

SUPPLEMENTARY INFORMATION

doi:10.1038/nature13545

Supplementary Tables

Supplementary Table 1 Details of the 58 studies contributing to the expanded meta-analysis of genome-wide association studies for age at menarche	2
Supplementary Table 2 Results from GCTA analyses to identify secondary signals at menarche loci...6	6
Supplementary Table 3 Follow up of 123 menarche SNPs in the EPIC-InterAct study.....8	8
Supplementary Table 4 Follow up of menarche-associated SNPs for association with puberty timing in the EGG consortium.....10	10
Supplementary Table 5 Parent of Origin analyses of loci in imprinted regions in the DeCODE study.14	14
Supplementary Table 6 eQTL analyses across multiple tissues.....17	17
Supplementary Table 7 eQTL analyses in Whole Blood.....17	17
Supplementary Table 8 Candidate genes at or near menarche loci.....18	18
Supplementary Table 9 Association of menarche signals with BMI and height in the GIANT consortium	22

Supplementary Note

Full study acknowledgments, author contributions and website details.....24	24
---	----

Supplementary Table 1 | Details of the 58 studies that contributed to the discovery phase for age at menarche.

Type ^a	Study Name / acronym	Full Study name	N	GC Factor	Mean age (SD)	Mean AAM (SD)	Mean birth year	SNP array	Imputation program	Analysis program	Specific Menarche question
Discovery - previous	WGH5	Women's Genome Health Study	22028	1.10	54.7 (7.1)	12.4 (1.4)	1939	Illumina HumanHap300 Duo "4"	MACH	MACH2QTL	At what age did your menstrual periods begin?" with response categories "9 or younger; 10-11; 12-13; 14; 15; 16; 17 or older."
Discovery - previous	deCODE	deCODE Genetics, Iceland	15864	1.28	-	13.2 (1.3)	1948	Illumina HumanHap 300K and 370K CNV	IMPUTE	In house	How old were you when your menstruation started?
Discovery - previous	ARIC	Atherosclerosis Risk in Communities Study	4247	1.03	53.9 (5.7)	12.9 (1.5)	-	Affymetrix 6.0	MACH	ProABEL	"At approximately what age were you when your menstrual periods started?"
Discovery - previous	FHS	Framingham Heart Study	3801	1.01	42.5 (10.1)	12.8 (1.5)	1952	Affymetrix 500K + Affymetrix 50K Illumina Human610-Quad v1 and 370K	MACH	R-packages MERLIN-fastaSOC	"Age at start of menses" and "How old were you when you had your first menstrual period (menses)?" "About how old were you when you had your first menstrual period?"
Discovery - previous	QIMR	Queensland Institute of Medical Research	3528	1.03	-	13.1 (1.3)	1964	-	MACH	MACH	"How old were you when you had your first menstrual period?"
Discovery - previous	RS1	Rotterdam Study 1	3175	1.02	69.6 (9.3)	13.5 (1.6)	1922	Illumina HumanHap 550K	MACH	MACH2QTL	"How old were you when you had your first menstrual period?"
Discovery - previous	NHS-HU	Nurses' Health Study	3090	1.02	55.7 (6.7)	12.5 (1.4)	-	Affymetrix 6.0	MACH	ProABEL	"At what age did you have your menstrual periods begin?"
Discovery - previous	Colaus	Cohort Lausannoise	2797	1.00	53.4 (10.8)	13.2 (1.6)	1954	Affymetrix 500K	IMPUTE	In house	"At what age did you have your first period?"
Discovery - previous	NFBC	Northern Finland Birth Cohort 1966	2648	1.03	31.2 (0.4)	12.9 (1.3)	-	Illumina Infinium 370CNV Duo	MACH	ProABEL	"How old were you when you started menstruating
Discovery - previous	TWINSUK	Twins UK	2276	1.01	58.2 (12.7)	13.0 (1.6)	1951	Illumina HumanHap 300K	IMPUTE	GenABEL	"How old were you when you had your first menstrual period?"
Discovery - previous	NHS-CGEMS/BBCA	Nurses' Health Study	2270	1.03	56.8 (6.4)	12.5 (1.4)	-	Illumina HumanHap 550K	MACH	ProABEL	"At what age did you have your menstrual periods begin?"
Discovery - previous	SardinIA	SardinIA Study	2158	1.23	43.9 (17.2)	13.2 (1.6)	1960	Affymetrix 10K, 50K BeadChip	MACH	IERLINFESTASS	"At what age did you have your menstrual periods begin?"
Discovery - previous	AGEs-Reykjavik	Age, Gene/Environment Susceptibility Study	1849	1.03	76.3 (5.5)	13.6 (1.3)	-	Illumina Human6160W-Quad	MACH	PLINK	"At what age did you have your menstrual periods begin?"
Discovery - previous	DNC	Danish National Birth Cohort, Preterm delivery study	1748	1.02	30.0 (4.3)	13.3 (1.3)	1970	Illumina HumanHap 370K CNV version 1B	MACH	MACH2QTL	How old were you when you had your first menstrual period?
Discovery - previous	Indiana	Indiana University premenopausal Caucasian women peak BMD study	1497	1.01	33.3 (7.2)	12.6 (1.4)	1966	Illumina HumanHap 610 Quad version 1B	IMPUTE	IERLINFESTASS	"At what age did your periods begin? _____ years old."
Discovery - previous	SAGE	Study of Addiction: Genetics and Environment	1291	1.00	38.4 (9.1)	12.8 (1.6)	-	Illumina Human1Mv1_C	IMPUTE	SNPTEST	"At what age did you have your first menstrual period?"
Discovery - previous	EPIC_Cohort	European Prospective Investigation into Cancer and Nutrition - Obesity study cohort	1215	0.97	58.7 (9.0)	12.9 (1.8)	1936	Affymetrix GeneChip 500K	IMPUTE	SNPTEST	How old were you when you had your first menstrual period?
Discovery - previous	RS2	Rotterdam Study 2	1119	1.00	65.1 (8.4)	13.3 (1.6)	1935	Illumina HumanHap 550K	MACH	MACH2QTL	"How old were you when you had your first menstrual period?"
Discovery - previous	RS3	Rotterdam Study 3	1112	1.01	56.2 (6.1)	13.1 (1.6)	1951	Illumina HumanHap 550K	MACH	MACH2QTL	"How old were you when you had your first menstrual period?"
Discovery - previous	ERF	Erasmus Rucphen Family study	1103	1.03	47.5 (14.3)	13.1 (1.7)	-	Illumina 6K_318K_370K, Affymetrix 250K	MACH	ProABEL	"At what age did your menstrual periods begin?"
Discovery - previous	NTR	Netherlands Twin Register	1051	1.01	44.6 (13.6)	13.2 (1.4)	1961	Affymetrix 500K Perlegen	IMPUTE	SNPTEST	"How old were you when you had your first menstrual period?"
Discovery - previous	TWINSUKII	Twins UK	1016	1.00	62.4 (11.6)	12.9 (1.5)	1946	Illumina Hap610Quad Illumina HumanHap610 quad (modified)	MACH	GenABEL	"How old were you when you had your first menstrual period?"
Discovery - previous	HBCS	Helsinki Birth Cohort Study	976	1.01	61.5 (3.0)	12.8 (1.5)	-	Illumina Hap610Quad	IMPUTE	ProABEL	"At what age did you have your first menstrual periods start?"
Discovery - previous	TWINSUKII	Twins UK	671	1.06	55.4 (14.6)	13.1 (1.6)	1953	Illumina Hap610Quad	IMPUTE	GenABEL	"How old were you when you had your first menstrual period?"
Discovery - previous	EPIC_Cases	European Prospective Investigation into Cancer and Nutrition - Obesity study cases	625	0.96	58.8 (8.8)	12.7 (2.0)	1936	Affymetrix GeneChip 500K	IMPUTE	SNPTEST	How old were you when you had your first menstrual period?
Discovery - previous	InCHIANTI	Invecchiare in Chianti, aging in the Chianti area	597	1.04	68.2 (15.5)	13.3 (1.5)	1930	Illumina HumanHap 550K	IMPUTE	SNPTEST	How old were you when you had your first menstrual period?
Discovery - previous	HAPI	The older-order Amish population	557	1.05	49.1 (3.7)	13.1 (1.3)	1953	Affymetrix 500K and 6.0	MACH	MMAP	How old were you when you had your first menstrual period?
Discovery - previous	Health 2000 (Gennets)	Health2000 cohort - control subsample	465	1.023	51.9 (11.6)	13.4 (1.6)	-	Illumina HumanHap610 quad (modified)	MACH	ProABEL	How old were you when your periods started?
Discovery - previous	Health 2000 (Gennets)	Health2000 cohort - case subsample	457	0.999	51.8 (11.5)	13.4 (1.5)	-	Illumina HumanHap610 quad (modified)	MACH	ProABEL	How old were you when your periods started?

Supplementary Table 1 (continued) | Details of the 58 studies that contributed to the discovery phase for age at menarche.

Type ^a	Study Name / acronym	Full Study name	N	GC Factor	Mean age (SD)	Mean AAM (SD)	Mean birth year	SNP array	Imputation program	Analysis program	Specific Menarche question
Discovery - new	ALSPAC (Children / Mothers)	Avon Longitudinal Study of Parents and Children	9315	1.00	-	12.7(1.1)/ 12.8(1.5)	-	Illumina HumanHap550 quad / Illumina Human600W-quad	MACH	GEMMA	How old were you / was your daughter when she had her first period?
Discovery - new	Lifelines	The Lifelines Cohort Study and Biobank	7483	1.05	47.74(10.9)	13.1(1.5)	1961	Illumina CytoSNP v.2.0-300K	Beagle 3.3	Plink-module dosage est	How old were you when you had your first menstrual period?
Discovery - new	Twingene	TwinGene	4922	1.03	64.52(8.2)	13.5(1.4)	-	Illumina OmniExpress bead chip	IMPUTE	SNPTEST	How old were you when you got your first menstruation?
Discovery - new	EGCUT_omni	Estonian Genome Center, University of Tartu	3570	1.03	52.8(21.2)	13.7(1.4)	1955	Illumina HumanOmniExpress	IMPUTE	Age 16, parental reply to question about cohort member: "At what age did she have her first menstrual period"	How old were you at the time your menstruations started?
Discovery - new	B5.8C	British 1958 birth cohort	2480	1.02	45.2 (0.4)	12.8(1.3)	1958	Illumina HumanHap550K / 610K (3 deposits)	MACH	ProbABEL	How old were you when you had your first menstrual period?
Discovery - new	GOYA_cases	Genomics in Obesity in Young Adults - case sample	1782	1.00	29(4.3)	12.8(1.33)	1970	Illumina MaP10Quad	MACH 1.0	ProbABEL-0.2.0	How old were you when you had your first menstrual period?
Discovery - new	GOYA_ctrls	Genomics in Obesity in Young Adults - control sample	1746	1.01	29(4.3)	13.3(1.29)	1970	Illumina MaP10Quad	MACH 1.0	ProbABEL-0.2.0	How old were you when you had your first menstrual period?
Discovery - new	NHS2_KS	Nurses' Health Study	1685	1.00	52.6(6.6)	12.5(1.4)	1936	Illumina Human610-Quadv1	MACH	ProbABEL	At what age did your menstrual periods begin?
Discovery - new	NHS2_KS	Nurses' Health Study	1685	1.00	37.0(4.6)	12.4(1.4)	1953	Illumina Human610K	MACH	ProbABEL	At what age did your menstrual periods begin?
Discovery - new	EGCUT_370K	Estonian Genome Center, University of Tartu	1177	1.02	40.4(15.6)	13.3(1.4)	1968	Illumina Human370CNV	IMPUTE	SNPTEST	How old were you at the time your menstruations started?
Discovery - new	NHS_CC	Nurses' Health Study	1168	1.00	58.0(6.6)	12.6(1.4)	1929	Illumina Infinium Omni Express	MACH	ProbABEL	At what age did your menstrual periods begin?
Discovery - new	NHS_CHD	Nurses' Health Study	1146	1.00	58.4(6.3)	12.6(1.4)	1930	Affymetrix 6.0	MACH	ProbABEL	At what age did your menstrual periods begin?
Discovery - new	KORA4	Cooperative Health Research in the Region of Augsburg	898	1.01	55.0(8.8)	13.5(1.5)	1946	Affymetrix 6.0	IMPUTE	QUICKTEST	"At what age did you have the first menstrual period (menarche)?"
Discovery - new	KORA33	Cooperative Health Research in the Region of Augsburg	809	1.01	52.8(10.1)	13.7(1.5)	1942	Affymetrix 50K	IMPUTE	QUICKTEST	"At what age did you have the first menstrual period (menarche)?"
Discovery - new	NHS_GA	Nurses' Health Study	804	1.01	57.4(6.2)	12.6(1.4)	1931	Illumina Human600W_Quad_v1_A	MACH	ProbABEL	At what age did your menstrual periods begin?
Discovery - new	NHS_MD	Nurses' Health Study	794	1.00	55.8(6.5)	12.5(1.3)	1932	Illumina Infinium Omni Express	MACH	ProbABEL	At what age did your menstrual periods begin?
Discovery - new	NHS_EC	Nurses' Health Study	744	0.99	55.4(6.9)	12.5(1.4)	1933	Illumina Infinium Omni Express	MACH	ProbABEL	At what age did your menstrual periods begin?
Discovery - new	CAHRES_cases	Cancer Hormone Replacement Epidemiology in Sweden - cases	724	1.00	78.75(6.3)	13.5(1.4)	-	SNP!llumina HumanHap500	IMPUTE	PLINK/quicktest	How old were you when you had your first menstruation?
Discovery - new	NHS_GO	Nurses' Health Study	711	1.01	55.8(6.3)	12.5(1.4)	1932	Illumina Infinium Omni Express	MACH	ProbABEL	At what age did your menstrual periods begin?
Discovery - new	CAHRES_ctrls	Cancer Hormone Replacement Epidemiology in Sweden - controls	677	0.99	79.08(6.4)	13.6(1.4)	-	SNP!llumina HumanHap500	IMPUTE	PLINK/quicktest	How old were you when you had your first menstruation?
Discovery - new	INGI_FVG	Italian Network on Genetic Isolates - Friuli Venezia Giulia	679	1.03	50.6(16.7)	13.1(1.6)	-	Illumina Infinium 370	MACH	ProbABEL	How old were you when you had your first menstrual period?
Discovery - new	TRAILS_Pop	Tracking Adolescents' Individual Lives Survey -Population cohort	671	1.00	16.2 (0.7)	12.8(1.2)	1990	Illumina CytosNP12 v2	IMPUTE2	SNPTEST	How old were you when you had your first menstrual period (and... months)?
Discovery - new	Raine	Western Australian Pregnancy (Raine) Study	614	1.01	22.77(0.7)	12.8 (1.1)	1990	Illumina 660 Quad	MACH	skameta/proBABEL	Primary care giver recorded dates and duration of first three menses and returned to study coordinator.
Discovery - new	SHIP-TREND	Study of Health in Pomerania -TREND	543	1.00	50.1(13.2)	13.3(1.5)	1959	Illumina Human Omni 2.5	IMPUTE	QUICKTEST	At what age did your menstrual periods begin?
Discovery - new	INGI_Carantino	Italian Network on Genetic Isolates - Carantino	314	1.03	47.2 (17.2)	12.9 (1.6)	-	Illumina Infinium 370	MACH	ProbABEL	How old were you when you had your first menstrual period?
Discovery - new	NHS2_BRCA	Nurses' Health Study	298	1.01	38.0(4.1)	12.4(1.4)	1952	Illumina Humanmap610K	MACH	ProbABEL	At what age did your menstrual periods begin?
Discovery - new	IUBC	Italy	227	1.04	50.8 (12.9)	12.5(1.4)	1958	Illumina HumanOmni2.5-8v1_A	1.0-1.8	ProbABEL 0.1	"how old were you when you had your first menstrual years old?"
Discovery - new	TRAILS_CC	Tracking Adolescents' Individual Lives Survey	95	0.967	15.8 (0.6)	12.7 (1.2)	1994	Illumina CytosNP12 v2	IMPUTE2	SNPTEST	How old were you when you had your first menstrual period (and...months)?

Supplementary Table 1 (continued) | Details of the 58 studies that contributed to the discovery phase for age at menarche.

Type ^a	Study Name / acronym	Full Study name	N	GC Factor	Mean age (SD)	Mean IAM (SD)	Mean birth year	SNP array	Imputation program	Analysis program	Specific Menarche question
Breast Cancer Association Consortium (cases):											
Discovery-iCOGs	BCAC Cases;	Ansterdam Breast Cancer Study	27645	1.06	45.8 (6.7)	13.1 (1.4)	1964	Illumina iSelect "ICOGS"	IMPUTEv2	In House	At what age did you have your first period?
Discovery-iCOGs	ABCs	Bavarian Breast Cancer Cases	507	60.3 (12.1)	13.4 (1.5)	1947	Illumina iSelect "ICOGS"				At what age you had your first menstrual period?
Discovery-iCOGs	BBCC	British Breast Cancer Study	525	55.6 (8.8)	12.6 (1.5)	1949	Illumina iSelect "ICOGS"				How old were you when your periods began?
Discovery-iCOGs	BBCS	CECILE Breast Cancer Study	204	54.4 (10.6)	12.9 (1.6)	1951	Illumina iSelect "ICOGS"				What year did you have your first period?
Discovery-iCOGs	CECILE	Copenhagen General Population Study	1002	61.3 (12.3)	13.5 (1.4)	1943	Illumina iSelect "ICOGS"				At what age did you have your first menstruation? ____ years old
Discovery-iCOGs	CGPS	Spanish National Cancer Centre Breast Cancer Study	1625	54.7 (12.1)	12.8 (1.4)	1951	Illumina iSelect "ICOGS"				At what age did you have your first period?
Discovery-iCOGs	CNIO-BCS	California Teachers Study	108	55.7 (8.1)	12.5 (1.4)	1939	Illumina iSelect "ICOGS"				At what age did you have your first period?
Discovery-iCOGs	CTS	ESTHER Breast Cancer Study	50	56.1 (12.1)	13.4 (1.6)	1941	Illumina iSelect "ICOGS"				At what age did your regular bleeding began?
Discovery-iCOGs	GENICA	Gene Environment Interaction and Breast Cancer in Germany	458	57.0 (10.9)	13.3 (1.5)	1945	Illumina iSelect "ICOGS"				At what age did the first menstrual period (menarche)?
Discovery-iCOGs	HEBCS	Helsinki Breast Cancer Study	361	57.4 (12.1)	13.2 (1.5)	1943	Illumina iSelect "ICOGS"				At what age did your periods start?
Discovery-iCOGs	KARBAC	Karolinska Breast Cancer Study	356	60.2 (11.8)	13.4 (1.4)	1938	Illumina iSelect "ICOGS"				At what age did you have your first period?
Discovery-iCOGs	KBCP	Kuopio Breast Cancer Project	402	58.4 (14.0)	13.7 (1.5)	1934	Illumina iSelect "ICOGS"				At what age did you have your first periods?
Discovery-iCOGs	kConFab/AOCS	Kathleen Cunningham Foundation Consortium for Research into Familial Breast Cancer/Australian Ovarian Cancer Study	575	53.5 (10.9)	13.0 (1.4)	1950	Illumina iSelect "ICOGS"				Age at first menstrual period:
Discovery-iCOGs	LMBC	Leuven Multidisciplinary Breast Centre	1797	56.8 (12.1)	13.3 (1.5)	1950	Illumina iSelect "ICOGS"				Age of menarche
Discovery-iCOGs	MARIE	Mammary Carcinoma Risk Factor Investigation	1581	62.3 (6.2)	13.5 (1.6)	1941	Illumina iSelect "ICOGS"				How old were you at the time of your first period?
Discovery-iCOGs	MBCSG	Milan Breast Cancer Study Group	177	45.4 (11.8)	12.4 (1.5)	1960	Illumina iSelect "ICOGS"				Age at first menstruation
Discovery-iCOGs	MBCS	Mayo Clinic Breast Cancer Study	1653	57.2 (12.4)	12.7 (1.4)	1948	Illumina iSelect "ICOGS"				How old were you when you had your first menstrual period?
Discovery-iCOGs	MCCS	Melbourne Collaborative Cohort Study	608	56.7 (8.1)	13.1 (1.5)	1936	Illumina iSelect "ICOGS"				How old were you when you had your first menstrual period?
Discovery-iCOGs	OBCS	Oulu Breast Cancer Study	412	56.2 (11.5)	13.4 (1.5)	1947	Illumina iSelect "ICOGS"				At what age did you have your first period?
Discovery-iCOGs	OFBCR	Ontario Family Breast Cancer Registry	985	56.8 (10.3)	12.6 (1.4)	1942	Illumina iSelect "ICOGS"				At what age did you have your first menstrual period?
Discovery-iCOGs	ORIGO	Leiden University Medical Centre Breast Cancer Study	266	57.2 (10.8)	13.2 (1.6)	1944	Illumina iSelect "ICOGS"				At what age did you have your first period?
Discovery-iCOGs	PBCS	NCI Polish Breast Cancer Study	506	56.4 (10.0)	13.5 (1.6)	1945	Illumina iSelect "ICOGS"				At what age did you have your first menstrual period?
Discovery-iCOGs	pKARMA	Karolinska Mammography Project for Risk Prediction of Breast Cancer - prevalent cases	5273	62.9 (9.8)	13.2 (1.4)	1946	Illumina iSelect "ICOGS"				At what age did you have your first menstruation?
Discovery-iCOGs	RBCS	Rotterdam Breast Cancer Study	175	41.9 (8.5)	13.0 (1.5)	1953	Illumina iSelect "ICOGS"				At what age did you first menstruate?
Discovery-iCOGs	SASBAC	Singapore and Sweden Breast Cancer Study	1059	63.4 (6.5)	13.5 (1.4)	1931	Illumina iSelect "ICOGS"				How old were you...at menarche?
Discovery-iCOGs	SBCS	Sheffield Breast Cancer Study	820	62.8 (11.9)	13.0 (1.6)	1937	Illumina iSelect "ICOGS"				Age at menarche
Discovery-iCOGs	SEARCH	Study of Epidemiology and Risk factors in Cancer Heredity	5928	55.4 (8.9)	12.8 (1.5)	1946	Illumina iSelect "ICOGS"				How old were you when you had your first menstrual period?
Discovery-iCOGs	SKDKFZ	Städtisches Klinikum Karlsruhe Deutsches Krebsforschungszentrum Study	102	58.0 (12.8)	13.7 (1.4)	1943	Illumina iSelect "ICOGS"				How old were you when you had your first menstrual period?

Supplementary Table 1 (continued) | Details of the 58 studies that contributed to the discovery phase for age at menarche.

Type ^a	Study Name / acronym	Full Study name	N	GC Factor	Mean age (SD)	Mean AAM (SD)	Mean birth year	SNP array	Imputation program	Analysis program	Specific Menarche question
Breast Cancer Association Consortium (controls):											
Discovery-iCOGs	ABCFS	Australian Breast Cancer Family Study	549		41.9 (9.3)	12.9 (1.6)	1954	Illumina iSelect "iCOGS"	IMPUTEv2	In House	Age at first menstrual period: _____ years
Discovery-iCOGs	ABCS	Amsterdam Breast Cancer Study	1159		47.7 (12.2)	13.0 (1.4)	1962	Illumina iSelect "iCOGS"			At what age did you have your first period?
Discovery-iCOGs	BBCC	Bavarian Breast Cancer Controls	371		57.5 (10.9)	13.4 (1.4)	1951	Illumina iSelect "iCOGS"			At what age you had your first menstrual period?
Discovery-iCOGs	BBCS	British Breast Cancer Study	210		51.5 (12.0)	12.8 (1.4)	1953	Illumina iSelect "iCOGS"			How old were you when your periods began?
Discovery-iCOGs	CECILE	CECILE Breast Cancer Study	978		54.6 (11.0)	13.0 (1.6)	1951	Illumina iSelect "iCOGS"			What year did you have your first period?
Discovery-iCOGs	CTS	California Teachers Study	44		55.6 (9.5)	12.5 (1.2)	1939	Illumina iSelect "iCOGS"			At what age did you have your first period?
Discovery-iCOGs	GENICA	Gene Environment Interaction and Breast Cancer in Germany	415		57.1 (11.8)	13.6 (1.6)	1945	Illumina iSelect "iCOGS"			At what age did the first menstrual period (menarche)?
Discovery-iCOGs	KBCP	Kuopio Breast Cancer Project	241		52.6 (11.5)	13.6 (1.4)	1940	Illumina iSelect "iCOGS"			At what age did you have your first periods?
Discovery-iCOGs	kConFab/AOCS	Kathleen Cushingham Foundation Consortium for research into Familial Breast Cancer/Australian Ovarian Cancer Study	486		61.7 (8.8)	13.1 (1.5)	1944	Illumina iSelect "iCOGS"			Age at first menstrual period:
Discovery-iCOGs	MARIE	Mammary Carcinoma Risk Factor Investigation	1474		61.6 (6.1)	13.5 (1.6)	1941	Illumina iSelect "iCOGS"			How old were you at the time of your first period?
Discovery-iCOGs	MBCS	Mayo Clinic Breast Cancer Study	1679		56.3 (14.0)	12.8 (1.3)	1950	Illumina iSelect "iCOGS"			How old were you when you had your first menstrual period?
Discovery-iCOGs	MCCS	Melbourne Collaborative Cohort Study	508		56.3 (8.3)	12.9 (1.5)	1937	Illumina iSelect "iCOGS"			How old were you when you had your first menstrual period?
Discovery-iCOGs	OFBCR	Ontario Familial Breast Cancer Registry	501		52.0 (9.2)	12.6 (1.6)	1947	Illumina iSelect "iCOGS"			At what age did you have your first menstrual period?
Discovery-iCOGs	PBCS	NCL Polish Breast Cancer Study	409		56.2 (9.9)	13.6 (1.6)	1945	Illumina iSelect "iCOGS"			At what age did you have your first menstrual period?
Discovery-iCOGs	pKARMA	Karolinska Mammography Project for Risk Prediction of Breast Cancer - prevalent cases	5337		53.9 (9.5)	13.1 (1.4)	1957	Illumina iSelect "iCOGS"			At what age did you have your first menstruation?
Discovery-iCOGs	SASBAC	Singapore and Sweden Breast Cancer Study	1251		63.2 (6.4)	13.5 (1.4)	1931	Illumina iSelect "iCOGS"			How old were you...at menarche?
Discovery-iCOGs	SBCS	Sheffield Breast Cancer Study	835		57.5 (5.7)	13.0 (1.7)	1943	Illumina iSelect "iCOGS"			Age at menarche
Discovery-iCOGs	SEARCH	Study of Epidemiology and Risk factors in Cancer Heredity	4556		59.5 (8.2)	12.9 (1.6)	1939	Illumina iSelect "iCOGS"			How old were you when you had your first menstrual period?
Replication	InterAct	EPIC-InterAct	8869	n/a	53.7 (8.7)	13.1 (1.6)	1941	Illumina Human660W-Quad BeadChip / Illumina HumanCoreExome	IMPUTE	GCTA (LMM)	How old were you when you had your first menstrual period?

*Discovery previous refers to studies contributing to the last ReproGen GWAS meta-analysis for age at menarche [Elks et al Nat Genet 2010 Dec;42(12):1077-8]. Discovery-new refers to additional studies contributing to GWAS meta-analysis. AAM: Age at menarche studies genotyped with the Illumina iSelect "iCOGS" array. GC factor: genomic control factor. AAM: Age at menarche

Supplementary Table 4 (continued) | Follow up of menarche-associated SNPs for association with puberty timing in the EGGG consortium.

Locus	SNP	Location	Consensus Gene	Association of menarche raising allele with Tanner Stage in both sexes				Association of menarche raising allele with Tanner Stage in boys				Association of menarche raising allele with Tanner Stage in girls					
				Menarche Raising Allele	Menarche Other Allele	n	Beta	SE	P	n	Beta	SE	P	n	Beta	SE	
61	rs7853970	985905386	RM1(N), NTRK2 (C)	t	c	9912	-0.018	0.014	0.186	3769	-0.005	0.024	0.835	6143	-0.025	0.017	0.139
62a	rs10816359	9-10797491	TMEM38B (N)	t	g	9914	-0.036	0.020	0.083	3769	-0.059	0.035	0.088	6145	-0.023	0.025	0.361
62b	rs10453225	9-107960041	TMEM38B (N)	g	t	9916	-0.060	0.014	2.490E-05	3769	-0.074	0.025	0.003	6147	-0.053	0.017	0.002
62c	rs10739221	9-108100651	TMEM38B (N)	c	t	9914	-0.048	0.016	0.003	3769	-0.056	0.029	0.048	6145	-0.044	0.019	0.023
63	rs11792861	9-110849116	TMEM245 (N,E)	a	c	9914	-0.007	0.015	0.651	3769	0.013	0.026	0.601	6145	-0.016	0.018	0.358
64	rs10980854	9-113090178	ZNF483 / OR2R2 (N)	a	g	9916	-0.020	0.029	0.489	3769	-0.025	0.050	0.619	6147	-0.018	0.035	0.617
64	rs10980921	9-113319733	ZNF483 / OR2R2 (N)	c	t	9915	0.003	0.024	0.893	3769	-0.001	0.043	0.939	6146	0.005	0.030	0.864
65	rs1874984	10-1721871	ADARB2 (N)	c	g	9913	-0.015	0.014	0.283	3769	-0.029	0.025	0.243	6144	-0.009	0.017	0.613
66	rs12571664	10-121698919	SEC23IP (N,E)	t	c	9914	-0.019	0.017	0.249	3769	-0.022	0.029	0.451	6145	-0.018	0.020	0.376
67	rs1915146	10-126836204	CTBP2 (N,C)	g	a	9913	-0.007	0.014	0.605	3769	0.009	0.024	0.701	6144	-0.015	0.017	0.366
68	rs7104764	11-219977	SIRT3 (N,E,C)	g	a	9916	-0.042	0.015	0.006	3769	-0.027	0.026	0.306	6147	-0.050	0.019	0.008
69	rs4929947	11-85956570	TRIM66 (N,E,F)	g	c	9915	-0.017	0.014	0.215	3769	-0.014	0.024	0.554	6146	-0.019	0.017	0.269
70	rs110227256	11-132272015	ARNTL (N), PTH (C)	a	c	9915	-0.044	0.015	0.003	3769	-0.045	0.025	0.078	6146	-0.043	0.018	0.017
71	rs7103411	11-21656701	BDNF (N,C), LGR4 (C)	c	t	9916	-0.021	0.016	0.202	3769	-0.038	0.028	0.169	6147	-0.012	0.020	0.556
72	rs16918636	11-29080758	FSHB (N[1'mbj]C)	t	c	9837	-0.015	0.016	0.350	3759	-0.038	0.028	0.170	6138	-0.003	0.020	0.861
73	rs4756059	11-46107195	PHF21A (N)	t	c	9914	-0.021	0.025	0.409	3769	-0.022	0.044	0.633	6145	-0.020	0.030	0.504
74	rs2063730	11-77726172	GAB2 (N), THrsp (C)	c	a	9915	0.000	0.018	0.989	3769	-0.036	0.030	0.230	6146	0.018	0.022	0.398
75	rs10895140	11-109494931	TRPC6 (N), FGRC (C)	g	a	9915	-0.004	0.014	0.774	3769	-0.003	0.024	0.886	6146	-0.004	0.017	0.801
76	rs11215400	11-114557845	CADM1 (N)	c	a	9914	-0.013	0.015	0.401	3769	0.017	0.026	0.527	6145	-0.027	0.019	0.141
77	rs1461503	11-122350285	BSX (N,C)	c	a	9915	-0.026	0.013	0.053	3769	-0.007	0.023	0.765	6146	-0.035	0.016	0.030
78	rs7955374	12-46166416	VDR (C)	t	c	9914	-0.024	0.021	0.252	3769	-0.010	0.036	0.775	6145	-0.031	0.026	0.228
79	rs7158833	12-48533735	BCDIN3B (N)	g	a	9913	-0.028	0.014	0.041	3766	-0.037	0.023	0.114	6147	-0.023	0.017	0.164
80	rs6553739	13-33137785	C0G6 (N,E)	g	t	9916	-0.010	0.014	0.490	3769	-0.019	0.024	0.434	6147	-0.005	0.017	0.768
81	rs1324913	13-73533589	KLF12 (N)	g	t	9914	-0.009	0.014	0.517	3769	0.050	0.024	0.041	6145	-0.038	0.017	0.027
82	rs9560113	13-11081349	TEX29 (N)	g	a	9915	-0.034	0.015	0.021	3769	-0.026	0.025	0.34	6146	-0.039	0.018	0.033
83	rs1254337	14-59990278	SX6 (N)	t	a	9915	-0.008	0.015	0.580	3769	0.013	0.025	0.618	6146	-0.019	0.018	0.302
84	rs1958560	14-65106548	FUT8 (N,E)	a	g	9914	-0.011	0.013	0.395	3769	-0.027	0.023	0.250	6145	-0.004	0.016	0.812
85a	rs10144321	14-99953158	DLK1 (C)	a	g	9914	-0.028	0.016	0.075	3769	-0.024	0.027	0.380	6145	-0.030	0.019	0.117
85b	rs7141210	14-100252223	DLK1 (N,E,C)	t	c	9914	0.004	0.014	0.769	3769	0.036	0.025	0.157	6145	-0.011	0.018	0.531
86	rs12148769	15-21703187	MKRN3 (C), MAGEL2 (C)	g	a	9914	-0.025	0.022	0.265	3768	-0.032	0.038	0.405	6146	-0.021	0.027	0.434
87	rs3743266	15-58568805	RORA (N,C)	t	c	9912	-0.048	0.014	0.001	3769	-0.033	0.025	0.175	6143	-0.055	0.017	0.002
88	rs802675	15-65746518	MAP2KS (N)	t	c	9915	0.006	0.013	0.660	3769	0.014	0.023	0.541	6146	0.002	0.016	0.912
89	rs2915845	15-858343471	DEF1 (N,E)	c	t	9911	-0.039	0.013	0.004	3769	-0.073	0.023	0.002	6142	-0.022	0.016	0.176
90	rs246185	16-14302933	MKL2 (N)	c	t	9913	-0.057	0.015	1.970E-04	3769	-0.131	0.026	5.010E-07	6144	-0.019	0.019	0.309

Supplementary Table 4 (continued) | Follow up of menarche-associated SNPs for association with puberty timing in the EGG consortium.

Locus	SNP	Location	Consensus Gene	Menarche Raising Allele		Menarche Other Allele		Association of menarche raising allele with Tanner Stage in both sexes		Association of menarche raising allele with Tanner Stage in boys		Association of menarche raising allele with Tanner Stage in girls					
				n	Beta	n	SE	P	n	Beta	SE	P	n				
91	rs12446632	16-19842890	GPRC5B(N,C)	a	g	9914	-0.058	0.019	0.003	3768	-0.085	0.033	0.011				
92	rs1129700	16-29825535	KCTD13(N),TRX6(E,C)	t	c	9907	-0.016	0.014	0.248	3768	-0.012	0.023	0.610	6139	-0.018	0.017	0.288
93	rs050136	16-52373776	FTO(N,C)	c	a	9914	-0.028	0.013	0.040	3769	0.001	0.023	0.982	6145	-0.042	0.016	0.011
94a	rs1364063	16-58146073	COG4(C),NFAT5(N)	c	t	9913	-0.014	0.013	0.301	3769	-0.022	0.023	0.350	6144	-0.010	0.016	0.538
94b	rs929843	16-68603249	COG4(C),WWP2(N)	a	c	9914	0.003	0.017	0.881	3769	0.035	0.030	0.242	6145	-0.013	0.021	0.532
95	rs7215990	17-5975555	WSD1(N,E),ALOX15B(E)	g	a	9915	-0.008	0.016	0.604	3769	-0.025	0.028	0.372	6146	0.000	0.019	0.990
96	rs9635759	17-46968784	CA10(N)	a	g	9912	-0.017	0.015	0.254	3769	0.039	0.026	0.129	6143	-0.046	0.018	0.013
97	rs244293	17-50585721	STXBP4(N,E)	g	a	9915	-0.023	0.014	0.087	3769	-0.039	0.023	0.093	6146	-0.015	0.017	0.355
98	rs12607903	18-3807134	DLGAP1(N)	c	t	9914	-0.008	0.015	0.608	3769	-0.029	0.026	0.264	6145	0.003	0.018	0.889
99	rs12137289	18-43006123	SKOR2(N)	a	g	9914	-0.006	0.014	0.664	3769	0.007	0.024	0.752	6145	-0.012	0.017	0.453
100	rs652260	19-7806562	EV15L(N),RETIN(C)	t	c	9912	-0.014	0.013	0.290	3769	0.009	0.023	0.710	6143	-0.026	0.016	0.118
101	rs889122	19-9856667	OLFML2(N),RDR8(C)	g	t	9914	-0.032	0.015	0.030	3769	-0.042	0.026	0.109	6145	-0.028	0.018	0.123
102	rs10423674	19-18678903	CRTC1(N,C)	a	c	9915	-0.031	0.014	0.030	3769	-0.002	0.024	0.951	6146	-0.045	0.017	0.009
103	rs852069	20-17070593	PCSK2(N,C)	g	a	9916	-0.006	0.014	0.658	3769	0.011	0.024	0.632	6147	-0.015	0.017	0.375
104	rs2836950	21-3956299	BRWD1(N,C)	c	g	9914	-0.023	0.014	0.093	3769	-0.020	0.024	0.396	6145	-0.025	0.017	0.142
105	rs13053505	22-37575564	NPTXR(N,E),CBX7(C)	g	t	9913	-0.025	0.017	0.152	3769	-0.004	0.030	0.898	6144	-0.035	0.021	0.095
106	rs6009583	22-48063650	C22orf34(N)	c	t	9916	-0.019	0.015	0.211	3769	-0.042	0.026	0.110	6147	-0.008	0.018	0.673

* Together, the 106 menarche loci explained 2.15% of the variance in Tanner stage in boys and girls in ALSPAC, the largest single study in the EGG Consortium (n=3009 girls and n=2373 boys at age 14 years).

Supplementary Table 5 | Parent-of-Origin specific associations with age at menarche for loci in imprinted / non-imprinted regions, in the DeCODE study.

Position	SNP	Ref allele	Paternal		Maternal		P_{het}	Imputation status	Imputed region
			Beta	P value	Beta	P value			
14-100252223	rs7141210	T	0.06 (0.03 - 0.08)	2.1E-04	-0.02 (-0.05 - 0.01)	1.2E-01	0.00021	Imprinted	99763005-100897120
19-18678903	rs10423674	C	-0.05 (-0.08 - -0.02)	1.5E-03	0.03 (0 - 0.06)	7.7E-02	0.00047	Not Imprinted	-
8-140720961	rs1469039	G	-0.01 (-0.04 - -0.03)	7.4E-01	-0.08 (-0.12 - -0.05)	5.7E-06	0.0029	Imprinted	140182263-141284481
15-21703187	rs12148769	G	0.11 (0.07 - 0.16)	2.5E-06	0.02 (-0.03 - 0.07)	4.3E-01	0.0058	Imprinted	20861546-21983542
7-41436618	rs1079866	C	-0.02 (-0.06 - -0.02)	3.2E-01	-0.1 (-0.14 - -0.06)	1.4E-06	0.0061	Not Imprinted	-
14-99952158	rs10144321	A	0.07 (0.04 - 0.11)	3.1E-05	0.01 (-0.02 - 0.05)	4.7E-01	0.015	Imprinted	99763005-100897120
13-110981349	rs9560113	A	-0.05 (-0.08 - -0.02)	1.1E-03	0 (-0.03 - 0.03)	8.8E-01	0.027	Not Imprinted	-
1-176156103	rs543874	A	0 (-0.03 - 0.04)	8.4E-01	0.06 (0.02 - 0.09)	2.4E-03	0.045	Not Imprinted	-
3-129377916	rs2687729	A	-0.03 (-0.06 - 0)	5.9E-02	0.01 (-0.02 - 0.05)	3.9E-01	0.052	Not Imprinted	-
3-134098154	rs2600959	A	0.04 (0.01 - 0.07)	4.7E-03	0 (-0.03 - 0.03)	7.7E-01	0.073	Not Imprinted	-
2-199983454	rs17266097	C	-0.02 (-0.05 - 0.01)	1.2E-01	-0.06 (-0.09 - -0.03)	4.5E-05	0.074	Not Imprinted	-
5-43152587	rs1532331	G	0 (-0.03 - 0.03)	9.4E-01	0.03 (0 - 0.06)	2.5E-02	0.099	Not Imprinted	-
13-73533589	rs1324913	G	0 (-0.03 - 0.03)	9.0E-01	0.03 (0 - 0.06)	3.0E-02	0.1	Not Imprinted	-
6-128432673	rs6938574	T	0.05 (0.01 - 0.08)	1.6E-02	0 (-0.03 - 0.04)	8.8E-01	0.11	Not Imprinted	-
11-8596570	rs4929947	G	0.05 (0.02 - 0.08)	2.9E-03	0.01 (-0.02 - 0.04)	4.3E-01	0.12	Not Imprinted	-
16-19842890	rs12446632	G	-0.02 (-0.07 - 0.02)	3.2E-01	-0.08 (-0.12 - -0.03)	1.4E-03	0.12	Not Imprinted	-
14-65106548	rs1958560	G	0 (-0.03 - 0.03)	9.5E-01	-0.03 (-0.06 - 0)	3.0E-02	0.13	Not Imprinted	-
12-48533735	rs7138803	G	0.01 (-0.02 - 0.04)	3.4E-01	0.05 (0.02 - 0.08)	2.3E-03	0.14	Not Imprinted	-
2-156460705	rs17236969	C	-0.01 (-0.06 - 0.03)	5.7E-01	-0.06 (-0.1 - -0.01)	9.0E-03	0.15	Not Imprinted	-
21-39526299	rs2836950	C	0.04 (0.01 - 0.07)	2.2E-02	0 (-0.03 - 0.03)	8.0E-01	0.15	Not Imprinted	-
6-41998960	rs2479724	T	0.04 (0.02 - 0.07)	2.4E-03	0.01 (-0.01 - 0.04)	3.1E-01	0.16	Not Imprinted	-
1-163661506	rs466639	T	-0.07 (-0.11 - -0.03)	1.2E-03	-0.03 (-0.07 - 0.01)	1.8E-01	0.18	Not Imprinted	-
18-43006123	rs2137289	G	-0.06 (-0.09 - -0.03)	2.7E-05	-0.03 (-0.06 - -0.01)	1.9E-02	0.19	Imprinted	42308570-43310446
4-95426711	rs13135934	G	-0.04 (-0.07 - -0.01)	1.1E-02	-0.01 (-0.04 - 0.02)	4.8E-01	0.19	Not Imprinted	-
2-59734549	rs268067	G	-0.04 (-0.08 - -0.01)	1.8E-02	-0.01 (-0.04 - 0.02)	5.4E-01	0.21	Not Imprinted	-
10-121698919	rs12571664	T	0 (-0.04 - 0.03)	8.5E-01	0.03 (-0.01 - 0.06)	1.2E-01	0.22	Not Imprinted	-
9-85905386	rs7853970	T	0.03 (0 - 0.06)	6.7E-02	0.05 (0.02 - 0.08)	4.0E-04	0.23	Not Imprinted	-
1-98148036	rs11165924	A	0.02 (-0.01 - 0.05)	1.2E-01	0 (-0.03 - 0.03)	9.2E-01	0.24	Not Imprinted	-
18-3807134	rs12607903	C	0.06 (0.03 - 0.09)	2.9E-04	0.03 (0 - 0.06)	5.1E-02	0.24	Not Imprinted	-
3-119045126	rs11715566	C	-0.02 (-0.05 - 0.01)	1.3E-01	-0.05 (-0.08 - -0.02)	1.4E-03	0.24	Not Imprinted	-
2-141944979	rs12472911	C	0.03 (0 - 0.07)	5.5E-02	0.06 (0.03 - 0.09)	4.6E-04	0.26	Not Imprinted	-
11-46107195	rs4756059	C	-0.09 (-0.15 - -0.03)	2.4E-03	-0.05 (-0.11 - 0.02)	1.5E-01	0.27	Not Imprinted	-
16-68603249	rs929843	A	0.02 (-0.01 - 0.05)	2.6E-01	0.04 (0.01 - 0.08)	8.3E-03	0.29	Not Imprinted	-
9-7164673	rs913588	G	0.02 (-0.01 - 0.05)	1.8E-01	0.04 (0.01 - 0.07)	4.5E-03	0.29	Not Imprinted	-
3-158281469	rs900400	T	0 (-0.03 - 0.03)	9.0E-01	0.02 (-0.01 - 0.05)	1.1E-01	0.3	Not Imprinted	-
11-27656701	rs7103411	C	0.04 (0 - 0.08)	2.8E-02	0.07 (0.03 - 0.11)	3.2E-04	0.32	Not Imprinted	-
10-126836204	rs1915146	A	-0.02 (-0.05 - 0.01)	1.2E-01	0 (-0.03 - 0.03)	8.7E-01	0.33	Not Imprinted	-
3-49485935	rs7647973	G	-0.04 (-0.07 - -0.01)	2.0E-02	-0.02 (-0.05 - 0.02)	3.1E-01	0.35	Not Imprinted	-
5-167302841	rs9647570	T	-0.01 (-0.05 - 0.03)	6.2E-01	-0.04 (-0.08 - 0)	6.8E-02	0.35	Not Imprinted	-
4-104860552	rs3733631	G	-0.07 (-0.11 - -0.03)	2.5E-04	-0.05 (-0.09 - -0.01)	2.1E-02	0.36	Not Imprinted	-

4-44877284	rs10938397	A	0.04 (0.01 - 0.07)	3.6E-03	0.02 (0 - 0.05)	1.0E-01	0.37	Not Imprinted	-
2-604168	rs2947411	A	0.05 (0.01 - 0.08)	2.0E-02	0.07 (0.03 - 0.11)	3.2E-04	0.38	Not Imprinted	-
9-113319733	rs10980921	T	-0.09 (-0.14 - -0.04)	7.0E-04	-0.06 (-0.11 - 0)	3.2E-02	0.38	Not Imprinted	-
15-65746518	rs8032675	T	0.04 (0.01 - 0.07)	6.5E-03	0.06 (0.03 - 0.09)	8.9E-05	0.39	Not Imprinted	-
16-68146073	rs1364063	T	-0.03 (-0.06 - 0)	4.8E-02	-0.05 (-0.08 - -0.02)	1.4E-03	0.39	Not Imprinted	-
6-54864267	rs988913	C	0.03 (0 - 0.06)	5.5E-02	0.01 (-0.02 - 0.04)	4.5E-01	0.41	Not Imprinted	-
19-9856867	rs889122	G	0.05 (0.02 - 0.08)	2.4E-03	0.03 (0 - 0.06)	5.4E-02	0.43	Not Imprinted	-
6-30030719	rs16896742	A	-0.04 (-0.07 - -0.01)	9.8E-03	-0.02 (-0.05 - 0.01)	1.4E-01	0.44	Not Imprinted	-
9-107797491	rs10816359	T	0.04 (0 - 0.08)	5.0E-02	0.02 (-0.02 - 0.06)	3.6E-01	0.46	Not Imprinted	-
11-114557845	rs11215400	A	-0.02 (-0.05 - 0.01)	2.1E-01	-0.04 (-0.07 - -0.01)	2.3E-02	0.47	Not Imprinted	-
4-28361152	rs1038903	C	0 (-0.04 - 0.03)	8.0E-01	0.01 (-0.02 - 0.04)	4.3E-01	0.47	Not Imprinted	-
1-203951975	rs951366	T	0.02 (-0.01 - 0.05)	2.2E-01	0.03 (0 - 0.06)	2.9E-02	0.5	Not Imprinted	-
11-13272015	rs11022756	A	0.05 (0.01 - 0.08)	5.2E-03	0.03 (0 - 0.06)	6.7E-02	0.5	Not Imprinted	-
6-105455237	rs2153127	T	0.06 (0.04 - 0.09)	9.3E-06	0.05 (0.02 - 0.08)	5.1E-04	0.5	Not Imprinted	-
1-72523773	rs3101336	T	0.02 (-0.01 - 0.04)	2.9E-01	0.03 (0 - 0.06)	4.5E-02	0.51	Not Imprinted	-
3-138472681	rs13067731	C	-0.03 (-0.06 - 0.01)	1.6E-01	-0.01 (-0.05 - 0.03)	6.3E-01	0.51	Not Imprinted	-
6-126823127	rs4895808	C	0.03 (0 - 0.05)	8.2E-02	0.04 (0.01 - 0.07)	8.9E-03	0.53	Not Imprinted	-
17-46968784	rs9635759	G	-0.06 (-0.09 - -0.03)	8.5E-05	-0.05 (-0.08 - -0.02)	2.3E-03	0.54	Not Imprinted	-
1-198064962	rs6427782	G	0 (-0.03 - 0.03)	8.7E-01	-0.01 (-0.04 - 0.01)	3.2E-01	0.56	Not Imprinted	-
5-133928412	rs13179411	G	-0.05 (-0.08 - -0.01)	1.5E-02	-0.06 (-0.1 - -0.02)	1.3E-03	0.56	Not Imprinted	-
9-107960041	rs10453225	G	0.08 (0.05 - 0.12)	2.4E-07	0.07 (0.04 - 0.1)	1.5E-05	0.56	Not Imprinted	-
3-24686017	rs6770162	G	-0.03 (-0.06 - 0)	4.4E-02	-0.02 (-0.05 - 0.01)	2.3E-01	0.57	Not Imprinted	-
2-199346935	rs1400974	G	-0.02 (-0.05 - 0.01)	1.9E-01	-0.03 (-0.06 - 0)	3.7E-02	0.59	Not Imprinted	-
8-4821198	rs7463166	A	0.04 (0.01 - 0.07)	1.1E-02	0.05 (0.02 - 0.08)	1.0E-03	0.59	Not Imprinted	-
9-110849116	rs11792861	A	0.03 (-0.01 - 0.06)	1.1E-01	0.01 (-0.02 - 0.05)	4.2E-01	0.59	Not Imprinted	-
15-86843471	rs12915845	C	0.02 (-0.01 - 0.05)	1.1E-01	0.03 (0.01 - 0.06)	2.0E-02	0.6	Not Imprinted	-
6-105207901	rs4946632	T	0.01 (-0.05 - 0.06)	8.4E-01	0.02 (-0.03 - 0.07)	3.6E-01	0.62	Not Imprinted	-
3-187118379	rs16860328	A	-0.04 (-0.07 - -0.01)	6.1E-03	-0.03 (-0.06 - 0)	4.1E-02	0.63	Not Imprinted	-
6-100222813	rs9321659	G	-0.03 (-0.07 - 0.02)	2.5E-01	-0.04 (-0.08 - 0)	6.9E-02	0.63	Not Imprinted	-
3-185528493	rs939317	A	-0.06 (-0.09 - -0.03)	1.5E-04	-0.05 (-0.08 - -0.02)	1.8E-03	0.64	Not Imprinted	-
9-10264080	rs7865468	G	-0.03 (-0.06 - 0)	8.0E-02	-0.02 (-0.05 - 0.01)	2.7E-01	0.64	Not Imprinted	-
17-50585721	rs244293	A	-0.01 (-0.04 - 0.02)	3.9E-01	-0.02 (-0.05 - 0.01)	1.3E-01	0.65	Not Imprinted	-
5-137752902	rs17171818	C	0.04 (0.01 - 0.08)	1.6E-02	0.03 (0 - 0.07)	7.6E-02	0.65	Not Imprinted	-
13-39137785	rs6563739	G	0.04 (0.01 - 0.07)	1.4E-02	0.03 (0 - 0.06)	7.0E-02	0.66	Not Imprinted	-
5-95871610	rs17086188	A	-0.01 (-0.09 - 0.07)	8.2E-01	0.02 (-0.06 - 0.09)	7.0E-01	0.67	Not Imprinted	-
7-121947978	rs11767400	C	-0.02 (-0.06 - 0.01)	1.3E-01	-0.02 (-0.05 - 0.02)	3.4E-01	0.7	Not Imprinted	-
20-17070593	rs852069	A	-0.03 (-0.06 - 0)	5.3E-02	-0.02 (-0.05 - 0.01)	1.6E-01	0.71	Not Imprinted	-
22-48063650	rs6009583	C	0.03 (0 - 0.06)	6.9E-02	0.02 (-0.01 - 0.06)	1.9E-01	0.71	Not Imprinted	-
6-100315159	rs4840086	A	0.05 (0.02 - 0.08)	5.4E-04	0.04 (0.01 - 0.07)	3.5E-03	0.71	Not Imprinted	-
8-3754618	rs2688325	T	0.05 (0.02 - 0.08)	2.1E-03	0.04 (0.01 - 0.07)	1.1E-02	0.71	Not Imprinted	-
14-59990278	rs1254337	A	-0.02 (-0.05 - 0.01)	1.6E-01	-0.03 (-0.06 - 0)	5.5E-02	0.72	Not Imprinted	-
6-101240798	rs239198	C	-0.03 (-0.06 - 0)	6.6E-02	-0.02 (-0.05 - 0.01)	1.9E-01	0.72	Not Imprinted	-
1-65589155	rs10789181	A	0.02 (-0.01 - 0.05)	2.3E-01	0.02 (0 - 0.05)	9.8E-02	0.74	Not Imprinted	-
10-1721871	rs1874984	G	-0.03 (-0.06 - 0)	4.2E-02	-0.02 (-0.05 - 0.01)	1.2E-01	0.74	Not Imprinted	-

12-46166416	rs7955374	C	-0.04 (-0.09 - 0)	3.1E-02	-0.04 (-0.08 - 0.01)	8.7E-02	0.75	Not Imprinted	-
6-151845447	rs6933660	C	0.03 (-0.01 - 0.06)	1.0E-01	0.03 (0 - 0.07)	3.6E-02	0.75	Not Imprinted	-
7-73739845	rs6964833	T	0.02 (-0.02 - 0.05)	3.3E-01	0.01 (-0.02 - 0.04)	6.1E-01	0.75	Not Imprinted	-
6-105485647	rs7759938	C	0.09 (0.06 - 0.12)	5.3E-09	0.1 (0.07 - 0.13)	3.7E-10	0.76	Not Imprinted	-
6-100866891	rs13196561	C	0.02 (-0.02 - 0.05)	3.5E-01	0.02 (-0.01 - 0.06)	1.8E-01	0.77	Not Imprinted	-
9-6932940	rs7037266	C	-0.02 (-0.05 - 0.01)	2.9E-01	-0.02 (-0.05 - 0.01)	1.4E-01	0.77	Not Imprinted	-
1-74779308	rs7514705	T	-0.03 (-0.06 - 0.01)	1.8E-02	-0.03 (-0.06 - 0)	5.0E-02	0.78	Not Imprinted	-
3-86999572	rs7642134	A	-0.03 (-0.06 - 0)	3.0E-02	-0.04 (-0.07 - 0.01)	1.0E-02	0.78	Not Imprinted	-
3-88323964	rs9849248	C	0.02 (-0.02 - 0.06)	3.6E-01	0.02 (-0.01 - 0.06)	2.0E-01	0.8	Not Imprinted	-
5-168682315	rs6555855	G	0.02 (-0.01 - 0.05)	2.7E-01	0.01 (-0.02 - 0.05)	4.5E-01	0.8	Not Imprinted	-
5-110887696	rs251130	G	0.04 (0.01 - 0.07)	8.3E-03	0.05 (0.02 - 0.08)	3.1E-03	0.82	Not Imprinted	-
11-219977	rs7104764	G	0.04 (0 - 0.07)	4.0E-02	0.04 (0.01 - 0.08)	1.9E-02	0.84	Not Imprinted	-
19-7806562	rs652260	C	-0.02 (-0.05 - 0.01)	1.8E-01	-0.02 (-0.04 - 0.01)	3.0E-01	0.84	Not Imprinted	-
8-53931766	rs16918254	A	0.05 (0 - 0.1)	6.1E-02	0.06 (0 - 0.11)	3.3E-02	0.84	Not Imprinted	-
8-144944399	rs4875053	C	-0.01 (-0.04 - 0.02)	5.1E-01	-0.01 (-0.03 - 0.02)	7.1E-01	0.84	Not Imprinted	-
22-37575564	rs13053505	G	0.04 (0 - 0.08)	4.2E-02	0.03 (0 - 0.07)	7.9E-02	0.85	Not Imprinted	-
2-199352283	rs17233066	C	0.03 (-0.03 - 0.09)	3.1E-01	0.02 (-0.04 - 0.08)	4.4E-01	0.86	Not Imprinted	-
15-58568805	rs3743266	T	0.02 (-0.02 - 0.05)	3.3E-01	0.02 (-0.01 - 0.05)	2.3E-01	0.87	Not Imprinted	-
16-29825535	rs1129700	T	0.01 (-0.02 - 0.04)	5.1E-01	0.01 (-0.02 - 0.03)	6.8E-01	0.87	Not Imprinted	-
17-5975555	rs7215990	G	0.04 (0.01 - 0.07)	1.5E-02	0.04 (0 - 0.07)	2.8E-02	0.87	Not Imprinted	-
8-4547489	rs7828501	A	-0.03 (-0.06 - 0)	4.2E-02	-0.03 (-0.06 - 0)	7.2E-02	0.87	Not Imprinted	-
11-100941931	rs10895140	A	-0.04 (-0.07 - 0.01)	2.0E-02	-0.03 (-0.06 - 0)	3.4E-02	0.89	Not Imprinted	-
16-52373776	rs8050136	C	0.05 (0.02 - 0.08)	3.5E-04	0.05 (0.02 - 0.08)	7.7E-04	0.89	Not Imprinted	-
2-105231258	rs6758290	T	0.03 (0 - 0.06)	2.4E-02	0.04 (0.01 - 0.06)	1.4E-02	0.89	Not Imprinted	-
3-50068213	rs6762477	G	0.01 (-0.02 - 0.04)	4.4E-01	0.01 (-0.01 - 0.04)	3.4E-01	0.9	Not Imprinted	-
1-102349609	rs11578152	A	-0.02 (-0.05 - 0.01)	1.5E-01	-0.02 (-0.05 - 0.01)	1.1E-01	0.91	Not Imprinted	-
6-56888700	rs9475752	C	0.02 (-0.02 - 0.05)	3.5E-01	0.02 (-0.02 - 0.06)	2.7E-01	0.91	Imprinted	56790380-57799120
5-153527602	rs7701886	G	-0.01 (-0.04 - 0.02)	6.5E-01	-0.01 (-0.04 - 0.02)	5.4E-01	0.92	Not Imprinted	-
11-29080758	rs16918636	T	0.03 (-0.01 - 0.06)	1.4E-01	0.03 (-0.01 - 0.07)	1.1E-01	0.93	Not Imprinted	-
11-122350285	rs1461503	A	-0.03 (-0.06 - 0)	5.9E-02	-0.03 (-0.05 - 0)	8.0E-02	0.93	Not Imprinted	-
2-56441253	rs6747380	G	-0.08 (-0.12 - -0.04)	7.9E-05	-0.08 (-0.12 - -0.04)	4.8E-05	0.93	Not Imprinted	-
9-108100651	rs10739221	T	-0.07 (-0.1 - -0.03)	2.9E-04	-0.06 (-0.1 - -0.03)	4.9E-04	0.94	Not Imprinted	-
11-77726172	rs2063730	A	-0.04 (-0.08 - 0)	5.5E-02	-0.04 (-0.08 - 0)	4.5E-02	0.95	Not Imprinted	-
2-156835210	rs4369815	T	0.04 (-0.01 - 0.09)	1.2E-01	0.04 (-0.01 - 0.09)	1.2E-01	0.97	Not Imprinted	-
9-113090178	rs10980854	G	-0.04 (-0.1 - 0.02)	1.7E-01	-0.04 (-0.1 - 0.02)	1.8E-01	0.97	Not Imprinted	-
16-14302933	rs246185	T	-0.04 (-0.07 - -0.01)	5.9E-03	-0.04 (-0.07 - -0.01)	5.4E-03	0.98	Not Imprinted	-
1-43894144	rs2274465	C	0.03 (0 - 0.06)	4.1E-02	0.03 (0 - 0.06)	4.4E-02	0.99	Not Imprinted	-
6-77224806	rs9447700	C	0.01 (-0.02 - 0.04)	3.6E-01	0.01 (-0.02 - 0.04)	3.5E-01	0.99	Not Imprinted	-
8-78256392	rs7821178	C	0.02 (-0.01 - 0.05)	2.3E-01	0.02 (-0.01 - 0.05)	2.3E-01	1	Not Imprinted	-

Supplementary Table 6 | eQTL results across multiple tissues.

See data file

Supplementary Table 7 | eQTL results in whole blood.

See data file

Supplementary Table 8 | Candidate genes at or near menarche loci.

Locus	SNP	Location	Consensus Gene	Gene name: function	Role in Hormone Function
1	rs2274465	1-43894144	KDM4A (N,C), PTPRF (E,C)	i) Lysine (K)-specific demethylase 4A: Stimulates ERalpha activity. Inhibits Ras-mediated CHDS induction, reducing p53 pathway activity. ii) Protein tyrosine phosphatase, receptor type, F: May contribute to pathogenesis of insulin resistance.	Y
2	rs10789181	1-65589155	LEPR (C)	Leptin receptor: Mediates adipocyte signalling on regulation of appetite and reproductive function.	Y
3	rs3101336	1-72523773	NEGR1 (N,C)	Neuronal growth regulator 1: May be a trans-neuronal growth-promoting factor in regenerative axons. BMI locus	
4	rs7514705	1-74779308	TNNI3K (N), TYW3 (E)	i) TNNI3 interacting kinase: MAP kinase kinase kinase. BMI locus. ii) Probable S-adenosyl-L-methionine-dependent methyltransferase that acts as a component of the wybutosine biosynthesis pathway	
5	rs11165924	1-98148036	DPYD (N)	Dihydropyrimidine dehydrogenase: Involved in uracil and thymidine catabolism.	
6	rs11578152	1-102349609	OLFM3 (N)	Olfactomedin 3: Possible role in brain and eye development, including cell migration and axon growth.	
7	rs466639	1-163661506	RXRG (N,C)	Retinoid X receptor, gamma: Nuclear hormone receptor; mediates the anti-proliferative effects of retinoic acid, and forms dimers with the retinoic acid, thyroid hormone and vitamin D receptors.	Y
8	rs543874	1-176156103	SEC16B (N)	SEC16 homolog B (<i>S. cerevisiae</i>): Organization of transitional endoplasmic reticulum sites and protein export. BMI locus	
9	rs6427782	1-198064962	NR5A2 (N,C)	Nuclear receptor subfamily 5, group A, member 2: Expressed in ovarian granulosa cells, role in steroidogenesis and ovulation. Interacts with the regulator of reproductive function NR5A1 (Steroidogenic factor 1)	Y
10	rs951366	1-203951975	NUCKS1 (N,E), RAB7L1 (E)	i) Nuclear casein kinase and cyclin-dependent kinase substrate 1: Encodes a nuclear protein which is phosphorylated by Cdk1 during mitosis. ii) RAB7, member RAS oncogene family-like 1: Unknown function	
11	rs2947411	2-604168	TMEM18 (N,C)	Transmembrane protein 18: Transcription repressor. Enhances the migration of neural stem and precursor cells. BMI locus	
12	rs6747380	2-56441253	CCDC85A (N)	Coiled-coil domain containing 85A: Unknown function	
13	rs268067	2-59734549	BCL11A (N[~800kb])	B-Cell CLL/Lymphoma 11A (Zinc Finger Protein): Functions as a myeloid and B-cell proto-oncogene	
14	rs6758290	2-105231258	GPR45 (N)	G protein-coupled receptor 45: Possible role in central nervous system signalling	
15	rs12472911	2-141944979	LRP1B (N)	Low density lipoprotein receptor-related protein 1B: Member of the low density lipoprotein (LDL) receptor gene family	
16	rs17236969	2-156460705	NR4A2 (N,C)	Nuclear receptor subfamily 4, group A, member 2: Transcription factor essential for differentiation of dopaminergic neurons in substantia nigra. Mutations in Parkinson's disease	
16	rs4369815	2-156835210	NR4A2 (N,C)	(as above)	
17a	rs1400974	2-199346935	SATB2 (N)	SATB homeobox 2: DNA binding protein that specifically binds nuclear matrix attachment regions. Mutations in cleft palate and mental retardation.	
17b	rs17233066	2-199352283	SATB2 (N)	(as above)	
17c	rs17266097	2-199983454	SATB2 (N)	(as above)	
18	rs6770162	3-24686017	THRHB (N,C)	Thyroid hormone receptor, beta: Nuclear hormone receptor for triiodothyronine. Mutations in thyroid hormone resistance	Y
19	rs7647973	3-49485935	WDR6 (E,C), UBA7 (C)	i) WD Repeat Domain 6: Enhances serine/threonine kinase 11-induced cell growth suppression; negative regulator of amino acid starvation-induced autophagy. ii) Ubiquitin-Like Modifier Activating Enzyme 7: A RAR retinoic acid target that activates ubiquitin (as above)	
19	rs6762477	3-50068213	WDR6 (E,C), UBA7 (C)	(as above)	
20	rs7642134	3-86999572	POU1F1 (PIT1) (C)	POU class 1 homeobox 1: Regulates pituitary development. Mutations in combined pituitary hormone deficiency	Y
21	rs9849248	3-88323964	ZNF654 (N,E,F), HTR1F (C)	i) Zinc finger protein 654: May be involved in transcriptional regulation. ii) 5-Hydroxytryptamine (Serotonin) Receptor 1F, G Protein-Coupled: Unknown physiological role	
22	rs11715566	3-119045126	IGSF11 (N[~1Mb])	Immunoglobulin Superfamily Member 11: Cell adhesion molecule. Stimulates cell growth	
23	rs2687729	3-129377916	EEFSEC (N,E)	Eukaryotic elongation factor, selenocysteine-tRNA-specific: Translation factor necessary for the incorporation of selenocysteine into proteins	
24	rs2600959	3-134098154	ACAD11 (E)	Acyl-CoA dehydrogenase family, member 11: Exhibits maximal activity towards saturated C22-Co-enzyme A	
25	rs13067731	3-138472681	IL20RB (N)	Interleukin 20 Receptor Beta: Forms receptor for interleukin-19, 20 and 24.	
26	rs900400	3-158281469	LEKR1 (N,E), CCNL1 (C)	i) Leucine, glutamate and lysine rich 1: Unknown function. ii) Cyclin L1: Transcriptional regulator, role in pre-mRNAs splicing. Birth weight locus.	
27	rs939317	3-185528493	EIF4G1 (N)	Eukaryotic translation initiation factor 4 gamma, 1: Regulates protein synthesis. Mutations in Parkinson's disease	
28	rs16860328	3-187118379	TRA2B (N), IGF2BP2 (C)	i) Transformer 2 beta homolog (Drosophila): Regulates mRNA processing, splicing and gene expression. ii) Insulin-like growth factor 2 mRNA binding protein 2: May regulate translation of target mRNAs.	
29	rs1038903	4-28361152	PCDH7 (N[~2Mb])	Protocadherin 7: An integral membrane protein thought to function in cell-cell recognition and adhesion	
30	rs10938397	4-44877284	GNPDA2 (N)	Glucosamine-6-phosphate deaminase 2: Catalyzes the conversion of D-glucosamine-6-phosphate to D-fructose-6-phosphate. BMI locus	
31	rs13135934	4-95426711	SMARDCA1 (N,E,F)	SWI/SNF-Related, Matrix-Associated Actin-Dependent Regulator Of Chromatin, Subfamily A, Containing DEAD/H Box 1: Encodes a helicase protein, with role in chromatin remodelling. Mutations in adermatoglyphia	
32	rs3733631	4-104860552	TACR3 (N,C)	Tachykinin receptor 3: Receptor for neurokinin 3. Mutations in hypogonadotropic hypogonadism	Y
33	rs1532331	5-43152587	ZNF131 (N,E,C), GHR (C)	i) Zinc finger protein 131: Inhibits estrogen receptor-alpha signaling. ii) Growth hormone receptor. Mutations in Laron syndrome and GH insensitivity.	Y
34	rs17086188	5-95871610	PCSK1 (N,C)	Proprotein convertase type 1: Prohormone convertase. Mutations in severe obesity with hormone deficiencies.	Y
35	rs251130	5-110887696	STARD4 (N,E,C)	StAR-related lipid transfer domain containing 4: Regulates intra-cellular cholesterol trafficking	Y

Supplementary Table 8 (continued) | Candidate genes at or near menarche loci.

Locus	SNP	Location	Consensus Gene	Gene name: function	Role in Hormone Function
36	rs13179411	5-133928412	PHF15 (N), TCF7 (E)	PHD zinc finger protein transcription factor: Component of the HBO1 complex which has a histone H4-specific acetyltransferase activity. ii) Transcription factor 7 (T-cell specific, HMG-box): Feedback repressor of TCF7L2. Regulates self-renewal of hematopoietic stem cells.	
37	rs17171818	5-137752902	KDM3B (N,C), BRD8 (C)	Lysine (K)-specific demethylase 3B: Histone H3K9 demethylase. ii) Bromodomain containing 8: Coactivator of hormone-activated nuclear receptors, including the thyroid hormone receptor.	Y
38	rs7701886	5-153527602	GALNT10 (N)	UDP-N-acetyl-alpha-D-galactosamine:polypeptide N-acetylgalactosaminyltransferase 10: Involved in synthesis of oligosaccharides. BMI locus in African Americans.	
39	rs9647570	5-167302841	TENM2 (N,C)	Teneurin transmembrane protein 2: Expressed in developing brain, and is induced by the Kallmann syndrome gene FGFR8 product.	Y
40	rs6555855	5-168682315	SLT3 (N)	Slit homolog 3: Involved in cell migration. Height locus (but not in LD with the height SNP [rs4282339])	
41	rs16896742	6-30030719	HLA-A (N)	Major histocompatibility complex, class I, A: Central role in immune regulation	
42	rs2479724	6-41998960	BYSL (N,E), FRS3 (C)	i) Bystin-Like: Possible role in trophoblast-dependent regulation of cell adhesion during embryo implantation. ii) Fibroblast growth factor receptor substrate 3: Involved in signalling of the Kallmann syndrome gene FGFR1 product.	Y
43	rs988913	6-54864267	FAM83B (N), HCRTR2 (C)	i) Family with sequence similarity 83, member A: Unknown function. ii) Hypocretin (orexin) receptor 2: G-protein coupled receptor involved in the regulation of feeding behavior	
44	rs9475752	6-56888700	DST (N), BEND6 (E)	i) Dystonin: Anchors neural intermediate filaments to the actin cytoskeleton. DST-deficient mice have sensory ataxia. ii) BEN domain containing 6: Unknown function.	
45	rs9447700	6-77224806	IMPG1 (N)	Interphotoreceptor matrix proteoglycan 1: A neuronal proteoglycan	
46a	rs9321659	6-100222813	SIM1 (C), MCHR2 (C)	i) Single-minded homolog 1 (Drosophila): Peak expression during neurogenesis in Drosophila. Mutations in severe obesity. ii) Melanin concentrating hormone receptor 2: Orphan G protein-coupled receptor, high affinity binding to the neuropeptide melanin-concentrating hormone (MCH), which regulates energy homeostasis and mood via MCHR1.	Y
46b	rs4840086	6-100315159	SIM1 (C), MCHR2 (C)	(as above)	
46c	rs13196561	6-100866891	SIM1 (N,C), MCHR2 (C)	(as above)	
46d	rs239198	6-101240798	SIM1 (C), ASC3 (N,E,F)	i) Single-minded homolog 1 (Drosophila): Peak expression during neurogenesis in Drosophila. Mutations in severe obesity. ii) Activating signal cointegrator 1 complex subunit 3: DNA helicase, unwinds DNA to allow ALKBH3-mediated DNA repair	
47a	rs4946632	6-105207901	LIN28B (C)	Lin-28 homolog B (<i>C. elegans</i>): Repression of let-7 microRNAs; cell stemness factor	
47b	rs2153127	6-105455237	LIN28B (E,C)	(as above)	
47c	rs7759938	6-105485647	LIN28B (N,C)	(as above)	
48	rs4895808	6-126823127	CENPW (N,E), NCOA7 (C)	i) Centromere protein W: RNA-associated nuclear matrix protein. ii) Nuclear receptor co-activator 7: Enhances transcriptional activity of ESR1, THR, PPARG, RARA and other nuclear receptors. T1D locus.	Y
49	rs6938574	6-128432673	PTPRK (N)	Protein tyrosine phosphatase, receptor type, K: Negatively regulates the transactivating function of beta-catenin.	
50	rs6933660	6-151845447	ESR1 (C)	Estrogen receptor 1: Essential for sexual development and reproductive function. Locus for Breast cancer and Bone mineral density	Y
51	rs1079866	7-41436618	INHBA (N,C)	Inhibin, beta A: Inhibitor of pituitary FSH secretion	
52	rs6964833	7-73739845	GTF2I (N,C)	General transcription factor III: Multifunctional phosphoprotein with roles in transcription and signal transduction. Deleted in the early puberty-associated Williams-Beuren syndrome.	
53	rs11767400	7-121947978	CADPS2 (N)	Calcium-dependent secretion activator 2: Deletions and impairment lead to autism by reducing axonal BDNF release.	
54a	rs2688325	8-3754618	CSMD1 (N)	CUB and Sushi multiple domains 1:	
54b	rs7828501	8-4547489	CSMD1 (N)	(as above)	
54c	rs7463166	8-4821198	CSMD1 (N)	(as above)	
55	rs16918254	8-53931766	NPBWR1 (N,C)	Neuropeptides B/W receptor 1: G protein-coupled receptor for neuropeptides B and W, which are implicated in feeding behavior, energy homeostasis, neuroendocrine function and stress response.	Y
56	rs7821178	8-78256392	PEX2 (N)	Peroxisomal biogenesis factor 2: Mutations result in Zellweger syndrome and infantile Refsum disease. Adult height locus.	
57	rs1469039	8-140720961	KCNK9 (N)	Potassium channel, subfamily K: Imprinted in fetal brain, preferential maternal expression. Encodes a two-pore domain potassium channel that regulates the resting membrane potential, firing frequency of neurons and aldosterone secretion.	
58	rs4875053	8-144944399	SCRIB (N), PARP10 (E)	i) Scribbled planar cell polarity protein: Mutations in the severe neural tube defect, craniorachischisis. ii) Poly (ADP-ribose) polymerase family, member 10: Regulates gene transcription by altering chromatin organization, possible role in cancer, diabetes, and CVD.	
59a	rs7037266	9-6932940	KDM4C (N)	Lysine (K)-specific demethylase 4C: Histone demethylase JMD2C	
59b	rs913588	9-7164673	KDM4C (N,F,C)	(as above)	
60	rs7865468	9-10264080	PTPRD (N)	Protein tyrosine phosphatase, receptor type, D: May promote neurite growth and regulate neuronal axon guidance	
61	rs7853970	9-85905386	RMI1 (N), NTRK2 (C)	i) RecQL mediated genome instability 1, homolog (<i>S. cerevisiae</i>): Component of the RMI complex, limits DNA crossover formation. ii) Neurotrophic tyrosine kinase, receptor, type 2: Receptor for BDNF. Mutations associated with obesity and mood disorders	
62a	rs10816359	9-107797491	TMEM38B (N)	Transmembrane protein 38B: Intracellular monovalent cation channel. Regulates intracellular calcium release. Mutations in osteogenesis imperfecta.	
62b	rs10453225	9-107960041	TMEM38B (N)	(as above)	
62c	rs10739221	9-108100651	TMEM38B (N)	(as above)	
63	rs11792861	9-110849116	TMEM245 (N,E)	Transmembrane protein 245: Unknown function	
64	rs10980854	9-113090178	ZNF483 / OR2K2 (N)	i) Zinc finger protein 483: May be involved in transcriptional regulation. ii) Olfactory receptor, family 2, subfamily K, member 2: G-protein-coupled receptor which triggers the perception of smell	
64	rs10980921	9-113319733	ZNF483 / OR2K2 (N)	(as above)	
65	rs1874984	10-1721871	ADARB2 (N)	Adenosine deaminase B2: Regulatory role in RNA editing. Candidate gene for longevity in humans and in <i>C. elegans</i> .	

Supplementary Table 8 (continued) | Candidate genes at or near menarche loci.

Locus	SNP	Location	Consensus Gene	Gene name: function	Role in Hormone Function
66	rs12571664	10-121698919	SEC23IP (N,E)	SEC23 interacting protein: Facilitates endoplasmic reticulum export. Implicated in Waardenburg syndrome	
67	rs1915146	10-126836204	CTBP2 (N,C)	C-terminal binding protein 2: Transcriptional repressor. Expression associated with decreased PTEN expression and PI3-kinase pathway activation. Coactivator of RAR/RXR signalling	
68	rs7104764	11-219977	SIRT3 (N,E,C)	Sirtuin 3: Unknown function in humans. Induced by caloric restriction, considered to be anti-ageing.	
69	rs4929947	11-8596570	TRIM66 (N,E,F)	Tripartite motif containing 66: May function as a transcription repressor. BMI locus	
70	rs11022756	11-13272015	ARNTL (N), PTH (C)	Aryl hydrocarbon receptor nuclear translocator-like: Forms a heterodimer with CLOCK to activate circadian rhythm-associated genes. Locus for PAI-1. ii) Parathyroid hormone: Regulates calcium metabolism.	Y
71	rs7103411	11-27656701	BDNF (N,C), LGR4 (C)	Brain-derived neurotrophic factor: Nerve growth factor. Regulation of stress response and mood. BMI locus. ii) Leucine-rich repeat containing G protein-coupled receptor 4: key regulator of stem cell differentiation via SOX2. Mutations in late menarche.	
72	rs16918636	11-29080758	FSHB (N[~1mb],C)	Follicle stimulating hormone, beta polypeptide: Beta subunit of follicle-stimulating hormone.	Y
73	rs4756059	11-46107195	PHF21A (N)	PHD finger protein 21A: Component of a BRAF35/histone deacetylase complex that represses neuron-specific genes	
74	rs2063730	11-77726172	GAB2 (N), THRSP (C)	i) GRB2-associated binding protein 2: Encodes the principal activator of PI3-kinase. ii) Thyroid hormone responsive: Biosynthesis of triglycerides with medium-length fatty acid chains. May modulate the activity of THRB.	Y
75	rs10895140	11-100941931	TRPC6 (N), PGR (C)	Transient receptor potential cation channel, subfamily C, member 6: PI3/PTEN pathway and calcium signalling, activated by diacylglycerol. Mutations in focal segmental glomerulosclerosis 2. ii) Progesterone receptor: Mediates the effects of the reproductive hormone progesterone.	Y
76	rs11215400	11-114557845	CADM1 (N)	Cell adhesion molecule 1: Possible driver of synapse assembly neuronal migration, axon growth, and pathfinding.	
77	rs1461503	11-122350285	BSX (N,C)	Brain specific homeobox: DNA binding protein and transcriptional activator. Expressed specifically in mouse pineal gland, telencephalic septum, hypothalamic pre-mammillary body and arcuate nucleus. Is necessary for postnatal growth.	
78	rs7955374	12-46166416	VDR (C)	Vitamin D (1,25-dihydroxyvitamin D3) receptor: Nuclear hormone receptor for vitamin D3	Y
79	rs7138803	12-48533735	BCDIN3D (N)	BCDIN3 domain containing: RNA methyltransferase, regulates microRNA-145 processing. BMI locus	
80	rs6563739	13-39137785	COG6 (N,E)	Component of oligomeric golgi complex 6: Maintains normal structure and activity of the Golgi apparatus	
81	rs1324913	13-73533589	KLF12 (N)	Kruppel-like factor 12: Locus for phospho- and sphingolipids; overexpression represses secretion of prolactin and IGFBP1	Y
82	rs9560113	13-110981349	TEX29 (N)	Testis expressed 29: Unknown function.	
83	rs1254337	14-59990278	SIX6 (N)	SIX homeobox 6: Homeobox protein involved in eye development. Mutations in isolated microphthalmia with cataract type 2.	
84	rs1958560	14-65106548	FUT8 (N,E)	Fucosyltransferase 8: Encodes alpha-1,6-Fucosyltransferase, involved in the biosynthesis of asparagine-linked glycoprotein oligosaccharides.	
85a	rs10144321	14-99952158	DLK1 (C),	Delta-like 1 homolog (Drosophila): Epidermal growth factor involved in cell differentiation. Imprinted fetal growth gene with preferential paternal expression. Maternal UPD14 is associated with growth retardation and advanced puberty	
85b	rs7141210	14-100252223	DLK1 (N,E,C)	(as above)	
86	rs12148769	15-21703187	MKRN3 (C), MAGEL2 (C)	i) Makorin ring finger protein 3: Deletions in precocious puberty. ii) Melanoma Antigen Family (MAGE)-like 2: patients with truncating mutations on the paternal allele have clinical and behavioural features of Prader Willi syndrome. NDN, MKRN3 and MAGEL2 are imprinted genes with paternal specific expression, in the Prader-Willi syndrome deleted region.	Y
87	rs3743266	15-58568805	RORA (N,C)	RAR-related orphan receptor A: Nuclear hormone receptor; unknown function.	
88	rs8032675	15-65746518	MAP2K5 (N)	Mitogen-activated protein kinase kinase 5: Activates MAPK7/ERK5, involved in growth factor stimulated cell proliferation. BMI locus	
89	rs12915845	15-86843471	DET1 (N,E)	De-Etiolated Homolog 1 (Arabidopsis): Component of the E3 ubiquitin ligase DCX DET1-COP1 complex, required for ubiquitination and degradation of target proteins.	
90	rs246185	16-14302933	MKL2 (N)	MKL/myocardin-like 2: Transcriptional coactivator of serum response factor. Required for skeletal myogenic differentiation.	
91	rs12446632	16-19842890	GPRC5B (N,C)	G protein-coupled receptor, family C, group 5, member B: May mediate the cellular effects of retinoic acid. BMI locus	
92	rs1129700	16-29825535	KCTD13 (N), TBX6 (E,C)	Potassium channel tetramerization domain containing 13: Responsible for the micro/macrocerebral phenotype of 16p11.2. 16p11.2 also has a mirrored obese phenotype due to SH2B1 (but is 1 Mb away).	
93	rs8050136	16-52373776	FTO (N,C)	Fat mass and obesity associated: 2-oxoglutarate-dependent oxygenase. BMI locus	
94a	rs1364063	16-68146073	COG4 (C), NFAT5 (N)	i) Component of oligomeric golgi complex 4: Role in Golgi apparatus function. Mutations may cause glycosylation defect type Iij. ii) Nuclear factor of activated T-cells 5, tonicity-responsive: Regulates gene expression induced by osmotic stress.	
94b	rs929843	16-68603249	COG4 (C), WWP2 (N)	i) Component of oligomeric golgi complex 4: Role in Golgi apparatus function. Mutations may cause glycosylation defect type Iij. ii) WW domain containing E3 ubiquitin protein ligase 2: Ubiquitin protein ligase for PTEN, promotes degradation of pluripotency factor OCT4 in human embryonic stem cells.	
95	rs7215990	17-5975555	WSCD1 (N,E), ALOX15B (E)	i) WSC Domain Containing 1: Unknown function. ii) Arachidonate 15-Lipoxygenase: Metabolises arachidonic acid.	

Supplementary Table 8 (continued) | Candidate genes at or near menarche loci.

Locus	SNP	Location	Consensus Gene	Gene name: function	Role in Hormone Function
96	rs9635759	17-46968784	CA10 (N)	Carbonic anhydrase X: A catalytic member of the alpha-carbonic anhydrase subgroup. May play a role in brain development.	
97	rs244293	17-50585721	STXBP4 (N,E)	Syntaxin binding protein 4: Insulin-regulated syntaxin 4-binding protein directly involved in the control of glucose transport and GLUT4 vesicle translocation.	
98	rs12607903	18-3807134	DLGAP1 (N)	Discs, large (Drosophila) homolog-associated protein 1: Part of the postsynaptic scaffold in neuronal cells.	
99	rs2137289	18-43006123	SKOR2 (N)	SKI family transcriptional corepressor: Acts as a TGF-beta antagonist in the nervous system.	
100	rs652260	19-7806562	EVI5L (N), RETN (C)	i) Ecotropic viral integration site 5-like: Functions as a GTPase-activating protein with broad specificity. ii) Resistin: Suppresses insulin-mediated glucose uptake by adipocytes.	Y
101	rs889122	19-9856867	OLFM2 (N), RDH8 (C)	i) Olfactomedin 2: Neuronal olfactomedin related ER localized protein. ii) Retinol dehydrogenase 8: Visual cycle enzyme.	
102	rs10423674	19-18678903	CRTC1 (N,C)	CREB regulated transcription coactivator 1: Transcriptional coactivator for CREB1. Murine deletions cause hyperphagia and delayed puberty.	Y
103	rs852069	20-17070593	PCSK2 (N,C)	Proprotein convertase type 2: Prohormone convertase.	Y
104	rs2836950	21-39526299	BRWD1 (N,C)	Bromodomain and WD repeat domain containing 1: Involved in a variety of cellular processes including cell cycle progression, signal transduction, apoptosis, and gene regulation. In the Down syndrome critical region-2. Murine deletions cause impaired oocyte maturation.	Y
105	rs13053505	22-37575564	NPTXR (N,E), CBX7 (C)	i) Neuronal pentraxin receptor: CSF marker of degeneration. ii) Chromobox Homolog 7: Part of the Polycomb group (PcG) of transcriptional silencers that repress the onset of reproductive maturity via Kiss1. Pubertal onset in mice is triggered by DNA methylation of the PcG complex, including at the Cbx7 promotor region, accompanied by decreased hypothalamic Cbx7 expression.	
106	rs6009583	22-48063650	C22orf34 (N)	Chromosome 22 open reading frame 34: Unknown function.	

Gene refers to the consensus gene(s) reported at that locus mapped using 4 approaches: (N) Nearest, (C) Candidate, (F) 1000 Genomes missense variant in high LD ($r^2 > 0.8$), (E) gene expression linked by eQTL.

Supplementary Table 9 (continued) | Association of menarche signals with BMI and height in the GIANT consortium.

Locus	SNP	Location	Consensus Gene	Raising Allele	Menarche Other Allele	Association of menarche raising allele with BMI			Association of menarche raising allele with height		
						N	Direction	p value	N	Direction	p value
61	rs7853970	9-85905386	RMI1 (N), NTRK2 (C)	t	c	123841	-	0.076	132021	+	0.167
62a	rs10816359	9-107797491	TMEM38B (N)	t	g	123861	-	0.068	132041	+	1.000
62b	rs10453225	9-107960041	TMEM38B (N)	g	t	123863	-	0.013	133832	+	3.022E-05
62c	rs10739221	9-108100651	TMEM38B (N)	c	t	123863	-	0.004	133818	+	0.009
63	rs11792861	9-110849116	TMEM245 (N,E)	a	c	123864	+	0.023	133821	+	0.007
64	rs10980854	9-113090178	ZNF483 / OR2K2 (N)	a	g	123300	-	0.077	133251	+	0.176
64	rs10980921	9-113319733	ZNF483 / OR2K2 (N)	c	t	123805	-	0.477	133779	+	0.001
65	rs1874984	10-1721871	ADARB2 (N)	c	g	123841	-	0.658	132021	+	0.630
66	rs12571664	10-121698919	SEC23IP (N,E)	t	c	123864	-	0.636	133753	+	0.187
67	rs1915146	10-126836204	CTBP2 (N,C)	g	a	123836	-	0.874	133768	+	0.004
68	rs7104764	11-219977	SIRT3 (N,E,C)	g	a	123865	-	0.219	133845	+	0.015
69	rs4929947	11-8596570	TRIM66 (N,E,F)	g	c	123846	-	2.040E-06	133818	+	0.344
70	rs11022756	11-13272015	ARNTL (N), PTH (C)	a	c	123858	-	0.014	133837	+	0.007
71	rs7103411	11-27656701	BDNF (N,C), LGR4 (C)	c	t	123864	-	9.150E-13	133828	+	0.597
72	rs16918636	11-29080758	FSHB (N[~1mb],C)	t	c	123859	-	0.001	133787	+	0.382
73	rs4756059	11-46107195	PHF21A (N)	t	c	123818	+	0.506	133808	+	0.022
74	rs2063730	11-77726172	GAB2 (N), THRSR (C)	c	a	123865	-	0.003	132045	+	0.841
75	rs10895140	11-100941931	TRPC6 (N), PGR (C)	g	a	123864	-	0.624	133798	+	0.576
76	rs11215400	11-114557845	CADM1 (N)	c	a	116688	-	0.005	126566	+	0.746
77	rs1461503	11-122350285	BSX (N,C)	c	a	123863	-	0.614	133805	+	4.144E-04
78	rs7955374	12-46166416	VDR (C)	t	c	123118	+	0.678	131291	+	0.011
79	rs7138803	12-48533735	BCDIN3D (N)	g	a	123799	-	3.960E-11	133757	-	0.176
80	rs6563739	13-39137785	COG6 (N,E)	g	t	123864	-	0.050	133855	+	0.078
81	rs1324913	13-73533589	KLF12 (N)	g	t	123837	-	0.944	133756	+	0.747
82	rs9560113	13-110981349	TEX29 (N)	g	a	123858	-	0.098	133816	-	0.845
83	rs1254337	14-59990278	SIX6 (N)	t	a	123864	+	0.896	133845	+	5.050E-10
84	rs1958560	14-65106548	FUT8 (N,E)	a	g	123864	-	0.087	133676	+	0.209
85a	rs10144321	14-99952158	DLK1 (C),	a	g	123864	-	0.389	133766	+	0.039
85b	rs7141210	14-100252223	DLK1 (N,E,C)	t	c	123858	-	0.981	133802	+	0.115
86	rs12148769	15-21703187	MKRN3 (C), MAGEL2 (C)	g	a	123855	+	0.754	133745	+	0.073
87	rs3743266	15-58568805	RORA (N,C)	t	c	123689	-	0.908	133613	+	0.042
88	rs8032675	15-65746518	MAP2K5 (N)	t	c	123730	-	4.120E-07	133689	+	0.757
89	rs12915845	15-86843471	DET1 (N,E)	c	t	123852	+	0.913	133839	+	0.061
90	rs246185	16-14302933	MKL2 (N)	c	t	123849	+	0.321	132029	+	7.510E-06
91	rs12446632	16-19842890	GPRC5B (N,C)	a	g	123853	-	4.610E-11	133795	+	0.072
92	rs1129700	16-29825535	KCTD13 (N), TBX6 (E,C)	t	c	119582	-	0.206	127211	-	0.331
93	rs8050136	16-52373776	FTO (N,C)	c	a	123729	-	1.040E-59	133662	+	0.055
94a	rs1364063	16-68146073	COG4 (C), NFAT5 (N)	c	t	123864	-	0.034	133850	+	5.913E-05
94b	rs929843	16-68603249	COG4 (C), WWF2 (N)	a	c	123859	-	0.227	132039	+	0.001
95	rs7215990	17-5975555	WSCD1 (N,E), ALOX15B (E)	g	a	123848	+	0.974	133836	+	0.502
96	rs9635759	17-46968784	CA10 (N)	a	g	123843	-	0.122	132023	+	0.412
97	rs244293	17-50585721	STXBP4 (N,E)	g	a	123863	-	0.009	133808	+	0.300
98	rs12607903	18-3807134	DLGAP1 (N)	c	t	123855	-	0.728	132035	+	0.119
99	rs2137289	18-43006123	SKOR2 (N)	a	g	123862	+	0.894	133786	+	0.061
100	rs652260	19-7806562	EVI5L (N), RETN (C)	t	c	123795	-	0.978	131952	+	0.036
101	rs889122	19-9856867	OLFM2 (N), RDH8 (C)	g	t	123783	-	0.508	133721	+	0.765
102	rs10423674	19-18678903	CRTC1 (N,C)	a	c	123825	-	0.016	131993	+	0.489
103	rs852069	20-17070593	PCSK2 (N,C)	g	a	123795	-	0.879	131973	-	0.621
104	rs2836950	21-39526299	BRWD1 (N,C)	c	g	123407	+	0.823	131459	+	0.135
105	rs13053505	22-37575564	NPTXR (N,E), CBX7 (C)	g	t	123900	-	0.124	132035	+	0.016
106	rs6009583	22-48063650	C22orf34 (N)	c	t	122954	+	0.105	132868	+	0.985

Supplementary Note

1. Acknowledged consortia members and affiliations

Early Growth Genetics (EGG) Consortium

Diana L. Cousminer¹, Evangelia Stergiakouli², Diane J. Berry³, Wei Ang⁴, Maria M. Groen-Blokhus^{5,6}, Antje Körner⁷, Niina Sijtonen⁸, Ioanna Ntalla^{9,10}, Marcella Marinelli^{11,12,13}, John R. B. Perry¹⁴, Johannes Kettunen^{1,15}, Rick Jansen^{16,17}, Ida Surakka^{1,15}, Nicholas J. Timpson², Susan Ring¹⁸, George McMahon¹⁸, Chris Power³, Carol Wang⁴, Mika Kähönen¹⁹, Jorma Viikari²⁰, Terho Lehtimäki²¹, Christel M. Middeldorp^{5,16,17}, Hilleke E. Hulshoff Pol²², Madlen Neef⁷, Sebastian Weise⁷, Katja Pahkala^{8,23}, Harri Niinikoski^{8,24}, Eleftheria Zeggini²⁵, Kalliope Panoutsopoulou²⁵, Mariiona Bustamante^{11,12,13}, Brenda W. J. H. Penninx^{6,16,17}, the ReproGen Consortium³³, Joanne Murabito²⁶, Maties Torrent¹³, George V. Dedoussis⁹, Wieland Kiess⁷, Dorret I. Boomsma^{5,6,17}, Craig E. Pennell⁴, Olli T. Raitakari^{8,27}, Elina Hyppönen^{3,28}, George Davey Smith², Samuli Ripatti^{1,25,29}, Mark I. McCarthy^{30,31,32}, and Elisabeth Widén^{*1}

¹Institute for Molecular Medicine, Finland (FIMM), University of Helsinki, Helsinki, Finland

²MRC Integrative Epidemiology Unit at the University of Bristol, Bristol, UK

³Centre for Paediatric Epidemiology and Biostatistics, UCL Institute of Child Health, London, UK

⁴School of Women's and Infants' Health, The University of Western Australia, Perth, WA, Australia

⁵Department of Biological Psychology, VU University, Amsterdam, the Netherlands

⁶EMGO+ Institute for Health and Care Research, VU University, Amsterdam, the Netherlands

⁷Center of Pediatric Research, Dept. of Women's & Child Health, University of Leipzig, Germany

⁸Research Centre of Applied and Preventive Cardiovascular Medicine, University of Turku, Finland

⁹Department of Nutrition and Dietetics, Harokopio University of Athens, Greece

¹⁰University of Leicester, Genetic Epidemiology Group, Department of Health Sciences, Leicester, UK

¹¹Center for Research in Environmental Epidemiology (CREAL), Barcelona, Catalonia, Spain

¹²Hospital del Mar Research Institute (IMIM), Barcelona, Catalonia, Spain

¹³Spanish consortium for Research on Epidemiology and Public Health (CIBERESP), Barcelona, Catalonia, Spain

¹⁴MRC Epidemiology Unit, Institute of Metabolic Science, Addenbrooke's Hospital, Cambridge, United Kingdom

¹⁵Department of Chronic Disease Prevention, National Institute for Health and Welfare, Helsinki, Finland

¹⁶Department of Psychiatry, VU medical center / GGZ inGeest, Amsterdam, the Netherlands

¹⁷Neuroscience Campus Amsterdam, VU University, Amsterdam, the Netherlands

¹⁸Avon Longitudinal Study of Parents and Children (ALSPAC), School of Social and Community Medicine, University of Bristol, Bristol, UK

¹⁹Department of Clinical Physiology, Tampere University Hospital and University of Tampere School of Medicine, Tampere 33521, Finland

²⁰Department of Medicine, University of Turku and Turku University Hospital, Turku 20521, Finland

²¹Department of Clinical Chemistry, Fimlab Laboratories and University of Tampere School of Medicine, Tampere 33520, Finland

²²Brain Center Rudolf Magnus, University Medical Center Utrecht, Department of Psychiatry, the Netherlands

²³Paavo Nurmi Centre, Sports & Exercise Medicine Unit, Department of Physical Activity and Health, University of Turku, Finland

²⁴Department of Pediatrics, University of Turku, Turku, Finland

²⁵Wellcome Trust Sanger Institute, Cambridge, United Kingdom

²⁶Department of Medicine, Section of General Internal Medicine, Boston University School of Medicine, Boston, MA, USA

²⁷Department of Clinical Physiology and Nuclear Medicine, Turku University Hospital, Turku, Finland

²⁸School of Population Health, University of South Australia, North Terrace, Adelaide, Australia

²⁹Hjelt Institute, University of Helsinki, Helsinki, Finland

³⁰Oxford Centre for Diabetes, Endocrinology and Metabolism, University of Oxford, Churchill Hospital, Old Road, Headington, Oxford, United Kingdom

³¹Wellcome Trust Centre for Human Genetics, University of Oxford, Oxford, United Kingdom

³²Oxford NIHR Biomedical Research Centre, Churchill Hospital, Oxford, United Kingdom

LifeLines Cohort Study

Behrooz Z Alizadeh (1), Rudolf A de Boer (2), H Marike Boezen (1), Marcel Bruinenberg (3), Lude Franke (4), Pim van der Harst (2), Hans L Hillege (1,2), Melanie M van der Klaauw (5), Gerjan Navis (6), Johan Ormel (7), Dirkje S Postma (8), Judith GM Rosmalen (7), Joris P Slaets (9), Harold Snieder (1), Ronald P Stolk (1), Bruce HR Wolffensbuttel (5), Cisca Wijmenga (4)

- (1) Department of Epidemiology, University of Groningen, University Medical Center Groningen, The Netherlands
- (2) Department of Cardiology, University of Groningen, University Medical Center Groningen, The Netherlands
- (3) LifeLines Cohort Study, University of Groningen, University Medical Center Groningen, The Netherlands
- (4) Department of Genetics, University of Groningen, University Medical Center Groningen, The Netherlands
- (5) Department of Endocrinology, University of Groningen, University Medical Center Groningen, The Netherlands
- (6) Department of Internal Medicine, Division of Nephrology, University of Groningen, University Medical Center Groningen, The Netherlands
- (7) Interdisciplinary Center of Psychopathology of Emotion Regulation (ICPE), Department of Psychiatry, University of Groningen, University Medical Center Groningen, The Netherlands
- (8) Department of Pulmonology, University of Groningen, University Medical Center Groningen, The Netherlands
- (9) University Center for Geriatric Medicine, University of Groningen, University Medical Center Groningen, The Netherlands

InterAct Consortium

Nita G Forouhi(1), Nicola D Kerrison(1), Claudia Langenberg(1), Robert A Scott(1), Stephen J Sharp(1), Matt Sims(1), Inês Barroso(2,3), Panos Deloukas(2), Mark I McCarthy(4,5,6), Larraitz Arriola(7,8,9), Beverley Balkau(10,11), Aurelio Barricarte(12,9), Heiner Boeing(13), Paul W Franks(14,15), Carlos Gonzalez(16), Sara Grioni(17), Rudolf Kaaks(18), Timothy J Key(19), Carmen Navarro(20,9,21), Peter M Nilsson(14), Kim Overvad(22,23), Domenico Palli(24), Salvatore Panico(25), J. Ramón Quirós(26), Olov Rolandsson(15), Carlotta Sacerdote(27,28), María-José Sánchez(29,9,30), Nadia Slimani(31), Anne Tjønneland(32), Rosario Tumino(33,34), Daphne L van der A(35), Yvonne T van der Schouw(36), Elio Riboli(37), Nicholas J Wareham(1)

- (1) MRC Epidemiology Unit, University of Cambridge, Cambridge, United Kingdom, (2) The Wellcome Trust Sanger Institute, Cambridge, United Kingdom, (3) University of Cambridge Metabolic Research Laboratories, Cambridge, UK, (4) Oxford Centre for Diabetes, Endocrinology and Metabolism (OCDEM), University of Oxford, UK, (5) Wellcome Trust Centre for Human Genetics, University of Oxford, Oxford, UK, (6) Oxford NIHR Biomedical Research Centre, Oxford, UK, (7) Public Health Division of Gipuzkoa, San Sebastian, Spain, (8) Instituto BIO-Donostia, Basque Government, San Sebastian, Spain, (9) CIBER Epidemiología y Salud Pública (CIBERESP), Spain, (10) Inserm, CESP, U1018, Villejuif, France, (11) Univ Paris-Sud, UMRS 1018, Villejuif, France, (12) Navarre Public Health Institute (ISPN), Pamplona, Spain, (13) German Institute of Human Nutrition Potsdam-Rehbruecke, Germany, (14) Lund University, Malmö, Sweden, (15) Umeå University, Umeå, Sweden, (16) Catalan Institute of Oncology (ICO), Barcelona, Spain, (17) Epidemiology and Prevention Unit, Milan, Italy, (18) German Cancer Research Centre (DKFZ), Heidelberg, Germany, (19) University of Oxford, United Kingdom, (20) Department of Epidemiology, Murcia Regional Health Council, Murcia, Spain, (21) Unit of Preventive Medicine and Public Health, School of Medicine, University of Murcia, Spain, (22) Department of Public Health, Section for Epidemiology, Aarhus University, Aarhus, Denmark, (23) Aalborg University Hospital, Aalborg, Denmark, (24) Cancer Research and Prevention Institute (ISPO), Florence, Italy, (25) Dipartimento di Medicina Clinica e Chirurgia, Federico II University, Naples, Italy, (26) Public Health Directorate, Asturias, Spain, (27) Unit of Cancer Epidemiology, Città' della Salute e della Scienza Hospital-University of Turin and Center for Cancer Prevention (CPO), Torino, Italy, (28) Human Genetics Foundation (HuGeF), Torino, Italy, (29) Andalusian School of Public Health, Granada, Spain, (30) Instituto de Investigación Biosanitaria de Granada (Granada.ibs), Granada (Spain), (31) International Agency for Research on Cancer, Lyon, France, (32) Danish Cancer Society Research Center, Copenhagen, Denmark, (33) ASP Ragusa, Italy, (34) Aire Onlus, Ragusa, Italy, (35) National Institute for Public Health and the Environment (RIVM), Bilthoven, The Netherlands, (36) University Medical Center Utrecht, Utrecht, the Netherlands, (37) School of Public Health, Imperial College London, UK

Australian Ovarian Cancer Study (AOCS)

David D Bowtell¹, Adele C Green², Georgia Chenevix-Trench², Anna deFazio³, Dorota Gertig⁴, Penelope M Webb².

¹ Peter MacCallum Cancer Centre, St Andrews Place, East Melbourne, Victoria, Australia

² QIMR Berghofer Medical Research Institute, Brisbane, Australia

³ Westmead Institute for Cancer Research, Westmead, New South Wales, Australia

⁴ Victorian Cervical Cytology Registry, Carlton South Victoria, Australia

Gene Environment Interaction and Breast Cancer in Germany (GENICA) network Consortium members

Hiltrud Brauch, Christina Justenhoven Dr. Margarete Fischer-Bosch Institute of Clinical Pharmacology, Stuttgart, and University Tübingen, Germany

Ute Hamann Molecular Genetics of Breast Cancer, Deutsches Krebsforschungszentrum (DKFZ), Heidelberg, Germany

Yon-Dschun Ko, Christian Baisch Department of Internal Medicine, Evangelische Kliniken Bonn gGmbH, Johanniter Krankenhaus, Bonn, Germany

Hans-Peter Fischer Institute of Pathology, University of Bonn, Bonn, Germany

Beate Pesch, Sylvia Rabstein, Anne Spickenheuer Institute for Prevention and Occupational Medicine of the German Social Accident Insurance(IPA), Bochum, Germany

Volker Harth Institute and Outpatient Clinic of Occupational Medicine, Saarland University Medical Center and Saarland University Faculty of Medicine, Homburg, Germany

kConFab Consortium members

Morteza Aghmesheh Medical Oncology Department, Illawarra Cancer Care Centre, Wollongong Hospital, Wollongong, NSW 2500

David Amor Genetic Health Services Victoria, Royal Children's Hospital, Melbourne VIC 3050

Lesley Andrews Hereditary Cancer Clinic, Prince of Wales Hospital, Randwick NSW 2031

Yoland Antill Dept. Haem and Medical Oncology, Peter MacCallum Cancer Centre, St Andrews Place, East Melbourne VIC 3002

Shane Armitage Molecular Genetics Lab, Royal Brisbane and Women's Hospital

Leanne Arnold Molecular Genetics Lab, Royal Brisbane and Women's Hospital

Rosemary Balleine Department of Translational Oncology, C/- Department of Medical Oncology, Westmead Hospital, Westmead NSW 2145

Agnes Bankier PO Box 5444, HEIDELBERG WEST, C/o The Austin Hospital, 3081 Australia

Patti Bastick St George Hospital, Medical Oncology Dept, Gray Street, Kogarah NSW 2000, Australia

Jonathan Beesley Queensland Institute of Medical Research, Herston Road, Herston Qld 4002, Australia

John Beilby Pathology Centre, Queen Elizabeth Medical Centre, Nedlands WA 6009

Ian Bennett Silverton Place, 101 Wickham Terrace, Brisbane QLD 4000

Barbara Bennett Hereditary Cancer Clinic, Prince of Wales Hospital, Randwick NSW 2031

Geoffrey Berry Dept of Public Health and Community Medicine, University of Sydney, Sydney NSW 2006

Anneke Blackburn John Curtin School of Medical Research, Australian National University, P.O. Box 334, Canberra ACT 2601

Michael Bogwitz Familial Cancer Centre, The Royal Melbourne Hospital, Grattan Street, Parkville Victoria 3050, Australia

Meagan Brennan NSW Breast Cancer Institute, PO Box 143, Westmead NSW 2145

Melissa Brown Department of Biochemistry, University of Queensland, St. Lucia QLD 4072

Michael Buckley Molecular and Cytogenetics Unit, Prince of Wales Hospital, Randwick NSW 2031

- Matthew Burgess** Clinical Genetics Service, Austin Health, Victoria 3084, Australia
- Jo Burke** Royal Hobart Hospital, GPO Box 1061L, Hobart TAS 7001
- Phyllis Butow** Medical Psychology Unit, Royal Prince Alfred Hospital, Camperdown NSW 2204
- Keith Byron** Australian Genome Research Facility, Walter & Eliza Hall Medical Research Institute, Royal Melbourne Hospital, Parkville VIC 3050
- David Callen** Dame Roma Mitchell Cancer Research Laboratories, University of Adelaide/Hanson Institute, P.O. Box 14, Rundle Mall SA 5000
- Ian Campbell** Peter MacCallum Cancer Centre, St Andrew's Place, East Melbourne VIC 3002
- Deepa Chauhan** School of Psychology, Brennan McCallum (Building A18), University of Sydney 2006
- Georgia Chenevix-Trench** Queensland Institute of Medical Research, Royal Brisbane Hospital, Herston QLD 4029
- Alice Christian** Genetics Department, Central Region Genetics Service, Wellington Hospital, New Zealand
- Christine Clarke** Westmead Institute for Cancer Research, University of Sydney, Westmead Hospital, Westmead NSW 2145
- Alison Colley** Department of Clinical Genetics, Liverpool Health Service, PO Box 103, Liverpool NSW 2170
- Dick Cotton** Mutation Research Centre, St Vincent's Hospital, Victoria Parade, Fitzroy VIC 3065
- Ashley Crook** Department of Clinical Genetics, Level 3E, Royal North Shore Hospital, St Leonards NSW 2065
- James Cui** Epidemiology and Preventive Medicine, Monash University, Prahan Vic 3004, Australia
- Bronwyn Culling** Molecular and Clinical Genetics, Level 1 Building 65, Royal Prince Alfred Hospital, Camperdown NSW 2050
- Margaret Cummings** Department of Pathology, University of Queensland Medical School, Herston NSW 4006
- Sarah-Jane Dawson** Molecular Genetics Department, Cambridge University, England
- Anna deFazio** Dept. Gynaecological Oncology, Westmead Institute for Cancer Research, Westmead Hospital, Westmead NSW 2145
- Martin Delatycki** Clinical Genetics, Austin Health, Heidelberg Repatriation Hospital, PO Box 5444, Heidelberg West Vic 3081, Australia
- Rebecca Dickson** Level 2, Block 51, Royal North Shore Hospital, North Shore NSW 2408
- Joanne Dixon** Central Regional Genetic Services, Wellington Hospital, Private bag 7902, Wellington, New Zealand
- Alexander Dobrovic** Molecular Pathology, Department of Pathology, Peter MacCallum Cancer Centre, St Andrew's Place, East Melbourne VIC 3002
- Tracy Dudding** Hunter Genetics, Hunter Area Health Service, PO Box 84, Waratah, 2298 NSW
- Ted Edkins** Clinical Chemistry, Princess Margaret Hospital for Children, Box D184, Perth WA 6001
- Stacey Edwards** Department of Biochemistry and Molecular Biology, University of Queensland, St Lucia Qld 4072, Australia
- Maurice Eisenbruch** Department of Multicultural Health, University of Sydney, NSW 2052
- Gelareh Farshid** Tissue Pathology, IMVS, Adelaide SA 5000
- Susan Fawcett** Family Cancer Clinic, Monash Medical Centre, Clayton VIC 3168
- Andrew Fellows** Molecular Diagnostic Development, Pathology Department, Peter MacCallum Cancer Centre, Melbourne, East Melbourne Vic 3002
- Georgina Fenton** South West Family Cancer Clinic, Liverpool Hospital, Liverpool BC NSW 1871
- Michael Field** Royal North Shore Hospital, Level 2, Vindin House, St Leonards NSW 2065
- Frank Firgaira** GTG, 60 - 66 Hanover Street, Fitzroy, 3065
- James Flanagan** Epigenetics Unit, Department of Surgery and Oncology, Imperial College London, London W12 0NN, England
- Jean Fleming** Eskitis Institute of Cell & Molecular Therapies, School of Biomolecular and Biomedical Sciences, Griffith University, Nathan QLD 4111
- Peter Fong** Medical Oncology Department, Regional Cancer and Blood Services, Level 1 Building 7, Auckland City Hospital, 2 Park Rd. Grafton, Auckland 1023, New Zealand
- John Forbes** Surgical Oncology, University of Newcastle, Newcastle Mater Hospital, Waratah NSW 2298

- Stephen Fox** Pathology Department, Level 1, Peter MacCallum Cancer Centre, St Andrew's Place, East Melbourne Vic 3002
- Juliet French** School of Molecular and Microbial Sciences, University of Queensland, St Lucia Qld 4072
- Michael Friedlander** Department of Medical Oncology, Prince of Wales Hospital, Randwick NSW 2031
- Clara Gaff** Victorian Clinical Genetics Service, Royal Melbourne Hospital, Parkville VIC 3052
- Mac Gardner** Genetic Health Services Victoria, 10th Floor The Murdoch Institute, Royal Children's Hospital, Parkville VIC 3052
- Mike Gattas** Queensland Clinical Genetic Service, Royal Children's Hospital, Bramston Terrace, Herston QLD 4020
- Peter George** Clinical Biochemistry Unit, Canterbury Health Labs, PO Box 151, Christchurch, New Zealand
- Graham Giles** Cancer Epidemiology Centre, Anti Cancer Council of Victoria, 1 Rathdowne Street, Carlton South VIC 3052
- Grantley Gill** Department of Surgery, Royal Adelaide Hospital, Adelaide SA 5000
- Jack Goldblatt** Genetic Services Of WA, King Edward Memorial Hospital, 374 Bagot Road, Subiaco WA 6008
- Sian Greening** Illawarra Cancer Centre, Wollongong Hospital, Private Mail Bag 8808, South Coast Mail Centre, NSW 2521
- Scott Grist** Department of Haematology and Genetic Pathology, SouthPath , Flinders Medical Centre , SA
- Eric Haan** Department of Medical Genetics, Women's and Children's Hospital, North Adelaide SA 5006
- Kate Hardie** Room 430 Bldg 76, School of Chemistry and Molecular Biosciences, University of Queensland, St Lucia QLD 4072
- Marion Harris** Familial Cancer Clinic, Peter MacCallum Cancer Centre, St Andrews Place, East Melbourne VIC 3002
- Stewart Hart** Breast and Ovarian Cancer Genetics, Monash Medical Centre, 871 Centre Road, Bentleigh East VIC, 3165
- Nick Hayward** Queensland Institute for Medical Research, Royal Brisbane Hospital Post Office, Herston QLD 4029
- Sue Healey** Queensland Institute of Medical Research (QIMR), 300 Herston Road, Herston Qld Q4006
- Louise Heiniger** Medical Psychology Research Unit, The University of Sydney, Sydney NSW 2006
- John Hopper** Centre for M.E.G.A. Epidemiology, University of Melbourne, Level 1, 723 Swanston Street, Carlton VIC 3010
- Evelyn Humphrey** Royal Hobart Hospital, GPO Box 1061L, Hobart TAS 7001
- Clare Hunt** Southern Health Familial Cancer Centre, Monash Medical Centre, Special Medicine Building, 246 Clayton Rd, Clayton Victoria 3168, Australia
- Paul James** Genetic Health Sevices, Monash Medical Centre, Clayton Vic
- Mark Jenkins** Centre for M.E.G.A. Epidemiology, The University of Melbourne, 723 Swanston Street, Carlton VIC 3053
- Alison Jones** Molecular Genetics Lab, Royal Brisbane and Women's Hospital, QLD
- Rick Kefford** Medical Oncology, Westmead Hospital, Westmead NSW 2145
- Alexa Kidd** Clinical Genetics Departments, Central Regional Genetics Service, Wellington Hospital, New Zealand
- Belinda Kiely** NHMRC Clinical Trials Centre, University of Sydney, Locked Bag 77, Camperdown Sydney NSW 1450
- Judy Kirk** Familial Cancer Service, Department of Medicine, Westmead Hospital, Westmead NSW 2145
- Jessica Koehler** Hereditary Cancer Clinic, Prince of Wales Hospital, Randwick NSW 2031
- James Kollias** Breast Endocrine and Surgical Unit, Royal Adelaide Hospital, North Terrace SA 5000
- Serguei Kovalenko** Genetic Technologies Limited, 60-66 Hanover Street, Fitzroy Vic 3065
- Sunil Lakhani** UQ Centre for Clinical Research, Level 6 Building 71/918, University of Queensland, The Royal Brisbane & Women's Hospital Herston, 4029
- Amanda Leaming** Wesley Breast Clinic, Chasely Street, Auchenflower, Brisbane Qld 4066
- Jennifer Leary** Familial Cancer Laboratory, Westmead Hospital, Westmead NSW 2145

Jacqueline Lim Dept of Psychological Medicine, Royal North Shore Hospital, St Leonards NSW 2065

Geoff Lindeman Breast Cancer Laboratory, Walter and Eliza Hall Institute, PO Royal Melbourne Hospital, Parkville VIC 3050

Lara Lipton Medical Oncology and Clinical Haematology Unit, Western Hospital, Footscray VIC

Liz Lobb Medical Psychology Research Unit, Room 332, Brennan MacCallum Building (A18), The University of Sydney, Camperdown, 2006

Graham Mann Westmead Institute for Cancer Research, Westmead Millennium Institute, Westmead NSW 2145

Deborah Marsh Kolling Institute of Medical Research, Royal North Shore Hospital, St Leonards NSW 2065

Sue Anne McLachlan Department of Oncology, St Vincent's Hospital, 41 Victoria Parade, Fitzroy VIC 3065

Bettina Meiser Hereditary Cancer Clinic, Prince of Wales Hospital, Randwick NSW 2031

Cliff Meldrum Molecular Pathology Dept, 1st Floor, Peter MacCallum Cancer Centre, St Andrew's Place, East Melbourne Vic 3002

Roger Milne Centro Nacional de Investigaciones Oncologicas, C/ Melchor Fernández Almagro, 3, E-28029 Madrid, Spain

Gillian Mitchell Family Cancer Clinic, Peter MacCallum Cancer Centre, St Andrew's Place, East Melbourne VIC 3002

Beth Newman School of Public Health Road, Queensland University of Technology, Victoria Park, Kelvin Grove QLD 4059

Shona O'Connell Southern Health Familial Cancer Centre, Special Medicine Building, 246 Clayton Road, Clayton Vic 3168

Imelda O'Loughlin St Vincent's Breast Clinic, PO Box 4751, Toowoomba QLD 4350

Richard Osborne Dept of Public Health and Community Medicine, 200 Berkeley Street, Carlton VIC 3053

Nick Pachter Familial Cancer and Clinical Genetics, Royal Melbourne Hospital, Grattan Street, Parkville VIC 3050, Australia

Briony Patterson Tas Clinical Genetics Service, Royal Hobart Hospital, GPO Box 1061, Hobart Tasmania 7001, Australia

Lester Peters Radiation Oncology, Peter MacCallum Cancer Centre, St Andrew's Place, East Melbourne VIC 3002

Kelly Phillips Department of Medical Oncology, Peter MacCallum Cancer Centre, St Andrew's Place, East Melbourne VIC 3002

Melanie Price Medical Psychology, University of Sydney, Sydney, 2006

Lynne Purser The Centre for Genetics Education NSW Health, PO Box 317, St Leonards NSW 1590, Australia

Jeanne Reeve Northern Regional Genetic Service, Auckland Hospital, New Zealand

Tony Reeve Cancer Genetics Laboratory, University of Otago, PO Box 56, Dunedin, New Zealand

Robert Richards Dept of Cytogenetics and Molecular Genetics, Women and Children's Hospital, Adelaide SA 5006

Edwina Rickard Familial Cancer centre, Westmead Hospital, Westmead NSW 2145

Bridget Robinson Oncology Service, Christchurch Hospital, Private Bag 4710, Christchurch, New Zealand

Barney Rudzki Molecular Pathology Department, The University of Melbourne, Parkville Vic 3050

Mona Saleh Centre for Genetic Education, Prince of Wales Hospital, Randwick NSW 2031

Elizabeth Salisbury Anatomical Pathology, UNSW, Prince of Wales Hospital, Randwick, 2031 NSW

Joe Sambrook Peter MacCallum Cancer Centre, St Andrew's Place, East Melbourne VIC 3002

Christobel Saunders School of Surgery and Pathology, QE11 Medical Centre, M block 2nd Floor, Nedlands WA 6907

Jodi Saunus Breast Pathology, University of Queensland Centre for Clinical Research, Building 71/918 Royal Brisbane and Women's Hospital, Herston Qld 4029

Robyn Sayer Gynaecological Cancer Centre, Royal Hospital for Women, Randwick NSW 2011

Elizabeth Scott South View Clinic, Suite 13, Level 3 South Street, Kogarah NSW 2217

Rodney Scott Hunter Area Pathology Service, John Hunter Hospital, Locked Bag 1 Regional Mail Centre, NSW 2310

Clare Scott Research Department, WEHI, C/o Royal Melbourne Hospital, Parkville, 3050

Ram Seshadri Department of Haematology, Flinders Medical Centre, Bedford Park SA 5042

Adrienne Sexton Familial Cancer Centre, Royal Melbourne Hospital, Grattan Street, Parkville Vic 3050

Raghwa Sharma Dept of Tissue Pathology, Westmead Hospital, Westmead NSW 2145

Andrew Shelling Obstetrics and Gynaecology, University of Auckland, New Zealand

Peter Simpson The University of Queensland, Building 71/918, RBWH Campus, Herston Qld 4029

Melissa Southey Genetic Epidemiology Laboratory, Departemnt of Pathology, University of Melbourne, VIC 3010

Amanda Spurdle Cancer Unit, Queensland Institute of Medical Research, Herston QLD 4029

Graeme Suthers South Australian Clinical Genetics Service, Centre for Medical Genetics, Women and Children's Hospital, North Adelaide SA 5006

Pamela Sykes Molecular Pathology, Flinders Medical Centre, Flinders Drive, Bedford Park, 5042, Australia

Donna Taylor Department of Radiology, Royal Perth Hospital, Perth WA

Jessica Taylor Familial Cancer and Genetics Medicine, Royal Melbourne Hospital, 2nd Floor Grattan Street, Parkville Vic 3050, Australia

Benjamin Thierry Ian Wark Research Institute, University of South Australia, Adelaide SA 5095 SA

Ella Thompson Cancer Genetics, Research Department, 3rd level, Peter MacCallum Cancer Centre, East Melbourne, Vic 3002

Heather Thorne Research Department, Peter MacCallum Cancer Centre, St Andrew's Place, East Melbourne VIC 3002

Sharron Townshend Genetic Services of Western Australia, 3rd Floor Agnes Walsh House, 374 Bagot Rd, Subiaco WA 6008

Alison Trainer University of NSW, Prince of Wales Hospital, Barker Street, Randwick NSW 2031

Lan Tran Medical Psychology Unit, University of Sydney, NSW 2006

Kathy Tucker Heredity Cancer Clinic, Prince of Wales Hospital, Randwick NSW 2031

Janet Tyler Hereditary Cancer Clinic, Prince of Wales Hospital, Randwick NSW 2031

Jane Visvader The Walter and Eliza Hall Institute of Medical Research, Post Office Royal Melbourne Hospital, Parkville VIC 3050

Logan Walker Molecular Cancer Epidemiology Laboratory, Queensland Institute of Medical Research, P.O. Royal Brisbane Hospital, Herston, Qld 4027, Australia

Ian Walpole Genetic Services of WA, King Edward Memorial Hospital, Subiaco WA 6008, Robin Ward Department of Medical Oncology, Prince of Wales Hospital, Randwick NSW 2031

Paul Waring Department of Pathology, University of WA, 35 Stirling Highway, CRAWLEY WA 6009 Perth,

Bev Warner Cabrini Hospital, 183 Wattletree Rd, Malvern VIC 3144.

Graham Warren St Vincent's Breast Clinic, PO Box 4751, Toowoomba QLD 4350

Rachael Williams Family Cancer Clinic, St Vincent's Hospital, Darlinghurst NSW 2010

Judy Wilson Block 4, Level 5, Royal North Shore Hospital, St Leonards NSW 2065

Ingrid Winship Department of Genetics, Royal Melbourne Hospital, Parkville, 3050

Kathy Wu Familial Cancer Centre, Westmead Hospital, Darcy Street, Westmead NSW 2045

Mary Ann Young Familial Cancer Clinic, Peter MacCallum Cancer Centre, St Andrew's Place, East Melbourne VIC 3002

2. Study Acknowledgements and disclosures

Study	Sources of funding and acknowledgements
ABCFS	The ABCFS, NC-BCFR and OFBCR work was supported by the United States National Cancer Institute, National Institutes of Health (NIH) under RFA-CA-06-503 and through cooperative agreements with members of the Breast Cancer Family Registry (BCFR) and Principal Investigators, including Cancer Care Ontario (U01 CA69467), Northern California Cancer Center (U01 CA69417), University of Melbourne (U01 CA69638). Samples from the NC-BCFR were processed and distributed by the Coriell Institute for Medical Research. The content of this manuscript does not necessarily reflect the views or policies of the National Cancer Institute or any of the collaborating centers in the BCFR, nor does mention of trade names, commercial products, or organizations imply endorsement by the US Government or the BCFR. The ABCFS was also supported by the National Health and Medical Research Council of Australia, the New South Wales Cancer Council, the Victorian Health Promotion Foundation (Australia) and the Victorian Breast Cancer Research Consortium. J.L.H. is a National Health and Medical Research Council (NHMRC) Australia Fellow and a Victorian Breast Cancer Research Consortium Group Leader. M.C.S. is a NHMRC Senior Research Fellow and a Victorian Breast Cancer Research Consortium Group Leader. The authors thank Maggie Angelakos, Judi Maskiell and Gillian Dite for their support.
ABCS	The ABCS study was supported by the Dutch Cancer Society [grants NKI 2007-3839; 2009 4363]; BBMRI-NL, which is a Research Infrastructure financed by the Dutch government (NWO 184.021.007); and the Dutch National Genomics Initiative. The authors thank Sten Cornelissen, Richard van Hien, Linde Braaf, Frans Hogervorst, Senno Verhoef, Laura van 't Veer, Emiel Rutgers, Ellen van der Schoot and Femke Atsma for their support.
AGES-Reykjavik	The Age, Gene/Environment Susceptibility-Reykjavik Study is funded by NIH contract N01-AG-12100, the NIA Intramural Research Program, Hjartavernd (the Icelandic Heart Association), and the Althingi (the Icelandic Parliament). Genotyping was conducted at the NIA IRP Laboratory of Neurogenetics.
ALSPAC	A grant from the Wellcome Trust funded collection of genome-wide data on the ALSPAC mothers (WT088806). We thank the Sanger Centre, Centre National de Génotypage, and 23andMe for generating the ALSPAC genome-wide data. The UK Medical Research Council and Wellcome Trust (092731), together with the University of Bristol, provide core support for the ALSPAC study. DAL, NJT and GDS work in a unit that receives funding from the University of Bristol and the UK Medical Research Council (MC_UU_12013/1-9). We are extremely grateful to all of the families who took part in this study, the midwives for recruiting them, and the whole ALSPAC team, which includes interviewers, computer and laboratory technicians, clerical workers, research scientists, volunteers, managers, receptionists and nurses.
Amish	Supported by NIH research grants: R01 AG18728, R01HL088119, R01AR046838, U01 HL084756, U01 HL105198, U01 GM074518, P30DK072488, and F2AR059469. Construction and maintenance of the Anabaptist Genealogy Database (AGDB) is covered under an IRB-approved protocol at the National Institutes of Health (Dr. Leslie Biesecker, Principal Investigator). We would also like to thank the Amish liaisons, field staff and participants for their important contributions to this project.
AOCS	We thank D. Bowtell, A. deFazio, D. Gertig, A. Green, P. Parsons, N. Hayward, P. Webb, and D. Whiteman (AUS). AOCS was supported by the US Department of Defense DAMD17-01-1-0729, Cancer Council Victoria, Queensland Cancer Fund, Cancer Council New South Wales, Cancer Council South Australia, Cancer Foundation of Western Australia, Cancer Council Tasmania, and the National Health and Medical Research Council of Australia.
ARIC	The Atherosclerosis Risk in Communities Study is carried out as a collaborative study supported by National Heart, Lung, and Blood Institute contracts (HHSN268201100005C, HHSN268201100006C, HHSN268201100007C, HHSN268201100008C, HHSN268201100009C, HHSN268201100010C, HHSN268201100011C, and HHSN268201100012C), and the National Genome Research Institute contract U01-HG-004402. The authors thank the staff and participants of the ARIC study for their important contributions.
B58C	We acknowledge use of phenotype and genotype data from the British 1958 Birth Cohort DNA collection, funded by the Medical Research Council grant G0000934 and the Wellcome Trust grant 068545/Z/02. (http://www.b58c.gen.sugr.ac.uk/). Genotyping for the B58C-WTCCC subset was funded by the Wellcome Trust grant 076113/B/04/Z. The B58C-T1DGC genotyping utilized resources provided by the Type 1 Diabetes Genetics Consortium, a collaborative clinical study sponsored by the National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK), National Institute of Allergy and Infectious Diseases (NIAID), National Human Genome Research Institute (NHGRI), National Institute of Child Health and Human Development (NICHD), and Juvenile Diabetes Research Foundation International (JDRF) and supported by U01 DK062418. B58C-T1DGC GWAS data were deposited by the Diabetes and Inflammation Laboratory, Cambridge Institute for Medical Research (CIMR), University of Cambridge, which is funded by Juvenile Diabetes Research Foundation International, the Wellcome Trust and the National Institute for Health Research Cambridge Biomedical Research Centre; the CIMR is in receipt of a Wellcome Trust Strategic Award (079895). The B58C-GABRIEL genotyping was supported by a contract from the European Commission Framework Programme 6 (018996) and grants from the French Ministry of Research. This work was also supported by Framework VII (ENGAGE: HEALTH-F4-2007-201413) and the Wellcome Trust grant 098381. M. McCis is a Wellcome Trust Senior Investigator Scientist
BBCC	The work of the BBCC was partly funded by ELAN-Fond of the University Hospital of Erlangen.
BBCS	The BBCS is funded by Cancer Research UK and Breakthrough Breast Cancer and acknowledges NHS funding to the NIHR Biomedical Research Centre, and the National Cancer Research Network (NCRN). The authors thank Eileen Williams, Elaine Ryder-Mills and Kara Sargus for their support.
BCAC	The BCAC is funded by CR-UK (C1287/A10118 and C1287/A12014). Meetings of the BCAC have been funded by the European Union COST programme (BM0606). D.F.E. is a Principal Research Fellow of CR-UK.
CAHRES	The CAHRES study was supported by funding from the Agency for Science, Technology and Research of Singapore (A*STAR), the United States National Institute of Health (NIH) and the Susan G. Komen Breast Cancer Foundation. E.I. was supported by grants from the Swedish Research Council, the Swedish Heart-Lung Foundation, the Swedish Society of Medicine, the Swedish Foundation for Strategic Research, and the Royal Swedish Academy of Science while working with this article.
CECILE	The CECILE study was funded by Fondation de France, Institut National du Cancer (INCa), Ligue Nationale contre le Cancer, Ligue contre le Cancer Grand Ouest, Agence Nationale de Sécurité Sanitaire (ANSES), Agence Nationale de la Recherche (ANR)
CGPS	The CGPS was supported by the Chief Physician Johan Bøserup and Use Bøserup Fund, the Danish Medical Research Council and Herlev Hospital. The authors thank staff and participants of the Copenhagen General Population Study. For the excellent technical assistance: Dorthe Uldall Andersen, Maria Birna Arnadottir, Anne Bank, Dorthe Kjeldgård Hansen.
CoLaus	The CoLaus study was and is supported by research grants from GlaxoSmithKline, the Faculty of Biology and Medicine of Lausanne, Switzerland, and by the Swiss National Science Foundation Grants #33CS0-122661 and 33CS30-139468.
CNIO-BCS	The CNIO-BCS was supported by the Genome Spain Foundation, the Red Temática de Investigación Cooperativa en Cáncer and grants from the Asociación Española Contra el Cáncer and the Fondo de Investigación Sanitaria (PI11/00923 and PI081120). The Human Genotyping-CEGEN Unit (CNIO) is supported by the Instituto de Salud Carlos III. The authors thank Guillermo Pita, Charo Alonso, Daniel Herrero, Nuria Álvarez, Pilar Zamora, Primitiva Menéndez, the Human Genotyping-CEGEN Unit (CNIO).
CTS	The CTS was supported by the California Breast Cancer Act of 1993; National Institutes of Health (grants R01 CA77398 and the Lon V Smith Foundation [LVS39420]); and the California Breast Cancer Research Fund (contract 97-10500). Collection of cancer incidence data used in this study was supported by the California Department of Public Health as part of the statewide cancer reporting program mandated by California Health and Safety Code Section 103885.
DNBC	Funding support for the DNBC was provided by the Danish National Research Foundation, the Danish Pharmacists' Fund, the Egmont Foundation, the March of Dimes Birth Defects Foundation, the Augustinus Foundation and the Health Fund of the Danish Health Insurance Societies. The generation of GWAS genotype data for the DNBC samples was carried out within the GENEVA consortium with funding provided through the NIH Genes, Environment and Health Initiative (GEI) (U01HG004423). Assistance with phenotype harmonization and genotype cleaning, as well as with general study coordination, was provided by the GENEVA Coordinating Center (U01HG004446). Genotyping was performed at Johns Hopkins University Center for Inherited Disease Research, with support from the NIH GEI (U01HG004438). We thank the women participating in the Danish National Birth Cohort (DNBC).

EGCUT	This work was supported by the Targeted Financing from the Estonian Ministry of Science and Education [SF0180142s08]; the Development Fund of the University of Tartu (grant SP1GVARENG); the European Regional Development Fund to the Centre of Excellence in Genomics (EXCEGEN; grant 3.2.0304.11-0312); and through FP7 grant 313010.
EPIC	The EPIC Norfolk Study is funded by Cancer Research United Kingdom and the Medical Research Council. This work was supported by the Medical Research Council [U106179472; MC_U106179472; U106179471; MC_U106179471]. The authors acknowledge the support of all EPIC-Norfolk staff and participants.
ESTHER	The ESTHER study was supported by a grant from the Baden Württemberg Ministry of Science, Research and Arts. Additional cases were recruited in the context of the VERDI study, which was supported by a grant from the German Cancer Aid (Deutsche Krebshilfe). The authors thank Hartwig Ziegler, Sonja Wolf and Volker Hermann for their support.
FHS	The Framingham Heart Study phenotype-genotype analyses were supported by the National Institute of Aging (Genetics of Reproductive Life Period and Health Outcomes, R21AG032598; JMM, KL, DEK, DPK, R01AG29451; JMM, KL and R01AR41398; DPK). The Framingham Heart Study of the National Heart Lung and Blood Institute of the National Institutes of Health and Boston University School of Medicine was supported by the National Heart, Lung and Blood Institute's Framingham Heart Study Contract No. N01-HC-25195 and its contract with Affymetrix, Inc for genotyping services (Contract No. N02-HL-6-4278). Analyses reflect intellectual input and resource development from the Framingham Heart Study investigators participating in the SNP Health Association Resource (SHARE) project. A portion of this research was conducted using the Linux Cluster for Genetic Analysis (LinGA-II) funded by the Robert Dawson Evans Endowment of the Department of Medicine at Boston University School of Medicine and Boston Medical Center. The authors thank the Framingham study participants and staff.
GENICA	The GENICA was funded by the Federal Ministry of Education and Research (BMBF) Germany grants 01KW9975/5, 01KW9976/8, 01KW9977/0 and 01KW0114, the Robert Bosch Foundation, Stuttgart, Deutsches Krebsforschungszentrum (DKFZ), Heidelberg, Institute for Prevention and Occupational Medicine of the German Social Accident Insurance, Institute of the Ruhr University Bochum (IPA), as well as the Department of Internal Medicine, Evangelische Kliniken Bonn gGmbH, Johanniter Krankenhaus, Bonn, Germany. The authors acknowledge the support of The GENICA Network: Dr. Margarete Fischer-Bosch-Institute of Clinical Pharmacology, Stuttgart, and University of Tübingen, Germany; [HB, Wing-Yee Lo, Christina Justenhoven], Department of Internal Medicine, Evangelische Kliniken Bonn gGmbH, Johanniter Krankenhaus, Bonn, Germany [Yon-Dschun Ko, Christian Baisch], Institute of Pathology, University of Bonn, Germany [Hans-Peter Fischer], Molecular Genetics of Breast Cancer, Deutsches Krebsforschungszentrum (DKFZ), Heidelberg, Germany [Ute Hamann] and Institute for Prevention and Occupational Medicine of the German Social Accident Insurance, Institute of the Ruhr University Bochum (IPA), Bochum, Germany [Thomas Brünig, Beate Pesch, Sylvia Rabstein, Anne Lotz]; Institute of Occupational Medicine and Maritime Medicine, University Medical Center Hamburg-Eppendorf, Germany [Volker Harth].
GOYA (Genomics of Obesity in Young Adults)	The genotyping for GOYA was funded by the Wellcome Trust (WT 084762). GOYA is a nested study within The Danish National Birth Cohort which was established with major funding from the Danish National Research Foundation. Additional support for this cohort has been obtained from the Pharmacy Foundation, the Egmont Foundation, The March of Dimes Birth Defects Foundation, the Augustinus Foundation, and the Health Foundation. The GOYA study was conducted as part of the activities of the Danish Obesity Research Centre (DanORC, www.danorc.dk) and The MRC centre for Causal Analyses in Translational Epidemiology (MRC CAITE).
Health2000/GENMETS	The Health 2000 Study is funded by the National Institute for Health and Welfare (THL), the Finnish Centre for Pensions (ETK), The Social Insurance Institution of Finland (KELA), The Local Government Pensions Institution (KEVA) and other organizations listed on the website of the survey (http://www.terveys2000.fi). GWAS genotyping was supported by the Wellcome Trust Sanger Institute. VS was supported by the Academy of Finland (grant number 139635).
HEBCS	The HEBCS was financially supported by the Helsinki University Central Hospital Research Fund, Academy of Finland (132473), the Finnish Cancer Society, The Nordic Cancer Union and the Sigrid Juselius Foundation. The authors acknowledge the support of Kristiina Aittomäki, Kirsimari Aaltonen, Karl von Smitten, Taru A. Muranen and Irja Erkkilä.
iCOGS	Funding for the iCOGS infrastructure came from: the European Community's Seventh Framework Programme under grant agreement n° 223175 (HEALTH-F2-2009-223175) (COGS), Cancer Research UK (C1287/A10118, C1287/A10710, C12292/A11174, C1281/A12014, C5047/A8384, C5047/A15007, C5047/A10692), the National Institutes of Health (CA128978) and Post-Cancer GWAS initiative (1U19 CA148537, 1U19 CA148065 and 1U19 CA148112 - the GAME-ON initiative), the Department of Defence (W81XWH-10-1-0341), the Canadian Institutes of Health Research (CIHR) for the CIHR Team in Familial Risks of Breast Cancer, Komen Foundation for the Cure, the Breast Cancer Research Foundation, and the Ovarian Cancer Research Fund. This study would not have been possible without the contributions of the following: Andrew Berchuck (OCAC), Rosalind A. Eeles, Ali Amin Al Olama, Zsofia Kote-Jarai, Sara Beniloch (PRACTICAL), Antonis Antoniou, Lesley McGuffog, Ken Offit (CIMBA), Andrew Lee, and Ed Dicks, Craig Luccarini and the staff of the Centre for Genetic Epidemiology Laboratory, Anna Gonzalez-Neira and the staff of the CINQ genotyping unit, Jacques Simard and Daniel C. Tessier, Francois Bacot, Daniel Vincent, Sylvie LaBoissière and Frederic Robidoux and the staff of the McGill University and Génome Québec Innovation Centre, Sune F. Nielsen, Borge G. Nordestgaard, and the staff of the Copenhagen DNA laboratory, and Julie M. Cunningham, Sharon A. Windebank, Christopher A. Hilkert, Jeffrey Meyer and the staff of Mayo Clinic Genotyping Core Facility. We thank all the individuals who took part in these studies and all the researchers, clinicians, technicians and administrative staff who have enabled this work to be carried out.
InChianti	InCHIANTI: The InCHIANTI study baseline (1998-2000) was supported as a "targeted project" (ICS110.1/RF97.71) by the Italian Ministry of Health and in part by the U.S. National Institute on Aging (Contracts: 263 MD 9164 and 263 MD 821336); the InCHIANTI Follow-up 1 (2001-2003) was funded by the U.S. National Institute on Aging (Contracts: N1-A-AG-1-1 and N1-A-AG-1-2111); the InCHIANTI Follow-ups 2 and 3 studies (2004-2010) were financed by the U.S. National Institute on Aging (Contract: N01-A-5-0002); supported in part by the Intramural research program of the National Institute on Aging, National Institutes of Health, Baltimore, Maryland.
Indiana [Indiana University premenopausal Caucasian women peak BMD study]	Funded by US NIH NIA R01AG041517
INGI-Carlantino	We thank Anna Morgan and Angela D'Eustacchio for technical support. We are very grateful to the municipal administrators for their collaboration on the project and for logistic support. We would like to thank all participants to this study.
INGI-FVG	We thank Anna Morgan and Angela D'Eustacchio for technical support. We are very grateful to the municipal administrators for their collaboration on the project and for logistic support. We would like to thank all participants to this study. The study was funded Regione FVG (L.26.2008)
InterAct	We thank all EPIC participants and staff for their contribution to the study. We thank staff from the Technical, Field Epidemiology and Data Functional Group Teams of the MRC Epidemiology Unit in Cambridge, UK, for carrying out sample preparation, DNA provision and quality control, genotyping and data-handling work. The EPIC-InterAct study received funding from the European Union (Integrated Project LSHM-CT-2006-037197 in the Framework Programme 6 of the European Community).
IUBC	The IUBC study is supported by the Indiana Clinical and Translational Sciences Institute (CTSI) Program Development Team Award, and the IUBC genome-wide genotyping is supported by the Expression Analysis and Illumina co-sponsored GWAS grant. We acknowledge donors to the Susan G. Komen for the Cure® Tissue Bank at the Indiana University Simon Cancer Center for their contribution in making this work possible.
KARBAC	Financial support for KARBAC was provided through the regional agreement on medical training and clinical research (ALF) between Stockholm County Council and Karolinska Institutet, the Swedish Cancer Society, The Gustav V Jubilee foundation and Bert von Kantows foundation.
KBCP	The KBCP was financially supported by the special Government Funding (EVO) of Kuopio University Hospital grants, Cancer Fund of North Savo, the Finnish Cancer Organizations, the Academy of Finland and by the strategic funding of the University of Eastern Finland. The authors acknowledge the support of Eija Myöhänen and Helena Kemiläinen.
kConFab	kConFab is supported by a grant from the National Breast Cancer Foundation, and previously by the National Health and Medical Research Council (NHMRC), the Queensland Cancer Fund, the Cancer Councils of New South Wales, Victoria, Tasmania and South Australia, and the Cancer Foundation of Western Australia. We wish to thank Heather Thorne, Eveline Niedermayr, all the kConFab research nurses and staff, the heads and staff of the Family Cancer Clinics, and the Clinical Follow Up Study (which has received funding from the NHMRC, the National Breast Cancer Foundation, Cancer Australia, and the National Institute of Health (USA) for their contributions to this resource, and the many families who contribute to kConFab.

KORA	The KORA research platform (KORA, Cooperative Health Research in the Region of Augsburg) was initiated and financed by the Helmholtz Zentrum München - German Research Center for Environmental Health, which is funded by the German Federal Ministry of Education and Research and by the State of Bavaria. Furthermore, KORA research was supported within the Munich Center of Health Sciences (MC Health), Ludwig-Maximilians-Universität, as part of LMUinnovativ. We thank all KORA study participants and all members of the field staff in Augsburg who planned and conducted the study.
LifeLines	The LifeLines Cohort Study is supported by the Netherlands Organization of Scientific Research NWO (grant 175.010.2007.006), the Economic Structure Enhancing Fund (FES) of the Dutch government, the Ministry of Economic Affairs, the Ministry of Education, Culture and Science, the Ministry for Health, Welfare and Sports, the Northern Netherlands Collaboration of Provinces (SNN), the Province of Groningen, University Medical Center Groningen, the University of Groningen, Dutch Kidney Foundation and Dutch Diabetes Research Foundation. The authors wish to acknowledge the services of the LifeLines Cohort Study and the contributing research centers delivering data to LifeLines. We thank Behrooz Z. Alizadeh, Annete Boesjes, Marcel Bruijnenberg, Noortje Festen, Pim van der Harst, Iija Nolte, Lude Franke, Mitra Valimohammadi for their help in creating the GWAS database, and Rob Bieringa, Joost Keers, René Ostergaard, Rosalie Visser, Judith Vonk for their work related to data-collection and validation. The authors are grateful to the study participants, the staff from the LifeLines Cohort Study and Medical Biobank Northern Netherlands, and the participating general practitioners and pharmacists.
LMBC	LMBC is supported by the 'Stichting tegen Kanker' (232-2008 and 196-2010). Diether Lambrechts is supported by the FWO and the KULP/FV/10/016-SymBioSysII. The authors acknowledge the support of Gillian Peuteman, Dominiek Smeets, Thomas Van Brussel and Kathleen Corthouts.
MARIE	The MARIE study was supported by the Deutsche Krebshilfe e.V. [70-2892-BR I], the Hamburg Cancer Society, the German Cancer Research Center and the Federal Ministry of Education and Research (BMBF) Germany [01KH0402]. The authors acknowledge the support of Judith Heinz, Nadia Obi, Alina Vrieling, Sabine Behrens, Ursula Eilber, Muhabbet Celik, Til Olchers, and Stefan Nickels.
MBCSG	MBCSG is supported by grants from the Italian Association for Cancer Research (AIRC) and by funds from the Italian citizens who allocated the 5/1000 share of their tax payment in support of the Fondazione IRCCS Istituto Nazionale Tumori, according to Italian laws (INT-Institutional strategic projects "5x1000"). The authors acknowledge the support of Shiranoush Manoukian, Bernard Peissel and Daniela Zaffaroni of the Fondazione IRCCS Istituto Nazionale dei Tumori (INT); Monica Barile and Irene Feroco of the Istituto Europeo di Oncologia (IEO) and the personnel of the Cogentech Cancer Genetic Test Laboratory.
MCBCS	The MCBCS was supported by the NIH grants [CA112340, CA128978], an NIH Specialized Program of Research Excellence (SPORE) in Breast Cancer [CA116201], the Breast Cancer Research Foundation, and the Komen Race for the Cure
MCCS	MCCS cohort recruitment in the was funded by VicHealth and Cancer Council Victoria. The MCCS was further supported by Australian NHMRC grants 209057, 251553 and 504711 and by infrastructure provided by Cancer Council Victoria.
NHS	The NHS GWAS were supported by grants from the National Institutes of Health [NCI (CA40356, CA087969, CA055075, CA98233, U01 CA137088, R01 CA059045, R01 CA137178, R01 CA082838, R01 CA131332), NIDDK (DK058845, DK070756), NHGRI (HG004399, HG004728), NHLBI (HL35464), NIAMS (R01 AR056291)]. We acknowledge the study participants in the NHS and NHS II for their contribution in making this study possible.
NTR	Funding was obtained from the Netherlands Organization for Scientific Research (NWO: MagW/ZonMW): Genetic basis of anxiety and depression (904-61-090); Genetics of individual differences in smoking initiation and persistence (NWO 985-10-002); Resolving cause and effect in the association between exercise and well-being (904-61-193); Twin family database for behavior genetics studies (480-04-004); Twin research focusing on behavior (400-05-717); Genetic determinants of risk behavior in relation to alcohol use and alcohol use disorder (Addiction-31160008); Genotype/phenotype database for behavior genetic and genetic epidemiological studies (40-0056-98-9032); Spinozapremie (SP1 56-464-14192); CMSB: Center for Medical Systems Biology (NWO Genomics); NBIC/BioAssist/RK/2008.024); BBMRI –NL: Biobanking and Biomolecular Resources Research Infrastructure (184.021.007); the VU University: Institute for Health and Care Research (EMGO+) and Neuroscience Campus Amsterdam (NCA); the European Science Foundation (ESF): Genomewide analyses of European twin and population cohorts (EU/QLRT-2001-01254); European Community's Seventh Framework Program (FP7/2007-2013): ENGAGE (HEALTH-F4-2007-201413); the European Science Council (ERC) Genetics of Mental Illness (230374); Rutgers University Cell and DNA Repository cooperative agreement (NIMH U24 MH068457-06); Collaborative study of the genetics of DZ twinning (NIH R01D0042157-01A); the Genetic Association Information Network, a public–private partnership between the NIH and Pfizer Inc., Affymetrix Inc. and Abbott Laboratories. The NTR study would like to thank all of our study participants for their continuous voluntary contributions to our scientific efforts as well as the SURF SARA institute for their computational resources.
OBCS	The OBCS was supported by research grants from the Finnish Cancer Foundation, the Academy of Finland, the University of Oulu, and the Oulu University Hospital. The authors acknowledge the support of Meeri Otsukka and Kari Mononen.
OFBCR	This work was supported by grant UM1 CA164920 from the National Cancer Institute. The content of this manuscript does not necessarily reflect the views or policies of the National Cancer Institute or any of the collaborating centers in the Breast Cancer Family Registry (BCFR), nor does mention of trade names, commercial products, or organizations imply endorsement by the US Government or the BCFR. The authors acknowledge the support of Teresa Selander, Nayana Weerasooriya.
ORIGO	The ORIGO study was supported by the Dutch Cancer Society (RUL 1997-1505) and the Biobanking and Biomolecular Resources Research Infrastructure (BBMRI-NL CP16). We thank E. Krol-Warmerdam, and J. Blom for patient accrual, administering questionnaires, and managing clinical information. The LUMC survival data were retrieved from the Leiden hospital-based cancer registry system (ONCDOC) with the help of Dr. J. Molenaar.
PBCS	The PBCS was funded by Intramural Research Funds of the National Cancer Institute, Department of Health and Human Services, USA. The authors acknowledge the support of Louise Brinton, Mark Sherman, Stephen Chanock, Neonila Szczesnia-Dabrowska, Beata Peplonska, Witold Zatonski, Pei Chao and Michael Stagner.
pKARMA	The Swedish Medical Research Council. The pKARMA study was supported by Märta and Hans Rausing's Initiative Against Breast Cancer.
QIMR	Funding was provided by the Australian National Health and Medical Research Council (241944, 339462, 389927, 389875, 389891, 389892, 389938, 442915, 442981, 496739, 552485, 552498, Australian Research Council (A7960034, A79906588, A79801419, DP0770096, DP0212016, DP0343921), the FP-5 GenomEUtwin Project (QLG2-CT-2002-01254), and the U.S. National Institutes of Health (NIH grants AA07535, AA10248, AA13320, AA13321, AA13326, AA14041, MH62602). We thank the twins and their families for their participation. We also thank Dixie Statham, Ann Eldridge, Marlene Grace, Kerrie McAloone (sample collection); Lisa Bowdler, Steven Crooks (DNA processing); Sarah Medland, Dale Nyholt and Scott Gordon (Imputation and genotyping QC). E.M.B. is supported by an NHMRC Early Career Fellowship.
RBCS	The RBCS was funded by the Dutch Cancer Society (DDHK 2004-3124, DDHK 2009-4318). The authors acknowledge the support of Petra Bos, Jannet Blom, Ellen Crepin, Elisabeth Huijkens, Annette Heemskerk and the Erasmus MC Family Cancer Clinic.
Rotterdam Study I, II and III	The generation and management of GWAS genotype data for the Rotterdam Study are supported by the Netherlands Organisation of Scientific Research NWO Investments (nr. 175.010.2005.011, 911-03-012). This study is funded by the Research Institute for Diseases in the Elderly (014-93-015; RIDE2), the Netherlands Genomics Initiative (NGI)/Netherlands Organisation for Scientific Research (NWO) project nr. 050-060-810, and funding from the European Commission (HEALTH-F2-2008-201865, GEFOS; HEALTH-F2-2008-35627, TREAT-OA). The Rotterdam Study is funded by Erasmus Medical Center and Erasmus University, Rotterdam, Netherlands Organization for the Health Research and Development (ZonMw), the Research Institute for Diseases in the Elderly (RIDE), the Ministry of Education, Culture and Science, the Ministry for Health, Welfare and Sports, the European Commission (DG XII), and the Municipality of Rotterdam. We thank Pascal Arp, Mila Jhamai, Dr Michael Moorhouse, Marijn Verkerk, and Sander Bervoets for their help in creating the GWAS database. The authors are grateful to the study participants, the staff from the Rotterdam Study and the participating general practitioners and pharmacists. We would like to thank Dr. Tobias A. Knoll, Anis Abuseiris, Karol Estrada, and Rob de Graaf as well as their institutions Biophysical Genomics, Erasmus MC Rotterdam, The Netherlands, and especially the national German MediGRID and Services@MediGRID part of the German D-Grid, both funded by the German Bundesministerium für Forschung und Technologie under grants #01 AK 803 A-H and #01 IG 07015 G for access to their grid resources.

SAGE	Funding support for the Study of Addiction: Genetics and Environment (SAGE) was provided through the NIH Genes, Environment and Health Initiative [GEI] (U01 HG004422). Assistance with phenotype harmonization and genotype cleaning, as well as with general study coordination, was provided by the GENEVA Coordinating Center (U01 HG004446). Support for collection of datasets and samples was provided by the Collaborative Study on the Genetics of Alcoholism (COGA; U10 AA008401), the Collaborative Genetic Study of Nicotine Dependence (COGEND; P01 CA089392), and the Family Study of Cocaine Dependence (FSCD; R01 DA013423, R01 DA019963). Funding support for genotyping, which was performed at the Johns Hopkins University Center for Inherited Disease Research, was provided by the NIH GEI (U01HG004438), the National Institute on Alcohol Abuse and Alcoholism, the National Institute on Drug Abuse, and the NIH contract "High throughput genotyping for studying the genetic contributions to human disease" (HHSN268200782096C).
SardiNIA	The SardiNIA study is supported by the National Institute on Aging, NIH, contract N01-AG-1-2109 to the SardiNIA ("ProgeNIA") team. The authors are grateful to all of the volunteers who participated in this study, the Bishop of Ogliastra, mayors and citizens of the Sardinian towns (Lanusei, Ilbono, Arzana, and Elini), the head of the Public Health Unit ASL4 for their volunteerism and cooperation, and the team of physicians, nurses, biologists and the recruitment personnel.
SASBAC	The SASBAC study was supported by funding from the Agency for Science, Technology and Research of Singapore (A*STAR), the US National Institute of Health (NIH) and the Susan G. Komen Breast Cancer Foundation. The authors thank the The Swedish Medical Research Council.
SBCS	The SBCS was supported by Yorkshire Cancer Research S295, S299, S305PA. The authors acknowledge the support of Malcolm W.R. Reed, Simon S. Cross, Sabapathy Balasubramanian, Sue Higham, Helen Cramp, and Dan Connley.
SEARCH	SEARCH is funded by a programme grant from Cancer Research UK [C490/A10124] and supported by the UK National Institute for Health Research Biomedical Research Centre at the University of Cambridge. The authors acknowledge the support of the SEARCH and EPIC study teams.
SHIP-TREND	SHIP is part of the Community Medicine Research net of the University of Greifswald, Germany, which is funded by the Federal Ministry of Education and Research (grants no. 01ZZ9603, 01ZZ0103, 01ZZ0403 and 03IS2061A), the Ministry of Cultural Affairs as well as the Social Ministry of the Federal State of Mecklenburg-West Pomerania, and the network 'Greifswald Approach to Individualized Medicine (GANI_MED)' funded by the Federal Ministry of Education and Research (grant 03IS2061A). The University of Greifswald is a member of the 'Center of Knowledge Interchange' program of the Siemens AG and the Caché Campus program of the InterSystems GmbH.
SKDKFZS	We thank all study participants, clinicians, family doctors, researchers and technicians for their contributions and commitment to this study. SKDKFZS is supported by the DKFZ.
TRAILS	This research is part of the TRacking Adolescents' Individual Lives Survey (TRAILS). Participating centers of TRAILS include various departments of the University Medical Center and University of Groningen, the Erasmus University Medical Center Rotterdam, the University of Utrecht, the Radboud Medical Center Nijmegen, and the Parnassia Bavo group, all in the Netherlands. TRAILS has been financially supported by various grants from the Netherlands Organization for Scientific Research NWO (Medical Research Council program grant GB-MW 940-38-011; ZonMW Brainpower grant 100-001-004; ZonMw Risk Behavior and Dependence grants 60-60600-97-118; ZonMw Culture and Health grant 261-98-710; Social Sciences Council medium-sized investment grants GB-MaGW 480-01-006 and GB-MaGW 480-07-001; Social Sciences Council project grants GB-MaGW 452-04-314 and GB-MaGW 452-06-004; NWO large-sized investment grant 175.010.2003.005; NWO Longitudinal Survey and Panel Funding 481-08-013), the Dutch Ministry of Justice (WODC), the European Science Foundation (EuroSTRESS project FP-006), Biobanking and Biomolecular Resources Research Infrastructure BBMRI-NL (CP 32), and the participating universities. We are grateful to all adolescents, their parents and teachers who participated in this research and to everyone who worked on this project and made it possible. Statistical analyses were carried out on the Genetic Cluster Computer (http://www.geneticcluster.org), which is financially supported by the Netherlands Scientific Organization (NWO 480-05-003) along with a supplement from the Dutch Brain Foundation.
TWINGENE	This work was supported by grants from the Ministry for Higher Education, the Swedish Research Council (M-2005-1112 and 2009-2298), GenomEUtwin (EU/QLRT-2001-01254; QLG2-CT-2002-01254), NIH grant DK U01-066134, The Swedish Foundation for Strategic Research (SSF; ICA08-0047).
TwinsUK	TwinsUK. The study was funded by the Wellcome Trust; European Community's Seventh Framework Programme (FP7/2007-2013). The study also receives support from the National Institute for Health Research (NIHR) BioResource Clinical Research Facility and Biomedical Research Centre based at Guy's and St Thomas' NHS Foundation Trust and King's College London. Tim Spector is holder of an ERC Advanced Principal Investigator award. SNP Genotyping was performed by The Wellcome Trust Sanger Institute and National Eye Institute via NIH/CIDR.
Western Australian Pregnancy (Raine) Cohort	Core funding for the Raine Study is provided by The Raine Medical Research Foundation at the University of Western Australia, The University of Western Australia, the Women and Infants Research Foundation, Curtin University and the Telethon Institute for Child Health Research. Project specific funding was provided by the National Health and Medical Research Council (NHMRC) APP572613. We are extremely grateful to all the families who took part in this study and the whole Raine Study team, which includes data collectors, cohort managers, data managers, clerical staff, research scientists and volunteers.
Women's Genome Health Study (WGHS)	The WGHS is supported by HL043851 and HL080467 from the National Heart, Lung, and Blood Institute and CA047988 from the National Cancer Institute with collaborative scientific support and funding for genotyping provided by Amgen.

Disclosures:

The University of Groningen has received money for Professor Postma regarding an unrestricted educational grant for research from Astra Zeneca, Chiesi. Travel to ERS and/or ATS has been partially funded by Astra Zeneca, Chiesi, GSK, Takeda. Fees for consultancies were given to the University of Groningen by Astra Zeneca, Boehringer Ingelheim, Chiesi, GSK, Takeda and REVA. Travel and lectures in China paid by Chiesi. Laura J. Bierut is listed as an inventor on Issued U.S. Patent 8,080,371, "Markers for Addiction" covering the use of certain SNPs in determining the diagnosis, prognosis, and treatment of addiction.

3. Author Contributions

Overall project management

JRBP, FD, CEE, PS, DJT, DFE, KS, JMM, KKO

Core analyses

JRBP, FD, CEE, PS, TF, DJT, DIC, TE, GT

Individual study analysts

AAR, AD, AG, AJ, AT, AVS, BZA, BF, CEE, DFG, DIC, DJT, DLC, DLK, EA, EKW, EM, EMB, ET, FD, GM, GmM, IMN, JAV, JD, JH, JRBP, JT, JZ, KLL, KM, LLP, LMR, LMY, LS, MM, NF, NTs, PK, PS, RM, SK, SS, SSU, TC, TE, TF, TFo, THP, WQA, ZK

Individual study data management and generation

AAR, ACH, AD, ADC, AGU, AJO, AMS, AMu, AP, APo, BAO, CAH, DC, DIC, DJH, DK, DLw, DPK, DPS, DS, EAN, EP, EW, FA, FBH, FG, FR, GD, GE, GGW, HS, HW, ID, JC, JH, JPR, LF, LFr, LM, LMR, MEG, MJS, MJW, MKB, MMb, MP, MW, NA, NJT, NLP, PKM, QW, RH, SB, SC, SG, SL, SR, SSU, TE, US, UT, VS, WLM

Individual study PIs

AC, AGU, AH, AJO, AKD, AL, AM, AMD, AMm, AMu, AR, BB, BZA, BHRW, CB, CEP, CG, CH, CMv, DIB, DF, DFE, DJH, DL, DLw, DSP, DPS, DSs, EAS, EB, EEJd, EI, EW, EWD, FBH, FJC, GC, GD, GGG, GW, GWM, HA, HAB, HB, HBe, HF, HN, HS, HV, ID, ILA, JAK, JB, JCC, JGE, JEB, JLH, JMC, JMM, JP, KC, KK, KKO, KP, KS, LC, LF, LJb, MCS, MG, MIM, MJ, MJE, MJH, MJS, MKS, MWB, MZ, NGM, NJW, PAF, PD, PDPP, PFM, PG, PH, PK, PMR, PN, PP, PPG, PR, PV, RJFL, RLM, RW, SB, SBm, SC, SEB, TBH, TDS, TIAS, UH, VG, VK, VS

3. Website

After publication, the ReproGen Consortium website (www.reprogen.org) will host a publicly accessible version of the GWAS summary statistics, along with additional locus zoom plots of the regions highlighted in this manuscript.