List of genes and variants that were selected for meta-analysis (sorted by gene pathway)

Gene	Variant	rs number	Cases vs. controls (number of samples)	Ref allele	Ref allele freq cases	Ref allele freq controls	Attributable familial risk	Result of most recent meta-analysis; cases/ controls (samples) (reference)	Other Meta-analyses
Adhesion molecules									
CDH1	C-160A	rs16260	7493 vs. 7329 (5*)	C	0·74	0·72	0·19%	No assoc [1]	
MMP1	G-1607GG	rs1799750	1007 vs. 1032 (5)	G	0·44	0·47	0·62%	Positive assoc; 1343/1590 (7) [2]	[3]
MMP3	AAAAA-1612AAAAAA	rs3025058	857 vs. 932 (4)	5A	0·38	0·41	0·03%	No assoc; 1296/1543 (7) [2]	[3]
MMP9	1562C/T[1]	rs3918242	575 vs. 836 (4)	C				No assoc; 575/836 (4) [3]	[3]
Alcohol metabolism									
ADH1B	Arg47His	rs1229984	1931 vs. 2898 (5)	His	0·75	0·76	0·32%	n/a	
ADH1C	Ile349Val (1045A>G)	rs698	3168 vs. 6229 (7)	Ile (A)	0·69	0·64	0·14%	n/a	
ALDH2	Glu487Lys	rs671	2209 vs. 3383 (8)	Glu (G)	0·76	0·74	0·02%	Inverse assoc; 1960/3163 (6) [4]	
Angiogenesis									
VEGF	936 C>T	rs3025039	1317 vs. 1192 (4)	C	0·84	0·83	0·60%	n/a	
VEGF	G634C[2]	rs2010963	1508 vs. 1308 (4)	C			0·00%a	No assoc; 1508/1308 (4) [5]	

Base-excision repair									
MGMT	Leu84Phe[3]	rs12917	1524 vs. 4646 (5)	C	0·89	0·88	0·14%	No assoc; 1524/4646 (5) [6]	
MGMT	Il3143Val[3]	rs2308321	1326 vs. 3520 (4)	A	0·89	0·87	0·19%	No assoc; 1326/3520 (4) [6]	
MUTYH	G396D	rs36053993	26592 vs. 19207 (15)	G	0·99	0·99	0·87%	Positive assoc; 25616/18521 (15) [7]	[8, 9, 10, 11]
MUTYH	Y179C	rs34612342	26370 vs. 19042 (15)	A	1·00	1·00	0·00%	Positive assoc; 25392/18362 (15) [7]	[8, 9, 10, 11]
OGG1	Ser326Cys	rs1052133	4713 vs. 6165 (9)	Ser (C)	0·71	0·65	0·02%	n/a	
XRCC1	Arg194Trp	rs1799782	6635 vs. 8488 (11*)	C	0·91	0·88	0·16%	No assoc; 1709/3233 (9) [12]	[13]
XRCC1	Arg280His	rs25489	3114 vs. 3679 (5)	G	0·94	0·95	0·04%	No assoc; 931/1547 (4) [12]	
XRCC1	Arg399Gln	rs25487	7247 vs. 8786 (12*)	G	0·67	0·67	0·19%	No assoc; 2776/4402 (14) [12]	[13]
XRCC3	Thr241Met	rs861539	4484 vs. 5235 (10*)	T	0·72	0·71	0·00%	No assoc; 3183/3926 (7) [13]	
Inflammation/immune response									
IL6	174G>C	rs1800795	6676 vs. 7942 (10*§)	G	0·61	0·60	0·05%	n/a	
IL8	251T/A	rs4073	3228 vs. 3772 (7*)	T	0·54	0·54	0·02%	n/a	
IL10	1082G/A	rs1800896	2964 vs. 3621 (5*)	A	0·52	0·51	0·04%	n/a	
PPAR-gamma	C1431T	rs3856806	5574 vs. 7035 (7*)	C	0·87	0·87	0·08%	No assoc;	[15]

								486/941 (3) [14]	
PPAR-gamma	Pro12Ala	rs1801282	15091 vs. 18690 (17*§)	C	0·88	0·88	0·06%	Inverse assoc; 6878/9391 (10) [14]	
PTGS2/COX2	A1195G	rs689466	4756 vs. 6030 (7*)	A	0·73	0·74	0·03%	Positive assoc; 1196/ 1691 (2) [16]	
PTGS2/COX2	A1803G	rs4648298	4229 vs. 4279 (5*)	A	0·97	0·97	0·00%b	No assoc; 480/657 (2) [16]	
PTGS2/COX2	C427T	rs5275	4745 vs. 5756 (7*)	T	0·66	0·66	0·01%	n/a	
PTGS2/COX2	G306C	rs5277	4269 vs. 4735 (5*)	G	0·85	0·84	0·24%	n/a	
PTGS2/COX2	G765C	rs20417	5459 vs. 7272 (11*)	G	0·86	0·88	0·26%	Positive assoc; 3322/5166 (10) [17]	[16]
PTGS2/COX2	T1532C	rs5273	2843 vs. 3216 (5*)	val/val	1·00	1·00		No assoc; 670/1113 (4) [16]	
TNF-alpha	308G>A	rs1800629	3843 vs. 4098 (9*)	G	0·80	0·81	0·02%	No assoc; 1372/1458 (7) [18]	
NOD2	3020incC	rs5743293	4222 vs. 2988 (8)	G	0·96	0·97	0·95%	Positive assoc; 2571/1856 (7) [19]	
NOD2	G908R	rs2066845	4541 vs. 3820 (6*)	G	0·99	0·99		Positive assoc; 1442/1109 (5) [19]	
NOD2	R702W	rs2066844	3445 vs. 2731 (6*)	C	0·96	0·97	0·00%	Positive	

								assoc; 1436/1109 (5) [19]	
Inhibition of cell growth									
CCND1	870A	rs17852153	4747 vs. 6783 (13)	G	0·46	0·46	0·18%	Positive assoc; 2289/3232 (13) [20]	
TGFB1	C509T[4]	rs1800469	994 vs. 2335 (5)	T	0·42	0·48	1·99%	No assoc; 994/2335 (5) [21]	
TGFBR1	TGFBR1*6A	rs11466445	3217 vs. 4539 (8)	C	0·90	0·92	0·52%	Positive assoc; 5666/8450 (7) [22]	[23], [24]
Insulin related									
IGF1	CA-repeat	n/a	7900 vs. 9161 (6)	19/19	0·62	0·61	0·02%	No assoc; 3672/4125 (4) [25]	[26]
IGFBP3	202A>C	rs2854744	7296 vs. 10452 (6)	A	0·48	0·50	0·00%	No assoc; 2834/3520 (3) [25]	
Iron metabolism									
HFE	C282T	rs1800562	5177 vs. 6150 (6*§)	C	0·92	0·93	0·25%	n/a	
Lipid metabolism									
ApoE	e2	rs7412	5821 vs. 6754 (5*)	e3	0·81	0·81	0·80%	n/a	
ApoE	e4	rs429358	3808 vs. 4684 (5*)	e3	0·79	0·79	0·09%	n/a	
Mitotic control									
STK15	F31I	rs2273535	4860 vs. 4629 (4)	T	0·75	0·76	0·31%	Positive assoc; 2302/1769 (3) [27]	

One carbon metabolism									
MTHFR	C677T	rs1801133	27372 vs. 39867 (52§)	C	0·67	0·67	0·24%	Inverse assoc; 9345/18887 (37) [28]	[29, 30, 31, 32, 33, 34]
MTHFR	A1298C	rs1801131	17178 vs. 24792 (34§)	A	0·70	0·70	0·06%	Inverse assoc; 4764/6592 (9) [31]	
MTR	A2756G	rs1805087	11829 vs. 15975 (14§)	A	0·81	0·80	0·01%	No assoc; 7804/8184 (9) [35]	[34]
MTRR	A66G	rs1801394	6170 vs. 8732 (9)	A	0·59	0·61	0·02%	n/a	
TS	TSER	rs34743033	3519 vs. 5289 (5)	3R/3R	0·57	0·57	0·24%	n/a	
TS	Ts1494del6	rs34489327	3262 vs. 4518 (4)	ins/ins	0·67	0·67	0·03%	n/a	
Rare, high penetrance									
APC	E1317Q	rs1801166	6898 vs. 6668 (6)	G	0·99	0·99		No assoc; 3794/4484 (8) [36]	
APC	D1822V	rs459552	6282 vs. 7038 (6)	Asp	0·78	0·77	0·38%	n/a	
MLH1	I219V	rs1799977	2956 vs. 5071 (7*)	A	0·71	0·71	0·02%	n/a	
MLH1	-93 G>A	rs1800734	4524 vs. 5544 (6*)	G	0·77	0·78	0·12%	n/a	
Substrate metabolism									
CYP1A1	2454A>G	rs1048943	10274 vs. 11978 (13*§)	A	0·91	0·92	0·18%	Positive assoc; 5336/6226 (13) [37]	[34]
CYP1A1	3698T>C	rs4646903	4897 vs. 6559 (7)	T	0·84	0·83	0·20%	No assoc; 234/250 (2) [34]	
CYP1A2	163C>A	rs762551	3051 vs. 5326 (9)	A	0·68	0·68	0·00%	n/a	
CYP1B1	4326C>G	rs1056836	8514 vs. 9721 (6*)	C	0·53	0·53	0·00%	n/a	

CYP2C9	430C>T	rs1799853	5134 vs. 6164 (6*)	C	0·86	0·86	0·95%	n/a	
CYP2C9	1057A>C	rs1057910	5379 vs. 6531 (6*)	A	0·93	0·94	1·85%	n/a	
CYP2E1	1053C>T	rs2031920	4456 vs. 5077 (8§)	C	0·90	0·88	0·64%	n/a	
CYP2E1	1293G>C	rs3813867	3424 vs. 4686 (7)	G	0·94	0·93	1·06%	No assoc; 4979/6012 (10) [38]	
GSTA1	GSTA1*B allele[5]		1648 vs. 2039 (4)	A				No assoc; 1648/2039 (4) [39]	
GSTM1	Null variant	n/a	18845 vs. 26662 (43)	present	0·48	0·49		Positive assoc; 11998/17552 (44) [39]	[40], [41], [42], [43], [34]
GSTP1	IIe105Val	rs1695	9267 vs. 12902 (22*)	IA	0·71	0·72	0·09%	No assoc; 5421/7671 (19) [39]	[44], [34]
GSTP1	Ala114Val	rs1138272	5183 vs. 5457 (6*§)	C	0·92	0·92	0·22%	n/a	
GSTT1	Null variant	n/a	13410 vs. 20455 (35)	present	0·65	0·68		Positive assoc; 8596/13589 (34) [39]	[34], [45], [46]
NAT1	slow/rapid	n/a	4791 vs. 6628 (15)	slow	0·69	0·68	0·12%	No assoc; 520/433 (3) [34]	
NAT2	slow/rapid	n/a	12908 vs. 16483 (26)	slow	0·67	0·66	0·04%	No assoc; 6741/8015 (18) [34]	[47], [29], [48]
NQO1	Pro187Ser (C609T)	rs1800566	5084 vs. 5932 (8)	C	0·81	0·79	0·00%	Positive assoc; 1783/2494 (6) [49]	
Tumour supressor genes									
TP53	Arg72Pro[6]	rs1042522	7414 vs. 9872 (27)	G			0·01%	No assoc;	[51], [52], [53]

								7414/9872 (27) [50]	
TP53	intron 3 16bp[7]	rs17878362	1637 vs. 1874 (5)	Del			0·00%	No assoc; 1637/1874 (5) [54]	
MDM2	309 T/G[8]	rs2279744	2543 vs. 2115 (7)	G	0·46	0·43	0·42%	No assoc; 2543/2115 (7) [55]	[56]
Vit D and Ca metabolism									
VDR	BsmI (60890GA)	rs1544410	5607 vs. 6202 (7)	G	0·63	0·60	0·12%c	Inverse assoc; 3285/1497 (4) [57]	
VDR	FokI	rs10735810	7646 vs. 8968 (9§)	C	0·61	0·60	0·00%	No assoc; 1331/2943 (5) [57]	
VDR	TaqI	rs731236	946 vs. 1184 (4)	T	0·68	0·70	0·01%	n/a	
Common low penetrance									
SMAD7	rs4939827	rs4939827	37650 vs. 36154 (13§)	T	0·55	0·51	0·64%	n/a	
SMAD7	rs12953717	rs12953717	33771 vs. 32364 (11§)	C	0·63	0·65	0·30%	n/a	
SMAD7	rs4464148	rs4464148	15999 vs. 15216 (7*)	T	0·62	0·66	0·47%	n/a	
8q24	rs6983267	rs6983267	40604 vs. 42672 (19)	A	0·48	0·51	1·06%	Positive assoc; (17) [58]	
8q24	rs10505477	rs10505477	18580 vs. 20147 (14)	C	0·46	0·49	0·59%	n/a	
9p24	rs719725	rs719725	13290 vs. 14774 (13)	C	0·37	0·39	0·16%	Positive assoc; 14064/15933 (17) [59]	
19q13·1	rs10411210	rs10411210	25607 vs. 26477 (17)	C	0·89	0·88	0·09%	n/a	
16q22·1	rs9929218	rs9929218	26191 vs. 27409 (18)	G	0·74	0·72	0·23%	n/a	
15q14	rs4779584	rs4779584	13656 vs. 12635 (9)	C	0·64	0·65	0·84%	n/a	
1q41	rs6691170	rs6691170	17740 vs. 19776 (11)	G	0·62	0·64	0·18%	n/a	

3q26·2	rs10936599	rs10936599	17802 vs. 19795 (11)	C	0·77	0·75	0·12%	n/a	
12q13·13	rs11169552	rs11169552	17148 vs. 19739 (11)	C	0·74	0·72	0·76%	n/a	
20q13·33	rs4925386	rs4925386	17847 vs. 19832 (11)	C	0·71	0·68	0·39%	n/a	
14q22·2	rs4444235	rs4444235	18607 vs. 19576 (13)	T	0·53	0·55	0·21%	n/a	
20p12·3	rs961253	rs961253	18118 vs. 19006 (13)	C	0·66	0·68	0·22%	n/a	
8q23·3	rs16892766	rs16892766	17180 vs. 17840 (4*)	A	0·88	0·90	0·10%	n/a	
10p14	rs10795668	rs10795668	20026 vs. 20682 (6*)	G	0·72	0·69	0·52%	n/a	
11q23·1	rs3802842	rs3802842	33004 vs. 31654 (14)	A	0·67	0·70	0·37%	n/a	
a) ref allele frequency taken from 1000 genomes data									
b) OR for homozygote estimated as square of OR for heterozygotes									
c) Based on the white only analysis									
* Includes unpublished data from SOCCS									
§ Includes unpublished data from Ontario									
** Tomlinson 2008 was based on 10 samples									
[1]McColgan 2009									
[2]Liu 2010									
[3]Zhong 2010									
[4]Fang 2009									
[5]Ekonomopoulos 2010									
[6]Ekonomopoulos 2010									
[7]Hu 2010									
[8]Fang et al, 2010									

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