

Rozen et al., Table S4. Detected sequence variants in Y-chromosomal single-copy genes and pseudogenes.

Variant ID ^a	Gene or pseudogene name	Exon number	Se-quence class ^b	Sequence to left of variant	Variants	Sequence to right of variant	Mnemonic variant ID ^{c,d}
PAGE S00004	AMELY	2	I	GGCACTTTATGGTGCACATG	T C	TTGAGAAGAGATGAGAAAAG	AMELY-12-43
PAGE S00001	AMELY	4	4	TATCTTCTCTTAAGGTGCT	C A	ACCCCTTTGAAGTGGTACCA	AMELY+108
PAGE S00002	AMELY	4	0	ACCACCAAATCATCCCCGTG	G A	TGTCCCAACAGCACCCCTG	AMELY+208
PAGE S00003	AMELY	4	0	GCCTCCAATGTCCCCCTGC	G A	GCCCTGCCCCCATACTTC	AMELY+497
PAGE S00005	APXLP	4	P	GGTGGTCCCACCCGCTCTGC	A C	TCAGAGCCTGAAGACAGTTC	APXLY+640
PAGE S00006	APXLP	4	P	ACAGCCTGGACCCCTCTGGG	C T	GCCTCTTGGTGCCCAAGTCC	APXLY-875
PAGE S00010	ARSDP	2	P	GAATCTTTGCCGGTGTCTACT	G C	TTTTTATGCTTGTCTTCTGAA	ARSDP+72
PAGE S00008	ARSDP	3	P	ATGACATTTGTGTGCTTTGTC	T C	CAGAATGCCAAATATTGACC	ARSDP+201-4
PAGE S00009	ARSDP	8	P	GGTGGGAAGGGGATAGGAGG	A G	TGGGAAGATGGGATCCATGT	ARSDP+598
PAGE S00011	ARSDP	9	P	TCTGCTCTGTGTTCTCTTCA	C T	GGAATCAGGGTGATCGACAA	ARSDP+742-9
PAGE S00095	ARSDP	9	P	CTTACCGAATCAGGGTGAT	C T	GACACCCGAGCTTGGTGCC	ARSDP+748
PAGE S00007	ARSDP	10	P	AAGGGTAGGTGCCGCGTGT	C T	GAAGCATTGGCAGACCCCTGA	ARSDP+1089
PAGE S00025	BCORP	4	P	CAAGTGGGCCAACCTCCAAC	C T	GTGCCTGTCAAGCAAAAGCAG	KIAA1575Y+687
PAGE S00026	BCORP	4	P	TTTCTACCTGCCATCACCT	C T	GCTATTTTCAGTCCCACATC	KIAA1575Y-377
PAGE S00027	BCORP	4	P	GGCTCCTGTTCGCCAGAAC	A T	CTGTCTTCTAGTTTGAGCA	KIAA1575Y-856
PAGE S00106	BCORP	4	P	ACTGATCATGGACCACACAG	G A	CCTGAAACAGGAAGGGCTCC	KIAA1575Y-891
PAGE S00107	BCORP	4	P	AAATGCTTTGCACACAGCAG	A G	AACACTTGGCTTGGACAGGC	KIAA1575Y-631
PAGE S00108	BCORP	4	P	AGGTGATGGACTGGATGATA	A G	TACAACAGCAAAGTTCTTTT	KIAA1575Y+472
PAGE S00109	BCORP	4	P	CAGAGAAAGATCCCATTCCC	A C	TGAAAGAGCCTGCCACAAAG	KIAA1575Y+1552
PAGE S00019	BCORP	10	P	CCTAGAGCAGCTGCTGCTTC	C T	AGCTGGAAGCAGGGCTTGC	FLJ20285+47+42
PAGE S00020	BCORP	12	P	TGCATGAGTTTAAATCCCACC	G A	CACATCTCTTACTAGGCCTC	FLJ20285+48-16
PAGE S00018	BCORP	13	P	AAATGCTCTTGGGAGTCTCTA	C T	GGCAGCTCTGTCTGTGGTGA	FLJ20285+255
PAGE S00043	CYORF15A	3	0	GCAAGCCCTAAACACTCTTT	C T	GACTCCAGAGGAGAAGCTGG	M61/CYORF15A+326
PAGE S00096	CYORF15A	3	0	ATTATTTGATGCAAGCCCTAA	A C	CACCTTTTCGACTCCAGAGG	M159/CYORF15A+317
PAGE S00012	CYORF15B	2	I	GGTATTTACATATTTTTAAT	A T	GAATATTTATGTA AAAAGTAA	CYORF15B+126+20
PAGE S00098	CYORF15B	3	I	GAAGAGGTAAAGAGGTTTTA	C T	GTGACTAAATAATGAGTACT	M118/CYORF15B+222+15
M191	DDX3Y	2	I	TTTTCTTTACAACCTTGA	G T	ATGAAAATATGAGATATTTT	M191/DDX3Y+103+108
PAGE S00099	DDX3Y	5	0	TTAACCTTTGAGAAATATGAT	G A	ATATACCAGTAGAGGCAACC	DDX3Y+487
PAGE S00014	DDX3Y	7	I	AAACTAAATCAGACAGTTTA	G A	TTGGTTACTTCCATTAATAT	DDX3Y+538-124
PAGE S00015	DDX3Y	8	I	AGCCCTGTTGACTTTTCTAA	C T	GGATGCCAGATACACCTTAT	DDX3Y+759+72
PAGE S00029	DDX3Y	8	I	AATTC AAGGGCATTAGAAC	A C	CTTTGTCTCTGTTAATAT	M173/DDX3Y+674-52
PAGE S00100	DDX3Y	11	I	GTAATTAGTTTCTCAGATCT	A G	ATAATCCAGTATCAACTGAG	DDX3Y+1020-35
PAGE S00101	DDX3Y	11	0	GCCTTATAATTTTTCAGGGA	G C	TGATTCACTTACTTTAGTGT	DDX3Y+1313
PAGE S00102	EIF1AY	3	I	CTTTTCAATAAAAAATTTGCC	G A	CAAAAAATGTCTCTGCTTTA	EIF1AY+204+37
PAGE S00103	EIF1AY	4	I	TTTTTTTTTTCACACTTGGT	G A	AATAGTATACCTGCTTTGTG	EIF1AY+205-93
PAGE S00016	EIF1AY	5	I	AATTTGAAAGTAACTTGTGA	A C	ACAACCTGGTATATTTTGGT	EIF1AY+256-57
PAGE S00017	EIF1AY	5	I	TTTGGAAAAGATATTAATAA	C T	TGGAATCCTCTAATAAAAAC	EIF1AY+337+183
PAGE S00104	EIF1AY	6	I	AATTTGGTGGGACTACAGT	A G	TATAATCAGTATGTCTGTTA	EIF1AY+338-33
PAGE S00044	KDM5D	4	I	TTCATCCATATTTGTTCTTA	T C	ACTTTCAGATTGTGATTGAG	M64/KDM5D+352-9
PAGE S00061	KDM5D	7	I	CCATTTGCTCTTCTGCGCC	A G	TACCACACTCACGTA AAAA	KDM5D+658-160
PAGE S00045	KDM5D	10	I	GTCTTGCTGAAATATATTTT	A G	TTTCAGGAGTGTAAACAGCC	M69/KDM5D+1093-7
PAGE S00046	KDM5D	10	I	TTCTGTGTGGTAGTCTTAG	T G	TCTCATGGAGACATGAGTCC	M70/KDM5D+1212+51
PAGE S00116	KDM5D	10	I	TAGTTCTCATGGAGACATGA	G A	TCCAAAGTATAGTGGGCTAT	KDM5D+1213-48
PAGE S00117	KDM5D	12	I	ACTTGAGCTGTCTTCTGCT	A G	CGTGAGCAGGGGTGAGCCGA	KDM5D+1554-10
PAGE S00058	KDM5D	14	I	TCCTACATCTGTTACAGCAT	C G	TAGAGACATCTTTGAGTTCA	KDM5D+1717-76
PAGE S00059	KDM5D	23	0	TGCAGACGCTGGCTCAGACA	G A	CACCAAGCGTAGCCGTTGGA	KDM5D+3296
PAGE S00119	KDM5D	23	4	CCGTGTGCAGACGCTGGCTC	A G	GACAGCACCAAGCGTAGCCG	KDM5D+3291
M50	KDM5D	24	I	GGAAAGGGCTCTGGTAAGAC	A G	GGTGTGGTTTGGGTAGGCTG	M50/KDM5D+4069+8
M54	KDM5D	24	4	GTGGCTGCAGAGGCTGCC	C T	GTGCGGCTGCCTGAGGGTGA	M54/KDM5D+3768
PAGE S00041	KDM5D	24	4	CAAGGGCTGCTGGAGAATGG	A G	GACAGTGTGACCACTCTGA	M49/KDM5D+4023
PAGE S00042	KDM5D	24	I	CTCTGATCCCTGTGGAAGC	C T	TGTGTCTACTCTGCTTCAAG	M51/KDM5D+4000-114
PAGE S00060	KDM5D	24	0	GATTATGTCTCAGGTGGGCC	G A	AGAAGAAGAACATTATCAGG	KDM5D+4433
PAGE S00023	KALP	7	P	TGGGGTCTCCCGAGGAGCC	G A	GACATCCTTGTGCATCACTA	KALP-260
PAGE S00105	KALP	7	P	GACATCCTTGTGCATCACTA	C G	AATGTCTTTTGGAGCTGGAC	KALP-239
PAGE S00024	KALP	11	P	ATCCTTATTTGGATGGAATAT	G T	ATTTCAATTTCTGCCAGTGT	KALP-65-76
rs56217212	KALP	11	P	AATATTTATTTCAATTTCTTGC	C A	AGTGTTTTTTAATCCCTAAT	KALP-65-61
PAGE S00021	KALP	12	P	TTCTTCTGAGTGTATCTTTT	G C	CAAGTATTAACACACACAA	KALP+327+146
PAGE S00022	KALP	13	P	CCAGATGTGCAAGCTCCAC	C T	CTCTTCAGCACACAGTGAGT	KALP+455
PAGE S00048	NLGN4Y	3	I	ATGTAATAGCATTAGTCAAG	T A	TACTTAGAAACTCTTATCAA	NLGN4Y-32-164
PAGE S00047	NLGN4Y	4	4	TCTGTAGGGTTTTTAAGTAC	C T	GGTGACCAGGCAGAAAAGG	NLGN4Y+135
PAGE S00110	NLGN4Y	5	4	TGGACGAACCTCGCCAAAAC	T C	GGGTACGTTCTTCTTCAATG	NLGN4Y+1095
PAGE S00049	PRKY	2	I	CAGGAGACCTAATGACCTCT	G A	TCGATTTCTTCTCTCTCCAG	PRKY+167-21
PAGE S00050	PRKY	7	I	ATTCTGTACACTGACGACAC	G A	TGTTGTTTCTTTTGAAGAAC	PRKY+816-18
PAGE S00052	RPS4Y1	5	I	TATAACTTTGTTTTTGTGG	T G	TTTTTTTTTGTTTTTTTAGT	RPS4Y1+81+26
PAGE S00113	RPS4Y1	7	I	GACTCCTAGGCTGCTAAAAAC	G A	TTCTCAGGTTCAACCTGGCA	RPS4Y1+263-98
PAGE S00114	RPS4Y1	8	2	GATGCTCGAACCATCCGCTA	C T	CCAGATCCTGTCAATCAAGGT	RPS4Y1+447
PAGE S00051	RPS4Y1	10	4	GGCAATAAATTTGATTTTC	C T	CTGCCAGGGGAAAAGGGCAT	RPS4Y1+711
PAGE S00115	RPS4Y1	10	I	ATGTGTGTTTTGGTGGGATG	T C	TGTTTTTCTCTCTCCCTTT	RPS4Y1+691-58
PAGE S00054	RPS4Y2	2	I	CTCCTCCGATCCGGTATTAC	C T	CGTTAGCAGTTGTTACAGCT	RPS4Y2+4-87
PAGE S00053	RPS4Y2	3	I	AAGTAAAAGCTGGAAGGG	G A	TTGGTAATGTAATGGGATGA	RPS4Y2+262+195
PAGE S00056	RPS4Y2	3	4	CTACAGCACCTCTGCTCATC	G A	ACAGGTCCTCACAGCTGAG	RPS4Y2+96
PAGE S00055	RPS4Y2	7	4	GAGAGAGACAAGAGGCTGGC	T C	GCCAAACAGAGCAGTGGCTA	RPS4Y2+771

Variant ID ^a	Gene or pseudogene name	Exon number	Se-quence class ^b	Sequence to left of variant	Variants	Sequence to right of variant	Mnemonic variant ID ^{c,d}
PAGE S00063	SRY	1	2	CCCGCTTGGGTTACTCTGCAG	C T	GAAGTGCAACTGGACAACAG	SRY+465
PAGE S00068	STSP	3	P	ACGTTGGGGGTTGCAGTGAG	C T	GGAGATCGCACTACTGCCT	STSP-311-113
PAGE S00121	STSP	3	P	GGTTGCAGTGCAGCGGAGATC	G A	CACTACTGCCTCCATCCTG	STSP-311-105
PAGE S00069	STSP	5	P	CCCCCATTTGTCTTCTCAGA	G T	AAATGGCACATTGGGATAAG	STSP-78
PAGE S00066	STSP	6	P	GCTTGTCTTGTCTTACCTCC	A G	CGTGACATGGCCTTGTCT	STSP+382
PAGE S00067	STSP	6	P	GTGTGGGTATGTCTCTCCTC	G A	GTGAATACTTAGAAAGCTGC	STSP+477+15
PAGE S00122	TBL1Y	3	I	GGCTAATCATTTTTTTCTCT	C T	TTTATAGGCATTTGATGTCT	TBL1Y-265-8
PAGE S00073	TBL1Y	8	I	TATGCTCCACTTCCCCTGTC	C G T	TCTCCTGCCAGAGAGTAGA	TBL1Y+205-82
PAGE S00074	TBL1Y	8	I	CACATGAAATCAGTGAGTGC	G A	CAGGCTCTGGAAGTTGGTG	TBL1Y+457+9
PAGE S00075	TBL1Y	9	I	GCCACAATTTAGTTAGACTC	G A	TTAGAGGTAGTGTACTTAG	TBL1Y+458-118
PAGE S00076	TBL1Y	10	I	TGATAAAGACAGAAGAGCAC	A G	TGCCTACAATGTACCTGGC	TBL1Y+591-96
PAGE S00071	TBL1Y	16	I	GTTGGCAAGGTAAGGGCCGG	C A	AGCACAACTGGTACAGCTCC	TBL1Y+1280+12
PAGE S00072	TBL1Y	18	4	CACAGCTACCAAGGCATGG	C A	GGTATCTTCGAGGTGTGCTG	TBL1Y+1482
PAGE S00080	USP9Y	4	2	CCACAGCATGAAGATGAAGA	T G	CCTGCATTTCCACATACTGA	USP9Y+195
PAGE S00031	USP9Y	7	0	CTGAAGATGAATTATTTGCT	C T	GTTCTTCAGATCCCTCGATCA	M179/USP9Y+631
M70	USP9Y	8	I	TTACTTAAAAATCATTGTTC	A C	TTTTTTTTCAGTGTGGGTTGT	M170/USP9Y+773+131
PAGE S00078	USP9Y	8	I	GGTTTAAATAAATGATAGTT	G C	TTTGAACACTGTGAGGTAAC	USP9Y+1164+167
PAGE S00032	USP9Y	12	2	CAACTTGATCATCTTTTTGA	T C	TGCTTTAAGGTAGTAGCTTG	M181/USP9Y+1413
PAGE S00079	USP9Y	13	I	TCTCGCTTACTGCAAGCTC	C T	GCCCTCGGGTTCATGCCAT	USP9Y+1629+174
PAGE S00081	USP9Y	17	I	CACATTTTAAATTCAGATA	C T	GTGTTTAAACAAATATTTTTG	USP9Y+1989-149
PAGE S00033	USP9Y	19	4	CACAAGGAAAGATGATCT	A C	CCTATGTCGAGGTTTGTGTG	M183/USP9Y+2628
PAGE S00082	USP9Y	20	4	GCATTTCTGTGGCAACACCT	C T	TCTCTTATAGTTCGGTTTCC	USP9Y+2661
PAGE S00034	USP9Y	23	0	CAGCTGTAGAAAATTACGA	G A	CTGTTTGTGGACCATGCA	M184/USP9Y+3178
PAGE S00083	USP9Y	23	I	ATGTGAACATTAATAACAC	A G	TGAGTTACACTTTATTTTAG	USP9Y+3152-43
PAGE S00123	USP9Y	23	I	ATTTCTTAGCAATGATCAGA	G A	GAGAAATAGATGTTACTAAG	USP9Y+3283-72
PAGE S00084	USP9Y	25	2	ATTCCTAATCCCTCATCCGA	G A	TGCGTACTTAGAAATGAGTC	USP9Y+3636
PAGE S00070	USP9Y	29	I	AGTGTAGACTTGTGAATCA	T C	GTTGTTTTAATTTAATATTT	Tat/USP9Y+4093-24
M188	USP9Y	31	4	ATTCAGTCTGTAGTTCACC	C T	GTTACCATCAATGCCGGTTT	M188/USP9Y+4491
PAGE S00085	USP9Y	31	I	TTTTTGTGGGGTTTTGTTTT	G T	AGATATAATGAGATATTTAA	USP9Y+4387-122
PAGE S00086	USP9Y	31	I	TTACATTTATTAATGTACT	C T	GGGTTTTTTTGTGGGGTTTT	USP9Y+4387-148
PAGE S00124	USP9Y	32	I	TATTAATTTGTAGCCGAA	A C	ATGGTGAACAGAACTACTGT	USP9Y+4610-58
PAGE S00087	USP9Y	33	I	AATGTATTTTTTAACTATT	C T	TTTTATGATAACTTATCTGT	USP9Y+4831-204
PAGE S00125	USP9Y	36	I	TATATGAATGTGTGGCTTT	T C	TTGGTATAGTTATTTTAAAA	USP9Y+6088+157
PAGE S00030	USP9Y	38	4	GAATACCTTCTGGAGTGCC	T C	AGTGCAGAAGTGAGGGGTGC	M174/USP9Y+6327
PAGE S00126	USP9Y	40	I	TTTTAATTATTGCAGTACT	A C	CCCTCTTAGTTTTTTTTTCT	USP9Y+6569-50
rs13304344	USP9Y	40	I	TGCAGTACTTACCCTCTTAG	T C	TTTTTTTTCTACATATTATT	USP9Y+6569-40
PAGE S00035	USP9Y	43	I	CTGATACTGAAATCATCT	A G	AATTCTAAATAGTTTTTATT	M190/USP9Y+7222-31
PAGE S00088	USP9Y	43	I	ATGGCACATAATTAGGA	C G	AAATGTTAGTACTATTGGA	USP9Y+7065-54
PAGE S00089	USP9Y	43	4	TTGCATACAATACAGCCCTC	G A	AAGAATCACTATCAAAAACG	USP9Y+7131
PAGE S00090	USP9Y	43	I	TTTTTCTCTTATAAATTTGT	A G	GAAACCTCTGTCAAGTAA	USP9Y+7221+28
PAGE S00127	USP9Y	43	I	GATACTGAAAATCATCTAA	A G	TTCTAAATAGTTTTTATTTT	USP9Y+7222-29
PAGE S00028	USP9Y	45	I	CCAAACCCATTTTGTGCTT	T G	ACTTAAAAGTCTTCAATTA	M172/USP9Y+7530+48
PAGE S00036	UTY	1	4	TTTGTGTTTCCATGAAATC	C G	TGCGCAGTGTGCTCACTAC	M203/UTY+9
PAGE S00037	UTY	3	I	AAGGTATTGTTATCTCTTT	T C	TAAATTTCTTGCTTGACTTA	M207/UTY+325+18
PAGE S00038	UTY	5	I	AACTTAAAACATCTCGTTAC	A G	TGACTTCTTATTAATATATG	M213/UTY+376-78
PAGE S00093	UTY	9	I	AACATTTATAGTATGTAAT	C A	FTCATGTCTAGCTATTTTAA	UTY+646-206
PAGE S00129	UTY	11	I	ATTAATAAATCATATAAT	G A	TACATCGCAATGTAATAATC	UTY+867-155
PAGE S00039	UTY	12	I	AGACACTGTGTAACAAAC	A G	GAAAAATAAACGAAACGAAAG	M214/UTY+966-59
PAGE S00091	UTY	13	I	TGACGATCTTCCCCCAATT	C T	GAAAGCAGTAGAAACAGTAA	UTY+1278+33
PAGE S00040	UTY	14	2	CAGCTGGAACAGTTAGAAAG	T C	CAGTTTGTCTTAAATGCAGCA	M215/UTY+1353
PAGE S00130	UTY	23	I	ACTGCTCAGCAGAAATTTT	T A	AAAATGATTTGATTTCAAAAT	UTY+3389+124
PAGE S00092	UTY	25	I	AGAATTGGTATTGCAGTTGT	C T	ACCTGAGAGGTGATAAATGA	UTY+3578-84
PAGE S00131	ZFY	5	0	TTACTTCAACCTCAATGTCT	A G	TGCCAGAACATGTTTTAACG	ZFY+391
PAGE S00094	ZFY	9	I	ATAAAGCAGGTATAATTTAC	C T	GAGAAGTGGAGAAGTACCT	ZFY+1222+280

^a dbSNP submitter ID, dbSNP rs#, or YCC "M" ID.

^b 0=coding non-degenerate; 2=coding twofold degenerate; 4=coding fourfold degenerate; I=intron; P=pseudogene.

^c Mnemonic identifier, which is also used in Figures S1 through S4.

The purpose of these identifiers is to make it somewhat easier to determine at a glance the gene in which a particular variant occurs and to determine whether it is coding or intronic. However, the information submitted to dbSNP, rather than the positions represented in the mnemonic identifiers, provide the definitive references for the variants.

The mnemonic identifiers have three possible forms: $[Mm]/G+P$, $[Mm]/G+P+I_{after}$, or $[Mm]/G+P-I_{before}$, where

$[Mm]/$ is an optional YCC "M" ID for previously named variants (main text refs. 2 and 3) that we detected in our survey, followed by the character "/",

G is a gene or pseudogene name,

P is a nucleotide position in G 's coding sequence,

$+I_{after}$ indicates a position in the intron following the exon that ends at P , with position +1 assigned to the first nucleotide after the end of the exon, and

$-I_{before}$ indicates a position in the intron preceding the exon that starts at P , with the position of the variant reckoned backward (5') from the exon starting at P and with position -1 assigned to the last intronic nucleotide before the exon.

^d FLJ20285 and KIAA1575Y represent parts of BCORP, which we did not initially recognize as homologous to a single X-chromosome gene.