**LISTA DE PATOLOGIAS GENETICAS Y LOS GENES RELACIONADOS**

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| Acute myeloid leukemia (ARHGEF12, CBFB, CEBPA, FLT3, GATA2, JAK2, KIT, LPP, NPM1, NPM1, NSD1, PICALM, RUNX1, SH3GL1, TERT) |
| Aicardi-Goutieres syndrome (TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1) |
| Albinism & Hermansky-Pudlak syndrome (AP3B1, BLOC1S3, C10ORF11, DTNBP1, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LYST, MC1R, MLPH, MITF, MYO5A, OCA2, RAB27A, SLC45A2, TYR, TYRP1). Read the factsheet on Breda Genetics Academy. |
| Alport syndrome (COL4A3, COL4A4, COL4A5) |
| Alzheimer disease and other dementias (APOE, APP, C9orf72, CHMP2B, CSF1R, FUS, GRN, MAPT, PRNP, PSEN1, PSEN2, SIGMAR1, SORL1, TARDBP, TREM2, UBE3A, UBQLN2, VCP) |
| Amyotrophic lateral sclerosis (ALS2, ANG, ATXN2, C9orf72, CHMP2B, CHGB, DCTN1, FIG4, FUS, NEFH, OPTN, PFN1, PRPH2, SETX, SIGMAR1, SOD1, SPG20, TARDBP, UBQLN2, VAPB, VCP, VEGFA) |
| [Anophthalmia, microphthalmia isolated/syndromic (ALDH1A3, ALX1, BCOR, BMP4, COX14, COA5, CHD7, COX6B1, CRYBA4, CRYBB2, CRYGD, DPYD, ERCC6, ESCO2, EYA1, FASTKD2, FOXE3, FOXL2, FRAS1, FREM1, FREM2, GDF3, GDF6, GJA1, GLI3, HCCS, HESX1, HMX1, IKBKG, ISPD, KERA, KIF11, MAF, MBTPS2, MFRP, NDP, OCLN, OTX2, PAX6, POMT1, PORCN, PQBP1, RAB18, RAB3GAP1, RAB3GAP2, RAX, RIPK4, SALL4, SHH, SIX3, SIX6, SLC36A2, SLC6A19, SLC6A20, SMAD4, SMOC1, SOX2, STRA6, TFAP2A, VCAN, VSX2).](http://www.bredagenetics.com/academy/anophthalmia-and-microphthalmia-isolated-and-syndromic/)  |
| Aortic aneurysm, familial thoracic (MYH11, ACTA2, TGFBR1, TGFBR2, FBN1, COL3A1, SMAD3, FBN2, SLC2A10, MYLK, TGFB2) |
| Arrhythmia, hereditary (AKAP9, ANK2, CACNA1C, CACNB2, CASQ2, CAV3, DSC2, DSG2, DSP, GPD1L, JUP, KCNA5, KCNE1, KCNE2, KCNE3, KCNH2, KCNJ2, KCNQ1, NPPA, PKP2, PLN, RYR2, SCN1B, SCN3B, SCN4B, SCN5A, SNTA1, TGFB3, TMEM43) |
| Arrhythmogenic right ventricular cardiomyopathy (DSP, DSG2, DSC2, JUP, PKP2, RYR2, TGFB3, TMEM43) |
| Arthrogryposis (TPM2, MYBPC1, MYH3, TNNT3, TNNI2, MYH8, FBN2) |
| Ashkenazi (GBA, CFTR, HEXA, IKBKAP, ASPA, G6PC, ABCC8, MCOLN1, BCKDHB, FANCC, DLD, SMPD1, CLRN1, PCDH15, BLM, NEB, BRCA1, BRCA2) |
| Autism spectrum disorders (EN2, MECP2, NLGN3, NLGN4X, PDE8B, RPL10) |
| Bardet Biedl syndrome (ALMS1, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CCDC28B, CEP290, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP) |
| Bartter Syndrome (ATP6V1B1, BSND, CA2, CASR, CLCNKA, CLCNKB, CLDN16, CLDN19, FXYD2, HSD11B2, KCNJ1, KCNJ10, KLHL3, NR3C2, SCNN1A, SCNN1B, SCNN1G, SLC12A1, SLC12A3, SLC4A1, SLC4A4, WNK1, WNK4) |
| Bethlem myopathy (COL6A1, COL6A2, COL6A3, COL12A1) |
| Breast ovarian cancer (ATM, BARD1, BRCA1, BRCA2, BRIP1, CHEK2, EPCAM, MEN1, MLH1, MRE11A, MSH2, MSH6, MUTYH, NBN, PALB2, PMS1, PMS2, RAD50, RAD51C, RAD51D, STK11, TP53, XRCC2) |
| Brugada syndrome (CACNA1C, CACNB2, GPD1L, HCN4, KCNE3, SCN1B,SCN3B, SCN5A) |
| [Cantú syndrome, Berardinelli-Seip syndrome and their differential diagnosis [incl. mucopolysacchcaridosis I, II, IVA, mucolipidosis III, and alpha-mannosidosis and Beckwith-Wiedemann syndrome] (ABCC9, KCNJ8, AGPAT2, BSCL2, CDKN1C, IDUA, IDS, GALNS, GNPTAB, GNPTG, MAN2B1). Read the factsheet on Breda Genetics Academy.](http://www.bredagenetics.com/academy/cantu-syndrome/) |
| Cardiomyopathy, dilated (ABCC9, ACTC1, ACTN2, ANKRD1, BAG3, CSRP3, DES, DMD, DSG2, EYA4, FKTN, GATAD1, LAMP2, LDB3, LMNA, MYBPC3, MYH6, MYH7, NEXN, PLN, PSEN1, PSEN2, RBM20, SCN5A, SDHA, SGCD, TAZ, TCAP, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TTN, TTR, VCL) |
| Cardiomyopathy, hypertrophic (ACTC1, CALR3, CAV3, CSRP3, GLA, JPH2, LAMP2, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOZ2, NEXN, PLN, PRKAG2, SLC25A4, TNNC1, TNNI3, TNNT2, TPM1, TTN, TTR, VCL) |
| Cataract (AGK, CRYAA, CRYAB, CRYBB1, CRYBB3, CTDP1, FYCO1, GCNT2, GJA8, HSF4, LIM2, SIL1, TDRD7) |
| Catecholaminergic polymorphic ventricular tachycardia (RYR2, CASQ2, KCNJ2) |
| Central hypoventilation syndrome (RET, GDNF, EDN3, BDNF, ASCL1, PHOX2A, PHOX2B, ZEB2, GFRA1, ECE1, MECP2) |
| Cerebellar ataxia (ADCK3, APTX, COQ2, COQ9, DNMT1, FXN, PDSS1, PDSS2, POLG, SACS, SETX, SYNE1, TTPA, VLDLR) |
| Ceroid lipofuscinosis (CLN3, CLN5, CLN6, CLN8, CTSD, DNAJC5, MFSD8, PPT1, TPP1) |
| Charcot-Marie-Tooth neuropathy axonal/demyelinating & Dejerine Sottas syndrome (AARS, ARHGEF10, CTDP1, DNM2, EGR2, FGD4, FIG4, GAN, GARS, GDAP1, GJB1, DYNC1H1, HSPB8, HSPB1, LITAF, MTMR2, KIF1B, LMNA, MED25, MFN2, MPZ, NDRG1, NEFL, RAB7A, PMP22, PRPS1, PRX, SBF2, SH3TC2, SLC12A6, TRPV4, YARS) |
| Ciliary primary dyskinesia panel (DNAI1, DNAAF2, DNAAF3, DNAH5, HYDIN , NME8, DNAH11, DNAI2 , RSPH4A, RSPH9, DNAAF1, CCDC39 , CCDC40, DNAL1, CCDC103, HEATR2, LRRC6) |
| CMT neuropathy axonal/demyelinating (AARS, ARHGEF10, CTDP1, DNM2, EGR2, FGD4, FIG4, GAN, GARS, GDAP1, GJB1, DYNC1H1, HSPB8, HSPB1, LITAF, MTMR2, KIF1B, LMNA, MED25, MFN2, MPZ, NDRG1, NEFL, RAB7A, PMP22, PRPS1, PRX, SBF2, SH3TC2, SLC12A6, TRPV4, YARS) |
| Coffin-Siris syndrome (ARID1A, ARID1B, SMARCA4, SMARCB1, SMARCE1) |
| Colon & gastric cancer, with/without polyposis (APC, BMPR1A, CDH1, MLH1, MSH2, MSH6, MUTYH, PMS1, PMS2, PTEN, SMAD4, STK11 MSH2, MLH1, MSH6, PMS2, EPCAM) |
| Cone-rod and cone dystrophy (ABCA4, ADAM9, AIPL1, BEST1, C8ORF37, CABP4, CACNA1F, CACNA2D4, CDHR1, CERKL, CNGB3, CNNM4, CRX, GUCA1A, GUCY2D, KCNV2, PDE6C, PDE6H, PITPNM3, PROM1, PRPH2, RAX2, RDH5, RGS9, RGS9BP, RIMS1, RPGR, RPGRIP1, SEMA4A, UNC119) |
| Congenital ichthyosis (ALOX12B, ABCA12, ALOXE3, CYP4F22, LIPN, NIPAL4, PNPLA1, TGM1) |
| Congenital myasthenic syndrome (AGRN, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, COLQ, DOK7, GFPT1, LAMB2, MUSK, PLEC, RAPSN, SCN4A) |
| Cornelia de Lange syndrome (HDAC8, NIPBL, RAD21, SMC1A, SMC3) |
| Deafness, non-syndromic sensorineural autosomal dominant/recessive/X-linked (ACTG1, CCDC50, COCH, COL11A2, COL4A6, CRYM, CDH23, CLDN14, COL11A2, DFNA5, DFNB31, DFNB59, DIABLO, DIAPH1, DIAPH3, ESPN, ESRRB, EYA4, FOXI1, GIPC3, GJB2, GJB3, GJB6, GPSM2, GRXCR1, GRHL2, HGF, ILDR1, KCNJ10, KCNQ4, LHFPL5, LOXHD1, LRTOMT, MARVELD2, MIR96, MSRB3, MYH14, MYH9, MYO1A, MYO15A, MYO3A, MYO6, MYO7A, MYO6, MYO7A, OTOA, OTOF, PCDH15, POU3F4, POU4F3, PRPS1, PTPRQ, RDX, SERPINB6, SIX1, SLC12A1, SLC17A8, SMPX, SLC26A4, SLC26A5, SMPX, STRC, TECTA, TJP2, TMC1, TMIE, TMPRSS3, TPRN, TRIOBP, USH1C, WFS1) |
| [Dense deposit disease, membranoproliferative glomerulonephritis II, atypical hemolytic uremic syndrome & thrombotic thrombocytopenic purpura (ADAMTS13, C3, CD46, CFB, CFHR1, CFHR3, CFHR4, CFHR5, CFH, CFI, LMNA, LCAT, THBD). Read the factsheet on Breda Genetics Academy.](http://www.bredagenetics.com/academy/membranoproliferative-glomerulonephrites/) |
| Diabetes neonatal (ABCC8, FOXP3, G6PC2, GCK, GLIS3, INS, INSR, KCNJ11, NEUROG3, PDX1) |
| Diamond-Blackfan anemia (RPL11, RPL35A, RPL5, RPS10, RPS17, RPS19, RPS24, RPS26, RPS7) |
| Dravet syndrome (SCN1A, GABRG2, SCN2A, SCN9A) |
| Dystonia panel (TOR1A, THAP1, GCH1, TH, SPR, SLC2A1, PRRT2, PNKD, SGCE) |
| Early infantile epileptic encephalopathy (ARX, CDKL5, SLC25A22, STXBP1, SPTAN1, SCN1A, KCNQ2, ARHGEF9, PCDH19, PNKP, SCN2A, SCN8A, PLCB1) |
| Ehlers-Danlos syndrome, Marfan syndrome, Familial Thoracic Aortic Aneurysm Dissection & Arterial tortuosity syndrome (ACTA2, CBS, COL3A1, COL5A1, COL5A2, FBN1, FBN2, FLNA, MED12, MYH11, MYLK, PLOD1, SKI, SLC2A10, SMAD3, TGFB2, TGFBR1, TGFBR2, TNXB) |
| Epidermolysis bullosa (COL7A1, COL17A1, ITGA6, ITGB4, KRT5, KRT14, LAMA3, LAMB3, LAMC2, PLEC, CD151, CDSN, CHST8, CSTA, DSG1, DSG2, DSG4, DSP, DST, FERMT1, GRIP1, ITGA3 , MMP1, NID1, PKP1, TGM5) |
| Epilepsy, hereditary (CACNA1H, CACNB4, CHRNA2, CHRNA4, CHRNB2, CLCN2, CPA6, EFHC1, GABRA1, GABRB3, GABRD, GABRG2, JRK, KCNMA1, KCNQ2, KCNQ3, KCNT1, LGI1, SCN1A, SCN1B, SCN2A, SCN8A, SCN9A, SLC2A1, SRPX2) |
| Epileptic encephalopathy (ACY1, ADSL, ALDH7A1, AMT, ARHGEF9, ARX, CDKL5, CNTNAP2, CPT2, FOLR1, FOXG1, GABRG2, GAMT, GCSH, GLDC, GRIN2A, GRIN2B, KCNJ10, KCNQ2, MAGI2, MAPK10, MECP2, MTHFR, NRXN1, PCDH19, PLCB1, PNKP, PNPO, PRRT2, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, SCN1A, SCN1B, SCN2A, SCN8A, SCN9A, SLC25A22, SLC2A1, SLC9A6, SPTAN1, STXBP1, TBCE, TCF4, TREX1, UBE3A, ZEB2) |
| Episodic ataxia (CACNA1A, CACNB4, KCNA1, SLC1A3) |
| Familial hemiplegic migraine, CADASIL, retinal vasculopathy with cerebral leukodystrophy, hereditary hemorrhagic telangiectasia, familial cerebral cavernous malformations & alternating hemiplegia of childhood (ATP1A2, CACNA1A, SCN1A, NOTHC3, TREX1, ENG, ACVRL1, SMAD4, KRIT1, CCM2) |
| Familial hypercholesterolemia panel (APOB, GHR, LDLR, PCSK9) |
| Fanconi anemia (BRCA2, BRIP1, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, PALB2, SLX4, XRCC2) |
| Female infertility (CYP21A2, FSHR, LHCGR, BMP15, LHB, FMR1) |
| Fleck retina, choroideremia and age-related macular degeneration (ABCA4, CFH, CFB, CHM, EFEMP1, PLA2G5, RDH5, RLBP1, RS1, TIMP3, VPS13B) |
| Focal Glomerulonephrosis/Nephrotic syndrome (ACTN4, CD2AP, INF2, NPHS1, NPHS2, TRPC6, WT1, LAMB2, PLCE1) |
| Frontotemporal dementia (CHMP2B, GRN, VCP, FUS, TARDBP, C9orf72 , MAPT, SIGMAR1, UBQLN2) |
| Glycogen storage disease (GYS1, GYS2, G6PC, SLC37A4, GAA, AGL, GBE1, PYGM, PYGL, PFKM, PHKA2, PGAM2, LDHA, ALDOA, ENO3, PHKB, PHKA1, PGM1, GYG1, PRKAG2, PHKG2) |
| Glycogen/lipid storage myopathies, with/without rhabdomyolysis & other metabolic myopathies (ABHD5, ACADM, ACADL, ACAD9, ACADVL, AGL, AMPD1, C10orf2, COA5, COX6B1, COX10, COX14, CPT1B, CPT2, CTDP1, ENO3, ETFA, ETFB, ETFDH, FASTDK2, GAA, GBE1, GYG1, GYS1, HADHA, HADHB, ISCU, LDHA, LPIN1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PNPLA2, POLG, POLG2, PRKAG2, PUS1, PYGM, RRM2B, SLC16A1, SLC22A5, SLC25A20, SUCLA2, TACO1, TAZ, TK2, TYMP, YARS2) |
| Glycosylation disorders (ALG1, ALG2, ALG3, ALG6, ALG8, ALG9, ALG11, ALG12, ALG13, B4GALT1, COG1, COG4, COG5, COG6, COG7, COG8, DOLK, DPAGT1, DPM1, DPM3, MGAT2, MOGS, MPDU1, MPI, PMM2, RFT1, SLC35A1, SLC35C1) |
| [Hemophagocytic lymphohistiocytosis and its differential diagnosis (PRF1, UNC13D, STX11, STXBP2, RAB27A, XIAP, SH2D1A, CHS1). Read the factsheet on Breda Genetics Academy.](http://www.bredagenetics.com/academy/familial-hemophagocytic-lymphohistiocytosis/) |
| [Hereditary red cell membrane disorders [including: hereditary spherocytosis, elliptocytosis, pyropoikilocytosis, and stomatocystosis] (ABCB6, RHAG, RHCE, SLC2A1, ANK1, SPTB, SPTA1, EPB41, EPB42, ABCG5, ABCG8, SLC4A1, GYPC, HFE). Read the factsheet on Breda Genetics Academy.](http://www.bredagenetics.com/academy/hereditary-red-cell-membrane-disorders/) |
| Heterotaxy, visceral, Tetralogy of Fallot, VATER & VACTERL(X) associations (ACVR2B, CFC1, CRELD1, GJA1, GDF1, FOXH1, LEFTY2, NKX2-5, NODAL, ZIC3) |
| Hirschsprung disease (ECE1, EDN3, EDNRB, GDNF, KIAA1279, NRG1, NRTN, RET, SOX10, ZEB2) |
| [Histone-modification disorders and their differential diagnosis (incl. Wiedemann-Steiner, Kabuki, Coffin-Siris, Nicolaides-Baraitser, and Cornelia de Lange syndromes) (KMT2A, KMT2D, KDM6A, SMARCA4, SMARCA2, NIPBL, SMC1A, SMC3, RAD21, HDAC8). Read the factsheet on Breda Genetics Academy.](http://www.bredagenetics.com/academy/histone-modification-disorders/) |
| Holoprosencephaly (CDON, FGF8, GLI2, GLI3, PTCH1, SHH, SIX3, TGIF1, ZIC2) |
| Hyperekplexia (ARHGEF9, GLRA1, GLRB, GPHN, SLC6A5) |
| Kallmann syndrome (CHD7, FGFR1, FGF8, GNRHR, GNRH1, KAL1, KISS1R, PROK2, PROKR2, SEMA3A, TAC3, TACR3) |
| Leber congenital amaurosis (AIPL1, CABP4, CEP290, CRB1, CRX, GUCY2D, IMPDH1, IQCB1, KCNJ13 , LCA5, LRAT, NMNAT1, OTX2, RD3, RDH12, RPE65, RPGRIP1, SPATA7, TULP1) |
| [Leigh syndrome/NARP & their differential diagnosis (ADAR1, BCS1L, BTD, C12orf65, C20ORF7, C8ORF38, COX10, COX15, DLD, EARS2, ETHE1, FARS2, FOXRED1, GFM1, HIBCH, HLCS, LIAS, LIPT1, LRPPRC , MCEE, MMAA, MMAB, MMADHC,  MT-ATP6, MT-CO3, MTFMT, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND5, MT-ND6, MT-TK, MT-TV, MT-TW, MUT , NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA9, NDUFAF2, NDUFAF5, NDUFAF6, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NUP62, PANK2, PCCA , PCCB, PDHA1, PDHB, PDHX, PDSS2, PET100, POLG, RANBP2, SCO2, SDHA, SERAC1, SLC19A3, SLC25A19, SUCLA2, SUCLG1, SURF1, TACO1, TTC19, UQCRQ). Read the factsheet on Breda Genetics Academy.](http://www.bredagenetics.com/academy/leigh-syndrome/) |
| Leukodystrophy and peroxisome biogenesis disorders (ABCD1, AIMP1, ARSA, ASPA, BEST1, CSF1R, CYP27A1, DARS2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, FA2H, FAM126A, GALC, GFAP, GJC2, HEPACAM, HSPD1, MLC1, NDUFV1, NOTCH3, PEX1, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PLP1, POLR3A, POLR3B, PSAP, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, SAMHD1, SDHA, SLC16A2, SOX10, SUMF1, TREM2, TREX1, HSD17B4, LMNB1, PEX11B, PHYH, SCP2, SDHAF1, TYROBP) |
| Limb-girdle muscular dystrophy (ANO5, CAPN3, CAV3, DAG1, DNAJB6, DYSF, FKRP, FKTN, LAMA2, LMNA, MYOT, PLEC, POMGNT1, POMT1, POMT2, SEPN1, SGCA, SGCB, SGCD, SGCG, TCAP, TRIM32, TTN (hot Spot testing)) |
| Lissencephaly (ARX, DCX, NDE1, PAFAH1B1, POMT1, POMT2, RELN, TUBA1A, YWHAE) |
| Long QT syndrome (AKAP9, ANK2, CACNA1C, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, SCN4B, SCN5A, SNTA1) |
| Lysosomal storage disease (ARSA, FUCA1, GALC, GBA, GLB1, GNPTAB, GUSB, HEXA, HEXB, MAN2B1, MANBA, NAGA, SMPD1) |
| Male infertility (AR, CATSPER1, CFTR, FSHR, LHCGR, AZF) |
| Malignant hyperthermia – 1179 € – (CACNA1S, RYR1) |
| Maple syrup urine disease (BCKDHA, BCKDHB, DBT, DLD) |
| Marfan syndrome and related disorders (ACTA2, COL3A1, COL5A1, COL5A2, FBN1, FBN2, MYH11, SLC2A10, SMAD3, TGFBR1, TGFBR2) |
| [Meckel-Gruber & Joubert syndrome (AHI1, ARL13B, B9D1, B9D2, C5orf42, CC2D2A, CEP290, CEP41, KIF7, MKS1, NPHP1, NPHP3, OFD1, RPGRIP1L, TCTN1, TCTN2, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TTC21B). Read the factsheet on Breda Genetics Academy.](http://www.bredagenetics.com/academy/meckel-meckel-gruber-syndrome/) |
| Megaloblastic anemia with/without homocystinuria – disorders of intracellular cobalamin metabolism (AMN, CUBN, GIF, SLC19A2, DHFR, MTR, MMACHC, MMADHC, MTRR, LMBRD1, ABCD4) |
| Mental retardation, X-linked (ABCD1, ACSL4, AFF2, AGTR2, AP1S2, ARHGEF6, ARHGEF9, ARX, ATP6AP2, ATP7A, ATRX, BCOR, BRWD3, CASK, CDKL5, CUL4B, DCX, DKC1, DLG3, ELK1, FANCB, FGD1, FLNA, FMR1, FTSJ1, GDI1, GK, GPC3, GRIA3, HCCS, HPRT1, HSD17B10, HUWE1, IDS, IGBP1, IL1RAPL1, KIAA2022, KDM5C, L1CAM, LAMP2, MAGT1, MAOA, MBTPS2, MECP2, MED12, MID1, MTM1, NDP, NDUFA1, NHS, NLGN3, NLGN4X, NSDHL, NXF5, OCRL, OFD1, OPHN1, OTC, PAK3, PCDH19, PDHA1, PGK1, PHF6, PHF8, PLP1, PORCN, PQBP1, PRPS1, RAB39B, RPL10, RPS6KA3, SHROOM4, SLC16A2, SLC6A8, SLC9A6, SMC1A, SMS, SOX3, SRPX2, SYN1, SYP, TIMM8A, TSPAN7, UBE2A, UPF3B, ZCCHC12, ZDHHC9, ZDHHC15, ZNF41, ZNF81, ZNF674, ZNF711) |
| Metaphyseal dysplasia (ANKH, CDKN1C, FLNA, MMP9, MMP13, NKX3-2, RMRP, RUNX2) |
| Methylmalonic acidemia(ABCD4, ACSF3, CD320, LMBRD1, MCEE, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MTR, MTRR, MUT, SUCLA2, SUCLG1) |
| Microcephaly (AKT3, AP4M1, ASPM, CASK, CDK5, RAP2, CENPJ, CEP135, CEP152, CEP63, DNM1L, EFTUD2, IER3IP1, KIF11, MCPH1, MRE11A, MSMO1, NDE1, NHEJ1, NR2E1, PAFAH1B1, PCNT, PNKP, POMT1, SLC25A19, STIL, TUBB2B, TUBGCP6, WDR62) |
| Mitochondrial complex IV deficiency (COA5, COX6B1, COX10, COX14, FASTKD2, TACO1) |
| MODY (ABCC8, BLK, CEL, GCK, HNF1A, HNF1B, HNF4A, INS, KLF11, NEUROD1, PAX4, PDX1, RFX6, ZFP57) |
| Mucopolysaccharidosis (ARSB, GALNS, GLB1, GNPTAB, GNPTG, GNS, GUSB, HGSNAT, IDS, IDUA, NAGLU, SGSH) |
| Multiple epiphyseal dysplasia (COL2A1, COL9A1, COL9A2, COL9A3, COMP, MATN3, SLC26A2) |
| Myofibrillar myopathy, inclusion body myopathy 2, dyspherlinopathy & Laing distal myopathy (BAG3, CRYAB, DES, DNAJB6, DYSF, FHL1, GNE, FLNC, LDB3, MYH7, MYOT) |
| [Nemaline myopathy and other congenital myopathies (ACTA1, BIN1, CCDC78, CFL2, CNTN1, DNM2, FHL1, KBTBD13, MAMLD1, MTM1, MTMR14, MYF6, MYH7, NEB, RYR1, SEPN1, TNNT1, TPM2, TPM3). Read the factsheet on Breda Genetics Academy.](http://www.bredagenetics.com/academy/congenital-myopathies/) |
| Nephronophthisis (NPHP1, INVS, NPHP3, NPHP4, IQCB1, CEP290, GLIS2, RPGRIP1L, NEK8, SDCCAG8) |
| Nephrotic syndrome/Focal Segmental Glomerulosclerosis (LAMB2, NPHS1, NPHS2, PLCE1, WT1, ACTN4, CD2AP, INF2, TRPC6) |
| Neurofibromatosis (NF1, NF2, SPRED1) |
| Neuronal migration disorders (ACTB, ACTG1, ARFGEF2, ARX, COL18A1, COL4A1, CPT2, DCX, EMX2, EOMES, FGFR3, FH, FKRP, FKTN, FLNA, GPR56, IER3IP1, ISPD, LAMA2, LAMC3, LARGE, MED12, MEF2C, OCLN, PAFAH1B1, PAX6, PEX7, POMGNT1, POMT1, POMT2, PQBP1, RAB18, RAB3GAP1, RAB3GAP2, RELN, SNAP29, SRPX2, TUBA1A, TUBA8, TUBB2B, TUBB3, WDR62) |
| Noonan – CFC syndrome (BRAF, CBL, HRAS, MAP2K1, KRAS, MAP2K2, NF1, NRAS, RAF1, PTPN11, SHOC2, SOS1, SPRED1) |
| Oculomotor apraxia (APTX, PIK3R5, SETX) |
| Ophthalmoplegia, progressive external (C10ORF2, OPA1, POLG, POLG2, RRM2B, SLC25A4, TYMP) |
| Optic atrophy (AUH, C12ORF65, CISD2, NDUFS1, OPA1, OPA3, POLG, SPG7, TIMM8A, TMEM126A, WFS1) |
| Osteogenesis imperfecta (COL1A1, COL1A2, BMP1, CRTAP, FKBP10, IFITM5, LEPRE1, PLOD2, PPIB, SERPINH1, SP7) |
| Osteopetrosis panel (CA2, CLCN7, LRP5, OSTM1, PLEKHM1, SNX10, TCIRG1, TNFSF11, TNFRSF11A) |
| Pancreatic cancer (APC, ATM, BMPR1A, CDKN2A, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS1, PMS2, PRSS1, SMAD4, STK11) |
| Pancreatitis (PRSS1, SPINK1, CFTR, CTRC) |
| Pantothenate kinase-associated neurodegeneration (ATP13A2, C19orf12, CP, DCAF17, FA2H, FTL, PANK2, PLA2G6, WDR45) |
| Parkinsons disease (SNCA, LRRK2, VPS35, PARK2, PINK1, PARK7, ATP13A2, PLA2G6, FBXO7, DNAJC6) |
| Periodic fever syndrome (ELANE, LPIN2, MEFV, MVK, NLRP3, PSTPIP1, TNFRSF1A) |
| Pheochromocytoma (MAX, PRKAR1A, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL) |
| Polycystic kidney (BICC1, PKD1, PKD2, NOTCH2, PKHD1) |
| Pontocerebellar hypoplasia (CASK, TSEN2, TSEN34, TSEN54, OPHN1, RARS2, VRK1, EXOSC3) |
| Refsum disease (PEX1, PEX2, PEX26, PEX7, PHYH) |
| Renal cancer (EPCAM, FH, FLCN, HNF1A, HNF1B, MET, MITF, MLH1, MSH2, MSH6, PMS1, PMS2, PTEN, SDHB, SDHD, TSC1, TSC2, VHL, WT1) |
| Retinitis pigmentosa (ABCA4, ARL6, BBS1, BEST1, C2ORF71, C8ORF37, CA4, CERKL, CNGA1, CNGB1, CRB1, CRX, CLRN1, DHDDS, EYS, FAM161A, FLVCR1, FSCN2, GNPTG, GUCA1B, IDH3B, IMPG2, IMPDH1, KLHL7, LRAT, MAK, MERTK, NR2E3, NRL, PDE6A, PDE6B, PDE6G, PRCD, PROM1, PRPF3, PRPF31, PRPF6, PRPF8, PRPH2, RBP3, RDH12, RGR, RHO, RLBP1, ROM1, RP1, RP2, RP9, RPE65, RPGR, SAG, SEMA4A, SNRNP200, SPATA7, TTC8, TULP1, USH2A, TOPORS, ZNF513) |
| [RIDDLE syndrome and other radiosensitivities with immunodeficiency (incl. Nijmegen breakage syndrome, ataxia-telengectasia, severe combined immunodeficiency with Cernunnos, severe combined immunodeficiency Athabascan type, Immunodeficiency 26, LIG4 syndrome and X-linked agammaglobulinemia) (RNF168, NBN, ATM, NHEJ1, DCLRE1C, PRKDC, LIG4, BTK). Read the factsheet on Breda Genetics Academy.](http://www.bredagenetics.com/academy/riddle-syndrome-and-other-radiosensitivities-with-immunodeficiency/) |
| Spinocerebellar ataxia – SCA (ATXN1, ATXN2, ATXN3, ATXN7, ATXN8OS, ATXN10, BEAN1, ATN1, CACNA1A, NOP56, PPP2R2B, TBP, AFG3L2, DNMT1, FGF14, IFRD1, ITPR1, KCNC3, KCND3, PDYN, PRKCG, SPTBN2, TGM6, TTBK2) |
| Seckel syndrome (ATR, RBBP8, CENPJ, CEP152, CEP63) |
| Skin cancer (CDKN2A, EPCAM, MC1R, MITF, MLH1, MSH2, MSH6, PMS1, PMS2, PTCH1, XRCC3) |
| Spastic paraplegia, autosomal dominant/recessive (ATL1, AP4M1, BSCL2, CYP7B1, FA2H, GJC2, HSPD1, KIAA0196, KIF1A, KIF5A, NIPA1, PNPLA6, REEP1, RTN2, SLC33A1, SPAST, SPG7, SPG11, SPG20, SPG21, ZFYVE26, ZFYVE27) |
| Spherocytosis (ANK1, EPB42, SLC4A1, SPTA1, SPTB) |
| Stargardt disease (ABCA4, BEST1, C1QTNF5, CDH3, CNGB3, ELOVL4, FSCN2, PROM1, PRPH2, RDH12, RP1L1, RPGR, TIMP3) |
| Stickler syndrome (COL2A1, COL9A1, COL9A2, COL11A1, COL11A2) |
| Surfactant metabolism dysfunction (ABCA3, CSF2RA, CSF2RB, SFTPA1, SFTPB, SFTPC, SFTPD) |
| Thrombocytopenia (ADAMTS13, GATA1, GP1BA, GP1BB, GP9, ITGA2B, ITGB3, MASTL, MYH9, MPL, RUNX1, WAS) |
| Thyroid cancer (APC, PTEN, RET) |
| Tuberous sclerosis (TSC1, TSC2) |
| Ullrich muscular dystrophy (COL6A1, COL6A2, COL6A3) |
| Urea cycle disorder (ARG1, ASL, ASS1, CPS1, NAGS, OTC) |
| Usher syndrome (CDH23, CIB2, CLRN1, DFNB31, GPR98, MYO7A, PCDH15, PDZD7, USH1C, USH1G, USH2A) |
| Vitreoretinopathy and Wagner syndrome (COL2A1, FZD4, LRP5, NDP, TSPAN12, VCAN) |
| Waardenburg syndrome (EDN3, EDNRB, MITF, PAX3, SNAI2, SOX10, TYR) |
| Walker-Warburg syndrome (FKRP, FKTN, ISPD, LARGE, POMT1, POMT2) |
| Zellweger syndrome (PEX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26) |