

<b>Personal information</b>	
First name(s) / Surname(s)	<b>Laura Ottini</b>
Address(es)	Department of Molecular Medicine, V.le Regina Elena 324, 00161, Rome, Italy
Telephone(s)	0039 06 44918268
E-mail	<a href="mailto:laura.ottini@uniroma1.it">laura.ottini@uniroma1.it</a>
<b>Occupational field</b>	Molecular Epidemiology of Cancer
<b>Work experience</b>	
2020-Present	Full Professor, Department of Molecular Medicine, "Sapienza" University of Rome, Rome, Italy
2005-2019	Associate Professor, Department of Molecular Medicine, "Sapienza" University of Rome, Rome, Italy
2000-2004	Research Assistant ( <i>Assegnista di Ricerca</i> ), "Sapienza" University of Rome, Rome, Italy
1999-2000	FIRC ( <i>Italian Foundation for Cancer Research</i> ) -Fellow, The Burnham Institute, La Jolla, California, USA
1996-1998	AIRC ( <i>Italian Association for Cancer Research</i> ) -Fellow, University of Chieti "G. D'Annunzio", Chieti, Italy
1992-1995	Board in Oncology
1992-1993	Post-Doc, European Molecular Biology Laboratory, Heidelberg, Germany
1990	Student-Fellow, National Institute of Health (NIH), Bethesda, Maryland, USA
<b>Education and training</b>	
July 2017	Integrative Molecular Epidemiology AACR Workshop - Harvard School of Public Health. Boston, USA
January 2015	Translational Bioinformatics Workshop, King's College, London (UK)
1991-1995	Board in Oncology, Sapienza University of Rome
1986-1991	MD, Sapienza University of Rome
<b>Mother Language</b>	Italian
<b>Other language(s)</b>	English (C1)
<b>Main activities and responsibilities</b>	<p><b>Research Activity</b></p> <p>Prof. Ottini's research activity is mainly focused on genetics and molecular epidemiology of solid tumors, with specific interest in breast, ovary, prostate, gastrointestinal and skin cancers. Her most significant studies concern the identification and characterization of genetic risk factors for breast and ovarian cancers, in the frame of national and international collaborations. Her most recent research activity concerns the characterization of genetic susceptibility, molecular basis and pathology of male breast cancer. In these fields, she has authored more than 100 peer-reviewed publications listed on Medline-PubMed and is:</p> <ul style="list-style-type: none"> <li>• Head of the Molecular Epidemiology Research Unit, Dept. Molecular Medicine "Sapienza" University of Rome.</li> <li>• Coordinator of the Italian Multicentre Study on Male Breast Cancer.</li> <li>• Coordinator of the Male Breast Cancer Research Study for the Italian Consortium within the Consortium of Investigators of Modifiers of BRCA1/2 (CIMBA).</li> </ul> <p><b>Teaching Activity</b></p> <ul style="list-style-type: none"> <li>• Medicine and Surgery, Course "C", University of Rome "La Sapienza"</li> <li>• Sciences of Health Prevention, University of Rome "La Sapienza"</li> <li>• Techniques of Biomedical Laboratories, University of Rome "La Sapienza"</li> <li>• PhD in "Molecular Medicine" University of Rome "La Sapienza"</li> </ul>

**Research Support**

Year		
2019	Sapienza University of Rome Research Funding RM11916B7A575951, PI	10.000,00 €
2018	AIRC IG #21389 5 year-project, PI	872.000,00 €
2018	Sapienza University of Rome Research Funding RM118164360F928D, PI	10.000,00 €
2017	Sapienza University of Rome Research Funding RM11715C3F07FFB2, PI	14.000,00 €
2016	Sapienza University of Rome Research Funding RM116154C95A9DCB, PI	14.000,00 €
2015	AIRC IG #16933 3 year-project, PI	683.000,00 €
2015	Sapienza University of Rome Research Funding C26A15ZK5M, PI	10.000,00 €
2014	Sapienza University of Rome Research Funding C26A14K2W4, PI	10.000,00 €
2013	Sapienza University of Rome Research Funding C26A13B92X, PI	3.000,00 €
2012	AIRC IG #12780 3 year-project, PI	450.000,00 €
2012	Sapienza University of Rome Research Funding C26A12X8PW, PI	5.000,00 €
2011	Sapienza University of Rome Instrumentation dedicated Funding C26G11J8SF, PI	35.000,00 €
2010	Sapienza University of Rome Research Funding C26A10BWXL, PI	13.500,00 €
2009	AIRC IG #8713 3 year-project, PI	300.000,00 €
2008	PRIN 008YKH47X_002 2 year-project, I	40.000,00 €
2006	Ricerca Finalizzata - Ministero della Salute, I	56.500,00 €
2005	PRIN 2005067073_005 2 year-project, I	38.000,00 €
2005	AIRC #1282 3 year-project, PI	120.000,00 €
2004	AIRC #2837 1 year-project, PI	40.000,00 €
2003	AIRC #2087 1year-project, PI	25.000,00 €

**Scientific Publications**

Total papers **131**; IF (relative to the publication year) **721.55**  
 Total papers (last 10 years) **73**; IF (relative to the publication year) **507.5**  
 Total Papers as first/last author or corresponding author **62**  
 Total H-index (Scopus) **35**

- Yang X, Leslie G, Doroszuk A, Schneider S, Allen J, Decker B, Dunning AM, Redman J, Scarth J, Plaskocinska I, Luccarini C, Shah M, Pooley K, Dorling L, Lee A, Adank MA, Adlard J, Aittomäki K, Andrusilis IL, Ang P, Barwell J, Bernstein JL, Bobolis K, Borg Å, Blomqvist C, Claes KBM, Concannon P, Cuggia A, Culver JO, Damiola F, de Pauw A, Diez O, Dolinsky JS, Domchek SM, Engel C, Evans DG, Fostira F, Garber J, Golmard L, Goode EL, Gruber SB, Hahn E, Hake C, Heikkilä T, Hurley JE, Janavicius R, Kleibl Z, Kleiblova P, Konstantopoulou I, Kvist A, Laduca H, Lee ASG, Lesueur F, Maher ER, Mannermaa A, Manoukian S, McFarland R, McKinnon W, Meindl A, Metcalfe K, Mohd Taib NA, Moilanen J, Nathanson KL, Neuhausen S, Ng PS, Nguyen-Dumont T, Nielsen SM, Obermair F, Offit K, Olopade OI, Ottini L, Penkert J, Pylkäs K, Radice P, Ramus SJ, Rudaitis V, Side L, Silva-Smith R, Silvestri V, Skytte AB, Slavin T, Soukupova J, Tondini C, Trainer AH, Unzeitig G, Usha L, van Overeem Hansen T, Whitworth J, Wood M, Yip CH, Yoon SY, Yussuf A, Zogopoulos G, Goldgar D, Hopper JL, Chenevix-

Trench G, Pharoah P, George SHL, Balmaña J, Houdayer C, James P, El-Haffaf Z, Ehrencrona H, Janatova M, Peterlongo P, Nevanlinna H, Schmutzler R, Teo SH, Robson M, Pal T, Couch F, Weitzel JN, Elliott A, Southey M, Winqvist R, Easton DF, Foulkes WD, Antoniou AC, Tischkowitz M. *Cancer Risks Associated With Germline PALB2 Pathogenic Variants: An International Study of 524 Families* **J Clin Oncol** 38. 2019 DOI <https://doi.org/10.1200/JCO.19.01907>.

2. Nicolussi A, Belardinilli F, Silvestri V, Mahdavian Y, Valentini V, D'Inzeo S, Petroni M, Zani M, Ferraro S, Di Giulio S, Fabretti F, Fratini B, Gradilone A, Ottini L, Giannini G, Coppa A, Capalbo C. *Identification of novel BRCA1 large genomic rearrangements by a computational algorithm of amplicon-based Next-Generation Sequencing data*. **PeerJ**. 2019 Nov 15;7:e7972. doi: 10.7717/peerj.7972. eCollection 2019.
3. Patel VL, Busch EL, Friebel TM, Cronin A, Leslie G, McGuffog L, Adlard J, Agata S, Agnarsson BA, Ahmed M, Aittomäki K, Alducci E, Andrusil IL, Arason A, Arnold N, Artioli G, Arver B, Auber B, Azzollini J, Balmaña J, Barkardottir RB, Barnes DR, Barroso A, Barrowdale D, Belotti M, Benitez J, Bertelsen B, Blok MJ, Bodrogi I, Bonadona V, Bonanni B, Bondavalli D, Boonen SE, Borde J, Borg A, Bradbury AR, Brady A, Brewer C, Brunet J, Buecher B, Buys SS, Cabezas-Camarero S, Caldés T, Caliebe A, Caligo MA, Calvello M, Campbell IG, Carnevali I, Carrasco E, Chan TL, Chu ATW, Chung WK, Claes KBM, Collaborators GS, Collaborators E, Cook J, Cortesi L, Couch FJ, Daly MB, Damante G, Darder E, Davidson R, de la Hoya M, Puppa LD, Dennis J, Díez O, Ding YC, Ditsch N, Domchek SM, Donaldson A, Dworniczak B, Easton DF, Eccles DM, Eeles RA, Ehrencrona H, Ejlertsen B, Engel C, Evans DG, Faivre L, Faust U, Feliubadaló L, Foretova L, Fostira F, Fountzilas G, Frost D, García-Barberán V, Garre P, Gauthier-Villars M, Géczi L, Gehrig A, Gerdes AM, Gesta P, Giannini G, Glendon G, Godwin AK, Goldgar DE, Greene MH, Gutierrez-Barrera AM, Hahn E, Hamann U, Hauke J, Herold N, Hogervorst FBL, Honisch E, Hopper JL, Hulick PJ, Investigators K, Investigators H, Izatt L, Jager A, James P, Janavicius R, Jensen UB, Jensen TD, Johannsson OT, John EM, Joseph V, Kang E, Kast K, Kiiski JI, Kim SW, Kim Z, Ko KP, Konstantopoulou I, Kramer G, Krogh L, Kruse TA, Kwong A, Larsen M, Lasset C, Lautrup C, Lazaro C, Lee J, Lee JW, Lee MH, Lemke J, Lesueur F, Liljegren A, Lindblom A, Llovet P, Lopez-Fernández A, Lopez-Perolio I, Lorca V, Loud JT, Ma ESK, Mai PL, Manoukian S, Mari V, Martin L, Matricardi L, Mebirouk N, Medici V, Meijers-Heijboer HEJ, Meindl A, Mensenkamp AR, Miller C, Gomes DM, Montagna M, Mooij TM, Moserle L, Mouret-Fourme E, Mulligan AM, Nathanson KL, Navratilova M, Nevanlinna H, Niederacher D, Nielsen FCC, Nikitina-Zake L, Offit K, Olah E, Olopade OI, Ong KR, Osorio A, Ott CE, Palli D, Park SK, Parsons MT, Pedersen IS, Peissel B, Peixoto A, Pérez-Segura P, Peterlongo P, Petersen AH, Porteous ME, Pujana MA, Radice P, Ramser J, Rantala J, Rashid MU, Rhiem K, Rizzolo P, Robson ME, Rookus MA, Rossing CM, Ruddy KJ, Santos C, Saule C, Scarpitta R, Schmutzler RK, Schuster H, Senter L, Seynaeve CM, Shah PD, Sharma P, Shin VY, Silvestri V, Simard J, Singer CF, Skytte AB, Snape K, Solano AR, Soucy P, Southey MC, Spurdle AB, Steele L, Steinemann D, Stoppa-Lyonnet D, Stradella A, Sunde L, Sutter C, Tan YY, Teixeira MR, Teo SH, Thomassen M, Tibiletti MG, Tischkowitz M, Tognazzo S, Toland AE, Tommasi S, Torres D, Toss A, Trainer AH, Tung N, van Asperen CJ, van der Baan FH, van der Kolk LE, van der Luijt RB, van Hest LP, Varesco L, Varon-Mateeva R, Viel A, Vierstrate J, Villa R, von Wachenfeldt A, Wagner P, Wang-Gohrke S, Wappenschmidt B, Weitzel JN, Wieme G, Yadav S, Yannoukakos D, Yoon SY, Zanzottera C, Zorn KK, D'Amico AV, Freedman ML, Pomerantz MM, Chenevix-Trench G, Antoniou AC, Neuhausen SL, Ottini L, Nielsen HR, Rebbeck TR. *Association of Genomic Domains in BRCA1 and BRCA2 with Prostate Cancer Risk and Aggressiveness*. **Cancer Res**. 2019 Nov 13; pii:canres.1840.2019. doi: 10.1158/0008-5472.CAN-19-1840.
4. Figlioli G, Bogliolo M, Catucci I, Caleca L, Lasheras SV, Pujol R, Kiiski JI, Muranen TA, Barnes DR, Dennis J, Michailidou K, Bolla MK, Leslie G, Aalfs CM; ABCTB Investigators, Adank MA, Adlard J, Agata S, Cadoo K, Agnarsson BA, Ahearn T, Aittomäki K, Ambrosone CB, Andrews L, Anton-Culver H, Antonenкова NN, Arndt V, Arnold N, Aronson KJ, Arun BK, Asseryanis E, Auber B, Auvinen P, Azzollini J, Balmaña J, Barkardottir RB, Barrowdale D, Barwell J, Beane Freeman LE, Beauparlant CJ, Beckmann MW, Behrens S, Benitez J, Berger R, Bermisheva M, Blanco AM, Blomqvist C, Bogdanova NV, Bojesen A, Bojesen SE, Bonanni B, Borg A, Brady AF, Brauch H, Brenner H, Brüning T, Burwinkel B, Buys SS, Caldés T, Caliebe A, Caligo MA, Campa D, Campbell IG, Canzian F, Castelao JE, Chang-Claude J, Chanock SJ, Claes KBM, Clarke CL, Collavoli A, Conner TA, Cox DG, Cybulski C, Czene K, Daly MB, de la Hoya M, Devilee P, Diez O, Ding YC, Dite GS, Ditsch N, Domchek SM, Dorfling CM, Dos-Santos-Silva I, Durda K, Dwek M, Eccles DM, Ekici AB, Eliassen AH, Ellberg C, Eriksson M, Evans DG, Fasching PA, Figueira J, Flyger H, Foulkes WD, Friebel TM, Friedman E, Gabrielson M, Gaddam P, Gago-Dominguez M, Gao C, Gapstur SM, Garber J, García-Closas M, García-Sáenz JA, Gaudet MM, Gayther SA; GEMO Study Collaborators, Giles GG, Glendon G, Godwin AK, Goldgar DE, Gutierrez-Barrera AM, Haeberle L, Haiman CA, Häkansson N, Hall P, Hamann U, Harrington PA, Hein A, Heyworth J, Hillemanns P, Hollestelle A, Hopper JL, Hosgood HD 3rd, Howell A, Hu C, Hulick PJ, Hunter DJ, Imyanitov EN; KConFab, Isaacs C, Jakimovska M, Jakubowska A, James P, Janavicius R, Janni W, John EM, Jones ME, Jung A, Kaaks R, Karlan BY, Khusnutdinova E, Kitahara CM, Konstantopoulou I, Koutros S, Kraft P, Lambrechts D, Lazaro C, Le Marchand L, Lester J, Lesueur F, Lilyquist J, Loud JT, Lu KH, Luben RN, Lubinski J, Mannermaa A, Manoochehri M, Manoukian S, Margolin S, Martens JWM, Maurer T, Mavroudis D, Mebirouk N, Meindl A, Menon U, Miller A, Montagna M, Nathanson KL, Neuhausen SL, Newman WG, Nguyen-Dumont T, Nielsen FC, Nielsen S, Nikitina-Zake L, Offit K, Olah E, Olopade OI, Olshan AF, Olson JE, Olsson H, Osorio A, Ottini L, Peissel B, Peixoto A, Peto J, Plaseska-Karanfilska D, Pocza T, Presneau N, Pujana MA, Punie K, Rack B, Rantala J, Rashid MU, Rau-Murthy R, Rennert G, Lejbkowicz F, Rhenius V, Romero A, Rookus MA, Ross EA, Rossing M, Rudaitis V, Ruebner M, Saloustros E, Sanden K, Santamariña M, Scheuner MT, Schmutzler RK, Schneider M, Scott C, Senter L, Shah M, Sharma P, Shu XO, Simard J, Singer CF, Sohn C, Soucy P, Southey MC, Spinelli JJ, Steele L, Stoppa-Lyonnet D, Tapper WJ, Teixeira MR, Terry MB, Thomassen M, Thompson J, Thull DL, Tischkowitz M, Tollenaar RAEM, Torres D, Troester MA, Truong T, Tung N, Untch M, Vachon CM, van Rensburg EJ, van Veen EM, Vega A, Viel A, Wappenschmidt B, Weitzel JN, Wendt C, Wieme G,

- Wolk A, Yang XR, Zheng W, Ziogas A, Zorn KK, Dunning AM, Lush M, Wang Q, McGuffog L, Parsons MT, Pharoah PDP, Fostira F, Toland AE, Andrusilis IL, Ramus SJ, Swerdlow AJ, Greene MH, Chung WK, Milne RL, Chenevix-Trench G, Dörk T, Schmidt MK, Easton DF, Radice P, Hahnen E, Antoniou AC, Couch FJ, Nevanlinna H, Surrallés J, Peterlongo P. *The FANCM:p.Arg658\* truncating variant is associated with risk of triple-negative breast cancer.* **NPJ Breast Cancer.** 2019 Nov 1;5:38. doi: 10.1038/s41523-019-0127-5.
5. Rizzolo P, Zelli V, Silvestri V, Valentini V, Zanna I, Bianchi S, Masala G, Spinelli AM, Tibiletti MG, Russo A, Varesco L, Giannini G, Capalbo C, Calistri D, Cortesi L, Viel A, Bonanni B, Azzollini J, Manoukian S, Montagna M, Peterlongo P, Radice P, Palli D, Ottini L. *Insight into genetic susceptibility to male breast cancer by multigene panel testing: Results from a multicenter study in Italy.* **Int J Cancer.** 2019 Jul 15;145(2):390-400. doi: 10.1002/ijc.32106. Epub 2019 Jan 24. PubMed PMID: 30613976.
  6. Rizzolo P, Silvestri V, Valentini V, Zelli V, Bucalo A, Zanna I, Bianchi S, Tibiletti MG, Russo A, Varesco L, Tedaldi G, Bonanni B, Azzollini J, Manoukian S, Coppa A, Giannini G, Cortesi L, Viel A, Montagna M, Peterlongo P, Radice P, Palli D, Ottini L. *Evaluation of CYP17A1 and CYP1B1 polymorphisms in male breast cancer risk.* **Endocr Connect.** 2019 Jul 1. pii: EC-19-0225.R1. doi: 10.1530/EC-19-0225. [Epub ahead of print]
  7. Valentini V, Zelli V, Gaggiano E, Silvestri V, Rizzolo P, Bucalo A, Calvieri S, Grassi S, Frascione P, Donati P, Soda G, Ottini L, Richetta AG. *MiRNAs as Potential Prognostic Biomarkers for Metastasis in Thin and Thick Primary Cutaneous Melanomas.* **Anticancer Res.** 2019 Aug;39(8):4085-4093. doi: 10.21873/anticanres.13566.
  8. Qian F, Rookus MA, Leslie G, Risch HA, Greene MH, Aalfs CM, Adank MA, Adlard J, Agnarsson BA, Ahmed M, Aittomäki K, Andrusilis IL, Arnold N, Arun BK, Ausems MGEM, Azzollini J, Barrowdale D, Barwell J, Benitez J, Białkowska K, Bonadona V, Borde J, Borg A, Bradbury AR, Brunet J, Buys SS, Caldés T, Caligo MA, Campbell I, Carter J, Chiquette J, Chung WK, Claes KBM, Collée JM, Collonge-Rame MA, Couch FJ, Daly MB, Delnatta C, Diez O, Domchek SM, Dorfling CM, Eason J, Easton DF, Eeles R, Engel C, Evans DG, Faivre L, Feliubadaló L, Foretova L, Friedman E, Frost D, Ganz PA, Garber J, Garcia-Barberan V, Gehrig A, Glendon G, Godwin AK, Gómez García EB, Hamann U, Hauke J, Hopper JL, Hulick PJ, Imyanitov EN, Isaacs C, Izatt L, Jakubowska A, Janavicius R, John EM, Karlan BY, Kets CM, Laitman Y, Lázaro C, Leroux D, Lester J, Lesueur F, Loud JT, Lubinski J, Łukomska A, McGuffog L, Mebirouk N, Meijers-Heijboer HEJ, Meindl A, Miller A, Montagna M, Mooij TM, Mouret-Fourme E, Nathanson KL, Nehoray B, Neuhausen SL, Nevanlinna H, Nielsen FC, Offit K, Olah E, Ong KR, Oosterwijk JC, Ottini L, Parsons MT, Peterlongo P, Pfeiler G, Pradhan N, Radice P, Ramus SJ, Rantala J, Rennert G, Robson M, Rodriguez GC, Salani R, Scheuner MT, Schmutzler RK, Shah PD, Side LE, Simard J, Singer CF, Steinemann D, Stoppa-Lyonnet D, Tan YY, Teixeira MR, Terry MB, Thomassen M, Tischkowitz M, Tognazzo S, Toland AE, Tung N, van Asperen CJ, van Engelen K, van Rensburg EJ, Venat-Bouvet L, Vierstraete J, Wagner G, Walker L, Weitzel JN, Yannoukakos D; KConFab Investigators; HEBON Investigators; GEMO Study Collaborators; EMBRACE Collaborators, Antoniou AC, Goldgar DE, Olopade OI, Chenevix-Trench G, Rebbeck TR, Huo D; CIMBA. *Mendelian randomisation study of height and body mass index as modifiers of ovarian cancer risk in 22,588 BRCA1 and BRCA2 mutation carriers.* **Br J Cancer.** 2019 Jun 19. doi: 10.1038/s41416-019-0492-8. [Epub ahead of print] PubMed PMID: 31213659.
  9. Nicolussi A, Belardinilli F, Mahdavian Y, Colicchia V, D'Inzeo S, Petroni M, Zani M, Ferraro S, Valentini V, Ottini L, Giannini G, Capalbo C, Coppa A. *Next-generation sequencing of BRCA1 and BRCA2 genes for rapid detection of germline mutations in hereditary breast/ovarian cancer.* **PeerJ.** 2019 Apr 22;7:e6661. doi: 10.7717/peerj.6661. eCollection 2019. PubMed PMID: 31065452; PubMed Central PMCID: PMC6482939.
  10. Rizzolo P, Silvestri V, Bucalo A, Zelli V, Valentini V, Catucci I, Zanna I, Masala G, Bianchi S, Spinelli AM, Tommasi S, Tibiletti MG, Russo A, Varesco L, Coppa A, Calistri D, Cortesi L, Viel A, Bonanni B, Azzollini J, Manoukian S, Montagna M, Radice P, Palli D, Peterlongo P, Ottini L. *Contribution of MUTYH Variants to Male Breast Cancer Risk: Results From a Multicenter Study in Italy.* **Front Oncol.** 2018 Dec 4;8:583. doi: 10.3389/fonc.2018.00583. eCollection 2018. PubMed PMID: 30564557; PubMed Central PMCID: PMC6288482.
  11. Santi R, Rizzolo P, Pietragalla M, Valentini V, Zelli V, Galassi FM, Ottini L, Nesi G. *The antiquity of hydrocephalus: the first full palaeo-neuropathological description.* **Neurol Sci.** 2019 Jun;40(6):1315-1322. doi: 10.1007/s10072-018-3643-4. Epub 2018 Nov 23. PubMed PMID: 30471018.
  12. Qian F, Wang S, Mitchell J, McGuffog L, Barrowdale D, Leslie G, Oosterwijk JC, Chung WK, Evans DG, Engel C, Kast K, Aalfs CM, Adank MA, Adlard J, Agnarsson BA, Aittomäki K, Alducci E, Andrusilis IL, Arun BK, Ausems MGEM, Azzollini J, Barouk-Simonet E, Barwell J, Belotti M, Benitez J, Berger A, Borg A, Bradbury AR, Brunet J, Buys SS, Caldés T, Caligo MA, Campbell I, Caputo SM, Chiquette J, Claes KBM, Margriet Collée J, Couch FJ, Coupier I, Daly MB, Davidson R, Diez O, Domchek SM, Donaldson A, Dorfling CM, Eeles R, Feliubadaló L, Foretova L, Fowler J, Friedman E, Frost D, Ganz PA, Garber J, Garcia-Barberan V, Glendon G, Godwin AK, Gómez García EB, Gronwald J, Hahn E, Hamann U, Henderson A, Hendricks CB, Hopper JL, Hulick PJ, Imyanitov EN, Isaacs C, Izatt L, Izquierdo Á, Jakubowska A, Kaczmarek K, Kang E, Karlan BY, Kets CM, Kim SW, Kim Z, Kwong A, Laitman Y, Lasset C, Hyuk Lee M, Won Lee J, Lee J, Lester J, Lesueur F, Loud JT, Lubinski J, Mebirouk N, Meijers-Heijboer HEJ, Meindl A, Miller A, Montagna M, Mooij TM, Morrison PJ, Mouret-Fourme E, Nathanson KL, Neuhausen SL, Nevanlinna H, Niederacher D, Nielsen FC, Nussbaum RL, Offit K, Olah E, Ong KR, Ottini L, Park SK,

- Peterlongo P, Pfeiler G, Phelan CM, Poppe B, Pradhan N, Radice P, Ramus SJ, Rantala J, Robson M, Rodriguez GC, Schmutzler RK, Hutten Selkirk CG, Shah PD, Simard J, Singer CF, Sokolowska J, Stoppa-Lyonnet D, Sutter C, Yen Tan Y, Teixeira RM, Teo SH, Terry MB, Thomassen M, Tischkowitz M, Toland AE, Tucker KM, Tung N, van Asperen CJ, van Engelen K, van Rensburg EJ, Wang-Gohrke S, Wappenschmidt B, Weitzel JN, Yannoukakos D; GEMO Study Collaborators; HEBON; EMBRACE, Greene MH, Rookus MA, Easton DF, Chenevix-Trench G, Antoniou AC, Goldgar DE, Olopade OI, Rebbeck TR, Huo D. *Height and Body Mass Index as Modifiers of Breast Cancer Risk in BRCA1/2 Mutation Carriers: A Mendelian Randomization Study*. *J Natl Cancer Inst.* 2019 Apr 1;111(4):350-364. doi: 10.1093/jnci/djy132. PubMed PMID: 30312457; PubMed Central PMCID: PMC6449171.
13. Richetta AG, Valentini V, Marraffa F, Paolino G, Rizzolo P, Silvestri V, Zelli V, Carbone A, Di Mattia C, Calvieri S, Frascione P, Donati P, Ottini L. *Metastases risk in thin cutaneous melanoma: prognostic value of clinical-pathologic characteristics and mutation profile*. *Oncotarget.* 2018 Aug 14;9(63):32173-32181. doi: 10.18632/oncotarget.25864. eCollection 2018 Aug 14. PubMed PMID: 30181807; PubMed Central PMCID: PMC6114949.
  14. Valentini V, Zelli V, Rizzolo P, Silvestri V, Alimandi M, D'Aloia MM, Giustini S, Calvieri S, Richetta AG, Monteleone G, Ottini L. *PIK3CA c.3140A>G mutation in a patient with suspected Proteus Syndrome: a case report*. *Clin Case Rep.* 2018 Jun 2;6(7):1358-1363. doi: 10.1002/ccr3.1546. eCollection 2018 Jul. PubMed PMID: 29988677; PubMed Central PMCID: PMC6028416.
  15. Ceccarelli V, Valentini V, Ronchetti S, Cannarile L, Billi M, Riccardi C, Ottini L, Talesa VN, Grignani F, Vecchini A. *Eicosapentaenoic acid induces DNA demethylation in carcinoma cells through a TET1-dependent mechanism*. *FASEB J.* 2018 May 14:fj201800245R. doi: 10.1096/fj.201800245R. [Epub ahead of print] PubMed PMID: 29757674.
  16. Rizzolo P, Silvestri V, Valentini V, Zelli V, Zanna I, Masala G, Bianchi S, Palli D, Ottini L. *Gene-specific methylation profiles in BRCA-mutation positive and BRCA-mutation negative male breast cancers*. *Oncotarget.* 2018 Apr 13;9(28):19783-19792. doi: 10.18632/oncotarget.24856. eCollection 2018 Apr 13. PubMed PMID: 29731982; PubMed Central PMCID: PMC5929425.
  17. Zanna I, Silvestri V, Palli D, Magrini A, Rizzolo P, Saieva C, Zelli V, Bendinelli B, Vezzosi V, Valentini V, Bianchi S, Ottini L\*, Masala G. *Smoking and FGFR2 rs2981582 variant independently modulate male breast cancer survival: A population-based study in Tuscany, Italy*. *Breast.* 2018 Aug;40:85-91. doi: 10.1016/j.breast.2018.04.017. Epub 2018 Apr 28. PubMed PMID: 29709729. \*Co-last author
  18. Silvestri V, Rizzolo P, Zelli V, Valentini V, Zanna I, Bianchi S, Tibiletti MG, Varesco L, Russo A, Tommasi S, Coppa A, Capalbo C, Calistri D, Viel A, Cortesi L, Manoukian S, Bonanni B, Montagna M, Palli D, Radice P, Peterlongo P, Ottini L. *A possible role of FANCM mutations in male breast cancer susceptibility: Results from a multicenter study in Italy*. *Breast.* 2018 Apr;38:92-97. doi: 10.1016/j.breast.2017.12.013. Epub 2018 Jan 4. PubMed PMID: 29287190.
  19. Coppa A, Nicolussi A, D'Inzeo S, Capalbo C, Belardinilli F, Colicchia V, Petroni M, Zani M, Ferraro S, Rinaldi C, Buffone A, Bartolazzi A, Screpanti I, Ottini L, Giannini G. *Optimizing the identification of risk-relevant mutations by multigene panel testing in selected hereditary breast/ovarian cancer families*. *Cancer Med.* 2018 Jan;7(1):46-55. doi: 10.1002/cam4.1251. Epub 2017 Dec 22. PubMed PMID: 29271107; PubMed Central PMCID: PMC5773970.
  20. Milne RL, Kuchenbaecker KB, Michailidou K, Beesley J, Kar S, Lindström S, Hui S, Lemaçon A, Soucy P, Dennis J, Jiang X, Rostamianfar A, Finucane H, Bolla MK, McGuffog L, Wang Q, Aalfs CM; ABCTB Investigators, Adams M, Adlard J, Agata S, Ahmed S, Ahsan H, Aittomäki K, Al-Ejeh F, Allen J, Ambrosone CB, Amos CI, Andrusil IL, Anton-Culver H, Antonenkova NN, Arndt V, Arnold N, Aronson KJ, Auber B, Auer PL, Ausems MGEM, Azzollini J, Bacot F, Balmaña J, Barile M, Barjhoux L, Barkardottir RB, Barrdahl M, Barnes D, Barrowdale D, Baynes C, Beckmann MW, Benitez J, Bermisheva M, Bernstein L, Bignon YJ, Blazer KR, Blok MJ, Blomqvist C, Blot W, Bobolis K, Boeckx B, Bogdanova NV, Bojesen A, Bojesen SE, Bonanni B, Børresen-Dale AL, Bozsik A, Bradbury AR, Brand JS, Brauch H, Brenner H, Bressac-de Paillerets B, Brewer C, Brinton L, Broberg P, Brooks-Wilson A, Brunet J, Brüning T, Burwinkel B, Buys SS, Byun J, Cai Q, Caldés T, Caligo MA, Campbell I, Canzian F, Caron O, Carracedo A, Carter BD, Castelao JE, Castera L, Caux-Moncoutier V, Chan SB, Chang-Claude J, Chanock SJ, Chen X, Cheng TD, Chiquette J, Christiansen H, Claes KBM, Clarke CL, Conner T, Conroy DM, Cook J, Cordina-Duverger E, Cornelissen S, Coupier I, Cox A, Cox DG, Cross SS, Cuk K, Cunningham JM, Czene K, Daly MB, Damiola F, Darabi H, Davidson R, De Leeneer K, Devilee P, Dicks E, Diez O, Ding YC, Ditsch N, Doheny KF, Domchek SM, Dorfling CM, Dörk T, Dos-Santos-Silva I, Dubois S, Dugué PA, Dumont M, Dunning AM, Durcan L, Dwek M, Dworniczak B, Eccles D, Eeles R, Ehrencrrona H, Eilber U, Ejlersen B, Ekici AB, Eliassen AH; EMBRACE, Engel C, Eriksson M, Fachal L, Faivre L, Fasching PA, Faust U, Figueroa J, Flesch-Janys D, Fletcher O, Flyger H, Foulkes WD, Friedman E, Fritsch L, Frost D, Gabrielson M, Gaddam P, Gammon MD, Ganz PA, Gapstur SM, Garber J, Garcia-Barberan V, García-Sáenz JA, Gaudet MM, Gauthier-Villars M, Gehrig A; GEMO Study Collaborators, Georgoulias V, Gerdes AM, Giles GG, Glendon G, Godwin AK, Goldberg MS, Goldgar DE, González-Neira A, Goodfellow P, Greene MH, Alnæs GIG, Grip M, Gronwald J, Grundy A, Gschwantler-Kaulich D, Guénél P, Guo Q, Haaberle L, Hahnem E, Haiman CA, Häkansson N, Hallberg E, Hamann U, Hamel N, Hankinson S, Hansen TVO, Harrington P, Hart SN, Hartikainen JM, Healey CS; HEBON, Hein A, Helbig S, Henderson A, Heyworth J, Hicks B, Hillemanns P, Hodgson S, Hogervorst FB, Hollestelle A, Hooning MJ, Hoover B, Hopper JL, Hu C, Huang G, Hulick PJ, Humphreys K, Hunter DJ, Imyanitov EN, Isaacs C, Iwasaki M, Izatt L, Jakubowska A, James P, Janavicius R, Janni W, Jensen

UB, John EM, Johnson N, Jones K, Jones M, Jukkola-Vuorinen A, Kaaks R, Kabisch M, Kaczmarek K, Kang D, Kast K; kConFab/AOCS Investigators, Keeman R, Kerin MJ, Kets CM, Keupers M, Khan S, Khusnutdinova E, Kiiski JI, Kim SW, Knight JA, Konstantopoulou I, Kosma VM, Kristensen VN, Kruse TA, Kwong A, Lænholm AV, Laitman Y, Laloo F, Lambrechts D, Landsman K, Lasset C, Lazaro C, Le Marchand L, Lecarpentier J, Lee A, Lee E, Lee JW, Lee MH, Lejbkowicz F, Lesueur F, Li J, Lilyquist J, Lincoln A, Lindblom A, Lissowska J, Lo WY, Loibl S, Long J, Loud JT, Lubinski J, Luccarini C, Lush M, MacInnis RJ, Maishman T, Makalic E, Kostovska IM, Malone KE, Manoukian S, Manson JE, Margolin S, Martens JWM, Martinez ME, Matsuo K, Mavroudis D, Mazoyer S, McLean C, Meijers-Heijboer H, Menéndez P, Meyer J, Miao H, Miller A, Miller N, Mitchell G, Montagna M, Muir K, Mulligan AM, Mulot C, Nadesan S, Nathanson KL; NBSC Collaborators, Neuhausen SL, Nevanlinna H, Nevelsteen I, Niederacher D, Nielsen SF, Nordestgaard BG, Norman A, Nussbaum RL, Olah E, Olopade OI, Olson JE, Olswold C, Ong KR, Oosterwijk JC, Orr N, Osorio A, Pankratz VS, Papi L, Park-Simon TW, Paulsson-Karlsson Y, Lloyd R, Pedersen IS, Peissel B, Peixoto A, Perez JIA, Peterlongo P, Peto J, Pfeiler G, Phelan CM, Pinchev M, Plaseska-Karanfilska D, Poppe B, Porteous ME, Prentice R, Presneau N, Prokofieva D, Pugh E, Pujana MA, Pylkas K, Rack B, Radice P, Rahman N, Rantala J, Rappaport-Fuerhauser C, Rennert G, Rennert HS, Rhenius V, Rhiem K, Richardson A, Rodriguez GC, Romero A, Romm J, Rookus MA, Rudolph A, Ruediger T, Saloustros E, Sanders J, Sandler DP, Sangrajrang S, Sawyer EJ, Schmidt DF, Schoemaker MJ, Schumacher F, Schürmann P, Schwentner L, Scott C, Scott RJ, Seal S, Senter L, Seynaeve C, Shah M, Sharma P, Shen CY, Sheng X, Shimelis H, Shrubsole MJ, Shu XO, Side LE, Singer CF, Sohn C, Southey MC, Spinelli JJ, Spurdle AB, Stegmaier C, Stoppa-Lyonnet D, Sukienicki G, Surowy H, Sutter C, Swerdlow A, Szabo CI, Tamimi RM, Tan YY, Taylor JA, Tejada MI, Tengström M, Teo SH, Terry MB, Tessier DC, Teulé A, Thöne K, Thull DL, Tibiletti MG, Tihomirova L, Tischkowitz M, Toland AE, Tollenar RAEM, Tomlinson I, Tong L, Torres D, Tranchant M, Truong T, Tucker K, Tung N, Tyrer J, Ulmer HU, Vachon C, van Asperen CJ, Van Den Berg D, van den Ouwendijk AMW, van Rensburg EJ, Varesco L, Varon-Mateeva R, Vega A, Viel A, Vijai J, Vincent D, Vollenweider J, Walker L, Wang Z, Wang-Gohrke S, Wappenschmidt B, Weinberg CR, Weitzel JN, Wendt C, Wesseling J, Whittemore AS, Wijnen JT, Willett W, Winquist R, Wolk A, Wu AH, Xia L, Yang XR, Yannoukakos D, Zaffaroni D, Zheng W, Zhu B, Ziogas A, Ziv E, Zorn KK, Gago-Dominguez M, Mannermaa A, Olsson H, Teixeira MR, Stone J, Offit K, Ottini L, Park SK, Thomassen M, Hall P, Meindl A, Schmutzler RK, Droit A, Bader GD, Pharoah PDP, Couch FJ, Easton DF, Kraft P, Chenevix-Trench G, García-Closas M, Schmidt MK, Antoniou AC, Simard J. Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. *Nat Genet.* 2017 Dec;49(12):1767-1778. doi: 10.1038/ng.3785. Epub 2017 Oct 23. PubMed PMID: 29058716; PubMed Central PMCID: PMC5808456.

21. Neuhausen SL, Shani H, Boker LK, Steele L, Silverman BG, Ottini L, Silvestri V, Laitman Y, Korach J, Perri T, Friedman E. Primary Peritoneal Serous Carcinoma in Men: A Rare and Non-BRCA-associated Entity. *Anticancer Res.* 2017 Jun;37(6):3069-3072. PubMed PMID: 28551646.
22. Lecarpentier J, Silvestri V, Kuchenbaecker KB, Barrowdale D, Dennis J, McGuffog L, Soucy P, Leslie G, Rizzolo P, Navazio AS, Valentini V, Zelli V, Lee A, Amin Al Olama A, Tyrer JP, Southey M, John EM, Conner TA, Goldgar DE, Buys SS, Janavicius R, Steele L, Ding YC, Neuhausen SL, Hansen TVO, Osorio A, Weitzel JN, Toss A, Medici V, Cortesi L, Zanna I, Palli D, Radice P, Manoukian S, Peissel B, Azzollini J, Viel A, Cini G, Damante G, Tommasi S, Peterlongo P, Fostira F, Hamann U, Evans DG, Henderson A, Brewer C, Eccles D, Cook J, Ong KR, Walker L, Side LE, Porteous ME, Davidson R, Hodgson S, Frost D, Adlard J, Izatt L, Eeles R, Ellis S, Tischkowitz M; EMBRACE, Godwin AK, Meindl A, Gehrig A, Dworczak B, Sutter C, Engel C, Niederacher D, Steinemann D, Hahnem E, Hauke J, Rhiem K, Kast K, Arnold N, Ditsch N, Wang-Gohrke S, Wappenschmidt B, Wand D, Lasset C, Stoppa-Lyonnet D, Belotti M, Damiola F, Barjhoux L, Mazoyer S; GEMO Study Collaborators, Van Heetvelde M, Poppe B, De Leeneer K, Claes KBM, de la Hoya M, Garcia-Barberan V, Caldes T, Perez Segura P, Kiiski JI, Aittomäki K, Khan S, Nevanlinna H, van Asperen CJ; HEBON, Vaszko T, Kasler M, Olah E, Balmaña J, Gutiérrez-Enríquez S, Diez O, Teulé A, Izquierdo A, Darder E, Brunet J, Del Valle J, Feliubadalo L, Pujana MA, Lazaro C, Arason A, Agnarsson BA, Johannsson OT, Barkardottir RB, Alducci E, Tognazzo S, Montagna M, Teixeira MR, Pinto P, Spurdle AB, Holland H; KConFab Investigators, Lee JW, Lee MH, Lee J, Kim SW, Kang E, Kim Z, Sharma P, Rebbeck TR, Vijai J, Robson M, Lincoln A, Musinsky J, Gaddam P, Tan YY, Berger A, Singer CF, Loud JT, Greene MH, Mulligan AM, Glendon G, Andrusil IL, Toland AE, Senter L, Bojesen A, Nielsen HR, Skytte AB, Sunde L, Jensen UB, Pedersen IS, Krogh L, Kruse TA, Caligo MA, Yoon SY, Teo SH, von Wachenfeldt A, Huo D, Nielsen SM, Olopade OI, Nathanson KL, Domchek SM, Lorenchick C, Jankowitz RC, Campbell I, James P, Mitchell G, Orr N, Park SK, Thomassen M, Offit K, Couch FJ, Simard J, Easton DF, Chenevix-Trench G, Schmutzler RK, Antoniou AC, Ottini L. Prediction of Breast and Prostate Cancer Risks in Male BRCA1 and BRCA2 Mutation Carriers Using Polygenic Risk Scores. *J Clin Oncol.* 2017 Jul 10;35(20):2240-2250. doi: 10.1200/JCO.2016.69.4935. Epub 2017 Apr 27. PubMed PMID: 28448241; PubMed Central PMCID: PMC5501359.
23. Phelan CM, Kuchenbaecker KB, Tyrer JP, Kar SP, Lawrenson K, Winham SJ, Dennis J, Pirie A, Riggan MJ, Chornokur G, Earp MA, Lyra PC Jr, Lee JM, Coetzee S, Beesley J, McGuffog L, Soucy P, Dicks E, Lee A, Barrowdale D, Lecarpentier J, Leslie G, Aalfs CM, Aben KKH, Adams M, Adlard J, Andrusil IL, Anton-Culver H, Antonenkova N; AOCS study group, Aravantinos G, Arnold N, Arun BK, Arver B, Azzollini J, Balmaña J, Banerjee SN, Barjhoux L, Barkardottir RB, Bean Y, Beckmann MW, Beeghly-Fadiel A, Benitez J, Bermisheva M, Bernardini MQ, Birrer MJ, Bjorge L, Black A, Blankstein K, Blok MJ, Bodelon C, Bogdanova N, Bojesen A, Bonanni B, Borg Å, Bradbury AR, Brenton JD, Brewer C, Brinton L, Broberg P, Brooks-Wilson A, Bruinsma F, Brunet J, Buecher B, Butzow R, Buys SS, Caldes T, Caligo MA, Campbell I, Cannioto R, Carney ME, Cescon T, Chan SB, Chang-Claude J, Chanock S, Chen XQ, Chiew YE, Chiquette J, Chung WK, Claeis KBM, Conner T, Cook LS, Cook J, Cramer DW, Cunningham JM, D'Aloisio AA, Daly MB, Damiola F, Damirovna SD, Dansonka-

Mieszkowska A, Dao F, Davidson R, DeFazio A, Delnatte C, Doheny KF, Diez O, Ding YC, Doherty JA, Domchek SM, Dorfling CM, Dörk T, Dossus L, Duran M, Dürst M, Dworniczak B, Eccles D, Edwards T, Eeles R, Eilber U, Ejlertsen B, Ekici AB, Ellis S, Elvira M; EMBRACE Study, Eng KH, Engel C, Evans DG, Fasching PA, Ferguson S, Ferrer SF, Flanagan JM, Fogarty ZC, Fortner RT, Fostira F, Foulkes WD, Fountzilas G, Fridley BL, Friebel TM, Friedman E, Frost D, Ganz PA, Garber J, García MJ, Garcia-Barberan V, Gehrig A; GEMO Study Collaborators, Gentry-Maharaj A, Gerdes AM, Giles GG, Glasspool R, Glendon G, Godwin AK, Goldgar DE, Goranova T, Gore M, Greene MH, Gronwald J, Gruber S, Hahnen E, Haiman CA, Håkansson N, Hamann U, Hansen TVO, Harrington PA, Harris HR, Hauke J; HEBON Study, Hein A, Henderson A, Hildebrandt MAT, Hillemanns P, Hodgson S, Høgdall CK, Høgdall E, Hogervorst FBL, Holland H, Hooning MJ, Hosking K, Huang RY, Hulick PJ, Hung J, Hunter DJ, Huntsman DG, Huzarski T, Imyanitov EN, Isaacs C, Iversen ES, Izatt L, Izquierdo A, Jakubowska A, James P, Janavicius R, Jernetz M, Jensen A, Jensen UB, John EM, Johnatty S, Jones ME, Kannisto P, Karlan BY, Karnezis A, Kast K; KConFab Investigators, Kennedy CJ, Khusnutdinova E, Kiemeney LA, Kiiski JI, Kim SW, Kjaer SK, Köbel M, Kopperud RK, Kruse TA, Kupryjanczyk J, Kwong A, Laitman Y, Lambrechts D, Larrañaga N, Larson MC, Lazaro C, Le ND, Le Marchand L, Lee JW, Lele SB, Leminen A, Leroux D, Lester J, Lesueur F, Levine DA, Liang D, Liebrich C, Lilyquist J, Lipworth L, Lissowska J, Lu KH, Lubinński J, Luccarini C, Lundvall L, Mai PL, Mendoza-Fandiño G, Manoukian S, Massuger LFAG, May T, Mazoyer S, McAlpine JN, McGuire V, McLaughlin JR, McNeish I, Meijers-Heijboer H, Meindl A, Menon U, Mensenkamp AR, Merritt MA, Milne RL, Mitchell G, Modugno F, Moes-Sosnowska J, Moffitt M, Montagna M, Moysich KB, Mulligan AM, Musinsky J, Nathanson KL, Nedergaard L, Ness RB, Neuhausen SL, Nevanlinna H, Niederacher D, Nussbaum RL, Odunsi K, Olah E, Olopade OI, Olsson H, Olswold C, O'Malley DM, Ong KR, Onland-Moret NC; OPAL study group, Orr N, Orsulic S, Osorio A, Palli D, Papi L, Park-Simon TW, Paul J, Pearce CL, Pedersen IS, Peeters PHM, Peissel B, Peixoto A, Pejovic T, Pelttari LM, Permuth JB, Peterlongo P, Pezzani L, Pfeiler G, Phillips KA, Piedmonte M, Pike MC, Piskorz AM, Poblete SR, Pocza T, Poole EM, Poppe B, Porteous ME, Prieur F, Prokofyeva D, Pugh E, Pujana MA, Pujol P, Radice P, Rantala J, Rappaport-Fuerhauser C, Rennert G, Rhiem K, Rice P, Richardson A, Robson M, Rodriguez GC, Rodriguez-Antona C, Romm J, Rookus MA, Rossing MA, Rothstein JH, Rudolph A, Runnebaum IB, Salvesen HB, Sandler DP, Schoemaker MJ, Senter L, Setiawan VW, Severi G, Sharma P, Shelford T, Siddiqui N, Side LE, Sieh W, Singer CF, Sobol H, Song H, Southey MC, Spurdle AB, Stadler Z, Steinemann D, Stoppa-Lyonnet D, Sucheston-Campbell LE, Sukiennicki G, Sutphen R, Sutter C, Swerdlow AJ, Szabo CI, Szafron L, Tan YY, Taylor JA, Tea MK, Teixeira MR, Teo SH, Terry KL, Thompson PJ, Thomsen LCV, Thull DL, Tihomirova L, Tinker AV, Tischkowitz M, Tognazzo S, Toland AE, Tone A, Trabert B, Travis RC, Trichopoulou A, Tung N, Tworoger SS, van Altena AM, Van Den Berg D, van der Hout AH, van der Luijt RB, Van Heetvelde M, Van Nieuwenhuysen E, van Rensburg EJ, Vanderstichele A, Varon-Mateeva R, Vega A, Edwards DV, Vergote I, Vierkant RA, Vijai J, Vratimos A, Walker L, Walsh C, Wand D, Wang-Gohrke S, Wappenschmidt B, Webb PM, Weinberg CR, Weitzel JN, Wentzensen N, Whittemore AS, Wijnen JT, Wilkens LR, Wolk A, Woo M, Wu X, Wu AH, Yang H, Yannoukakos D, Ziogas A, Zorn KK, Narod SA, Easton DF, Amos CI, Schildkraut JM, Ramus SJ, Ottini L, Goodman MT, Park SK, Kelemen LE, Risch HA, Thomassen M, Offit K, Simard J, Schmutzler RK, Hazelett D, Monteiro AN, Couch FJ, Berchuck A, Chenevix-Trench G, Goode EL, Sellers TA, Gayther SA, Antoniou AC, Pharoah PDP. Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. *Nat Genet.* 2017 May;49(5):680-691. doi: 10.1038/ng.3826. Epub 2017 Mar 27. PubMed PMID: 28346442; PubMed Central PMCID: PMC5612337.

24. Rizzolo P, Silvestri V, Ottini L. Retesting BRCA1/BRCA2 mutation negative male breast cancer patients using next generation sequencing technologies. *Breast Cancer Res Treat.* 2017 Feb;162(1):199-200. doi: 10.1007/s10549-017-4108-9. Epub 2017 Jan 16. PubMed PMID: 28091860.
25. Rizzolo P, Navazio AS, Silvestri V, Valentini V, Zelli V, Zanna I, Masala G, Bianchi S, Scarnò M, Tommasi S, Palli D, Ottini L. Somatic alterations of targetable oncogenes are frequently observed in BRCA1/2 mutation negative male breast cancers. *Oncotarget.* 2016 Nov 8;7(45):74097-74106. doi: 10.18632/oncotarget.12272. PubMed PMID: 27765917; PubMed Central PMCID: PMC5342038.
26. Amos CI, Dennis J, Wang Z, Byun J, Schumacher FR, Gayther SA, Casey G, Hunter DJ, Sellers TA, Gruber SB, Dunning AM, Michailidou K, Fachal L, Doheny K, Spurdle AB, Li Y, Xiao X, Romm J, Pugh E, Coetzee GA, Hazelett DJ, Bojesen SE, Cag-Anan C, Haiman CA, Kamal A, Luccarini C, Tessier D, Vincent D, Bacot F, Van Den Berg DJ, Nelson S, Demetriadis S, Goldgar DE, Couch FJ, Forman JL, Giles GG, Conti DV, Bickeböller H, Risch A, Waldenberger M, Brüské-Hohlfeld I, Hicks BD, Ling H, McGuffog L, Lee A, Kuchenbaecker K, Soucy P, Manz J, Cunningham JM, Butterbach K, Kote-Jarai Z, Kraft P, FitzGerald L, Lindström S, Adams M, McKay JD, Phelan CM, Benlloch S, Kelemen LE, Brennan P, Riggan M, O'Mara TA, Shen H, Shi Y, Thompson DJ, Goodman MT, Nielsen SF, Berchuck A, Laboissiere S, Schmit SL, Shelford T, Edlund CK, Taylor JA, Field JK, Park SK, Offit K, Thomassen M, Schmutzler R, Ottini L, Hung RJ, Marchini J, Amin AI Olama A, Peters U, Eeles RA, Seldin MF, Gillanders E, Seminara D, Antoniou AC, Pharoah PD, Chenevix-Trench G, Chanock SJ, Simard J, Easton DF. The OncoArray Consortium: A Network for Understanding the Genetic Architecture of Common Cancers. *Cancer Epidemiol Biomarkers Prev.* 2017 Jan;26(1):126-135. doi: 10.1158/1055-9965.EPI-16-0106. Epub 2016 Oct 3. Review. PubMed PMID: 27697780; PubMed Central PMCID: PMC5224974.
27. Silvestri V, Zelli V, Valentini V, Rizzolo P, Navazio AS, Coppa A, Agata S, Oliani C, Barana D, Castrignanò T, Viel A, Russo A, Tibiletti MG, Zanna I, Masala G, Cortesi L, Manoukian S, Azzollini J, Peissel B, Bonanni B, Peterlongo P, Radice P, Palli D, Giannini G, Chillemi G, Montagna M, Ottini L. Whole-exome sequencing and targeted gene sequencing provide insights into the role of PALB2 as a male breast cancer susceptibility gene. *Cancer.* 2017 Jan 1;123(2):210-218. doi: 10.1002/cncr.30337. Epub 2016 Sep 20. PubMed PMID: 27648926.

28. Navazio AS, Rizzolo P, Silvestri V, Valentini V, Zelli V, Zanna I, Masala G, Bianchi S, Tommasi S, Palli D, Ottini L. *EMSY copy number variation in male breast cancers characterized for BRCA1 and BRCA2 mutations*. *Breast Cancer Res Treat*. 2016 Nov;160(1):181-186. Epub 2016 Sep 15. PubMed PMID: 27628328.
29. Chandramouli B, Silvestri V, Scarno M, Ottini L, Chillemi G. *Smyd3 open & closed lock mechanism for substrate recruitment: The hinge motion of C-terminal domain inferred from μ-second molecular dynamics simulations*. *Biochim Biophys Acta*. 2016 Jul;1860(7):1466-74. doi: 10.1016/j.bbagen.2016.04.006. Epub 2016 Apr 13. PubMed PMID: 27085704.
30. Dunning AM, Michailidou K, Kuchenbaecker KB, Thompson D, French JD, Beesley J, Healey CS, Kar S, Pooley KA, Lopez-Knowles E, Dicks E, Barrowdale D, Sinnott-Armstrong NA, Sallari RC, Hillman KM, Kaufmann S, Sivakumaran H, Moradi Marjaneh M, Lee JS, Hills M, Jarosz M, Drury S, Canisius S, Bolla MK, Dennis J, Wang Q, Hopper JL, Southey MC, Broeks A, Schmidt MK, Lophatananon A, Muir K, Beckmann MW, Fasching PA, Dos-Santos-Silva I, Peto J, Sawyer EJ, Tomlinson I, Burwinkel B, Marme F, Guénel P, Truong T, Bojesen SE, Flyger H, González-Neira A, Perez JI, Anton-Culver H, Eunjung L, Arndt V, Brenner H, Meindl A, Schmutzler RK, Brauch H, Hamann U, Aittomäki K, Blomqvist C, Ito H, Matsuo K, Bogdanova N, Dörk T, Lindblom A, Margolin S, Kosma VM, Mannermaa A, Tseng CC, Wu AH, Lambrechts D, Wildiers H, Chang-Claude J, Rudolph A, Peterlongo P, Radice P, Olson JE, Giles GG, Milne RL, Haiman CA, Henderson BE, Goldberg MS, Teo SH, Yip CH, Nord S, Borresen-Dale AL, Kristensen V, Long J, Zheng W, Pylkäs K, Winqvist R, Andrusis IL, Knight JA, Devilee P, Seynaeve C, Figueroa J, Sherman ME, Czene K, Darabi H, Hollestelle A, van den Ouweland AM, Humphreys K, Gao YT, Shu XO, Cox A, Cross SS, Blot W, Cai Q, Ghoussaini M, Perkins BJ, Shah M, Choi JY, Kang D, Lee SC, Hartman M, Kabisch M, Torres D, Jakubowska A, Lubinski J, Brennan P, Sangrajrang S, Ambrosone CB, Toland AE, Shen CY, Wu PE, Orr N, Swerdlow A, McGuffog L, Healey S, Lee A, Kapuscinski M, John EM, Terry MB, Daly MB, Goldgar DE, Buys SS, Janavicius R, Tihomirova L, Tung N, Dorfling CM, van Rensburg EJ, Neuhausen SL, Ejlersen B, Hansen TV, Osorio A, Benitez J, Rando R, Weitzel JN, Bonanni B, Peissel B, Manoukian S, Papi L, Ottini L, Konstantopoulou I, Apostolou P, Garber J, Rashid MU, Frost D; EMBRACE, Izatt L, Ellis S, Godwin AK, Arnold N, Niederacher D, Rhiem K, Bogdanova-Markov N, Sagne C, Stoppa-Lyonnet D, Damiola F; GEMO Study Collaborators, Sinilnikova OM, Mazoyer S, Isaacs C, Claes KB, De Leeneer K, de la Hoya M, Caldes T, Nevanlinna H, Khan S, Mensenkamp AR; HEBON, Hooning MJ, Rookus MA, Kwong A, Olah E, Diez O, Brunet J, Pujana MA, Gronwald J, Huzarski T, Barkardottir RB, Laframboise R, Soucy P, Montagna M, Agata S, Teixeira MR; kConFab Investigators, Park SK, Lindor N, Couch FJ, Tischkowitz M, Foretova L, Vijai J, Offit K, Singer CF, Rappaport C, Phelan CM, Greene MH, Mai PL, Rennert G, Imyanitov EN, Hulick PJ, Phillips KA, Piedmonte M, Mulligan AM, Glendon G, Bojesen A, Thomassen M, Caligo MA, Yoon SY, Friedman E, Laitman Y, Borg A, von Wachenfeldt A, Ehrencrona H, Rantala J, Olopade OI, Ganz PA, Nussbaum RL, Gayther SA, Nathanson KL, Domchek SM, Arun BK, Mitchell G, Karlan BY, Lester J, Maskarinec G, Woolcott C, Scott C, Stone J, Apicella C, Tamimi R, Luben R, Khaw KT, Helland Å, Haakensen V, Dowsett M, Pharoah PD, Simard J, Hall P, García-Closas M, Vachon C, Chenevix-Trench G, Antoniou AC, Easton DF, Edwards SL. *Breast cancer risk variants at 6q25 display different phenotype associations and regulate ESR1, RMND1 and CCDC170*. *Nat Genet*. 2016 Apr;48(4):374-86. doi: 10.1038/ng.3521. Epub 2016 Feb 29. PubMed PMID: 26928228; PubMed Central PMCID: PMC4938803.
31. Silvestri V, Barrowdale D, Mulligan AM, Neuhausen SL, Fox S, Karlan BY, Mitchell G, James P, Thull DL, Zorn KK, Carter NJ, Nathanson KL, Domchek SM, Rebbeck TR, Ramus SJ, Nussbaum RL, Olopade OI, Rantala J, Yoon SY, Caligo MA, Spugnesi L, Bojesen A, Pedersen IS, Thomassen M, Jensen UB, Toland AE, Senter L, Andrusis IL, Glendon G, Hulick PJ, Imyanitov EN, Greene MH, Mai PL, Singer CF, Rappaport-Fuerhauser C, Kramer G, Vijai J, Offit K, Robson M, Lincoln A, Jacobs L, Machackova E, Foretova L, Navratilova M, Vasickova P, Couch FJ, Hallberg E, Ruddy KJ, Sharma P, Kim SW; kConFab Investigators, Teixeira MR, Pinto P, Montagna M, Matricardi L, Arason A, Johannsson OT, Barkardottir RB, Jakubowska A, Lubinski J, Izquierdo A, Pujana MA, Balmaña J, Diez O, Ivady G, Papp J, Olah E, Kwong A; Hereditary Breast and Ovarian Cancer Research Group Netherlands (HEBON), Nevanlinna H, Aittomäki K, Perez Segura P, Caldes T, Van Maerken T, Poppe B, Claes KB, Isaacs C, Elan C, Lasset C, Stoppa-Lyonnet D, Barjhoux L, Belotti M, Meindl A, Gehrig A, Sutter C, Engel C, Niederacher D, Steinemann D, Hahnens E, Kast K, Arnold N, Varon-Mateeva R, Wand D, Godwin AK, Evans DG, Frost D, Perkins J, Adlard J, Izatt L, Platte R, Eeles R, Ellis S; EMBRACE, Hamann U, Garber J, Fostira F, Fountzilas G, Pasini B, Giannini G, Rizzolo P, Russo A, Cortesi L, Papi L, Varesco L, Palli D, Zanna I, Savarese A, Radice P, Manoukian S, Peissel B, Barile M, Bonanni B, Viel A, Pensotti V, Tommasi S, Peterlongo P, Weitzel JN, Osorio A, Benitez J, McGuffog L, Healey S, Gerdes AM, Ejlersen B, Hansen TV, Steele L, Ding YC, Tung N, Janavicius R, Goldgar DE, Buys SS, Daly MB, Bane A, Terry MB, John EM, Southey M, Easton DF, Chenevix-Trench G, Antoniou AC, Ottini L. *Male breast cancer in BRCA1 and BRCA2 mutation carriers: pathology data from the Consortium of Investigators of Modifiers of BRCA1/2*. *Breast Cancer Res*. 2016 Feb 9;18(1):15. doi: 10.1186/s13058-016-0671-y. PubMed PMID: 26857456; PubMedCentral PMCID: PMC4746828.
32. Deb S, Lakhani SR, Ottini L, Fox SB. *The cancer genetics and pathology of male breast cancer*. *Histopathology*. 2016 Jan;68(1):110-8. doi: 10.1111/his.12862. Review. PubMed PMID: 26768033.
33. Silvestri V, Rizzolo P, Scarnò M, Chillemi G, Navazio AS, Valentini V, Zelli V, Zanna I, Saieva C, Masala G, Bianchi S, Manoukian S, Barile M, Pensotti V, Peterlongo P, Varesco L, Tommasi S, Russo A, Giannini G, Cortesi L, Viel A, Montagna M, Radice P, Palli D, Ottini L. *Novel and known genetic variants for male breast cancer risk at 8q24.21, 9p21.3, 11q13.3 and 14q24.1: results from a multicenter study in Italy*. *Eur J Cancer*. 2015 Nov;51(16):2289-95. doi: 10.1016/j.ejca.2015.07.020. Epub 2015 Aug 3. PubMed PMID: 26248686.

34. Peterlongo P, Catucci I, Colombo M, Caleca L, Mucaki E, Bogliolo M, Marin M, Damiola F, Bernard L, Pensotti V, Volorio S, Dall'Olio V, Meindl A, Bartram C, Sutter C, Surowy H, Sornin V, Dondon MG, Eon-Marchais S, Stoppa-Lyonnet D, Andrieu N, Sinilnikova OM; GENESIS, Mitchell G, James PA, Thompson E; kConFab; SWE-BRCA, Marchetti M, Verzeroli C, Tartari C, Capone GL, Putignano AL, Genuardi M, Medici V, Marchi I, Federico M, Tognazzo S, Matricardi L, Agata S, Dolcetti R, Della Puppa L, Cini G, Gismondi V, Viassolo V, Perfumo C, Mencarelli MA, Baldassarri M, Peissel B, Roversi G, Silvestri V, Rizzolo P, Spina F, Vivianet C, Tibiletti MG, Caligo MA, Gambino G, Tommasi S, Pilato B, Tondini C, Corna C, Bonanni B, Barile M, Osorio A, Benitez J, Balestrino L, Ottini L, Manoukian S, Pierotti MA, Renieri A, Varesco L, Couch FJ, Wang X, Devilee P, Hilbers FS, van Asperen CJ, Viel A, Montagna M, Cortesi L, Diez O, Balmaña J, Hauke J, Schmutzler RK, Papi L, Pujana MA, Lázaro C, Falanga A, Offit K, Vijai J, Campbell I, Burwinkel B, Kvist A, Ehrencrona H, Mazoyer S, Pizzamiglio S, Verderio P, Surralles J, Rogan PK, Radice P. *FANCM c.5791C>T nonsense mutation (rs144567652) induces exon skipping, affects DNA repair activity and is a familial breast cancer risk factor.* **Hum Mol Genet.** 2015 Sep 15;24(18):5345-55. doi: 10.1093/hmg/ddv251. Epub 2015 Jun 30. PubMed PMID: 26130695; PubMed Central PMCID: PMC4550823.
35. Ovarian Cancer Association Consortium, Breast Cancer Association Consortium, and Consortium of Modifiers of BRCA1 and BRCA2, Hollestelle A, van der Baan FH, Berchuck A, Johnatty SE, Aben KK, Agnarsson BA, Aittomäki K, Alducci E, Andrulius IL, Anton-Culver H, Antonenkova NN, Antoniou AC, Apicella C, Arndt V, Arnold N, Arun BK, Arver B, Ashworth A; Australian Ovarian Cancer Study Group, Baglietto L, Balleine R, Bandera EV, Barrowdale D, Bean YT, Beckmann L, Beckmann MW, Benitez J, Berger A, Berger R, Beuselinck B, Bisogna M, Bjorge L, Blomqvist C, Bogdanova NV, Bojesen A, Bojesen SE, Bolla MK, Bonanni B, Brand JS, Brauch H; Breast Cancer Family Register, Brenner H, Brinton L, Brooks-Wilson A, Bruinsma F, Brunet J, Brüning T, Budzilowska A, Bunker CH, Burwinkel B, Butzow R, Buys SS, Caligo MA, Campbell I, Carter J, Chang-Claude J, Chanock SJ, Claes KBM, Collée JM, Cook LS, Couch FJ, Cox A, Cramer D, Cross SS, Cunningham JM, Cybulski C, Czene K, Damiola F, Dansonka Mieszkowska A, Darabi H, de la Hoya M, deFazio A, Dennis J, Devilee P, Dicks EM, Diez O, Doherty JA, Domchek SM, Dorfling CM, Dörk T, Silva IDS, du Bois A, Dumont M, Dunning AM, Duran M, Easton DF, Eccles D, Edwards RP, Ehrencrona H, Ejlerksen B, Ekici AB, Ellis SD; EMBRACE, Engel C, Eriksson M, Fasching PA, Feliubadalo L, Figueira J, Flesch-Janys D, Fletcher O, Fontaine A, Fortuzzi S, Fostira F, Fridley BL, Friebel T, Friedman E, Friel G, Frost D, Garber J, García-Closas M, Gayther SA; GEMO Study Collaborators; GENICA Network, Gentry-Maharaj A, Gerdes AM, Giles GG, Glasspool R, Glendon G, Godwin AK, Goodman MT, Gore M, Greene MH, Grip M, Gronwald J, Gschwantler Kaulich D, Guénél P, Guzman SR, Haeberle L, Haiman CA, Hall P, Halverson SL, Hamann U, Hansen TVO, Harter P, Hartikainen JM, Healey S; HEBON, Hein A, Heitz F, Henderson BE, Herzog J, T Hildebrandt MA, Høgdall CK, Høgdall E, Hogervorst FBL, Hopper JL, Humphreys K, Huzarski T, Imyanitov EN, Isaacs C, Jakubowska A, Janavicius R, Jaworska K, Jensen A, Jensen UB, Johnson N, Jukkola-Vuorinen A, Kabisch M, Karlan BY, Kataja V, Kauff N; KConFab Investigators, Kelemen LE, Kerin MJ, Kiemeney LA, Kjaer SK, Knight JA, Knol-Bout JP, Konstantopoulou I, Kosma VM, Krakstad C, Kristensen V, Kuchenbaecker KB, Kupryjanczyk J, Laitman Y, Lambrechts D, Lambrechts S, Larson MC, Lasa A, Laurent-Puig P, Lazaro C, Le ND, Le Marchand L, Leminen A, Lester J, Levine DA, Li J, Liang D, Lindblom A, Lindor N, Lissowska J, Long J, Lu KH, Lubinski J, Lundvall L, Lurie G, Mai PL, Mannermaa A, Margolin S, Mariette F, Marme F, Martens JWM, Massuger LFAG, Maugard C, Mazoyer S, McGuffog L, McGuire V, McLean C, McNeish I, Meindl A, Menegaux F, Menéndez P, Menkiszak J, Menon U, Mensenkamp AR, Miller N, Milne RL, Modugno F, Montagna M, Moysich KB, Müller H, Mulligan AM, Muranen TA, Narod SA, Nathanson KL, Ness RB, Neuhausen SL, Nevanlinna H, Neven P, Nielsen FC, Nielsen SF, Nordestgaard BG, Nussbaum RL, Odunsi K, Offit K, Olah E, Olopade OI, Olson JE, Olson SH, Oosterwijk JC, Orlow I, Orr N, Orsulic S, Osorio A, Ottini L, Paul J, Pearce CL, Pedersen IS, Peissel B, Pejovic T, Peittari LM, Perkins J, Permuth-Wey J, Peterlongo P, Peto J, Phelan CM, Phillips KA, Piedmonte M, Pike MC, Platte R, Plsiecka-Halasa J, Poole EM, Poppe B, Pylkäs K, Radice P, Ramus SJ, Rebbeck TR, Reed MWR, Rennert G, Risch HA, Robson M, Rodriguez GC, Romero A, Rossing MA, Rothstein JH, Rudolph A, Runnebaum I, Salani R, Salvesen HB, Sawyer EJ, Schildkraut JM, Schmidt MK, Schmutzler RK, Schneeweiss A, Schoemaker MJ, Schrauder MG, Schumacher F, Schwaab I, Scuvera G, Sellers TA, Severi G, Seynaeve CM, Shah M, Shrubsole M, Siddiqui N, Sieh W, Simard J, Singer CF, Sinilnikova OM, Smeets D, Sohn C, Soller M, Song H, Soucy P, Southey MC, Stegmaier C, Stoppa-Lyonnet D, Sucheston L; SWE-BRCA, Swerdlow A, Tangen IL, Tea MK, Teixeira MR, Terry KL, Terry MB, Thomassen M, Thompson PJ, Tihomirova L, Tischkowitz M, Toland AE, Tollenaar RAEM, Tomlinson I, Torres D, Truong T, Tsimiklis H, Tung N, Tworoger SS, Tyrer JP, Vachon CM, Van 't Veer LJ, van Altena AM, Van Asperen CJ, van den Berg D, van den Ouwendijk AMW, van Doorn HC, Van Nieuwenhuysen E, van Rensburg EJ, Vergote I, Verhoef S, Vierkant RA, Vijai J, Vitonis AF, von Wachenfeldt A, Walsh C, Wang Q, Wang-Gohrke S, Wappenschmidt B, Weischer M, Weitzel JN, Weltens C, Wentzensen N, Whittemore AS, Wilkens LR, Winquist R, Wu AH, Wu X, Yang HP, Zaffaroni D, Pilar Zamora M, Zheng W, Ziogas A, Chenevix-Trench G, Pharoah PDP, Rookus MA, Hooning MJ, Goode EL. *No clinical utility of KRAS variant rs61764370 for ovarian or breast cancer.* **Gynecol Oncol.** 2016 May;141(2):386-401. doi: 10.1016/j.ygyno.2015.04.034. Epub 2015 May 2. Review. PubMed PMID: 25940428; PubMed Central PMCID: PMC4630206.
36. Blein S, Bardel C, Danjean V, McGuffog L, Healey S, Barrowdale D, Lee A, Dennis J, Kuchenbaecker KB, Soucy P, Terry MB, Chung WK, Goldgar DE, Buys SS; Breast Cancer Family Registry, Janavicius R, Tihomirova L, Tung N, Dorfling CM, van Rensburg EJ, Neuhausen SL, Ding YC, Gerdes AM, Ejlerksen B, Nielsen FC, Hansen TV, Osorio A, Benitez J, Conejero RA, Segota E, Weitzel JN, Thelander M, Peterlongo P, Radice P, Pensotti V, Dolcetti R, Bonanni B, Peissel B, Zaffaroni D, Scuvera G, Manoukian S, Varesco L, Capone GL, Papi L, Ottini L, Yannoukakos D, Konstantopoulou I, Garber J, Hamann U, Donaldson A, Brady A, Brewer C, Foo C, Evans DG, Frost D, Eccles D; EMBRACE, Douglas F, Cook J, Adlard J, Barwell J, Walker L, Izatt L, Side LE, Kennedy MJ, Tischkowitz M, Rogers MT, Porteous ME, Morrison PJ, Platte R, Eeles R, Davidson R, Hodgson S, Cole T, Godwin AK, Isaacs C, Claes K, De Leeneer K, Meindl A, Gehrig A, Wappenschmidt B, Sutter C, Engel C,

- Niederacher D, Steinemann D, Plendl H, Kast K, Rhiem K, Ditsch N, Arnold N, Varon-Mateeva R, Schmutzler RK, Preisler-Adams S, Markov NB, Wang-Gohrke S, de Pauw A, Lefol C, Lasset C, Leroux D, Rouleau E, Damiola F; GEMO Study Collaborators, Dreyfus H, Barjhoux L, Golmard L, Uhrhammer N, Bonadona V, Sornin V, Bignon YJ, Carter J, Van Le L, Piedmonte M, DiSilvestro PA, de la Hoya M, Caldes T, Nevanlinna H, Aittomäki K, Jager A, van den Ouweland AM, Kets CM, Aalfs CM, van Leeuwen FE, Hogervorst FB, Meijers-Heijboer HE; HEBON, Oosterwijk JC, van Roozendaal KE, Rookus MA, Devilee P, van der Luijt RB, Olah E, Diez O, Teulé A, Lazaro C, Blanco I, Del Valle J, Jakubowska A, Sukiennicki G, Gronwald J, Lubinski J, Durda K, Jaworska-Bieniek K, Agnarsson BA, Maugard C, Amadori A, Montagna M, Teixeira MR, Spurdle AB, Foulkes W, Olswold C, Lindor NM, Pankratz VS, Szabo CI, Lincoln A, Jacobs L, Corines M, Robson M, Vijai J, Berger A, Fink-Retter A, Singer CF, Rappaport C, Kaulich DG, Pfeiler G, Tea MK, Greene MH, Mai PL, Rennert G, Imyanitov EN, Mulligan AM, Glendon G, Andrulis IL, Tchatchou S, Toland AE, Pedersen IS, Thomassen M, Kruse TA, Jensen UB, Caligo MA, Friedman E, Zidan J, Laitman Y, Lindblom A, Melin B, Arver B, Loman N, Rosenquist R, Olopade OI, Nussbaum RL, Ramus SJ, Nathanson KL, Domchek SM, Rebbeck TR, Arun BK, Mitchell G, Karlan BY, Lester J, Orsulic S, Stoppa-Lyonnet D, Thomas G, Simard J, Couch FJ, Offit K, Easton DF, Chenevix-Trench G, Antoniou AC, Mazoyer S, Phelan CM, Sinilnikova OM, Cox DG. *An original phylogenetic approach identified mitochondrial haplogroup T1a1 as inversely associated with breast cancer risk in BRCA2 mutation carriers.* **Breast Cancer Res.** 2015 Apr 25;17:61. doi: 10.1186/s13058-015-0567-2. PubMed PMID: 25925750; PubMed Central PMCID: PMC4478717.
37. Blanco I, Kuchenbaecker K, Cuadras D, Wang X, Barrowdale D, de Garibay GR, Librado P, Sánchez-Gracia A, Rozas J, Bonifaci N, McGuffog L, Pankratz VS, Islam A, Mateo F, Berenguer A, Petit A, Català I, Brunet J, Feliubadaló L, Tornero E, Benítez J, Osorio A, Ramón y Cajal T, Nevanlinna H, Aittomäki K, Arun BK, Toland AE, Karlan BY, Walsh C, Lester J, Greene MH, Mai PL, Nussbaum RL, Andrulis IL, Domchek SM, Nathanson KL, Rebbeck TR, Barkardottir RB, Jakubowska A, Lubinski J, Durda K, Jaworska-Bieniek K, Claes K, Van Maerken T, Diez O, Hansen TV, Jønson L, Gerdes AM, Ejlertsen B, de la Hoya M, Caldés T, Dunning AM, Oliver C, Fineberg E, Cook M, Peock S, McCann E, Murray A, Jacobs C, Pichert G, Laloo F, Chu C, Dorkins H, Paterson J, Ong KR, Teixeira MR; Teixeira, Hogervorst FB, van der Hout AH, Seynaeve C, van der Luijt RB, Ligtenberg MJ, Devilee P, Wijnen JT, Rookus MA, Meijers-Heijboer HE, Blok MJ, van den Ouweland AM, Aalfs CM, Rodriguez GC, Phillips KA, Piedmonte M, Nerenstone SR, Bae-Jump VL, O'Malley DM, Ratner ES, Schmutzler RK, Wappenschmidt B, Rhiem K, Engel C, Meindl A, Ditsch N, Arnold N, Plendl HJ, Niederacher D, Sutter C, Wang-Gohrke S, Steinemann D, Preisler-Adams S, Kast K, Varon-Mateeva R, Gehrig A, Bojesen A, Pedersen IS, Sunde L, Jensen UB, Thomassen M, Kruse TA, Foretova L, Peterlongo P, Bernard L, Peissel B, Scuvera G, Manoukian S, Radice P, Ottini L, Montagna M, Agata S, Maugard C, Simard J, Soucy P, Berger A, Fink-Retter A, Singer CF, Rappaport C, Geschwantler-Kaulich D, Tea MK, Pfeiler G; BCFR, John EM, Miron A, Neuhausen SL, Terry MB, Chung WK, Daly MB, Goldgar DE, Janavicius R, Dorfling CM, van Rensburg EJ, Fostira F, Konstantopoulou I, Garber J, Godwin AK, Olah E, Narod SA, Rennert G, Paluch SS, Laitman Y, Friedman E; SWEBRCA, Liljegren A, Rantala J, Stenmark-Askmalm M, Loman N, Imyanitov EN, Hamann U; kConFab Investigators, Spurdle AB, Healey S, Weitzel JN, Herzog J, Margileth D, Gorrini C, Esteller M, Gómez A, Sayols S, Vidal E, Heyn H; GEMO, Stoppa-Lyonnet D, Léoné M, Barjhoux L, Fassy-Colcombet M, de Pauw A, Lasset C, Ferrer SF, Castera L, Berthet P, Cornelis F, Bignon YJ, Damiola F, Mazoyer S, Sinilnikova OM, Maxwell CA, Vijai J, Robson M, Kauff N, Corines MJ, Villano D, Cunningham J, Lee A, Lindor N, Lázaro C, Easton DF, Offit K, Chenevix-Trench G, Couch FJ, Antoniou AC, Pujana MA. *Assessing associations between the AURKA-HMMR-TPX2-TUBG1 functional module and breast cancer risk in BRCA1/2 mutation carriers.* **PLoS One.** 2015 Apr 1;10(4):e0120020. doi: 10.1371/journal.pone.0120020. eCollection 2015. PubMed PMID: 25830658; PubMed Central PMCID: PMC4382299.
38. Ottini L, Rizzolo P, Siniscalchi E, Zijno A, Silvestri V, Crebelli R, Marcon F. *Gene promoter methylation and DNA repair capacity in monozygotic twins with discordant smoking habits.* **Mutat Res Genet Toxicol Environ Mutagen.** 2015 Feb;779:57-64. doi: 10.1016/j.mrgentox.2015.01.006. Epub 2015 Jan 15. PubMed PMID: 25813726.
39. Kuchenbaecker KB, Ramus SJ, Tyrer J, Lee A, Shen HC, Beesley J, Lawrenson K, McGuffog L, Healey S, Lee JM, Spindler TJ, Lin YG, Pejovic T, Bean Y, Li Q, Coetzee S, Hazelett D, Miron A, Southey M, Terry MB, Goldgar DE, Buys SS, Janavicius R, Dorfling CM, van Rensburg EJ, Neuhausen SL, Ding YC, Hansen TV, Jønson L, Gerdes AM, Ejlertsen B, Barrowdale D, Dennis J, Benitez J, Osorio A, Garcia MJ, Komenaka I, Weitzel JN, Ganschow P, Peterlongo P, Bernard L, Viel A, Bonanni B, Peissel B, Manoukian S, Radice P, Papi L, Ottini L, Fostira F, Konstantopoulou I, Garber J, Frost D, Perkins J, Platte R, Ellis S; EMBRACE, Godwin AK, Schmutzler RK, Meindl A, Engel C, Sutter C, Sinilnikova OM; GEMO Study Collaborators, Damiola F, Mazoyer S, Stoppa-Lyonnet D, Claes K, De Leeneer K, Kirk J, Rodriguez GC, Piedmonte M, O'Malley DM, de la Hoya M, Caldés T, Aittomäki K, Nevanlinna H, Collée JM, Rookus MA, Oosterwijk JC; Breast Cancer Family Registry, Tihomirova L, Tung N, Hamann U, Isaccs C, Tischkowitz M, Imyanitov EN, Caligo MA, Campbell IG, Hogervorst FB; HEBON, Olah E, Diez O, Blanco I, Brunet J, Lazaro C, Pujana MA, Jakubowska A, Gronwald J, Lubinski J, Sukiennicki G, Barkardottir RB, Plante M, Simard J, Soucy P, Montagna M, Tognazzo S, Teixeira MR; kConFab Investigators, Pankratz VS, Wang X, Lindor N, Szabo CI, Kauff N, Vijai J, Aghajanian CA, Pfeiler G, Berger A, Singer CF, Tea MK, Phelan CM, Greene MH, Mai PL, Rennert G, Mulligan AM, Tchatchou S, Andrulis IL, Glendon G, Toland AE, Jensen UB, Kruse TA, Thomassen M, Bojesen A, Zidan J, Friedman E, Laitman Y, Soller M, Liljegren A, Arver B, Einbeigi Z, Stenmark-Askmalm M, Olopade OI, Nussbaum RL, Rebbeck TR, Nathanson KL, Domchek SM, Lu KH, Karlan BY, Walsh C, Lester J; Australian Cancer Study (Ovarian Cancer Investigators); Australian Ovarian Cancer Study Group, Hein A, Ekici AB, Beckmann MW, Fasching PA, Lambrechts D, Van Nieuwenhuysen E, Vergote I, Lambrechts S, Dicks E, Doherty JA, Wicklund KG, Rossing MA, Rudolph A, Chang-Claude J, Wang-Gohrke S, Eilber U, Moysich KB, Odunsi K, Sucheston L, Lele S, Wilkens LR, Goodman MT, Thompson PJ, Shvetsov YB, Runnebaum IB, Dürst M, Hillemanns P, Dörk T, Antonenková N, Bogdanova N, Leminen A, Pelttari LM, Butzow R,

- Modugno F, Kelley JL, Edwards RP, Ness RB, du Bois A, Heitz F, Schwaab I, Harter P, Matsuo K, Hosono S, Orsulic S, Jensen A, Kjaer SK, Hogdall E, Hasmad HN, Azmi MA, Teo SH, Woo YL, Fridley BL, Goode EL, Cunningham JM, Vierkant RA, Bruinsma F, Giles GG, Liang D, Hildebrandt MA, Wu X, Levine DA, Bisogna M, Berchuck A, Iversen ES, Schildkraut JM, Concannon P, Weber RP, Cramer DW, Terry KL, Poole EM, Tworoger SS, Bandera EV, Orlow I, Olson SH, Krakstad C, Salvesen HB, Tangen IL, Bjorge L, van Altena AM, Aben KK, Kiemeney LA, Massuger LF, Kellar M, Brooks-Wilson A, Kelemen LE, Cook LS, Le ND, Cybulski C, Yang H, Lissowska J, Brinton LA, Wentzensen N, Hogdall C, Lundvall L, Nedergaard L, Baker H, Song H, Eccles D, McNeish I, Paul J, Carty K, Siddiqui N, Glasspool R, Whittemore AS, Rothstein JH, McGuire V, Sieh W, Ji BT, Zheng W, Shu XO, Gao YT, Rosen B, Risch HA, McLaughlin JR, Narod SA, Monteiro AN, Chen A, Lin HY, Permuth-Wey J, Sellers TA, Tsai YY, Chen Z, Ziogas A, Anton-Culver H, Gentry-Maharaj A, Menon U, Harrington P, Lee AW, Wu AH, Pearce CL, Coetze G, Pike MC, Dansonka-Mieszkowska A, Timorek A, Rzepecka IK, Kupryjanczyk J, Freedman M, Noushmehr H, Easton DF, Offit K, Couch FJ, Gayther S, Pharoah PP, Antoniou AC, Chenevix-Trench G; Consortium of Investigators of Modifiers of BRCA1 and BRCA2. *Identification of six new susceptibility loci for invasive epithelial ovarian cancer*. **Nat Genet.** 2015 Feb;47(2):164-71. doi: 10.1038/ng.3185. Epub 2015 Jan 12. PubMed PMID: 25581431; PubMed Central PMCID: PMC4445140.
40. Di Vincenzo F, Carbone I, Ottini L, Profico A, Ricci F, Tafuri MA, Fornaciari G, Manzi G. *Modern beams for ancient mummies computerized tomography of the holocene mummified remains from wadi takarkori (acacus, south-western libya; Middle pastoral)*. **Med Secoli.** 2015;27(2):575-88. PubMed PMID: 26946601.
  41. Ottini L, Rizzolo P, Zanna I, Silvestri V, Saieva C, Falchetti M, Masala G, Navazio AS, Capalbo C, Bianchi S, Manoukian S, Barile M, Peterlongo P, Caligo MA, Varesco L, Tommasi S, Russo A, Giannini G, Cortesi L, Cini G, Montagna M, Radice P, Palli D. *Association of SULT1A1 Arg<sup>213</sup>His polymorphism with male breast cancer risk: results from a multicenter study in Italy*. **Breast Cancer Res Treat.** 2014 Dec;148(3):623-8. doi: 10.1007/s10549-014-3193-2. Epub 2014 Nov 11. PubMed PMID: 25385181.
  42. Ottini L. *Male breast cancer: a rare disease that might uncover underlying pathways of breast cancer*. **Nat Rev Cancer.** 2014 Oct;14(10):643. doi: 10.1038/nrc3806. PubMed PMID: 25379579.
  43. Osorio A, Milne RL, Kuchenbaecker K, Vaclová T, Pita G, Alonso R, Peterlongo P, Blanco I, de la Hoya M, Duran M, Díez O, Ramón Y Cajal T, Konstantopoulou I, Martínez-Bouzas C, Andrés Conejero R, Soucy P, McGuffog L, Barrowdale D, Lee A, Swe-Brca, Arver B, Rantala J, Loman N, Ehrencrona H, Olopade OI, Beattie MS, Domchek SM, Nathanson K, Rebbeck TR, Arun BK, Karlan BY, Walsh C, Lester J, John EM, Whittemore AS, Daly MB, Southey M, Hopper J, Terry MB, Buys SS, Janavicius R, Dorfling CM, van Rensburg EJ, Steele L, Neuhausen SL, Ding YC, Hansen TV, Jønson L, Ejlertsen B, Gerdes AM, Infante M, Herráez B, Moreno LT, Weitzel JN, Herzog J, Weeman K, Manoukian S, Peissel B, Zaffaroni D, Scuvera G, Bonanni B, Mariette F, Volorio S, Viel A, Varesco L, Papi L, Ottini L, Tibiletti MG, Radice P, Yannoukakos D, Garber J, Ellis S, Frost D, Platte R, Fineberg E, Evans G, Laloo F, Izatt L, Eeles R, Adlard J, Davidson R, Cole T, Eccles D, Cook J, Hodgson S, Brewer C, Tischkowitz M, Douglas F, Porteous M, Side L, Walker L, Morrison P, Donaldson A, Kennedy J, Foo C, Godwin AK, Schmutzler RK, Wappenschmidt B, Rhiem K, Engel C, Meindl A, Ditsch N, Arnold N, Plendl HJ, Niederacher D, Sutter C, Wang-Gohrke S, Steinemann D, Preisler-Adams S, Kast K, Varon-Mateeva R, Gehrig A, Stoppa-Lyonnet D, Sinilnikova OM, Mazoyer S, Damiola F, Poppe B, Claes K, Piedmonte M, Tucker K, Backes F, Rodríguez G, Brewster W, Wakeley K, Rutherford T, Caldés T, Nevanlinna H, Aittomäki K, Rookus MA, van Os TA, van der Kolk L, de Lange JL, Meijers-Heijboer HE, van der Hout AH, van Asperen CJ, Gómez Garcia EB, Hoogerbrugge N, Collée JM, van Deurzen CH, van der Luijt RB, Devilee P, Hebon, Olah E, Lázaro C, Teulé A, Menéndez M, Jakubowska A, Cybulski C, Gronwald J, Lubinski J, Durda K, Jaworska-Bieniek K, Johannsson OT, Maugard C, Montagna M, Tognazzo S, Teixeira MR, Healey S, Investigators K, Olswold C, Guidugli L, Lindor N, Slager S, Szabo CI, Vijai J, Robson M, Kauff N, Zhang L, Rau-Murthy R, Fink-Retter A, Singer CF, Rappaport C, Geschwantler Kaulich D, Pfeiler G, Tea MK, Berger A, Phelan CM, Greene MH, Mai PL, Lejbkowicz F, Andrusis I, Mulligan AM, Glendon G, Toland AE, Bojesen A, Pedersen IS, Sunde L, Thomassen M, Kruse TA, Jensen UB, Friedman E, Laitman Y, Shimon SP, Simard J, Easton DF, Offit K, Couch FJ, Chenevix-Trench G, Antoniou AC, Benitez J. *DNA glycosylases involved in base excision repair may be associated with cancer risk in BRCA1 and BRCA2 mutation carriers*. **PLoS Genet.** 2014 Apr 3;10(4):e1004256. doi: 10.1371/journal.pgen.1004256. eCollection 2014 Apr. PubMed PMID: 24698998; PubMed Central PMCID: PMC3974638.
  44. Richetta AG, Silvestri V, Giancristoforo S, Rizzolo P, D'Epiro S, Graziano V, Mattozzi C, Navazio AS, Falchetti M, Calvieri S, Ottini L. *Mutational profiling in melanocytic tumors: multiple somatic mutations and clinical implications*. **Oncology.** 2014;86(2):104-8. doi: 10.1159/000356878. Epub 2014 Jan 18. PubMed PMID: 24457427.
  45. Richetta AG, Silvestri V, Giancristoforo S, Rizzolo P, D'Epiro S, Graziano V, Mattozzi C, Navazio AS, Campoli M, D'Amico C, Scarnò M, Calvieri S, Ottini L. *A-1012G promoter polymorphism of vitamin D receptor gene is associated with psoriasis risk and lower allele-specific expression*. **DNA Cell Biol.** 2014 Feb;33(2):102-9. doi: 10.1089/dna.2013.2217. Epub 2013 Dec 9. PubMed PMID: 24320988.
  46. Rizzolo P, Silvestri V, Tommasi S, Pinto R, Danza K, Falchetti M, Gulino M, Frati P, Ottini L. *Male breast cancer: genetics, epigenetics, and ethical aspects*. **Ann Oncol.** 2013 Nov;24 Suppl 8:vii75-viii82. doi: 10.1093/annonc/mdt316. Review. PubMed PMID: 24131976.

47. Palli D, Rizzolo P, Zanna I, Silvestri V, Saieva C, Falchetti M, Navazio AS, Graziano V, Masala G, Bianchi S, Russo A, Tommasi S, **Ottini L**. *SULT1A1 gene deletion in BRCA2-associated male breast cancer: a link between genes and environmental exposures?* *J Cell Mol Med.* 2013 May;17(5):605-7. doi: 10.1111/jcmm.12043. PubMed PMID: 23711090; PubMed Central PMCID: PMC3822812.
48. Couch FJ, Wang X, McGuffog L, Lee A, Olswold C, Kuchenbaecker KB, Soucy P, Fredericksen Z, Barrowdale D, Dennis J, Gaudet MM, Dicks E, Kosei M, Healey S, Sinilnikova OM, Lee A, Bacot F, Vincent D, Hogervorst FB, Peock S, Stoppa-Lyonnet D, Jakubowska A; kConFab Investigators, Radice P, Schmutzler RK; SWE-BRCA, Domchek SM, Piedmonte M, Singer CF, Friedman E, Thomassen M; Ontario Cancer Genetics Network, Hansen TV, Neuhausen SL, Szabo CI, Blanco I, Greene MH, Karlan BY, Garber J, Phelan CM, Weitzel JN, Montagna M, Olah E, Andrusilis IL, Godwin AK, Yannoukakos D, Goldgar DE, Caldes T, Nevanlinna H, Osorio A, Terry MB, Daly MB, van Rensburg EJ, Hamann U, Ramus SJ, Toland AE, Caligo MA, Olopade OI, Tung N, Claes K, Beattie MS, Southey MC, Imyanitov EN, Tischkowitz M, Janavicius R, John EM, Kwong A, Diez O, Balmaña J, Barkardottir RB, Arun BK, Rennert G, Teo SH, Ganz PA, Campbell I, van der Hout AH, van Deurzen CH, Seynaeve C, Gómez Garcia EB, van Leeuwen FE, Meijers-Heijboer HE, Gille JJ, Ausems MG, Blok MJ, Ligtenberg MJ, Rookus MA, Devilee P, Verhoef S, van Os TA, Wijnen JT; HEBON; EMBRACE, Frost D, Ellis S, Fineberg E, Platte R, Evans DG, Izatt L, Eeles RA, Adlard J, Eccles DM, Cook J, Brewer C, Douglas F, Hodgson S, Morrison PJ, Side LE, Donaldson A, Houghton C, Rogers MT, Dorkins H, Eason J, Gregory H, McCann E, Murray A, Calender A, Hardouin A, Berthet P, Delnatte C, Nogues C, Lasset C, Houdayer C, Leroux D, Rouleau E, Prieur F, Damiola F, Sobol H, Couper I, Venat-Bouvet L, Castera L, Gauthier-Villars M, Léoné M, Pujol P, Mazoyer S, Bignon YJ; GEMO Study Collaborators, Ziolkowska-Perłowska E, Gronwald J, Lubinski J, Durda K, Jaworska K, Huzarski T, Spurdle AB, Viel A, Peissel B, Bonanni B, Melloni G, **Ottini L**, Papi L, Varesco L, Tibiletti MG, Peterlongo P, Volorio S, Manoukian S, Pensotti V, Arnold N, Engel C, Deissler H, Gadzicki D, Gehrig A, Kast K, Rhiem K, Meindl A, Niederacher D, Ditsch N, Plendl H, Preisler-Adams S, Engert S, Sutter C, Varon-Mateeva R, Wappenschmidt B, Weber BH, Arver B, Stenmark-Askmalm M, Loman N, Rosenquist R, Einbeigi Z, Nathanson KL, Rebbeck TR, Blank SV, Cohn DE, Rodriguez GC, Small L, Friedlander M, Bae-Jump VL, Fink-Retter A, Rappaport C, Gschwantler-Kaulich D, Pfeiler G, Tea MK, Lindor NM, Kaufman B, Shimon Paluch S, Laitman Y, Skytte AB, Gerdes AM, Pedersen IS, Moeller ST, Kruse TA, Jensen UB, Vijai J, Sarrel K, Robson M, Kauff N, Mulligan AM, Glendon G, Ozcelik H, Ejlertsen B, Nielsen FC, Jønson L, Andersen MK, Ding YC, Steele L, Foretova L, Teulé A, Lazaro C, Brunet J, Pujana MA, Mai PL, Loud JT, Walsh C, Lester J, Orsulic S, Narod SA, Herzog J, Sand SR, Tognazzo S, Agata S, Vaszko T, Weaver J, Stavropoulou AV, Buys SS, Romero A, de la Hoya M, Aittomäki K, Muranen TA, Duran M, Chung WK, Lasa A, Dorfling CM, Miron A; BCFR, Benitez J, Senter L, Huo D, Chan SB, Sokolenko AP, Chiquette J, Tihomirova L, Friebel TM, Agnarsson BA, Lu KH, Lejbkowicz F, James PA, Hall P, Dunning AM, Tessier D, Cunningham J, Slager SL, Wang C, Hart S, Stevens K, Simard J, Pastinen T, Pankratz VS, Offit K, Easton DF, Chenevix-Trench G, Antoniou AC; CIMBA. *Genome-wide association study in BRCA1 mutation carriers identifies novel loci associated with breast and ovarian cancer risk.* *PLoS Genet.* 2013;9(3):e1003212. doi: 10.1371/journal.pgen.1003212. Epub 2013 Mar 27. PubMed PMID: 23544013; PubMed Central PMCID: PMC3609646.
49. **Ottini L**, Silvestri V, Saieva C, Rizzolo P, Zanna I, Falchetti M, Masala G, Navazio AS, Graziano V, Bianchi S, Manoukian S, Barile M, Peterlongo P, D'Amico C, Varesco L, Tommasi S, Russo A, Giannini G, Cortesi L, Viel A, Montagna M, Radice P, Palli D. *Association of low-penetrance alleles with male breast cancer risk and clinicopathological characteristics: results from a multicenter study in Italy.* *Breast Cancer Res Treat.* 2013 Apr;138(3):861-8. doi: 10.1007/s10549-013-2459-4. Epub 2013 Mar 7. PubMed PMID: 23468243.
50. Falchetti M, **Ottini L**. *Fire worship on Soratte: form prehistory to present days.* *Med Secoli.* 2013;25(2):609-36. PubMed PMID: 25807785.
51. **Ottini L**, Franzia A, Rizzolo P, Falchetti M, Santi R, Nesi G. *Medical research through historical resources. Talking objects: a case of a parasitic perineal monstrosity.* *Med Secoli.* 2013;25(1):267-94. PubMed PMID: 25807709.
52. Pinto R, Pilato B, **Ottini L**, Lambo R, Simone G, Paradiso A, Tommasi S. *Different methylation and microRNA expression pattern in male and female familial breast cancer.* *J Cell Physiol.* 2013 Jun;228(6):1264-9. doi: 10.1002/jcp.24281. PubMed PMID: 23160909.
53. Orr N, Lemnrau A, Cooke R, Fletcher O, Tomczyk K, Jones M, Johnson N, Lord CJ, Mitsopoulos C, Zvelebil M, McDade SS, Buck G, Blancher C; KConFab Consortium, Trainer AH, James PA, Bojesen SE, Bokmand S, Nevanlinna H, Mattson J, Friedman E, Laitman Y, Palli D, Masala G, Zanna I, **Ottini L**, Giannini G, Hollestelle A, Ouwendijk AM, Novaković S, Krajc M, Gago-Dominguez M, Castelao JE, Olsson H, Hedenfalk I, Easton DF, Pharoah PD, Dunning AM, Bishop DT, Neuhausen SL, Steele L, Houlston RS, Garcia-Closas M, Ashworth A, Swerdlow AJ. *Genome-wide association study identifies a common variant in RAD51B associated with male breast cancer risk.* *Nat Genet.* 2012 Nov;44(11):1182-4. doi: 10.1038/ng.2417. Epub 2012 Sep 23. PubMed PMID: 23001122; PubMed Central PMCID: PMC3722904.
54. **Ottini L**, Silvestri V, Rizzolo P, Falchetti M, Zanna I, Saieva C, Masala G, Bianchi S, Manoukian S, Barile M, Peterlongo P, Varesco L, Tommasi S, Russo A, Giannini G, Cortesi L, Viel A, Montagna M, Radice P, Palli D. *Clinical and pathologic characteristics of BRCA-positive and BRCA-negative male breast cancer patients: results from a collaborative multicenter*

55. Antoniou AC, Kuchenbaecker KB, Soucy P, Beesley J, Chen X, McGuffog L, Lee A, Barrowdale D, Healey S, Sinilnikova OM, Caligo MA, Loman N, Harbst K, Lindblom A, Arver B, Rosenquist R, Karlsson P, Nathanson K, Domchek S, Rebbeck T, Jakubowska A, Lubinski J, Jaworska K, Durda K, Złowowcka-Perlowska E, Osorio A, Durán M, Andrés R, Benítez J, Hamann U, Hogervorst FB, van Os TA, Verhoef S, Meijers-Heijboer HE, Wijnen J, Gómez Garcia EB, Ligtenberg MJ, Krieger M, Collée JM, Ausems MG, Oosterwijk JC, Peacock S, Frost D, Ellis SD, Platé R, Fineberg E, Evans DG, Laloo F, Jacobs C, Eeles R, Adlard J, Davidson R, Cole T, Cook J, Paterson J, Douglas F, Brewer C, Hodgson S, Morrison PJ, Walker L, Rogers MT, Donaldson A, Dorkins H, Godwin AK, Bove B, Stoppani-Lyonnet D, Houdayer C, Buecher B, de Pauw A, Mazoyer S, Calender A, Léoné M, Bressac-de Paillerets B, Caron O, Sobol H, Frenay M, Prieur F, Ferrer SU, Mortemousque I, Buys S, Daly M, Miron A, Terry MU, Hopper JL, John EM, Southey M, Goldgar D, Singer CF, Fink-Retter A, Tea MK, Kaulich DU, Hansen TV, Nielsen FC, Barkardottir RB, Gaudet M, Kirchhoff T, Joseph V, Dutra-Clarke A, Offit K, Piedmonte M, Kirk J, Cohn D, Hurteau J, Byron J, Fiorica J, Toland AE, Montagna M, Oliani C, Imanitov E, Isaacs C, Tihomirova L, Blanco I, Lazaro C, Teulé A, Valle JD, Gayther SA, Odunsi K, Gross J, Karlan BY, Olah E, Teo SH, Ganz PA, Beattie MS, Dorfling CM, van Rensburg EU, Diez O, Kwong A, Schmutzler RK, Wappenschmidt B, Engel C, Meindl A, Ditsch N, Arnold N, Heidemann S, Niederacher D, Preisler-Adams S, Gadzicki D, Varon-Mateeva R, Deissler H, Gehrig A, Sutter C, Kast K, Fiebig B, Schäfer D, Caldes T, de la Hoya M, Nevanlinna H, Muranen TA, Lespérance B, Spurdle AB, Neuhausen SL, Ding YC, Wang X, Fredericksen Z, Pankratz VS, Lindor NM, Peterlongo P, Manoukian S, Peissel B, Zaffaroni D, Bonanni B, Bernard L, Dolcetti R, Papi L, Ottini L, Radice P, Greene MH, Loud JT, Andrusik IL, Ozcelik H, Mulligan AU, Glendon G, Thomassen M, Gerdes AM, Jensen UB, Skytte AB, Kruse TA, Chenevix-Trench G, Couch FJ, Simard J, Easton DF; CIMBA, SWE-BRCA; HEBON; EMBRACE; GEMO Collaborators Study; kConFab Investigators. Common variants at 12p11, 12q24, 9p21, 9q31.2 and in ZNF365 are associated with breast cancer risk for BRCA1 and/or BRCA2 mutation carriers. *Breast Cancer Res*. 2012 Feb 20;14(1):R33. PubMed PMID: 22348646; PubMed Central PMCID: PMC3496151.
56. Ramus SJ, Kartsonaki C, Gayther SA, Pharoah PD, Sinilnikova OM, Beesley J, Chen X, McGuffog L, Healey S, Couch FJ, Wang X, Fredericksen Z, Peterlongo P, Manoukian S, Peissel B, Zaffaroni D, Roversi G, Barile M, Viel A, Allavena A, Ottini L, Papi L, Gismondi V, Capra F, Radice P, Greene MH, Mai PL, Andrusik IL, Glendon G, Ozcelik H; OCGN, Thomassen M, Gerdes AM, Kruse TA, Cruger D, Jensen UB, Caligo MA, Olsson H, Kristoffersson U, Lindblom A, Arver B, Karlsson P, Stenmark Askalm M, Borg A, Neuhausen SL, Ding YC, Nathanson KL, Domchek SM, Jakubowska A, Lubinski J, Huzarski T, Byrski T, Gronwald J, Górska B, Cybulski C, Dębniak T, Osorio A, Durán M, Tejada MI, Benítez J, Hamann U, Rookus MA, Verhoef S, Tilanus-Linthorst MA, Vreeswijk MP, Bodmer D, Ausems MG, van Os TA, Asperen CJ, Blok MJ, Meijers-Heijboer HE; HEBON; EMBRACE, Peacock S, Cook M, Oliver C, Frost D, Dunning AM, Evans DG, Eeles R, Pichert G, Cole T, Hodgson S, Brewer C, Morrison PJ, Porteous M, Kennedy MJ, Rogers MT, Side LE, Donaldson A, Gregory H, Godwin A, Stoppani-Lyonnet D, Moncoutier V, Castera L, Mazoyer S, Barjhoux L, Bonadona V, Leroux D, Faivre L, Lidereau R, Nogues C, Bignon YJ, Prieur F, Collonge-Rame MA, Venat-Bouvet L, Fert-Ferrer S; GEMO Study Collaborators, Miron A, Buys SS, Hopper JL, Daly MB, John EM, Terry MB, Goldgar D; BCFR, Hansen TV, Jønson L, Ejlersen B, Agnarsson BA, Offit K, Kirchhoff T, Vijai J, Dutra-Clarke AV, Przybylo JA, Montagna M, Casella C, Imanitov EN, Janavicius R, Blanco I, Lázaro C, Moysich KB, Karlan BY, Gross J, Beattie MS, Schmutzler R, Wappenschmidt B, Meindl A, Ruehl I, Fiebig B, Sutter C, Arnold N, Deissler H, Varon-Mateeva R, Kast K, Niederacher D, Gadzicki D, Caldes T, de la Hoya M, Nevanlinna H, Aittomäki K, Simard J, Soucy P; kConFab Investigators, Spurdle AB, Holland H, Chenevix-Trench G, Easton DF, Antoniou AC; Consortium of Investigators of Modifiers of BRCA1/2. Ovarian cancer susceptibility alleles and risk of ovarian cancer in BRCA1 and BRCA2 mutation carriers. *Hum Mutat*. 2012 Apr;33(4):690-702. doi: 10.1002/humu.22025. Epub 2012 Feb 14. PubMed PMID: 22253144; PubMed Central PMCID: PMC3458423.
57. Mavaddat N, Barrowdale D, Andrusik IL, Domchek SM, Eccles D, Nevanlinna H, Ramus SJ, Spurdle A, Robson M, Sherman M, Mulligan AM, Couch FJ, Engel C, McGuffog L, Healey S, Sinilnikova OM, Southey MC, Terry MB, Goldgar D, O'Malley F, John EM, Janavicius R, Tihomirova L, Hansen TV, Nielsen FC, Osorio A, Stavropoulou A, Benítez J, Manoukian S, Peissel B, Barile M, Volorio S, Pasini B, Dolcetti R, Putignano AL, Ottini L, Radice P, Hamann U, Rashid MU, Hogervorst FB, Krieger M, van der Luijt RB; HEBON, Peacock S, Frost D, Evans DG, Brewer C, Walker L, Rogers MT, Side LE, Houghton C; EMBRACE, Weaver J, Godwin AK, Schmutzler RK, Wappenschmidt B, Meindl A, Kast K, Arnold N, Niederacher D, Sutter C, Deissler H, Gadzicki D, Preisler-Adams S, Varon-Mateeva R, Schönbuchner I, Gevensleben H, Stoppani-Lyonnet D, Belotti M, Barjhoux L; GEMO Study Collaborators, Isaacs C, Peshkin BN, Caldes T, de la Hoya M, Cañadas C, Heikkilä T, Heikkilä P, Aittomäki K, Blanco I, Lazaro C, Brunet J, Agnarsson BA, Arason A, Barkardottir RB, Dumont M, Simard J, Montagna M, Agata S, D'Andrea E, Yan M, Fox S; kConFab Investigators, Rebbeck TR, Rubinstein W, Tung N, Garber JE, Wang X, Fredericksen Z, Pankratz VS, Lindor NM, Szabo C, Offit K, Sakr R, Gaudet MM, Singer CF, Tea MK, Rappaport C, Mai PL, Greene MH, Sokolenko A, Imanitov E, Toland AE, Senter L, Sweet K, Thomassen M, Gerdes AM, Kruse T, Caligo M, Aretini P, Rantala J, von Wachenfeld A, Henriksson K; SWE-BRCA Collaborators, Steele L, Neuhausen SL, Nussbaum R, Beattie M, Odunsi K, Sucheston L, Gayther SA, Nathanson K, Gross J, Walsh C, Karlan B, Chenevix-Trench G, Easton DF, Antoniou AC; Consortium of Investigators of Modifiers of BRCA1/2. Pathology of breast and ovarian cancers among BRCA1 and BRCA2 mutation carriers: results from the Consortium of Investigators of Modifiers of BRCA1/2 (CIMBA). *Cancer Epidemiol Biomarkers Prev*. 2012 Jan;21(1):134-47. doi: 10.1158/1055-9965.EPI-11-0775. Epub 2011 Dec 5. PubMed PMID: 22144499; PubMed Central PMCID: PMC3272407.

58. Mulligan AM, Couch FJ, Barrowdale D, Domchek SM, Eccles D, Nevanlinna H, Ramus SJ, Robson M, Sherman M, Spurdle AB, Wappenschmidt B, Lee A, McGuffog L, Healey S, Sinilnikova OM, Janavicius R, Hansen Tv, Nielsen FC, Ejlertsen B, Osorio A, Muñoz-Repeta I, Durán M, Godino J, Pertesi M, Benítez J, Peterlongo P, Manoukian S, Peissel B, Zaffaroni D, Cattaneo E, Bonanni B, Viel A, Pasini B, Papi L, Ottini L, Savarese A, Bernard L, Radice P, Hamann U, Verheus M, Meijers-Heijboer HE, Wijnen J, Gómez García EB, Nelen MR, Kets CM, Seynaeve C, Tilanus-Linthorst MM, van der Luijt RB, van Os T, Rookus M, Frost D, Jones JL, Evans DG, Laloo F, Eeles R, Izatt L, Adlard J, Davidson R, Cook J, Donaldson A, Dorkins H, Gregory H, Eason J, Houghton C, Barwell J, Side LE, McCann E, Murray A, Peock S, Godwin AK, Schmutzler RK, Rhie K, Engel C, Meindl A, Ruehl I, Arnold N, Niederacher D, Sutter C, Deissler H, Gadzicki D, Kast K, Preisler-Adams S, Varon-Mateeva R, Schoenbuchner I, Fiebig B, Heinritz W, Schäfer D, Gevensleben H, Caux-Moncoutier V, Fassy-Colcomet M, Cornelis F, Mazoyer S, Léoné M, Boutry-Kryza N, Hardouin A, Berthet P, Muller D, Fricker JP, Mortemousque I, Pujol P, Couper I, Lebrun M, Kientz C, Longy M, Sevenet N, Stoppa-Lyonnet D, Isaacs C, Caldes T, de la Hoya M, Heikkinen T, Aittomäki K, Blanco I, Lazaro C, Barkardottir RB, Soucy P, Dumont M, Simard J, Montagna M, Tognazzo S, D'Andrea E, Fox S, Yan M, Rebbeck T, Olopade O, Weitzel JN, Lynch HT, Ganz PA, Tomlinson GE, Wang X, Frederickson Z, Pankratz VS, Lindor NM, Szabo C, Offit K, Sakr R, Gaudet M, Bhatia J, Kauff N, Singer CF, Tea MK, Gschwantler-Kaulich D, Fink-Retter A, Mai PL, Greene MH, Imyanitov E, O'Malley FP, Ozcelik H, Glendon G, Toland AE, Gerdes AM, Thomassen M, Kruse TA, Jensen UB, Skytte AB, Caligo MA, Soller M, Henriksson K, Wachenfeldt vA, Arver B, Stenmark-Askmalm M, Karlsson P, Ding YC, Neuhausen SL, Beattie M, Pharoah PD, Moysich KB, Nathanson KL, Karlan BY, Gross J, John EM, Daly MB, Buys SM, Southey MC, Hopper JL, Terry MB, Chung W, Miron AF, Goldgar D, Chenevix Trench G, Easton DF, Andrulis IL, Antoniou AC; Breast Cancer Family Registry; EMBRACE; GEMO Study Collaborators; HEBON; kConFab Investigators; Ontario Cancer Genetics Network; SWE-BRCA; CIMBA. *Common breast cancer susceptibility alleles are associated with tumour subtypes in BRCA1 and BRCA2 mutation carriers: results from the Consortium of Investigators of Modifiers of BRCA1/2.* **Breast Cancer Res.** 2011;13(6):R110. doi: 10.1186/bcr3052. Epub 2011 Nov 2. PubMed PMID: 22053997; PubMed Central PMCID: PMC3326552.
59. Tommasi S, Mangia A, Iannelli G, Chiarappa P, Rossi E, Ottini L, Mottolese M, Zoli W, Zuffardi O, Paradiso A. *Gene copy number variation in male breast cancer by aCGH.* **Cell Oncol (Dordr).** 2011 Oct;34(5):467-73. doi: 10.1007/s13402-011-0041-9. Epub 2011 May 6. Erratum in: *Cell Oncol (Dordr).* 2011 Oct;34(5):509-10. PubMed PMID: 21547577.
60. Martrat G, Maxwell CM, Tominaga E, Porta-de-la-Riva M, Bonifaci N, Gómez-Baldó L, Bogliolo M, Lázaro C, Blanco I, Brunet J, Aguilar H, Fernández-Rodríguez J, Seal S, Renwick A, Rahman N, Kühl J, Neveling K, Schindler D, Ramírez MJ, Castellà M, Hernández G; EMBRACE, Easton DF, Peock S, Cook M, Oliver CT, Frost D, Platte R, Evans DG, Laloo F, Eeles R, Izatt L, Chu C, Davidson R, Ong KR, Cook J, Douglas F, Hodgson S, Brewer C, Morrison PJ, Porteous M, Peterlongo P, Manoukian S, Peissel B, Zaffaroni D, Roversi G, Barile M, Viel A, Pasini B, Ottini L, Putignano AL, Savarese A, Bernard L, Radice P, Healey S, Spurdle A, Chen X, Beesley J; kConFab, Rookus MA, Verhoef S, Tilanus-Linthorst MA, Vreeswijk MP, Asperen CJ, Bodmer D, Ausems MG, van Os TA, Blok MJ, Meijers-Heijboer HE, Hogervorst FB; HEBON, Goldgar DE, Buys S, John EM, Miron A, Southey M, Daly MB; BCFR; SWE-BRCA, Harbst K, Borg A, Rantala J, Barbany-Bustinza G, Ehrencreutz H, Stenmark-Askmalm M, Kaufman B, Laitman Y, Milgrom R, Friedman E, Domchek SM, Nathanson KL, Rebbeck TR, Johannsson OT, Couch FJ, Wang X, Frederickson Z, Cuadras D, Moreno V, Pientka FK, Depping R, Caldés T, Osorio A, Benítez J, Bueren J, Heikkinen T, Nevanlinna H, Hamann U, Torres D, Caligo MA, Godwin AK, Imyanitov EN, Janavicius R; GEMO Study Collaborators, Sinilnikova OM, Stoppa-Lyonnet D, Mazoyer S, Verny-Pierre C, Castera L, de Pauw A, Bignon YJ, Uhrhammer N, Peyrat JP, Vennin P, Ferrer SF, Collonge-Rame MA, Mortemousque I, McGuffog L, Chenevix-Trench G, Pereira-Smith OM, Antoniou AC, Cerón J, Tominaga K, Surrallés J, Pujana MA. *Exploring the link between MORF4L1 and risk of breast cancer.* **Breast Cancer Res.** 2011 Apr 5;13(2):R40. doi: 10.1186/bcr2862. PubMed PMID: 21466675; PubMed Central PMCID: PMC3219203.
61. Silvestri V, Rizzolo P, Falchetti M, Zanna I, Masala G, Palli D, Ottini L. *Mutation screening of RAD51C in male breast cancer patients.* **Breast Cancer Res.** 2011 Feb 8;13(1):404. doi: 10.1186/bcr2823. PubMed PMID: 21392410; PubMed Central PMCID: PMC3109583.
62. Palmirotta R, Guadagni F, Savonarola A, Ludovici G, De Marchis ML, Palli D, Falchetti M, Ottini L. *PRKCSH GAG trinucleotide repeat is a mutational target in gastric carcinomas with high-level microsatellite instability.* **Clin Genet.** 2011 Apr;79(4):397-8; author reply 399-400. doi: 10.1111/j.1399-0004.2010.01536.x. PubMed PMID: 21371016.
63. Palmirotta R, Guadagni F, Savonarola A, Ludovici G, Nesi G, Palli D, Falchetti M, Ottini L. *PAI-1 4G/5G repeat is a target in gastric carcinomas with microsatellite instability.* **Dig Liver Dis.** 2011 Jun;43(6):454-8. doi: 10.1016/j.dld.2010.12.002. Epub 2011 Jan 8. PubMed PMID: 21216681.
64. Rizzolo P, Silvestri V, Falchetti M, Ottini L. *Inherited and acquired alterations in development of breast cancer.* **Appl Clin Genet.** 2011 Nov 14;4:145-58. doi: 10.2147/TACG.S13226. Print 2011. PubMed PMID: 23776375; PubMed Central PMCID: PMC3681186.
65. Falchetti M, Ottini L. *Water cults on Soratte Mount.* **Med Secoli.** 2011;23(3):947-62. PubMed PMID: 23057207.

66. Ramus SJ, Kartsonaki C, Gayther SA, Pharoah PD, Sinilnikova OM, Beesley J, Chen X, McGuffog L, Healey S, Couch FJ, Wang X, Fredericksen Z, Peterlongo P, Manoukian S, Peissel B, Zaffaroni D, Roversi G, Barile M, Viel A, Allavena A, **Ottini L**, Papi L, Gismondi V, Capra F, Radice P, Greene MH, Mai PL, Andrulis IL, Glendon G, Ozcelik H; OCGN, Thomassen M, Gerdes AM, Kruse TA, Cruger D, Jensen UB, Caligo MA, Olsson H, Kristoffersson U, Lindblom A, Arver B, Karlsson P, Stenmark Askalm M, Borg A, Neuhausen SL, Ding YC, Nathanson KL, Domchek SM, Jakubowska A, Lubinski J, Huzarski T, Byrski T, Gronwald J, Górska B, Cybulski C, Dębniak T, Osorio A, Durán M, Tejada MI, Benítez J, Hamann U, Rookus MA, Verhoef S, Tilanus-Linthorst MA, Vreeswijk MP, Bodmer D, Ausems MG, van Os TA, Asperen CJ, Blok MJ, Meijers-Heijboer HE; HEBON; EMBRACE, Peacock S, Cook M, Oliver C, Frost D, Dunning AM, Evans DG, Eeles R, Pichert G, Cole T, Hodgson S, Brewer C, Morrison PJ, Porteous M, Kennedy MJ, Rogers MT, Side LE, Donaldson A, Gregory H, Godwin A, Stoppa-Lyonnet D, Moncoubier V, Castera L, Mazoyer S, Barjhoux L, Bonadona V, Leroux D, Faivre L, Lidereau R, Nogues C, Bignon YJ, Prieur F, Collonge-Rame MA, Venat-Bouvet L, Fert-Ferrer S; GEMO Study Collaborators, Miron A, Buys SS, Hopper JL, Daly MB, John EM, Terry MB, Goldgar D; BCFR, Hansen Tv, Jönson L, Ejlertsen B, Agnarsson BA, Offit K, Kirchhoff T, Vijai J, Dutra-Clarke AV, Przybylo JA, Montagna M, Casella C, Imanitov EN, Janavicius R, Blanco I, Lázaro C, Moysich KB, Karlan BY, Gross J, Beattie MS, Schmutzler R, Wappenschmidt B, Meindl A, Ruehl I, Fiebig B, Sutter C, Arnold N, Deissler H, Varon-Mateeva R, Kast K, Niederacher D, Gadzicki D, Caldes T, de la Hoya M, Nevanlinna H, Aittomäki K, Simard J, Soucy P; kConFab Investigators, Spurdle AB, Holland H, Chenevix-Trench G, Easton DF, Antoniou AC; Consortium of Investigators of Modifiers of BRCA1/2. *Genetic variation at 9p22.2 and ovarian cancer risk for BRCA1 and BRCA2 mutation carriers*. **J Natl Cancer Inst.** 2011 Jan 19;103(2):105-16. doi: 10.1093/jnci/djq494. Epub 2010 Dec 17. PubMed PMID: 21169536; PubMed Central PMCID: PMC3107565.
67. Silvestri V, Rizzolo P, Falchetti M, Zanna I, Masala G, Bianchi S, Palli D, **Ottini L**. *Mutation analysis of BRIP1 in male breast cancer cases: a population-based study in Central Italy*. **Breast Cancer Res Treat.** 2011 Apr;126(2):539-43. doi: 10.1007/s10549-010-1289-x. Epub 2010 Dec 17. PubMed PMID: 21165771.
68. **Ottini L**, Falchetti M, Marinozzi S, Angeletti LR, Fornaciari G. *Gene-environment interactions in the pre Industrial Era: the cancer of King Ferrante I of Aragon (1431-1494)*. **Hum Pathol.** 2011 Mar;42(3):332-9. doi: 10.1016/j.humpath.2010.07.010. Epub 2010 Dec 15. PubMed PMID: 21111451.
69. Palli D, Polidoro S, D'Errico M, Saieva C, Guarnera S, Calcagnile AS, Sera F, Allione A, Gemma S, Zanna I, Filomena A, Testai E, Caini S, Moretti R, Gomez-Miguel MJ, Nesi G, Luzzi I, **Ottini L**, Masala G, Matullo G, Dogliotti E. *Polymorphic DNA repair and metabolic genes: a multigenic study on gastric cancer*. **Mutagenesis.** 2010 Nov;25(6):569-75. doi: 10.1093/mutage/geq042. Epub 2010 Sep 3. PubMed PMID: 20817763.
70. Guastadisegni C, Colafranceschi M, **Ottini L**, Dogliotti E. *Microsatellite instability as a marker of prognosis and response to therapy: a meta-analysis of colorectal cancer survival data*. **Eur J Cancer.** 2010 Oct;46(15):2788-98. doi: 10.1016/j.ejca.2010.05.009. Epub 2010 Jun 4. Review. PubMed PMID: 20627535.
71. Zanna I, Rizzolo P, Sera F, Falchetti M, Aretini P, Giannini G, Masala G, Gulino A, Palli D, **Ottini L**. *The BRCAPRO 5.0 model is a useful tool in genetic counseling and clinical management of male breast cancer cases*. **Eur J Hum Genet.** 2010 Jul;18(7):856-8. doi: 10.1038/ejhg.2010.29. Epub 2010 Mar 17. PubMed PMID: 20234394; PubMed Central PMCID: PMC2987355.
72. Silvestri V, Rizzolo P, Zanna I, Falchetti M, Masala G, Bianchi S, Papi L, Giannini G, Palli D, **Ottini L**. *PALB2 mutations in male breast cancer: a population-based study in Central Italy*. **Breast Cancer Res Treat.** 2010 Jul;122(1):299-301. doi: 10.1007/s10549-010-0797-z. Epub 2010 Feb 24. PubMed PMID: 20180015.
73. **Ottini L**, Capalbo C, Rizzolo P, Silvestri V, Bronte G, Rizzo S, Russo A. *HER2-positive male breast cancer: an update*. **Breast Cancer** (Dove Med Press). 2010 Oct 4;2:45-58. doi: 10.2147/BCTT.S6519. Review. PubMed PMID: 24367166; PubMed Central PMCID: PMC3846466.
74. **Ottini L**, Falchetti M. *When history meets molecular medicine: molecular history of human tuberculosis*. **Med Secoli.** 2010;22(1-3):611-32. PubMed PMID: 21563490.
75. **Ottini L**, Palli D, Rizzo S, Federico M, Bazan V, Russo A. *Male breast cancer*. **Crit Rev Oncol Hematol.** 2010 Feb;73(2):141-55. doi: 10.1016/j.critrevonc.2009.04.003. Epub 2009 May 7. Review. PubMed PMID: 19427229.
76. D'Errico M, de Rinaldis E, Blasi MF, Viti V, Falchetti M, Calcagnile A, Sera F, Saieva C, **Ottini L**, Palli D, Palombo F, Giuliani A, Dogliotti E. *Genome-wide expression profile of sporadic gastric cancers with microsatellite instability*. **Eur J Cancer.** 2009 Feb;45(3):461-9. doi: 10.1016/j.ejca.2008.10.032. Epub 2008 Dec 8. PubMed PMID: 19081245.
77. Papi L, Putignano AL, Congregati C, Zanna I, Sera F, Morrone D, Falchetti M, Turco MR, **Ottini L**, Palli D, Genuardi M. *Founder mutations account for the majority of BRCA1-attributable hereditary breast/ovarian cancer cases in a population from Tuscany, Central Italy*. **Breast Cancer Res Treat.** 2009 Oct;117(3):497-504. doi: 10.1007/s10549-008-0190-3. Epub 2008 Sep 27. PubMed PMID: 18821011.

78. Ottini L, Rizzolo P, Zanna I, Falchetti M, Masala G, Ceccarelli K, Vezzosi V, Gulino A, Giannini G, Bianchi S, Sera F, Palli D. *BRCA1/BRCA2 mutation status and clinical-pathologic features of 108 male breast cancer cases from Tuscany: a population-based study in central Italy.* *Breast Cancer Res Treat.* 2009 Aug;116(3):577-86. doi: 10.1007/s10549-008-0194-z. Epub 2008 Sep 26. PubMed PMID: 18819001.
79. Giannini G, Capalbo C, Ottini L, Buffone A, De Marchis L, Margaria E, Vitolo D, Ricevuto E, Rinaldi C, Zani M, Ferraro S, Marchetti P, Cortesi E, Frati L, Screpanti I, Gulino A. *Clinical classification of BRCA1 DNA missense variants: H1686Q is a novel pathogenic mutation occurring in the ontogenetically invariant THV motif of the N-terminal BRCT domain.* *J Clin Oncol.* 2008 Sep 1;26(25):4212-4; author reply 4214-5. doi: 10.1200/JCO.2008.18.2089. PubMed PMID: 18757339.
80. Falchetti M, Lupi R, Ottini L. *From cellular biology to molecular biology: Golgi apparatus from the discovery to nowadays.* *Med Secoli.* 2007;19(1):119-44. PubMed PMID: 18447171.
81. Falchetti M, Saieva C, Lupi R, Masala G, Rizzolo P, Zanna I, Ceccarelli K, Sera F, Mariani-Costantini R, Nesi G, Palli D, Ottini L. *Gastric cancer with high-level microsatellite instability: target gene mutations, clinicopathologic features, and long-term survival.* *Hum Pathol.* 2008 Jun;39(6):925-32. doi: 10.1016/j.humpath.2007.10.024. Epub 2008 Apr 28. PubMed PMID: 18440592.
82. Di Lernia S, Manzi G, Ricci F, Passariello R, Sedati P, Angeletti LR, Marrazzo S, Ottini L, Fornaciari G, Giuffra V, Ventura L, Anag G. *The mummies of the Roman age, from Giarabub (II century D.C.).* *Med Secoli.* 2006;18(3):953-8. PubMed PMID: 18175631.
83. Falchetti M, Lupi R, Ottini L. *Molecular analysis of a colorectal carcinoma from a mummy of the XVth century.* *Med Secoli.* 2006;18(3):943-51. PubMed PMID: 18175630.
84. Verginelli F, Capelli C, Coia V, Musiani M, Falchetti M, Ottini L, Palmirota R, Tagliacozzo A, Mazzorin Ide G, Mariani-Costantini R. *The origins of dogs: archaeozoology, genetics, and ancient DNA.* *Med Secoli.* 2006;18(3):741-54. PubMed PMID: 18175620.
85. Gargano G, Calcaro D, Corsale S, Agnese V, Intrivici C, Fulfaro F, Pantuso G, Cajozzo M, Morello V, Tomasino RM, Ottini L, Colucci G, Bazan V, Russo A. *Aberrant methylation within RUNX3 CpG island associated with the nuclear and mitochondrial microsatellite instability in sporadic gastric cancers. Results of a GOIM (Gruppo Oncologico dell'Italia Meridionale) prospective study.* *Ann Oncol.* 2007 Jun;18 Suppl 6:vi103-9. PubMed PMID: 17591800.
86. Palli D, Falchetti M, Masala G, Lupi R, Sera F, Saieva C, D'Amico C, Ceroti M, Rizzolo P, Caligo MA, Zanna I, Ottini L. *Association between the BRCA2 N372H variant and male breast cancer risk: a population based case-control study in Tuscany, Central Italy.* *BMC Cancer.* 2007 Sep 3;7:170. PubMed PMID: 17767707; PubMed Central PMCID: PMC2001195.
87. Falchetti M, Lupi R, Rizzolo P, Ceccarelli K, Zanna I, Calò V, Tommasi S, Masala G, Paradiso A, Gulino A, Giannini G, Russo A, Palli D, Ottini L. *BRCA1/BRCA2 rearrangements and CHEK2 common mutations are infrequent in Italian male breast cancer cases.* *Breast Cancer Res Treat.* 2008 Jul;110(1):161-7. Epub 2007 Jul 28. PubMed PMID: 17661168.
88. Falchetti M, Lupi R, Ottini L. *Prevention of mediterranean anemia in Latium: Ida Bianco archives.* *Med Secoli.* 2006;18(1):241-61. PubMed PMID: 17526287.
89. Buffone A, Capalbo C, Ricevuto E, Sidoni T, Ottini L, Falchetti M, Cortesi E, Marchetti P, Scambia G, Tomao S, Rinaldi C, Zani M, Ferraro S, Frati L, Screpanti I, Gulino A, Giannini G. *Prevalence of BRCA1 and BRCA2 genomic rearrangements in a cohort of consecutive Italian breast and/or ovarian cancer families.* *Breast Cancer Res Treat.* 2007 Dec;106(2):289-96. Epub 2007 Feb 28. PubMed PMID: 17333342.
90. Cattaneo F, Venesio T, Molatore S, Russo A, Fiocca R, Frattini M, Scovassi AI, Ottini L, Bertario L, Ranzani GN. *Functional analysis and case-control study of -160C/A polymorphism in the E-cadherin gene promoter: association with cancer risk.* *Anticancer Res.* 2006 Nov-Dec;26(6B):4627-32. PubMed PMID: 17201188.
91. Pepe C, Guidugli L, Sensi E, Aretini P, D'Andrea E, Montagna M, Manoukian S, Ottini L, Radice P, Viel A, Bevilacqua G, Caligo MA. *Methyl group metabolism gene polymorphisms as modifier of breast cancer risk in Italian BRCA1/2 carriers.* *Breast Cancer Res Treat.* 2007 May;103(1):29-36. Epub 2006 Dec 7. PubMed PMID: 17151928.
92. Ottini L, Falchetti M, Lupi R, Rizzolo P, Agnese V, Colucci G, Bazan V, Russo A. *Patterns of genomic instability in gastric cancer: clinical implications and perspectives.* *Ann Oncol.* 2006 Jun;17 Suppl 7:vii97-102. PubMed PMID: 16760303.
93. Gargano G, Agnese V, Calò V, Corsale S, Augello C, Bruno L, La Paglia L, Gullo A, Ottini L, Russo A, Fulfaro F, Rinaldi G, Crosta A, Cicero G, Majorana O, Palmeri L, Cipolla C, Agrusa A, Gulotta G, Morello V, Di Fede G, Adamo V, Colucci G,

- Tomasino RM, Valerio MR, Bazan V, Russo A; Gruppo Oncologico dell'Italia Meridionale. *Detection and quantification of mammaglobin in the blood of breast cancer patients: can it be useful as a potential clinical marker? Preliminary results of a GOIM (Gruppo Oncologico dell'Italia Meridionale) prospective study.* *Ann Oncol.* 2006 Jun;17 Suppl 7:vii41-5. PubMed PMID: 16760290.
94. Rocco A, Caruso R, Toracchio S, Rigoli L, Verginelli F, Catalano T, Neri M, Curia MC, Ottini L, Agnese V, Bazan V, Russo A, Pantuso G, Colucci G, Mariani-Costantini R, Nardone G. *Gastric adenomas: relationship between clinicopathological findings, Helicobacter pylori infection, APC mutations and COX-2 expression.* *Ann Oncol.* 2006 Jun;17 Suppl 7:vii103-8. PubMed PMID: 16760271.
95. Ottini L, Lupi R, Falchetti M, Fornaciari G, Mariani-Costantini R, Angeletti LR. *Molecular paleopathology: a novel perspective for biomedical history.* *Med Secoli.* 2005;17(1):181-91. PubMed PMID: 16285089.
96. Bianchi S, Palli D, Falchetti M, Saieva C, Masala G, Mancini B, Lupi R, Noviello C, Omerovic J, Paglierani M, Vezzosi V, Alimandi M, Mariani-Costantini R, Ottini L. *ErbB-receptors expression and survival in breast carcinoma: a 15-year follow-up study.* *J Cell Physiol.* 2006 Mar;206(3):702-8. PubMed PMID: 16245316.
97. Palli D, Saieva C, Luzzi I, Masala G, Topa S, Sera F, Gemma S, Zanna I, D'Errico M, Zini E, Guidotti S, Valeri A, Fabbrucci P, Moretti R, Testai E, del Giudice G, Ottini L, Matullo G, Dogliotti E, Gomez-Miguel MJ. *Interleukin-1 gene polymorphisms and gastric cancer risk in a high-risk Italian population.* *Am J Gastroenterol.* 2005 Sep;100(9):1941-8. PubMed PMID: 16128937.
98. Verginelli F, Capelli C, Coia V, Musiani M, Falchetti M, Ottini L, Palmirota R, Tagliacozzo A, De Grossi Mazzorin I, Mariani-Costantini R. *Mitochondrial DNA from prehistoric canids highlights relationships between dogs and South-East European wolves.* *Mol Biol Evol.* 2005 Dec;22(12):2541-51. Epub 2005 Aug 24. PubMed PMID: 16120801.
99. Lazzereschi D, Nardi F, Turco A, Ottini L, D'Amico C, Mariani-Costantini R, Gulino A, Coppa A. *A complex pattern of mutations and abnormal splicing of Smad4 is present in thyroid tumours.* *Oncogene.* 2005 Aug 11;24(34):5344-54. PubMed PMID: 15940269.
100. Passali D, De Benedetto M, Giordano C, Ottini L, Patrignani P, Piemonte M, Damiani V, Passali GC, Passali FM, Bellussi L. *Rationale of the use of COX-2 inhibitors in ENT pathologies.* *Clin Ter.* 2004 Oct;155(10):439-42. Review. PubMed PMID: 15702656.
101. Ottini L, Angeletti LR, Pantano WB, Falchetti M, Minozzi S, Fortini P, Catalano P, Mariani-Costantini R. *Possible human sacrifice at the origins of Rome: novel skeletal evidences.* *Med Secoli.* 2003;15(3):459-68. PubMed PMID: 15682539.
102. Palli D, Masala G, Mariani-Costantini R, Zanna I, Saieva C, Sera F, Decarli A, Ottini L. *A gene environment interaction between occupation and BRCA1/BRCA2 mutations in male breast cancer?* *Eur J Cancer.* 2004 Nov;40(16):2474-9. PubMed PMID: 15519522.
103. Ottini L, Falchetti M, Saieva C, De Marco M, Masala G, Zanna I, Paglierani M, Giannini G, Gulino A, Nesi G, Mariani Costantini R, Palli D. *MRE11 expression is impaired in gastric cancer with microsatellite instability.* *Carcinogenesis.* 2004 Dec;25(12):2337-43. Epub 2004 Aug 19. PubMed PMID: 15319296.
104. Migliavacca M, Ottini L, Bazan V, Agnese V, Corsale S, Macaluso M, Lupi R, Dardanoni G, Valerio MR, Pantuso G, Di Fede G, Tomasino RM, Gebbia N, Mariani-Costantini R, Russo A. *TP53 in gastric cancer: mutations in the I3 loop and LSH motif DNA-binding domains of TP53 predict poor outcome.* *J Cell Physiol.* 2004 Sep;200(3):476-85. PubMed PMID: 15254976.
105. Ottini L. *Physiopathology of the inflammatory process with the involvement of the upper respiratory tract.* *Acta Otorhinolaryngol Ital.* 2003 Aug;23(4 Suppl 74):7-12, 37-40. PubMed PMID: 15058603.
106. Aretini P, D'Andrea E, Pasini B, Viel A, Mariani Costantini R, Cortesi L, Ricevuto E, Agata S, Bisegna R, Boiocchi M, Caligo MA, Chieco-Bianchi L, Cipollini G, Crucianelli R, D'Amico C, Federico M, Ghimenti C, De Giacomi C, De Nicolo A, Della Puppa L, Ferrari S, Ficarella C, Iandolo D, Manoukian S, Marchetti P, Marroni F, Menin C, Montagna M, Ottini L, Pensotti V, Pierotti M, Radice P, Santarosa M, Silingardi V, Turchetti D, Bevilacqua G, Presciuttini S. *Different expressivity of BRCA1 and BRCA2: analysis of 179 Italian pedigrees with identified mutation.* *Breast Cancer Res Treat.* 2003 Sep;81(1):71-9. PubMed PMID: 14531499.
107. Verginelli F, Ottini L, Esposito D, Cama A, Battista P, Ciranni R, Zavaglia K, Mariani-Costantini R, Fornaciari G. *Ancient DNA studies: from paleopathology to population genetics.* *Med Secoli.* 2002;14(2):587-607. PubMed PMID: 14510002.
108. Rocco A, Staibano S, Ottini L, Mezza E, Somma P, Mariani-Costantini R, Budillon G, Nardone G. *Is there a link between environmental factors and a genetic predisposition to cancer? A lesson from a familial cluster of gastric cancers.* *Eur J Cancer.* 2003 Jul;39(11):1619-24. PubMed PMID: 12855270.

109. Ottini L, Masala G, D'Amico C, Mancini B, Saieva C, Aceto G, Gestri D, Vezzosi V, Falchetti M, De Marco M, Paglierani M, Cama A, Bianchi S, Mariani-Costantini R, Palli D. *BRCA1 and BRCA2 mutation status and tumor characteristics in male breast cancer: a population-based study in Italy*. *Cancer Res.* 2003 Jan 15;63(2):342-7. PubMed PMID: 12543786.
110. Ottini L, Mariani-Costantini R, Costantini AM. *Malnutrition in XIXth century Naples*. *Med Secoli.* 2001;13(1):93-114. PubMed PMID: 12362938.
111. Lotti LV, Ottini L, D'Amico C, Gradini R, Cama A, Belleudi F, Frati L, Torrisi MR, Mariani-Costantini R. *Subcellular localization of the BRCA1 gene product in mitotic cells*. *Genes Chromosomes Cancer.* 2002 Nov;35(3):193-203. PubMed PMID: 12353262.
112. Giannini G, Ristori E, Cerignoli F, Rinaldi C, Zani M, Viel A, Ottini L, Crescenzi M, Martinotti S, Bignami M, Frati L, Scropanti I, Gulino A. *Human MRE11 is inactivated in mismatch repair-deficient cancers*. *EMBO Rep.* 2002 Mar;3(3):248-54. Epub 2002 Feb 15. PubMed PMID: 11850399; PubMed Central PMCID: PMC1084012.
113. Mariani-Costantini R, Ottini L, Caramiello S, Palmirota R, Mallegni F, Rossi A, Frati L, Capasso L. *Taphonomy of the fossil hominid bones from the Acheulean site of Castel di Guido near Rome, Italy*. *J Hum Evol.* 2001 Sep;41(3):211-25. PubMed PMID: 11535000.
114. Ottini L, Minozzi S, Pantano WB, Maucci C, Gazzaniga V, Angeletti LR, Catalano P, Mariani-Costantini R. *A subject with abnormally short stature from Imperial Rome*. *J Endocrinol Invest.* 2001 Jul-Aug;24(7):546-8. PubMed PMID: 11508791.
115. Richetta A, Ottini L, Falchetti M, Innocenzi D, Bottoni U, Faiola R, Mariani-Costantini R, Calvieri S. *Instability at sequence repeats in melanocytic tumours*. *Melanoma Res.* 2001 Jun;11(3):283-9. PubMed PMID: 11468517.
116. Palli D, Russo A, Ottini L, Masala G, Saieva C, Amorosi A, Cama A, D'Amico C, Falchetti M, Palmirota R, Decarli A, Mariani Costantini R, Fraumeni JF Jr. *Red meat, family history, and increased risk of gastric cancer with microsatellite instability*. *Cancer Res.* 2001 Jul 15;61(14):5415-9. PubMed PMID: 11454685.
117. Ottini L, Falchetti M, D'Amico C, Amorosi A, Saieva C, Palli D, Mariani-Costantini R. *Gastric cancer with mutator phenotype: molecular bases and mechanisms of progression*. *Dig Liver Dis.* 2000 Dec;32 Suppl 3:S188-9. PubMed PMID: 11245291.
118. Ottini L, D'Amico C, Noviello C, Lauro S, Lalle M, Fornarini G, Colantuoni OA, Pizzi C, Cortesi E, Carlini S, Guadagni F, Bianco AR, Frati L, Contegiacomo A, Mariani-Costantini R. *BRCA1 and BRCA2 mutations in central and southern Italian patients*. *Breast Cancer Res.* 2000;2(4):307-10. Epub 2000 Mar 31. PubMed PMID: 11056688; PubMed Central PMCID: PMC13918.
119. Ottini L, Palli D, Falchetti M, D'Amico C, Noviello C, Saieva C, Russo A, Corsi AM, Masala G, Paglierani M, Vezzosi V, Bianchi S, Mariani-Costantini R. *Instability at dinucleotide and trinucleotide repeats in breast cancer*. *Int J Oncol.* 2000 Oct;17(4):819-26. PubMed PMID: 10995897.
120. Lazzereschi D, Palmirota R, Ranieri A, Ottini L, Veri MC, Cama A, Cetta F, Nardi F, Colletta G, Mariani Costantini R. *Microsatellite instability in thyroid tumours and tumour-like lesions*. *Br J Cancer.* 1999 Jan;79(2):340-5. PubMed PMID: 9888478; PubMed Central PMCID: PMC2362211.
121. Ottini L, Falchetti M, D'Amico C, Amorosi A, Saieva C, Masala G, Frati L, Cama A, Palli D, Mariani Costantini R. *Mutations at coding mononucleotide repeats in gastric cancer with the microsatellite mutator phenotype*. *Oncogene.* 1998 May 28;16(21):2767-72. PubMed PMID: 9652743.
122. Ottini L, D'Amico C, Noviello C, Pizzi C, Pagliarulo C, Curia MC, Limite G, Bianco AR, Frati L, Caramia FG, Cama A, Contegiacomo A, Mariani-Costantini R. *Novel deletion at codon 1254 of the BRCA1 gene in an Italian breast cancer kindred*. *Hum Mutat.* 1998;Suppl 1:S237-9. PubMed PMID: 9452097.
123. Ottini L, Palli D, Falchetti M, D'Amico C, Amorosi A, Saieva C, Calzolari A, Cimoli F, Tatarelli C, De Marchis L, Masala G, Mariani-Costantini R, Cama A. *Microsatellite instability in gastric cancer is associated with tumor location and family history in a high-risk population from Tuscany*. *Cancer Res.* 1997 Oct 15;57(20):4523-9. PubMed PMID: 9377564.
124. Richetta A, Silipo V, Calvieri S, Frati L, Ottini L, Cama A, Mariani-Costantini R. *Microsatellite instability in primary and metastatic melanoma*. *J Invest Dermatol.* 1997 Jul;109(1):119-20. PubMed PMID: 9204967.
125. De Marchis L, Contegiacomo A, D'Amico C, Palmirota R, Pizzi C, Ottini L, Mastranzo P, Figliolini M, Petrella G, Amanti C, Battista P, Bianco AR, Frati L, Cama A, Mariani-Costantini R. *Microsatellite instability is correlated with lymph node-positive breast cancer*. *Clin Cancer Res.* 1997 Feb;3(2):241-8. PubMed PMID: 9815679.

126. Ottini L, Marziali G, Conti A, Charlesworth A, Sorrentino V. *Alpha and beta isoforms of ryanodine receptor from chicken skeletal muscle are the homologues of mammalian RyR1 and RyR3*. *Biochem J.* 1996 Apr 1;315 ( Pt 1):207-16. PubMed PMID: 8670108; PubMed Central PMCID: PMC1217172.
127. Ottini L, Esposito DL, Richetta A, Carlesimo M, Palmirota R, Veri MC, Battista P, Frati L, Caramia FG, Calvieri S, Cama A, Mariani-Costantini R. *Alterations of microsatellites in neurofibromas of von Recklinghausen's disease*. *Cancer Res.* 1995 Dec 1;55(23):5677-80. PubMed PMID: 7585653.
128. Carlesimo M, Giustini S, Richetta A, Ottini L, Mariani-Costantini R, Falcioni R, Calvieri S. *Integrin b4 expression in peripheral-nerve tumors*. *Oncol Rep.* 1995 Nov;2(6):997-1000. PubMed PMID: 21597841.
129. Contegiacomo A, Pizzi C, De Marchis L, Alimandi M, Delrio P, Di Palma E, Petrella G, Ottini L, French D, Frati L, Bianco AR, Mariani-Costantini R. *High cell kinetics is associated with amplification of the int-2, bcl-1, myc and erbB-2 proto-oncogenes and loss of heterozygosity at the DF3 locus in primary breast cancers*. *Int J Cancer.* 1995 Mar 29;61(1):1-6. PubMed PMID: 7705920.
130. Cama A, de la Luz Sierra M, Quon MJ, Ottini L, Gorden P, Taylor SI. *Substitution of glutamic acid for alanine 1135 in the putative "catalytic loop" of the tyrosine kinase domain of the human insulin receptor. A mutation that impairs proteolytic processing into subunits and inhibits receptor tyrosine kinase activity*. *J Biol Chem.* 1993 Apr 15;268(11):8060-9. PubMed PMID: 8096518.
131. Cama A, de la Luz Sierra M, Ottini L, Kadawaki T, Gorden P, Imperato-McGinley J, Taylor SI. *A mutation in the tyrosine kinase domain of the insulin receptor associated with insulin resistance in an obese woman*. *J Clin Endocrinol Metab.* 1991 Oct;73(4):894-901. PubMed PMID: 1890161.

Textbooks  
(Chapters)

1. Ottini L., Capalbo, C. Male breast cancer. pp. 753-762. In Breast Cancer: Innovations in Research and Management. Springer International Publishing AG 2017 U. Veronesi et al. (eds.), Breast Cancer, DOI 10.1007/978-3-319-48848-6\_63. 2017
2. Ottini L, Falchetti M, Nesi G. Gene Signatures in Gastric Cancer. Chapter 7 pp. 95-113. In: Diagnostic, Prognostic and Therapeutic Value of Gene Signatures. Current Clinical Pathology. Springer Science Editor. 2012
3. Ottini L, Falchetti M. Microsatellite Instability in Gastric Cancer, pp193-205. Chapter 5. In: Genome Research Advances. Alberto P. Bellini Editor. 2007

Rome, 13/02/2020

Signature

