

## Publikationen 2010

Antoniou, A.C., J. Beesley, L. McGuffog, O.M. Sinilnikova, S. Healey, S.L. Neuhausen, Y.C. Ding, T.R. Rebbeck, J.N. Weitzel, H.T. Lynch, C. Isaacs, P.A. Ganz, G. Tomlinson, O.I. Olopade, F.J. Couch, X. Wang, N.M. Lindor, V.S. Pankratz, P. Radice, S. Manoukian, B. Peissel, D. Zaffaroni, M. Barile, A. Viel, A. Allavena, V. Dall'Olio, P. Peterlongo, C.I. Szabo, M. Zikan, K. Claes, B. Poppe, L. Foretova, P.L. Mai, M.H. Greene, G. Rennert, F. Lejbkowitz, G. Glendon, H. Ozelik, I.L. Andrulis, N. Ontario Cancer Genetics, M. Thomassen, A.M. Gerdes, L. Sunde, D. Cruger, U. Birk Jensen, M. Caligo, E. Friedman, B. Kaufman, Y. Laitman, R. Milgrom, M. Dubrovsky, S. Cohen, A. Borg, H. Jernstrom, A. Lindblom, J. Rantala, M. Stenmark-Askmal, B. Melin, B. Swe, K. Nathanson, S. Domchek, A. Jakubowska, J. Lubinski, T. Huzarski, A. Osorio, A. Lasa, M. Duran, M.I. Tejada, J. Godino, J. Benitez, U. Hamann, M. Kriege, N. Hoogerbrugge, R.B. van der Luijt, C.J. van Asperen, P. Devilee, E.J. Meijers-Heijboer, M.J. Blok, C.M. Aalfs, F. Hogervorst, M. Rookus, Hebon, M. Cook, C. Oliver, D. Frost, D. Conroy, D.G. Evans, F. Lalloo, G. Pichert, R. Davidson, T. Cole, J. Cook, J. Paterson, S. Hodgson, P.J. Morrison, M.E. Porteous, L. Walker, M.J. Kennedy, H. Dorkins, S. Peock, Embrace, A.K. Godwin, D. Stoppa-Lyonnet, A. de Pauw, S. Mazoyer, V. Bonadona, C. Lasset, H. Dreyfus, D. Leroux, A. Hardouin, P. Berthet, L. Faivre, Gemo, C. Loustalot, T. Noguchi, H. Sobol, E. Rouleau, C. Nagues, M. Frenay, L. Venat-Bouvet, Gemo, J.L. Hopper, M.B. Daly, M.B. Terry, E.M. John, S.S. Buys, Y. Yassin, A. Miron, D. Goldgar, R. Breast Cancer Family, C.F. Singer, A.C. Dressler, D. Gschwantler-Kaulich, G. Pfeiler, T.V. Hansen, L. Jonson, B.A. Agnarsson, T. Kirchhoff, K. Offit, V. Devlin, A. Dutra-Clarke, M. Piedmonte, G.C. Rodriguez, K. Wakeley, J.F. Boggess, J. Basil, P.E. Schwartz, S.V. Blank, A.E. Toland, M. Montagna, C. Casella, E. Imyanitov, L. Tihomirova, I. Blanco, C. Lazaro, S.J. Ramus, L. Sucheston, B.Y. Karlan, J. Gross, R. Schmutzler, B. Wappenschmidt, C. Engel, A. Meindl, M. Lochmann, N. Arnold, S. Heidemann, R. Varon-Mateeva, D. Niederacher, C. Sutter, H. Deissler, D. Gadzicki, S. Preisler-Adams, K. Kast, I. Schonbuchner, T. Caldes, M. de la Hoya, K. Aittomaki, H. Nevanlinna, J. Simard, A.B. Spurdle, H. Holland, X. Chen, kConFab, R. Platte, G. Chenevix-Trench, D.F. Easton and Cimba (2010) Common breast cancer susceptibility alleles and the risk of breast cancer for BRCA1 and BRCA2 mutation carriers: implications for risk prediction. *Cancer Res*, 70(23): p. 9742-54.

Antoniou, A.C., X. Wang, Z.S. Fredericksen, L. McGuffog, R. Tarrell, O.M. Sinilnikova, S. Healey, J. Morrison, C. Kartsonaki, T. Lesnick, M. Ghousaini, D. Barrowdale, Embrace, S. Peock, M. Cook, C. Oliver, D. Frost, D. Eccles, D.G. Evans, R. Eeles, L. Izatt, C. Chu, F. Douglas, J. Paterson, D. Stoppa-Lyonnet, C. Houdayer, S. Mazoyer, S. Giraud, C. Lasset, A. Remenieras, O. Caron, A. Hardouin, P. Berthet, G.S. Collaborators, F.B. Hogervorst, M.A. Rookus, A. Jager, A. van den Ouweland, N. Hoogerbrugge, R.B. van der Luijt, H. Meijers-Heijboer, E.B. Gomez Garcia, Hebon, P. Devilee, M.P. Vreeswijk, J. Lubinski, A. Jakubowska, J. Gronwald, T. Huzarski, T. Byrski, B. Gorski, C. Cybulski, A.B. Spurdle, H. Holland, kConFab, D.E. Goldgar, E.M. John, J.L. Hopper, M. Southey, S.S. Buys, M.B. Daly, M.B. Terry, R.K. Schmutzler, B. Wappenschmidt, C. Engel, A. Meindl, S. Preisler-Adams, N. Arnold, D. Niederacher, C. Sutter, S.M. Domchek, K.L. Nathanson, T. Rebbeck, J.L. Blum, M. Piedmonte, G.C. Rodriguez, K. Wakeley, J.F. Boggess, J. Basil, S.V. Blank, E. Friedman, B. Kaufman, Y. Laitman, R. Milgrom, I.L. Andrulis, G. Glendon, H. Ozelik, T. Kirchhoff, J. Vijai, M.M. Gaudet, D. Altshuler, C. Guiducci, B. Swe, N. Loman, K. Harbst, J. Rantala, H. Ehrencrona, A.M. Gerdes, M. Thomassen, L. Sunde, P. Peterlongo, S.

Manoukian, B. Bonanni, A. Viel, P. Radice, T. Caldes, M. de la Hoya, C.F. Singer, A. Fink-Retter, M.H. Greene, P.L. Mai, J.T. Loud, L. Guidugli, N.M. Lindor, T.V. Hansen, F.C. Nielsen, I. Blanco, C. Lazaro, J. Garber, S.J. Ramus, S.A. Gayther, C. Phelan, S. Narod, C.I. Szabo, S. Mod, J. Benitez, A. Osorio, H. Nevanlinna, T. Heikkinen, M.A. Caligo, M.S. Beattie, U. Hamann, A.K. Godwin, M. Montagna, C. Casella, S.L. Neuhausen, B.Y. Karlan, N. Tung, A.E. Toland, J. Weitzel, O. Olopade, J. Simard, P. Soucy, W.S. Rubinstein, A. Arason, G. Rennert, N.G. Martin, G.W. Montgomery, J. Chang-Claude, D. Flesch-Janys, H. Brauch, Genica, G. Severi, L. Baglietto, A. Cox, S.S. Cross, P. Miron, S.M. Gerty, W. Tapper, D. Yannoukakos, G. Fountzilas, P.A. Fasching, M.W. Beckmann, I. Dos Santos Silva, J. Peto, D. Lambrechts, R. Paridaens, T. Rudiger, A. Forsti, R. Winqvist, K. Pylkas, R.B. Diasio, A.M. Lee, J. Eckel-Passow, C. Vachon, F. Blows, K. Driver, A. Dunning, P.P. Pharoah, K. Offit, V.S. Pankratz, H. Hakonarson, G. Chenevix-Trench, D.F. Easton and F.J. Couch (2010) A locus on 19p13 modifies risk of breast cancer in BRCA1 mutation carriers and is associated with hormone receptor-negative breast cancer in the general population. *Nat Genet*, 42(10): p. 885-92.

Audeh, M.W., J. Carmichael, R.T. Penson, M. Friedlander, B. Powell, K.M. Bell-McGuinn, C. Scott, J.N. Weitzel, A. Oaknin, N. Loman, K. Lu, R.K. Schmutzler, U. Matulonis, M. Wickens, and A. Tutt (2010) Oral poly(ADP-ribose) polymerase inhibitor olaparib in patients with BRCA1 or BRCA2 mutations and recurrent ovarian cancer: a proof-of-concept trial. *Lancet*, 376(9737): p. 245-51.

Catucci, I., R. Yang, P. Verderio, S. Pizzamiglio, L. Heesen, K. Hemminki, C. Sutter, B. Wappenschmidt, M. Dick, N. Arnold, P. Bugert, D. Niederacher, A. Meindl, R.K. Schmutzler, C.C. Bartram, F. Ficarazzi, L. Tizzoni, D. Zaffaroni, S. Manoukian, M. Barile, M.A. Pierotti, P. Radice, B. Burwinkel, and P. Peterlongo (2010) Evaluation of SNPs in miR-146a, miR196a2 and miR-499 as low-penetrance alleles in German and Italian familial breast cancer cases. *Hum Mutat*, 31(1): p. E1052-7.

Engel, C., B. Versmold, B. Wappenschmidt, J. Simard, D.F. Easton, S. Peock, M. Cook, C. Oliver, D. Frost, R. Mayes, D.G. Evans, R. Eeles, J. Paterson, C. Brewer, C. Epidemiological Study of Familial Breast, L. McGuffog, A.C. Antoniou, D. Stoppa-Lyonnet, O.M. Sinilnikova, L. Barjhoux, M. Frenay, C. Michel, D. Leroux, H. Dreyfus, C. Toulas, L. Gladiëff, N. Uhrhammer, Y.J. Bignon, A. Meindl, N. Arnold, R. Varon-Mateeva, D. Niederacher, S. Preisler-Adams, K. Kast, H. Deissler, C. Sutter, D. Gadzicki, G. Chenevix-Trench, A.B. Spurdle, X. Chen, J. Beesley, C. Kathleen Cuninghame Foundation Consortium for Research into Familial Breast, H. Olsson, U. Kristoffersson, H. Ehrencrona, A. Liljegren, S. Swedish Breast Cancer Study, R.B. van der Loo, T.A. van Os, F.E. van Leeuwen, B. Hereditary, N. Ovarian cancer group, S.M. Domchek, T.R. Rebbeck, K.L. Nathanson, A. Osorio, T. Ramon y Cajal, I. Konstantopoulou, J. Benitez, E. Friedman, B. Kaufman, Y. Laitman, P.L. Mai, M.H. Greene, H. Nevanlinna, K. Aittomaki, C.I. Szabo, T. Caldes, F.J. Couch, I.L. Andrulis, A.K. Godwin, U. Hamann, R.K. Schmutzler, and B. Consortium of Investigators of Modifiers of (2010) Association of the variants CASP8 D302H and CASP10 V410I with breast and ovarian cancer risk in BRCA1 and BRCA2 mutation carriers. *Cancer Epidemiol Biomarkers Prev*, 19(11): p. 2859-68.

Fletcher, O., N. Johnson, I. dos Santos Silva, N. Orr, A. Ashworth, H. Nevanlinna, T. Heikkinen, K. Aittomaki, C. Blomqvist, B. Burwinkel, C.R. Bartram, A. Meindl,

R.K. Schmutzler, A. Cox, I. Brock, G. Elliott, M.W. Reed, M.C. Southey, L. Smith, A.B. Spurdle, J.L. Hopper, F.J. Couch, J.E. Olson, X. Wang, Z. Fredericksen, P. Schurmann, R. Waltes, M. Bremer, T. Dork, P. Devilee, C.J. van Asperen, R.A. Tollenaar, C. Seynaeve, P. Hall, K. Czene, K. Humphreys, J. Liu, S. Ahmed, A.M. Dunning, M. Maranian, P.D. Pharoah, G. Chenevix-Trench, I. kConFab, A. Group, J. Beesley, N.V. Bogdanova, N.N. Antonenkova, I.V. Zalutsky, H. Anton-Culver, A. Ziogas, H. Brauch, Y.D. Ko, U. Hamann, G. Consortium, P.A. Fasching, R. Strick, A.B. Ekici, M.W. Beckmann, G.G. Giles, G. Severi, L. Baglietto, D.R. English, R.L. Milne, J. Benitez, J.I. Arias, G. Pita, B.G. Nordestgaard, S.E. Bojesen, H. Flyger, D. Kang, K.Y. Yoo, D.Y. Noh, A. Mannermaa, V. Kataja, V.M. Kosma, M. Garcia-Closas, S. Chanock, J. Lissowska, L.A. Brinton, J. Chang-Claude, S. Wang-Gohrke, A. Broeks, M.K. Schmidt, F.E. van Leeuwen, L.J. Van't Veer, S. Margolin, A. Lindblom, M.K. Humphreys, J. Morrison, R. Platte, D.F. Easton, J. Peto, and C. Breast Cancer Association (2010) Missense variants in ATM in 26,101 breast cancer cases and 29,842 controls. *Cancer Epidemiol Biomarkers Prev*, 19(9): p. 2143-51.

Flucke, U., M.T. Flucke, L. Hoy, E. Breuer, R. Goebbels, K. Rhiem, R. Schmutzler, H. Winzenried, M. Braun, S. Steiner, R. Buettner, and H. Gevensleben (2010) Distinguishing medullary carcinoma of the breast from high-grade hormone receptor-negative invasive ductal carcinoma: an immunohistochemical approach. *Histopathology*, 56(7): p. 852-9.

Gaudet, M.M., T. Kirchhoff, T. Green, J. Vijai, J.M. Korn, C. Guiducci, A.V. Segre, K. McGee, L. McGuffog, C. Kartsonaki, J. Morrison, S. Healey, O.M. Sinilnikova, D. Stoppa-Lyonnet, S. Mazoyer, M. Gauthier-Villars, H. Sobol, M. Longy, M. Frenay, G.S. Collaborators, F.B. Hogervorst, M.A. Rookus, J.M. Collee, N. Hoogerbrugge, K.E. van Roozendaal, H.S. Collaborators, M. Piedmonte, W. Rubinstein, S. Nerenstone, L. Van Le, S.V. Blank, T. Caldes, M. de la Hoya, H. Nevanlinna, K. Aittomaki, C. Lazaro, I. Blanco, A. Arason, O.T. Johannsson, R.B. Barkardottir, P. Devilee, O.I. Olopade, S.L. Neuhausen, X. Wang, Z.S. Fredericksen, P. Peterlongo, S. Manoukian, M. Barile, A. Viel, P. Radice, C.M. Phelan, S. Narod, G. Rennert, F. Lejbkowitz, A. Flugelman, I.L. Andrulis, G. Glendon, H. Ozelik, Ocg, A.E. Toland, M. Montagna, E. D'Andrea, E. Friedman, Y. Laitman, A. Borg, M. Beattie, S.J. Ramus, S.M. Domchek, K.L. Nathanson, T. Rebbeck, A.B. Spurdle, X. Chen, H. Holland, kConFab, E.M. John, J.L. Hopper, S.S. Buys, M.B. Daly, M.C. Southey, M.B. Terry, N. Tung, T.V. Overeem Hansen, F.C. Nielsen, M.H. Greene, P.L. Mai, A. Osorio, M. Duran, R. Andres, J. Benitez, J.N. Weitzel, J. Garber, U. Hamann, Embrace, S. Peock, M. Cook, C. Oliver, D. Frost, R. Platte, D.G. Evans, F. Lalloo, R. Eeles, L. Izatt, L. Walker, J. Eason, J. Barwell, A.K. Godwin, R.K. Schmutzler, B. Wappenschmidt, S. Engert, N. Arnold, D. Gadzicki, M. Dean, B. Gold, R.J. Klein, F.J. Couch, G. Chenevix-Trench, D.F. Easton, M.J. Daly, A.C. Antoniou, D.M. Altshuler and K. Offit (2010) Common genetic variants and modification of penetrance of BRCA2-associated breast cancer. *PLoS Genet*, 6(10): p. e1001183.

Hemminki, K., B. Muller-Myhsok, P. Lichtner, C. Engel, B. Chen, B. Burwinkel, A. Forsti, C. Sutter, B. Wappenschmidt, H. Hellebrand, T. Illig, N. Arnold, D. Niederacher, B. Dworniczak, H. Deissler, K. Kast, D. Gadzicki, T. Meitinger, H.E. Wichmann, M. Kiechle, C.R. Bartram, R.K. Schmutzler, and A. Meindl (2010) Low-risk variants FGFR2, TNRC9 and LSP1 in German familial breast cancer patients. *Int J Cancer*, 126(12): p. 2858-62.

Meindl, A., H. Hellebrand, C. Wiek, V. Erven, B. Wappenschmidt, D. Niederacher, M. Freund, P. Lichtner, L. Hartmann, H. Schaal, J. Ramser, E. Honisch, C. Kubisch, H.E. Wichmann, K. Kast, H. Deissler, C. Engel, B. Muller-Myhsok, K. Neveling, M. Kiechle, C.G. Mathew, D. Schindler, R.K. Schmutzler, and H. Hanenberg (2010) Germline mutations in breast and ovarian cancer pedigrees establish RAD51C as a human cancer susceptibility gene. *Nat Genet*, 42(5): p. 410-4.

Rhiem, K., U. Todt, B. Wappenschmidt, A. Klein, E. Wardelmann, and R.K. Schmutzler (2010) Sporadic breast carcinomas with somatic BRCA1 gene deletions share genotype/phenotype features with familial breast carcinomas. *Anticancer Res*, 30(9): p. 3445-9.

Rhiem K, S.R. (2010) Das familiäre Mammakarzinom *Der Gynäkologe* 43: p. 79-86.

Schmutzler, R.K., C. Engel, and I. Schreer (2010) Screening in women at elevated risk for breast cancer. *J Clin Oncol*, 28(30): p. e607-8; author reply e609-10.

Schmutzler RK, K.K., Familiäres Mammakarzinom – Beratung und Betreuung betroffener Familien., in *Mammakarzinom Interdisziplinär*, M.V. Kreienberg R, Jonat W, Kühn T Editor. 2010, Springer Verlag.

Tchatchou, S., A. Riedel, S. Lyer, J. Schmutzhard, O. Strobel-Freidekind, S. Gronert-Sum, C. Mietag, M. D'Amato, B. Schlehe, K. Hemminki, C. Sutter, N. Ditsch, A. Blackburn, L.Z. Hill, D.J. Jerry, P. Bugert, B.H. Weber, D. Niederacher, N. Arnold, R. Varon-Mateeva, B. Wappenschmidt, R.K. Schmutzler, C. Engel, A. Meindl, C.R. Bartram, J. Mollenhauer, and B. Burwinkel (2010) Identification of a DMBT1 polymorphism associated with increased breast cancer risk and decreased promoter activity. *Hum Mutat*, 31(1): p. 60-6.

Tutt, A., M. Robson, J.E. Garber, S.M. Domchek, M.W. Audeh, J.N. Weitzel, M. Friedlander, B. Arun, N. Loman, R.K. Schmutzler, A. Wardley, G. Mitchell, H. Earl, M. Wickens, and J. Carmichael (2010) Oral poly(ADP-ribose) polymerase inhibitor olaparib in patients with BRCA1 or BRCA2 mutations and advanced breast cancer: a proof-of-concept trial. *Lancet*, 376(9737): p. 235-44.  
Verderio, P., S. Pizzamiglio, M.C. Southey, A.B. Spurdle, J.L. Hopper, X. Chen, J. Beesley, K. Australian Ovarian Cancer Study Group, R.K. Schmutzler, C. Engel, B. Burwinkel, P. Bugert, F. Ficarazzi, S. Manoukian, M. Barile, B. Wappenschmidt, G. Chenevix-Trench, P. Radice, and P. Peterlongo (2010) A BRCA1 promoter variant (rs11655505) and breast cancer risk. *J Med Genet*, 47(4): p. 268-70.

Walker, L.C., Z.S. Fredericksen, X. Wang, R. Tarrell, V.S. Pankratz, N.M. Lindor, J. Beesley, S. Healey, X. Chen, kConFab, D. Stoppa-Lyonnet, C. Tirapo, S. Giraud, S. Mazoyer, D. Muller, J.P. Fricker, C. Delnatte, G.S. Collaborators, R.K. Schmutzler, B. Wappenschmidt, C. Engel, I. Schonbuchner, H. Deissler, A. Meindl, F.B. Hogervorst, M. Verheus, M.J. Hooning, A.M. van den Ouweland, M.R. Nelen, M.G. Ausems, C.M. Aalfs, C.J. van Asperen, P. Devilee, M.M. Gerrits, Q. Waisfisz, Hebon, C.I. Szabo, ModSquaD, D.F. Easton, S. Peock, M. Cook, C.T. Oliver, D. Frost, P. Harrington, D.G. Evans, F. Lalloo, R. Eeles, L. Izatt, C. Chu, R. Davidson, D. Eccles, K.R. Ong, J. Cook, Embrace, T. Rebbeck, K.L. Nathanson, S.M. Domchek, C.F. Singer, D. Gschwantler-Kaulich, A.C. Dressler, G. Pfeiler,

A.K. Godwin, T. Heikkinen, H. Nevanlinna, B.A. Agnarsson, M.A. Caligo, H. Olsson, U. Kristoffersson, A. Liljegren, B. Arver, P. Karlsson, B. Melin, B. Swe, O.M. Sinilnikova, L. McGuffog, A.C. Antoniou, G. Chenevix-Trench, A.B. Spurdle, and F.J. Couch (2010) Evidence for SMAD3 as a modifier of breast cancer risk in BRCA2 mutation carriers. *Breast Cancer Res*, 12(6): p. R102.

Wang, X., V.S. Pankratz, Z. Fredericksen, R. Tarrell, M. Karas, L. McGuffog, P.D. Pharoah, B.A. Ponder, A.M. Dunning, S. Peock, M. Cook, C. Oliver, D. Frost, Embrace, O.M. Sinilnikova, D. Stoppa-Lyonnet, S. Mazoyer, C. Houdayer, Gemo, F.B. Hogervorst, M.J. Hoening, M.J. Ligtenberg, Hebon, A. Spurdle, G. Chenevix-Trench, kConFab, R.K. Schmutzler, B. Wappenschmidt, C. Engel, A. Meindl, S.M. Domchek, K.L. Nathanson, T.R. Rebbeck, C.F. Singer, D. Gschwantler-Kaulich, C. Dressler, A. Fink, C.I. Szabo, M. Zikan, L. Foretova, K. Claes, G. Thomas, R.N. Hoover, D.J. Hunter, S.J. Chanock, D.F. Easton, A.C. Antoniou, and F.J. Couch (2010) Common variants associated with breast cancer in genome-wide association studies are modifiers of breast cancer risk in BRCA1 and BRCA2 mutation carriers. *Hum Mol Genet*, 19(14): p. 2886-97.

Yang, R., B. Schlehe, K. Hemminki, C. Sutter, P. Bugert, B. Wappenschmidt, J. Volkmann, R. Varon, B.H. Weber, D. Niederacher, N. Arnold, A. Meindl, C.R. Bartram, R.K. Schmutzler, and B. Burwinkel (2010) A genetic variant in the pre-miR-27a oncogene is associated with a reduced familial breast cancer risk. *Breast Cancer Res Treat*, 121(3): p. 693-702.