

Supplemental Table 1

Allele frequencies for all analyzed subjects and variants. AIMSS: Aromatase inhibitor associated musculoskeletal syndrome; MAF: minor allele frequency; No: number; SNP: single nucleotide polymorphism; Vt: variant; Wt: wildtype

Gene	SNP	MAF	Wt/Wt			Wt/Vt			Vt/Vt			Missing	
			Geno- type	%	No	Geno- type	%	No	Geno- type	%	No	%	No
ARVCF	rs165815	0.19	TT	64.4%	278	TC	29.2%	126	CC	4.2%	18	2.3%	10
COMT	rs165599	0.35	AA	41.2%	178	AG	44.4%	192	GG	12.3%	53	2.1%	9
COMT	rs165774	0.30	GG	50.2%	217	GA	37.3%	161	AA	10.4%	45	2.1%	9
COMT	rs2097903	0.48	AA	26.2%	113	AT	50.0%	216	TT	21.3%	92	2.5%	11
COMT	rs4633	0.48	CC	27.1%	117	CT	22.5%	207	TT	22.5%	97	2.5%	11
COMT	rs4646310	0.17	GG	64.8%	280	GA	25.7%	111	AA	3.2%	14	6.3%	27
COMT	rs4680	0.47	GG	27.3%	118	GA	44.4%	192	AA	22.0%	95	6.3%	27
COMT	rs4818	0.39	CC	33.1%	143	CG	41.9%	181	GG	14.1%	61	10.9%	47
COMT	rs6269	0.40	AA	36.6%	158	AG	45.8%	198	GG	16.9%	73	0.7%	3
COMT	rs933271	0.31	TT	46.8%	202	TC	41.7%	180	CC	9.0%	39	2.5%	11
CYP19A1	rs10046	0.47	GG	26.4%	114	GA	50.7%	219	AA	21.3%	92	1.6%	7
CYP19A1	rs1008805	0.39	AA	33.1%	143	AG	47.7%	206	GG	12.7%	55	6.5%	28
CYP19A1	rs10459592	0.46	GG	30.1%	130	GT	46.1%	199	TT	22.2%	96	1.6%	7
CYP19A1	rs1062033	0.41	CC	34.3%	148	CG	46.8%	202	GG	16.0%	69	3.0%	13
CYP19A1	rs11575899	0.34	TCT	45.0%	192	TCT/-	42.2%	180	-/	12.9%	55	1.2%	5
CYP19A1	rs2008691	0.19	AA	64.1%	277	AG	31.0%	134	GG	3.7%	16	1.2%	5
CYP19A1	rs2289105	0.48	TT	26.2%	113	TC	49.5%	214	CC	21.5%	93	2.8%	12
CYP19A1	rs2414096	0.46	GG	24.1%	104	GA	41.9%	181	AA	17.1%	74	16.9%	73
CYP19A1	rs28566535	0.10	AA	80.8%	349	AC	16.0%	69	CC	2.1%	9	1.2%	5
CYP19A1	rs28757184#	0.02	GG	92.8%	401	GA	4.9%	21	AA	0.0%	0	2.3%	10
CYP19A1	rs3759811	0.45	TT	26.2%	113	TC	48.1%	208	CC	16.2%	70	9.5%	41
CYP19A1	rs4646	0.28	CC	48.8%	211	CA	42.1%	182	AA	6.7%	29	2.3%	10
CYP19A1	rs4774585	0.22	GG	53.0%	229	GA	31.9%	138	AA	3.9%	17	11.1%	48
CYP19A1	rs4775936	0.42	CC	32.2%	139	CT	49.5%	214	TT	17.1%	74	1.2%	5
CYP19A1	rs6493497	0.13	GG	67.4%	291	GA	21.5%	93	AA	0.9%	4	10.2%	44
CYP19A1	rs700518	0.45	TT	28.2%	122	TC	51.6%	223	CC	18.5%	80	1.6%	7
CYP19A1	rs7181886	0.09	CC	77.5%	335	CG	13.4%	58	GG	1.4%	6	7.6%	33
CYP19A1	rs727479	0.34	AA	43.8%	189	AC	41.0%	177	CC	12.7%	55	2.5%	11
CYP19A1	rs730154	0.19	TT	55.3%	239	TC	24.8%	107	CC	3.9%	17	16.0%	69
CYP19A1	rs749292	0.43	GG	30.1%	130	GA	50.2%	217	AA	16.9%	73	2.8%	12
CYP19A1	rs752760	0.48	CC	24.8%	107	CT	44.4%	192	TT	21.5%	93	9.3%	40
CYP19A1	rs936308	0.20	CC	63.4%	274	CG	30.6%	132	GG	4.6%	20	1.4%	6
EP300	rs20551	0.27	AA	52.3%	226	AG	38.7%	167	GG	6.7%	29	2.3%	10
ESR1	rs1062577	0.08	TT	83.1%	359	TA	13.4%	58	AA	0.7%	3	2.8%	12
ESR1	rs11963577#	0.00	CC	99.1%	428	CT	0.2%	1	TT	0.0%	0	0.7%	3
ESR1	rs1709181	0.45	TT	25.5%	110	TC	42.8%	185	CC	17.1%	74	14.6%	63
ESR1	rs1709182	0.37	TT	36.1%	156	TC	40.3%	174	CC	13.2%	57	10.4%	45
ESR1	rs1884049	0.24	CC	54.6%	236	CT	38.4%	166	TT	4.2%	18	2.8%	12
ESR1	rs2077647	0.49	CC	26.2%	113	CT	45.1%	195	TT	24.8%	107	3.9%	17
ESR1	rs2228480	0.20	GG	61.6%	266	GA	30.8%	133	AA	3.9%	17	3.7%	16
ESR1	rs2234693	0.49	TT	23.8%	103	TC	44.7%	193	CC	22.7%	98	8.8%	38
ESR1	rs2347868	0.22	TT	57.9%	250	TC	34.3%	148	CC	3.9%	17	3.9%	17
ESR1	rs2347869	0.42	AA	33.3%	144	AC	47.0%	203	CC	17.4%	75	2.3%	10
ESR1	rs2747648#	0.03	TT	91.2%	394	TC	5.6%	24	CC	0.5%	2	2.8%	12
ESR1	rs2813543	0.24	GG	58.6%	253	GA	33.8%	146	AA	7.4%	32	0.2%	1
ESR1	rs2982734	0.37	GG	38.4%	166	GA	44.9%	194	AA	13.4%	58	3.2%	14
ESR1	rs3003917	0.22	AA	52.8%	228	AG	33.8%	146	GG	2.8%	12	10.6%	46
ESR1	rs3020368	0.09	CC	79.4%	343	CT	16.0%	69	TT	0.5%	2	4.2%	18
ESR1	rs3020371	0.36	CC	39.8%	172	CT	44.2%	191	TT	13.2%	57	2.8%	12
ESR1	rs3020376*	0.27	AA	52.8%	228	AG	24.3%	105	GG	12.3%	53	10.6%	46
ESR1	rs3020377	0.37	AA	38.2%	165	AG	45.8%	198	GG	13.4%	58	2.5%	11
ESR1	rs3020410	0.16	CC	68.3%	295	CA	29.2%	126	AA	1.2%	5	1.4%	6
ESR1	rs3778610*	0.06	AA	83.1%	359	AG	6.5%	28	GG	2.3%	10	8.1%	35
ESR1	rs3798565*	0.23	CC	61.3%	265	CT	24.1%	104	TT	9.5%	41	5.1%	22
ESR1	rs3798577	0.46	TT	29.4%	127	TC	48.1%	208	CC	20.8%	90	1.6%	7
ESR1	rs3798758#	0.03	CC	92.1%	398	CA	5.6%	24	AA	0.2%	1	2.1%	9
ESR1	rs3853250	0.50	TT	25.0%	108	TG	46.8%	202	GG	24.5%	106	3.7%	16
ESR1	rs4870056	0.49	GG	23.4%	101	GA	45.4%	196	AA	22.0%	95	9.3%	40
ESR1	rs4870061	0.22	CC	60.0%	259	CT	31.9%	138	TT	4.9%	21	3.2%	14
ESR1	rs4986934#	0.04	CC	90.5%	391	CT	7.2%	31	TT	0.0%	0	2.3%	10
ESR1	rs6909026#	0.02	AA	91.2%	394	AG	4.2%	18	GG	0.2%	1	4.4%	19
ESR1	rs6920483#	0.02	GG	91.4%	395	GA	4.2%	18	AA	0.0%	0	4.4%	19
ESR1	rs6930355	0.15	TT	69.2%	299	TC	23.8%	103	CC	2.8%	12	4.2%	18
ESR1	rs750686	0.37	GG	38.0%	164	GA	44.2%	191	AA	13.0%	56	4.9%	21
ESR1	rs867239#	0.03	CC	94.2%	407	CG	5.6%	24	GG	0.0%	0	0.2%	1
ESR1	rs926848#	0.04	CC	87.5%	378	CT	7.2%	31	TT	0.2%	1	5.1%	22
ESR1	rs9322331	0.34	CC	44.7%	193	CT	42.6%	184	TT	12.3%	53	0.5%	2
ESR1	rs9322335	0.27	CC	51.9%	224	CT	41.4%	179	TT	5.6%	24	1.2%	5

ESR1	rs9322336	0.22	TT	55.8%	241	TC	31.7%	137	CC	4.2%	18	8.3%	36
ESR1	rs9322338*	0.06	GG	84.3%	364	GA	6.9%	30	AA	2.5%	11	6.3%	27
ESR1	rs9322348#	0.00	CC	98.6%	426	CT	0.0%	0	TT	1.4%	6	0.0%	0
ESR1	rs9340799	0.37	AA	36.1%	156	AG	41.7%	180	GG	12.7%	55	9.5%	41
ESR1	rs9340817#	0.03	CC	86.3%	373	CA	4.2%	18	AA	0.9%	4	8.6%	37
ESR1	rs9340835	0.35	GG	36.8%	159	GA	43.8%	189	AA	10.0%	43	9.5%	41
ESR1	rs9340941*#	0.10	CC	82.4%	356	CT	0.0%	0	TT	8.8%	38	8.8%	38
ESR1	rs9340958	0.07	CC	81.0%	350	CT	12.5%	54	TT	0.5%	2	6.0%	26
ESR1	rs9341004	0.11	CC	75.0%	324	CG	18.3%	79	GG	1.2%	5	5.6%	24
ESR1	rs9341066	0.07	GG	78.9%	341	GA	12.5%	54	AA	0.2%	1	8.3%	36
ESR1	rs9341070#	0.01	CC	97.9%	423	CT	0.9%	4	TT	0.2%	1	0.9%	4
ESR1	rs9341072#	0.00	GG	93.5%	404	GA	0.0%	0	AA	0.2%	1	6.3%	27
ESR1	rs9341077#	0.04	TT	90.7%	392	TC	8.1%	35	CC	0.2%	1	0.9%	4
ESR1	rs9479188	0.17	GG	68.8%	297	GA	25.9%	112	AA	3.9%	17	1.4%	6
ESR2	rs10140457#	0.03	CC	93.5%	404	CA	5.1%	22	AA	0.2%	1	1.2%	5
ESR2	rs1256031	0.45	AA	29.2%	126	AG	44.0%	190	GG	19.2%	83	7.6%	33
ESR2	rs1256049#	0.04	CC	89.8%	388	CT	7.4%	32	TT	0.0%	0	2.8%	12
ESR2	rs1256059	0.42	GG	32.9%	142	GA	43.5%	188	AA	17.4%	75	6.3%	27
ESR2	rs1256062	0.15	TT	72.5%	313	TC	23.8%	103	CC	2.8%	12	0.9%	4
ESR2	rs1256064*	0.16	AA	72.5%	313	AG	20.1%	87	GG	5.1%	22	2.3%	10
ESR2	rs17766755	0.34	GG	41.0%	177	GA	41.9%	181	AA	11.1%	48	6.0%	26
ESR2	rs1952585*	0.13	AA	75.0%	324	AG	17.6%	76	GG	3.2%	14	4.2%	18
ESR2	rs2274705	0.12	AA	65.3%	282	AC	16.0%	69	CC	1.9%	8	16.9%	73
ESR2	rs3020450	0.32	CC	44.4%	192	CT	44.9%	194	TT	9.0%	39	1.6%	7
ESR2	rs4986938	0.36	CC	41.9%	181	CT	42.6%	184	TT	13.7%	59	1.9%	8
ESR2	rs8003490	0.08	GG	79.2%	342	GA	14.6%	63	AA	0.2%	1	6.0%	26
ESR2	rs8006145	0.27	CC	53.9%	233	CA	38.0%	164	AA	7.4%	32	0.7%	3
ESR2	rs867443	0.25	GG	55.3%	239	GA	36.1%	156	AA	6.7%	29	1.9%	8
ESR2	rs944045	0.09	TT	79.2%	342	TC	13.9%	60	CC	2.1%	9	4.9%	21
ESR2	rs944460#	0.05	GG	82.9%	358	GC	7.2%	31	CC	0.7%	3	9.3%	40
EZH2	rs2302427	0.07	CC	84.5%	365	CG	13.4%	58	GG	0.2%	1	1.9%	8
HCRT	rs41513044#	0.01	AA	95.4%	412	AC	2.3%	10	CC	0.0%	0	2.3%	10
HCRT	rs9902709	0.16	AA	66.4%	287	AG	26.9%	116	GG	2.3%	10	4.4%	19
HCRTR1	rs11806980	0.05	TT	88.7%	383	TC	9.0%	39	CC	0.5%	2	1.9%	8
HCRTR1	rs2176807	0.46	CC	28.9%	125	CA	47.9%	207	AA	21.3%	92	1.9%	8
HCRTR1	rs2271933	0.39	GG	35.9%	155	GA	45.8%	198	AA	15.3%	66	3.0%	13
HCRTR2	rs2653349	0.18	CC	66.2%	286	CT	28.2%	122	TT	3.2%	14	2.3%	10
HTR1A	rs1800046#	0.00	GG	98.6%	426	GC	0.5%	2	CC	0.0%	0	0.9%	4
HTR1A	rs6295	0.47	CC	25.5%	110	CG	50.0%	216	GG	20.1%	87	4.4%	19
HTR2A	rs1328685	0.10	AA	77.3%	334	AG	18.5%	80	GG	0.5%	2	3.7%	16
HTR2A	rs1805055*#	0.13	GG	81.9%	354	GT	6.0%	26	TT	10.0%	43	2.1%	9
HTR2A	rs2274639	0.13	CC	73.6%	318	CG	22.2%	96	GG	2.1%	9	2.1%	9
HTR2A	rs2296972	0.36	CC	42.1%	182	CA	40.5%	175	AA	14.8%	64	2.5%	11
HTR2A	rs3742278	0.13	AA	75.0%	324	AG	19.4%	84	GG	3.5%	15	2.1%	9
HTR2A	rs594242	0.19	GG	65.0%	281	GC	28.2%	122	CC	3.9%	17	2.8%	12
HTR2A	rs6306	0.07	GG	82.9%	358	GA	12.7%	55	AA	0.2%	1	4.2%	18
HTR2A	rs6308#	0.00	GG	96.5%	417	GA	0.7%	3	AA	0.0%	0	2.8%	12
HTR2A	rs6311	0.43	CC	30.6%	132	CT	47.9%	207	TT	17.6%	76	3.9%	17
HTR2A	rs6313	0.43	GG	29.9%	129	GA	50.0%	216	AA	17.1%	74	3.0%	13
HTR2A	rs731779	0.17	AA	65.3%	282	AC	29.6%	128	CC	1.9%	8	3.2%	14
HTR2A	rs7997012	0.37	GG	38.7%	167	GA	43.5%	188	AA	14.4%	62	3.5%	15
HTR2A	rs9567746	0.17	AA	66.0%	285	AG	29.4%	127	GG	1.9%	8	2.8%	12
NCOA1	rs11125744	0.14	CC	72.2%	312	CG	20.4%	88	GG	2.8%	12	4.6%	20
NCOA1	rs17737058	0.24	CC	55.3%	239	CG	35.0%	151	GG	5.1%	22	4.6%	20
NCOA1	rs1804645#	0.04	CC	90.0%	389	CT	8.3%	36	TT	0.0%	0	1.6%	7
NCOA1	rs2033861	0.47	GG	25.5%	110	GA	38.4%	166	AA	20.6%	89	15.5%	67
NCOA1	rs2083389	0.26	TT	49.3%	213	TC	35.4%	153	CC	5.6%	24	9.7%	42
NCOA1	rs3731628	0.07	AA	85.9%	371	AT	13.0%	56	TT	0.0%	0	1.2%	5
NCOA1	rs920418	0.45	GG	30.3%	131	GC	45.1%	195	CC	21.5%	93	3.0%	13
NCOA2	rs1380896	0.20	CC	56.5%	244	CT	24.8%	107	TT	5.1%	22	13.7%	59
NCOA2	rs2228591#	0.05	CC	89.1%	385	CG	8.8%	38	GG	0.5%	2	1.6%	7
NCOA2	rs2272670	0.25	TT	53.2%	230	TC	33.3%	144	CC	6.9%	30	6.5%	28
NCOA2	rs3088092	0.06	AA	83.8%	362	AT	9.5%	41	TT	0.5%	2	6.3%	27
NCOA2	rs4738079	0.09	GG	75.0%	324	GA	15.5%	67	AA	0.2%	1	9.3%	40
NCOA3	rs2076546	0.10	AA	78.2%	338	AG	18.5%	80	GG	0.7%	3	2.5%	11
NCOA3	rs2230782	0.07	GG	82.4%	356	GC	13.0%	56	CC	0.5%	2	4.2%	18
NCOA3	rs365602	0.12	CC	66.0%	285	CT	16.0%	69	TT	1.9%	8	16.2%	70
NCOA3	rs396221	0.45	TT	26.9%	116	TG	51.6%	223	GG	17.1%	74	4.4%	19
NCOA3	rs6094752	0.06	CC	85.4%	369	CT	10.6%	46	TT	0.5%	2	3.5%	15
NCOR1	rs1065822	0.47	TT	29.2%	126	TA	45.1%	195	AA	23.8%	103	1.9%	8
NCOR1	rs1073093#	0.03	CC	90.7%	392	CT	6.5%	28	TT	0.0%	0	2.8%	12
NCOR1	rs61753150#	0.02	TT	90.7%	392	TG	3.2%	14	GG	0.0%	0	6.0%	26
NCOR2	rs1812730	0.44	AA	29.6%	128	AG	49.8%	215	GG	17.8%	77	2.8%	12
NCOR2	rs2229840	0.16	CC	68.1%	294	CT	27.8%	120	TT	1.9%	8	2.3%	10

NCOR2	rs7978237	0.13	GG	71.8%	310	GA	19.4%	84	AA	2.8%	12	6.0%	26
NCOR2	rs837935	0.34	TT	41.7%	180	TC	39.8%	172	CC	12.5%	54	6.0%	26
NRIP1	rs2229742	0.11	GG	77.8%	336	GC	18.1%	78	CC	1.4%	6	2.8%	12
PELP1	rs9436	0.21	TT	63.2%	273	TA	28.7%	124	AA	6.7%	29	1.4%	6
PGR	rs1042838	0.14	CC	73.1%	316	CA	22.7%	98	AA	2.3%	10	1.9%	8
PGR	rs11571171	0.31	AA	44.9%	194	AG	42.8%	185	GG	8.3%	36	3.9%	17
PGR	rs500760	0.26	TT	52.8%	228	TC	33.6%	145	CC	7.2%	31	6.5%	28
PGR	rs506487	0.33	CC	44.9%	194	CT	41.2%	178	TT	11.8%	51	2.1%	9
PGR	rs516693	0.14	CC	70.8%	306	CT	24.8%	107	TT	1.2%	5	3.2%	14
PGR	rs518162	0.10	GG	74.1%	320	GA	16.0%	69	AA	1.4%	6	8.6%	37
PGR	rs660149	0.28	CC	48.8%	211	CG	41.0%	177	GG	6.9%	30	3.2%	14
PGR	rs7116336	0.09	TT	78.0%	337	TA	14.1%	61	AA	1.6%	7	6.3%	27
SLC6A4	rs140700	0.08	CC	83.1%	359	CT	15.0%	65	TT	0.5%	2	1.4%	6
SLC6A4	rs16965628	0.09	GG	81.0%	350	GC	16.2%	70	CC	0.5%	2	2.3%	10
SLC6A4	rs25531#	0.03	AA	91.2%	394	AG	4.9%	21	GG	0.0%	0	3.9%	17
SLC6A4	rs25532	0.14	CC	63.4%	274	CT	20.1%	87	TT	1.6%	7	14.8%	64
TCL1A	rs11849538	0.14	CC	73.1%	316	CG	24.5%	106	GG	1.9%	8	0.5%	2
TCL1A	rs2369049	0.18	AA	68.1%	294	AG	26.9%	116	GG	4.9%	21	0.2%	1
TCL1A	rs7158782	0.20	AA	65.7%	284	AG	28.9%	125	GG	5.1%	22	0.2%	1
TCL1A	rs7159713	0.19	AA	66.9%	289	AG	28.0%	121	GG	4.9%	21	0.2%	1

	# repeats	None		Any			
		%	No	%	No		
CYP19A1	rs60271534	TTTA repeats	7	19.9%	85	80.1%	342
			8	81.5%	348	18.5%	79
			10	98.1%	419	1.9%	8
			11	48.7%	208	51.3%	219
			12	93.9%	401	6.1%	26

* not in Hardy-Weinberg equilibrium

excluded from analysis (plus 8 additional SNPs not listed because MAF 0)