

Table S1. Genes within mapped homozygous regions of families A3471, F874 and KKESH001-7. Note that only 1 gene (red) in each family had a homozygous variant that passed Polyphen 1 and 2 scores, segregated within the family, and was absent from >270 controls.

Family A3471 (2 sibs calculated together):

Chr	Start (hg19)	End (hg19)	Size (Mb)	List of genes in the region
11	87,286,975	95,192,462	7.91	RAB38, CTC5, GRM5, TYR, NOX4, FOLH1B, TRIM77P, TRIM49, TRIM53P, LOC399940, TRIM64B, TRIM49L1, LOC399939, TRIM64, LOC399940, TRIM53P, TRIM49L2, UBTF1, NAALAD2, CHORDC1, MIR4490, FAT3, MTNR1B, SLC36A4, CCDC67, C11orf75, KIAA1731, SCARNA9, SNORA25, SNORA32, SNORD6, SNORA1, SNORA8, SNORD5, SNORA18, MIR1304, SNORA40, TAF1D, C11orf54, MED17, VSTM5, HEPHL1, PANX1, FOLR4, GPR83, MRE11A , ANKRD49, LOC643037, FUT4, PIWIL4, AMOTL1, CWC15, KDM4D, KDM4DL, SRSF8, ENDD1, SESN3
15	67,533,543	78,711,803	11.18	AAGAB, IQCH, LOC100506886, C15orf61, MAP2K5, SKOR1, PIAS1, CALML4, CLN6, FEM1B, ITGA11, CORO2B, ANP32A, MIR4312, ANP32A-IT1, MIR548H4, SPESP1, NOX5, LINC00277, GLCE, PAQR5, KIF23, RPLP1, LOC145837, C15orf50, TLE3, MIR629, UACA, LARP6, THAP10, LRRC49, CT62, THSD4, NR2E3, MYO9A, SENP8, GRAMD2, PKM2, PAPP6, CELF6, HEXA, C15orf34, TMEEM202, ARIH1, MIR630, GOLGA6B, HIGD2B, BBS4, ADPGK, LOC100287559, NEO1, HCN4, C15orf60, NPTN, CD276, C15orf59, TBC1D21, LOC100287616, LOXL1, STOML1, PML, GOLGA6A, LOC283731, ISLR2, ISLR, STRA6, CCDC33, CYP11A1, LOC729739, SEMA7A, UBL7, LOC440288, ARID3B, CLK3, EDC3, CYP1A1, CYP1A2, CSK, MIR4513, LMAN1L, CPLX3, ULK3, SCAMP2, MPI, C15orf17, COX5A, RPP25, SCAMP5, PPCDC, C15orf39, GOLGA6C, GOLGA6D, COMMD4, NEL1, MIR631, MAN2C1, SIN3A, PTPN9, SNUPN, IMP3, SNX33, CSPG4, ODF3L1, DNMI1P35, MIR4313, UBE2Q2, FBXO22, FBXO22-AS1, NRG4, C15orf27, ETF4, TYRO3P, ISL2, SCAPER, RCN2, PSTPIP1, TSPAN3, PEAK1, C15orf5, HMG20A, LINGO1, LOC253044, LOC645752, LOC91450, TBC1D2B, SH2D7, CIB2, IDH3A, ACSBG1, DNAJA4, WDR61, CRABP1

Family F874:

Chr	Start (hg19)	End (hg19)	Size (Mb)	List of genes in the region
6	60,001	10,269,064	10.38	DUSP22, IRF4, EXOC2, HUSB1, LOC285768, FOXQ1, FOXF2, FOXC1, GMD5, C6orf195, MYLK4, WRNIP1, SERPINB1, MIR4645, MGC39372, SERPINB9, DKFZP688I152I7, NQO2, LOC401233, RIPK1, BPHL, TUBB2A, LOC100507194, TUBB2B, PSMG4, SLC22A23, C6orf145, FAM50B, PRPF46, C6orf146, C6orf201, EC12, CDYL, RPP40, PPP1R3G, LYRM4, MIR3691, FARS2, NRN1, F13A1, LY86-AS1, LY86, RREB1, SSR1, CAGE1, CAGE1, R1OK1, DSP, SNRNP48, BMP6, TXNDC5, MUTED-TXNDC5, PIP5K1P1, MUTED, EEF1E1-MUTED, EEF1E1, SCARNA27, SLC35B3, LOC100506207, HULC
7	9,952,849	13,986,939	4.03	NDUFA4, PHF14, THSD7A, TMEM106B, VWDE, SCIN, ARL4A, ETV1
8	127,063,064	128,762,864	1.70	FAM84B, PCAT1, POU5F1B, LOC727677, MYC
9	36,906,780	81,096,302	44.19	PAX5, LOC100506710, ZCCHC7, GRHRP, ZBTB5, POLR1E, FBXO10, TOMM5, FRMPD1, RG9MTD3, EXOSC3, DCAF10, MCART1, SHB, ALDH1B1, IGF1BP1, ANKRD18A, FAM201A, CNTNAP3, LOC653501, ZNF658B, FAM75A2, FAM75A1, FAM74A1, FAM75A3, FAM74A3, ZNF658, FAM75A4, MGC21881, KGFPL2, ANKRD20A2, ANKRD20A3, FAM95B1, FOXD4L4, FOXD4L2, AQP7P3, LOC642929, FAM75A6, CNTNAP3B, FAM27C, FAM27A, KGFPL1, FAM74A2, FAM74A4, LOC442421, LOC100133920, AQP7P1, FAM27B, ANKRD20A1, LOC642236, LOC100132352, PGM5P2, LOC440896, FOXD4L6, CBWD6, ANKRD20A4, FOXD4L5, CBWD3, CBWD5, FOXD4L3, LOC572558, PGM5, C9orf71, PIP5K1B, FAM122A, PRKACG, FXN, TJP2, FAM189A2, APBA1, PTAR1, LOC494558, C9orf135, MAMDC2, LOC100507244, LOC100507299, SMC5, KLF9, TRPM3, MIR204, TMEM2, FAM108B1, C9orf85, C9orf57, GDA, ZFAND5, TMC1, ALDH1A1, ANXA1, RORB, TRPM6, C9orf40, C9orf41, C9orf95, OSTF1, MIR548H3, PCSK5, RFK, RPSAP9, GCNT1, PRUNE2, PCA3, FOXB2, LOC100286938, VPS13A, GNA14, GNAQ, CEP7B, PSAT1
11	99,307,416	103,920,105	4.61	CNTN5, ARHGAP42, TMEM133, PGR, TRPC6, MIR3920, ANGPTL5, KIAA1377, C11orf70, YAP1, BIRC3, BIRC2, TMEM123, MMP7, MMP20, MMP27, MMP8, MMP10, LOC100288077, MMP1, MMP3, MMP12, MMP13, DCUN1D5, DYNC2H1, MIR4693, PDGFG, DDI1
13	80,878,033	87,062,889	6.18	SPRY2, SLITRK1, SLITRK6
15	89,327,344	97,069,505	7.74	ACAN, HAPLN3, MFG8, ABHD2, RLBP1, FANCI, POLG, MIR9-3, LOC254559, RHCG, LOC283761, C15orf42, KIF7, PLIN1, PEX11A, WDR93, MESP1, MESP2, ANPEP, AP3S2, C15orf38-AP3S2, C15orf38, ZNF710, IDH2, SEMA4B, CIB1, C15orf58, TLL13, NGRN, GABARAPL3, ZNF774, IQGAP1, CRTCS, BLM, FURIN, FES, MAN2A2, UNC45A, HDDC3, RCCD1, PRC1, VPS33B, SV2B, SLC03A1, ST8SIA2, C15orf32, LOC100144604, FAM174B, ASB9P1, LOC100507217, CHD2, MIR3175, RGMA, MCTP2, LOC400456, LOC145820, MIR1469, NR2F2
16	27,139,829	80,352,195	53.21	JMJD5, NSMCE1, FLJ21408, IL4R, IL21R1, LOC283888, GTF3C1, KIAA0556, GSG1L, XPO6, SBK1, EIF3CL, EIF3C, CLN3, APOBR, IL27, NUPR1, CCDC101, SULT1A2, SULT1A1A, ATXN2L, TUFM, MIR4721, SH2B1, ATP2A1, RABEP2, CD19, NFATC2IP, MIR4517, SPNS1, LAT, RRN3P2, RUNCDC2C, LOC606724, BOLA2B, BOLA2, SLX1B, SLX1A, SLX1B-SULT1A4, SLX1A-SULT1A3, SULT1A3, SULT1A4, LOC388242, LOC613038, LOC440354, SLC7A5P1, SPN, QPRT, C16orf54, ZG16, KIF22, MAZ, PRRT2, C16orf53, MVP, CDIPT, LOC440356, SEZ6L2, ASPHD1, KCTD13, TMEM219, TAOK2, HIRIP3, INO80E, DOC2A, C16orf92, FAM57B, ALDOA, PPP4C, TBX6, YPEL3, GDDP3, MAPK3, LOC100271831, CORO1A, LOC613037, LOC595101, CD2BP2, TBC1D10B, MYLPP, 1-Sep, ZNF48, ZNF771, DCTPP1, SEPHS2, ITGAL, MIR4518, ZNF768, ZNF747, ZNF764, ZNF688, ZNF785, ZNF689, PRR14, FBRS, SRCAP, SNORA30, PHKG2, C16orf93, RNF40, ZNF629, MIR4519, BCL7C, MIR762, CTF1, FBXL19-AS1, FBXL19, ORAI3, SETD1A, HSD3B7, STX1B, STX4, ZNF668, ZNF646, PRSS53, VKORC1, BCKDK, KAT8, PRSS8, PRSS36, FUS, PYCARD, TRIM27, PYDC1, ITGAM, ITGAX, ITGAD, COX6A2, ZNF843, ARMC5, TGFBI1, SLC5A2, C16orf58, AHSP, CSDAP1, KIAA0664L3, ZNF720, ZNF267, HERC2P4, LOC729264, LOC330705, TP53TG3, SLC6A10P, LOC653550, TP53TG3B, LINC00273, UBE2MP1, LOC283914, LOC146481, LOC100130700, FLJ26245, ANKRD28P1, SHCBP1, VPS35, ORC6, MYLK3, C16orf87, GPT2, DNAJA2, NETO2, ITFG1, PHKB, ABCC12, ABCC11, MIR548E2, LONP2, LOC100507577, SIAH1, N4BP1, CBLN1, C16orf78, ZNF423 , TMEM188, HEATR3, PAPDS, ADCY7, BRD7, NKD1, SNX20, NOD2, CYLD, SALL1, LOC388276, LOC100505619, TOX3, LOC643714, CHD9, LOC643802, RBL2, AKTIP, RPRGRI1P, FTO, IRX3, CRNDE, IRX5, IRX6, MMP2, LPTA2, CAPNS2, SLC6A2, CES1P2, CES1P1, CES1, CESSA, LOC283856, GNAO1, DKFZP434H168, MIR3935, AMFR, NUDT21, OGFD01, BBS2, MT4, MT3, MT2A, MT1L, MT1M, MT1J, MT1P, MT1A, MT1D, MT1B, MT1F, MT1G, MT1H, MT1I, MT1X, NUP93, MIR138-2, SLC12A3, HERPUD1, CETP, NLRCS, CPN2E, FAM192A, RSPRY1, ARL2BP, PLLP, CCL22, CX3CL1, CCL17, CIAPIN1, COO9, POLR2C, DOK4, CCDC102A, GPR114, GPR56, GPR97, CCDC135, KATNB1, KIFC3, CNGB1, TEPP, ZNF319, C16orf57, MMP15, C16orf80, CSNK2A2, CCDC113, PRSS54, GINS3, NDRG4, SETD6, CNOT1, SNORA46, SNORA50, SLC38A7, G0T2, LOC644649, CDH8, CDH11, LOC283867, CDH5, BEAN1, TK2, CKLF, CKLF-CMTM1, CMTM1, CMTM2, CMTM3, CMTM4, DYNC1L2, CCDC79, NAE1, CA7, PDP2, CDH16, RRAD, FAM96B, CES2, CES3, CES4A, CBF6, C16orf70, B3GNT9, TRADD, FBXL8, HSF4, NOL3, KIAA0895L, EXOC3L1, E2F4, ELMO3, MIR328, LRRC29, TMEM208, FHOD1, SLC9A5, PLEKHG4, KCTD19, LRRC36, TPPP3, ZDHHC7, HSD11B2, ATP6VD01, AGRP, FAM65A, CTCF, RLTPR, ACD, PARD6A, C16orf48, C16orf86, GFOD2, RANBP10, TSNAXIP1, CENPT, THAP11, NUTF2, EDC4, NRN1L, PSKH1, CTRL, PSMB10, LCAT, SLC12A4, DPEP3, DPEP2, DDX28, DUS2L, NFATC3, ESRP2, PLA2G15, SLC7A6, SLC7A6OS, PRMT7, SMPD3, ZFP90, CDH3, CDH1, TMC07, HAS3, CHTF8, CIRH1A, SNTB2, VPS4A, COG8, PDF, NIP7, TMED6, TERF2, CYB5B, MIR1538, NFAT5, NQO1, NOB1, WWP2, MIR140, CLEC18A, PDXC2P, MIR1972-1, MIR1972-2, PDPR, CLEC18C, LOC729513, EXOSC6, AARS, DDX19B, LOC100506083, DDX19B, ST3GAL2, FUK, COG4, SF3B3, SNORD111B, SNORD111, IL34, MTSS1L, VAC14, LOC100130894, HYDIN, FTSJD1, CALB2, ZNF23, ZNF19, CHST4, TAT, MARVELD3, PHLPP2, SNORA70D, AP1G1, SNORD71, ATXN1L, ZNF821, IST1, PKD1L3, DHODH, HP, HPR, TXNL4B, DHX38, PMFBP1, ZFHX3, HTA, LOC100506172, PSMDD7, LOC283922, CLEC18B, GLG1, RFWD3, MLKL, FA2H, WDR59, ZNRF1, LDHD, ZFP1, CTRB2, CTRB1, BCAR1, CFPD1, TMEM170A, CHST6, CHST5, TMEM231, GABARAPL2, ADAT1, KARS, TERF2IP, CNTNAP4, MIR4719, MON1B, SYCE1L, ADAMTS18, NUDT7, VAT1L, CLEC3A, WWOX, MAF
18	65,627,833	73,086,805	7.46	TMX3, CCDC102B, DOK6, CD226, RTTN, SOCS6, LOC100505776, CBLN2, NETO1, LOC400655, LOC100505817, FBXO15, C18orf55, CYB5A, C18orf63, FAM69C, CNDP2, CNDP1, LOC400657, ZNF407, ZADH2, TSHZ1

Family KKESH001-7:

Chr	Start (hg19)	End (hg19)	Size (Mb)	List of genes in the region
1	30,581,567	43,171,822	12.59	MATN1, LOC100129196, LAPTM5, MIR4420, SDC3, PUM1, SNORD103A, SNORD103B, SNORD85, PRO0611, NKAIN1, SNRNP40, ZCCHC17, FABP3, SERINC2, LOC149086, LOC284551, TINAGL1, HCRTR1, PEF1, COL16A1, BAI2, MIR4254, SPOCD1, PTP4A2, KHDRBS1, TMEM39B, KPNIA6, TXLNA, CCDC28B, IQCC, CCDC2B, TMEM234, EIF3I, MTMR9LP, FAM167B, LCK, HDAC1, MARCKSL1, LOC100128071, TSSK3, BSDC1, ZBTB88, ZBTB8A, ZBTB80S, RBBP4, SYNC, KIAA1522, YARS, S100PBP, FNDCC5, HPCA, TMEM54, RNF19B, AK2, ADC, TRIM62, ZNF362, PHC2, MIR3605, ZSCAN20, CSMD2, HMG84, LOC402779, C1orf94, GJB5, GJB4, GJB3, GJA4, C1orf121, DLGAP3, LOC653160, ZMYM6NB, ZMYM6, ZMYM1, SFPO, ZMYM4, KIAA0319L, NCDN, TFAP2E, PSMB2, C1orf126, CLSPN, EIF2C4, EIF2C1, EIF2C3, TEK2, ADPRHL2, COL8A2, TRAPP3C, MAP7D1, THRAPP3, SH3D21, FAM176B, STK40, LSM10, OSCP1, MRPS15, CSF3R, GRIK3, MIR4255, LOC728431, ZC3H12A, MEAF6, SINP1, DNALI1, HNTA, RSP01, C1orf109, CDCAB, EPHA10, MANEAL, YRDC, C1orf122, MTF1, INPP5B, SF3A3, FHL3, UTP11L, POU3F1, MIR3659, LOC339442, RRAGC, GJA9-MYCBP, GJA9, MYCBP, RHBDL2, AKRIN1, NDUFS5, MACF1, KIAA0754, BMP8A, PPIEL, PABPC4, SNORA55, HEYL, NTSICA1, HPCAL4, PPIE, BMP8B, OXC72, TRIT1, MYCL1, MFSDD2A, CAP1, PPT1, RLF, TMC02, ZMPSTE24, COL9A2, SMAP2, ZNF643, ZNF642, DEM1, ZNF684, RIMS3, LOC100130557, NFYC, MIR30E, MIR30C1, KCNQ4, CITED4, CTPS, LOC100507178, SLFN1L, SCMH1, EDN2, HIVEP3, GUCAC2B, GUCAC2A, FOXJ3, RIMKLA, ZMYND12, PCCS, CCDC30, PPIH, YBX1

19	48,317,892	56,863,306	8.55	<p>CRX, SULT2A1, SNAR-A3, SNAR-A4, SNAR-A5, SNAR-A7, SNAR-A11, SNAR-A9, SNAR-A6, SNAR-A8, SNAR-A10, SNAR-A14, SNAR-A1, SNAR-A2, SNAR-C4, SNAR-A12, SNAR-A13, SNAR-C3, SNAR-C2, SNAR-C5, SNAR-C1, BSPH1, ELSBPB1, CABP5, PLA2G4C, LIG1, CARD8, LOC100505812, ZNF114, CCDC114, EMP3, TMEM143, SYNGR4, KDELR1, GRIN2D, GRWD1, KCNU14, CYTH2, LMTK3, SULT2B1, FAM83E, SPACA4, RPL18, SPHK2, DBP, CA11, SEC1, NTN5, FUT2, MAMSTR, RASIP1, IZUMO1, FUT1, FGF21, BCAT2, HSD17B14, PLEKHA4, PPP1R15A, TULP2, NUCB1, DHDH, BAX, FTL, GYS1, RUVBL2, LHB, CGB, SNAR-G2, CGB2, CGB1, SNAR-G1, CGB5, CGB8, CGB7, NTF4, KCNA7, SNRNP70, LIN7B, C19orf73, PPIA3, HRC, TRPM4, SLC6A16, MIR4324, CD37, TEAD2, DKK1, CCDC155, PTH2, LOC100507003, SLC17A7, PIH1D1, ALDH16A1, FLT3LG, RPL13A, RPL13AP5, SNORD32A, SNORD33, SNORD34, SNORD35A, RPS11, SNORD35B, MIR150, FCGRT, RCN3, NOSIP, PRRG2, PRR12, RRAS, SCAF1, IRF3, BCL2L12, PRMT1, C19orf76, CPT1C, TSKS, AP2A1, FUZ, MED25, LOC100506033, PTOV1, MIR4749, PINKP, AKT1S1, TBC1D17, MIR4750, IL4I1, NUP62, ATF5, MIR4751, SIGLEC11, SIGLEC16, VRK3, ZNF473, FLJ26850, SNAR-B2, SNAR-B1, SNAR-D, IZUMO2, MYH14, KCNC3, NAPS8, NAPS4, NR1H2, POLD1, SPIB, MYBPC2, FAM71E1, C19orf63, JOSD2, ASPDH, LRRC4B, SNAR-F, SYT3, C19orf81, SHANK1, CLEC11A, GPR32, ACPT, C19orf48, SNORD88B, SNORD88A, SNORD88C, MGC45922, KLK1, KLK15, KLK3, KLK2, KLKP1, KLK4, KLK5, KLK6, KLK7, KLK8, KLK9, KLK10, KLK11, KLK12, KLK13, KLK14, CTU1, SIGLEC9, SIGLEC7, SIGLECP3, CD33, C19orf75, IGLON5, VSIG10L, ETFB, CLDN2, NKG7, LIM2, LOC147646, SIGLEC10, LOC100129083, SIGLEC8, CEACAM18, SIGLEC12, SIGLEC6, ZNF175, FLJ30403, SIGLEC5, SIGLEC14, MIR99B, MIRLETTE, MIR125A, LINC00085, HAS1, FPR1, FPR2, FPR3, ZNF577, ZNF649, ZNF613, ZNF350, ZNF615, ZNF614, ZNF432, ZNF841, ZNF616, ZNF836, PPP2R1A, ZNF766, MIR643, ZNF480, ZNF610, ZNF880, ZNF528, ZNF534, ZNF578, ZNF808, ZNF701, ZNF137P, ZNF83, ZNF611, ZNF600, ZNF28, ZNF468, ZNF320, ZNF816-ZNF321P, ZNF321P, ZNF816, ZNF702P, ERVV-1, ERVV-2, ZNF160, ZNF415, ZNF347, ZNF665, ZNF677, VN1R2, VN1R4, LOC646508, BIRC8, ZNF845, ZNF525, ZNF765, LOC147804, ZNF761, ZNF813, ZNF331, LOC284379, DPRX, MIR512-2, MIR512-1, MIR1323, MIR498, MIR520E, MIR519E, MIR520F, MIR515-1, MIR515-2, MIR519C, MIR1283-1, MIR520A, MIR526B, MIR519B, MIR525, MIR523, MIR518F, MIR520B, MIR518B, MIR526A1, MIR520C, MIR518C, MIR524, MIR517A, MIR519D, MIR521-2, MIR520D, MIR517B, MIR520G, MIR516B2, MIR526A2, MIR518E, MIR518A1, MIR518D, MIR516B1, MIR518A2, MIR517C, MIR520H, MIR521-1, MIR522, MIR519A1, MIR527, MIR516A1, MIR1283-2, MIR516A2, MIR519A2, MIR371, MIR371B, MIR372, MIR373, NLRP12, MYADM, PRKCG, CACNG7, CACNG8, MIR935, CACNG6, VSTM1, TARM1, OSCAR, NDUFA3, TFPT, PRPF31, CNOT3, LENG1, TMC4, MBOAT7, TSEN34, RPS9, LILRB3, LILRA6, LILRB5, LILRB2, MIR4752, LILRA3, LILRA5, LILRA4, LAIR1, TTYH1, LENG8, LENG9, CDC42EP5, LAIR2, KIR3DX1, LILRA2, LILRA1, LILRB1, LILRB4, LILRP2, KIR3DL3, KIR2DL3, KIR2DL1, KIR2DL4, KIR3DL1, KIR2DS4, KIR3DL2, FCAR, NCR1, NLRP7, NLRP2, GP6, RDH13, EPS8L1, PPP1R12C, TNNT1, TNNT3, C19orf51, SYT5, PTPRH, TMEM86B, PPP6R1, HSPBP1, BRSK1, TMEM150B, SUV420H2, COX6B2, FAM71E2, IL11, TMEM190, TMEM238, RPL28, UBE2S, SHISA7, ISOC2, ZNF628, NAT14, SSC5D, SBK2, SGK110, ZNF579, FIZ1, ZNF524, ZNF685, ZNF784, ZNF580, ZNF581, CCDC106, U2AF2, EPN1, NLRP9, RFLP4A, NLRP11, NLRP4, NLRP13, NLRP8, NLRP5, ZNF787, ZNF444, GALP, ZSCAN5B, ZSCAN5A</p>
20	4,155,948	19,897,944	15.74	<p>SMOX, LOC728228, ADRA1D, PRNP, PRND, PRNT, RASSF2, SLC23A2, C20orf30, PCNA, PCNA-AS1, CDS2, PROKR2, LOC100507629, LOC643406, LOC149837, GPCPD1, C20orf196, CHGB, TRMT6, MCM8, CRLS1, LRRN4, FERMT1, BMP2, HAO1, TMX4, PLCB1, PLCB4, C20orf103, PAK7, LOC100131208, ANKRD5, SNAF25, MKKS, C20orf94, JAG1, LOC339593, BTBD3, SPTLC3, ISM1, LOC100505536, TASP1, ESF1, C20orf7, SEL1L2, FLRT3, MACROD2-AS1, MACROD2, SNRPB2, OTOR, PCSK2, Bfsp1, DSTN, RRPB1, BANF2, SNX5, SNORD17, C20orf72, OVOL2, PET117, CSR2BP, ZNF133, DZANK1-AS1, DZANK1, POLR3F, MIR3192, RBBP9, SEC23B, LINC00493, DTD1, HSPC072, LOC100270804, LOC100128496, C20orf79, SLC24A3, LOC100130264, RIN2</p>
	47,831,904	52,868,496	5.04	<p>DDX27, ZNF1, ZNF1-AS1, SNORD12C, SNORD12B, SNORD12, KCNB1, PTGIS, B4GALT5, SLC9A8, SPATA2, RNF114, SNAI1, UBE2V1, TMEM189-UBE2V1, TMEM189, CEBPB, LOC284751, PTPN1, MIR645, FAM65C, PARD6B, BCAS4, ADNP, DPM1, MOCS3, KCNG1, NFATC2, MIR3194, ATP9A, SALL4, ZFP64, TSH22, ZNF217, SUMO1P1, BCAS1, MIR4756, CYP24A1, PFDN4</p>

TABLE S2: Topline statistics on evaluation of 32 wholehuman exomes, including the exomes described in the accompanying paper.

Statistics are shown for genetic variants grouped by both, novelty (absence from dbSNP132) and filtration status. (See tables on page 2 for legends on abbreviations.)

<u>CompOverlap</u>										
CompOverlap	CompRod	EvalRod	Filter	Novelty	nEvalVariants	novelSites	nVariantsAtComp	compRate	nConcordant	concordantRate
CompOverlap	dbSNP	eval	called	all	267927	76951	190976	71.2791171	188044	98.46472855
CompOverlap	dbSNP	eval	called	known	190976	0	190976	100	188044	98.46472855
CompOverlap	dbSNP	eval	called	novel	76951	76951	0	0	0	0
CompOverlap	dbSNP	eval	filtered	all	86826	64593	22233	25.8063852	20514	92.2682499
CompOverlap	dbSNP	eval	filtered	known	22233	0	22233	100	20514	92.2682499
CompOverlap	dbSNP	eval	filtered	novel	64593	64593	0	0	0	0
CompOverlap	dbSNP	eval	raw	all	354753	141544	213209	60.1006898	208558	97.81857239
CompOverlap	dbSNP	eval	raw	known	213209	0	213209	100	208558	97.81857239
CompOverlap	dbSNP	eval	raw	novel	141544	141544	0	0	0	0

<u>CountVariants</u>																												
CountVariants	CompRod	EvalRod	Filter	Novelty	nProcessedLoci	nCalledLoci	nRefLoci	nVariantLoci	variantRate	variantRatePerBp	nSNPs	nMNs	nInsertions	nDeletions	nComplex	nMixed	nNoCalls	nHets	nHomRef	nHomVar	nSingletons	nHomozygous	nHeterozygous	heterozygosityPerBp	hetHomRatio	indelRatePerBp	indelRateRatio	deletionInsertionRatio
CountVariants	dbSNP	eval	called	all	3101804739	267927	0	267927	8.64E-05	11577	235409	0	14766	17752	0	0	62270	1553758	5901022	1056614	79063	0	0.000501	1996	1.470507	1.05E-05	95387	1.202221
CountVariants	dbSNP	eval	called	known	3101804739	190976	0	190976	6.16E-05	16241	178423	0	5556	6997	0	0	42236	1328752	3753984	986260	30485	0	0.000428	2334	1.347263	4.05E-06	247096	1.259359
CountVariants	dbSNP	eval	called	novel	3101804739	76951	0	76951	2.48E-05	40308	56986	0	9210	10755	0	0	20034	225006	2147038	70354	48578	0	7.25E-05	13785	3.198198	6.44E-06	155362	1.167752
CountVariants	dbSNP	eval	filtered	all	3101804739	86826	0	86826	2.8E-05	35724	60496	0	14042	12288	0	0	82831	772901	1836747	85953	17179	0	0.000249	4013	8.992135	8.49E-06	117804	0.875089
CountVariants	dbSNP	eval	filtered	known	3101804739	22233	0	22233	7.17E-06	139513	19176	0	1054	2003	0	0	14140	260374	375571	61371	1873	0	8.39E-05	11912	4.242623	9.9E-07	1014656	1.90038
CountVariants	dbSNP	eval	filtered	novel	3101804739	64593	0	64593	2.08E-05	48020	41320	0	12988	10285	0	0	68691	512527	1461176	24582	15306	0	0.000165	6051	20.84969	7.5E-06	133279	0.791885
CountVariants	dbSNP	eval	raw	all	3101804739	354753	0	354753	0.000114	8743	295905	0	28808	30040	0	0	145101	2326659	7737769	1142567	96242	0	0.000075	1333	2.036344	1.9E-05	52708	1.042766
CountVariants	dbSNP	eval	raw	known	3101804739	213209	0	213209	6.87E-05	14548	197599	0	6610	9000	0	0	56376	1589126	4129555	1047631	32358	0	0.000512	1951	1.516876	5.03E-06	198706	1.361573
CountVariants	dbSNP	eval	raw	novel	3101804739	141544	0	141544	4.56E-05	21914	98306	0	22198	21040	0	0	88725	737533	3608214	94936	63884	0	0.000238	4205	7.768739	1.39E-05	71737	0.947833

<u>TiTvVariantEvaluator</u>													
TiTvVariantEvaluator	CompRod	EvalRod	Filter	Novelty	nTi	nTv	tiTvRatio	nTilnComp	nTvlnComp	TiTvRatioStandard	nTiDerive	nTvDerive	tiTvDerivedRatio
TiTvVariantEvaluator	dbSNP	eval	called	all	168111	67298	2.49800886	128769	52921	2.43323066	0	0	0
TiTvVariantEvaluator	dbSNP	eval	called	known	127923	50500	2.53312871	127746	51671	2.47229587	0	0	0
TiTvVariantEvaluator	dbSNP	eval	called	novel	40188	16798	2.39242767	1023	1250	0.8184	0	0	0
TiTvVariantEvaluator	dbSNP	eval	filtered	all	31482	29014	1.08506238	12736	9367	1.35966692	0	0	0
TiTvVariantEvaluator	dbSNP	eval	filtered	known	11274	7902	1.42672741	11683	8066	1.44842549	0	0	0
TiTvVariantEvaluator	dbSNP	eval	filtered	novel	20208	21112	0.95718075	1053	1301	0.8093774	0	0	0
TiTvVariantEvaluator	dbSNP	eval	raw	all	199593	96312	2.07235858	1480263	8700435	1.70130149	0	0	0
TiTvVariantEvaluator	dbSNP	eval	raw	known	139197	58402	2.38342865	139429	59737	2.33404758	0	0	0
TiTvVariantEvaluator	dbSNP	eval	raw	novel	60396	37910	1.59314165	14662634	8640698	1.69692703	0	0	0

LEGENDS TO ABBREVIATIONS

1) CompOverlap : The overlap between eval and comp sites column descriptions:

nEvalSNPs	number of eval SNP sites
nCompSNPs	number of comp SNP sites
novelSites	number of eval sites outside of comp sites
nVariantsAtComp	number of eval sites at comp sites (that is, sharing the same locus as a variant in the comp track, regardless of whether the alternate allele is the same)
compRate	percentage of eval sites at comp sites
nConcordant	number of concordant sites (that is, for the sites that share the same locus as a variant in the comp track, those that have the same alternate allele)
concordantRate	the concordance rate

3) TiTvVariantEvaluator : Ti/Tv (Transition/Transversion) Variant Evaluator column descriptions:

nTi	number of transition loci
nTv	number of transversion loci
tiTvRatio	the transition to transversion ratio
nTiInComp	number of comp transition sites
nTvInComp	number of comp transversion sites
TiTvRatioStandard	the transition to transversion ratio for comp sites

2) CountVariants : Counts different classes of variants in the sample column descriptions:

nProcessedLoci	Number of processed loci
nCalledLoci	Number of called loci
nRefLoci	Number of reference loci
nVariantLoci	Number of variant loci
variantRate	Variants per loci rate
variantRatePerBp	Number of variants per base
nSNPs	Number of snp loci
nInsertions	Number of insertion
nDeletions	Number of deletions
nComplex	Number of complex loci
nNoCalls	Number of no calls loci
nHets	Number of het loci
nHomRef	Number of hom ref loci
nHomVar	Number of hom var loci
nSingletons	Number of singletons
heterozygosity	heterozygosity per locus rate
heterozygosityPerBp	heterozygosity per base pair
hetHomRatio	heterozygosity to homozygosity ratio
indelRate	indel rate (insertion count + deletion count)
indelRatePerBp	indel rate per base pair
deletionInsertionRatio	deletion to insertion ratio