | *RPL5, RPS10, RPL11, RPL35A, RPS7, RPS19, RPS24, RPS26, GATA1, RPS17* |
| Shwachman-Diamond Syndrome | *SBDS* |
| Dyskeratosis Congenita | *DKC1, CTC1, NHP2, NOP10, TERT, TINF2, WRAP53* |
| Myelodysplastic Syndrome | *SRP72, GATA2* |
| Neutropenia | *ELANE, GFI1, HAX1, G6PC3, WAS* |

## HAEMATOLOGY – EDINBURGH HAEMATOLOGY (RIE)

**NB: Samples should be sent to Western General Genetics laboratory, Edinburgh**

| Subpanel | Genes |
|---|---|
| **Erythrocytosis**<br><br>**Testing criteria**<br>Idiopathic erythrocytosis with:<br>• No acquired *JAK2* variants<br>• Secondary causes excluded<br>• Young onset and/or family history | *BPGM, EGLN1, EPAS1, EPO, EPOR, HBA1, HBA2, HBB, JAK2, SH2B3, VHL* |
| **Iron Regulation**<br>Criteria:<br>• Juvenile Haemochromatosis (<30years) with severe iron overload in liver AND/OR heart. Raised serum ferritin >1000ug/L and transferrin saturation >90%<br>• Juvenile Haemochromatosis >30 years with unexplained severe haemochromatosis and HFE negative<br>• Ferroportin disease: raised serum ferritin with normal transferrin saturation and evidence of reticuloendothelial iron staining on liver biopsy or splenic iron overload on MRI and HFE mutations negative<br>• Haemochromatosis: raised serum ferritin and transferrin saturation C282Y negative<br>• Hereditary Hyperferritinemia cataract syndrome: High and constant levels of serum ferritin unresponsive to iron depletion and no signs of iron overload and no relevant clinical symptoms apart from visual impairment by cataract<br>• Biochemical evidence of unexplained iron overload and lack of homozygous/compound homozygous HFE mutations<br>• Iron Refractory Iron Deficiency Anaemia (IRIDA): Very low mean corpuscular volume (MCV) and low serum iron and low transferrin saturation, normal ferritin or ferritin levels in the lower limits of normal, no response to oral iron treatment | *ABCB7, ALAS2, ATP7B, CP, CYBRD1, FTL, GBA, GLRX5, HAMP, HFE, HFE2, SLC11A2, SLC25A38, SLC40A1, TF, TFR2, TMPRSS6* |
| **Red Cell Membranopathy**<br><br>**Suitability depends on phenotype** - contact RIE Haem if required | *ADD1, AK1, ANK1, APOB, EPB41, EPB42, MTTP, RhAG, SLC2A1, SLC4A1, SPTA1, SPTB, TPM3, XK* |

## HEARING LOSS - DUNDEE

| Subpanel | Genes |
|---|---|
| Hearing Loss, Syndromic and Non Syndromic | *ACTG1, ADGRV1 (GPR98), ATP2B2, BDP1, CABP2, CCDC50, CDH23, CEACAM16, CIB2, CLDN14, CLIC5, CLPP, CLRN1, COCH, COL11A2, COL4A6, CRYM, DIABLO, DIAPH1, DIAPH3, EDN3, EDNRB, EPS8, ESPN, ESRRB, EYA1, EYA4, GATA3, GIPC3, GJB2, GJB3, GJB6, GPSM2, GRHL2, GRXCR1, GSDME (DFNA5), HARS, HARS2, HGF, HSD17B4, ILDR1, KARS, KCNE1, KCNJ10, KCNQ1, KCNQ4, KIT, LARS2, LHFPL5, LOXHD1, LRTOMT, MARVELD2, MITF, MSRB3, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, NARS2, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PAX3, PCDH15, PDZD7, PJVK (DFNB59), PNPT1, POU3F4, POU4F3, PRPS1, PTPRQ, RDX, RPGR, SERPINB6, SIX1, SIX5, SLC17A8, SLC26A4, SLC26A5, SLC4A11, SMPX, SNAI2, SOX10, STRC, SYNE4, TBC1D24, TECTA, TJP2, TMC1, TMIE, TMPRSS3, TNC, TPRN, TRIOBP, TSPEAR, USH1C, USH1G, USH2A, WFS1, WHRN (DFNB31)* |
| Waardenburg Syndrome type 1, 2, 3 and 4 | *PAX3, MITF, SOX10, SNAI2, EDNRB, EDN3* |

## INHERITED CANCER - GLASGOW

| Subpanel | Genes |
|---|---|
| **Gorlin syndrome (Basal Cell Nevus syndrome)**<br>Referral/approval by Clinical Genetics | *PTCH1, SUFU* |
| **DICER1 syndrome**<br>Referral/approval by Clinical Genetics | *DICER1* |

# METABOLIC - ABERDEEN

| Subpanel | Genes |
|---|---|
| Amino Acid Disorders and Disorders of Neurotransmission | *ABAT, ALDH18A1, ALDH5A1, ALDH7A1, AMT, ASPA, CBS, CTH, D2HGDH, DBH, DDC, FAH, GABRG2, GCDH, GCH1, GLDC, GLRA1, HGD, L2HGDH, MAT1A, OAT, PAH, PCBD1, PNPO, QDPR, SLC25A22, SLC6A19, SLC7A7, SUOX* |
| Disorders of Carbohydrate Metabolism (including Glycogen Storage Disorders) | *AGL, ALDOA, ENO3, EPM2A, FBP1, G6PC, G6PC3, GAA, GALE, GALK1, GALT, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, NHLRC1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PYGL, PYGM, SLC2A2, SLC37A4* |
| Disorders associated with Hyperammonaemia/Fatty Acid Oxidation/Ketogenesis/Ketolysis | *ACADM, ACADS, ACADVL, ARG1, ASL, ASS1, CPS1, CPT1A, CPT2, ETFA, ETFB, ETFDH, GLUD1, HADHA, HADHB, HMGCL, HMGCS2, IVD, MMAA, MMAB, MMACHC, MMADHC, MUT, NAGS, OAT, OTC, OXCT1, PCCA, PCCB, SLCC2A5, SLC25A13, SLC25A15, SLC25A20, SLC7A7, SLC52A3* |
| Fatty Acid Oxidation | *ACADM, ACADS, ACADVL, CPT1A, CPT2, ETFA, ETFB, ETFDH, HADHA, HADHB, HMGCL, HMGCS2, IVD, MMAA, MMAB, MMACHC, MMADHC, OXCT1, SLC22A5, SLC25A20* |
| Glycogen storage disease type 2 (Pompe disease) | *GAA* |
| Homocysteinuria | *CBS, MMADHC, MTHR, MTR, MTRR* |
| Hypobetalipoproteinaemia | *APOB* |
| Mucolipidosis II and III Alpha/Beta | *GNPTAB* |
| Multiple Acyl-CoA Dehydrogenase (MAAD) | *ETFDH, ETFA, ETFB, SLC52A2, SLC52A3* |
| Neuronal ceroid lipofuscinosis (NCL) | *CLN3, CLN5, CLN6, CLN8, CTSD, DNAJC5, KCTD7, MFSD8, PPT1, TPP1* |
| Niemann Pick Disease | *NPC1, NPC2, SMPD1* |
| Niemann Pick Disease Types C1 and C2 | *NPC1, NPC2* |
| Non Ketotic Hyperglycinaemia | *ALH7A1, AMT, GLDC, PPT1, TPP1* |
| Organic Acidaemias and Cofactor/Vitamin Disorders | *ABCD4, ACSF3, AMN, AUH, BCKDHA, BCKDHB, BTD, CUBN, DBT, DHFR, DNAJC19, FOLR1, GIF, HLCS, IVD, LMBRD1, MCCC1, MCCC2, MCEE, MLYCD MMAA, MMAB, MMACHC, MMADHC, MTHFR, MTR, MTRR, MUT, OPA3, PCCA, PCCB, SLC19A3, SLC46A1, SLC52A3, SUCLA2, SUCLG1, TAZ, TMEM70* |
| Peroxisomal disorders | *ABCD1, ACOX1, AGPS, AGXT, AMACR, CAT, DNM1L, GNPAT, HSD17B4, PEX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26, PHYH, SCP2* |
| Proprionic Anaemia | *PCCA, PCCB* |
| Refsum disease | *PEX7, PHYH* |
| Vitamin B deficiency | *AMN, GIF, CUBN* |
| VLCAD deficiency | *ACADVL* |

**A clinical exome proforma is not required for Sanger sequencing referrals**

| Sanger sequencing | Genes |
|---|---|
| Batten's disease (NCL) | *TPP1* |
| Carnitine Palmitoyltransferase II Deficiency | *CPT2* |
| Cerebral folate transport deficiency | *FOLR1* |
| Citrullinaemia type 1 | *ASS1* |
| Fanconi Bickel Syndrome | *SLC2A2* |
| Galactosaemia | *GALT* |
| Gaucher disease (B-glucocerebrosidase deficiency) | *GBA* |
| Glutaric Acidaemia Type 1 | *GCDH* |
| Glycogen storage disease 1A | *G6PC* |
| Metachromatic leukodystrophy | *ARSA* |
| Muccopolysaccharidosis type 1 (Hurler/Scheie syndrome) | *IDUA* |
| Niemann Pick Disease Types A and B | *SMPD1* |
| Ornithine Aminotransferase Deficiency | *OAT* |
| Succinic semialdehyde dehydrogenase deficiency (SSADH) | *ALDH5A1* |
| Tay-Sachs disease | *HEXA* |

# MUSCULOSKELETAL

## MUSCULOSKELETAL - EDINBURGH GENETICS

| Subpanel | Genes |
|---|---|
| **Osteogenesis imperfecta**<br>Multiple fractures of long bones without significant trauma **AND** at least two of the following:<br>• Wormian bones<br>• Blue/grey sclera<br>• Hearing loss<br>• Ribs, broad and beaded, thin and irregular<br>• Short stature<br>• Dentinogenesis imperfecta<br>• Triangular face and narrow thorax<br>• Round facies and short barrel-shaped chest | *BMP1, COL1A1, COL1A2, CRTAP, FKBP10, IFITM5, LEPRE1 (P3H1), PLOD2, PPIB, SERPINF1, SERPINH1, SP7* |
| **Skeletal dysplasia (113 gene panel)**<br>• Antenatal evidence (Ultrasound or other imaging modality) or Postnatal evidence of skeletal dysplasia (X ray and clinical examination)<br>• Multiple joint involvements (e.g. ephyseal or metaphyseal abnormalities)<br>• Short limbs (Long bone length-3SD below mean or serial measurement at or below 5th centile)<br>• Narrow thorax<br>• Poly and/or Oligodactyly<br>• Syndactyly<br>• Limb reduction defects<br>• Fractures of long bones<br>• Poor mineralisation of calvarium or spine | *ACAN, ACP5, ADAMTS10, ADAMTSL2, AGPS, ALPL, ANKH, ARSE, B3GALT6, BMP1, BMPR1B, CA2, CANT1, CDC6, CDKN1C, CDT1, CHST3, CLCN7, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, COL9A1, COL9A2, COL9A3, COMP, CRTAP, CTSK, CUL7, CYP27B1, DHCR24, DLL3, DYM, DYNC2H1, EBP, EIF2AK3, ENPP1, ESCO2, EVC, EVC2, FAM20C, FGF23, FGFR1, FGFR2, FGFR3, FKBP10, FLNA, FLNB, GDF5, GNPAT, GPC6, HSPG2, IFT122, IFT140, IFT43, IFT80, IHH, KAT6B, LBR, LEPRE1, LIFR, LMX1B, LRP5, LTBP2, MATN3, MMP9, NEK1, NPR2, OBSL1, ORC1, ORC4, ORC6, OSTM1, PAPSS2, PCNT, PEX7, PHEX, PLOD2, PPIB, PTH1R, RMRP, RNU4ATAC, ROR2, RUNX2, SBDS, SERPINF1, SERPINH1, SHOX, SLC26A2, SLC34A3, SLC35D1, SLC39A13, SMAD4, SMARCAL1, SNX10, SOX9, TCIRG1, TGFB1, TNFRSF11A, TNFRSF11B, TNFSF11, TRAPPC2, TRIP11, TRPV4, TTC21B, VDR, WDR19, WDR35, WISP3, WNT5A, XYLT1* |

MUSCULOSKELETAL - GLASGOW

| Subpanel | Genes |
|---|---|
| **Chondrodysplasia Punctata**<br><br>Referral/approval by Clinical Genetics<br><br>**Criteria:** Stippling involving the epiphyses of the long bones and vertebrae, the trachea and distal ends of the ribs seen on x-ray OR rhizomelia with stippling involving the epiphyses knee, hip, elbow, and shoulder OR biochemical evidence of Chondrodysplasia punctata. | *AGPS, ARSE, EBP, GNPAT, PEX7* |
| **Ellis-Van Creveld syndrome**<br><br>Referral/approval by Clinical Genetics | *EVC, EVC2* |
| **Osteopetrosis**<br><br>Referrals from Clinical Genetics, Consultant Paediatricians (specialising in bone marrow transplantation, haematology, metabolic disease or orthopaedics) and Adult Orthopaedic Consultants | *AMER1, ANKH, CA2, CLCN7, CTSK, FAM20C, FERMT3, LEMD3, LRP5, OSTM1, PTH1R, RASGRP2, SNX10, SOST, TCIRG1, TGFB1, TNFRSF11A, TNFSF11, TYROBP* |
| **Hereditary Multiple Osteochondromas/Multiple Exostoses**<br><br>Referrals from Clinical Genetics, Consultant Paediatricians, Orthopaedic Consultant/Surgeon | *EXT1, EXT2* |

# NEUROLOGY

## NEUROLOGY - DUNDEE

| Subpanel | Genes |
|---|---|
| Episodic movement, migraine and epileptic disorders (Brain Channelopathies) | *ATP1A2, ATP1A3, CACNB4, CACNA1A, PNKD, KCNK18, KCNA1, PRRT2, SCN1A, SLC1A3, SLC2A1* |
| Leukoencephalopathy with vanishing white matter | *EIF2B1, EIF2B2, EIF2B4, EIF2B5, EIF2B3* |
| Aicardi-Goutieres Syndrome | *ADAR, IFIH1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, TREX1* |
| Porencephaly | *COL4A1, COL4A2* |
| Cortical Brain Malformations | *ACTB, ACTG1, ADGRG1 (GPR56), AKT3, ARFGEF2, ARX, ASPM, CASK, CCND2, COL4A1, COL4A2, DAG1, DCX, DYNC1H1, FKRP, FKTN, FLNA, GPSM2, GRIN1, ISPD, KIF1BP (KIAA1279), LAMA2, LAMB1, LAMC3, LARGE1 (LARGE), NDE1, NEDD4L, OCLN, OPHN1, PAFAH1B1, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PIK3CA, PIK3R2, POMGNT1 (GTDC2), POMGNT2, POMT1, POMT2, RELN, SCN3A, SRPX2, TMEM5, TUBA1A, TUBA8, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, VLDLR, WDR62* |
| Dementia | *APP, CHMP2B, CHCHD10, CSF1R, CYP27A1, DNMT1, DCTN1, FUS, GRN, HNRNPA2B1, HTRA1, ITM2B, MATR3, MAPT, NOTCH3, PSEN1, PSEN2, PRNP, SQSTM1, SPG21, TBK1, TARDBP, TREM2, TUBA4A, TYROBP, UBQLN2, VCP* |
| Holoprosencephaly Disorders | *CDON, DHCR7, DISP1, FGF8, FGFR1, GLI2, PTCH1, SHH, SIX3, TGIF1, ZIC2* |
| Pain disorders (congenitcal erythromelalgia/ small fibre neuropathy, Indifference to Pain, Congenital, Autosomal Recessive, Neuropathy/Paroxysmal Extreme Pain Disorder (congenital insensitivity to pain) | *ATL1, ATL3, ELP1, GLA, KIF1A, NGF, NTRK1, PRNP, RAB7A, RETREG1, SCN10A, SCN11A, SCN9A, SEPT9, SPTLC1, SPTLC2, TRPA1, TTR, WNK1* |
| Dystonia (Please note testing of genes associated with Parkinson's disease only, including copy number analysis, is available via our targeted panel) | *AFG3L2, ANO3, ATP7B, CYP27A1, FA2H, GNAL, PANK2, PNKD, PRRT2, PRKCG, SGCE, SPR, SLC16A2, SLC2A1, THAP1, WDR45* |
| Dystonia & Parkinsonism (Please note testing of genes associated with Parkinson's disease only, including copy number analysis, is available via our targeted panel) | *AFG3L2, ATP7B, ATP1A3, CYP27A1, FA2H, FTL, GBA, GCH1, LRRK2, MAPT, PANK2, PARK2, PARK7, PNKD, PRRT2, PRKCG, PINK1, SGCE, SPR, SLC16A2, SLC2A1, SPG11, SNCA, THAP1, TH, WDR45* |

NEUROLOGY - EDINBURGH GENETICS

| Subpanel | Genes |
|---|---|
| **Hereditary Ataxia (35 gene panel) – East of Scotland patients**<br>• Exclusion of metabolic, neoplastic, alcohol, and drugrelated<br>• causes<br>• Normal/routine neurological bloods, and vitamin E<br>• Testing<br>• Negative spinocerebellar ataxia repeat expansion<br>• panel, including FXTAS and FA testing<br>• MRI neuroimaging normal, or isolated cerebellar<br>• atrophy<br>• Family history of ataxia, or young age of onset (<50) | *ADCK3, AFG3L2, APTX, ATM, ABCB7, ATP7B, ATP1A3, CACNB4, CACNA1A, CYP27A1, FTL, FGF14, FXN, GBA, ITPR1, IFRD1, MTPAP, KCNA1, KCNC3, KCND3, PDYN, PRRT2, PRKCG, SACS, SETX, SIL1, SLC1A3, SLC16A2, SLC2A1, SPTBN2, SPG7, SYNE1, TGM6, TTBK2, TTPA* |
| **Hereditary Spastic Paraplegia (20 gene panel) – East of Scotland patients**<br>See referral criteria at:<br>(www.tinyurl.com/Edinburghgenelab) | *ATL1, BSCL2, CYP7B1, FA2H, GJC2, HSPD1, KIAA0196, KIF5A, L1CAM, NIPA1, PLP1, REEP1, SLC33A1, SPG11, SPG20, SPG21, SPAST, SPG7, ZEB2, ZFYVE27* |

NEUROLOGY - GLASGOW

| Subpanel | Genes |
|---|---|
| **Hereditary Ataxia**<br>Referral by Consultant Neurologist or Clinical Genetics<br><br>**Criteria:** West of Scotland patients only (Edinburgh testing East of Scotland patients). | *AAAS, ABCB7, ABHD12, AFG3L2, AMPD2, ANO10, AP1S2, APTX, ARSA, ATCAY, ATM, ATP1A3, CA8, CACNA1A, CACNA1G, CAMTA1, CASK, CHMP1A, CLN6, COQ8A, COX20, CP, CWF19L1, CYP27A1, CYP2U1, DARS2, DDHD2, DNAJC5, DNMT1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL4, EPM2A, EXOSC3, FGF14, FLVCR1, FOLR1, FXN, GBA2, GJC2, GOSR2, GRID2, GRM1, HEXA, HEXB, ITPR1, KCNA1, KCNC3, KCND3, KCNJ10, KIF1C, MARS2, MMACHC, MRE11A, MTTP, NHLRC1, NPC1, NPC2, OPHN1, PAX6, PDYN, PEX16, PLA2G6, PMPCA, PNKP, PNPLA6, POLG, POLR3A, PRKCG, PRNP, PRRT2, RARS2, RNF170, RNF216, SACS, SAR1B, SEPSECS, SETX, SIL1, SLC1A3, SLC2A1, SLC9A6, SNX14, SPG7, SPTBN2, SRD5A3, STUB1, SYNE1, TGM6, TMEM240, TPP1, TSEN2, TSEN54, TTBK2, TTC19, TTPA, TUBB4A, TWNK, VLDLR, VRK1, WDR73, WDR81, WFS1, WWOX* |

| Subpanel | Genes |
|---|---|
| **Hereditary Spastic Paraplegia**<br>Referral by Consultant Neurologist or Clinical Genetics<br><br>**Criteria:** West of Scotland patients only (Edinburgh testing East of Scotland patients). | **Stage 1:** *ATL1, BSCL2, CYP7B1, FA2H, GJC2, HSPD1, KIAA0196 (WASHC5), KIF5A, L1CAM, NIPA1, PLP1, REEP1, SLC33A1, SPG11, SPG20 (SPART), SPG21, SPAST, SPG7, ZEB2, ZFYVE27 (ALT1 & SPAST by MLPA also)*<br><br>**Stage 2:** *ABCD1, ADAR, AFG3L2, AIMP1, ALDH18A1, ALS2, AP4B1, AP4E1, AP4M1, AP4S1, ARG1, ATP13A2, B4GALNT1, C12orf65, C19orf12, CAPN1, CYP27A1, CYP2U1, DDHD1, DDHD2, ERLIN1, ERLIN2, FARS2, GBA2, HACE1, KIDINS220, KIF1A, NT5C2, OPA3, PNPLA6, POLR3A, RTN2, SACS, SERAC1, SLC16A2, SLC1A4, SLC25A46, SLC2A1, TUBB4A, WDR45B, ZFYVE26*<br>(performed if stage 1 is negative) |
| **Neurodegeneration with Brain Iron Accumulation (NBIA)**<br><br>Referral by Consultant Neurologist or Clinical Genetics<br><br>**Criteria:** Suspected clinical diagnosis in patients with hallmark findings of NBIA, or further assessment of patients with clinical diagnosis of idiopathic NBIA who have had mutations ruled out in other genes. | *ATP13A2, C19ORF12, COASY, CP, DCAF17, FA2H, FTL, FUCA1, KIF1A, KMT2B, MECR, PANK2, PLA2G6, PSEN1, SCP2, SLC39A14, SQSTM1, TRIM32, UBTF, VPS13A, WDR45* |
| **Familial Hemiplegic Migraine**<br><br>Referral from Clinical Genetics or Consultant Neurologists<br><br>*Please note this is a subpanel from the established epilepsy panel. The latest Epilepsy proforma should be completed for these cases (see lab website), and not the clinical exome proforma.* | *ATP1A2, CACNA1A, PRRT2, SCN1A, SLC2A1* |
| **Charcot Marie Tooth / HMSN**<br><br>Referral by Clinical Genetics or Consultant Neurologist and approved Clinical Genetics<br><br>**Criteria:** Tier 1 testing (performed in Aberdeen) must be performed first. Specific proforma in development. | *AARS, ATL1, ATP7A, BICD2, BSCL2, CCT5, DCTN1, DNM2, DNMT1, DYNC1H1, EGR2, FAM134B, FGD4, FIG4, GARS, GDAP1, HINT1, HSPB1, HSPB3, HSPB8, IGHMBP2, IKBKAP, INF2, KIF1A, LITAF, LMNA, LRSAM1, MARS, MTMR2, NDRG1, NEFL, NGF, NTRK1, PLEKHG5, PRPS1, PRX, RAB7A, REEP1, SBF2, SCN9A, SETX, SH3TC2, SLC52A1, SLC52A2, SLC52A3, SORD, SPTLC1, SPTLC2, TRPV4, VCP, WNK1, YARS* |

| Subpanel | Genes |
|---|---|
| **Spinal Muscular Atrophy**<br><br>Referral by Consultant Neurologist or Clinical Genetics<br><br>**Criteria:**<br><br>1. dHMN/SMA clinical phenotype<br>AND<br>2. Compatible neurophysiology (not required in infants)<br>AND<br>3. 5q linked SMA excluded (not required in infants)<br><br>Please note SMN1 sequencing is not available. | *AARS, ASAH1, ATP7A, BICD2, BSCL2, CHCHD10, DCTN1, DNAJB2, DYNC1H1, EXOSC3, EXOSC8, FBXO38, FIG4, GARS, HEXA, HSPB1, HSPB3, HSPB8, IGHMBP2, LAS1L, MATR3, MFN2, PLEKHG5, REEP1, SCO2, SETX, SIGMAR1, SLC52A2, SLC52A3, SLC5A7, SOD1, SYT2, TRPV4, UBA1, VAPB, VCP, VRK1* |
| **Rhabdomyolysis & Metabolic Myopathies**<br><br>Referral by Consultant in Neurology, Clinical Genetics or Metabolic Medicine<br><br>*This panel is intended for patients with isolated skeletal muscle symptoms. Patients with multisystem disease may be more appropriately tested on alternative panels*<br><br>**Criteria:**<br><br>**Single episode rhabdomyolysis**<br><br>ALL MUST FULFIL 2 essential criteria<br><br>1. CK documented >10,000IU/L associated with muscle pain<br>2. Mitochondrial myopathy/PEO considered and excluded where appropriate<br><br>IN ADDITION PATIENTS AGED >10years must fulfil at least one of the following three criteria<br><br>1. No environmental cause AND *Accustomed* exercise (NOT too much, too fast, too soon)<br>2. High risk features- exercise intolerance preceding rhabdo +/OR weakness on examination >4mths after event +/OR family history documented rhabdo +/OR biochemistry classical of VLCAD, MADD, or CPT2 +/OR cardiomyopathy<br>3. CK>500 IU/L >6 months after rhabdo episode<br><br>**Recurrent rhabdomyolysis**<br><br>All must fulfil 3 essential criteria<br><br>1. CK documented >10,000IU/L associated with muscle pain on at least one occasion<br><br>2. At least one further episode of acute muscle pain associated with documented CK rise or pigmenturia<br><br>3. Mitochondrial myopathy/PEO considered and excluded where appropriate<br><br>**Other criteria for rhabdo panel testing**<br><br>Clinical suspicion metabolic myopathy AND any of<br><br>- Moderate to profound XS lipid or glycogen on biopsy<br>- Cores/minicores on biopsy<br><br>Muscle MRI characteristic of RYR1 | *ACADVL, AGL, ALDOA, ANO5, CACNA1S, CAPN3, CAV3, CPT2, DMD, DYSF, ENO3, ETFA, ETFB, ETFDH, FKRP, GAA, GBE1, GMPPB, GYG1, GYS1, HADHA, HADHB, ISCU, LDHA, LPIN1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PNPLA2, PYGM, RBCK1, RYR1, SLC22A5, TANGO2* |

| Subpanel | Genes |
| --- | --- |
| **Arthrogryposis**<br><br>Referral by Clinical Genetics<br><br>**Criteria:** Antenatally detected joint contractures of more than two *different* joints OR Born with joint contractures of more than two *different* joints. All cases should have DM1 testing before panel testing<br><br>**Exclusion:** Isolated talipes. Finger contractures/camptodactyly with no other joint contractures<br><br>*Please consider alternative appropriate panels in children with definite cognitive involvement, particularly those where arthrogryposis is mild or additional clinical features are present.* | *ACTA1, ADAMTS10, ANTXR2, ASCC1, ASXL1, B3GALNT2, B4GAT1, BICD2, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CNTNAP1, COL12A1, COL6A1, COL6A2, COL6A3, COLQ, DAG1, DNM2, DOK7, DPAGT1, DYNC1H1, ECEL1, ERCC6, ERCC8, EXOSC3, FAM20C, FBN2, FGFR2, FKBP10, FKRP, FKTN, GBA, GBE1, GLDN, GLE1, GMPPB, ADGRG6, HSPG2, ISPD, KLHL40, KLHL41, LAMA2, LARGE1, LMOD3, MAGEL2, MPZ, MTM1, MUSK, MYBPC1, MYH2, MYH3, MYH7, MYH8, NALCN, NEB, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PIEZO2, PLOD1, PLOD2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POR, PRG4, RAPSN, RYR1, SCARF2, SCN4A, SKI, SLC5A7, SMAD4, STAC3, SYNE1, TMEM5, TNNI2, TNNT1, TNNT3, TPM2, TPM3, TRPV4, TSEN54, UBA1, VAMP1, VIPAS39, VPS33B, ZC4H2* |

# RENAL - DUNDEE

| Subpanel | Genes |
|---|---|
| Ciliopathy (Polycystic Kidney Disease, Nephronophthisis and Related Disorders Sub Panel) | *ALMS1, ACVR2B, AHI1, ANKS6, ARL13B, ARL6, ARMC4, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C21orf59 (CFAP298), C2CD3, C5orf42 (CPLANE1), CC2D2A, CCDC103, CCDC114, CCDC151, CCDC28B, CCDC39, CCDC40, CCDC65, CCNO, CEP120, CEP164, CEP290, CEP41, CEP83, CFAP53, CFC1, CRELD1, CSPP1, DDX59, DNAAF1, DNAAF2, DNAAF3, DNAAF4 (DYX1C1), DNAAF5, DNAH11, DNAH5, DNAI1, DNAI2, DNAL1, DRC1, DYNC2H1, EVC, EVC2, GDF1, GLI3, GLIS2, HNF1B, HYDIN, HYLS1, IFT122*, IFT140, IFT172, IFT27, IFT43, IFT80, INVS, IQCB1, KIF7, LBR, LRRC6, LZTFL1, MKKS, MKS1, MUC1, NEK1, NEK8, NME8, NODAL, NPHP1, NPHP3, NPHP4, OFD1, PDE6D, PKD1, PKD2, PKHD1, POC1B, PRKCSH, RPGR, RPGRIP1L, RSPH1, RSPH4A, RSPH9, SBDS, SCLT1, SDCCAG8, SEC63, SPAG1, TCTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TRIM32, TTC21B, TTC8, UMOD, WDPCP, WDR19, WDR34, WDR35, WDR60, ZIC3, ZMYND10, ZNF423* |
| Bartter Syndrome and Gitelman Syndrome | *BSND, CLCNKB, KCNJ1, SLC12A1, SLC12A3* |
| Steroid Resistant Nephrotic Syndrome (SRNS) | *ACTN4, ALG1 , ALMS1 , ANKS6 , ANLN, APOL1 , ARHGAP24, ARHGDIA, CD151, CD2AP, CFH, CLK1 (COQ7), CLCN5, COL4A1, COL4A3, COL4A4, COL4A5, COQ2, COQ6, COQ8B, COQ9, CRB2, CUBN, CYP11B2, DGKE, EMP2, FAT1, GLA, INF2, ITGA3, ITGB4, KANK1, KANK2, KANK4, LAMB2, LMNA, LMX1B, MAGI2, MEFV, MUC1, MYH9, MYO1E, NEIL1, NPHP4, NPHS1, NPHS2, NUP107, NXF5, OCRL, PAX2, PDSS2, PLCE1, PMM2, PODXL, PTPRO, SCARB2, SMARCAL1, SYNPO, TRPC6, TTC21B, VIPAS39, VPS33B, WDR73, WT1, XPO5, ZMPSTE24* |
| Gitelman Syndrome | *SLC12A3, CLCNKB* |
| Branchiootorenal syndrome 1 and 2 | *EYA1, SIX1, SIX5* |
| Autosomal Dominant Tubulointerstitial Kidney Disease (ADTKD) (previously known as Hyperuricemic Nephropathy, Familial Juvenile, type 1 and 2) | *REN, UMOD* |
| Renal Cancer | *BAP1, FH, FLCN, MET, PTEN, SDHB, VHL* |
| Primary hyperoxaluria | *AGXT, GRHPR, HOGA1* |

# RESPIRATORY - GLASGOW

| Subpanel | Genes |
|---|---|
| **Surfactant Metabolism Dysfunction**<br>Referrals from Clinical Geneticists & Consultant Intensivists.<br>**Criteria:** Neonatal respiratory insufficiency of disproportionate severity for advanced gestation, with clinical and X-ray features consistent with pulmonary surfactant deficiency AND no other obvious cause for respiratory distress e.g. no difficult delivery, no infection, not premature | *ABCA3, NKX2-1, SFTPB, SFTPC* |
| **Primary Ciliary Dyskinesia**<br>Referrals from Clinical Geneticists, Consultant Paediatricians or Consultants in Respiratory Medicine.<br>**Criteria**:<br>Neonate - at least one of the following:<br>a) Situs inversus plus lower airway or nasal symptoms<br>b) Persistent respiratory distress where other causes have been excluded<br>c) Persistent rhinorrhoea and cough distress where other causes have been excluded<br>d) Sibling with PCD<br>Childhood – at least one of the following:<br>a) Persistent lifelong wet cough (cystic fibrosis excluded)<br>b) Unexplained bronchiectasis (cystic fibrosis excluded)<br>c) Serious otitis media in association with recurrent lower and upper airway symptoms<br>Adults – symptoms as above since early childhood, often associated with infertility or subfertility | *ARMC4, C21ORF59, CCDC39, CCDC40, CCDC65, CCDC103, CCDC114, CCDC151, CCNO, DNAAF1, DNAAF2, DNAAF3, DNAAF5, DNAH5, DNAH11, DNAI1, DNAI2, DNAL1, DRC1, DYX1C1, GAS8, LRRC6, MCIDAS, RPGR, RSPH1, RSPH4A, RSPH9, SPAG1, ZMYND10* |

# SKIN - DUNDEE

| Subpanel | Genes |
|---|---|
| Ichthyosis and erythrokeratoderma (includes ARCI subpanel) | *AAGAB, ABCA12, ALOX12B, ALOXE3, AQP5, CARD14, CAST, CERS3, CLDN1, CYP4F22, DSC2, DSG1, DSP, ENPP1, FLG, GJA1, GJB2, GJB3, GJB4, GJB6, JUP, KDSR, KRT1, KRT10, KRT14, KRT16, KRT17, KRT2, KRT6A*, KRT6B*, KRT6C*, KRT9, LOR, MSMO1, NIPAL4, PIGL, PNPLA1, RSPO1, RHBDF2, SERPINB7, SLC27A4, SLURP1, SMARCAD1, SNAP29, SPINK5, ST14, STS, SULT2B1, TAT, TGM1, TRPV3* |
| Epidermolysis bullosa | *COL17A1, COL7A1, DSP, DST, EXPH5, FERMT1, ITGA3, ITGA6, ITGB4, JUP, KRT14, KRT5, LAMA3, LAMB3, LAMC2, PKP1, PLEC, TGM5* |
| Palmoplantar keratodermas | *AAGAB, ABCA12, ABHD5, ADAM17, ALDH3A2, ALOX12B, ALOXE3, AP1S1, AQP5, ARSE, CAST, CDSN, CERS3, CLDN1, CSTA, CTSC, CYP4F22, DSC2, DSC3, DSG1, DSG4, DSP, EBP, ELOVL4, ENPP1, FLG, GJA1, GJB2, JUP, KANK2, KDSR, KRT1, KRT10, KRT2, KRT6C*, KRT9, LIPN, MBTPS2, MVK, LOR, NIPAL4, NSDHL, PEX7, PHYH, PKP1, PNPLA1, POMP, RHBDF2, RSPO1, SERPINB7, SLC27A4, SLURP1, SNAP29, SPINK5, ST14, STK11, STS, SULT2B1, TGM1, TRPV3, VPS33B* |
| Vascular skin disorders<br>Under review | *ACVRL1, ADAMTS13, ALAS2, ATM, ATR, CCBE1, ENG, EPHB4, F12, FECH, FLT4, FOXC2, GLMN, KRIT1, PIK3CA, PIK3R2, RASA1, SCN9A, SMAD4, SOX18, TEK, TMEM173* |
| Rare genetic inflammatory skin disorders | *ABCC6, ADA2, ADAMTS2, AGPAT2, AIRE, ANTXR2, ATP6V0A2, ATP7A, ATP7B, CARD11, CARD14, CARD9, COL1A1, COL1A2, COL3A1, COL4A3, COL4A4, COL4A5, COL5A1, COL5A2, CSTA, CYBB, DCLRE1C, DOCK8, EFEMP2, EGFR, ELN, FBLN5, FGF23, FLG, FLT4, FMO3, FOXC2, GALNT3, GGCX, GJA1, GJB3, IKBKG, IL1RN, GJB4, IL36RN, KIT, KRT1, KRT10, LYST, NCSTN, NLRP1, NLRP3, NOD2, NSDHL, OSMR, PSEN1, PSENEN, RAG1, RAG2, SAMHD1, SH3PXD2B, SLC39A4, STAT3, TMEM173, TREX1* |
| Ectodermal dysplasia | *APCDD1, CDH3, CDSN, DSG4, EDA, EDAR, EDARADD, FGF2, FGF3, FGF10, GJB2, GJB6, GRHL2, HLA-DRA, HOXC13, HR, IKBKG, KRT14, KRT71, KRT74, KRT81, KRT83, KRT85, LIPH, LPAR6, MBTPS2, MSX1, NECTIN1, NECTIN4, NFKB2, NFKBIA, PKP1, PORCN, RSPO4, SNRPE, TP63, TSPEAR, WNT10A* |
| Hair disorders | *APCDD1, ATP7A, CDH3, CDSN, DSC3, DSG4, EDAR, ERCC2, GJB2, GJB6, HOXC13, HR, JUP, KRT71, KRT74, KRT81, KRT83, KRT85, KRT86, LIPH, LPAR6, MBTPS2, RIPK4, SNRPE, SPINK5, VDR* |