**Supplementary Note**

## Detailed Description of the Case and Control Sample Sets

Subjects were drawn from 11 studies, comprising 6,054 endometrial cancer cases and 14,125 controls. A summary of the studies included in stage1 and stage 2 is shown in Supplementary Table 2, with additional details provided below. All studies were predominantly of women of European ancestry, with the exception of SECS, which included women of Asian ancestry from China. All studies have the relevant IRB approval in each country in accordance with the principles embodied in the Declaration of Helsinki, and informed consent was obtained from all participants.

### Australian Studies, incorporating ANECS/ARCBS and NECS

The Australian National Endometrial Cancer Study (ANECS) is an Australian population-based case-control-family study of cancer of the uterine corpus (1). Women aged 18-79, newly diagnosed with histologically confirmed primary cancer of the endometrium between July 2005 and December 2007 were identified through major hospitals nationally, and also from state-based cancer registries. Excluding women who could not be contacted (mostly due to death, illness or failure to contact), case participation rate was 63%. Cases who reported a family history of cancer were also asked to invite their relatives (with and without cancer) to participate in the family component of the study. Relatives were not genotyped for this SNP study. Female controls, with no personal history of endometrial cancer or hysterectomy were recruited using two sources: A population-based control group comprised women randomly selected using the Australian Electoral Roll (voting is compulsory in Australia) and matched to the age and geographic distribution of the cases (53% participation among those contacted); and a control group of female blood donors recruited with the aid of the Australian Red Cross Blood Service in Queensland (>99% participation rate) (2). Participants completed a detailed questionnaire providing clinical and epidemiological information, including ethnicity of all four grandparents. Information on tumor pathology characteristics was abstracted in standardized format from clinical pathology reports for all patients.

The Newcastle Endometrial Cancer Study (NECS) includes histologically confirmed endometrial cancer cases consecutively recruited from 1992 up to 2005 at the Hunter Centre for Gynaecological Cancer, John Hunter Hospital, Newcastle, New South Wales, Australia (3). The final analysis included 194 endometrial cancer patients. Data on reproductive and environmental risk factors including ethnicity, was collected using self reported questionnaires. Information regarding recurrence, stage, grade and histology of endometrial cancer was collected from medical records. Patients presenting at this hospital-based site were captured by ANECS recruitment from 2005 onwards.

### SEARCH

The Studies of Epidemiology and Risk factors in Cancer Heredity (SEARCH) is an ongoing population-based study with cases ascertained through the Eastern Cancer Registration and Information Centre (http://www.ecric.org.uk). All women diagnosed with endometrial cancer between the ages of 18-69 years (average age diagnosis 58 years) from 31 July 2001 to 1st September 2007 were eligible for inclusion. Approximately 54% of eligible patients have enrolled in the study. Women taking part in the study were asked to provide a 20ml blood sample for DNA analysis, and to complete a comprehensive epidemiological questionnaire. Controls were also drawn from SEARCH (http://www.srl.cam.ac.uk/search/Homepage.htm), but had no prior history of cancer at the time of recruitment. They were female, also between the ages of 18-69 at the time of recruitment and matched to cases in geographical profile. Approximately 35% of eligible controls enrolled in the study. All participants reported Caucasian ethnicity. Information on tumor pathology characteristics was provided by the Eastern Cancer Registration and Information Centre and was derived from clinical pathology reports for all patients.

**WTCCC**
Controls utilized for stage 1 analysis were genotyped as part of the Wellcome Trust Case Control Consortium (WTCCC2) (4). These controls are drawn from two sources: 2,922 controls from the 1958 Birth Cohort (1958BC), a population-based study in the United Kingdom of individuals born in 1 week in 1958 (5); and 2,737 controls identified through the UK National Blood Service (NBS) (4). The analyses presented here are based on 2,694 1958BC and 2,496 NBS controls for which valid genotype data were available at the time of analysis.

**BECS**
The Bavarian Endometrial Cancer Cases and Controls Study (BECS) is a single-center case-control study, conducted between 2002 and 2008, with the aim of investigating genetic and epidemiological risk factors for endometrial cancer. Cases were either incident cases referred to the University Hospital Erlangen by surrounding practioners (66% of the case sample set), or prevalent cases that were outpatients in follow-up care approached within 6.2 (±4.6 SD years after treatment for primary endometrial cancer in the same hospital (34% of the case sample set). Controls were age-matched individuals invited by newspaper advertisement to take part in this case-control study. Epidemiological information for cases and controls was collected by a structured questionnaire, which was completed during an interview and clinical data for the cases was obtained from clinical health records.

**LES**
The Leuven Endometrial Study (LES) is a hospital based case-control study. Eligible cases, identified by active surveillance of electronic patient files at the Leuven University Hospital, were white women aged 27-80 years diagnosed with endometrial cancer. Clinical data for endometrial cancer patients were recorded during interview at the time of diagnosis, and from pathology reports. All medical records were reviewed by trained abstractors and pathology reports compatible with primary, invasive, epithelial endometrial adenocarcinoma of all stages (I –IV) and all grades were consulted. A control group of healthy female blood donors was recruited with the aid of the Red Cross Blood Service in the University Hospital. Participants completed a detailed questionnaire providing epidemiological information, including self-reported Belgian (Flemish) ethnicity for 3 generations. Participation rates exceeded 95% for both cases and controls.

**MoMaTEC**
Molecular Markers in Treatment of Endometrial Cancer (MoMaTEC) cases were recruited from an unselected patient population primarily treated for endometrial carcinoma at Haukeland University Hospital, Bergen during 2001-2009. This is the referral hospital for Hordaland county; the area is demographically well defined, with about 450,000 inhabitants, representing approximately 10% of the Norwegian population and with a similar incidence rate and prognosis as the total Norwegian population of endometrial cancers (6-8). Clinical Information for cases regarding age, FIGO stage, histologic subtype, grade and prognosis was extracted from medical records. Controls were healthy female Caucasian blood donors with no known cancer disease, and with known age at sampling. DNA was extracted from peripheral blood samples for cases and controls.

**NSECG**
National Study of the Genetics of Endometrial Cancer (NSECG) cases were identified from collaborating clinicians throughout the UK from 2008 to present, taking care not to recruit from centres involved in SEARCH. Inclusion criteria were adenocarcinomas of the uterus presenting at 70 years of age or younger. Almost all cases were incident and sampled within 6 months of diagnosis. Peripheral blood was collected from each participant and DNA extracted using standard methods. Tumour histology was confirmed from routine hospital reports and further details of histopathology and other tumor pathology characteristic was abstracted from these clinical pathology reports. Cases were

genotyped for stage 2 SNPs using sequenom iplex methodology, as indicated in Supplementary Table 2. Controls (45% males, 55% females) were drawn from the UK1/CORGI colorectal cancer sample set (9), and were spouses or partners of colorectal cancer cases unaffected by cancer and without a personal family history (to 2nd degree relative level) of colorectal neoplasia. All cases and controls were of white UK ethnic origin. Control genotype data for stage 2 SNPs were extracted from existing Illumina 550K genome-wide scan data (9), or genotyped using the Kaspar method if the SNP was not present on the Illumina 550K platform.

**PECS**

The Polish Endometrial Cancer Study (PECS) is a large, population-based, case–control study (10) conducted among women residing in two Polish cities, Warsaw and Lodz, during 2001–2003. Eligible cases were women aged 20–74 newly diagnosed with histologically or cytologically confirmed invasive endometrial cancer, identified through a rapid identification system at participating hospitals and also through the local cancer registries. Eligible controls with no history of endometrial cancer or hysterectomy were identified through the Polish Electronic System (PESEL), a database of all Polish residents, during case accrual. In total, 551 (79.3%) of 695 eligible cases and 1,925 (67.7%) of 2,843 eligible controls agreed to complete an in-person interview, and 88% of participating cases and 94% of participating controls donated blood. Controls with available blood DNA were randomly selected within age and study site strata for genetic studies. All cases and controls were considered to be of Caucasian ethnicity. Medical records of case participants were abstracted for diagnostic parameters. Case and control genotype data for stage 2 SNPs were extracted from existing Illumina 660W-Quad genome-wide scan data.

**SASBAC**

Details of the population selection process for the Singapore and Sweden Breast/Endometrial Cancer Study (SASBAC) have been published previously (11). Briefly, this population based case-control study was conducted among Swedish women aged 50-74 years, who were residing in Sweden between January 1st 1994 and December 31st 1995. Endometrial cancer cases were identified through the nation-wide cancer registries in Sweden. Controls, frequency-matched for age, were randomly selected from the Swedish Registry of Total Population. The study was restricted to postmenopausal women with an intact uterus and no previous diagnosis of endometrial cancer. All participants provided detailed questionnaire information. For endometrial cancer, histological specimens were reviewed and re-classified by the study pathologist. All participants reported Caucasian ethnicity. Genomic analyses were conducted by the Singapore node of the study.

**SECGS**

The Shanghai Endometrial Cancer Genetic Study (SECGS) includes 832 endometrial cancer cases who were recruited to the Shanghai Endometrial Cancer Study (SECS) and 2,049 controls who were recruited to the Shanghai Breast Cancer Study (SBCS). As described in detail elsewhere, both SECS and SBCS are two population-based case-control studies that were conducted in parallel in Shanghai during same period using an identical study protocol (12, 13). Briefly, 1199 women aged between 30 and 69 with newly diagnosed with EC 1997 and 2003 were identified through the population-based tumor registry and recruited to the SECS (response rate 83%). The SBCS controls were randomly selected from the general population using the Shanghai Resident Registry with response rate of 74%. Women with prior hysterectomies were not eligible for inclusion in this study. Participants completed a detailed in-person interview at the time of enrollment and provided a blood or buccal cell sample. Case and control genotype data for stage 2 SNPs, or for correlated SNPs with $R^2>0.8$, were extracted from existing Affymetrix 6.0 genome-wide scan data.

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**11. Weiderpass E, Adami HO, Baron JA, et al. Risk of endometrial cancer following estrogen replacement with and without progestins. J Natl Cancer Inst 1999; 91:1131-7.**
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## Study Collaborators

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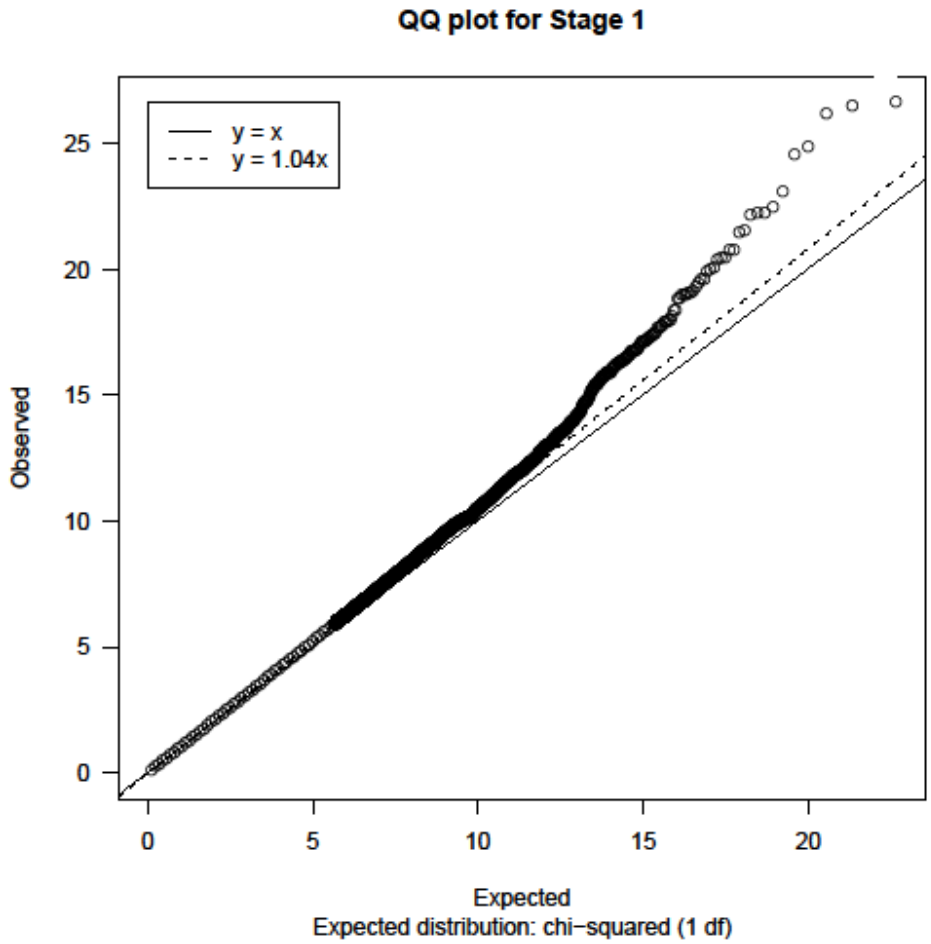
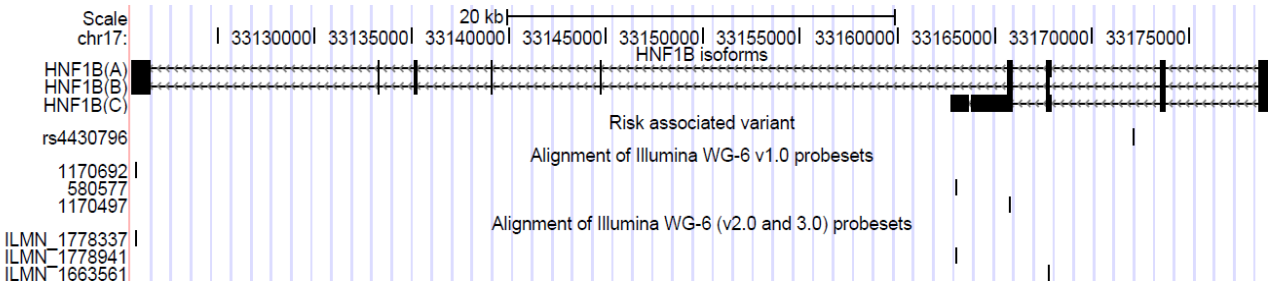
**Supplementary Figure 1: Quantile-quantile plot of the ranked trend test statistics.**
Y-value: observed test statistic (adjusted for the first three principal components – see methods).
X-value: expected statistic under the global null hypothesis of no association.
Markers denote the test statistics for each SNP. Line denotes the expectation under no association. SNPs which showed poor clustering or poor concordance between stages 1 and 2 are excluded from this plot (see methods).



**Supplementary Figure 2: Association of rs4430796 genotype with *HNF1B* gene expression.**

**A. Location of Illumina expression probe sequences in relation to *HNF1B* isoforms.**
Screenshot of UCSC Genome Browser showing *HNF1B* isoform A (NM_000458), isoform B (NM_001165923) and isoform C (NM_006481), the location of rs4430796, and the location of the Illumina expression probe sequences for *HNF1B* on 3 versions of the Human WG-6 array.




**B. Association of rs4430796 genotype with *HNF1B* expression assessed by Spearman rank analysis across three studies***

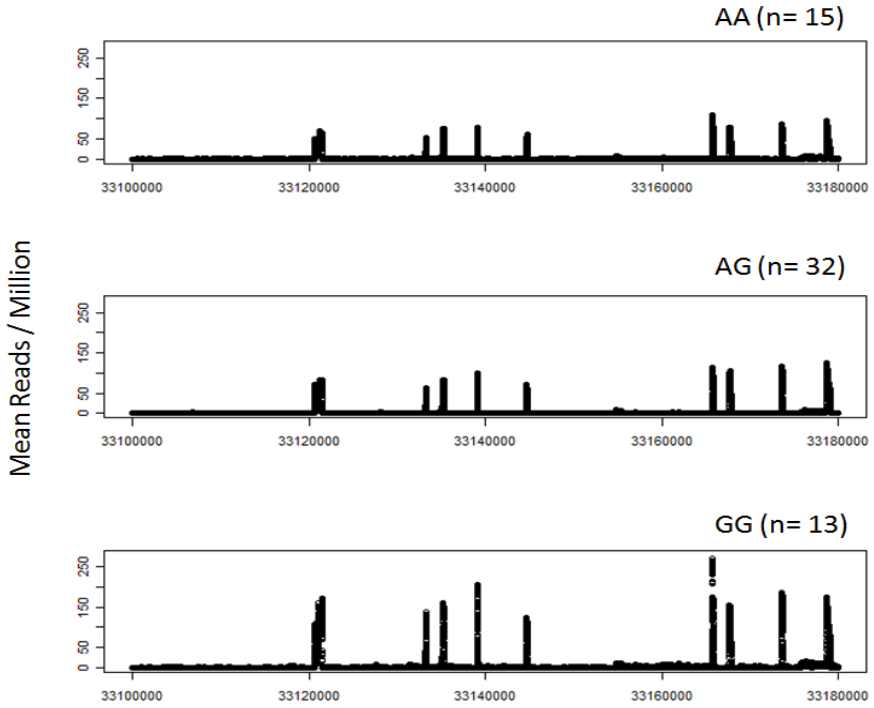
| Reference | Tissue source | Population group | Cohort size | Platform | Probe | rho | P-value |
|---|---|---|---|---|---|---|---|
| Stranger et al[1] | LCLs from peripheral blood | CEU | 60 | Illumina WG-6 v1 | 1170692 | 0.166 | 0.20 |
| | | | | | 580577 | 0.260 | **0.04** |
| | | | | | 1170497 | 0.179 | 0.18 |
| Dimas et al[2] | LCLs from umbilical cord blood | Western European | 75 | Illumina WG-6 v3 | ILMN_1778337 | -0.247 | **0.03** [a] |
| | | | | | ILMN_1778941 | ND | ND |
| | | | | | ILMN_1663561 | -0.291 | 0.11 |
| Dermitzakis et al[3] | LCLs from peripheral blood | CEU | 109 | Illumina WG-6 v2 | ILMN_1778337 | 0.218 | **0.02** |
| | | | | | ILMN_1778941 | ND | ND |
| | | | | | ILMN_1663561 | 0.051 | 0.60 |
| | | YRI | 108 | Illumina WG-6 v2 | ILMN_1778337 | 0.073 | 0.46 |
| | | | | | ILMN_1778941 | ND | ND |
| | | | | | ILMN_1663561 | 0.036 | 0.71 |

* There was known overlap of individuals between the studies of Stranger et al, and Dermitzakis et al: 56/60 CEU individuals assayed by Stranger et al were included in the analysis of 109 CEU individuals by Dermitzakis et al.

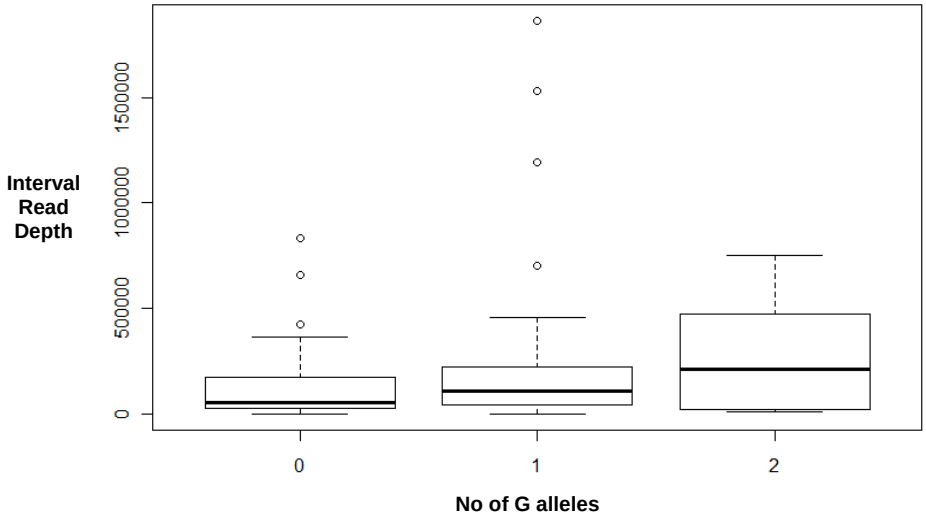
[1] Stranger BE, et al. Relative impact of nucleotide and copy number variation on gene expression phenotypes. Science. 2007;315:848–853.
[2] Dimas AS, et al. Common regulatory variation impacts gene expression in a cell type-dependent manner. Science. 2009;325:1246–1250.
[3] Dermitzakis, Montgomery, personal communication.
[a] This eQTL was not present in Fibroblasts or T-cells interrogated in the same study likely because the gene was lowly or not-expressed, with mean expression at background detection level in T-cells (6.55) and fibroblasts (6.70), compared to LCLs (8.24).

Abbreviations: LCL, lymphoblastoid cell lines; CEU, Utah residents with Northern and Western European ancestry from the Centre de'Etude du Polymorphism Humain (CEPH) collection; YRI, Yoruban in Ibadan, Nigeria; ND, no data.

**C. Association of rs4430796 genotype with *HNF1B* expression as assessed by RNA sequencing.** Expression of *HNF1B* was collected using RNA-Sequencing data from 60 CEU HapMap individuals, Caucasians of northern and western European ancestry living in Utah USA[1]. *(Note, a large subset of this sample of 60 individuals was assayed previously using Illumina expression array platforms: 43 individuals overlapped with the study by Stranger et al[2], and 55 individuals with the study by Dermitzakis et al[3].)* None of the three *HNF1B* isoforms or well-quantified exons (annotated using Gencode v3b) were significantly associated with rs4430796, however all 8 exon quantification of *HNF1B* expression by rs4430796 using Spearman rank tests showed some evidence of association (mean p-value 0.22, sd 0.11). Reduced power and increased difficulty in dissecting the relative contributions of individual transcripts may have prevented detection of a significant association. To further investigate possible associations, the mean reads/million across the interval containing *HNF1B* were stratified by genotypic class for the 60 individuals. There was a non-significant increase in read abundance in the interval of *HNF1B* conditioned on the G-allele (P=0.3).




Suggestion of a trend was also seen by analysis of a plot of interval read depth by genotype, with an increase in read depth with increasing number of G alleles.



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2. Stranger BE, et al. Relative impact of nucleotide and copy number variation on gene expression phenotypes. Science. 2007;315:848–853.
3. Dermitzakis, Montgomery, personal communication.

**Supplementary Table 1: Stage 1, stage 2 and combined results for 47 SNPs selected for replication.**

| | | | | Stage 1 | | | | Stage 2 | | | | Combined | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| SNP | Alleles | Chromosome | Position | cases/controls | MAF* | P (1df) | OR (95% CI) | cases/controls | MAF* | P (1df) | OR (95% CI) | P (1df) | OR (95% CI) |
| rs673604 | A/G | 1 | 35460402 | 1265/5188 | 0.08 | 6.11E-07 | 1.45 (1.25-1.67) | 3741/6151 | 0.09 | 4.10E-02 | 1.11 (1.00-1.23) | 5.88E-06 | 1.21 (1.12-1.32) |
| rs12117146 | G/A | 1 | 227871836 | 1264/5186 | 0.13 | 1.11E-05 | 1.32 (1.17-1.49) | 3743/6065 | 0.13 | 6.98E-01 | 1.02 (0.93-1.11) | 4.77E-03 | 1.11 (1.03-1.19) |
| rs1453194 | A/C | 2 | 19736284 | 1265/5187 | 0.12 | 2.26E-05 | 0.72 (0.61-0.84) | 3804/5864 | 0.12 | 7.85E-01 | 0.99 (0.90-1.08) | 1.93E-02 | 0.91 (0.84-0.99) |
| rs990270 | C/A | 2 | 40435853 | 1265/5189 | 0.39 | 7.13E-07 | 1.25 (1.15-1.37) | 3816/6217 | 0.43 | 2.31E-01 | 1.04 (0.98-1.10) | 1.80E-04 | 1.10 (1.05-1.15) |
| rs6544324 | G/A | 2 | 40436562 | 1263/5185 | 0.38 | 3.24E-05 | 1.21 (1.11-1.32) | 3798/5897 | 0.41 | 3.12E-01 | 1.03 (0.97-1.09) | 1.90E-03 | 1.08 (1.03-1.14) |
| rs1477004 | G/A | 2 | 40465576 | 1265/5187 | 0.31 | 4.63E-05 | 1.21 (1.10-1.33) | 3806/6255 | 0.34 | 7.05E-01 | 1.01 (0.95-1.08) | 1.11E-02 | 1.07 (1.02-1.12) |
| rs1124787 | A/C | 2 | 46168439 | 1264/5185 | 0.40 | 1.63E-04 | 0.84 (0.77-0.92) | 3815/6220 | 0.38 | 1.40E-02 | 1.08 (1.02-1.14) | 9.57E-01 | 1.00 (0.95-1.05) |
| rs10167555 | C/A | 2 | 46174610 | 1263/5177 | 0.20 | 5.39E-05 | 0.78 (0.70-0.88) | 3699/5900 | 0.18 | 1.50E-02 | 1.10 (1.02-1.18) | 8.93E-01 | 1.00 (0.93-1.06) |
| rs4954564 | A/G | 2 | 136611978 | 1265/5190 | 0.26 | 5.79E-03 | 1.15 (1.04-1.26) | 3778/6159 | 0.30 | 6.56E-01 | 1.02 (0.95-1.08) | 5.58E-02 | 1.05 (1.00-1.11) |
| rs4663132 | A/G | 2 | 234970071 | 1264/5189 | 0.45 | 1.25E-04 | 1.19 (1.09-1.30) | 3727/6121 | 0.55 | 6.22E-01 | 1.02 (0.96-1.08) | 1.12E-02 | 1.06 (1.01-1.12) |
| rs12489038 | A/C | 3 | 12986140 | 1265/5189 | 0.17 | 5.52E-04 | 1.22 (1.09-1.36) | 3799/6205 | 0.19 | 2.21E-01 | 1.05 (0.97-1.13) | 3.59E-03 | 1.10 (1.03-1.16) |
| rs720625 | A/G | 3 | 45785098 | 1264/5188 | 0.36 | 2.54E-05 | 1.21 (1.11-1.33) | 3757/6196 | 0.38 | 9.31E-01 | 1.00 (0.94-1.06) | 1.63E-02 | 1.06 (1.01-1.12) |
| rs798741 | G/A | 4 | 1682211 | 1265/5188 | 0.19 | 9.68E-06 | 1.27 (1.14-1.41) | 3755/6192 | 0.21 | 6.56E-01 | 1.02 (0.95-1.09) | 4.66E-03 | 1.09 (1.03-1.16) |
| rs1320071 | A/G | 4 | 7979091 | 1265/5179 | 0.39 | 1.56E-04 | 0.84 (0.76-0.92) | 3749/6161 | 0.38 | 4.42E-01 | 1.02 (0.96-1.09) | 1.53E-01 | 0.96 (0.92-1.01) |
| rs10008860 | G/A | 4 | 8020494 | 1265/5190 | 0.43 | 1.25E-05 | 1.22 (1.11-1.33) | 3812/6216 | 0.44 | 6.65E-01 | 1.01 (0.96-1.07) | 5.47E-03 | 1.07 (1.02-1.12) |
| rs6537310 | G/A | 4 | 145856182 | 1265/5187 | 0.44 | 7.67E-06 | 0.81 (0.74-0.89) | 3772/6129 | 0.42 | 2.76E-01 | 0.97 (0.91-1.03) | 8.05E-04 | 0.92 (0.88-0.97) |
| rs1425542 | A/G | 5 | 113902191 | 1264/5188 | 0.21 | 4.29E-05 | 1.24 (1.12-1.38) | 3119/4622 | 0.21 | 2.95E-01 | 0.96 (0.89-1.04) | 9.34E-02 | 1.06 (0.99-1.12) |
| rs2548975 | G/A | 5 | 133438999 | 1264/5189 | 0.19 | 2.07E-05 | 1.26 (1.13-1.40) | 3810/6211 | 0.20 | 9.81E-01 | 1.00 (0.93-1.08) | 1.80E-02 | 1.07 (1.01-1.14) |
| rs986359 | A/C | 5 | 136675003 | 1264/5183 | 0.36 | 4.59E-05 | 1.20 (1.10-1.32) | 3811/6211 | 0.37 | 9.38E-01 | 1.00 (0.94-1.06) | 2.46E-02 | 1.06 (1.01-1.11) |
| rs10515787 | G/A | 5 | 158407789 | 1265/5187 | 0.01 | 9.58E-04 | 1.75 (1.25-2.44) | 3774/6218 | 0.02 | 4.15E-01 | 1.10 (0.87-1.39) | 1.04E-02 | 1.28 (1.06-1.55) |
| rs2493046 | G/A | 6 | 488134 | 1264/5184 | 0.11 | 6.78E-05 | 1.31 (1.15-1.49) | 3816/6223 | 0.12 | 4.38E-01 | 1.04 (0.95-1.13) | 4.67E-03 | 1.11 (1.03-1.20) |
| rs4149366 | G/A | 6 | 2960488 | 1265/5189 | 0.07 | 4.29E-05 | 0.66 (0.55-0.81) | 2312/3779 | 0.07 | 1.20E-01 | 1.12 (0.97-1.30) | 2.28E-01 | 0.93 (0.83-1.05) |
| rs559901 | G/A | 6 | 153899629 | 1264/5187 | 0.28 | 6.97E-05 | 1.21 (1.10-1.33) | 3767/5902 | 0.30 | 5.28E-01 | 1.02 (0.96-1.09) | 6.56E-03 | 1.08 (1.02-1.14) |
| rs2049449 | A/C | 7 | 14665378 | 1265/5188 | 0.14 | 6.63E-05 | 1.28 (1.13-1.44) | 3810/6225 | 0.15 | 2.06E-01 | 1.05 (0.97-1.14) | 1.09E-03 | 1.12 (1.04-1.19) |
| rs9969635 | A/C | 8 | 10369046 | 1265/5188 | 0.10 | 3.06E-05 | 1.33 (1.17-1.53) | 3288/5295 | 0.11 | 3.83E-01 | 1.05 (0.95-1.15) | 1.59E-03 | 1.14 (1.05-1.23) |
| rs11784168 | A/G | 8 | 28083277 | 1264/5187 | 0.13 | 4.38E-04 | 1.25 (1.10-1.41) | 3811/6218 | 0.15 | 6.30E-02 | 0.92 (0.85-1.00) | 6.66E-01 | 1.02 (0.95-1.09) |
| rs17589981 | A/G | 9 | 9109229 | 1265/5184 | 0.32 | 1.32E-04 | 1.20 (1.09-1.31) | 3805/6187 | 0.33 | 5.65E-01 | 0.98 (0.92-1.04) | 1.04E-01 | 1.04 (0.99-1.10) |
| rs4295736 | G/A | 9 | 81072159 | 1263/5188 | 0.06 | 6.27E-05 | 1.42 (1.19-1.68) | 3816/6189 | 0.07 | 7.31E-01 | 0.98 (0.87-1.10) | 4.61E-02 | 1.10 (1.00-1.22) |
| rs11138152 | G/A | 9 | 81156461 | 1265/5188 | 0.08 | 7.38E-06 | 1.40 (1.21-1.62) | 3762/6204 | 0.09 | 1.39E-01 | 0.93 (0.84-1.03) | 1.84E-01 | 1.06 (0.97-1.15) |
| rs1932658 | A/G | 9 | 86005570 | 1258/5167 | 0.26 | 8.54E-05 | 1.22 (1.10-1.34) | 3775/6089 | 0.28 | 7.45E-01 | 0.99 (0.93-1.06) | 6.04E-02 | 1.05 (1.00-1.11) |
| rs7358426 | A/G | 11 | 22079689 | 1265/5189 | 0.31 | 4.09E-05 | 0.81 (0.74-0.90) | 3766/6197 | 0.31 | 1.35E-01 | 0.95 (0.89-1.02) | 5.65E-04 | 0.91 (0.86-0.96) |
| rs521319 | G/A | 11 | 63928607 | 1265/5188 | 0.30 | 2.92E-04 | 1.19 (1.08-1.30) | 3805/6216 | 0.32 | 2.13E-01 | 0.96 (0.90-1.02) | 3.38E-01 | 1.03 (0.97-1.08) |
| rs2238136 | G/A | 12 | 46563980 | 1265/5188 | 0.25 | 6.01E-06 | 1.25 (1.14-1.38) | 3814/6224 | 0.27 | 7.91E-01 | 0.99 (0.93-1.06) | 2.25E-02 | 1.07 (1.01-1.12) |
| rs7132324 | G/A | 12 | 46593576 | 1264/5187 | 0.33 | 5.90E-05 | 1.21 (1.10-1.33) | 3744/6186 | 0.34 | 4.87E-01 | 1.02 (0.96-1.09) | 5.06E-03 | 1.08 (1.02-1.13) |
| rs10870680 | G/A | 13 | 19638681 | 1265/5186 | 0.20 | 2.68E-04 | 0.81 (0.72-0.90) | 3815/6221 | 0.19 | 2.15E-01 | 0.95 (0.89-1.03) | 2.50E-03 | 0.91 (0.85-0.97) |
| rs11846930 | G/A | 14 | 61627081 | 1264/5187 | 0.07 | 8.25E-05 | 1.38 (1.18-1.63) | 3801/6229 | 0.07 | 7.50E-01 | 1.02 (0.91-1.14) | 1.25E-02 | 1.13 (1.03-1.23) |
| rs2123458 | A/G | 14 | 64128910 | 1265/5178 | 0.29 | 8.54E-05 | 1.21 (1.10-1.33) | 3516/5519 | 0.30 | 6.07E-01 | 1.02 (0.95-1.09) | 7.41E-03 | 1.08 (1.02-1.14) |
| rs1568221 | G/A | 15 | 34434869 | 1265/5187 | 0.33 | 3.01E-05 | 1.21 (1.11-1.33) | 3811/6220 | 0.35 | 1.07E-01 | 1.05 (0.99-1.12) | 2.36E-04 | 1.10 (1.04-1.15) |
| rs7191629 | A/G | 16 | 5689444 | 1265/5185 | 0.27 | 5.88E-05 | 1.22 (1.11-1.34) | 3641/6074 | 0.28 | 5.18E-01 | 1.02 (0.96-1.09) | 4.88E-03 | 1.08 (1.02-1.14) |
| rs4782024 | G/A | 16 | 17166439 | 1265/5185 | 0.04 | 1.18E-04 | 0.55 (0.41-0.75) | 3770/5848 | 0.03 | 2.02E-01 | 1.11 (0.95-1.31) | 4.80E-01 | 0.95 (0.82-1.10) |
| rs4430796 | A/G | 17 | 33172153 | 1262/5179 | 0.48 | 3.06E-07 | 0.79 (0.73-0.87) | 2332/4349 | 0.48 | 2.00E-04 | 0.87 (0.81-0.94) | 7.11E-10 | 0.84 (0.79-0.89) |
| rs4239217 | A/G | 17 | 33173100 | 1265/5190 | 0.41 | 2.48E-07 | 0.79 (0.72-0.86) | 2342/4307 | 0.40 | 2.00E-03 | 0.89 (0.82-0.96) | 1.19E-08 | 0.84 (0.80-0.90) |
| rs7501939 | G/A | 17 | 33175269 | 1263/5187 | 0.40 | 2.17E-06 | 0.80 (0.73-0.88) | 3337/5234 | 0.40 | 1.00E-03 | 0.90 (0.84-0.96) | 5.35E-08 | 0.86 (0.82-0.91) |
| rs1212721 | G/A | 18 | 5245109 | 1264/5183 | 0.21 | 1.27E-04 | 0.80 (0.72-0.90) | 3760/6195 | 0.20 | 9.73E-01 | 1.00 (0.93-1.08) | 4.23E-02 | 0.94 (0.88-1.00) |
| rs364418 | G/A | 18 | 7803239 | 1264/5190 | 0.39 | 4.61E-05 | 1.20 (1.10-1.32) | 3770/6207 | 0.43 | 5.03E-01 | 1.02 (0.96-1.08) | 5.51E-03 | 1.07 (1.02-1.13) |
| rs918171 | G/A | 19 | 3287539 | 1265/5188 | 0.07 | 1.41E-04 | 1.36 (1.16-1.59) | 3769/6208 | 0.07 | 1.70E-02 | 1.14 (1.03-1.28) | 3.70E-05 | 1.21 (1.11-1.32) |
| rs1557203 | A/G | 20 | 33627877 | 1265/5187 | 0.07 | 1.53E-04 | 1.36 (1.16-1.60) | 3817/6224 | 0.08 | 2.86E-01 | 1.06 (0.95-1.17) | 2.92E-03 | 1.14 (1.05-1.24) |

* MAF=minor allele frequency in controls

*NB: Not all of the p-values for Stage 1 are less than 1E-04. This is because Stage 2 SNP selection was based on a slightly earlier version of the dataset and was not corrected by PCA.*

| **Supplementary Table 2: Endometrial cancer sample sets included in Stage 1 and Stage 2 analyses.*** | | | | | |
|---|---|---|---|---|---|
| | | | | | |
| **Study** | **Abbreviation** | **General Setting** | **Cases** | **Controls** | **Genotyping platform**** |
| | | | | | |
| **Stage 1 Sample Sets** | | | | | |
| Australian National Endometrial Cancer Study | ANECS | Australia; population based case-control study | 599 | - | Illumina 610K |
| Study of Epidemiology and Risk Factors in Cancer Heredity | SEARCH | England; population based case-control study | 666 | - | Illumina 610K |
| Wellcome Trust Case-Control Consortium | WTCCC | UK: sample from 1958 Birth Cohort and UK Blood Donors from NBS | - | 5,190 | Illumina 1.2M |
| | | | | | |
| **Stage 2 Sample Sets** | | | | | |
| | | | | | |
| Australian National Endometrial Cancer Study*** | ANECS | Australia; population based case-control study | 607 | 587 | Sequenom iPLEX |
| Australian Red Cross Blood Service*** | ARCBS | Brisbane, Australia; female donor controls | | 577 | Sequenom iPLEX |
| Newcastle Endometrial Cancer Study*** | NECS | Newcastle, Australia; hospital-based cases | 194 | - | Sequenom iPLEX |
| Study of Epidemiology and Risk Factors in Cancer Heredity | SEARCH | England; population based case-control study | 688 | 1,600 | ABI Fluidigm |
| Bavarian Endometrial Cancer Study | BECS | Germany; population based case-control study | 222 | 396 | Sequenom iPLEX |
| Leuven Endometrial Cancer Study | LES | Belgium; hospital based case-control study | 238 | 671 | Sequenom iPLEX |
| Molecular Markers in Treatment of Endometrial Cancer | MoMaTEC | Norway; population based case-control study | 213 | 138 | Sequenom iPLEX |
| National Study of the Genetics of Endometrial Cancer | NSECG | United Kingdom; population based case-control study | 829 | 998 | Sequenom iPLEX (cases); Illumina 550K or Kaspar for selected SNPs (controls) |
| Polish Endometrial Cancer Study | PECS | Poland, Warsaw and Lodz; population based case-control study | 466 | 897 | Illumina 660W-Quad |
| Singapore and Swedish Breast/Endometrial Cancer Study | SASBAC | Sweden; population based cohort study | 500 | 1,022 | Sequenom iPLEX |
| Shanghai Endometrial Cancer Genetic Study | SECS | Shanghai, China; population based case-control studies | 832 | 2,049 | Affymetrix 6.0 |
| ***Total*** | | | ***6,054*** | ***14,125*** | |
| | | | | | |

*The number of cases and controls represents the maximum number of genotypes from cases and controls of reported Caucasian ethnicity, with the exception of SECS samples of Chinese ancestry.

**Fluidigm genotyping was performed in Cambridge using primers and probes supplied by Applied Biosystems as Assays-By-Design. Sequenom's MassARRAY system and iPLEX technology (Sequenom Inc, San Diego, CA) was used for genotyping for most other studies, using matrix-assisted laser desorption/ionization time of flight mass spectrometry for the determination of allele-specific primer extension products. Oligonucleotides were designed using MassARRAY Assay Design software (version 3.1, Sequenom Inc) in Brisbane for use at other sites undertaking iplex genotyping (Leuven – LES, Singapore Genome Institute – SASBAC), and results were analysed using TYPER software (version 3.4, Sequenom Inc).

***Australian stage 2 samples considered as a single group for statistical analysis

**Supplementary Table 3: Stage 1, stage 2 (endometrioid histology cases only) and combined results for 47 SNPs selected for replication.**

| | | | | Stage 1 | | | | Stage 2: endometrioid histology only | | | | Combined: endometrioid histology only | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| SNP | Alleles | Chromosome | Position | cases/controls | MAF* | P (1df) | OR (95% CI) | cases/controls | MAF* | P (1df) | OR (95% CI) | P (1df) | OR (95% CI) |
| rs673604 | A/G | 1 | 35460402 | 1265/5188 | 0.08 | 6.11E-07 | 1.45 (1.25-1.67) | 2948/6151 | 0.09 | 4.90E-02 | 1.12 (1.00-1.25) | 4.58E-06 | 1.23 (1.12-1.34) |
| rs12117146 | G/A | 1 | 227871836 | 1264/5186 | 0.13 | 1.11E-05 | 1.32 (1.17-1.49) | 2951/6065 | 0.13 | 6.93E-01 | 1.02 (0.93-1.12) | 3.17E-03 | 1.12 (1.04-1.20) |
| rs1453194 | A/C | 2 | 19736284 | 1265/5187 | 0.12 | 2.26E-05 | 0.72 (0.61-0.84) | 2977/5864 | 0.12 | 5.93E-01 | 0.97 (0.89-1.07) | 7.19E-03 | 0.89 (0.82-0.97) |
| rs990270 | C/A | 2 | 40435853 | 1265/5189 | 0.39 | 7.13E-07 | 1.25 (1.15-1.37) | 2989/6217 | 0.43 | 2.22E-01 | 1.04 (0.98-1.11) | 9.76E-05 | 1.11 (1.05-1.17) |
| rs6544324 | G/A | 2 | 40436562 | 1263/5185 | 0.38 | 3.24E-05 | 1.21 (1.11-1.32) | 2975/5897 | 0.42 | 3.30E-01 | 1.03 (0.97-1.10) | 1.46E-03 | 1.09 (1.03-1.15) |
| rs1477004 | G/A | 2 | 40465576 | 1265/5187 | 0.31 | 4.63E-05 | 1.21 (1.10-1.33) | 2977/6255 | 0.34 | 6.70E-01 | 1.01 (0.95-1.08) | 6.95E-03 | 1.08 (1.02-1.14) |
| rs1124787 | A/C | 2 | 46168439 | 1264/5185 | 0.40 | 1.63E-04 | 0.84 (0.77-0.92) | 2985/6220 | 0.38 | 2.70E-02 | 1.08 (1.01-1.15) | 6.72E-01 | 0.99 (0.94-1.04) |
| rs10167555 | C/A | 2 | 46174610 | 1263/5177 | 0.20 | 5.39E-05 | 0.78 (0.70-0.88) | 2907/5900 | 0.19 | 5.90E-02 | 1.08 (1.00-1.17) | 4.53E-01 | 0.97 (0.91-1.04) |
| rs4954564 | A/G | 2 | 136611978 | 1265/5190 | 0.26 | 5.79E-03 | 1.15 (1.04-1.26) | 2955/6159 | 0.30 | 6.79E-01 | 1.02 (0.95-1.09) | 4.93E-02 | 1.06 (1.00-1.12) |
| rs4663132 | A/G | 2 | 234970071 | 1264/5189 | 0.45 | 1.25E-04 | 1.19 (1.09-1.30) | 2927/6121 | 0.55 | 7.89E-01 | 1.01 (0.95-1.07) | 1.39E-02 | 1.07 (1.01-1.12) |
| rs12489038 | A/C | 3 | 12986140 | 1265/5189 | 0.17 | 5.52E-04 | 1.22 (1.09-1.36) | 2970/6205 | 0.19 | 3.62E-01 | 1.04 (0.96-1.12) | 6.21E-03 | 1.09 (1.03-1.17) |
| rs720625 | A/G | 3 | 45785098 | 1264/5188 | 0.36 | 2.54E-05 | 1.21 (1.11-1.33) | 2951/6196 | 0.38 | 7.30E-01 | 1.01 (0.95-1.08) | 6.14E-03 | 1.08 (1.02-1.13) |
| rs798741 | G/A | 4 | 1682211 | 1265/5188 | 0.19 | 9.68E-06 | 1.27 (1.14-1.41) | 2952/6192 | 0.21 | 8.43E-01 | 0.99 (0.92-1.07) | 1.46E-02 | 1.08 (1.02-1.15) |
| rs1320071 | A/G | 4 | 7979091 | 1265/5179 | 0.39 | 1.56E-04 | 0.84 (0.76-0.92) | 2946/6161 | 0.38 | 9.64E-01 | 1.00 (0.94-1.07) | 3.12E-02 | 0.94 (0.89-0.99) |
| rs10008860 | G/A | 4 | 8020494 | 1265/5190 | 0.43 | 1.25E-05 | 1.22 (1.11-1.33) | 2984/6216 | 0.44 | 4.89E-01 | 1.02 (0.96-1.09) | 1.85E-03 | 1.09 (1.03-1.14) |
| rs6537310 | G/A | 4 | 145856182 | 1265/5187 | 0.44 | 7.67E-06 | 0.81 (0.74-0.89) | 2949/6129 | 0.42 | 9.50E-02 | 0.95 (0.89-1.01) | 7.96E-05 | 0.90 (0.86-0.95) |
| rs1425542 | A/G | 5 | 113902191 | 1264/5188 | 0.21 | 4.29E-05 | 1.24 (1.12-1.38) | 2434/4622 | 0.21 | 4.80E-01 | 0.97 (0.89-1.06) | 3.64E-02 | 1.07 (1.00-1.15) |
| rs2548975 | G/A | 5 | 133438999 | 1264/5189 | 0.19 | 2.07E-05 | 1.26 (1.13-1.40) | 2984/6211 | 0.20 | 9.76E-01 | 1.00 (0.92-1.08) | 1.41E-02 | 1.08 (1.02-1.15) |
| rs986359 | A/C | 5 | 136675003 | 1264/5183 | 0.36 | 4.59E-05 | 1.20 (1.10-1.32) | 2982/6211 | 0.37 | 8.63E-01 | 0.99 (0.93-1.06) | 2.11E-02 | 1.06 (1.01-1.12) |
| rs10515787 | G/A | 5 | 158407789 | 1265/5187 | 0.01 | 9.58E-04 | 1.75 (1.25-2.44) | 2965/6218 | 0.02 | 2.42E-01 | 1.16 (0.91-1.49) | 3.53E-03 | 1.34 (1.10-1.64) |
| rs2493046 | G/A | 6 | 488134 | 1264/5184 | 0.11 | 6.78E-05 | 1.31 (1.15-1.49) | 2988/6223 | 0.12 | 3.06E-01 | 1.05 (0.96-1.16) | 1.75E-03 | 1.13 (1.05-1.22) |
| rs4149366 | G/A | 6 | 2960488 | 1265/5189 | 0.07 | 4.29E-05 | 0.66 (0.55-0.81) | 2435/3779 | 0.07 | 2.18E-01 | 1.11 (0.94-1.30) | 1.00E-01 | 0.90 (0.80-1.02) |
| rs559901 | G/A | 6 | 153899629 | 1264/5187 | 0.28 | 6.97E-05 | 1.21 (1.10-1.33) | 2963/5902 | 0.30 | 4.62E-01 | 1.03 (0.96-1.10) | 3.65E-03 | 1.09 (1.03-1.15) |
| rs2049449 | A/C | 7 | 14665378 | 1265/5188 | 0.14 | 6.63E-05 | 1.28 (1.13-1.44) | 2981/6225 | 0.16 | 3.32E-01 | 1.04 (0.96-1.14) | 1.77E-03 | 1.12 (1.04-1.20) |
| rs9969635 | A/C | 8 | 10369046 | 1265/5188 | 0.10 | 3.06E-05 | 1.33 (1.17-1.53) | 2587/5423 | 0.11 | 5.41E-01 | 1.03 (0.93-1.15) | 2.20E-03 | 1.14 (1.05-1.24) |
| rs11784168 | A/G | 8 | 28083277 | 1264/5187 | 0.13 | 4.38E-04 | 1.25 (1.10-1.41) | 2984/6218 | 0.14 | 1.18E-01 | 0.93 (0.85-1.02) | 4.16E-01 | 1.03 (0.96-1.11) |
| rs17589981 | A/G | 9 | 9109229 | 1265/5184 | 0.32 | 1.32E-04 | 1.20 (1.09-1.31) | 2977/6187 | 0.33 | 4.52E-01 | 0.98 (0.91-1.04) | 1.05E-01 | 1.05 (0.99-1.10) |
| rs4295736 | G/A | 9 | 81072159 | 1263/5188 | 0.06 | 6.27E-05 | 1.42 (1.19-1.68) | 2988/6189 | 0.07 | 5.10E-01 | 0.96 (0.84-1.09) | 5.94E-02 | 1.10 (1.00-1.22) |
| rs11138152 | G/A | 9 | 81156461 | 1265/5188 | 0.08 | 7.38E-06 | 1.40 (1.21-1.62) | 2955/6204 | 0.09 | 1.02E-01 | 0.91 (0.82-1.02) | 1.65E-01 | 1.06 (0.97-1.16) |
| rs1932658 | A/G | 9 | 86005570 | 1258/5167 | 0.26 | 8.54E-05 | 1.22 (1.10-1.34) | 2951/6089 | 0.28 | 5.61E-01 | 1.02 (0.95-1.09) | 6.17E-03 | 1.08 (1.02-1.15) |
| rs7358426 | A/G | 11 | 22079689 | 1265/5189 | 0.31 | 4.09E-05 | 0.81 (0.74-0.90) | 2958/6197 | 0.30 | 1.53E-01 | 0.95 (0.89-1.02) | 4.84E-04 | 0.90 (0.85-0.96) |
| rs521319 | G/A | 11 | 63928607 | 1265/5188 | 0.30 | 2.92E-04 | 1.19 (1.08-1.30) | 2985/6216 | 0.32 | 4.89E-01 | 0.98 (0.91-1.05) | 1.23E-01 | 1.04 (0.99-1.10) |
| rs2238136 | G/A | 12 | 46563980 | 1265/5188 | 0.25 | 6.01E-06 | 1.25 (1.14-1.38) | 2986/6224 | 0.27 | 6.45E-01 | 1.02 (0.95-1.09) | 2.55E-03 | 1.09 (1.03-1.16) |
| rs7132324 | G/A | 12 | 46593576 | 1264/5187 | 0.33 | 5.90E-05 | 1.21 (1.10-1.33) | 2947/6186 | 0.34 | 4.19E-01 | 1.03 (0.96-1.10) | 2.66E-03 | 1.09 (1.03-1.15) |
| rs10870680 | G/A | 13 | 19638681 | 1265/5186 | 0.20 | 2.68E-04 | 0.81 (0.72-0.90) | 2985/6221 | 0.19 | 5.99E-01 | 0.98 (0.90-1.06) | 1.17E-02 | 0.92 (0.86-0.98) |
| rs11846930 | G/A | 14 | 61627081 | 1264/5187 | 0.07 | 8.25E-05 | 1.38 (1.18-1.63) | 2976/6229 | 0.07 | 9.03E-01 | 1.01 (0.89-1.14) | 1.39E-02 | 1.13 (1.03-1.25) |
| rs2123458 | A/G | 14 | 64128910 | 1265/5178 | 0.29 | 8.54E-05 | 1.21 (1.10-1.33) | 2770/5519 | 0.30 | 3.74E-01 | 1.03 (0.96-1.11) | 2.07E-03 | 1.09 (1.03-1.16) |
| rs1568221 | G/A | 15 | 34434869 | 1265/5187 | 0.33 | 3.01E-05 | 1.21 (1.11-1.33) | 2984/6220 | 0.35 | 1.01E-01 | 1.06 (0.99-1.13) | 1.47E-04 | 1.11 (1.05-1.17) |
| rs7191629 | A/G | 16 | 5689444 | 1265/5185 | 0.27 | 5.88E-05 | 1.22 (1.11-1.34) | 2880/6074 | 0.28 | 7.02E-01 | 1.01 (0.94-1.09) | 6.72E-03 | 1.08 (1.02-1.15) |
| rs4782024 | G/A | 16 | 17166439 | 1265/5185 | 0.04 | 1.18E-04 | 0.55 (0.41-0.75) | 2962/5848 | 0.03 | 3.55E-01 | 1.09 (0.91-1.30) | 2.47E-01 | 0.91 (0.79-1.06) |
| rs4430796 | A/G | 17 | 33172153 | 1262/5179 | 0.48 | 3.06E-07 | 0.79 (0.73-0.87) | 1786/4349 | 0.47 | 2.56E-05 | 0.84 (0.77-0.91) | 4.28E-11 | 0.82 (0.77-0.87) |
| rs4239217 | A/G | 17 | 33173100 | 1265/5190 | 0.41 | 2.48E-07 | 0.79 (0.72-0.86) | 1753/4307 | 0.39 | 5.90E-04 | 0.86 (0.79-0.94) | 1.27E-09 | 0.83 (0.78-0.88) |
| rs7501939 | G/A | 17 | 33175269 | 1263/5187 | 0.40 | 2.17E-06 | 0.80 (0.73-0.88) | 2562/5234 | 0.38 | 3.30E-04 | 0.88 (0.82-0.94) | 7.57E-09 | 0.85 (0.80-0.90) |
| rs1212721 | G/A | 18 | 5245109 | 1264/5183 | 0.21 | 1.27E-04 | 0.80 (0.72-0.90) | 2953/6195 | 0.20 | 8.67E-01 | 1.01 (0.93-1.09) | 4.14E-02 | 0.93 (0.88-1.00) |
| rs364418 | G/A | 18 | 7803239 | 1264/5190 | 0.39 | 4.61E-05 | 1.20 (1.10-1.32) | 2962/6207 | 0.43 | 8.09E-01 | 0.99 (0.93-1.06) | 3.17E-02 | 1.06 (1.01-1.12) |
| rs918171 | G/A | 19 | 3287539 | 1265/5188 | 0.07 | 1.41E-04 | 1.36 (1.16-1.59) | 2960/6208 | 0.07 | 7.10E-02 | 1.12 (0.99-1.26) | 1.94E-04 | 1.20 (1.09-1.32) |
| rs1557203 | A/G | 20 | 33627877 | 1265/5187 | 0.07 | 1.53E-04 | 1.36 (1.16-1.60) | 2989/6224 | 0.08 | 3.24E-01 | 1.06 (0.95-1.19) | 2.62E-03 | 1.15 (1.05-1.26) |

* MAF=minor allele frequency in controls

*NB: All stage 1 cases were chosen on the basis of endometrioid histology*

*NB: Not all of the p-values for Stage 1 are less than 1E-04. This is because Stage 2 SNP selection was based on a slightly earlier version of the dataset and was not corrected by PCA.*

**Supplementary Table 4: Stage 2 results for 47 SNPs selected for replication, by histology**

| | | | | | endometrioid only (case control) | | | non-endometrioid only (case control) | | | non-endometrioid vs endometrioid (case only) | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| SNP | Alleles | Chromosome | Position | Nctrls | Ncases | P (1df) | OR (95% CI) | Ncases | P (1df) | OR (95% CI) | P (1df) | OR (95% CI) |
| rs673604 | A/G | 1 | 35460402 | 6151 | 2948 | **4.90E-02** | 1.12 (1.00-1.25) | 697 | 5.04E-01 | 1.07 (0.88-1.30) | 8.57E-01 | 0.98 (0.80-1.21) |
| rs12117146 | G/A | 1 | 227871836 | 6065 | 2951 | 6.93E-01 | 1.02 (0.93-1.12) | 677 | 6.44E-01 | 1.04 (0.88-1.23) | 8.07E-01 | 1.02 (0.86-1.22) |
| rs1453194 | A/C | 2 | 19736284 | 5864 | 2977 | 5.93E-01 | 0.97 (0.89-1.07) | 711 | 5.25E-01 | 1.06 (0.89-1.25) | 7.71E-01 | 1.03 (0.86-1.23) |
| rs990270 | C/A | 2 | 40435853 | 6217 | 2989 | 2.22E-01 | 1.04 (0.98-1.11) | 711 | 2.34E-01 | 1.07 (0.96-1.20) | 8.79E-01 | 1.01 (0.90-1.14) |
| rs6544324 | G/A | 2 | 40436562 | 5897 | 2975 | 3.30E-01 | 1.03 (0.97-1.10) | 708 | 2.64E-01 | 1.07 (0.95-1.19) | 9.04E-01 | 1.01 (0.89-1.14) |
| rs1477004 | G/A | 2 | 40465576 | 6255 | 2977 | 6.70E-01 | 1.01 (0.95-1.08) | 714 | 5.99E-01 | 1.03 (0.92-1.16) | 6.50E-01 | 0.97 (0.86-1.10) |
| rs1124787 | A/C | 2 | 46168439 | 6220 | 2985 | **2.70E-02** | 1.08 (1.01-1.15) | 714 | 2.34E-01 | 1.07 (0.96-1.20) | 7.69E-01 | 0.98 (0.87-1.11) |
| rs10167555 | C/A | 2 | 46174610 | 5900 | 2907 | 5.90E-02 | 1.08 (1.00-1.17) | 683 | **7.00E-03** | 1.22 (1.06-1.40) | 1.61E-01 | 1.11 (0.96-1.29) |
| rs4954564 | A/G | 2 | 136611978 | 6159 | 2955 | 6.79E-01 | 1.02 (0.95-1.09) | 707 | 5.59E-01 | 1.04 (0.92-1.17) | 6.69E-01 | 0.97 (0.85-1.11) |
| rs4663132 | A/G | 2 | 234970071 | 6121 | 2927 | 7.89E-01 | 1.01 (0.95-1.07) | 690 | 4.56E-01 | 1.04 (0.93-1.17) | 3.53E-01 | 1.06 (0.94-1.20) |
| rs12489038 | A/C | 3 | 12986140 | 6205 | 2970 | 3.62E-01 | 1.04 (0.96-1.12) | 713 | 5.71E-01 | 1.04 (0.90-1.20) | 6.00E-01 | 1.04 (0.89-1.22) |
| rs720625 | A/G | 3 | 45785098 | 6196 | 2951 | 7.30E-01 | 1.01 (0.95-1.08) | 695 | 4.79E-01 | 0.96 (0.85-1.08) | 3.63E-01 | 0.94 (0.83-1.07) |
| rs798741 | G/A | 4 | 1682211 | 6192 | 2952 | 8.43E-01 | 0.99 (0.92-1.07) | 692 | 3.59E-01 | 1.07 (0.93-1.22) | 2.74E-01 | 1.09 (0.94-1.26) |
| rs1320071 | A/G | 4 | 7979091 | 6161 | 2946 | 9.64E-01 | 1.00 (0.94-1.07) | 694 | 1.11E-01 | 1.10 (0.98-1.23) | 1.21E-01 | 1.10 (0.97-1.25) |
| rs10008860 | G/A | 4 | 8020494 | 6216 | 2984 | 4.89E-01 | 1.02 (0.96-1.09) | 712 | 7.71E-01 | 0.98 (0.88-1.10) | 6.68E-01 | 0.97 (0.86-1.10) |
| rs6537310 | G/A | 4 | 145856182 | 6129 | 2949 | 9.50E-02 | 0.95 (0.89-1.01) | 708 | 3.37E-01 | 1.06 (0.95-1.18) | **1.30E-02** | 1.17 (1.03-1.32) |
| rs1425542 | A/G | 5 | 113902191 | 4622 | 2434 | 4.80E-01 | 0.97 (0.89-1.06) | 569 | 5.30E-01 | 0.95 (0.81-1.11) | 8.10E-01 | 0.98 (0.83-1.16) |
| rs2548975 | G/A | 5 | 133438999 | 6211 | 2984 | 9.76E-01 | 1.00 (0.92-1.08) | 710 | 6.51E-01 | 1.03 (0.90-1.18) | 3.98E-01 | 1.07 (0.92-1.23) |
| rs986359 | A/C | 5 | 136675003 | 6211 | 2982 | 8.63E-01 | 0.99 (0.93-1.06) | 714 | 9.61E-01 | 1.00 (0.89-1.12) | 8.02E-01 | 1.02 (0.90-1.15) |
| rs10515787 | G/A | 5 | 158407789 | 6218 | 2965 | 2.42E-01 | 1.16 (0.91-1.49) | 698 | 8.81E-01 | 1.04 (0.65-1.64) | 2.83E-01 | 0.77 (0.48-1.24) |
| rs2493046 | G/A | 6 | 488134 | 6223 | 2988 | 3.06E-01 | 1.05 (0.96-1.16) | 712 | 8.36E-01 | 1.02 (0.86-1.21) | 5.81E-01 | 0.95 (0.79-1.14) |
| rs4149366 | G/A | 6 | 2960488 | 3779 | 2435 | 2.18E-01 | 1.11 (0.94-1.30) | 566 | 1.30E-01 | 1.22 (0.94-1.57) | 5.07E-01 | 1.09 (0.84-1.41) |
| rs559901 | G/A | 6 | 153899629 | 5902 | 2963 | 4.62E-01 | 1.03 (0.96-1.10) | 695 | 5.04E-01 | 0.96 (0.84-1.09) | 5.14E-01 | 0.96 (0.84-1.09) |
| rs2049449 | A/C | 7 | 14665378 | 6225 | 2981 | 3.32E-01 | 1.04 (0.96-1.14) | 713 | 1.08E-01 | 1.13 (0.97-1.31) | 4.13E-01 | 1.07 (0.91-1.25) |
| rs9969635 | A/C | 8 | 10369046 | 5423 | 2587 | 5.41E-01 | 1.03 (0.93-1.15) | 598 | 5.78E-01 | 1.06 (0.87-1.28) | 7.48E-01 | 0.97 (0.79-1.19) |
| rs11784168 | A/G | 8 | 28083277 | 6218 | 2984 | 1.18E-01 | 0.93 (0.85-1.02) | 712 | 4.60E-01 | 0.94 (0.80-1.11) | 6.71E-01 | 0.96 (0.81-1.15) |
| rs17589981 | A/G | 9 | 9109229 | 6187 | 2977 | 4.52E-01 | 0.98 (0.91-1.04) | 712 | 6.27E-01 | 1.03 (0.92-1.16) | 2.96E-01 | 1.07 (0.94-1.21) |
| rs4295736 | G/A | 9 | 81072159 | 6189 | 2988 | 5.10E-01 | 0.96 (0.84-1.09) | 713 | 9.28E-01 | 1.01 (0.80-1.27) | 6.04E-01 | 1.07 (0.84-1.36) |
| rs11138152 | G/A | 9 | 81156461 | 6204 | 2955 | 1.02E-01 | 0.91 (0.82-1.02) | 697 | 5.37E-01 | 0.94 (0.77-1.15) | 9.77E-01 | 1.00 (0.81-1.25) |
| rs1932658 | A/G | 9 | 86005570 | 6089 | 2951 | 5.61E-01 | 1.02 (0.95-1.09) | 708 | 7.50E-02 | 0.89 (0.79-1.01) | **1.50E-02** | 0.85 (0.74-0.97) |
| rs7358426 | A/G | 11 | 22079689 | 6197 | 2958 | 1.53E-01 | 0.95 (0.89-1.02) | 697 | 4.26E-01 | 0.95 (0.84-1.08) | 9.73E-01 | 1.00 (0.87-1.14) |
| rs521319 | G/A | 11 | 63928607 | 6216 | 2985 | 4.89E-01 | 0.98 (0.91-1.05) | 704 | 1.98E-01 | 0.92 (0.82-1.04) | 3.14E-01 | 0.94 (0.82-1.07) |
| rs2238136 | G/A | 12 | 46563980 | 6224 | 2986 | 6.45E-01 | 1.02 (0.95-1.09) | 712 | 2.03E-01 | 0.92 (0.81-1.05) | 1.81E-01 | 0.91 (0.79-1.05) |
| rs7132324 | G/A | 12 | 46593576 | 6186 | 2947 | 4.19E-01 | 1.03 (0.96-1.10) | 689 | 6.03E-01 | 0.97 (0.86-1.09) | 3.27E-01 | 0.94 (0.83-1.07) |
| rs10870680 | G/A | 13 | 19638681 | 6221 | 2985 | 5.99E-01 | 0.98 (0.90-1.06) | 714 | 5.20E-02 | 0.86 (0.74-1.00) | 5.10E-02 | 0.85 (0.73-1.00) |
| rs11846930 | G/A | 14 | 61627081 | 6229 | 2976 | 9.03E-01 | 1.01 (0.89-1.14) | 709 | 7.93E-01 | 0.97 (0.78-1.21) | 7.74E-01 | 0.97 (0.76-1.23) |
| rs2123458 | A/G | 14 | 64128910 | 5519 | 2770 | 3.74E-01 | 1.03 (0.96-1.11) | 637 | 7.72E-01 | 0.98 (0.86-1.12) | 5.13E-01 | 0.95 (0.83-1.10) |
| rs1568221 | G/A | 15 | 34434869 | 6220 | 2984 | 1.01E-01 | 1.06 (0.99-1.13) | 712 | 4.79E-01 | 1.04 (0.93-1.17) | 8.36E-01 | 0.99 (0.87-1.12) |
| rs7191629 | A/G | 16 | 5689444 | 6074 | 2880 | 7.02E-01 | 1.01 (0.94-1.09) | 652 | 5.41E-01 | 1.04 (0.92-1.18) | 5.79E-01 | 1.04 (0.91-1.19) |
| rs4782024 | G/A | 16 | 17166439 | 5848 | 2962 | 3.55E-01 | 1.09 (0.91-1.30) | 697 | 3.76E-01 | 1.15 (0.85-1.55) | 6.55E-01 | 1.08 (0.78-1.49) |
| rs4430796 | A/G | 17 | 33172153 | 4349 | 1786 | **2.56E-05** | 0.84 (0.77-0.91) | 484 | 3.74E-01 | 0.94 (0.82-1.08) | 2.20E-01 | 1.10 (0.95-1.28) |
| rs4239217 | A/G | 17 | 33173100 | 4307 | 1753 | **5.90E-04** | 0.86 (0.79-0.94) | 588 | 6.18E-01 | 0.97 (0.85-1.10) | 7.60E-02 | 1.14 (0.99-1.31) |
| rs7501939 | G/A | 17 | 33175269 | 5234 | 2562 | **3.30E-04** | 0.88 (0.82-0.94) | 690 | 4.29E-01 | 0.95 (0.85-1.07) | 1.68E-01 | 1.09 (0.96-1.24) |
| rs1212721 | G/A | 18 | 5245109 | 6195 | 2953 | 8.67E-01 | 1.01 (0.93-1.09) | 696 | 4.62E-01 | 0.95 (0.82-1.09) | 6.10E-01 | 0.96 (0.82-1.12) |
| rs364418 | G/A | 18 | 7803239 | 6207 | 2962 | 8.09E-01 | 0.99 (0.93-1.06) | 698 | **9.00E-03** | 1.16 (1.04-1.30) | **1.70E-02** | 1.16 (1.03-1.31) |
| rs918171 | G/A | 19 | 3287539 | 6208 | 2960 | 7.10E-02 | 1.12 (0.99-1.26) | 698 | **2.40E-02** | 1.27 (1.03-1.56) | 2.35E-01 | 1.14 (0.92-1.42) |
| rs1557203 | A/G | 20 | 33627877 | 6224 | 2989 | 3.24E-01 | 1.06 (0.95-1.19) | 712 | 4.13E-01 | 1.09 (0.89-1.32) | 8.05E-01 | 1.03 (0.83-1.27) |

*NB: the numbers of cases with and without endometrioid histology do not sum to the total number of Stage 2 cases in Supplementary Table 1, because the main analysis also included a small number of cases for whom we did not have access to histology data.*

**Supplementary Table 5: Endometrial cancer case-control analysis using genotypes imputed from 1000 Genomes Project for SNPs within the rs4430796 linkage disequilibrium block.**

| SNP name | Position | Alleles | MAF | Rsq | $r^2$ with rs4430796 | P-value | Genotyped in stage 1? |
|---|---|---|---|---|---|---|---|
| rs7405776 | 33167135 | G/A | 0.38 | 0.66 | 0.51 | **6.07E-07** | |
| c17_pos33169359 | 33169359 | T/G | 0.05 | 0.61 | 0.04 | 6.63E-01 | |
| rs2005705 | 33170413 | G/A | 0.45 | 0.89 | 0.83 | **2.35E-07** | |
| c17_pos33170534 | 33170534 | T/C | 0.14 | 0.35 | 0.18 | 4.29E-03 | |
| rs757211 | 33170591 | C/T | 0.49 | 0.66 | 0.46 | **9.31E-07** | |
| rs757210 | 33170628 | C/T | 0.37 | 0.99 | 0.57 | **1.27E-06** | yes |
| **rs4430796** | **33172153** | **A/G** | **0.47** | **1.00** | **-** | **2.92E-07** | **yes** |
| rs4239217 | 33173100 | A/G | 0.40 | 1.00 | 0.80 | **2.60E-07** | yes |
| rs11651755 | 33173953 | T/C | 0.47 | 0.99 | 1.00 | **1.82E-07** | |
| c17_pos33173971 | 33173971 | G/C | 0.07 | 0.96 | 0.06 | 7.08E-01 | |
| rs10908278 | 33174065 | A/T | 0.47 | 0.95 | 0.96 | **2.25E-07** | |
| rs11657964 | 33174880 | G/A | 0.41 | 0.95 | 0.77 | **9.67E-07** | |
| rs7501939 | 33175269 | C/T | 0.40 | 1.00 | 0.80 | **1.16E-06** | yes |
| rs8064454 | 33175699 | C/A | 0.47 | 0.98 | 1.00 | **4.72E-07** | |
| rs12601991 | 33175746 | G/T | 0.45 | 0.76 | 0.64 | **1.38E-06** | |
| rs11263762 | 33176039 | G/A | 0.46 | 0.77 | 0.67 | **1.22E-06** | |
| rs7405696 | 33176148 | G/C | 0.46 | 0.77 | 0.67 | **1.22E-06** | |
| rs11651052 | 33176494 | G/A | 0.47 | 0.97 | 1.00 | **4.63E-07** | |
| rs757209 | 33176946 | G/A | 0.49 | 0.77 | 0.78 | **6.15E-07** | |
| rs9901746 | 33177262 | G/A | 0.44 | 0.74 | 0.62 | **1.69E-06** | |
| rs11263763 | 33177678 | A/G | 0.49 | 0.87 | 0.96 | **3.71E-07** | |
| rs11658063 | 33177985 | G/C | 0.39 | 0.96 | 0.80 | **8.59E-07** | |
| rs12453443 | 33178234 | C/G | 0.44 | 0.74 | 0.60 | **3.49E-06** | |
| c17_pos33178389 | 33178389 | C/G | 0.06 | 0.77 | 0.05 | 6.57E-01 | |
| c17_pos33178417 | 33178417 | C/G | 0.07 | 0.91 | 0.06 | 6.55E-01 | |
| rs34443065 | 33179971 | C/T | 0.13 | 0.46 | 0.11 | 1.02E-01 | |
| rs9913260 | 33180010 | G/A | 0.28 | 0.55 | 0.40 | 1.30E-05 | |
| rs3760511 | 33180426 | T/G | 0.32 | 0.99 | 0.30 | 1.45E-03 | yes |
| c17_pos33181982 | 33181982 | T/C | 0.06 | 0.75 | 0.05 | 6.60E-01 | |
| c17_pos33182146 | 33182146 | T/G | 0.07 | 0.88 | 0.06 | 6.61E-01 | |
| c17_pos33182283 | 33182283 | A/G | 0.07 | 0.86 | 0.06 | 6.58E-01 | |
| rs67143603 | 33182344 | T/C | 0.33 | 0.92 | 0.27 | 2.19E-03 | |
| rs17626423 | 33182480 | T/C | 0.21 | 0.99 | 0.21 | 3.71E-02 | yes |

***Notes:***

Genotypes were imputed for all 1265 endometrial cancer cases and 5190 controls included in stage 1 of the study

1000 Genomes Project data (CEU population) was used as the reference panel.

MAF=minor allele frequency (i.e. frequency of the allele listed second)

Rsq is an estimate of the squared correlation between the imputed and true genotypes for this SNP (SNPs with Rsq<0.3 are not shown)

P-values <1e-5 are shown in bold.

**Supplementary Table 6: Frequency of common *HNF1B* locus haplotypes in Stage 1 endometrial cancer cases and controls***

| Haplotype | Frequency | Case Frequency | Control Frequency | p-value |
|---|---|---|---|---|
| **AGGAAA** | **0.343** | **0.304** | **0.353** | **5.39E-06** |
| GAAGCA | 0.310 | 0.333 | 0.304 | 5.70E-03 |
| GAAGAG | 0.180 | 0.192 | 0.177 | 8.08E-02 |
| **GGAGAA** | **0.071** | **0.068** | **0.071** | **5.20E-01** |
| GAAGAA | 0.029 | 0.036 | 0.027 | 2.55E-02 |
| **GGGAAG** | **0.023** | **0.025** | **0.023** | **6.60E-01** |
| **GGGAAA** | **0.014** | **0.011** | **0.015** | **2.11E-01** |

* Common (>1%) haplotypes defined by tag SNPs rs757210, rs4430796, rs4239217, rs7501939, rs3760511, and rs1762642, respectively

Rare allele of rs4430796 lies only on haplotypes shown in bold

**Supplementary Table 7: Stage 1 Endometrial cancer risk estimates for GWAS-identified prostate cancer loci.**

| SNP | Region | Chromosome | Build36 Position | Reference* | Endometrial Cancer Stage 1 P-value | *Proxy SNP and $r^2$ with prostate SNP (if SNP not typed in endometrial study)* | Notes: LD with other prostate cancer SNPs |
|---|---|---|---|---|---|---|---|
| rs1465618 | THADA | 2 | 43407453 | Eeles et al 2009 | 1.00 | | |
| rs721048 | EHBP1 | 2 | 62985235 | Gudmundsson et al 2008 | 0.48 | *rs2710646; $r^2$=1.00* | |
| rs2710646 | EHBP1 | 2 | 62988383 | Gudmundsson et al 2008 | 0.48 | | $r^2$=1.00 with rs721048 |
| rs12621278 | ITGA6 | 2 | 173019799 | Eeles et al 2009 | 0.26 | | |
| rs2660753 | 3p12.1 | 3 | 87193364 | Eeles et al 2008 | 0.16 | | |
| rs10934853 | 3q21.3 | 3 | 129521063 | Gudmundsson et al 2009 | 0.27 | | |
| rs12500426 | PDLIM5 | 4 | 95733632 | Eeles et al 2009 | 0.59 | | |
| rs17021918 | PDLIM5 | 4 | 95781900 | Eeles et al 2009 | 0.73 | | |
| rs7679673 | TET2 | 4 | 106280983 | Eeles et al 2009 | 0.83 | *rs4698932; $r^2$=0.69* | |
| rs9364554 | SLC22A3 | 6 | 160753654 | Eeles et al 2008 | 0.77 | | |
| rs10486567 | JAZF1 | 7 | 27943088 | Thomas et al 2008 | 0.81 | | |
| rs6465657 | LMTK2 | 7 | 97654263 | Eeles et al 2008 | 0.51 | | |
| rs2928679 | NKX3-1 | 8 | 23494920 | Eeles et al 2009 | 0.90 | | |
| rs1512268 | NKX3-1 | 8 | 23582408 | Eeles et al 2009 | 0.33 | | |
| rs10086908 | 8q24 (1) | 8 | 128081119 | Amin Al Olama et al 2009 | 0.38 | | |
| rs1016343 | 8q24 (2) | 8 | 128162479 | Amin Al Olama et al 2009 | 0.85 | | |
| rs13254738 | 8q24 (2) | 8 | 128173525 | Haiman et al 2007 | 0.48 | *rs1456315; $r^2$=0.68* | |
| rs6983561 | 8q24 (2) | 8 | 128176062 | Amin Al Olama et al 2009 | 0.45 | *rs16901966; $r^2$=1.00* | |
| rs16901979 | 8q24 (2) | 8 | 128194098 | Gudmundsson et al 2007 | 0.42 | *rs10505483; $r^2$=1.00* | $r^2$=0.84 with rs6983561 |
| rs620861 | 8q24 (3) | 8 | 128404855 | Amin Al Olama et al 2009 | 0.26 | | |
| rs6983267 | 8q24 (4) | 8 | 128482487 | Yeager et al 2007 | 0.60 | | |
| rs1447295 | 8q24 (5) | 8 | 128554220 | Amundadottir et al 2006 | 0.68 | | |
| rs4242382 | 8q24 (5) | 8 | 128586755 | Eeles et al 2008 | 0.78 | | $r^2$=1.00 with rs1447295 |
| rs10090154 | 8q24 (5) | 8 | 128601319 | Amin Al Olama et al 2009 | 0.68 | *rs1447295; $r^2$=1.00* | $r^2$=1.00 with rs1447295 |
| rs10993994 | MSMB | 10 | 51219502 | Eeles et al 2008 | 0.19 | | |
| rs4962416 | CTBP2 | 10 | 126686862 | Thomas et al 2008 | 0.43 | | |
| rs7127900 | 11p15 | 11 | 2190150 | Eeles et al 2009 | 0.58 | | |
| rs7931342 | 11q13.2 | 11 | 68751073 | Eeles et al 2008 | 0.56 | | |
| rs10896449 | 11q13.2 | 11 | 68751243 | Thomas et al 2008 | 0.61 | | $r^2$=0.97 with rs7931342 |
| rs11649743 | 17q12 | 17 | 33149092 | Sun et al 2008 | 0.76 | | |
| **rs4430796** | **HNF1B** | **17** | **33172153** | **Gudmundsson et al 2007a** | **3.06E-07** | | |
| rs1859962 | 17q24.3 | 17 | 66620348 | Gudmundsson et al 2007a | 0.23 | | |
| rs8102476 | 19q13.2 | 19 | 43427453 | Gudmundsson et al 2009 | 0.41 | | |
| rs2735839 | KLK3 | 19 | 56056435 | Eeles et al 2008 | 0.51 | | |
| rs5759167 | 22q13.2 | 22 | 41830156 | Eeles et al 2009 | 0.20 | | |
| rs5945572 | NUDT10/11 | X | 51246423 | Gudmundsson et al 2008 | 0.69 | | $r^2$=0.91 with rs5945619 |
| rs5945619 | NUDT10/11 | X | 51258412 | Eeles et al 2008 | 0.56 | | |

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