

Supplementary Table 1; List of analyzed genes

| HGNC symbol | HGNC ID | Description |
|-------------|---------|---|
| ABCA12 | 14637 | ATP binding cassette subfamily A member 12 |
| ABCC9 | 60 | ATP binding cassette subfamily C member 9 |
| ACAN | 319 | aggrecan |
| ACOX1 | 119 | acyl-CoA oxidase 1 |
| ADK | 257 | adenosine kinase |
| AGGF1 | 24684 | angiogenic factor with G-patch and FHA domains 1 |
| AHI1 | 21575 | Abelson helper integration site 1 |
| AKT1 | 391 | AKT serine/threonine kinase 1 |
| AKT3 | 393 | AKT serine/threonine kinase 3 |
| ALX3 | 449 | ALX homeobox 3 |
| ALX4 | 450 | ALX homeobox 4 |
| AMER1 | 26837 | APC membrane recruitment protein 1 |
| ANKH | 15492 | ANKH inorganic pyrophosphate transport regulator |
| ANTXR2 | 21732 | ANTXR cell adhesion molecule 2 |
| AP1S2 | 560 | adaptor related protein complex 1 subunit sigma 2 |
| APC2 | 24036 | APC2 WNT signaling pathway regulator |
| ARL13B | 25419 | ADP ribosylation factor like GTPase 13B |
| ARL6 | 13210 | ADP ribosylation factor like GTPase 6 |
| ARMC9 | 20730 | armadillo repeat containing 9 |
| ARSB | 714 | arylsulfatase B |
| ARVCF | 728 | ARVCF delta catenin family member |
| ASPA | 756 | aspartoacylase |
| ASXL2 | 23805 | ASXL transcriptional regulator 2 |
| B3GALNT2 | 28596 | beta-1 3-N-acetylgalactosaminyltransferase 2 |
| B3GALT6 | 17978 | beta-1 3-galactosyltransferase 6 |
| B3GLCT | 20207 | beta 3-glucosyltransferase |
| B4GALT1 | 924 | beta-1 4-galactosyltransferase 1 |
| B4GALT7 | 930 | beta-1 4-galactosyltransferase 7 |
| B4GAT1 | 15685 | beta-1 4-glucuronyltransferase 1 |
| B9D1 | 24123 | B9 domain containing 1 |
| B9D2 | 28636 | B9 domain containing 2 |
| BBIP1 | 28093 | BBSome interacting protein 1 |
| BBS1 | 966 | Bardet-Biedl syndrome 1 |
| BBS10 | 26291 | Bardet-Biedl syndrome 10 |
| BBS12 | 26648 | Bardet-Biedl syndrome 12 |
| BBS2 | 967 | Bardet-Biedl syndrome 2 |
| BBS4 | 969 | Bardet-Biedl syndrome 4 |
| BBS5 | 970 | Bardet-Biedl syndrome 5 |
| BBS7 | 18758 | Bardet-Biedl syndrome 7 |
| BBS9 | 30000 | Bardet-Biedl syndrome 9 |
| BGN | 1044 | biglycan |
| BHLHA9 | 35126 | basic helix-loop-helix family member a9 |
| BLM | 1058 | BLM RecQ like helicase |
| BMP2 | 1069 | bone morphogenetic protein 2 |
| BMP4 | 1071 | bone morphogenetic protein 4 |
| BMPRI1B | 1077 | bone morphogenetic protein receptor type 1B |
| BRAF | 1097 | B-Raf proto-oncogene serine/threonine kinase |
| BRCA2 | 1101 | BRCA2 DNA repair associated |

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|----------|-------|---|
| BRWD3 | 17342 | bromodomain and WD repeat domain containing 3 |
| BUB1 | 1148 | BUB1 mitotic checkpoint serine/threonine kinase |
| BUB1B | 1149 | BUB1 mitotic checkpoint serine/threonine kinase B |
| BUB3 | 1151 | BUB3 mitotic checkpoint protein |
| C12orf57 | 29521 | chromosome 12 open reading frame 57 |
| C2CD3 | 24564 | C2 calcium dependent domain containing 3 |
| C8orf37 | 27232 | chromosome 8 open reading frame 37 |
| CAMTA1 | 18806 | calmodulin binding transcription activator 1 |
| CC2D2A | 29253 | coiled-coil and C2 domain containing 2A |
| CCDC22 | 28909 | coiled-coil domain containing 22 |
| CCND2 | 1583 | cyclin D2 |
| CD96 | 16892 | CD96 molecule |
| CDC45 | 1739 | cell division cycle 45 |
| CDC73 | 16783 | cell division cycle 73 |
| CDCA7 | 14628 | cell division cycle associated 7 |
| CDKN1C | 1786 | cyclin dependent kinase inhibitor 1C |
| CEP104 | 24866 | centrosomal protein 104 |
| CEP120 | 26690 | centrosomal protein 120 |
| CEP164 | 29182 | centrosomal protein 164 |
| CEP290 | 29021 | centrosomal protein 290 |
| CEP41 | 12370 | centrosomal protein 41 |
| CEP55 | 1161 | centrosomal protein 55 |
| CEP57 | 30794 | centrosomal protein 57 |
| CFC1 | 18292 | cripto FRL-1 cryptic family 1 |
| CHD4 | 1919 | chromodomain helicase DNA binding protein 4 |
| CHD7 | 20626 | chromodomain helicase DNA binding protein 7 |
| CHN1 | 1943 | chimerin 1 |
| CHRNA7 | 1960 | cholinergic receptor nicotinic alpha 7 subunit |
| CLCN7 | 2025 | chloride voltage-gated channel 7 |
| COG6 | 18621 | component of oligomeric golgi complex 6 |
| COL25A1 | 18603 | collagen type XXV alpha 1 chain |
| COL2A1 | 2200 | collagen type II alpha 1 chain |
| COL4A1 | 2202 | collagen type IV alpha 1 chain |
| COLEC10 | 2220 | collectin subfamily member 10 |
| COMT | 2228 | catechol-O-methyltransferase |
| CPLX1 | 2309 | complexin 1 |
| CRB2 | 18688 | crumbs cell polarity complex component 2 |
| CREBBP | 2348 | CREB binding protein |
| CSPP1 | 26193 | centrosome and spindle pole associated protein 1 |
| CTBP1 | 2494 | C-terminal binding protein 1 |
| CUL4B | 2555 | cullin 4B |
| CWC27 | 10664 | CWC27 spliceosome associated protein homolog |
| D2HGDH | 28358 | D-2-hydroxyglutarate dehydrogenase |
| DACT1 | 17748 | dishevelled binding antagonist of beta catenin 1 |
| DAG1 | 2666 | dystroglycan 1 |
| DDX59 | 25360 | DEAD-box helicase 59 |
| DEAF1 | 14677 | DEAF1 transcription factor |
| DHCR24 | 2859 | 24-dehydrocholesterol reductase |
| DHCR7 | 2860 | 7-dehydrocholesterol reductase |
| DICER1 | 17098 | dicer 1 ribonuclease III |
| DIS3L2 | 28648 | DIS3 like 3'-5' exoribonuclease 2 |

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| DLL3 | 2909 | delta like canonical Notch ligand 3 |
| DMRT3 | 13909 | doublesex and mab-3 related transcription factor 3 |
| DNMT3A | 2978 | DNA methyltransferase 3 alpha |
| DNMT3B | 2979 | DNA methyltransferase 3 beta |
| DOCK6 | 19189 | dedicator of cytokinesis 6 |
| DPYD | 3012 | dihydropyrimidine dehydrogenase |
| DVL1 | 3084 | dishevelled segment polarity protein 1 |
| DVL3 | 3087 | dishevelled segment polarity protein 3 |
| DYNC2H1 | 2962 | dynein cytoplasmic 2 heavy chain 1 |
| DYNC2LI1 | 24595 | dynein cytoplasmic 2 light intermediate chain 1 |
| EBP | 3133 | EBP cholesterol delta-isomerase |
| EED | 3188 | embryonic ectoderm development |
| EFNB1 | 3226 | ephrin B1 |
| EFTUD2 | 30858 | elongation factor Tu GTP binding domain containing 2 |
| EIF2B1 | 3257 | eukaryotic translation initiation factor 2B subunit alpha |
| EIF2B2 | 3258 | eukaryotic translation initiation factor 2B subunit beta |
| EIF2B3 | 3259 | eukaryotic translation initiation factor 2B subunit gamma |
| EIF2B4 | 3260 | eukaryotic translation initiation factor 2B subunit delta |
| EIF2B5 | 3261 | eukaryotic translation initiation factor 2B subunit epsilon |
| EML1 | 3330 | EMAP like 1 |
| EP300 | 3373 | E1A binding protein p300 |
| ERF | 3444 | ETS2 repressor factor |
| ERMARD | 21056 | ER membrane associated RNA degradation |
| ETFA | 3481 | electron transfer flavoprotein subunit alpha |
| ETFB | 3482 | electron transfer flavoprotein subunit beta |
| ETFDH | 3483 | electron transfer flavoprotein dehydrogenase |
| EVC | 3497 | EvC ciliary complex subunit 1 |
| EVC2 | 19747 | EvC ciliary complex subunit 2 |
| EXT2 | 3513 | exostosin glycosyltransferase 2 |
| EZH2 | 3527 | enhancer of zeste 2 polycomb repressive complex 2 subunit |
| FAM111A | 24725 | family with sequence similarity 111 member A |
| FANCB | 3583 | FA complementation group B |
| FBLN1 | 3600 | fibulin 1 |
| FBN1 | 3603 | fibrillin 1 |
| FGF10 | 3666 | fibroblast growth factor 10 |
| FGFR1 | 3688 | fibroblast growth factor receptor 1 |
| FGFR2 | 3689 | fibroblast growth factor receptor 2 |
| FGFR3 | 3690 | fibroblast growth factor receptor 3 |
| FGFRL1 | 3693 | fibroblast growth factor receptor like 1 |
| FH | 3700 | fumarate hydratase |
| FIBP | 3705 | FGF1 intracellular binding protein |
| FKRP | 17997 | fukutin related protein |
| FKTN | 3622 | fukutin |
| FLI1 | 3749 | Fli-1 proto-oncogene ETS transcription factor |
| FLII | 3750 | FLII actin remodeling protein |
| FLNA | 3754 | filamin A |
| FMR1 | 3775 | fragile X mental retardation 1 |
| FOXP1 | 3823 | forkhead box P1 |
| FOXRED1 | 26927 | FAD dependent oxidoreductase domain containing 1 |
| FZD2 | 4040 | frizzled class receptor 2 |
| GABRD | 4084 | gamma-aminobutyric acid type A receptor delta subunit |

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| GATA4 | 4173 | GATA binding protein 4 |
| GATA6 | 4174 | GATA binding protein 6 |
| GCDH | 4189 | glutaryl-CoA dehydrogenase |
| GDF1 | 4214 | growth differentiation factor 1 |
| GDF5 | 4220 | growth differentiation factor 5 |
| GDF6 | 4221 | growth differentiation factor 6 |
| GFAP | 4235 | glial fibrillary acidic protein |
| GJA1 | 4274 | gap junction protein alpha 1 |
| GJB6 | 4288 | gap junction protein beta 6 |
| GLI1 | 4317 | GLI family zinc finger 1 |
| GLI2 | 4318 | GLI family zinc finger 2 |
| GLI3 | 4319 | GLI family zinc finger 3 |
| GNAI3 | 4387 | G protein subunit alpha i3 |
| GNAQ | 4390 | G protein subunit alpha q |
| GNAS | 4392 | GNAS complex locus |
| GP1BB | 4440 | glycoprotein Ib platelet subunit beta |
| GPC3 | 4451 | glypican 3 |
| GPC4 | 4452 | glypican 4 |
| GRIA3 | 4573 | glutamate ionotropic receptor AMPA type subunit 3 |
| GUSB | 4696 | glucuronidase beta |
| H19 | 4713 | H19 imprinted maternally expressed transcript |
| HDAC4 | 14063 | histone deacetylase 4 |
| HDAC6 | 14064 | histone deacetylase 6 |
| HELLS | 4861 | helicase lymphoid specific |
| HEPACAM | 26361 | hepatic and glial cell adhesion molecule |
| HERC1 | 4867 | HECT and RLD domain containing E3 ubiquitin protein ligase family member 1 |
| HES7 | 15977 | hes family bHLH transcription factor 7 |
| HESX1 | 4877 | HESX homeobox 1 |
| HEXB | 4879 | hexosaminidase subunit beta |
| HIRA | 4916 | histone cell cycle regulator |
| HIST1H1E | 4718 | histone cluster 1 H1 family member e |
| HNRNPK | 5044 | heterogeneous nuclear ribonucleoprotein K |
| HOXA13 | 5102 | homeobox A13 |
| HOXD13 | 5136 | homeobox D13 |
| HRAS | 5173 | HRas proto-oncogene GTPase |
| HSD17B4 | 5213 | hydroxysteroid 17-beta dehydrogenase 4 |
| HUWE1 | 30892 | HECT UBA and WWE domain containing 1 E3 ubiquitin protein ligase |
| HYLS1 | 26558 | HYLS1 centriolar and ciliogenesis associated |
| ICK | 21219 | intestinal cell kinase |
| IDS | 5389 | iduronate 2-sulfatase |
| IDUA | 5391 | iduronidase alpha-L- |
| IFT140 | 29077 | intraflagellar transport 140 |
| IFT172 | 30391 | intraflagellar transport 172 |
| IFT27 | 18626 | intraflagellar transport 27 |
| IFT43 | 29669 | intraflagellar transport 43 |
| IFT52 | 15901 | intraflagellar transport 52 |
| IFT74 | 21424 | intraflagellar transport 74 |
| IFT80 | 29262 | intraflagellar transport 80 |
| IFT81 | 14313 | intraflagellar transport 81 |
| IGBP1 | 5461 | immunoglobulin binding protein 1 |
| IGF2 | 5466 | insulin like growth factor 2 |

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| IHH | 5956 | Indian hedgehog signaling molecule |
| INPP5E | 21474 | inositol polyphosphate-5-phosphatase E |
| INPPL1 | 6080 | inositol polyphosphate phosphatase like 1 |
| INTU | 29239 | inturned planar cell polarity protein |
| IQSEC2 | 29059 | IQ motif and Sec7 domain 2 |
| ISPD | 37276 | isoprenoid synthase domain containing |
| ITCH | 13890 | itchy E3 ubiquitin protein ligase |
| JMJD1C | 12313 | jumonji domain containing 1C |
| KCNAB2 | 6229 | potassium voltage-gated channel subfamily A regulatory beta subunit 2 |
| KCNJ1 | 6255 | potassium voltage-gated channel subfamily J member 1 |
| KCNJ8 | 6269 | potassium voltage-gated channel subfamily J member 8 |
| KCNQ1 | 6294 | potassium voltage-gated channel subfamily Q member 1 |
| KCNQ1OT1 | 6295 | KCNQ1 opposite strand/antisense transcript 1 |
| KDM1A | 29079 | lysine demethylase 1A |
| KDM5C | 11114 | lysine demethylase 5C |
| KIAA0556 | 29068 | KIAA0556 |
| KIAA0586 | 19960 | KIAA0586 |
| KIAA0753 | 29110 | KIAA0753 |
| KIF22 | 6391 | kinesin family member 22 |
| KIF7 | 30497 | kinesin family member 7 |
| KLLN | 37212 | killin p53 regulated DNA replication inhibitor |
| KPTN | 6404 | kaptin actin binding protein |
| KRAS | 6407 | KRAS proto-oncogene GTPase |
| L1CAM | 6470 | L1 cell adhesion molecule |
| L2HGDH | 20499 | L-2-hydroxyglutarate dehydrogenase |
| LAMB1 | 6486 | laminin subunit beta 1 |
| LARGE1 | 6511 | LARGE xylosyl- and glucuronyltransferase 1 |
| LBR | 6518 | lamin B receptor |
| LETM1 | 6556 | leucine zipper and EF-hand containing transmembrane protein 1 |
| LFNG | 6560 | LFNG O-fucosylpeptide 3-beta-N-acetylglucosaminyltransferase |
| LHX4 | 21734 | LIM homeobox 4 |
| LMBR1 | 13243 | limb development membrane protein 1 |
| LRP2 | 6694 | LDL receptor related protein 2 |
| LRP4 | 6696 | LDL receptor related protein 4 |
| LRP5 | 6697 | LDL receptor related protein 5 |
| LZTFL1 | 6741 | leucine zipper transcription factor like 1 |
| MAB21L2 | 6758 | mab-21 like 2 |
| MAFB | 6408 | MAF bZIP transcription factor B |
| MAN2B1 | 6826 | mannosidase alpha class 2B member 1 |
| MAP2K1 | 6840 | mitogen-activated protein kinase kinase 1 |
| MAP2K2 | 6842 | mitogen-activated protein kinase kinase 2 |
| MAP3K1 | 6848 | mitogen-activated protein kinase kinase kinase 1 |
| MAPK10 | 6872 | mitogen-activated protein kinase 10 |
| MBTPS2 | 15455 | membrane bound transcription factor peptidase site 2 |
| MECP2 | 6990 | methyl-CpG binding protein 2 |
| MED12 | 11957 | mediator complex subunit 12 |
| MEGF8 | 3233 | multiple EGF like domains 8 |
| MESP2 | 29659 | mesoderm posterior bHLH transcription factor 2 |
| MGAT2 | 7045 | mannosyl (alpha-1 6-)-glycoprotein beta-1 2-N-acetylglucosaminyltransferase |
| MITF | 7105 | melanocyte inducing transcription factor |
| MKKS | 7108 | McKusick-Kaufman syndrome |

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| MKS1 | 7121 | Meckel syndrome type 1 |
| MLC1 | 17082 | megalencephalic leukoencephalopathy with subcortical cysts 1 |
| MOCS1 | 7190 | molybdenum cofactor synthesis 1 |
| MOCS2 | 7193 | molybdenum cofactor synthesis 2 |
| MPDZ | 7208 | multiple PDZ domain crumbs cell polarity complex component |
| MSX2 | 7392 | msh homeobox 2 |
| MTM1 | 7448 | myotubularin 1 |
| MTOR | 3942 | mechanistic target of rapamycin kinase |
| MYH8 | 7578 | myosin heavy chain 8 |
| NDUFA1 | 7683 | NADH:ubiquinone oxidoreductase subunit A1 |
| NDUFA11 | 20371 | NADH:ubiquinone oxidoreductase subunit A11 |
| NDUFAF1 | 18828 | NADH:ubiquinone oxidoreductase complex assembly factor 1 |
| NDUFAF2 | 28086 | NADH:ubiquinone oxidoreductase complex assembly factor 2 |
| NDUFAF3 | 29918 | NADH:ubiquinone oxidoreductase complex assembly factor 3 |
| NDUFAF4 | 21034 | NADH:ubiquinone oxidoreductase complex assembly factor 4 |
| NDUFAF5 | 15899 | NADH:ubiquinone oxidoreductase complex assembly factor 5 |
| NDUFB3 | 7698 | NADH:ubiquinone oxidoreductase subunit B3 |
| NDUFB9 | 7704 | NADH:ubiquinone oxidoreductase subunit B9 |
| NDUFS1 | 7707 | NADH:ubiquinone oxidoreductase core subunit S1 |
| NDUFS2 | 7708 | NADH:ubiquinone oxidoreductase core subunit S2 |
| NDUFS3 | 7710 | NADH:ubiquinone oxidoreductase core subunit S3 |
| NDUFS4 | 7711 | NADH:ubiquinone oxidoreductase subunit S4 |
| NDUFS6 | 7713 | NADH:ubiquinone oxidoreductase subunit S6 |
| NDUFV1 | 7716 | NADH:ubiquinone oxidoreductase core subunit V1 |
| NDUFV2 | 7717 | NADH:ubiquinone oxidoreductase core subunit V2 |
| NEK1 | 7744 | NIMA related kinase 1 |
| NEK9 | 18591 | NIMA related kinase 9 |
| NELFA | 12768 | negative elongation factor complex member A |
| NF1 | 7765 | neurofibromin 1 |
| NFIA | 7784 | nuclear factor I A |
| NFIX | 7788 | nuclear factor I X |
| NKX2-5 | 2488 | NK2 homeobox 5 |
| NKX2-6 | 32940 | NK2 homeobox 6 |
| NKX3-2 | 951 | NK3 homeobox 2 |
| NLRC4 | 16412 | NLR family CARD domain containing 4 |
| NLRP3 | 16400 | NLR family pyrin domain containing 3 |
| NOTCH2 | 7882 | notch 2 |
| NPHP1 | 7905 | nephrocystin 1 |
| NPHP3 | 7907 | nephrocystin 3 |
| NR0B1 | 7960 | nuclear receptor subfamily 0 group B member 1 |
| NR5A1 | 7983 | nuclear receptor subfamily 5 group A member 1 |
| NRAS | 7989 | NRAS proto-oncogene GTPase |
| NSD1 | 14234 | nuclear receptor binding SET domain protein 1 |
| NSD2 | 12766 | nuclear receptor binding SET domain protein 2 |
| NUBPL | 20278 | nucleotide binding protein like |
| NXN | 18008 | nucleoredoxin |
| OFD1 | 2567 | OFD1 centriole and centriolar satellite protein |
| OPHN1 | 8148 | oligophrenin 1 |
| OTX2 | 8522 | orthodenticle homeobox 2 |
| PALB2 | 26144 | partner and localizer of BRCA2 |
| PAX6 | 8620 | paired box 6 |

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| PDE6D | 8788 | phosphodiesterase 6D |
| PDSS1 | 17759 | decaprenyl diphosphate synthase subunit 1 |
| PEX1 | 8850 | peroxisomal biogenesis factor 1 |
| PEX10 | 8851 | peroxisomal biogenesis factor 10 |
| PEX11B | 8853 | peroxisomal biogenesis factor 11 beta |
| PEX12 | 8854 | peroxisomal biogenesis factor 12 |
| PEX13 | 8855 | peroxisomal biogenesis factor 13 |
| PEX14 | 8856 | peroxisomal biogenesis factor 14 |
| PEX16 | 8857 | peroxisomal biogenesis factor 16 |
| PEX19 | 9713 | peroxisomal biogenesis factor 19 |
| PEX2 | 9717 | peroxisomal biogenesis factor 2 |
| PEX26 | 22965 | peroxisomal biogenesis factor 26 |
| PEX3 | 8858 | peroxisomal biogenesis factor 3 |
| PEX5 | 9719 | peroxisomal biogenesis factor 5 |
| PEX6 | 8859 | peroxisomal biogenesis factor 6 |
| PHF21A | 24156 | PHD finger protein 21A |
| PHF6 | 18145 | PHD finger protein 6 |
| PHF8 | 20672 | PHD finger protein 8 |
| PIBF1 | 23352 | progesterone immunomodulatory binding factor 1 |
| PIGA | 8957 | phosphatidylinositol glycan anchor biosynthesis class A |
| PIGN | 8967 | phosphatidylinositol glycan anchor biosynthesis class N |
| PIGT | 14938 | phosphatidylinositol glycan anchor biosynthesis class T |
| PIK3CA | 8975 | phosphatidylinositol-4 5-bisphosphate 3-kinase catalytic subunit alpha |
| PIK3R2 | 8980 | phosphoinositide-3-kinase regulatory subunit 2 |
| PITX1 | 9004 | paired like homeodomain 1 |
| PLCB4 | 9059 | phospholipase C beta 4 |
| PLG | 9071 | plasminogen |
| PNPLA6 | 16268 | patatin like phospholipase domain containing 6 |
| POC1A | 24488 | POC1 centriolar protein A |
| POLE | 9177 | DNA polymerase epsilon catalytic subunit |
| POMGNT1 | 19139 | protein O-linked mannose N-acetylglucosaminyltransferase 1 (beta 1 2-) |
| POMGNT2 | 25902 | protein O-linked mannose N-acetylglucosaminyltransferase 2 (beta 1 4-) |
| POMK | 26267 | protein-O-mannose kinase |
| POMT1 | 9202 | protein O-mannosyltransferase 1 |
| POMT2 | 19743 | protein O-mannosyltransferase 2 |
| POP1 | 30129 | POP1 homolog ribonuclease P/MRP subunit |
| PORCN | 17652 | porcupine O-acyltransferase |
| POU1F1 | 9210 | POU class 1 homeobox 1 |
| POU6F2 | 21694 | POU class 6 homeobox 2 |
| PPP2R5D | 9312 | protein phosphatase 2 regulatory subunit B' delta |
| PRDM16 | 14000 | PR/SET domain 16 |
| PROP1 | 9455 | PROP paired-like homeobox 1 |
| PTCH1 | 9585 | patched 1 |
| PTCH2 | 9586 | patched 2 |
| PTDSS1 | 9587 | phosphatidylserine synthase 1 |
| PTEN | 9588 | phosphatase and tensin homolog |
| PTH1H | 9607 | parathyroid hormone like hormone |
| RAB23 | 14263 | RAB23 member RAS oncogene family |
| RAB39B | 16499 | RAB39B member RAS oncogene family |
| RAC1 | 9801 | Rac family small GTPase 1 |
| RAI1 | 9834 | retinoic acid induced 1 |

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| RBM10 | 9896 | RNA binding motif protein 10 |
| RERE | 9965 | arginine-glutamic acid dipeptide repeats |
| REST | 9966 | RE1 silencing transcription factor |
| RIPPLY2 | 21390 | rippy transcriptional repressor 2 |
| RIT1 | 10023 | Ras like without CAAX 1 |
| RNF125 | 21150 | ring finger protein 125 |
| RNF135 | 21158 | ring finger protein 135 |
| ROR2 | 10257 | receptor tyrosine kinase like orphan receptor 2 |
| RPGRIP1 | 13436 | RPGR interacting protein 1 |
| RPGRIP1L | 29168 | RPGRIP1 like |
| RREB1 | 10449 | ras responsive element binding protein 1 |
| RUNX2 | 10472 | runt related transcription factor 2 |
| SALL1 | 10524 | spalt like transcription factor 1 |
| SALL4 | 15924 | spalt like transcription factor 4 |
| SC5D | 10547 | sterol-C5-desaturase |
| SDCCAG8 | 10671 | serologically defined colon cancer antigen 8 |
| SDHB | 10681 | succinate dehydrogenase complex iron sulfur subunit B |
| SDHC | 10682 | succinate dehydrogenase complex subunit C |
| SDHD | 10683 | succinate dehydrogenase complex subunit D |
| SEC23A | 10701 | Sec23 homolog A coat complex II component |
| SEC23B | 10702 | Sec23 homolog B coat complex II component |
| SEC24C | 10705 | SEC24 homolog C COPII coat complex component |
| SEC24D | 10706 | SEC24 homolog D COPII coat complex component |
| SEMA3E | 10727 | semaphorin 3E |
| SERPINH1 | 1546 | serpin family H member 1 |
| SETBP1 | 15573 | SET binding protein 1 |
| SETD2 | 18420 | SET domain containing 2 |
| SH2B1 | 30417 | SH2B adaptor protein 1 |
| SHANK3 | 14294 | SH3 and multiple ankyrin repeat domains 3 |
| SHH | 10848 | sonic hedgehog signaling molecule |
| SHOC2 | 15454 | SHOC2 leucine rich repeat scaffold protein |
| SHPK | 1492 | sedoheptulokinase |
| SIM1 | 10882 | SIM bHLH transcription factor 1 |
| SKI | 10896 | SKI proto-oncogene |
| SLC25A1 | 10979 | solute carrier family 25 member 1 |
| SLC26A2 | 10994 | solute carrier family 26 member 2 |
| SLC29A3 | 23096 | solute carrier family 29 member 3 |
| SLC2A10 | 13444 | solute carrier family 2 member 10 |
| SLC35D1 | 20800 | solute carrier family 35 member D1 |
| SMARCB1 | 11103 | SWI/SNF related matrix associated actin dependent regulator of chromatin subfamily b member 1 |
| SMO | 11119 | smoothened frizzled class receptor |
| SMOC1 | 20318 | SPARC related modular calcium binding 1 |
| SNX10 | 14974 | sorting nexin 10 |
| SNX14 | 14977 | sorting nexin 14 |
| SOS1 | 11187 | SOS Ras/Rac guanine nucleotide exchange factor 1 |
| SOST | 13771 | sclerostin |
| SOX9 | 11204 | SRY-box 9 |
| SPINT2 | 11247 | serine peptidase inhibitor Kunitz type 2 |
| SPRED1 | 20249 | sprouty related EVH1 domain containing 1 |
| SRY | 11311 | sex determining region Y |
| STRADA | 30172 | STE20 related adaptor alpha |

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| SUFU | 16466 | SUFU negative regulator of hedgehog signaling |
| SUMF1 | 20376 | sulfatase modifying factor 1 |
| SUZ12 | 17101 | SUZ12 polycomb repressive complex 2 subunit |
| SYN1 | 11494 | synapsin I |
| TBCK | 28261 | TBC1 domain containing kinase |
| TBX1 | 11592 | T-box 1 |
| TBX15 | 11594 | T-box 15 |
| TBX3 | 11602 | T-box 3 |
| TBX6 | 11605 | T-box 6 |
| TCF12 | 11623 | transcription factor 12 |
| TCIRG1 | 11647 | T cell immune regulator 1 ATPase H ⁺ transporting V0 subunit a3 |
| TCTEX1D2 | 28482 | Tctex1 domain containing 2 |
| TCTN1 | 26113 | tectonic family member 1 |
| TCTN2 | 25774 | tectonic family member 2 |
| TCTN3 | 24519 | tectonic family member 3 |
| TFAP2A | 11742 | transcription factor AP-2 alpha |
| TFAP2B | 11743 | transcription factor AP-2 beta |
| TGFBR1 | 11772 | transforming growth factor beta receptor 1 |
| TGFBR2 | 11773 | transforming growth factor beta receptor 2 |
| THRA | 11796 | thyroid hormone receptor alpha |
| TIMMDC1 | 1321 | translocase of inner mitochondrial membrane domain containing 1 |
| TMCO1 | 18188 | transmembrane and coiled-coil domains 1 |
| TMEM107 | 28128 | transmembrane protein 107 |
| TMEM126B | 30883 | transmembrane protein 126B |
| TMEM138 | 26944 | transmembrane protein 138 |
| TMEM216 | 25018 | transmembrane protein 216 |
| TMEM231 | 37234 | transmembrane protein 231 |
| TMEM237 | 14432 | transmembrane protein 237 |
| TMEM67 | 28396 | transmembrane protein 67 |
| TNFRSF11A | 11908 | TNF receptor superfamily member 11a |
| TNFRSF11B | 11909 | TNF receptor superfamily member 11b |
| TNFSF11 | 11926 | TNF superfamily member 11 |
| TP53 | 11998 | tumor protein p53 |
| TRAF3IP1 | 17861 | TRAF3 interacting protein 1 |
| TRIM32 | 16380 | tripartite motif containing 32 |
| TRIM37 | 7523 | tripartite motif containing 37 |
| TRIP11 | 12305 | thyroid hormone receptor interactor 11 |
| TRIP13 | 12307 | thyroid hormone receptor interactor 13 |
| TTC21B | 25660 | tetratricopeptide repeat domain 21B |
| TTC8 | 20087 | tetratricopeptide repeat domain 8 |
| TWIST1 | 12428 | twist family bHLH transcription factor 1 |
| UBE2A | 12472 | ubiquitin conjugating enzyme E2 A |
| UBE3A | 12496 | ubiquitin protein ligase E3A |
| UFD1 | 12520 | ubiquitin recognition factor in ER associated degradation 1 |
| UPF3B | 20439 | UPF3B regulator of nonsense mediated mRNA decay |
| UQCC2 | 21237 | ubiquinol-cytochrome c reductase complex assembly factor 2 |
| USP9X | 12632 | ubiquitin specific peptidase 9 X-linked |
| VAMP7 | 11486 | vesicle associated membrane protein 7 |
| WASHC5 | 28984 | WASH complex subunit 5 |
| WDPCP | 28027 | WD repeat containing planar cell polarity effector |
| WDR19 | 18340 | WD repeat domain 19 |

| | | |
|--------|-------|--|
| WDR34 | 28296 | WD repeat domain 34 |
| WDR35 | 29250 | WD repeat domain 35 |
| WDR60 | 21862 | WD repeat domain 60 |
| WNT5A | 12784 | Wnt family member 5A |
| WNT7A | 12786 | Wnt family member 7A |
| WT1 | 12796 | Wilms tumor 1 |
| WWOX | 12799 | WW domain containing oxidoreductase |
| XYLT1 | 15516 | xylosyltransferase 1 |
| YME1L1 | 12843 | YME1 like 1 ATPase |
| ZBTB20 | 13503 | zinc finger and BTB domain containing 20 |
| ZBTB24 | 21143 | zinc finger and BTB domain containing 24 |
| ZBTB42 | 32550 | zinc finger and BTB domain containing 42 |
| ZDHHC9 | 18475 | zinc finger DHHC-type containing 9 |
| ZFPM2 | 16700 | zinc finger protein FOG family member 2 |
| ZIC1 | 12872 | Zic family member 1 |
| ZIC3 | 12874 | Zic family member 3 |
| ZNF141 | 12926 | zinc finger protein 141 |
| ZNF423 | 16762 | zinc finger protein 423 |
| ZNF469 | 23216 | zinc finger protein 469 |
| ZSWIM6 | 29316 | zinc finger SWIM-type containing 6 |

Supplementary Table 2; Sequence variants correlating to patient phenotype

| HGNC symbol, sequence variant |
|--|
| DICER1, NM_030621 (ENST00000393063) c.4031C>T p.Ser1344Leu, heterozygous |
| GLI2, NM_005270 (ENST00000361492) c.1760C>T p.Thr587Met, heterozygous |
| LRP5, NM_002335 (ENST00000294304) c.3552C>T p.=, heterozygous |
| LRP5, NM_002335 (ENST00000294304) c.4466C>T p.Thr1489Met, heterozygous |