

eTable 1. Characteristics of Individual Studies

Study	Group	Region	Age Range	Sample Size
CHEK2*110delC				
Chekmariova et al., 2006	Controls	Russia	17-74	448
	Unilateral BC	Russia	25-86	660
	Bilateral BC	Russia	26-87	155
Vahteristo et al., 2002	Controls	Finland	18-65	1885
	Unilateral BC	Finland	Unselected	1035
	Bilateral BC	Finland	16-87	33
Johnson et al., 2005	Controls	UK	24-81	637
	Bilateral BC	UK	26-65	469
Meijers-Heijboer et al., 2002	Controls	Southwestern Netherlands	Matched to cases	909
CHEK2 Consortium 2004	Unilateral BC	Southwestern Netherlands	Unselected	2851
Broeks et al. 2007, 2004	Bilateral BC	Southwestern Netherlands	Majority cases dx < age 50	247
Justenhoven et al., 2004	Controls	Germany	24-80	1251
Pesch et al., 2005	Bilateral BC	Germany	29-89	106
Rashid et al., 2005				
Kilpivaara et al., 2005	Unilateral BC	Finland	26-97	1229
	Bilateral BC	Finland	-	68
Mellemkjaer et al., 2008	Unilateral BC	USA, Denmark	Unselected	1395
Bernstein JL et al., 2006	Bilateral BC	USA, Denmark	Unselected	708
CHEK2 Consortium 2004	Controls	East Anglia, UK	-	3749
Day et al., 1999	Unilateral BC	East Anglia, UK	45-74	2886
CHEK2 Consortium 2004	Controls	UK	Matched	288
Meijers-Heijboer et al., 2002	Unilateral BC	UK	36-45	564
	Bilateral BC	UK	-	-
CHEK2 Consortium 2004	Controls	Finland	Matched to cases	447
Mitrunen et al. 2001	Unilateral BC	Finland	-	464
CHEK2 Consortium 2004	Controls	Southern Germany	Matched to cases	650
Chang-Claude et al. 2000	Unilateral BC	Southern Germany	<51	604
CHEK2 Consortium 2004	Controls	Germany (Hannover)	-	401
Dork et al. 2001	Unilateral BC	Germany (Hannover)	-	985
CHEK2 Consortium 2004	Controls	Australia	Matched to cases	736
Spurdle et al. 2002	Unilateral BC	Australia	40-59	1474
De Jong et al., 2004	Controls	The Netherlands	-	184

	Unilateral BC	The Netherlands	Unselected	962
Pereira et al., 2004	Control	USA	-	1030
	Unilateral BC	USA	-	801
Van Binst et al., 2004	Controls	Belgium	-	103
	Unilateral BC	Belgium	-	52
Baeyens et al., 2005	Controls	Belgium	-	50
	Unilateral BC	Belgium	Unselected	100
Kleibl et al 2005	Controls	Czech Republic	-	688
	Unilateral BC	Czech Republic	-	688
Cybulski et al., 2007	Controls	Poland	-	2748
	Unilateral BC	Poland	>51	1978
Einarsdottir et al., 2006	Controls	Swedish	Matched	1334
	Unilateral BC	Swedish	50-74	1509
Weischer et al., 2007	Controls	Denmark	31-83	4633
	Unilateral BC	Denmark	36-85	1101
BRCA1/BRCA2 truncating mix				
Abeliovich et al., 1997	Unilateral BC	Non-Ashkenazi		36
	Bilateral BC	Non-Ashkenazi		3
Ahn et al., 2004	Unilateral BC	Korean	-	109
	Bilateral BC	Korean	-	15
Anglian Group (2000)	Unilateral BC	East Anglia	-	1220
Anton-Culver et al., 2000	Unilateral BC	US (4.5% Ashkenazi)	Unselected (29% <50 years)	590
	Bilateral BC	US (4.5% Ashkenazi)	Unselected (29% <50 years)	83
Apicella C et al., 2007	Unilateral BC	Ashkenazi (US)	Unselected	659
	Bilateral BC	Ashkenazi (US)		105
Backe et al., 1999	Unilateral BC	German	21-70	800
Bergthorsson JT et al., 2001	Controls	Danish	Unselected	180
	Bilateral BC	Danish	24-45 (selected <46 years)	58
Fodor et al., 1998	Controls	Ashkenazi	-	1715
	Unilateral BC	Ashkenazi	35-90	268
Gomes et al., 2007	Unilateral BC	Brazil	25-67	402
Gorski et al., 2005	Controls	Polish	-	4000
	Unilateral BC	Polish	26-85	2012
King et al., 2003	Unilateral BC	Ashkenazi (US)	-	1008

Roa et al, 1996	Controls	Ashk (3120); Non-Ashk (1045)	-	-
Rogozińska-Szczepka et al., 2004	Bilateral BC	Polish	28-76, median 47	108
Satagopan et al., 2001	Controls	Ashkenazi (USA)	Matched to cases	3434
	Unilateral BC	Ashk (USA 603, Canada 209)	Unselected	782
Sokolenko et al., 2007	Controls	Russian	18-74	478
Sokolenko et al., 2006	Unilateral BC	Russian	25-86	857
	Bilateral BC	Russian	25-85	152
Steinmann D et al., 2001	Unilateral BC	Germany	Matched to BiBC	-
	Bilateral BC	Germany	Median age: 54 years	75
Tang et al., 1999	Unilateral BC	Hong Kong	Unselected	130
Van Der Looij et al., 2000	Controls	Hungary	-	350
	Unilateral	Hungary	-	500
Warner et al., 1999	Controls	Ashkenazi (Canada)	25-88	380
	Unilateral BC	Ashkenazi (Canada)	-	412
FGFR2 rs2981582T/C				
Gorodnova et al., 2009 ¹	Controls	Russia, St. Petersburg	75-80 (174); 32-70(353)	524
	Unilateral BC	Russia, St. Petersburg	26-85	532
	Bilateral BC	Russia, St. Petersburg	28-78	100
McInerney et al., 2008	Controls	West of Ireland	>60 years	1016
	Unilateral BC	West of Ireland	24-90	988
	Bilateral BC	West of Ireland	Mean age: 53.5years	40
Huijts et al., 2007 ²	Controls	The Netherlands	-	590
	Unilateral BC	The Netherlands	21-87	1114
	Bilateral BC	The Netherlands	-	102

1. The data from this study have not yet been published. Two of our authors are co-investigators on this study (EK, EI).
2. The authors of this article supplied us with data on genotype frequencies that are unavailable in the published article.

eTable 2

	Frequencies of At-Risk Genotypes			Mantel-Haenszel Pooled Odds Ratio (95% CI)		
	Controls (%)	UBC (%)	BiBCs (%)	UBC vs. Control	BiBC vs. UBC	BiBC vs. Control
CHEK2*1100delC						
Chekmariova (2006)	1/448 (0.2)	14/660 (2.1)	8/155 (5.2)	9.7 (1.3-73.9)	2.5 (1.0-6.1)	20.7 (2.6-167)
Vahteristo (2002)	26/188 (1.4)	12/594 (2.0)	4/33 (12.1)	1.5 (0.74-2.9)	6.7 (2.0-22.0)	9.9 (3.2-30.1)
Johnson (2005)	4/637 (0.6)	-	11/583 (1.9)	-	-	3.4 (1.1-10.7)
Meijers-Heijboer (2004); Broeks (2007)	9/909 (1.0)	102/2851 (3.6)	15/247 (6.1)	3.7 (1.9-7.4)	1.7 (1.0-3.0)	6.5 (2.8-15.0)
Justenhoven (2004); Pesch (2005) Rashid (2005)	6/1251 (0.5)	-	2/106 (1.9)	-	-	4.0 (0.8-20.0)
Kilpivaara (2005)	-	26/1229 (2.1)	6/68 (8.8)	-	4.5 (1.8-11.3)	-
Bernstein (2004); Mellemkjaer (2008)	-	10/1395 (0.7)	7/708 (1.0)	-	1.4 (0.5-3.7)	-
CHEK2 Consortium (2004); Day (1999)	20/3749 (0.5)	35/2886 (1.2)	-	2.3 (1.3-4.0)	-	-
Chek2 Cons. (2004); Meijers-Heijboer (2002)	1/288 (0.4)	7/564 (1.2)	-	3.6 (0.44-29.5)	-	-
CHEK2 Consortium (2004); Mitrunen (2001)	5/447 (1.1)	13/464 (2.8)	-	2.5 (0.90-7.2)	-	-
CHEK2 Consortium (2004); Chang-Claude (2000)	1/650 (0.2)	2/601 (0.3)	-	2.2 (0.20-23.9)	-	-
CHEK2 Consortium (2004); Dork (2001)	1/401 (0.3)	11/985 (1.1)	-	4.5 (0.58-35.1)	-	-
CHEK2 Consortium (2004); Spurdle (2002)	1/736 (0.1)	10/1474 (0.7)	-	5.0 (0.64-39.3)	-	-
De Jong (2004)	3/184 (1.6)	28/962 (2.9)	-	1.8 (0.54-6.0)	-	-
Pereira (2004)	4/1030 (0.4)	9/801 (1.1)	-	2.9 (0.89-9.5)	-	-
Van Binst (2004)	1/103 (1.0)	0/52 (0.0)	-	0.7 (0.03-16.4)	-	-
Baeyens (2005)	0/50 (0.0)	1/100 (1.0)	-	1.5 (0.06-38.1)	-	-
Kleibl (2005)	2/730 (0.3)	3/688 (0.4)	-	1.6 (0.27-9.6)	-	-
Cybulski (2006)	6/2748 (0.2)	10/1978 (0.5)	-	2.3 (0.84-6.4)	-	-
Einarsdo' ttir (2006)	8/1334 (0.6)	19/1509 (1.3)	-	2.1 (0.92-4.8)	-	-
Weischer (2007)	22/4633 (0.5)	16/1374 (1.2)	-	2.5 (1.3-4.7)	-	-
Total frequency	121/22213 (0.5)	328/21167 (1.5)	53/1900 (2.8)	2.6 (2.0-3.3)	2.2 (1.5-3.2)	6.0 (3.5-10.2)
Heterogeneity	P=0.002	P<0.001	P<0.001	P=0.95	P=0.12	P=0.51
BRCA1 mix (Ashkenazi)						
Abeliovich (1997)	-	13/160 (8.1)	3/18 (16.7)	-	2.3 (0.58-8.8)	-

Apicella (2007)		73/659 (11.1)	24/105 (22.9)		2.4 (1.4-4.0)	
Satagopan (2001)	32/3440 (0.9)	57/782 (7.2)	-	8.4 (5.4-13.0)	-	-
Fodor (1998)	20/1715 (1.2)	10/268 (3.7)	-	3.3 (1.5-7.1)	-	-
King (2003)	-	67/1008 (6.7)	-	-	-	-
Warner (1999)	-	34/412 (8.3)	-	-	-	-
Roa (1996)	38/3116 (1.2)	-	-	-	-	-
Total frequency	90/8271 (1.1)	254/3289 (7.7)	27/127 (21.3)	6.7 (4.6-9.8)	2.2 (1.5-3.8)	
Heterogeneity	P=0.48	P=0.003	P=0.23	P=0.04	P=0.95	-
BRCA1 mix (non-Ashkenazi)						
Steinmann (2001)	-	3/75 (4.0)	1/75 (1.3)		0.3 (0.03-3.2)	-
Bergthorsson (2001)	0/180 (0.0)	-	9/58 (15.5)		-	107 (6.3-1835)
Sokolenko (2007, 2006)	-	13/150 (8.7)	18/152 (11.8)		1.4 (0.7-3.0)	-
Ahn (2004)	-	2/108 (1.9)	2/15 (13.3)		8.2 (1.1-62.9)	-
Anton-Culver (2000)	-	10/590 (1.7)	1/83 (1.2)		0.7 (0.09-5.6)	-
Anglian Breast Cancer Study Group (2000)	-	8/1220 (0.7)	-		-	-
Syrjakoski (2000)	-	4/1035 (0.4)	-		-	-
Van Der Looij (2000)	-	17/500 (3.4)	-		-	-
Gomes (2007)	-	6/402 (1.5)	-		-	-
Tang (1999)	-	5/130 (3.9)	-		-	-
Gorski (2005)	17/4000 (0.4)	59/2012 (2.9)	-	7.1 (4.1-12.2)	-	-
Roa (1996)	0/1045 (0.0)	-	-	-	-	-
Rogozinska-Szczepka (2004)	-	-	31/108 (28.7)		-	-
Total frequency	17/5225 (0.3)	127/6222 (2.0)	62/491 (12.6)	7.1 (4.1-12.2)	1.3 (0.7-2.4)	107 (6.3-1835)
Heterogeneity	P=0.081	P<0.001	P=0.000	-	P=0.18	-
BRCA1 5382insC (Ashkenazi)						
Satagopan (2001)	11/3440 (0.3)	12/782 (1.5)		4.9 (2.1-11.1)	-	-
Fodor (1998)	2/1715 (0.1)	2/268 (0.8)	-	6.4 (0.9-45.9)	-	-
King (2003)	-	25/1008 (2.5)	-	-	-	-
Warner (1999)	-	8/412 (1.9)	-	-	-	-
Roa (1996)	4/3116 (0.1)	-	-	-	-	-

Total frequency	17/8271 (0.21)	47/2470 (1.9)		5.1 (2.4-10.8)		-
Heterogeneity	P=0.20	P=0.26		P=0.80		-
BRCA1 5382insC (non-Ashkenazi)						
Bergthorsson (2001)	0/180 (0.0)	-	1/58 (1.7)	-	-	9.4 (0.38-234)
Sokolenko (2007, 2006)	0/478 (0.0)	32/857 (3.7)	15/144(10.4)	37 (2.3-603)	3.0 (1.6-5.7)	114 (6.8-1927)
Van Der Looij (2000)	-	7/500 (1.4)	-	-	-	-
Gomes (2007)	-	5/402 (1.2)	-	-	-	-
Backe (1999)	-	8/800 (1.0)	-	-	-	-
Gorski (2005)	14/4000 (0.7)	43/2012 (2.1)	-	6.2 (3.4-11.4)	-	-
Roa (1996)	0/1045 (0.0)	-	-	-	-	-
Total frequency	14/5703 (0.4)	95/4571 (2.1)	16/202 (7.9)	8.2 (4.5-14.9)	3.0 (1.6-5.7)	58 (7.9-428)
Heterogeneity	P=0.212	P=0.002	P=0.044	P=0.168	-	P=0.228
BRCA2 mix (Ashkenazi)						
Abeliovich (1997)	-	5/160 (3.1)	3/18 (16.7)	-	6.2 (1.4-28.5)	-
Apicella (2007)		22/659 (3.3)	7/105 (6.7)		2.1 (0.9-5.0)	
Satagopan (2001)	30/3440 (0.9)	23/782 (2.9)	-	3.4 (2.0-6.0)	-	-
Fodor (1998)	18/1715 (1.1)	8/268 (3.0)	-	2.9 (1.2-6.7)	-	-
King (2003)	-	37/1008 (3.7)	-	-	-	-
Warner (1999)	-	15/412 (3.6)	-	-	-	-
Roa (1996)	47/3085 (1.5)	-	-	-	-	-
Total frequency	95/8240 (1.2)	110/3289 (3.3)	10/123 (8.1)	3.3 (2.1-5.2)	2.6 (1.2-5.5)	
BRCA2 mix (non-Ashkenazi)						
Steinmann (2001)	-	0/75 (0.0)	4/75 (5.3)	-	9.5 (0.50-179)	-
Bergthorsson (2001)	0/180 (0.0)	-	4/58 (6.9)	-	-	29.8 (1.6-562)
Sokolenko (2007, 2006)	-	0/150 (0.0)	1/152 (0.6)	-	3.0 (0.12-73.7)	-
Ahn (2004)	-	1/108 (0.9)	1/15 (6.7)	-	7.6 (0.45-129)	-
Anglian Breast Cancer Study Group (2000)	-	16/1220 (1.3)	-	-	-	-

Syrjakoski (2000)	-	15/1035 (1.5)	-	-	-	-
Van Der Looij (2000)	-	1/500 (0.2)	-	-	-	-
Gomes (2007)	-	3/402 (0.8)	-	-	-	-
Roa (1996)	0/1045 (0.0)	-	-	-	-	-
Rogozinska-Szczepka (2004)	-	-	1/108 (0.9)	-	-	-
Total frequency	0/1225 (0.0)	36/3490 (1.0)	11/408 (2.7)	-	6.4 (1.1-38.6)	29.8 (1.6-562)
Heterogeneity	-	P=0.27	P=0.044	-	P=0.86	-
FGFR2 rs2981582/TT vs. CC						
Gorodnova (2009)	64/524 (12.2)	77/532 (14.5)	24/100 (24.0)	1.5 (1.0-2.2)	1.7 (0.97-3.1)	2.5 (1.4-4.5)
McInerney (2008)	179/997 (18.0)	214/941 (22.7)	-	1.5 (1.1-1.9)	-	-
Huijts (2007)	84/590 (14.2)	217/1114 (19.5)	21/102 (20.6)	1.7 (1.2-2.3)		
Total Frequency	327/2111 (15.5)	508/2587 (19.6)	45/202 (22.3)	1.5 (1.3-1.8)	1.4 (0.94-2.1)	2.2 (1.5-3.4)
Heterogeneity	P<0.001	P<0.001	P=0.76	P=0.80	P=0.35	P=0.54
FGFR2 rs2981582/TC vs. CC						
Gorodnova et al.	210/ 460 (45.7)	251/455 (55.2)	39/76 (51.3)	1.5 (1.13-1.90)	0.9 (0.53-1.4)	1.3 (0.77-2.1)
McInerney (2008)	483/818 (59.0)	458/727 (63.0)	-	1.2 (0.96-1.45)		
Huijts (2007)	280/506 (55.3)	550/897 (61.3)	52/81 (64.2)	1.3 (1.03-1.60)	1.1 (0.70-1.8)	1.4 (0.89-2.4)
Total frequency	973/2111(46.1)	1259/2587(48.7)	91/202 (45.0)	1.3 (1.13-1.46)	1.0 (0.71-1.4)	1.3 (0.96-1.9)
Heterogeneity	P<0.001	P=0.02	P=0.10	P=0.44	P=0.42	P=0.68

References for Supplementary Tables

Abeliovich D, Kaduri L, Lerer I, Weinberg N, Amir G, Sagi M, Zlotogora J, Heching N, Peretz T. The founder mutations 185delAG and 5382insC in BRCA1 and 6174delT in BRCA2 appear in 60% of ovarian cancer and 30% of early-onset breast cancer patients among Ashkenazi women. *Am J Hum Genet.* 1997 Mar;60(3):505-14.

Ahn SH, Hwang UK, Kwak BS, Yoon HS, Ku BK, Kang HJ, Kim JS, Ko BK, Ko CD, Yoon KS, Cho DY, Kim JS, Son BH. Prevalence of BRCA1 and BRCA2 mutations in Korean breast cancer patients. *J Korean Med Sci.* 2004 Apr;19(2):269-74.

Anglian Breast Cancer Study Group (2000) Prevalence and penetrance of *BRCA1* and *BRCA2* in a population based series of breast cancer cases. *Br J Cancer* 83:1301–1308

Anton-Culver H, Cohen PF, Gildea ME, Ziogas A (2000) Characteristics of *BRCA1* mutations in a population-based case series of breast and ovarian cancer. *Eur J Cancer* 36:1200–1208

Antoniou A, Pharoah PD, Narod S, Risch HA, Eyfjord JE, Hopper JL, Loman N, Olsson H, Johannsson O, Borg A, Pasini B, Radice P, Manoukian S, Eccles DM, Tang N, Olah E, Anton-Culver H, Warner E, Lubinski J, Gronwald J, Gorski B, Tulinius H, Thorlacius S, Eerola H, Nevanlinna H, Syrjäkoski K, Kallioniemi OP, Thompson D, Evans C, Peto J, Lalloo F, Evans DG, Easton DF. Average risks of breast and ovarian cancer associated with BRCA1 or BRCA2 mutations detected in case Series unselected for family history: a combined analysis of 22 studies. *Am J Hum Genet.* 2003 May;72(5):1117-30. Epub 2003 Apr 3. Erratum in: *Am J Hum Genet.* 2003 Sep;73(3):709.

Apicella C, Dowty JG, Dite GS, Jenkins MA, Senie RT, Daly MB, Andrulis IL, John EM, Buys SS, Li FP, Glendon G, Chung W, Ozcelik H, Miron A, Kotar K, Southey MC, Foulkes WD, Hopper JL. Validation study of the LAMBDA model for predicting the BRCA1 or BRCA2 mutation carrier status of North American Ashkenazi Jewish women. *Clin Genet.* 2007 Aug;72(2):87-97.

Backe, S. Hofferbert and B. Skawran *et al.*, Frequency of BRCA1 mutation 5382insC in German breast cancer patients, *Gynecol Oncol* **72** (1999), pp. 402–406.

Baeyens A, Claes K, Willems P, et al: Chromosomal radiosensitivity of breast cancer with a *CHEK2* mutation. *Cancer Genet Cytogenet* 2005; 163:106- 112,

Bergthorsson JT, Ejlertsen B, Olsen JH, Borg A, Nielsen KV, Barkardottir RB, Klausen S, Mouridsen HT, Winther K, Fenger K, Niebuhr A, Harboe TL, Niebuhr E. BRCA1 and BRCA2 mutation status and cancer family history of Danish women affected with multifocal or bilateral breast cancer at a young age. *J Med Genet.* 2001 Jun;38(6):361-8.

Bernstein JL, Teraoka SN, John EM, et al: The *CHEK2**1100delC allelic variant and risk of breast cancer: Screening results from the Breast Cancer Family Registry. *Cancer Epidemiol Biomarkers Prev* 2006; 15:348-352,

Broeks A, Braaf LM, Huseinovic A, et al. Identification of women with an increased risk of developing radiation-induced breast cancer: a case only study. *Breast Cancer Res* 2007;9:R26.

Broeks A, de Witte L, Nooijen A, et al. Excess risk for contralateral breast cancer in *CHEK2**1100delC germline mutation carriers. *Breast Cancer Res Treat* 2004;83:91 – 3.

Chang-Claude J, Eby N, Kiechle M, Bastert G, Becher H (2000) Breastfeeding and breast cancer risk by age 50 among women in Germany. *Cancer Causes Control* 11:687–695

CHEK2 Breast Cancer Case-Control Consortium. *CHEK2**1100delC and susceptibility to breast cancer: a collaborative analysis involving 10,860 breast cancer cases and 9,065 controls from 10 studies. *Am J Hum Genet* 2004;74:1175 – 82.

Chekmariova EV, Sokolenko AP, Buslov KG, et al. *CHEK2* 1100delC mutation is frequent among Russian breast cancer patients. *Breast Cancer Res Treat* 2006;100:99 – 102

Cybulski C, Wokolorczyk D, Huzarski T, et al: A deletion in *CHEK2* of 5,395 bp predisposes to breast cancer in Poland. *Breast Cancer Res Treat* 2007; 102:119-122

Day N, Oakes S, Luben R, Khaw KT, Bingham S, Welch A, Wareham N (1999) EPIC-Norfolk: study design and characteristics of the cohort. *European Prospective Investigation of Cancer. Br J Cancer* 80 Suppl 1: 95–103

De Jong MM, Van der Graaf WTA, Nolte IM, et al: Increased *CHEK2* 1100delC genotype frequency (also) in unselected breast cancer patients. *J Clin Oncol* 2004; 22:844s, (suppl; abstr 9536)

Dork T, Bendix R, Bremer M, Rades D, Klopper K, Nicke M, Skawran B, Hector A, Yamini P, Steinmann D, Weise S, Stuhmann M, Karstens JH (2001) Spectrum of ATM gene mutations in a hospital-based series of unselected breast cancer patients. *Cancer Res* 61:7608–7615

Dufault MR, Betz B, Wappenschmidt B, et al: Limited relevance of the *CHEK2* gene in hereditary breast cancer. *Int J Cancer* 2004; 110:320-325,

Einarsdottir K, Humphreys K, Bonnard C, et al: Linkage disequilibrium mapping of *CHEK2*: Common variation and breast cancer risk. *PLoS Med* 3:e168, 2006

Fodor F.H., A. Weston and I.J. Bleiweiss *et al.*, Frequency and carrier risk associated with common BRCA1 and BRCA2 mutations in Ashkenazi Jewish breast cancer patients, *Am J Hum Genet* **63** (1998), pp. 45–51.

Gomes MC, Costa MM, Borojevic R, Monteiro AN, Vieira R, Koifman S, Koifman RJ, Li S, Royer R, Zhang S, Narod SA. Prevalence of BRCA1 and BRCA2 mutations in breast cancer patients from Brazil. *Breast Cancer Res Treat.* 2007 Jul;103(3):349-53.

Górski B, Cybulski C, Huzarski T, Byrski T, Gronwald J, Jakubowska A, Stawicka M, Gozdecka-Grodecka S, Szwiec M, Urbański K, Mituś J, Marczyk E, Dziuba J, Wandzel P, Surdyka D, Haus O, Janiszewska H, Debniak T, Tołoczko-Grabarek A, Medrek K, Masojć B, Mierzejewski M, Kowalska E, Narod SA, Lubiński J. Breast cancer predisposing alleles in Poland. *Breast Cancer Res Treat.* 2005 Jul;92(1):19-24.

Huijts PE, Vreeswijk MP, Kroeze-Jansema KH, Jacobi CE, Seynaeve C, Krol-Warmerdam EM, Wijers-Koster PM, Blom JC, Pooley KA, Klijn JG, Tollenaar RA, Devilee P, van Asperen CJ. Clinical correlates of low-risk variants in *FGFR2*, *TNRC9*, *MAP3K1*, *LSP1* and *8q24* in a Dutch cohort of incident breast cancer cases. *Breast Cancer Res.* 2007;9(6):R78.

Johnson N, Fletcher O, Naceur-Lombardelli C, et al. Interaction between *CHEK2**1100delC and other low-penetrance breast-cancer susceptibility genes: a familial study. *Lancet* 2005;366:1554 – 7.

Justenhoven C, Hamann U, Pesch B, et al. *ERCC2* genotypes and a corresponding haplotype are linked with breast cancer risk in a German population. *Cancer Epidemiol Biomarkers Prev* 2004;13: 2059 – 64.

Kilpivaara O, Bartkova J, Eerola H, Syrjakoski K, Vahteristo P, Lukas J, Blomqvist C, Holli K, Heikkila P, Sauter G, Kallioniemi OP, Bartek J, Nevanlinna H (2005) Correlation of *CHEK2* protein expression and c.1100delC mutation status with tumor characteristics among unselected breast cancer patients. *Int J Cancer* 113: 575 –580

King M.C., J.H. Marks and J.B. Mandell, New York Breast Cancer Study Group. Breast and ovarian cancer risks due to inherited mutations in BRCA1 and BRCA2, *Science* **302** (2003), pp. 643–646.

Kleibl Z, Novotny J, Bezdickova D, et al: The *CHEK2* c. 1100delC germline mutation rarely contributes to breast cancer development in the Czech Republic. *Breast Cancer Res Treat* 2005; 90:165- 167,

McInerney N, Colleran G, Rowan A, Walther A, Barclay E, Spain S, Jones AM, Tuohy S, Curran C, Miller N, Kerin M, Tomlinson I, Sawyer E. Low penetrance breast cancer predisposition SNPs are site specific. *Breast Cancer Res Treat*. 2008 Nov 13. [Epub ahead of print] PMID: 19005751

Meijers-Heijboer H, van den Ouweland A, Klijn J, et al. Low penetrance susceptibility to breast cancer due to *CHEK2*(*1100delC) in noncarriers of *BRCA1* or *BRCA2* mutations. *Nat Genet* 2002;31: 55 – 9

Mellemkjaer L, Dahl C, Olsen JH, Bertelsen L, Guldborg P, Christensen J, Børresen-Dale AL, Stovall M, Langholz B, Bernstein L, Lynch CF, Malone KE, Haile RW, Andersson M, Thomas DC, Concannon P, Capanu M, Boice JD Jr; WECARE Study Collaborative Group, Bernstein JL. Risk for contralateral breast cancer among carriers of the *CHEK2**1100delC mutation in the WECARE Study. *Br J Cancer*. 2008 Feb 26;98(4):728-33.

Mitrunen, K, Jourenkova N, Kataja V, Eskelinen M, Kosma VM, Benhamou S, Kang D, Vainio H, Uusitupa M, Hirvonen A (2001) Polymorphic catechol-O-methyltransferase gene and breast cancer risk. *Cancer Epidemiol Biomarkers Prev* 10:635–640

Pereira LH, Sigurdson AJ, Doody MM, et al: *CHEK2*:1100delC and female breast cancer in the United States. *Int J Cancer* 2004; 112:541- 543

Pesch B, Ko Y, Brauch H, et al. Factors modifying the association between hormone-replacement therapy and breast cancer risk. *Eur J Epidemiol* 2005;20:699 – 711.

Rashid MU, Jakubowska A, Justenhoven C, et al. German populations with infrequent *CHEK2**1100delC and minor associations with early-onset and familial breast cancer. *Eur J Cancer* 2005;41: 2896 – 903.

Roa B.B., A.A. Boyd, K. Volcik and C.S. Richards, Ashkenazi Jewish population frequencies for common mutations in *BRCA1* and *BRCA2*, *Nat Genet* **14** (1996), pp. 185–187

Rogozińska-Szczepka J, Utracka-Hutka B, Grzybowska E, Maka B, Nowicka E, Smok-Ragankiewicz A, Zientek H, Steffen J, Wojciechowska-Łacka A. *BRCA1* and *BRCA2* mutations as prognostic factors in bilateral breast cancer patients. *Ann Oncol*. 2004 Sep;15(9):1373-6.

Satagopan J.M., K. Offit and W. Foulkes *et al.*, The lifetime risks of breast cancer in Ashkenazi Jewish carriers of *BRCA1* and *BRCA2* mutations, *Cancer Epidemiol Biomarkers Prev* **10** (2001), pp. 467–473.

Sodha N, Bullock S, Taylor R, et al: *CHEK2* variants in susceptibility to breast cancer and evidence of retention of the wild type allele in tumours. *Br J Cancer* 2002; 87:1445-1448,

Sokolenko AP, Mitiushkina NV, Buslov KG, Bit-Sava EM, Iyevleva AG, Chekmariova EV, Kuligina ESh, Ulibina YM, Rozanov ME, Suspitsin EN, Matsko DE, Chagunava OL, Trofimov DY, Devilee P, Cornelisse C, Togo AV, Semiglazov VF, Imyanitov EN. High frequency of BRCA1 5382insC mutation in Russian breast cancer patients. *Eur J Cancer*. 2006 Jul;42(10):1380-4.

Sokolenko AP, Rozanov ME, Mitiushkina NV, Sherina NY, Iyevleva AG, Chekmariova EV, Buslov KG, Shilov ES, Togo AV, Bit-Sava EM, Voskresenskiy DA, Chagunava OL, Devilee P, Cornelisse C, Semiglazov VF, Imyanitov EN. Founder mutations in early-onset, familial and bilateral breast cancer patients from Russia. *Fam Cancer*. 2007;6(3):281-6.

Spurdle AB, Hopper JL, Chen X, McCredie MR, Giles GG, Venter DJ, Southey MC, Chenevix-Trench G (2002). The progesterone receptor exon 4 Val660Leu G/T polymorphism and risk of breast cancer in Australian women. *Cancer Epidemiol Biomarkers Prev* 11:439–443

Steinmann D, Bremer M, Rades D, Skawran B, Siebrands C, Karstens JH, Dörk T. Mutations of the BRCA1 and BRCA2 genes in patients with bilateral breast cancer. *Br J Cancer*. 2001 Sep 14;85(6):850-8.

Syrjäkoski K, Vahteristo P, Eerola H, Tamminen A, Kivinummi K, Sarantaus L, Holli K, Blomqvist C, Kallioniemi OP, Kainu T, Nevanlinna H (2000) Population-based study of BRCA1 and BRCA2 mutations in 1035 unselected Finnish breast cancer patients. *J Natl Cancer Inst* 92:1529–1531

Tang NL, Pang CP, Yeo W, Choy KW, Lam PK, Suen M, Law LK, King WW, Johnson P, Hjelm M (1999) Prevalence of mutations in the BRCA1 gene among Chinese patients with breast cancer. *J Natl Cancer Inst* 91:882–885

Vahteristo P, Bartkova J, Eerola H, Syrjäkoski K, Ojala S, Kilpivaara O, Tamminen A, Kononen J, Aittomaki K, Heikkila P, Holli K, Blomqvist C, Bartek J, Kallioniemi OP, Nevanlinna H. A CHEK2 genetic variant contributing to a substantial fraction of familial breast cancer. *Am J Hum Genet* 2002; 71:432–438

Van Binst T, Goelen G, Sermijn E, Van Hassel G, De Neef K, Bonduelle M, Fontaine C, Teugels E, and De Greve JJ: The *CHEK2**1100delC mutation predisposes for early-onset breast cancer in breast cancer families. *Breast Cancer Res Treat* 2004; 88:S151,. (suppl; abstr)

Van Der Looij M, Szabo C, Besznyak I, Liszka G, Csokay B, Pulay T, Toth J, Devilee P, King MC, Olah E (2000) Prevalence of founder BRCA1 and BRCA2 mutations among breast and ovarian cancer patients in Hungary. *Int J Cancer* 86:737–740

Warner, W. Foulkes and P. Goodwin *et al.*, Prevalence and penetrance of BRCA1 and BRCA2 gene mutations in unselected Ashkenazi Jewish women with breast cancer, *J Natl Cancer Inst* **91** (1999), pp. 1241–1247.

Weischer M, Bojesen SE, Tybjaerg-Hansen A, et al: Increased risk of breast cancer associated with *CHEK2**1100delC. *J Clin Oncol* 25:57-63, 2007