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Date: 16.03.2023

Report to:	<b>Order Number</b>	<b>20 2009 1234</b>
Requesting Physician Name	<b>Born</b>	DD/MM/YYYY
Address	<b>Sex</b>	
Contact Information	<b>Date test requested:</b>	27.02.2023
	<b>Sample collected:</b>	27.02.2023
	<b>Sample / Specimen:</b>	DNA from EDTA blood

**Order:** molecular genetic analysis of Intellectual disability\_3.2

**Additional Information /patient phenotype:** Global developmental delay, no speech, severe learning disability, epilepsy, unsteady gait. ID panel requested.

## **RESULT SUMMARY:**

### **NEGATIVE**

**No pathogenic/likely pathogenic or variant of uncertain significance (SNV) was identified.**

## **Conclusion**

In the examined genes, no pathogenic variant, likely pathogenic variant or variant of unclear significance could be detected which, according to the current state of knowledge, is or could be causally related to the present clinical symptoms of the patient.

## **Recommended action**

Reanalysis of the data may be performed

- with a focus on other genes in the exome in case of occurrence of new symptoms in the patient.
- after few years based on updated scientific knowledge.

Report released by

John Doe 16.03.2023

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**TEST METHODOLOGY**

Sequencing	Enrichment	SNV and CNV Data analysis	data evaluation	Reference genome
Next Generation Sequencing (Illumina)	Twist Human Core Exome plus RefSeq Spikeln	Illumina Dragen Bio-IT Platform VarSeq by GoldenHelix	VarSeq by GoldenHelix	hg38, NCBI GR38
Quality criteria	SNV detection sensitivity	Classification of variants	in silico algorithms	Databases
>30 (precision >99.9%) in min. 75% of bases	99.92 - 99.93 %; confirmation of reported SNV with Sanger sequencing, data analysis with SeqPilot	Richards et al. 2015, Genet Med 17:405; Ellard et al. "ACGS Best Practice Guidelines for Variant Classification 2020"	MaxEntScan, SpliceSiteFinder-like, REVEL	HGMD Professional release, ClinVar, gnomAD

**PERCENTAGE OF SEQUENCED BASES WITH COVERAGE >20X**

96.951%

**ANALYZED GENES**

RIT1(NM\_006912.5), BCS1L(NM\_001079866.1), ROR2(NM\_004560.3), RORA(NM\_134261.2), RPIA(NM\_144563.2), RPL10(NM\_001256577.2), CNM2(NM\_017649.4), RPS6KA3(NM\_004586.2), MSMO1(NM\_006745.4), SC5D(NM\_006918.4), BLM(NM\_000057.3), SCN1A(NM\_001165963.1), SCN2A(NM\_001040142.1), SCN3A(NM\_006922.3), SCN8A(NM\_001330260.1), SCO1(NM\_004589.3), SCO2(NM\_005138.2), AIMP1(NM\_001142416.1), CWC27(NM\_005869.3), SDCCAG8(NM\_006642.3), SDHA(NM\_004168.3), BMP4(NM\_001202.5), SET(NM\_001122821.1), SGPL1(NM\_003901.3), SGSH(NM\_000199.3), SHH(NM\_000193.3), ST3GAL3(NM\_006279.3), ST3GAL5(NM\_003896.3), STIL(NM\_001048166.1), SIX3(NM\_005413.3), SKI(NM\_003036.3), SLC12A6(NM\_133647.1), SLC16A2(NM\_006517.4), SLC17A5(NM\_012434.4), SLC1A2(NM\_004171.3), SLC1A4(NM\_003038.4), BRAF(NM\_004333.4), SLC25A1(NM\_005984.4), SLC25A12(NM\_003705.4), SLC25A15(NM\_014252.3), SLC2A1(NM\_006516.2), SLC35A1(NM\_006416.4), SLC35A2(NM\_001042498.2), SLC4A4(NM\_001098484.2), SLC6A1(NM\_003042.3), SLC6A3(NM\_001044.4), SLC6A8(NM\_005629.3), SLC6A9(NM\_201649.3), SLC9A6(NM\_001042537.1), SMARCA2(NM\_003070.4), SMARCA4(NM\_001128849.1), SMARCB1(NM\_003073.4), SMARCC2(NM\_003075.4), SMARCD1(NM\_003076.4), SMARCE1(NM\_003079.4), ARID1A(NM\_006015.4), SMC1A(NM\_006306.3), KDM5C(NM\_004187.3), SMPD1(NM\_000543.4), SMS(NM\_004595.4), SNAP25(NM\_130811.3), SNAP29(NM\_004782.3), SIK1(NM\_173354.4), SNRPB(NM\_003091.3), SON(NM\_138927.2), SOS1(NM\_005633.3), SOS2(NM\_006939.3), SOX10(NM\_006941.3), SOX11(NM\_003108.3), SOX2(NM\_003106.3), SOX3(NM\_005634.2), SOX4(NM\_003107.2), SOX5(NM\_006940.5), SOX9(NM\_000346.3), BTBD9(NM\_000060.4), SPG11(NM\_025137.3), ATL1(NM\_015915.4), SPR(NM\_003124.4), SPTAN1(NM\_001130438.2), SPTBN2(NM\_006946.2), SSR4(NM\_001204526.1), STAG1(NM\_005862.2), STAG2(NM\_001042750.1), PLK4(NM\_014264.4), BRSK2(NM\_001256627.1), CDKL5(NM\_003159.2), STXPB1(NM\_003165.3), SUCLG1(NM\_003849.3), SUOX(NM\_001032386.1), SURF1(NM\_003172.3), BUB1B(NM\_001211.5), SYN1(NM\_133499.2), SYNGAP1(NM\_006772.2), SYNJ1(NM\_003895.3), SYP(NM\_003179.2), SYT1(NM\_005639.2), TAF1(NM\_001286074.1), TAF6(NM\_005641.3), BRF1(NM\_001519.3), TAT(NM\_000353.2), TAZ(NM\_000116.4), TBCD(NM\_005993.4), TBCE(NM\_003193.4), TBR1(NM\_006593.3), TCF20(NM\_005650.3), TCF4(NM\_001083962.1), TCN2(NM\_000355.3), TGIF1(NM\_003244.3), TH(NM\_199292.2), THRA(NM\_199334.3), ACO2(NM\_001098.2), NKX2-1(NM\_001079668.2), C12orf4(NM\_020374.3), TLK2(NM\_001284333.1), TSPAN7(NM\_004615.3), ACOX1(NM\_004035.6), MED12(NM\_005120.2), TREX1(NM\_033629.4), TRIO(NM\_007118.3), TRIP12(NM\_004238.2), TRRAP(NM\_001244580.1), TSC1(NM\_000368.4), TSC2(NM\_000548.4), TSFM(NM\_005726.5), CEP41(NM\_018718.2), TSHB(NM\_000549.4), TUBB2A(NM\_001069.2), TUBG1(NM\_001070.4), TWIST1(NM\_000474.3), UBE2A(NM\_003336.3), UBE3A(NM\_130838.1), UBTF(NM\_014233.3), UMPS(NM\_000373.3), USP9X(NM\_001039590.2), KDM6A(NM\_021140.3), VAMP1(NM\_014231.4), VAMP2(NM\_014232.2), VARS1(NM\_006295.2), VLDLR(NM\_003383.4), VRK1(NM\_003384.2), WARS2(NM\_015836.3), WASF1(NM\_003931.2), NSD2(NM\_001042424.2), WWOX(NM\_016373.3), XRCC4(NM\_022406.3), YWHAG(NM\_012479.3), YY1(NM\_003403.4), ZIC2(NM\_007129.3), ZNF142(NM\_001105537.2), PCGF2(NM\_007144.2), KAT6A(NM\_006766.4), ZBTB18(NM\_205768.2), SCAPER(NM\_020843.2), ZNF711(NM\_001330574.1), ACTB(NM\_001101.3), ARL6(NM\_177976.3), BCL11A(NM\_022893.3), BCL11B(NM\_138576.3), FTSJ1(NM\_012280.3), HDAC8(NM\_018486.2), MCOLN1(NM\_020533.2), NSDHL(NM\_015922.2), PRUNE1(NM\_021222.2), RLIM(NM\_183353.2), UBE3B(NM\_130466.3), SAMD9(NM\_017654.3), ZBTB20(NM\_001164342.2), RXYLT1(NM\_014254.2), ATP8A2(NM\_016529.5), ERLIN2(NM\_007175.6), BRD4(NM\_058243.2), FBXO11(NM\_001190274.1), FBXL3(NM\_012158.2), FBXL4(NM\_001278716.1), FBXW11(NM\_012300.2),

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CAMK2B(NM\_001220.4), DEAF1(NM\_021008.3), COG5(NM\_006348.3), GTPBP3(NM\_032620.3), ZEB2(NM\_014795.3), CAR1(NM\_001014437.2), PIGT(NM\_015937.5), PPP1R15B(NM\_032833.4), CASK(NM\_003688.3), SNX14(NM\_153816.5), MIRC5(NM\_001195518.1), CBL(NM\_005188.3), SHOC2(NM\_007373.3), MBTPS2(NM\_015884.3), ANKH(NM\_054027.4), CBS(NM\_000071.2), MBOAT7(NM\_024298.4), TSEN34(NM\_024075.4), PUS1(NM\_025215.5), JAM3(NM\_032801.4), SETBP1(NM\_015559.2), TRIM8(NM\_030912.2), KLHL7(NM\_001031710.2), PHIP(NM\_017934.6), LRPPRC(NM\_133259.3), ADNP(NM\_015339.4), PIGU(NM\_080476.4), CCND2(NM\_001759.3), BSCL2(NM\_032667.6), ARFGEF2(NM\_006420.2), RTEL1(NM\_001283009.1), PLCB1(NM\_015192.3), SAMHD1(NM\_015474.3), TOE1(NM\_025077.3), ACTL6B(NM\_016188.4), MPLKIP(NM\_138701.3), PCNT(NM\_006031.5), TBC1D20(NM\_144628.3), SLC19A3(NM\_025243.3), PNPLA6(NM\_001166111.1), NFU1(NM\_001002755.2), TRIM32(NM\_012210.3), ZMIZ1(NM\_020338.3), RAB39B(NM\_171998.3), MRPS34(NM\_023936.1), BCAP31(NM\_001139441.1), 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ALKBH8(NM\_001301010.1), MMADHC(NM\_015702.2), AHDC1(NM\_001029882.3), CTSD(NM\_001909.4), DDX59(NM\_001031725.5), ARL13B(NM\_001174150.1), TANGO2(NM\_152906.6), PUS3(NM\_00131307.3), DDX2(NM\_018122.4), CUL4B(NM\_003588.3), PRMT7(NM\_019023.2), SETD5(NM\_001080517.2), ATAD3A(NM\_001170535.1), CUX1(NM\_001202543.1), VPS53(NM\_001128159.2), CWF19L1(NM\_018294.5), TBC1D23(NM\_001199198.2), OFD1(NM\_003611.2), RNASEH2B(NM\_024570.3), ADK(NM\_001123.3), PGAP1(NM\_024989.3), TCTN2(NM\_024809.4), DCAF17(NM\_025000.3), CYC1(NM\_001916.4), CPLANE1(NM\_023073.3), SRD5A3(NM\_024592.4), POMGNT2(NM\_032806.5), WDR73(NM\_032856.3), TRMT1(NM\_017722.3), PIGG(NM\_001127178.2), NSUN2(NM\_017755.5), TTC19(NM\_017775.3), PIGV(NM\_017837.3), PUS7(NM\_019042.4), TMEM70(NM\_017866.5), CTC1(NM\_025099.5), CSPP1(NM\_024790.6), FAR1(NM\_032228.5), TTI2(NM\_001102401.2), BBS10(NM\_024685.3), HEPACAM(NM\_152722.4), PTCHD1(NM\_173495.2), UROC1(NM\_144639.2), HGSNAT(NM\_152419.2), HYLS1(NM\_145014.2), UNC80(NM\_032504.1), WDR81(NM\_001163809.1), BBS12(NM\_152618.2), DAG1(NM\_004393.5), DARS1(NM\_001349.3), C12orf65(NM\_152269.4), AMER1(NM\_152424.3), CKAP2L(NM\_152515.4), TMTC3(NM\_181783.3), ERCC6L2(NM\_020207.4), FOXRED1(NM\_017547.3), KIAA1109(NM\_015312.3), DBT(NM\_001918.3), METTL23(NM\_001080510.4), DCX(NM\_178151.2), DDC(NM\_000790.3), ESCO2(NM\_001017420.2), DDOST(NM\_005216.4), IBA57(NM\_001010867.3), DDX11(NM\_030653.3), DDX3X(NM\_001356.4), DDX6(NM\_004397.5), TSEN54(NM\_207346.2), SLC6A19(NM\_001003841.2), WDPCP(NM\_015910.5), TBCK(NM\_001163435.2), D2HGDH(NM\_152783.4), THOC6(NM\_024339.4), TMEM67(NM\_153704.5), TRMT10A(NM\_152292.4), TSEN2(NM\_025265.3), MFSD8(NM\_152778.2), GLIS3(NM\_152629.3), DHCR24(NM\_014762.3), B3GALNT2(NM\_152490.4), DHCR7(NM\_001360.2), DHFR(NM\_000791.3), DIS3L2(NM\_152383.4), DHPS(NM\_001930.3), CYB5R3(NM\_000398.6), DIAPH1(NM\_005219.4), MED25(NM\_030973.3), P4HTM(NM\_177939.2), NIPBL(NM\_133433.3), DKC1(NM\_001363.4), DNAJC12(NM\_021800.2), CCDC22(NM\_014008.4), WDR45(NM\_007075.3), EMC1(NM\_015047.2), DLD(NM\_000108.4), TMEM94(NM\_014738.5), FRMPD4(NM\_014728.3), DLG3(NM\_021120.3), CEP290(NM\_025114.3), SPECC1L(NM\_015330.4), DLG4(NM\_001365.4), SZT2(NM\_015284.3), IQSEC2(NM\_001111125.2), KDM1A(NM\_001009999.2), CEP135(NM\_025009.4), TELO2(NM\_016111.3), ADSL(NM\_000026.3), DDHD2(NM\_015214.2), RPRGRI1L(NM\_015272.4), SETD1B(NM\_015048.1), TBC1D24(NM\_001199107.1), SVBP(NM\_199342.3), PRR12(NM\_020719.2), CC2D2A(NM\_001080522.2), TAOK1(NM\_020791.2), DMD(NM\_004006.2), CEP152(NM\_001194998.1), ZSWIM6(NM\_020928.1), EPG5(NM\_020964.2), ASXL3(NM\_030632.2), DMXL2(NM\_001174116.1), CCBE1(NM\_133459.3), NEXMIF(NM\_001008537.2), ROGD1(NM\_024589.2), GPSM2(NM\_013296.4), KIDINS220(NM\_020738.2), C12orf57(NM\_138425.3), TBL1XR1(NM\_024665.5), ARV1(NM\_022786.2), DYNC1H1(NM\_001376.4), MTFMT(NM\_139242.3), GNPTAB(NM\_024312.4), DNMI1(NM\_004408.3), DNMT3A(NM\_175629.2), DNMT3B(NM\_006892.3), DCPS(NM\_014026.4), HECW2(NM\_020760.2), NFASC(NM\_015090.3), NKAP(NM\_024528.3), DOCK3(NM\_004947.4), CSDE1(NM\_001007553.2), COASY(NM\_025233.6), DPAGT1(NM\_001382.3), BBS9(NM\_198428.2), DPH1(NM\_001383.4), PACS1(NM\_018026.3), DPM1(NM\_001317034.1), POLR3A(NM\_007055.3), DPP6(NM\_130797.3), DPYD(NM\_000110.3), STRADA(NM\_001003787.2), ATP13A2(NM\_022089.3), RFT1(NM\_052859.3), CC2D1A(NM\_017721.4), TUSC3(NM\_006765.3), PYCR2(NM\_013328.3), ATN1(NM\_001940.3), POLR3B(NM\_018082.5), IFT172(NM\_015662.2), KIF7(NM\_198525.2), SLC46A1(NM\_080669.5), DNAJC19(NM\_145261.3), PPP1R21(NM\_001135629.2), SEPSECS(NM\_016955.3), STRA6(NM\_022369.3), CLPB(NM\_001258392.2), TMX2(NM\_015959.3), TMEM165(NM\_018475.4), TRAPPC9(NM\_005879.2), GATAD2B(NM\_020699.3), NAA15(NM\_057175.3), CEP57(NM\_014679.4), TUBB2B(NM\_178012.4), TRAPPC9(NM\_001160372.3), EFTUD2(NM\_004247.3), ALG13(NM\_001099922.2), HUWE1(NM\_031407.6), DYRK1A(NM\_001396.4), LINS1(NM\_001040616.2), EBP(NM\_006579.2), SLC6A17(NM\_001010898.3), WDR37(NM\_014023.3), AFG3L2(NM\_006796.2), AGA(NM\_000027.3), EED(NM\_003797.4), EEF1A2(NM\_001958.3), ALG11(NM\_001004127.2), EIF2AK3(NM\_004836.6), EIF2S3(NM\_001415.3), AGPS(NM\_003659.3), EIF3F(NM\_003754.2), SMPD4(NM\_017951.4), EML1(NM\_004434.2), EMX2(NM\_004098.3), EP300(NM\_001429.3), SDHAF1(NM\_001042631.2), INPP5K(NM\_016532.3), ERCC1(NM\_202001.2), ERCC2(NM\_000400.3), ERCC3(NM\_000122.1), ERCC5(NM\_000123.3), ERCC6(NM\_000124.3), ERCC8(NM\_000082.3), ETFA(NM\_000126.3), ERCC(NM\_001985.2), ETFDH(NM\_004453.3), EXTL3(NM\_001440.3), EZH2(NM\_004456.4), ACSL4(NM\_004458.2), BPTF(NM\_182641.3), FKTN(NM\_006731.2), FGD1(NM\_004463.2), FGF12(NM\_021032.4), FH(NM\_000143.3), CRPPA(NM\_001101426.3), FLNA(NM\_001456.3), FMR1(NM\_002024.5), AFF2(NM\_002025.3), FOLR1(NM\_016725.2), FOXG1(NM\_005249.4), FOXP1(NM\_032682.5), AKT3(NM\_005465.4), MTOR(NM\_004958.3), FTCD(NM\_006657.2), FUCA1(NM\_000147.4), FUT8(NM\_178155.2), ALDH3A2(NM\_000382.2), ALDH4A1(NM\_003748.3), GABRA1(NM\_000806.5), GABRA2(NM\_001330690.1), GABRA5(NM\_000810.3), ALDH5A1(NM\_001080.3), GABRB2(NM\_021911.2), GABRB3(NM\_000814.5), GABRG2(NM\_000816.3), GALC(NM\_001201402.1), GALE(NM\_000403.3), B4GALNT1(NM\_001478.4), GALT(NM\_000155.3), GAMT(NM\_000156.5), GATM(NM\_001482.2), GCDH(NM\_000159.3),

GCH1(NM\_000161.2), GDI1(NM\_001493.2), GFAP(NM\_002055.4), GFER(NM\_005262.2), ALMS1(NM\_015120.4), GK(NM\_000167.5), GLB1(NM\_000404.3), GLDC(NM\_000170.2), GLI2(NM\_005270.4), GLI3(NM\_000168.5), GLUL(NM\_002065.6), GM2A(NM\_000405.4), GNAI1(NM\_002069.5), GNAO1(NM\_020988.2), GNAS(NM\_080425.3), GNB1(NM\_002074.4), GNB5(NM\_016194.3), GNPAT(NM\_014236.3), GNS(NM\_002076.3), GPAA1(NM\_003801.3), GPC3(NM\_004484.3), GABBR2(NM\_005458.7), ADGRG1(NM\_005682.6), GRIA2(NM\_000826.3), GRIA3(NM\_007325.4), GRIA4(NM\_000829.3), GRID2(NM\_001510.3), GRIK2(NM\_021956.4), GRIN1(NM\_007327.3), GRIN2A(NM\_000833.4), GRIN2B(NM\_000834.3), GRIN2D(NM\_000836.2), GRM1(NM\_001278064.1), GTPBP2(NM\_019096.4), AMPD2(NM\_004037.7), GUSB(NM\_000181.3), H1-4(NM\_005321.2), AMT(NM\_000481.3), HADH(NM\_005327.4), HSD17B10(NM\_004493.2), HADHA(NM\_000182.4), HCCS(NM\_005333.4), HCFC1(NM\_005334.2), HCN1(NM\_021072.3), HERC1(NM\_003922.3), HESX1(NM\_003865.2), HEXA(NM\_000520.5), HEXB(NM\_000521.3), HIBCH(NM\_014362.3), HIVEP2(NM\_006734.3), HK1(NM\_033500.2), HLCS(NM\_000411.6), HMGCL(NM\_000191.2), HNRNP2(NM\_019597.4), HNRNPK(NM\_002140.4), HNRNPR(NM\_005826.4), HNRNPU(NM\_031844.2), HOXA1(NM\_005522.4), HPD(NM\_002150.2), HPR1(NM\_000194.2), HRAS(NM\_005343.3), HSD17B4(NM\_000414.3), HSPD1(NM\_002156.4), IARS1(NM\_002161.5), IDH2(NM\_002168.3), IDS(NM\_000202.7), IDUA(NM\_000203.4), IGF1(NM\_000618.4), IGF1R(NM\_000875.4), AP1S1(NM\_001283.3), AP1S2(NM\_003916.4), AP2M1(NM\_004068.3), AP3B1(NM\_003664.4), AP3B2(NM\_001278512.1), AP4B1(NM\_006594.4), AP4E1(NM\_007347.4), AP4M1(NM\_004722.3), AP4S1(NM\_007077.4), IKBKG(NM\_003639.4), IL1RAPL1(NM\_014271.3), ABCC9(NM\_005691.3), ABCD1(NM\_000033.3), ITPA(NM\_033453.3), ITPR1(NM\_001168272.1), IVD(NM\_002225.3), KARS1(NM\_001130089.1), KCNA2(NM\_001204269.1), KCNB1(NM\_004975.3), KCNC1(NM\_001112741.1), KCNC3(NM\_004977.2), KCNH1(NM\_172362.2), KCNJ10(NM\_002241.4), KCNJ11(NM\_000525.3), KCNJ6(NM\_002240.4), KCNK9(NM\_001282534.1), KCNQ2(NM\_172107.3), KCNQ3(NM\_004519.3), KCNQ5(NM\_001160133.1), KIF2A(NM\_001098511.2), KIF5A(NM\_004984.2), KIF5C(NM\_004522.2), KIF11(NM\_004523.3), KPTN(NM\_007059.3), KRAS(NM\_004985.4), L1CAM(NM\_000425.4), LAMA1(NM\_005559.3), LAMA2(NM\_000426.3), LAMB1(NM\_002291.2), ARCN1(NM\_001655.4), LAMC3(NM\_006059.3), LAMP2(NM\_002294.2), LARGE1(NM\_004737.5), COG1(NM\_018714.2), LIG4(NM\_002312.3), ARG1(NM\_000045.3), LRP2(NM\_004525.2), LZTR1(NM\_006767.3), MAB21L1(NM\_005584.4), MAB21L2(NM\_006439.4), SMAD4(NM\_005359.5), MAF(NM\_005360.4), ABCD4(NM\_005050.3), MAGEL2(NM\_019066.4), MAN1B1(NM\_016219.4), MAN2B1(NM\_000528.3), MANBA(NM\_005908.3), MAOA(NM\_000240.3), MAP2K1(NM\_002755.3), MAP2K2(NM\_030662.3), MAPK8IP3(NM\_001318852.1), MASP1(NM\_139125.3), MAT1A(NM\_000429.2), MCCC1(NM\_020166.4), MCCC2(NM\_022132.4), MCM3AP(NM\_003906.4), MCPH1(NM\_024596.4), MDH2(NM\_005918.3), MECP2(NM\_004992.3), MEF2C(NM\_002397.4), MEIS2(NM\_170674.4), MGAT2(NM\_002408.3), MGP(NM\_000900.4), MID1(NM\_000381.3), MKKS(NM\_170784.2), MKS1(NM\_017777.3), ARSA(NM\_000487.5), KMT2A(NM\_001197104.1), KMT2D(NM\_003482.3), ARSB(NM\_000046.3), MLYCD(NM\_012213.2), ARSL(NM\_000047.2), MOCS1(NM\_005943.5), MOCS2(NM\_176806.3), MPDU1(NM\_004870.3), MPI(NM\_002435.2), ASAH1(NM\_177924.4), MSL3(NM\_078629.3), MTHFR(NM\_005957.4), ASL(NM\_000048.3), MTR(NM\_000254.2), MTRR(NM\_002454.2), MMUT(NM\_000255.3), ASNS(NM\_133436.3), MVK(NM\_000431.3), MYCN(NM\_005378.5), ASPA(NM\_000049.2), ASS1(NM\_000050.4), MYO5A(NM\_000259.3), MYT1L(NM\_015025.3), NAGA(NM\_000262.2), NAGLU(NM\_000263.3), NBEA(NM\_015678.4), NDP(NM\_000266.3), NDST1(NM\_001543.4), NDUFA1(NM\_004541.3), NDUFS1(NM\_005006.6), NDUFS4(NM\_002495.3), NDUFS7(NM\_024407.4), NDUFS8(NM\_002496.3), NDUFV1(NM\_007103.3), NEU1(NM\_000434.3), NF1(NM\_000267.3), NFIA(NM\_005595.4), NFIX(NM\_001271043.2), NHS(NM\_198270.3), NONO(NM\_007363.4), CNOT1(NM\_016284.4), CNOT2(NM\_014515.5), CNOT3(NM\_014516.3), NPC1(NM\_000271.4), NPHP1(NM\_001128178.1), ATIC(NM\_004044.6), ATM(NM\_000051.3), NR2F1(NM\_005654.5), NRAS(NM\_002524.4), ATP1A1(NM\_000701.7), NRXN1(NM\_001135659.2), NT5C2(NM\_012229.4), NTRK1(NM\_001012331.1), NTRK2(NM\_006180.4), OCLN(NM\_001205254.1), OCRL(NM\_000276.3), ODC1(NM\_002539.2), OGT(NM\_181672.2), OPA3(NM\_025136.3), OPHN1(NM\_002547.2), ATP6V1A(NM\_001690.3), OTC(NM\_000531.5), OTX2(NM\_172337.2), ATP6V1B2(NM\_001693.3), PAFAH1B1(NM\_000430.3), PAH(NM\_000277.1), PAK1(NM\_002576.4), PAK3(NM\_002578.4), PARN(NM\_002582.3), PAX6(NM\_000280.4), PAX8(NM\_003466.3), PBX1(NM\_002585.3), PC(NM\_001040716.1), PCCA(NM\_000282.3), PCCB(NM\_000532.4), PCDH12(NM\_016580.3), ATP7A(NM\_000052.6), CHMP1A(NM\_002768.4), PCYT2(NM\_002861.4), AIFM1(NM\_004208.3), ALDH7A1(NM\_001182.4), PDE4D(NM\_001104631.1), PDGFRB(NM\_002609.3), PDHA1(NM\_000284.3), ATR(NM\_001184.3), PEPD(NM\_000285.3), PEX1(NM\_000466.2), PEX10(NM\_153818.1), PEX11B(NM\_003846.2), PEX12(NM\_000286.2), PEX13(NM\_002618.3), PEX14(NM\_004565.2), PEX16(NM\_057174.2), PEX3(NM\_003630.2), PEX6(NM\_000287.3), ATRX(NM\_000489.4), PEX7(NM\_000288.3), KIF1A(NM\_004321.7), PGK1(NM\_000291.3), ACADM(NM\_000016.5), AUH(NM\_001698.2), PGM3(NM\_001199917.1), PHGDH(NM\_006623.3), PIGA(NM\_002641.3), PIGB(NM\_004855.4), PIGL(NM\_004278.3), PIGN(NM\_176787.4), PIK3CA(NM\_006218.3), PIK3R2(NM\_005027.3), ACADS(NM\_000017.3), PLA2G6(NM\_003560.2), PLAA(NM\_001031689.2), PLP1(NM\_000533.4), PMM2(NM\_000303.2), PMPCB(NM\_004279.2), PNKP(NM\_007254.3), POLA1(NM\_001330360.1), POLG(NM\_002693.2), POLR2A(NM\_000937.4), POMT1(NM\_007171.3), POU3F3(NM\_006236.2), CTSA(NM\_000308.3), PPM1D(NM\_003620.3), PPP1CB(NM\_002709.2), PPP2CA(NM\_002715.2), B4GALT7(NM\_0007255.2), PPP2R1A(NM\_014225.5), PPP2R5D(NM\_006245.3), PPP3CA(NM\_000944.4), PPT1(NM\_000310.3), PQBP1(NM\_005710.2), PRKD1(NM\_002742.2), PRODH(NM\_016335.4), PLPBP(NM\_007198.3), PRPS1(NM\_002764.3), PRSS12(NM\_003619.3), LONP1(NM\_004793.3), PSAP(NM\_002778.3), SLC33A1(NM\_004733.3), PSMD12(NM\_002816.4), PSPH(NM\_004577.3), PTCH1(NM\_000264.3), PTDSS1(NM\_014754.2), PTEN(NM\_000314.6), PTPN11(NM\_002834.4), BBS1(NM\_024649.4), BBS2(NM\_031885.3), PTS(NM\_000317.2), BBS4(NM\_033028.4), BBS5(NM\_152384.2), PURA(NM\_005859.4), PEX19(NM\_002857.3), PEX2(NM\_000318.2), PEX5(NM\_001131025.1), PYCR1(NM\_006907.3), ALDH18A1(NM\_002860.3), QARS1(NM\_005051.2), QDPR(NM\_000320.2), RAB11B(NM\_004218.3), RAC1(NM\_006908.4), RAC3(NM\_005052.2), RAD21(NM\_006265.2),

RAF1(NM\_002880.3), RAI1(NM\_030665.3), RALA(NM\_005402.3), BCKDHA(NM\_000709.3), RARB(NM\_001290216.2), BCKDHB(NM\_183050.3), RBBP8(NM\_002894.2), RBM10(NM\_005676.4), RELN(NM\_005045.3), DPF2(NM\_006268.4), RERE(NM\_001042681.1)

**LIST OF EXONS WITH COVERAGE <20X**

Chr.	Pos.	Gene	Exon	Transcript	Mean Coverage (Min/Max)
Chr1	1447644..1447858	ATAD3A	Exon 01	NM_001170535.1	3.47 (2/6)
Chr1	1470734..1470892	TMEM240	Exon 04	NM_001114748.1	56.40 (16/82)
Chr1	1475665..1475731	TMEM240	Exon 01	NM_001114748.1	13.88 (10/16)
Chr1	2160201..2161179	SKI	Exon 01	NM_003036.3	42.07 (0/81)
Chr1	2238011..2238209	SKI	Exon 07	NM_003036.3	34.44 (9/55)
Chr1	2343825..2343946	PEX10	Exon 01	NM_002617.3	3.51 (2/4)
Chr1	2343825..2343946	PEX10	Exon 01	NM_153818.1	3.51 (2/4)
Chr1	6845586..6845640	CAMTA1	Exon 01	NM_015215.3	17.56 (12/18)
Chr1	17338219..17338238	ATP13A2	Exon 01	NM_022089.3	5.60 (4/8)
Chr1	19228951..19229022	ALDH4A1	Exon 01	NM_003748.3	9.33 (1/22)
Chr1	23346085..23346445	KDM1A	Exon 01	NM_015013.3	25.29 (0/54)
Chr1	24194383..24194781	FUCA1	Exon 01	NM_000147.4	79.87 (13/128)
Chr1	27022890..27024036	ARID1A	Exon 01	NM_006015.4	22.38 (1/93)
Chr1	39875933..39880226	KIAA0754	Exon 01	NM_015038.1	123.27 (5/243)
Chr1	43424300..43424327	SLC2A1	Exon 01	NM_006516.2	6.36 (6/8)
Chr1	43919261..43919469	HYI	Exon 01	NM_001330526.1	9.85 (3/19)
Chr1	55352557..55352797	DHCR24	Exon 01	NM_014762.3	61.59 (19/94)
Chr1	63153893..63153940	DOCK7	Exon 01	NM_001271999.1	3.42 (2/4)
Chr1	63153893..63153940	DOCK7	Exon 01	NM_001272000.1	3.42 (2/4)
Chr1	63153893..63153940	DOCK7	Exon 01	NM_001272001.1	3.42 (2/4)
Chr1	63153893..63153940	DOCK7	Exon 01	NM_001272002.1	3.42 (2/4)
Chr1	63153893..63153940	DOCK7	Exon 01	NM_001330614.1	3.42 (2/4)
Chr1	63153893..63153940	DOCK7	Exon 01	NM_033407.3	3.42 (2/4)
Chr1	156830722..156830943	NTRK1	Exon 01	NM_001012331.1	52.54 (0/89)
Chr1	156830722..156830943	NTRK1	Exon 01	NM_002529.3	52.54 (0/89)
Chr1	202777225..202777438	KDM5B	Exon 01	NM_006618.4	38.46 (13/57)
Chr1	215741024..215741116	KCTD3	Exon 01	NM_016121.4	62.72 (17/94)
Chr1	228345455..228346784	GJC2	Exon 02	NM_020435.3	52.45 (0/131)
Chr1	228353513..228353863	IBA57	Exon 01	NM_001010867.3	18.17 (0/47)
Chr1	231126341..231126449	ARV1	Exon 04	NM_001346992.1	2.63 (2/3)

Chr1	235611980..235612082	<i>TBCE</i>	Exon 17	NM_001079515.2	25.90 (14/32)
Chr1	235611980..235612082	<i>TBCE</i>	Exon 18	NM_001287801.1	25.90 (14/32)
Chr1	235611980..235612082	<i>TBCE</i>	Exon 16	NM_001287802.1	25.90 (14/32)
Chr1	235611980..235612082	<i>TBCE</i>	Exon 17	NM_003193.4	25.90 (14/32)
Chr1	235667436..235667557	<i>B3GALNT2</i>	Exon 01	NM_152490.4	8.98 (1/17)
Chr1	240255405..240257029	<i>FMN2</i>	Exon 01	NM_020066.4	114.21 (8/202)
Chr1	245026914..245027614	<i>HNRNPU</i>	Exon 01	NM_031844.2	54.44 (0/112)
Chr2	3391390..3392446	<i>TRAPPC12</i>	Exon 02	NM_016030.5	77.05 (7/147)
Chr2	5832849..5834184	<i>SOX11</i>	Exon 01	NM_003108.3	73.66 (1/151)
Chr2	16082182..16082981	<i>MYCN</i>	Exon 02	NM_005378.5	94.82 (0/205)
Chr2	39347472..39347568	<i>SOS1</i>	Exon 01	NM_005633.3	25.75 (16/33)
Chr2	44222933..44223091	<i>LRPPRC</i>	Exon 01	NM_133259.3	37.82 (12/64)
Chr2	45169239..45170054	<i>SIX3</i>	Exon 01	NM_005413.3	119.46 (5/198)
Chr2	45171702..45171904	<i>SIX3</i>	Exon 02	NM_005413.3	49.21 (17/81)
Chr2	48132623..48132864	<i>FBXO11</i>	Exon 01	NM_001190274.1	13.19 (1/40)
Chr2	50573824..50574092	<i>NRXN1</i>	Exon 01	NM_138735.4	65.01 (0/122)
Chr2	60780346..60780410	<i>BCL11A</i>	Exon 01	NM_022893.3	22.45 (8/27)
Chr2	65216773..65217309	<i>SLC1A4</i>	Exon 01	NM_003038.4	57.20 (2/107)
Chr2	73114557..73114870	<i>SPR</i>	Exon 01	NM_003124.4	10.26 (2/21)
Chr2	74374943..74375006	<i>BOLA3</i>	Exon 01	NM_212552.2	11.16 (8/13)
Chr2	74692018..74692379	<i>MOGS</i>	Exon 01	NM_006302.2	49.95 (4/104)
Chr2	86115942..86116033	<i>ST3GAL5</i>	Exon 01	NM_003896.3	5.26 (2/7)
Chr2	88926480..88926797	<i>EIF2AK3</i>	Exon 01	NM_004836.6	14.32 (0/43)
Chr2	88991212..88991506	<i>RPIA</i>	Exon 01	NM_144563.2	62.37 (14/104)
Chr2	105471964..105473476	<i>POU3F3</i>	Exon 01	NM_006236.2	69.09 (0/213)
Chr2	121745779..121748256	<i>GLI2</i>	Exon 13	NM_005270.4	119.00 (0/208)
Chr2	162279875..162280743	<i>TBR1</i>	Exon 06	NM_006593.3	47.30 (0/114)
Chr2	172291083..172291218	<i>DCAF17</i>	Exon 01	NM_025000.3	21.85 (9/32)
Chr2	178257513..178257782	<i>AGPS</i>	Exon 01	NM_003659.3	33.68 (5/70)
Chr2	242674635..242674936	<i>D2HGDH</i>	Exon 02	NM_152783.4	51.94 (9/93)
Chr3	25824746..25824886	<i>NGLY1</i>	Exon 01	NM_001145293.1	43.35 (14/58)
Chr3	25824746..25824886	<i>NGLY1</i>	Exon 01	NM_001145295.1	43.35 (14/58)
Chr3	25824746..25824886	<i>NGLY1</i>	Exon 01	NM_018297.3	43.35 (14/58)
Chr3	43732480..43732536	<i>ABHD5</i>	Exon 01	NM_016006.4	24.14 (19/28)

Chr3	49027685..49028048	<i>P4HTM</i>	Exon 01	NM_177939.2	28.48 (0/63)
Chr3	50712690..50712736	<i>DOCK3</i>	Exon 01	NM_004947.4	15.94 (15/16)
Chr3	93715439..93715533	<i>ARL13B</i>	Exon 03	NM_001321328.1	0.00 (0/0)
Chr3	181430144..181431107	<i>SOX2</i>	Exon 01	NM_003106.3	174.14 (4/298)
Chr4	980868..981035	<i>IDUA</i>	Exon 01	NM_000203.4	11.79 (5/19)
Chr4	996052..996278	<i>IDUA</i>	Exon 08	NM_000203.4	40.87 (6/63)
Chr4	996515..996737	<i>IDUA</i>	Exon 09	NM_000203.4	7.21 (4/15)
Chr4	996819..996950	<i>IDUA</i>	Exon 10	NM_000203.4	11.60 (2/16)
Chr4	997128..997263	<i>IDUA</i>	Exon 11	NM_000203.4	13.25 (2/42)
Chr4	1242699..1242748	<i>CTBP1</i>	Exon 01	NM_001328.2	3.76 (2/4)
Chr4	15482816..15482841	<i>CC2D2A</i>	Exon 07	NM_020785.2	1.00 (1/1)
Chr4	15482816..15482914	<i>CC2D2A</i>	Exon 05	NM_001164720.1	1.05 (1/2)
Chr4	17513568..17513682	<i>QDPR</i>	Exon 01	NM_000320.2	50.39 (14/71)
Chr4	56212499..56212729	<i>SRD5A3</i>	Exon 01	NM_024592.4	43.98 (11/65)
Chr4	56262352..56262568	<i>TMEM165</i>	Exon 01	NM_018475.4	25.95 (2/40)
Chr4	103265596..103265824	<i>SLC39A8</i>	Exon 02	NM_001135146.1	60.52 (14/86)
Chr4	103265596..103265824	<i>SLC39A8</i>	Exon 01	NM_022154.5	60.52 (14/86)
Chr4	108852795..108853294	<i>CYP2U1</i>	Exon 01	NM_183075.2	93.14 (0/211)
Chr4	119273369..119273880	<i>PRSS12</i>	Exon 01	NM_003619.3	81.90 (1/157)
Chr5	218466..218538	<i>SDHA</i>	Exon 01	NM_004168.3	10.12 (5/16)
Chr5	6632992..6633097	<i>NSUN2</i>	Exon 01	NM_017755.5	6.15 (2/10)
Chr5	14143830..14143996	<i>TRIO</i>	Exon 01	NM_007118.3	0.86 (0/2)
Chr5	14487568..14488374	<i>TRIO</i>	Exon 48	NM_007118.3	54.63 (0/169)
Chr5	45695766..45696200	<i>HCN1</i>	Exon 01	NM_021072.3	40.89 (1/100)
Chr5	59188990..59189454	<i>PDE4D</i>	Exon 01	NM_001104631.1	41.55 (0/101)
Chr5	60628095..60628780	<i>ZSWIM6</i>	Exon 01	NM_020928.1	18.63 (0/72)
Chr5	68843752..68843933	<i>OCLN</i>	Exon 07	NM_002538.3	9.51 (6/12)
Chr5	68847366..68847417	<i>OCLN</i>	Exon 08	NM_002538.3	2.00 (2/2)
Chr5	78280755..78281076	<i>ARSB</i>	Exon 01	NM_000046.3	18.93 (0/41)
Chr5	78280755..78281076	<i>ARSB</i>	Exon 02	NM_198709.2	18.93 (0/41)
Chr5	79950542..79950788	<i>MSH3</i>	Exon 01	NM_002439.4	37.05 (8/64)
Chr5	92920725..92921197	<i>NR2F1</i>	Exon 01	NM_005654.5	65.07 (0/145)
Chr5	139493762..139494740	<i>PURA</i>	Exon 01	NM_005859.4	77.16 (0/142)
Chr5	140998360..140998486	<i>DIAPH1</i>	Exon 01	NM_005219.4	9.63 (0/16)



Chr5	161529555..161529684	<i>GABRG2</i>	Exon 06	NM_198903.2	0.00 (0/0)
Chr5	171433457..171433511	<i>FBXW11</i>	Exon 01	NM_012300.2	27.31 (19/34)
Chr5	177027207..177027266	<i>B4GALT7</i>	Exon 01	NM_007255.2	10.53 (6/12)
Chr6	3154092..3155162	<i>TUBB2A</i>	Exon 04	NM_001069.2	78.64 (7/179)
Chr6	3154092..3155184	<i>TUBB2A</i>	Exon 04	NM_001310315.1	79.17 (7/179)
Chr6	3224980..3226050	<i>TUBB2B</i>	Exon 04	NM_178012.4	92.34 (10/189)
Chr6	13281656..13281790	<i>LOC100130357</i>	Exon 04	NM_001242698.1	1.71 (1/2)
Chr6	21594761..21596195	<i>SOX4</i>	Exon 01	NM_003107.2	66.87 (1/182)
Chr6	24495220..24495583	<i>ALDH5A1</i>	Exon 01	NM_001080.3	18.01 (0/54)
Chr6	24495220..24495583	<i>ALDH5A1</i>	Exon 01	NM_170740.1	18.01 (0/54)
Chr6	42946002..42946893	<i>PEX6</i>	Exon 01	NM_000287.3	86.17 (0/220)
Chr6	43596709..43596904	<i>GTPBP2</i>	Exon 01	NM_019096.4	40.32 (9/63)
Chr6	73331913..73332320	<i>KCNQ5</i>	Exon 01	NM_001160133.1	37.62 (0/108)
Chr6	79787412..79787451	<i>PHIP</i>	Exon 03	NM_017934.6	7.00 (7/7)
Chr6	105307437..105307522	<i>HACE1</i>	Exon 01	NM_020771.3	21.45 (18/26)
Chr6	137143799..137143938	<i>PEX7</i>	Exon 01	NM_000288.3	16.26 (6/23)
Chr6	157099059..157100610	<i>ARID1B</i>	Exon 01	NM_001346813.1	42.23 (0/166)
Chr6	157099059..157100610	<i>ARID1B</i>	Exon 01	NM_020732.3	42.23 (0/166)
Chr7	193195..193809	<i>FAM20C</i>	Exon 01	NM_020223.3	33.09 (0/112)
Chr7	6414362..6414406	<i>RAC1</i>	Exon 01	NM_018890.3	13.38 (12/14)
Chr7	16460686..16460952	<i>CRPPA</i>	Exon 01	NM_001101426.3	35.18 (0/70)
Chr7	19156331..19156949	<i>TWIST1</i>	Exon 01	NM_000474.3	76.64 (0/187)
Chr7	39990236..39991456	<i>CDK13</i>	Exon 01	NM_003718.4	49.16 (0/123)
Chr7	44364951..44365025	<i>CAMK2B</i>	Exon 01	NM_001220.4	14.96 (10/16)
Chr7	66094047..66094200	<i>KCTD7</i>	Exon 01	NM_001167961.2	45.47 (12/62)
Chr7	66094047..66094200	<i>KCTD7</i>	Exon 01	NM_153033.4	45.47 (12/62)
Chr7	69064635..69064953	<i>AUTS2</i>	Exon 01	NM_015570.3	56.78 (3/119)
Chr7	101891687..101892327	<i>CUX1</i>	Exon 24	NM_001202543.1	15.47 (0/45)
Chr7	101891687..101892327	<i>CUX1</i>	Exon 24	NM_181552.3	15.47 (0/45)
Chr7	103629573..103629808	<i>RELN</i>	Exon 01	NM_005045.3	91.99 (17/136)
Chr7	103629573..103629808	<i>RELN</i>	Exon 01	NM_173054.2	91.99 (17/136)
Chr7	140624361..140624508	<i>BRAF</i>	Exon 01	NM_004333.4	13.32 (7/18)
Chr7	153749901..153750153	<i>DPP6</i>	Exon 01	NM_130797.3	20.23 (1/54)
Chr7	155595589..155596425	<i>SHH</i>	Exon 03	NM_000193.3	57.12 (1/176)

Chr8	42995635..42995762	<i>HGSNAT</i>	Exon 01	NM_152419.2	1.02 (0/2)
Chr8	100221815..100221901	<i>VPS13B</i>	Exon 18	NM_015243.2	5.68 (4/6)
Chr8	133492389..133492784	<i>KCNQ3</i>	Exon 01	NM_004519.3	52.13 (0/101)
Chr8	145137629..145137712	<i>GPAA1</i>	Exon 01	NM_003801.3	6.37 (5/8)
Chr8	145149998..145150136	<i>CYC1</i>	Exon 01	NM_001916.4	0.00 (0/0)
Chr9	214504..215401	<i>DOCK8-AS1</i>	Exon 01	NM_152569.2	58.90 (3/178)
Chr9	2047224..2047489	<i>SMARCA2</i>	Exon 05	NM_003070.4	30.51 (0/76)
Chr9	2622185..2622276	<i>VLDLR</i>	Exon 01	NM_001018056.2	12.04 (6/20)
Chr9	2622185..2622276	<i>VLDLR</i>	Exon 01	NM_001322225.1	12.04 (6/20)
Chr9	2622185..2622276	<i>VLDLR</i>	Exon 01	NM_001322226.1	12.04 (6/20)
Chr9	2622185..2622276	<i>VLDLR</i>	Exon 01	NM_003383.4	12.04 (6/20)
Chr9	6645240..6645504	<i>GLDC</i>	Exon 01	NM_000170.2	32.56 (0/76)
Chr9	94123905..94124176	<i>AUH</i>	Exon 01	NM_001306190.1	33.47 (8/64)
Chr9	94123905..94124176	<i>AUH</i>	Exon 01	NM_001698.2	33.47 (8/64)
Chr9	94712144..94712250	<i>ROR2</i>	Exon 01	NM_004560.3	11.36 (2/20)
Chr9	98270438..98270648	<i>PTCH1</i>	Exon 01	NM_000264.3	24.48 (2/60)
Chr9	101470694..101471024	<i>GABBR2</i>	Exon 01	NM_005458.7	64.56 (0/165)
Chr9	126128191..126128396	<i>CRB2</i>	Exon 03	NM_173689.6	74.80 (17/119)
Chr9	126128501..126128650	<i>CRB2</i>	Exon 04	NM_173689.6	49.01 (17/65)
Chr9	126135408..126136204	<i>CRB2</i>	Exon 10	NM_173689.6	15.67 (2/70)
Chr9	130965745..130965915	<i>DNM1</i>	Exon 01	NM_001005336.2	45.74 (18/59)
Chr9	130965745..130965915	<i>DNM1</i>	Exon 01	NM_001288737.1	45.74 (18/59)
Chr9	130965745..130965915	<i>DNM1</i>	Exon 01	NM_001288738.1	45.74 (18/59)
Chr9	130965745..130965915	<i>DNM1</i>	Exon 01	NM_001288739.1	45.74 (18/59)
Chr9	130965745..130965915	<i>DNM1</i>	Exon 01	NM_004408.3	45.74 (18/59)
Chr9	131012999..131013224	<i>DNM1</i>	Exon 21	NM_001005336.2	15.25 (0/37)
Chr9	131012999..131013224	<i>DNM1</i>	Exon 21	NM_001288737.1	15.25 (0/37)
Chr9	131012999..131013224	<i>DNM1</i>	Exon 21	NM_001288738.1	15.25 (0/37)
Chr9	131012999..131013224	<i>DNM1</i>	Exon 21	NM_001288739.1	15.25 (0/37)
Chr9	131012999..131013224	<i>DNM1</i>	Exon 21	NM_004408.3	15.25 (0/37)
Chr9	131709994..131710035	<i>NUP188</i>	Exon 01	NM_015354.2	21.79 (14/30)
Chr9	133884597..133884979	<i>LAMC3</i>	Exon 01	NM_006059.3	23.73 (0/68)
Chr9	135117258..135117503	<i>NTNG2</i>	Exon 08	NM_032536.2	9.23 (0/27)
Chr9	136223271..136223334	<i>SURF1</i>	Exon 01	NM_003172.3	4.72 (3/7)

Chr9	138606418..138606571	<i>KCNT1</i>	Exon 02	NM_020822.2	33.55 (5/44)
Chr9	138678038..138678372	<i>KCNT1</i>	Exon 28	NM_001272003.1	53.30 (10/85)
Chr9	138678038..138678372	<i>KCNT1</i>	Exon 29	NM_020822.2	53.30 (10/85)
Chr9	139333055..139333876	<i>INPP5E</i>	Exon 01	NM_001318502.1	44.97 (2/91)
Chr9	139333055..139333876	<i>INPP5E</i>	Exon 01	NM_019892.5	44.97 (2/91)
Chr9	139981447..139981675	<i>MAN1B1</i>	Exon 01	NM_016219.4	65.34 (8/111)
Chr9	140055503..140055654	<i>GRIN1</i>	Exon 09	NM_007327.3	43.66 (4/61)
Chr9	140513476..140513506	<i>EHMT1</i>	Exon 01	NM_024757.4	0.00 (0/0)
Chr9	140729220..140729410	<i>EHMT1</i>	Exon 27	NM_024757.4	37.16 (14/59)
Chr9	140772381..140772674	<i>CACNA1B</i>	Exon 01	NM_000718.3	35.68 (0/71)
Chr9	140917458..140918268	<i>CACNA1B</i>	Exon 19	NM_000718.3	26.40 (0/68)
Chr10	23481455..23482248	<i>PTF1A</i>	Exon 01	NM_178161.2	44.87 (0/127)
Chr10	26986636..26986774	<i>PDSS1</i>	Exon 01	NM_001321978.1	6.40 (4/9)
Chr10	26986636..26986774	<i>PDSS1</i>	Exon 01	NM_014317.4	6.40 (4/9)
Chr10	26991086..26991128	<i>PDSS1</i>	Exon 02	NM_001321978.1	0.00 (0/0)
Chr10	26991086..26991128	<i>PDSS1</i>	Exon 02	NM_014317.4	0.00 (0/0)
Chr10	71078699..71078771	<i>HK1</i>	Exon 01	NM_000188.2	23.34 (15/27)
Chr10	104404370..104404949	<i>TRIM8</i>	Exon 01	NM_030912.2	50.81 (18/88)
Chr10	104678233..104679863	<i>CNNM2</i>	Exon 01	NM_017649.4	75.57 (4/144)
Chr11	694754..695052	<i>DEAF1</i>	Exon 01	NM_021008.3	4.68 (0/12)
Chr11	695604..695851	<i>TMEM80</i>	Exon 01	NM_001276274.1	38.28 (3/76)
Chr11	797621..798278	<i>PANO1</i>	Exon 01	NM_001293167.1	66.32 (0/140)
Chr11	1411510..1411610	<i>BRSK2</i>	Exon 01	NM_001256627.1	10.44 (4/14)
Chr11	1481725..1481958	<i>BRSK2</i>	Exon 20	NM_001256627.1	19.39 (10/26)
Chr11	6651225..6652099	<i>DCHS1</i>	Exon 10	NM_003737.3	65.81 (5/187)
Chr11	17757545..17758124	<i>KCNC1</i>	Exon 01	NM_001112741.1	104.54 (14/206)
Chr11	65488053..65488234	<i>RNASEH2C</i>	Exon 01	NM_032193.3	55.26 (10/86)
Chr11	65837953..65838318	<i>PACS1</i>	Exon 01	NM_018026.3	7.23 (0/16)
Chr11	70766131..70766149	<i>SHANK2</i>	Exon 11	NM_012309.4	0.00 (0/0)
Chr11	70776121..70776141	<i>SHANK2</i>	Exon 10	NM_012309.4	0.00 (0/0)
Chr11	70776133..70776155	<i>SHANK2</i>	Exon 09	NM_012309.4	0.00 (0/0)
Chr11	70785644..70785662	<i>SHANK2</i>	Exon 08	NM_012309.4	0.11 (0/1)
Chr11	77813935..77813999	<i>ALG8</i>	Exon 13	NM_001007027.2	0.00 (0/0)
Chr11	118307223..118307664	<i>KMT2A</i>	Exon 01	NM_001197104.1	16.88 (0/42)

Chr11	118307223..118307664	<i>KMT2A</i>	Exon 01	NM_005933.3	16.88 (0/42)
Chr11	124791029..124791341	<i>HEPACAM</i>	Exon 07	NM_152722.4	19.53 (2/44)
Chr12	50479148..50479334	<i>SMARCD1</i>	Exon 01	NM_003076.4	23.48 (14/32)
Chr12	56583130..56583250	<i>SMARCC2</i>	Exon 01	NM_003075.4	28.75 (19/38)
Chr12	58185743..58185815	<i>TSM</i>	Exon 05	NM_001172696.1	0.58 (0/1)
Chr12	64173736..64173914	<i>RXYLT1</i>	Exon 01	NM_014254.2	38.76 (6/71)
Chr12	88502838..88502963	<i>CEP290</i>	Exon 23	NM_025114.3	34.67 (19/43)
Chr12	111757811..111758584	<i>CUX2</i>	Exon 17	NM_015267.3	76.35 (7/137)
Chr12	121163684..121163739	<i>ACADS</i>	Exon 01	NM_000017.3	11.46 (10/13)
Chr12	121163684..121163739	<i>ACADS</i>	Exon 01	NM_001302554.1	11.46 (10/13)
Chr12	122252007..122252841	<i>SETD1B</i>	Exon 06	NM_015048.1	79.81 (10/130)
Chr12	124179762..124179849	<i>TCTN2</i>	Exon 11	NM_001143850.2	17.32 (12/21)
Chr12	124179762..124179849	<i>TCTN2</i>	Exon 11	NM_024809.4	17.32 (12/21)
Chr12	132414447..132414685	<i>PUS1</i>	Exon 02	NM_025215.5	41.69 (2/79)
Chr13	25946346..25946431	<i>ATP8A2</i>	Exon 01	NM_016529.5	23.01 (13/26)
Chr13	31774217..31774296	<i>B3GLCT</i>	Exon 01	NM_194318.3	5.60 (4/8)
Chr13	35516953..35517256	<i>NBEA</i>	Exon 01	NM_015678.4	72.79 (14/136)
Chr13	51484208..51484281	<i>RNASEH2B</i>	Exon 01	NM_024570.3	8.89 (4/10)
Chr13	77574588..77575109	<i>CLN5</i>	Exon 04	NM_006493.2	103.50 (17/172)
Chr13	100634314..100635398	<i>ZIC2</i>	Exon 01	NM_007129.3	92.97 (2/227)
Chr13	100637572..100637941	<i>ZIC2</i>	Exon 03	NM_007129.3	22.51 (0/78)
Chr13	100741370..100741484	<i>PCCA</i>	Exon 01	NM_000282.3	27.27 (5/47)
Chr13	110959286..110959379	<i>COL4A1</i>	Exon 01	NM_001845.5	10.51 (1/15)
Chr14	29236481..29237960	<i>FOXP1</i>	Exon 01	NM_005249.4	108.33 (0/224)
Chr14	30396450..30396723	<i>PRKD1</i>	Exon 01	NM_002742.2	42.33 (1/90)
Chr14	31495105..31495396	<i>STRN3</i>	Exon 01	NM_001083893.1	24.78 (6/55)
Chr14	36986478..36987230	<i>NKX2-1</i>	Exon 03	NM_001079668.2	86.23 (2/187)
Chr14	36988185..36988580	<i>NKX2-1</i>	Exon 02	NM_001079668.2	72.94 (16/132)
Chr14	55369034..55369386	<i>GCH1</i>	Exon 01	NM_000161.2	39.90 (4/81)
Chr14	58907934..58908066	<i>KIAA0586</i>	Exon 06	NM_001244189.1	0.00 (0/0)
Chr14	58907934..58908066	<i>KIAA0586</i>	Exon 05	NM_001244192.1	0.00 (0/0)
Chr14	59010596..59010690	<i>KIAA0586</i>	Exon 33	NM_001244189.1	2.78 (1/4)
Chr14	59010596..59010690	<i>KIAA0586</i>	Exon 31	NM_001329944.1	2.78 (1/4)
Chr14	70346391..70346499	<i>SMOC1</i>	Exon 01	NM_001034852.2	47.26 (13/71)

Chr14	77491740..77494140	<i>IRF2BPL</i>	Exon 01	NM_024496.3	81.85 (0/161)
Chr14	77786772..77787029	<i>POMT2</i>	Exon 01	NM_013382.5	50.20 (16/91)
Chr14	88459309..88459513	<i>GALC</i>	Exon 01	NM_000153.3	17.96 (3/44)
Chr14	88459309..88459513	<i>GALC</i>	Exon 01	NM_001201401.1	17.96 (3/44)
Chr14	91883970..91884039	<i>CCDC88C</i>	Exon 01	NM_001080414.3	23.39 (14/30)
Chr14	99640483..99642537	<i>BCL11B</i>	Exon 04	NM_138576.3	61.28 (0/167)
Chr14	100259809..100259885	<i>EML1</i>	Exon 01	NM_004434.2	24.01 (18/26)
Chr14	100705577..100706265	<i>YY1</i>	Exon 01	NM_003403.4	32.26 (3/84)
Chr14	105715203..105715427	<i>BTBD6</i>	Exon 02	NM_033271.2	0.49 (0/2)
Chr14	105715489..105715589	<i>BTBD6</i>	Exon 03	NM_033271.2	20.29 (12/26)
Chr14	105766778..105766971	<i>BRF1</i>	Exon 01	NM_001519.3	22.56 (0/39)
Chr14	105781251..105781379	<i>PACS2</i>	Exon 01	NM_001100913.2	13.58 (0/22)
Chr15	23889135..23892894	<i>MAGEL2</i>	Exon 01	NM_019066.4	154.66 (4/327)
Chr15	27018025..27018114	<i>GABRB3</i>	Exon 01	NM_000814.5	19.92 (15/22)
Chr15	40902992..40903079	<i>KNL1</i>	Exon 07	NM_170589.4	1.00 (1/1)
Chr15	40903671..40903714	<i>KNL1</i>	Exon 08	NM_170589.4	7.77 (7/8)
Chr15	45670578..45670656	<i>GATM</i>	Exon 01	NM_001482.2	20.13 (16/25)
Chr15	61521247..61521422	<i>RORA</i>	Exon 01	NM_134261.2	26.02 (9/40)
Chr15	65321738..65321956	<i>MTFMT</i>	Exon 01	NM_139242.3	35.03 (8/49)
Chr15	68521835..68521927	<i>CLN6</i>	Exon 01	NM_017882.2	3.63 (2/6)
Chr15	83349219..83349512	<i>AP3B2</i>	Exon 07	NM_001278511.1	96.10 (11/143)
Chr15	83349219..83349512	<i>AP3B2</i>	Exon 08	NM_001278512.1	96.10 (11/143)
Chr15	83349219..83349512	<i>AP3B2</i>	Exon 08	NM_004644.4	96.10 (11/143)
Chr15	83378341..83378463	<i>AP3B2</i>	Exon 01	NM_001278511.1	24.43 (16/31)
Chr15	83378341..83378463	<i>AP3B2</i>	Exon 01	NM_001278512.1	24.43 (16/31)
Chr15	83378341..83378463	<i>AP3B2</i>	Exon 01	NM_004644.4	24.43 (16/31)
Chr15	90191481..90192010	<i>KIF7</i>	Exon 05	NM_198525.2	32.95 (0/98)
Chr15	90645503..90645627	<i>IDH2</i>	Exon 01	NM_002168.3	14.04 (4/25)
Chr16	1401962..1402023	<i>GNPTG</i>	Exon 01	NM_032520.4	0.76 (0/1)
Chr16	1822795..1823125	<i>MRPS34</i>	Exon 01	NM_023936.1	15.19 (6/27)
Chr16	2034215..2034482	<i>GFER</i>	Exon 01	NM_005262.2	15.87 (2/42)
Chr16	4852523..4852577	<i>ROGDI</i>	Exon 01	NM_024589.2	8.31 (5/11)
Chr16	46918623..46918875	<i>GPT2</i>	Exon 02	NM_133443.3	32.68 (0/86)
Chr16	53656126..53656273	<i>RPGRIP1L</i>	Exon 22	NM_001330538.1	29.32 (15/39)

Chr16	53656126..53656273	<i>RPGRIP1L</i>	Exon 23	NM_015272.4	29.32 (15/39)
Chr16	54966984..54967790	<i>IRX5</i>	Exon 03	NM_005853.5	78.85 (2/158)
Chr16	69363895..69364478	<i>PDF</i>	Exon 01	NM_022341.1	30.21 (2/90)
Chr16	69364737..69365003	<i>COG8</i>	Exon 05	NM_032382.4	66.72 (16/109)
Chr16	69373074..69373460	<i>COG8</i>	Exon 01	NM_032382.4	58.81 (15/98)
Chr16	74808379..74808658	<i>FA2H</i>	Exon 01	NM_024306.4	18.62 (2/42)
Chr16	79632677..79633804	<i>MAF</i>	Exon 01	NM_005360.4	94.28 (0/177)
Chr16	83932745..83933282	<i>MLYCD</i>	Exon 01	NM_012213.2	18.26 (1/56)
Chr16	89258608..89258857	<i>CDH15</i>	Exon 11	NM_004933.2	18.58 (9/33)
Chr16	89334881..89335076	<i>ANKRD11</i>	Exon 14	NM_001256182.1	52.35 (17/75)
Chr16	89334881..89335076	<i>ANKRD11</i>	Exon 13	NM_013275.5	52.35 (17/75)
Chr16	89345475..89352062	<i>ANKRD11</i>	Exon 10	NM_001256182.1	97.00 (8/151)
Chr16	89345475..89352062	<i>ANKRD11</i>	Exon 09	NM_013275.5	97.00 (8/151)
Chr17	7412846..7412956	<i>POLR2A</i>	Exon 22	NM_000937.4	23.91 (15/28)
Chr17	8066191..8066202	<i>VAMP2</i>	Exon 01	NM_014232.2	10.00 (10/10)
Chr17	12921015..12921269	<i>ELAC2</i>	Exon 01	NM_018127.6	61.18 (14/98)
Chr17	15903158..15903351	<i>TTC19</i>	Exon 01	NM_017775.3	6.22 (0/18)
Chr17	15903427..15903564	<i>TTC19</i>	Exon 02	NM_017775.3	28.33 (16/40)
Chr17	26732900..26733137	<i>SLC46A1</i>	Exon 01	NM_080669.5	44.38 (8/68)
Chr17	40688286..40688678	<i>NAGLU</i>	Exon 01	NM_000263.3	7.35 (0/24)
Chr17	48674086..48674289	<i>CACNA1G</i>	Exon 16	NM_001256324.1	46.31 (12/75)
Chr17	48674086..48674289	<i>CACNA1G</i>	Exon 16	NM_001256325.1	46.31 (12/75)
Chr17	48674086..48674289	<i>CACNA1G</i>	Exon 16	NM_001256326.1	46.31 (12/75)
Chr17	48674086..48674289	<i>CACNA1G</i>	Exon 16	NM_001256327.1	46.31 (12/75)
Chr17	48674086..48674289	<i>CACNA1G</i>	Exon 16	NM_001256328.1	46.31 (12/75)
Chr17	48674086..48674289	<i>CACNA1G</i>	Exon 16	NM_001256329.1	46.31 (12/75)
Chr17	48674086..48674289	<i>CACNA1G</i>	Exon 16	NM_001256330.1	46.31 (12/75)
Chr17	48674086..48674289	<i>CACNA1G</i>	Exon 16	NM_001256331.1	46.31 (12/75)
Chr17	48674086..48674289	<i>CACNA1G</i>	Exon 15	NM_001256332.1	46.31 (12/75)
Chr17	48674086..48674289	<i>CACNA1G</i>	Exon 16	NM_001256333.1	46.31 (12/75)
Chr17	48674086..48674289	<i>CACNA1G</i>	Exon 16	NM_001256334.1	46.31 (12/75)
Chr17	48674086..48674289	<i>CACNA1G</i>	Exon 16	NM_001256359.1	46.31 (12/75)
Chr17	48674086..48674289	<i>CACNA1G</i>	Exon 16	NM_001256360.1	46.31 (12/75)
Chr17	48674086..48674289	<i>CACNA1G</i>	Exon 16	NM_001256361.1	46.31 (12/75)

Chr17	48674086..48674289	<i>CACNA1G</i>	Exon 16	NM_018896.4	46.31 (12/75)
Chr17	48674086..48674289	<i>CACNA1G</i>	Exon 15	NM_198376.2	46.31 (12/75)
Chr17	48674086..48674289	<i>CACNA1G</i>	Exon 16	NM_198377.2	46.31 (12/75)
Chr17	48674086..48674289	<i>CACNA1G</i>	Exon 16	NM_198378.2	46.31 (12/75)
Chr17	48674086..48674289	<i>CACNA1G</i>	Exon 15	NM_198379.2	46.31 (12/75)
Chr17	48674086..48674289	<i>CACNA1G</i>	Exon 16	NM_198380.2	46.31 (12/75)
Chr17	48674086..48674289	<i>CACNA1G</i>	Exon 15	NM_198382.2	46.31 (12/75)
Chr17	48674086..48674289	<i>CACNA1G</i>	Exon 15	NM_198383.2	46.31 (12/75)
Chr17	48674086..48674289	<i>CACNA1G</i>	Exon 16	NM_198384.2	46.31 (12/75)
Chr17	48674086..48674289	<i>CACNA1G</i>	Exon 16	NM_198385.2	46.31 (12/75)
Chr17	48674086..48674289	<i>CACNA1G</i>	Exon 16	NM_198386.2	46.31 (12/75)
Chr17	48674086..48674289	<i>CACNA1G</i>	Exon 15	NM_198387.2	46.31 (12/75)
Chr17	48674086..48674289	<i>CACNA1G</i>	Exon 15	NM_198388.2	46.31 (12/75)
Chr17	48674086..48674289	<i>CACNA1G</i>	Exon 15	NM_198396.2	46.31 (12/75)
Chr17	58677771..58678252	<i>PPM1D</i>	Exon 01	NM_003620.3	64.72 (14/132)
Chr17	65821836..65822458	<i>BPTF</i>	Exon 01	NM_004459.6	42.55 (0/137)
Chr17	70119679..70120533	<i>SOX9</i>	Exon 03	NM_000346.3	120.93 (4/191)
Chr17	71189204..71189528	<i>COG1</i>	Exon 01	NM_018714.2	71.43 (0/118)
Chr17	73512637..73512702	<i>TSEN54</i>	Exon 01	NM_207346.2	0.00 (0/0)
Chr17	73512822..73512996	<i>TSEN54</i>	Exon 02	NM_207346.2	25.30 (7/46)
Chr17	78120587..78120765	<i>EIF4A3</i>	Exon 01	NM_014740.3	53.25 (16/75)
Chr17	79869138..79869236	<i>PCYT2</i>	Exon 01	NM_002861.4	3.91 (1/6)
Chr17	79989633..79989677	<i>RAC3</i>	Exon 01	NM_005052.2	0.00 (0/0)
Chr17	80710065..80710258	<i>TBCD</i>	Exon 01	NM_005993.4	21.85 (2/46)
Chr18	12376963..12377086	<i>AFG3L2</i>	Exon 01	NM_006796.2	3.73 (2/6)
Chr18	21166246..21166312	<i>NPC1</i>	Exon 01	NM_000271.4	16.28 (12/18)
Chr18	42643039..42643668	<i>SETBP1</i>	Exon 06	NM_015559.2	97.22 (3/188)
Chr18	77439943..77440266	<i>CTDP1</i>	Exon 01	NM_004715.4	7.72 (0/18)
Chr19	1401290..1401480	<i>GAMT</i>	Exon 01	NM_000156.5	23.95 (3/39)
Chr19	1456847..1457247	<i>APC2</i>	Exon 09	NM_005883.2	41.70 (9/75)
Chr19	1465149..1470217	<i>APC2</i>	Exon 15	NM_005883.2	41.33 (0/131)
Chr19	1912042..1913155	<i>ADAT3</i>	Exon 02	NM_138422.3	42.74 (4/106)
Chr19	4123776..4123877	<i>MAP2K2</i>	Exon 01	NM_030662.3	47.42 (16/62)
Chr19	5719710..5720148	<i>LONP1</i>	Exon 01	NM_004793.3	37.49 (10/89)

Chr19	7587632..7587672	<i>MCOLN1</i>	Exon 01	NM_020533.2	14.32 (11/18)
Chr19	11373068..11373121	<i>DOCK6</i>	Exon 01	NM_020812.3	18.26 (16/20)
Chr19	12984418..12985689	<i>MAST1</i>	Exon 26	NM_014975.2	75.13 (18/141)
Chr19	13318122..13318872	<i>CACNA1A</i>	Exon 47	NM_001127222.1	20.01 (0/53)
Chr19	13318122..13318872	<i>CACNA1A</i>	Exon 48	NM_023035.2	20.01 (0/53)
Chr19	13319565..13319828	<i>CACNA1A</i>	Exon 47	NM_000068.3	88.08 (16/147)
Chr19	13319565..13319828	<i>CACNA1A</i>	Exon 46	NM_001127221.1	88.08 (16/147)
Chr19	13319565..13319828	<i>CACNA1A</i>	Exon 46	NM_001127222.1	88.08 (16/147)
Chr19	13319565..13319828	<i>CACNA1A</i>	Exon 47	NM_001174080.1	88.08 (16/147)
Chr19	13319565..13319828	<i>CACNA1A</i>	Exon 47	NM_023035.2	88.08 (16/147)
Chr19	13409353..13410172	<i>CACNA1A</i>	Exon 19	NM_000068.3	61.48 (5/118)
Chr19	13409353..13410172	<i>CACNA1A</i>	Exon 19	NM_001127221.1	61.48 (5/118)
Chr19	13409353..13410172	<i>CACNA1A</i>	Exon 19	NM_001127222.1	61.48 (5/118)
Chr19	13409353..13410172	<i>CACNA1A</i>	Exon 19	NM_001174080.1	61.48 (5/118)
Chr19	13409353..13410172	<i>CACNA1A</i>	Exon 19	NM_023035.2	61.48 (5/118)
Chr19	13616741..13617043	<i>CACNA1A</i>	Exon 01	NM_000068.3	60.25 (12/109)
Chr19	13616741..13617043	<i>CACNA1A</i>	Exon 01	NM_001127221.1	60.25 (12/109)
Chr19	13616741..13617043	<i>CACNA1A</i>	Exon 01	NM_001127222.1	60.25 (12/109)
Chr19	13616741..13617043	<i>CACNA1A</i>	Exon 01	NM_001174080.1	60.25 (12/109)
Chr19	13616741..13617043	<i>CACNA1A</i>	Exon 01	NM_023035.2	60.25 (12/109)
Chr19	15355037..15355416	<i>BRD4</i>	Exon 13	NM_058243.2	73.08 (14/143)
Chr19	17448812..17449069	<i>GTPBP3</i>	Exon 02	NM_133644.3	52.20 (8/83)
Chr19	18266685..18267016	<i>PIK3R2</i>	Exon 02	NM_005027.3	40.70 (8/88)
Chr19	18271859..18272000	<i>PIK3R2</i>	Exon 05	NM_005027.3	58.06 (8/99)
Chr19	18272084..18272310	<i>PIK3R2</i>	Exon 06	NM_005027.3	3.09 (0/6)
Chr19	18279892..18280109	<i>PIK3R2</i>	Exon 16	NM_005027.3	44.51 (9/73)
Chr19	35617169..35617684	<i>LGI4</i>	Exon 08	NM_139284.2	58.96 (17/107)
Chr19	36486172..36486529	<i>SDHAF1</i>	Exon 01	NM_001042631.2	30.00 (7/72)
Chr19	47249401..47249692	<i>STRN4</i>	Exon 01	NM_013403.2	9.05 (0/27)
Chr19	47258703..47260200	<i>FKRP</i>	Exon 04	NM_001039885.2	63.66 (0/142)
Chr19	47258703..47260200	<i>FKRP</i>	Exon 04	NM_024301.4	63.66 (0/142)
Chr19	48901645..48902119	<i>GRIN2D</i>	Exon 02	NM_000836.2	8.28 (0/27)
Chr19	48945852..48947199	<i>GRIN2D</i>	Exon 13	NM_000836.2	6.78 (0/38)
Chr19	50097949..50101275	<i>PRR12</i>	Exon 04	NM_020719.2	89.67 (17/184)



Chr19	50823498..50823611	<i>KCNK3</i>	Exon 04	NM_004977.2	25.81 (15/35)
Chr19	50831465..50832344	<i>KCNK3</i>	Exon 01	NM_004977.2	25.58 (0/117)
Chr19	54695211..54695463	<i>TSEN34</i>	Exon 02	NM_024075.4	25.51 (4/47)
Chr20	442975..443054	<i>TBC1D20</i>	Exon 01	NM_144628.3	5.80 (2/7)
Chr20	30946574..30946640	<i>ASXL1</i>	Exon 01	NM_001164603.1	16.82 (16/18)
Chr20	30946574..30946640	<i>ASXL1</i>	Exon 01	NM_015338.5	16.82 (16/18)
Chr20	47538422..47538552	<i>ARFGEF2</i>	Exon 01	NM_006420.2	24.95 (19/30)
Chr20	48098446..48099022	<i>KCNB1</i>	Exon 01	NM_004975.3	89.17 (17/123)
Chr20	49557637..49557751	<i>DPM1</i>	Exon 07	NM_001317034.1	22.52 (10/32)
Chr20	49557661..49557751	<i>DPM1</i>	Exon 07	NM_001317035.1	20.41 (10/27)
Chr20	57428316..57430393	<i>GNAS</i>	Exon 01	NM_080425.3	137.64 (5/218)
Chr20	57466777..57466925	<i>GNAS</i>	Exon 01	NM_000516.5	18.07 (4/22)
Chr20	62103516..62103821	<i>KCNQ2</i>	Exon 01	NM_004518.5	24.11 (16/30)
Chr20	62103516..62103821	<i>KCNQ2</i>	Exon 01	NM_172106.2	24.11 (16/30)
Chr20	62103516..62103821	<i>KCNQ2</i>	Exon 01	NM_172107.3	24.11 (16/30)
Chr20	62103516..62103821	<i>KCNQ2</i>	Exon 01	NM_172108.4	24.11 (16/30)
Chr20	62103516..62103821	<i>KCNQ2</i>	Exon 01	NM_172109.2	24.11 (16/30)
Chr20	62119646..62119783	<i>EEF1A2</i>	Exon 08	NM_001958.3	8.81 (0/14)
Chr21	34100252..34100356	<i>SYNJ1</i>	Exon 01	NM_003895.3	15.39 (11/18)
Chr21	34921777..34927702	<i>SON</i>	Exon 03	NM_032195.2	119.44 (5/210)
Chr21	34921777..34927702	<i>SON</i>	Exon 03	NM_138927.2	119.44 (5/210)
Chr21	47565727..47565866	<i>FTCD</i>	Exon 09	NM_006657.2	25.11 (5/37)
Chr22	18561138..18561377	<i>PEX26</i>	Exon 02	NM_017929.5	50.00 (17/94)
Chr22	18923523..18923805	<i>PRODH</i>	Exon 02	NM_016335.4	5.60 (2/10)
Chr22	19165641..19165758	<i>SLC25A1</i>	Exon 02	NM_005984.4	51.46 (18/67)
Chr22	19166088..19166191	<i>SLC25A1</i>	Exon 01	NM_005984.4	6.15 (2/10)
Chr22	38379359..38379796	<i>SOX10</i>	Exon 02	NM_006941.3	84.25 (5/163)
Chr22	43045296..43045326	<i>CYB5R3</i>	Exon 01	NM_000398.6	2.00 (2/2)
Chr22	50964194..50964352	<i>TYMP</i>	Exon 10	NM_001953.4	16.46 (1/29)
Chr22	50964425..50964575	<i>TYMP</i>	Exon 09	NM_001953.4	9.38 (4/12)
Chr22	50964670..50964910	<i>TYMP</i>	Exon 08	NM_001953.4	12.27 (0/31)
Chr22	51020982..51021215	<i>CHKB</i>	Exon 01	NM_005198.4	51.96 (14/69)
Chr22	51113065..51113137	<i>SHANK3</i>	Exon 01	NM_033517.1	0.44 (0/2)
Chr22	51113471..51113684	<i>SHANK3</i>	Exon 02	NM_033517.1	40.79 (7/55)

Chr22	51135980..51135994	<i>SHANK3</i>	Exon 11	NM_033517.1	0.27 (0/2)
Chr22	51135987..51136148	<i>SHANK3</i>	Exon 12	NM_033517.1	6.56 (0/10)
Chr22	51158607..51160870	<i>SHANK3</i>	Exon 22	NM_033517.1	67.16 (0/154)
Chr22	51169144..51169745	<i>SHANK3</i>	Exon 23	NM_033517.1	25.67 (2/80)
ChrX	17393876..17394450	<i>NHS</i>	Exon 01	NM_198270.3	18.05 (0/57)
ChrX	18631267..18631400	<i>CDKL5</i>	Exon 16	NM_001037343.1	22.74 (15/26)
ChrX	18631267..18631400	<i>CDKL5</i>	Exon 14	NM_001323289.1	22.74 (15/26)
ChrX	18631267..18631400	<i>CDKL5</i>	Exon 15	NM_003159.2	22.74 (15/26)
ChrX	18646486..18646882	<i>CDKL5</i>	Exon 17	NM_001323289.1	71.11 (19/98)
ChrX	19379452..19379538	<i>MAP3K15</i>	Exon 28	NM_001001671.3	21.11 (18/25)
ChrX	20179757..20179884	<i>RPS6KA3</i>	Exon 20	NM_004586.2	28.02 (18/34)
ChrX	20206610..20206657	<i>RPS6KA3</i>	Exon 08	NM_004586.2	21.00 (19/22)
ChrX	20284677..20284755	<i>RPS6KA3</i>	Exon 01	NM_004586.2	20.27 (16/23)
ChrX	21958938..21958996	<i>SMS</i>	Exon 01	NM_004595.4	13.46 (10/16)
ChrX	25022782..25023032	<i>ARX</i>	Exon 05	NM_139058.2	19.10 (9/26)
ChrX	25025223..25025561	<i>ARX</i>	Exon 04	NM_139058.2	24.05 (6/45)
ChrX	25031034..25031920	<i>ARX</i>	Exon 02	NM_139058.2	29.92 (0/94)
ChrX	31526320..31526359	<i>DMD</i>	Exon 01	NM_004014.2	21.35 (18/24)
ChrX	38420795..38420885	<i>TSPAN7</i>	Exon 01	NM_004615.3	39.27 (16/50)
ChrX	40440313..40440359	<i>ATP6AP2</i>	Exon 01	NM_005765.2	10.72 (8/11)
ChrX	41000214..41000475	<i>USP9X</i>	Exon 08	NM_001039590.2	28.05 (18/39)
ChrX	41056612..41056768	<i>USP9X</i>	Exon 29	NM_001039590.2	45.61 (16/58)
ChrX	47433396..47433994	<i>SYN1</i>	Exon 12	NM_133499.2	13.39 (1/36)
ChrX	47466535..47466602	<i>SYN1</i>	Exon 02	NM_133499.2	20.38 (15/26)
ChrX	47478746..47479132	<i>SYN1</i>	Exon 01	NM_133499.2	15.04 (2/29)
ChrX	49056605..49056650	<i>SYP</i>	Exon 01	NM_003179.2	19.46 (16/23)
ChrX	53263396..53264371	<i>IQSEC2</i>	Exon 15	NM_001111125.2	56.91 (7/110)
ChrX	53349610..53350326	<i>IQSEC2</i>	Exon 01	NM_001111125.2	40.28 (0/86)
ChrX	53449436..53449554	<i>SMC1A</i>	Exon 01	NM_006306.3	20.27 (12/25)
ChrX	53565791..53566046	<i>HUWE1</i>	Exon 76	NM_031407.6	34.04 (18/45)
ChrX	54521554..54521870	<i>FGD1</i>	Exon 01	NM_004463.2	56.79 (1/111)
ChrX	69665047..69665413	<i>DLG3</i>	Exon 01	NM_021120.3	52.49 (12/76)
ChrX	70641154..70641231	<i>TAF1</i>	Exon 29	NM_004606.4	27.95 (15/37)
ChrX	77266668..77266754	<i>ATP7A</i>	Exon 08	NM_000052.6	31.49 (19/37)

ChrX	77266668..77266754	<i>ATP7A</i>	Exon 08	NM_001282224.1	31.49 (19/37)
ChrX	108912258..108912407	<i>ACSL4</i>	Exon 10	NM_004458.2	25.40 (17/31)
ChrX	110952188..110952281	<i>ALG13</i>	Exon 05	NM_001099922.2	18.37 (12/24)
ChrX	110952188..110952281	<i>ALG13</i>	Exon 05	NM_001257230.1	18.37 (12/24)
ChrX	110952188..110952281	<i>ALG13</i>	Exon 05	NM_001257231.1	18.37 (12/24)
ChrX	110952188..110952281	<i>ALG13</i>	Exon 05	NM_001257234.1	18.37 (12/24)
ChrX	110952188..110952281	<i>ALG13</i>	Exon 05	NM_001257237.1	18.37 (12/24)
ChrX	110952188..110952281	<i>ALG13</i>	Exon 05	NM_001324292.1	18.37 (12/24)
ChrX	110952188..110952281	<i>ALG13</i>	Exon 05	NM_001324293.1	18.37 (12/24)
ChrX	118715465..118715564	<i>UBE2A</i>	Exon 04	NM_003336.3	24.30 (16/31)
ChrX	118975034..118975226	<i>UPF3B</i>	Exon 07	NM_023010.3	27.45 (14/35)
ChrX	118975034..118975226	<i>UPF3B</i>	Exon 07	NM_080632.2	27.45 (14/35)
ChrX	119660611..119660716	<i>CUL4B</i>	Exon 22	NM_003588.3	12.02 (10/13)
ChrX	119677964..119678063	<i>CUL4B</i>	Exon 09	NM_003588.3	29.27 (18/35)
ChrX	122536840..122536954	<i>GRIA3</i>	Exon 08	NM_000828.4	14.85 (10/18)
ChrX	128674412..128674460	<i>OCRL</i>	Exon 01	NM_000276.3	19.47 (14/22)
ChrX	128674412..128674460	<i>OCRL</i>	Exon 01	NM_001318784.1	19.47 (14/22)
ChrX	128674412..128674460	<i>OCRL</i>	Exon 01	NM_001587.3	19.47 (14/22)
ChrX	132833918..132834061	<i>GPC3</i>	Exon 04	NM_004484.3	37.21 (18/45)
ChrX	133594337..133594373	<i>HPRT1</i>	Exon 01	NM_000194.2	6.35 (4/10)
ChrX	135067657..135067991	<i>SLC9A6</i>	Exon 01	NM_001042537.1	81.30 (2/115)
ChrX	135067657..135067991	<i>SLC9A6</i>	Exon 01	NM_006359.2	81.30 (2/115)
ChrX	135104740..135104861	<i>SLC9A6</i>	Exon 11	NM_001042537.1	21.37 (14/25)
ChrX	135104740..135104861	<i>SLC9A6</i>	Exon 12	NM_001177651.1	21.37 (14/25)
ChrX	135104740..135104861	<i>SLC9A6</i>	Exon 12	NM_001330652.1	21.37 (14/25)
ChrX	135104740..135104861	<i>SLC9A6</i>	Exon 11	NM_006359.2	21.37 (14/25)
ChrX	135115568..135115657	<i>SLC9A6</i>	Exon 14	NM_001042537.1	18.62 (16/23)
ChrX	135115568..135115657	<i>SLC9A6</i>	Exon 15	NM_001177651.1	18.62 (16/23)
ChrX	135115568..135115657	<i>SLC9A6</i>	Exon 15	NM_001330652.1	18.62 (16/23)
ChrX	135115568..135115657	<i>SLC9A6</i>	Exon 14	NM_006359.2	18.62 (16/23)
ChrX	139585880..139587230	<i>SOX3</i>	Exon 01	NM_005634.2	48.41 (0/137)
ChrX	147582613..147582669	<i>AFF2</i>	Exon 01	NM_002025.3	25.60 (16/32)
ChrX	152954025..152954296	<i>SLC6A8</i>	Exon 01	NM_001142805.1	10.75 (0/24)
ChrX	152954025..152954296	<i>SLC6A8</i>	Exon 01	NM_005629.3	10.75 (0/24)

ChrX	153200332..153200362	<i>NAA10</i>	Exon 01	NM_003491.3	2.00 (2/2)
ChrX	153363056..153363127	<i>MECP2</i>	Exon 01	NM_001110792.1	2.65 (2/3)
ChrX	153599236..153599618	<i>FLNA</i>	Exon 02	NM_001110556.1	41.08 (8/63)
ChrX	153599236..153599618	<i>FLNA</i>	Exon 02	NM_001456.3	41.08 (8/63)
ChrX	153640176..153640294	<i>TAZ</i>	Exon 01	NM_000116.4	16.35 (7/22)
ChrX	153640176..153640294	<i>TAZ</i>	Exon 01	NM_181311.3	16.35 (7/22)
ChrX	153640176..153640294	<i>TAZ</i>	Exon 01	NM_181312.3	16.35 (7/22)
ChrX	153640176..153640294	<i>TAZ</i>	Exon 01	NM_181313.3	16.35 (7/22)
ChrX	153640176..153640348	<i>TAZ</i>	Exon 01	NM_001303465.1	17.58 (7/25)
ChrX	153774999..153775090	<i>G6PD</i>	Exon 01	NM_000402.4	4.24 (2/6)
ChrX	153784375..153784596	<i>IKBKG</i>	Exon 03	NM_001099856.4	0.00 (0/0)
ChrX	153784375..153784596	<i>IKBKG</i>	Exon 03	NM_001099857.2	0.00 (0/0)
ChrX	153784375..153784596	<i>IKBKG</i>	Exon 03	NM_001145255.2	0.00 (0/0)
ChrX	153784375..153784596	<i>IKBKG</i>	Exon 03	NM_001321396.1	0.00 (0/0)
ChrX	153784375..153784596	<i>IKBKG</i>	Exon 03	NM_001321397.1	0.00 (0/0)
ChrX	153784375..153784596	<i>IKBKG</i>	Exon 03	NM_003639.4	0.00 (0/0)
ChrX	153786742..153786870	<i>IKBKG</i>	Exon 04	NM_001099856.4	0.00 (0/0)
ChrX	153786742..153786870	<i>IKBKG</i>	Exon 04	NM_001099857.2	0.00 (0/0)
ChrX	153786742..153786870	<i>IKBKG</i>	Exon 04	NM_001145255.2	0.00 (0/0)
ChrX	153786742..153786870	<i>IKBKG</i>	Exon 04	NM_001321396.1	0.00 (0/0)
ChrX	153786742..153786870	<i>IKBKG</i>	Exon 04	NM_003639.4	0.00 (0/0)
ChrX	153786745..153786870	<i>IKBKG</i>	Exon 04	NM_001321397.1	0.00 (0/0)
ChrX	153788617..153788779	<i>IKBKG</i>	Exon 05	NM_001099856.4	0.63 (0/1)
ChrX	153788617..153788779	<i>IKBKG</i>	Exon 05	NM_001099857.2	0.63 (0/1)
ChrX	153788617..153788779	<i>IKBKG</i>	Exon 05	NM_001321396.1	0.63 (0/1)
ChrX	153788617..153788779	<i>IKBKG</i>	Exon 05	NM_001321397.1	0.63 (0/1)
ChrX	153788617..153788779	<i>IKBKG</i>	Exon 05	NM_003639.4	0.63 (0/1)
ChrX	153789898..153790004	<i>IKBKG</i>	Exon 06	NM_001099856.4	0.00 (0/0)
ChrX	153789898..153790004	<i>IKBKG</i>	Exon 06	NM_001099857.2	0.00 (0/0)
ChrX	153789898..153790004	<i>IKBKG</i>	Exon 05	NM_001145255.2	0.00 (0/0)
ChrX	153789898..153790004	<i>IKBKG</i>	Exon 06	NM_001321396.1	0.00 (0/0)
ChrX	153789898..153790004	<i>IKBKG</i>	Exon 06	NM_001321397.1	0.00 (0/0)
ChrX	153789898..153790004	<i>IKBKG</i>	Exon 06	NM_003639.4	0.00 (0/0)
ChrX	153791020..153791173	<i>IKBKG</i>	Exon 07	NM_001099856.4	0.00 (0/0)

ChrX	153791020..153791173	<i>IKBKG</i>	Exon 07	NM_001099857.2	0.00 (0/0)
ChrX	153791020..153791173	<i>IKBKG</i>	Exon 07	NM_001321396.1	0.00 (0/0)
ChrX	153791020..153791173	<i>IKBKG</i>	Exon 07	NM_001321397.1	0.00 (0/0)
ChrX	153791020..153791173	<i>IKBKG</i>	Exon 07	NM_003639.4	0.00 (0/0)
ChrX	153791769..153791921	<i>IKBKG</i>	Exon 08	NM_001099856.4	0.00 (0/0)
ChrX	153791769..153791921	<i>IKBKG</i>	Exon 08	NM_001099857.2	0.00 (0/0)
ChrX	153791769..153791921	<i>IKBKG</i>	Exon 06	NM_001145255.2	0.00 (0/0)
ChrX	153791769..153791921	<i>IKBKG</i>	Exon 08	NM_001321396.1	0.00 (0/0)
ChrX	153791769..153791921	<i>IKBKG</i>	Exon 08	NM_001321397.1	0.00 (0/0)
ChrX	153791769..153791921	<i>IKBKG</i>	Exon 08	NM_003639.4	0.00 (0/0)
ChrX	153792169..153792240	<i>IKBKG</i>	Exon 09	NM_001099856.4	12.06 (10/14)
ChrX	153792169..153792240	<i>IKBKG</i>	Exon 09	NM_001099857.2	12.06 (10/14)
ChrX	153792169..153792240	<i>IKBKG</i>	Exon 07	NM_001145255.2	12.06 (10/14)
ChrX	153792169..153792240	<i>IKBKG</i>	Exon 09	NM_001321396.1	12.06 (10/14)
ChrX	153792169..153792240	<i>IKBKG</i>	Exon 09	NM_001321397.1	12.06 (10/14)
ChrX	153792169..153792240	<i>IKBKG</i>	Exon 09	NM_003639.4	12.06 (10/14)
ChrX	153792529..153792681	<i>IKBKG</i>	Exon 10	NM_001099856.4	22.06 (7/38)
ChrX	153792529..153792681	<i>IKBKG</i>	Exon 10	NM_001099857.2	22.06 (7/38)
ChrX	153792529..153792681	<i>IKBKG</i>	Exon 08	NM_001145255.2	22.06 (7/38)
ChrX	153792529..153792681	<i>IKBKG</i>	Exon 10	NM_001321396.1	22.06 (7/38)
ChrX	153792529..153792681	<i>IKBKG</i>	Exon 10	NM_001321397.1	22.06 (7/38)
ChrX	153792529..153792681	<i>IKBKG</i>	Exon 10	NM_003639.4	22.06 (7/38)
ChrX	154005069..154005147	<i>DKC1</i>	Exon 15	NM_001142463.2	16.42 (12/18)
ChrX	154005069..154005147	<i>DKC1</i>	Exon 15	NM_001363.4	16.42 (12/18)

## TECHNICAL LIMITATIONS

mosaics (<20%); indels >21bp; repeat expansions; repetitive regions; variants in: homopolymeric regions or regions of high sequence homology, unenriched regions (untranslated regions, introns, promoter and enhancer regions) or enriched but insufficiently covered regions; variants in mt-DNA (VAF<20%); determination of the phase of multiple variants in one gene; balanced genomic rearrangements. For this case, the number of CNVs called exceeded the standard significantly, indicating that CNV detection was unreliable. Therefore, CNVs could not be evaluated.

## CLASSES OF VARIANTS

- Class 5:** pathogenic variant – are reported, posterior probability >99 %
- Class 4:** likely pathogenic variant – are reported, posterior probability >90 %
- Class 3:** uncertain significance – only be listed in the report if posterior probability is >67.5 %
- Class 2:** likely benign – not reported, posterior probability <10 %
- Class 1:** benign – not reported, posterior probability <0,1 %

## ACMG CRITERIA

### 1. Criteria for pathogenic evidence

PVS1: Null variant in a gene where loss of function (LOF) is a known mechanism of disease; PS1: same amino acid change as a previously established pathogenic variant regardless of nucleotide change; PS2/PM6: de novo in a patient with the disease and no family history; PS3: well-established functional studies supportive of a damaging effect on the gene or gene product; PS4: the prevalence of the variant in affected individuals is significantly increased compared with the prevalence in controls/was identified in unrelated affected individuals; PM1: missense variant located in a mutational hot spot and/or critical and well-established functional domain; PM2: absent from controls (or at extremely low frequency) in Genome Aggregation Database (gnomAD); PM3: for recessive disorders, detected in homozygous state or together with another (not benign or likely benign) variant; PM4: protein length changes as a result of in-frame deletions/insertions in a non-repeat region or stop-loss variants; PM5: missense change at an amino acid residue where a different missense change determined to be (likely) pathogenic has been seen before; PP1: co-segregation with disease in multiple affected family members; PP2: missense variant in a gene that has a low rate of benign missense variation and in which missense variants are a common mechanism of disease; PP3: multiple lines of computational evidence support a deleterious effect on the gene or gene product; PP4: patient's phenotype or family history is (highly) is specific for variations in the affected gene; PP5: reputable source recently reports variant as pathogenic, but the evidence is not available to the laboratory to perform an independent evaluation.

### 2. Criteria for benign evidence

BA1: allele frequency is >5% if recessive and 0.5% if dominant in gnomAD; BS1: allele frequency is greater than expected for disorder; BS2: observed in a healthy adult individual for a recessive (homozygous), dominant (heterozygous), or X-linked (hemizygous) disorder, with full penetrance expected at an early age; BS3: well-established functional studies show no damaging effect on protein function or splicing; BS4: lack of segregation with disease; BP1: missense variant in a gene for which primarily truncating variants are known to cause disease OR for loss-of-function variants in a gene where the disease is caused by gain-of-function variants; BP2: observed in trans with a pathogenic variant for a fully penetrant dominant gene/disorder or observed in cis with a pathogenic variant in any inheritance pattern; BP3: in-frame deletions/insertions in a repetitive region without a known function; BP4: multiple lines of computational evidence suggest no impact on gene or gene product; BP5: variant found in a case with an alternate molecular basis for disease; BP6: reputable source recently reports variant as benign, but the evidence is not available to the laboratory to perform an independent evaluation; BP7: a synonymous (silent) variant for which splicing prediction algorithms predict no impact to the splice consensus sequence nor the creation of a new splice site AND the nucleotide is not highly conserved.

According to Ellard et al. 2020, the strength level of criteria PVS1, PS1, PS2, PS3, PS4, PM1, PM3, PM4, PM5, PP1, PP4, BP2, and BP4 can be modified depending on the cogency of the evidence.

Report released by

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