

Supplementary Table 1. Characteristics of variation in sex-steroid receptors (ESR1, ESR2, and PGR) and hormone-synthesizing genes (CYP19A1 and HSD17B2) in the Women's Health Study.

Gene/ Variant	LD block #	Region	Base change	Cases allele (%)	Controls allele (%)	P_{HWE}^*	$FDR P$
ESR1							
rs2077647	--	Exon 1	T → C	47	47	0.41	0.90
rs6902771	1	Intron 1	C → T	48	45	0.78	0.95
rs2234693	1	Intron 1	T → C	48	44	0.82	0.95
rs9340799	1	Intron 1	A → G	39	34	0.71	0.95
rs712221	--	Intron 2	A → T	43	40	0.20	0.88
rs1709183	--	Intron 2	T → C	30	27	0.39	0.90
rs2347923	2	Intron 3	A → C	29	31	0.17	0.88
rs12199722	2	Intron 3	A → G	35	31	0.86	0.96
rs6912184	3	Intron 3	A → G	20	22	0.85	0.96
rs4583998	3	Intron 3	G → A	30	31	0.30	0.88
rs1801132	3	Exon 4	C → G	20	22	0.61	0.93
rs3020314	4	Intron 4	T → C	29	33	0.30	0.88
rs3020401	4	Intron 4	A → G	28	31	0.28	0.88
rs2982685	5	Intron 4	A → G	33	37	0.52	0.93
rs2982683	5	Intron 4	C → T	33	29	0.60	0.93
rs3020407	--	Intron 4	A → G	30	32	0.25	0.88
rs9340954	--	Intron 4	T → G	24	26	0.78	0.95
rs2982712	6	Intron 5	T → G	42	41	0.004	0.41
rs3020434	6	Intron 5	C → T	16	17	0.40	0.90
rs3020363	6	Intron 5	T → C	33	33	0.10	0.88
rs3020370	--	Intron 5	G → C	21	22	0.87	0.96
rs3798577	--	3' UTR	T → C	46	48	0.33	0.90
ESR2							
rs1255953	--	Exon 5	T → C	18	14	0.51	0.77
rs928554	1	3' UTR	T → C	46	44	0.32	0.77
rs1256031	1	Intron 3	T → C	49	46	0.31	0.77
rs3020450	1	5' UTR	G → A	33	32	0.59	0.77
PGR							
rs473409	1	3' UTR	A → C	26	23	0.87	0.92
rs484389	--	3' UTR	T → C	23	22	0.60	0.91
rs500760	--	3' UTR	A → G	23	22	0.66	0.91
rs1042839	--	Exon 5	C → T	17	14	0.72	0.91
rs545835	1	Intron 4	C → T	48	49	0.62	0.91
rs1042838	1	Exon 4	G → T	17	14	0.71	0.91
rs1824128	1	Intron 3	C → A	19	15	0.36	0.91
rs477151	2	Intron 3	T → C	26	28	0.49	0.91
rs559700	2	Intron 2	A → G	17	14	0.82	0.91
rs516693	2	Intron 2	G → A	13	12	0.72	0.91
rs508533	2	Intron 2	A → C	37	38	0.79	0.91
rs572483	3	Intron 2	A → G	32	34	0.98	0.99
rs543215	3	Intron 2	T → C	19	17	0.77	0.91
rs613120	3	Intron 2	T → C	51	48	0.35	0.91
rs529359	4	Intron 2	A → G	38	38	0.65	0.91
rs481775	4	Intron 2	C → T	32	35	0.94	0.98
rs1379130	--	Exon 1	G → A	31	35	0.99	0.99
rs3740753	4	Exon 1	C → G	17	14	0.45	0.91
CYP19A1							

rs4646	1	Exon 10	G → T	26	27	0.51	0.93
rs10046	1	Exon 10	T → C	46	53	0.97	0.98
rs2414096	1	Intron 2	A → G	52	47	0.50	0.93
rs727479	1	Intron 2	T → G	38	35	0.57	0.93
rs1008805	2	Intron 1	T → C	46	43	0.86	0.98
rs749292	2	Intron 1	C → T	41	43	0.80	0.98
rs936306	3	Intron 1	G → A	14	15	0.95	0.98
rs3751591	3	Intron 1	T → C	18	17	0.97	0.98
rs1004984	4	Intron 1	C → T	36	35	0.26	0.91
rs2445762	4	Intron 1	A → G	25	26	0.57	0.93
rs2470144	4	Intron 1	A → G	44	46	0.79	0.98
rs2445765	4	5' UTR	C → G	15	16	0.13	0.88
rs2446405	4	5' UTR	A → T	14	17	0.13	0.88
HSD17B2							
rs4445895	1	Exon 1	C → T	38	35	0.87	0.93
rs6564961	1	Intron 1	A → G	40	37	0.88	0.93
rs2911422	1	Intron 3	G → C	48	42	0.18	0.51
rs2042429	1	Intron 3	T → C	50	44	0.33	0.62
rs723012	1	Intron 3	C → T	33	37	0.04	0.25
rs9939740	--	Intron 3	A → G	36	41	0.22	0.57

*. P values for tests of Hardy-Weinberg equilibrium (HWE) among controls.