

Supplementary Note

Acknowledged consortia members and affiliations

The PRACTICAL Consortium (<http://practical.ccge.medschl.cam.ac.uk/>):

Rosalind Eeles^{1,2}, Doug Easton³, Zsolt Kote-Jarai¹, Ali Amin Al Olama³, Sara Benlloch³, Kenneth Muir⁴, Graham G. Giles^{5,6}, Fredrik Wiklund⁷, Henrik Gronberg⁷, Christopher A. Haiman⁸, Johanna Schleutker^{9,10}, Maren Weischer¹¹, Ruth C. Travis¹², David Neal¹³, Paul Pharoah¹⁴, Kay-Tee Khaw¹⁵, Janet L. Stanford^{16,17}, William J. Blot¹⁸, Stephen Thibodeau¹⁹, Christiane Maier^{20,21}, Adam S. Kibel^{22,23}, Cezary Cybulski²⁴, Lisa Cannon-Albright²⁵, Hermann Brenner^{26,27}, Jong Park²⁸, Radka Kaneva²⁹, Jyotsna Batra³⁰, Manuel R. Teixeira³¹, Hardev Pandha³²

¹The Institute of Cancer Research, 15 Cotswold Road, Sutton, Surrey, SM2 5NG, UK, ²Royal Marsden NHS Foundation Trust, Fulham and Sutton, London and Surrey, UK, ³Centre for Cancer Genetic Epidemiology, Department of Public Health and Primary Care, University of Cambridge, Strangeways Laboratory, Worts Causeway, Cambridge, UK, ⁴University of Warwick, Coventry, UK, ⁵Cancer Epidemiology Centre, Cancer Council Victoria, 615 St Kilda Road, Melbourne Victoria, Australia, ⁶Centre for Epidemiology and Biostatistics, Melbourne School of Population and Global Health, The University of Melbourne, Victoria, Australia, ⁷Department of Medical Epidemiology and Biostatistics, Karolinska Institute, Stockholm, Sweden, ⁸Department of Preventive Medicine, Keck School of Medicine, University of Southern California/Norris Comprehensive Cancer Center, Los Angeles, California, USA, ⁹Department of Medical Biochemistry and Genetics, University of Turku, Turku, Finland, ¹⁰Institute of Biomedical Technology/BioMediTech, University of Tampere and FimLab Laboratories, Tampere, Finland, ¹¹Department of Clinical Biochemistry, Herlev Hospital, Copenhagen University Hospital, Herlev Ringvej 75, DK-2730 Herlev, Denmark, ¹²Cancer Epidemiology Unit, Nuffield Department of Clinical Medicine, University of Oxford, Oxford, UK, ¹³Surgical Oncology (Uro-Oncology: S4), University of Cambridge, Box 279, Addenbrooke's Hospital, Hills Road, Cambridge, UK and Cancer Research UK Cambridge Research Institute, Li Ka Shing Centre, Cambridge, UK, ¹⁴Centre for Cancer Genetic Epidemiology, Department of Oncology, University of Cambridge, Strangeways Laboratory, Worts Causeway, Cambridge, UK, ¹⁵Cambridge Institute of Public Health, University of Cambridge, Forvie Site, Robinson Way, Cambridge CB2 0SR, ¹⁶Division of Public Health Sciences, Fred Hutchinson Cancer Research Center, Seattle, Washington, USA, ¹⁷Department of Epidemiology, School of Public Health, University of Washington, Seattle, Washington, USA, ¹⁸International Epidemiology Institute, 1455 Research Blvd., Suite 550, Rockville, MD 20850, ¹⁹Mayo Clinic, Rochester, Minnesota, USA, ²⁰Department of Urology, University Hospital Ulm, Germany, ²¹Institute of Human Genetics University Hospital Ulm, Germany, ²²Brigham and Women's Hospital/Dana-Farber Cancer Institute, 45 Francis Street- ASB II-3, Boston, MA 02115, ²³Washington University, St Louis, Missouri, ²⁴International Hereditary Cancer Center, Department of Genetics and Pathology, Pomeranian Medical University, Szczecin, Poland, ²⁵Division of Genetic Epidemiology, Department of Medicine, University of Utah School of Medicine, ²⁶Division of Clinical Epidemiology and Aging Research & Division of Preventive Oncology, German Cancer Research Center, Heidelberg Germany, ²⁷German Cancer Consortium (DKTK), German Cancer Research Center (DKFZ), Heidelberg Germany, ²⁸Division of Cancer Prevention and Control, H. Lee Moffitt Cancer Center, 12902 Magnolia Dr., Tampa, Florida, USA, ²⁹Molecular Medicine Center and Department of Medical Chemistry and Biochemistry, Medical University - Sofia, 2 Zdrave St, 1431, Sofia, Bulgaria, ³⁰Australian Prostate Cancer Research Centre-Qld, Institute of Health and Biomedical Innovation and Schools of Life Science and Public Health, Queensland University of Technology, Brisbane, Australia, ³¹Department of Genetics, Portuguese Oncology Institute, Porto, Portugal and Biomedical Sciences Institute (ICBAS), Porto University, Porto, Portugal, ³²The University of Surrey, Guildford, Surrey, GU2 7XH, UK

LifeLines Cohort Study

Behrooz Z Alizadeh (1), Rudolf A de Boer (2), H Marike Boezen (1), Marcel Bruinenberg (3), Lude Franke (4), Pim van der Harst (2), Hans L Hillege (1,2), Melanie M van der Klauw (5), Gerjan Navis (6), Johan Ormel (7), Dirkje S Postma (8), Judith GM Rosmalen (7), Joris P Slaets (9), Harold Snieder (1), Ronald P Stolk (1), Bruce HR Wolffenbuttel (5), Cisca Wijmenga (4)

(1)Department of Epidemiology, University of Groningen, University Medical Center Groningen, The Netherlands
(2)Department of Cardiology, University of Groningen, University Medical Center Groningen, The Netherlands
(3)LifeLines Cohort Study, University of Groningen, University Medical Center Groningen, The Netherlands
(4)Department of Genetics, University of Groningen, University Medical Center Groningen, The Netherlands
(5)Department of Endocrinology, University of Groningen, University Medical Center Groningen, The Netherlands
(6)Department of Internal Medicine, Division of Nephrology, University of Groningen, University Medical Center

Groningen, The Netherlands (7)Interdisciplinary Center of Psychopathology of Emotion Regulation (ICPE), Department of Psychiatry, University of Groningen, University Medical Center Groningen, The Netherlands (8)Department of Pulmonology, University of Groningen, University Medical Center Groningen, The Netherlands (9)University Center for Geriatric Medicine, University of Groningen, University Medical Center Groningen, The Netherlands

InterAct Consortium

Nita G Forouhi(1), Nicola D Kerrison(1), Claudia Langenberg(1), Robert A Scott(1), Stephen J Sharp(1), Matt Sims(1), Inês Barroso(2,3), Panos Deloukas(2), Mark I McCarthy(4,5,6), Larraitz Arriola(7,8,9), Beverley Balkau(10,11), Aurelio Barricarte(12,9), Heiner Boeing(13), Paul W Franks(14,15), Carlos Gonzalez(16), Sara Grioni(17), Rudolf Kaaks(18), Timothy J Key(19), Carmen Navarro(20,9,21), Peter M Nilsson(14), Kim Overvad(22,23), Domenico Palli(24), Salvatore Panico(25), J. Ramón Quirós(26), Olov Rolandsson(15), Carlotta Sacerdote(27,28), María-José Sánchez(29,9,30), Nadia Slimani(31), Anne Tjonneland(32), Rosario Tumino(33,34), Daphne L van der A(35), Yvonne T van der Schouw(36), Elio Riboli(37), Nicholas J Wareham(1)

- (1) MRC Epidemiology Unit, University of Cambridge, Cambridge, United Kingdom, (2) The Wellcome Trust Sanger Institute, Cambridge, United Kingdom, (3) University of Cambridge Metabolic Research Laboratories, Cambridge, UK, (4) Oxford Centre for Diabetes, Endocrinology and Metabolism (OCDEM), University of Oxford, UK, (5) Wellcome Trust Centre for Human Genetics, University of Oxford, Oxford, UK, (6) Oxford NIHR Biomedical Research Centre, Oxford, UK, (7) Public Health Division of Gipuzkoa, San Sebastian, Spain, (8) Instituto BIO-Donostia, Basque Government, San Sebastian, Spain, (9) CIBER Epidemiología y Salud Pública (CIBERESP), Spain, (10) Inserm, CESP, U1018, Villejuif, France, (11) Univ Paris-Sud, UMRS 1018, Villejuif, France, (12) Navarre Public Health Institute (ISPN), Pamplona, Spain, (13) German Institute of Human Nutrition Potsdam-Rehbruecke, Germany, (14) Lund University, Malmö, Sweden, (15) Umeå University, Umeå, Sweden, (16) Catalan Institute of Oncology (ICO), Barcelona, Spain, (17) Epidemiology and Prevention Unit, Milan, Italy, (18) German Cancer Research Centre (DKFZ), Heidelberg, Germany, (19) University of Oxford, United Kingdom, (20) Department of Epidemiology, Murcia Regional Health Council, Murcia, Spain, (21) Unit of Preventive Medicine and Public Health, School of Medicine, University of Murcia, Spain, (22) Department of Public Health, Section for Epidemiology, Aarhus University, Aarhus, Denmark, (23) Aalborg University Hospital, Aalborg, Denmark, (24) Cancer Research and Prevention Institute (ISPO), Florence, Italy, (25) Dipartimento di Medicina Clinica e Chirurgia, Federico II University, Naples, Italy, (26) Public Health Directorate, Asturias, Spain, (27) Unit of Cancer Epidemiology, Citta' della Salute e della Scienza Hospital-University of Turin and Center for Cancer Prevention (CPO), Torino, Italy, (28) Human Genetics Foundation (HuGeF), Torino, Italy, (29) Andalusian School of Public Health, Granada, Spain, (30) Instituto de Investigación Biosanitaria de Granada (Granada.ibs), Granada (Spain), (31) International Agency for Research on Cancer, Lyon, France, (32) Danish Cancer Society Research Center, Copenhagen, Denmark, (33) ASP Ragusa, Italy, (34) Aire Onlus, Ragusa, Italy, (35) National Institute for Public Health and the Environment (RIVM), Bilthoven, The Netherlands, (36) University Medical Center Utrecht, Utrecht, the Netherlands, (37) School of Public Health, Imperial College London, UK.

Australian Ovarian Cancer Study (AOCS) members

David D Bowtell¹, Adele C Green², Georgia Chenevix-Trench², Anna deFazio³, Dorota Gertig⁴, Penelope M Webb².
1 Peter MacCallum Cancer Centre, St Andrews Place, East Melbourne, Victoria, Australia 2 QIMR Berghofer Medical Research Institute, Brisbane, Australia 3 Westmead Institute for Cancer Research, Westmead, New South Wales, Australia 4 Victorian Cervical Cytology Registry, Carlton South Victoria, Australia

kConFab Consortium members

Morteza Aghmesheh Medical Oncology Department, Illawarra Cancer Care Centre, Wollongong Hospital, Wollongong, NSW 2500

David Amor Genetic Health Services Victoria, Royal Children's Hospital, Melbourne VIC 3050

Lesley Andrews Hereditary Cancer Clinic, Prince of Wales Hospital, Randwick NSW 2031

Yoland Antill Dept. Haem and Medical Oncology, Peter MacCallum Cancer Centre, St Andrews Place, East Melbourne VIC 3002

Shane Armitage Molecular Genetics Lab, Royal Brisbane and Women's Hospital Leanne Arnold Molecular Genetics Lab, Royal Brisbane and Women's Hospital

Rosemary Balleine Department of Translational Oncology, C/- Department of Medical Oncology, Westmead Hospital, Westmead NSW 2145

Agnes Bankier PO Box 5444, HEIDELBERG WEST, C/o The Austin Hospital, 3081 Australia
 Patti Bastick St George Hospital, Medical Oncology Dept, Gray Street, Kogarah NSW 2000, Australia
 Jonathan Beesley Queensland Institute of Medical Research, Herston Road, Herston Qld 4002, Australia
 John Beilby Pathology Centre, Queen Elizabeth Medical Centre, Nedlands WA 6009
 Ian Bennett Silverton Place, 101 Wickham Terrace, Brisbane QLD 4000
 Barbara Bennett Hereditary Cancer Clinic, Prince of Wales Hospital, Randwick NSW 2031
 Geoffrey Berry Dept of Public Health and Community Medicine, University of Sydney, Sydney NSW 2006
 Anneke Blackburn John Curtin School of Medical Research, Australian National University, P.O. Box 334, Canberra ACT 2601
 Michael Bogwitz Familial Cancer Centre, The Royal Melbourne Hospital, Grattan Street, Parkville Victoria 3050, Australia
 Meagan Brennan NSW Breast Cancer Institute, POBox 143, Westmead NSW 2145
 Melissa Brown Department of Biochemistry, University of Queensland, St. Lucia QLD 4072
 Michael Buckley Molecular and Cytogenetics Unit, Prince of Wales Hospital, Randwick NSW 2031
 Matthew Burgess Clinical Genetics Service, Austin Health, Victoria 3084, Australia
 Jo Burke Royal Hobart Hospital, GPO Box 1061L, Hobart TAS 7001
 Phyllis Butow Medical Psychology Unit, Royal Prince Alfred Hospital, Camperdown NSW 2204
 Keith Byron Australian Genome Research Facility, Walter & Eliza Hall Medical Research Institute, Royal Melbourne Hospital, Parkville VIC 3050
 David Callen Dame Roma Mitchell Cancer Research Laboratories, University of Adelaide/Hanson Institute, P.O. Box 14, Rundle Mall SA 5000
 Ian Campbell Peter MacCallum Cancer Centre, St Andrew's Place, East Melbourne VIC 3002
 Deepa Chauhan School of Psychology, Brennan McCallum (Building A18), University of Sydney 2006
 Georgia Chenevix-Trench Queensland Institute of Medical Research, Royal Brisbane Hospital, Herston QLD 4029
 Alice Christian Genetics Department, Central Region Genetics Service, Wellington Hospital, New Zealand
 Christine Clarke Westmead Institute for Cancer Research, University of Sydney, Westmead Hospital, Westmead NSW 2145
 Alison Colley Department of Clinical Genetics, Liverpool Health Service, PO Box 103, Liverpool NSW 2170
 Dick Cotton Mutation Research Centre, St Vincent's Hospital, Victoria Parade, Fitzroy VIC 3065
 Ashley Crook Department of Clinical Genetics, Level 3E, Royal North Shore Hospital, St Leonards NSW 2065
 James Cui Epidemiology and Preventive Medicine, Monash University, Prahan Vic 3004, Australia
 Bronwyn Culling Molecular and Clinical Genetics, Level 1 Building 65, Royal Prince Alfred Hospital, Camperdown NSW 2050
 Margaret Cummings Department of Pathology, University of Queensland Medical School, Herston NSW 4006
 Sarah-Jane Dawson Molecular Genetics Department, Cambridge University, England
 Anna deFazio Dept. Gynaecological Oncology, Westmead Institute for Cancer Research, Westmead Hospital, Westmead NSW 2145
 Martin Delatycki Clinical Genetics, Austin Health, Heidelberg Repatriation Hospital, PO Box 5444, Heidelberg West Vic 3081, Australia
 Rebecca Dickson Level 2, Block 51, Royal North Shore Hospital, North Shore NSW 2408
 Joanne Dixon Central Regional Genetic Services, Wellington Hospital, Private bag 7902, Wellington, New Zealand
 Alexander Dobrovic Molecular Pathology, Department of Pathology, Peter MacCallum Cancer Centre, St Andrew's Place, East Melbourne VIC 3002
 Tracy Dudding Hunter Genetics, Hunter Area Health Service, PO Box 84, Waratah, 2298 NSW
 Ted Edkins Clinical Chemistry, Princess Margaret Hospital for Children, Box D184, Perth WA 6001
 Stacey Edwards Department of Biochemistry and Molecular Biology, University of Queensland, St Lucia Qld 4072, Australia
 Maurice Eisenbruch Department of Multicultural Health, University of Sydney, NSW 2052
 Gelareh Farshid Tissue Pathology, IMVS, Adelaide SA 5000 Susan Fawcett Family Cancer Clinic, Monash Medical Centre, Clayton VIC 3168
 Andrew Fellows Molecular Diagnostic Development, Pathology Department, Peter MacCallum Cancer Centre, Melbourne, East Melbourne Vic 3002
 Georgina Fenton South West Family Cancer Clinic, Liverpool Hospital, Liverpool BC NSW 1871
 Michael Field Royal North Shore Hospital, Level 2, Vindin House, St Leonards NSW 2065
 Frank Firgaira GTG, 60 - 66 Hanover Street, Fitzroy, 3065
 James Flanagan Epigenetics Unit, Department of Surgery and Oncology, Imperial College London, London W12 0NN, England

Jean Fleming Eskitis Institute of Cell & Molecular Therapies, School of Biomolecular and Biomedical Sciences, Griffith University, Nathan QLD 4111

Peter Fong Medical Oncology Department, Regional Cancer and Blood Services, Level 1 Building 7, Auckland City Hospital, 2 Park Rd. Grafton, Auckland 1023, New Zealand

John Forbes Surgical Oncology, University of Newcastle, Newcastle Mater Hospital, Waratah NSW 2298

Stephen Fox Pathology Department, Level 1, Peter MacCallum Cancer Centre, St Andrew's Place, East Melbourne Vic 3002

Juliet French School of Molecular and Microbial Sciences, University of Queensland, St Lucia Qld 4072

Michael Friedlander Department of Medical Oncology, Prince of Wales Hospital, Randwick NSW 2031

Clara Gaff Victorian Clinical Genetics Service, Royal Melbourne Hospital, Parkville VIC 3052

Mac Gardner Genetic Health Services Victoria, 10th Floor The Murdoch Institute, Royal Children's Hospital, Parkville VIC 3052

Mike Gattas Queensland Clinical Genetic Service, Royal Children's Hospital, Bramston Terrace, Herston QLD 4020

Peter George Clinical Biochemistry Unit, Canterbury Health Labs, PO Box 151, Christchurch, New Zealand

Graham Giles Cancer Epidemiology Centre, Anti Cancer Council of Victoria, 1 Rathdowne Street, Carlton South VIC 3052

Grantley Gill Department of Surgery, Royal Adelaide Hospital, Adelaide SA 5000

Jack Goldblatt Genetic Services Of WA, King Edward Memorial Hospital, 374 Bagot Road, Subiaco WA 6008

Sian Greening Illawarra Cancer Centre, Wollongong Hospital, Private Mail Bag 8808, South Coast Mail Centre, NSW 2521

Scott Grist Department of Haematology and Genetic Pathology, SouthPath , Flinders Medical Centre , SA

Eric Haan Department of Medical Genetics, Women's and Children's Hospital, North Adelaide SA 5006 Kate Hardie Room 430 Bldg 76, School of Chemistry and Molecular Biosciences, University of Queensland, St Lucia QLD 4072

Marion Harris Familial Cancer Clinic, Peter MacCallum Cancer Centre, St Andrews Place, East Melbourne VIC 3002

Stewart Hart Breast and Ovarian Cancer Genetics, Monash Medical Centre, 871 Centre Road, Bentleigh East VIC, 3165

Nick Hayward Queensland Institute for Medical Research, Royal Brisbane Hospital Post Office, Herston QLD 4029

Sue Healey Queensland Institute of Medical Research (QIMR), 300 Herston Road, Herston Qld Q4006

Louise Heiniger Medical Psychology Research Unit, The University of Sydney, Sydney NSW 2006

John Hopper Centre for M.E.G.A. Epidemiology, University of Melbourne, Level 1, 723 Swanston Street, Carlton VIC 3010

Evelyn Humphrey Royal Hobart Hospital, GPO Box 1061L, Hobart TAS 7001

Clare Hunt Southern Health Familial Cancer Centre, Monash Medical Centre, Special Medicine Building, 246 Clayton Rd, Clayton Victoria 3168, Australia

Paul James Genetic Health Seviles, Monash Medical Centre, Clayton Vic

Mark Jenkins Centre for M.E.G.A. Epidemiology, The University of Melbourne, 723 Swanston Street, Carlton VIC 3053

Alison Jones Molecular Genetics Lab, Royal Brisbane and Women's Hospital, QLD

Rick Kefford Medical Oncology, Westmead Hospital, Westmead NSW 2145

Alexa Kidd Clinical Genetics Departments, Central Regional Genetics Service, Wellington Hospital, New Zealand

Belinda Kiely NHMRC Clinical Trials Centre, University of Sydney, Locked Bag 77, Camperdown Sydney NSW 1450

Judy Kirk Familial Cancer Service, Department of Medicine, Westmead Hospital, Westmead NSW 2145

Jessica Koehler Hereditary Cancer Clinic, Prince of Wales Hospital, Randwick NSW 2031

James Kollias Breast Endocrine and Surgical Unit, Royal Adelaide Hospital, North Terrace SA 5000

Serguei Kovalenko Genetic Technologies Limited, 60-66 Hanover Street, Fitzroy Vic 3065

Sunil Lakhani UQ Centre for Clinical Research, Level 6 Building 71/918, University of Queensland, The Royal Brisbane & Women's Hospital Herston, 4029

Amanda Leaming Wesley Breast Clinic, Chasely Street, Auchenflower, Brisbane Qld 4066

Jennifer Leary Familial Cancer Laboratory, Westmead Hospital, Westmead NSW 2145 W W W . N A T U R E . C O M /

Jacqueline Lim Dept of Psychological Medicine, Royal North Shore Hospital, St Leonards NSW 2065

Geoff Lindeman Breast Cancer Laboratory, Walter and Eliza Hall Institute, PO Royal Melbourne Hospital, Parkville VIC 3050

Lara Lipton Medical Oncology and Clinical Haematology Unit, Western Hospital, Footscray VIC

Liz Lobb Medical Psychology Research Unit, Room 332, Brennan MacCallum Building (A18), The University of Sydney, Camperdown, 2006

Graham Mann Westmead Institute for Cancer Research, Westmead Millennium Institute, Westmead NSW 2145
 Deborah Marsh Kolling Institute of Medical Research, Royal North Shore Hospital, St Leonards NSW 2065 Sue Anne McLachlan Department of Oncology, St Vincent's Hospital, 41 Victoria Parade, Fitzroy VIC 3065
 Bettina Meiser Hereditary Cancer Clinic, Prince of Wales Hospital, Randwick NSW 2031
 Cliff Meldrum Molecular Pathology Dept, 1st Floor, Peter MacCallum Cancer Centre, St Andrew's Place, East Melbourne Vic 3002
 Roger Milne Centro Nacional de Investigaciones Oncologicas, C/ Melchor Fernández Almagro, 3, E-28029 Madrid, Spain
 Gillian Mitchell Family Cancer Clinic, Peter MacCallum Cancer Centre, St Andrew's Place, East Melbourne VIC 3002
 Beth Newman School of Public Health Road, Queensland University of Technology, Victoria Park, Kelvin Grove QLD 4059
 Shona O'Connell Southern Health Familial Cancer Centre, Special Medicine Building, 246 Clayton Road, Clayton Vic 3168
 Imelda O'Loughlin St Vincent's Breast Clinic, PO Box 4751, Toowoomba QLD 4350
 Richard Osborne Dept of Public Health and Community Medicine, 200 Berkeley Street, Carlton VIC 3053
 Nick Pachter Familial Cancer and Clinical Genetics, Royal Melbourne Hospital, Grattan Street, Parkville VIC 3050, Australia
 Briony Patterson Tas Clinical Genetics Service, Royal Hobart Hospital, GPO Box 1061, Hobart Tasmania 7001, Australia
 Lester Peters Radiation Oncology, Peter MacCallum Cancer Centre, St Andrew's Place, East Melbourne VIC 3002
 Kelly Phillips Department of Medical Oncology, Peter MacCallum Cancer Centre, St Andrew's Place, East Melbourne VIC 3002
 Melanie Price Medical Psychology, University of Sydney, Sydney, 2006
 Lynne Purser The Centre for Genetics Education NSW Health, PO Box 317, St Leonards NSW 1590, Australia
 Jeanne Reeve Northern Regional Genetic Service, Auckland Hospital, New Zealand
 Tony Reeve Cancer Genetics Laboratory, University of Otago, PO Box 56, Dunedin, New Zealand
 Robert Richards Dept of Cytogenetics and Molecular Genetics, Women and Children's Hospital, Adelaide SA 5006
 Edwina Rickard Familial Cancer centre, Westmead Hospital, Westmead NSW 2145 Bridget Robinson Oncology Service, Christchurch Hospital, Private Bag 4710, Christchurch, New Zealand
 Barney Rudzki Molecular Pathology Department, The University of Melbourne, Parkville Vic 3050 Mona Saleh Centre for Genetic Education, Prince of Wales Hospital, Randwick, NSW 2031
 Elizabeth Salisbury Anatomical Pathology, UNSW, Prince of Wales Hospital, Randwick, 2031 NSW
 Joe Sambrook Peter MacCallum Cancer Centre, St Andrew's Place, East Melbourne VIC 3002 Christobel Saunders School of Surgery and Pathology, QE11 Medical Centre, M block 2nd Floor, Nedlands WA 6907
 Jodi Saunus Breast Pathology, University of Queensland Centre for Clinical Research, Building 71/918 Royal Brisbane and Women's Hospital, Herston Qld 4029
 Robyn Sayer Gynaecological Cancer Centre, Royal Hospital for Women, Randwick NSW 2011
 Elizabeth Scott South View Clinic, Suite 13, Level 3 South Street, Kogarah NSW 2217
 Rodney Scott Hunter Area Pathology Service, John Hunter Hospital, Locked Bag 1 Regional Mail Centre, NSW 2310
 Clare Scott Research Department, WEHI, C/o Royal Melbourne Hospital, Parkville, 3050
 Ram Seshadri Department of Haematology, Flinders Medical Centre, Bedford Park SA 5042 Adrienne Sexton Familial Cancer Centre, Royal Melbourne Hospital, Grattan Street, Parkville Vic 3050
 Raghwa Sharma Dept of Tissue Pathology, Westmead Hospital, Westmead NSW 2145 Andrew Shelling Obstetrics and Gynaecology, University of Auckland, New Zealand
 Peter Simpson The University of Queensland, Building 71/918, RBWH Campus, Herston Qld 4029
 Melissa Southey Genetic Epidemiology Laboratory, Department of Pathology, University of Melbourne, VIC 3010
 Amanda Spurdle Cancer Unit, Queensland Institute of Medical Research, Herston QLD 4029 Graeme Suthers South Australian Clinical Genetics Service, Centre for Medical Genetics, Women and Children's Hospital, North Adelaide SA 5006
 Pamela Sykes Molecular Pathology, Flinders Medical Centre, Flinders Drive, Bedford Park, 5042, Australia Donna Taylor Department of Radiology, Royal Perth Hospital, Perth WA
 Jessica Taylor Familial Cancer and Genetics Medicine, Royal Melbourne Hospital, 2nd Floor Grattan Street, Parkville Vic 3050, Australia
 Benjamin Thierry Ian Wark Research Institute, University of South Australia, Adelaide SA 5095 SA
 Ella Thompson Cancer Genetics, Research Department, 3rd level, Peter MacCallum Cancer Centre, East Melbourne, VIC 3002
 Heather Thorne Research Department, Peter MacCallum Cancer Centre, St Andrew's Place, East Melbourne VIC 3002

Sharron Townshend Genetic Services of Western Australia, 3rd Floor Agnes Walsh House, 374 Bagot Rd, Subiaco WA 6008
Alison Trainer University of NSW, Prince of Wales Hospital, Barker Street, Randwick NSW 2031
Lan Tran Medical Psychology Unit, University of Sydney, NSW 2006 Kathy Tucker Heredity Cancer Clinic, Prince of Wales Hospital, Randwick NSW 2031
Janet Tyler Hereditary Cancer Clinic, Prince of Wales Hospital, Randwick NSW 2031
Jane Visvader The Walter and Eliza Hall Institute of Medical Research, Post Office Royal Melbourne Hospital, Parkville VIC 3050
Logan Walker Molecular Cancer Epidemiology Laboratory, Queensland Institute of Medical Research, P.O. Royal Brisbane Hospital, Herston, Qld 4027, Australia
Ian Walpole Genetic Services of WA, King Edward Memorial Hospital, Subiaco WA 6008, Robin Ward Department of Medical Oncology, Prince of Wales Hospital, Randwick NSW 2031
Paul Waring Department of Pathology, University of WA, 35 Stirling Highway, CRAWLEY WA 6009 Perth,
Bev Warner Cabrini Hospital, 183 Wattletree Rd, Malvern VIC 3144. Graham Warren St Vincent's Breast Clinic, PO Box 4751, Toowoomba QLD 4350
Rachael Williams Family Cancer Clinic, St Vincent's Hospital, Darlinghurst NSW 2010
Judy Wilson Block 4, Level 5, Royal North Shore Hospital, St Leonards NSW 2065
Ingrid Winship Department of Genetics, Royal Melbourne Hospital, Parkville, 3050
Kathy Wu Familial Cancer Centre, Westmead Hospital, Darcy Street, Westmead NSW 2045
Mary Ann Young Familial Cancer Clinic, Peter MacCallum Cancer Centre, St Andrew's Place, East Melbourne VIC 3002

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1) GWAS

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CARL	Cancer Aids Registries Linkage	We are very grateful to the municipal administrators for their collaboration on the project and for logistic support. We would like to thank all participants to this study. We thank Anna Morgan and Angela D’Eustacchio for technical support.
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Colaous	Etude Cohorte Lausannoise	
CROATIA-Korcula	CROATIA-Korcula	<p>We would like to acknowledge the contributions of the recruitment team in Korcula, the administrative teams in Croatia and Edinburgh and the people of Korcula. The SNP genotyping for the KORCULA cohort was performed in Helmholtz Zentrum München, Neuherberg, Germany. Some array genotyping was performed at the Wellcome Trust Clinical Research Facility Genetics Core at Western General Hospital, Edinburgh, UK. Christian Gieger is supported by Russian Foundation for Basic Research (RFBR)-Helmholtz research group program.</p>
CROATIA-Split	CROATIA-Split	<p>We would like to acknowledge the contributions of the recruitment team from the Croatian Centre for Global Health, University of Split, the administrative teams in Croatia and Edinburgh and the people of Split. The SNP genotyping for the CROATIA_Split cohort was performed by AROS Applied Biotechnology, Aarhus, Denmark. Medical Research Council UK and the Ministry of Science, Education and Sport in the Republic of Croatia (number 108-1080315-0302).</p>
CROATIA-Vis	CROATIA-Vis	<p>We would like to acknowledge the staff of several institutions in Croatia that supported the field work, including but not limited to The University of Split and Zagreb Medical Schools, Institute for Anthropological Research in Zagreb and Croatian Institute for Public Health. The SNP genotyping for the CROATIA_Vis cohort was performed in the core genotyping laboratory of the Wellcome Trust Clinical Research Facility at the Western General Hospital, Edinburgh, Scotland</p> <p>Medical Research Council UK and the Ministry of Science, Education and Sport in the</p>

		Republic of Croatia (number 108-1080315-0302).
deCODE		
EGCUT (370k)	Estonian Genome Center, University of Tartu	EGCUT work was supported by the Targeted Financing from the Estonian Ministry of Science and Education [SF0180142s08]; the US National Institute of Health [R01DK075787]; the Development Fund of the University of Tartu (grant SP1GVARENG); the European Regional Development Fund to the Centre of Excellence in Genomics (EXCEGEN; grant 3.2.0304.11-0312); and through FP7 grant 313010.
EGCUT OmniX	Estonian Genome Center, University of Tartu	EGCUT work was supported by the Targeted Financing from the Estonian Ministry of Science and Education [SF0180142s08]; the US National Institute of Health [R01DK075787]; the Development Fund of the University of Tartu (grant SP1GVARENG); the European Regional Development Fund to the Centre of Excellence in Genomics (EXCEGEN; grant 3.2.0304.11-0312); and through FP7 grant 313010.
ERF	Erasmus Rucphen Family study	We are grateful to all general practitioners for their contributions, to Petra Veraart for her help in genealogy, Jeannette Vergeer for the supervision of the laboratory work and Peter Snijders for his help in data collection. The ERF study as a part of EUROSPAN (European Special Populations Research Network) was supported by European Commission FP6 STRP grant number 018947 (LSHG-CT-2006-01947) and also received funding from the European Community's Seventh Framework Programme (FP7/2007-2013)/grant agreement HEALTH-F4-2007-201413 by the European Commission under the programme "Quality of Life and Management of the Living Resources" of 5th Framework Programme (no. QL2-CT-2002-01254). High-throughput analysis of the ERF data was supported by joint grant from Netherlands Organization for Scientific Research and the Russian Foundation for Basic Research (NWO-RFBR 047.017.043). Exome sequencing analysis in ERF was supported by the ZonMw grant (project 91111025). Najaf Amin is supported by the Hersenstichting Nederland (project number F2013(1)-28).
FHS	Framingham Heart Study	The authors thank the Framingham Heart Study participants and staff. The Framingham Heart Study phenotype-genotype analyses were supported by the National Institute of Aging (Genetics of Reproductive Life Period and Health Outcomes, R21AG032598; JMM, KL and R01AG29451 JMM, KL). The Framingham Heart Study of the National Heart Lung and Blood Institute of the National Institutes of Health and Boston University School of Medicine was supported by the National Heart, Lung and Blood Institute's Framingham Heart Study Contract No. N01-HC-25195 and its contract with Affymetrix, Inc for genotyping services (Contract No. N02-HL-6-4278). Analyses reflect intellectual input and resource development from the Framingham Heart Study investigators participating in the SNP Health Association Resource (SHARe) project. A portion of this research was conducted using the Linux Cluster for Genetic Analysis (LinGA-II) funded by the Robert Dawson Evans Endowment of the Department of Medicine at Boston University School of Medicine and Boston Medical Center. Genotyping, quality control and calling of the Illumina HumanExome BeadChip in the Framingham Heart Study was supported by funding from the National Heart, Lung and Blood Institute Division of Intramural Research (Daniel Levy and Christopher J. O'Donnell, Principal Investigators).
Generation Scotland	Generation Scotland: Scottish Family Health Study	We would like to acknowledge the contributions of the families who took part in the Generation Scotland: Scottish Family Health Study, the general practitioners and Scottish School of Primary Care for their help in recruiting them, and the whole Generation Scotland team, which includes academic researchers, IT staff, laboratory technicians, statisticians and research managers. Genotyping was performed at the Wellcome Trust Clinical Research Facility Genetics Core at Western General Hospital, Edinburgh, UK. Scottish Executive Health Department, Chief Scientist Office, grant number CZD/16/6. Exome array genotyping for GS:SFHS was funded by the Medical Research Council UK.
GENOA	Genetic Epidemiology Network of Arteriopathy	Support for the Genetic Epidemiology Network of Arteriopathy (GENOA) was provided by the National Heart, Lung and Blood Institute of the National Institutes of Health (HL054464, HL054457, HL054481, and HL087660). Genotyping was performed at the Mayo Clinic (Stephen Turner, Mariza de Andrade, Julie Cunningham) and was made possible by the University of Texas Health Sciences Center (Eric Boerwinkle, Megan Grove-Gaona). We would also like to thank the families that participated in the GENOA study. HL054464, HL054457, HL054481, and HL087660.

HAPI Heart Study	Heredity and Phenotype Intervention (HAPI) Heart Study	U01-HL72515, U01-HL84756, R01-088119, P30-DK072488, K01-HL116770
HealthABC	The Health, Aging, and Body Composition Study	The Health ABC Study was supported by NIA contracts N01AG62101, N01AG62103, and N01AG62106 and, in part, by the NIA Intramural Research Program. The genome-wide association study was funded by NIA grant 1R01AG032098-01A1 to Wake Forest University Health Sciences and genotyping services were provided by the Center for Inherited Disease Research (CIDR). CIDR is fully funded through a federal contract from the National Institutes of Health to The Johns Hopkins University, contract number HHSN268200782096C. This study utilized the high-performance computational capabilities of the Biowulf Linux cluster at the National Institutes of Health, Bethesda, Md. (http://biowulf.nih.gov). see acknowledgement statement
HRS	Health and Retirement Study	HRS is supported by the National Institute on Aging (NIA U01AG009740). The genotyping was funded separately by the National Institute on Aging (RC2 AG036495, RC4 AG039029). Our genotyping was conducted by the NIH Center for Inherited Disease Research (CIDR) at Johns Hopkins University. Genotyping quality control and final preparation of the data were performed by the Genetics Coordinating Center at the University of Washington. U01 AG009740, RC2 AG036495, RC4 AG039029
InChianti	Invecchiare in Chianti	The InCHIANTI study baseline (1998-2000) was supported as a “targeted project” (ICS110.1/RF97.71) by the Italian Ministry of Health and in part by the U.S. National Institute on Aging (Contracts: 263 MD 9164 and 263 MD 821336); the InCHIANTI Follow-up 1 (2001-2003) was funded by the U.S. National Institute on Aging (Contracts: N.1-AG-1-1 and N.1-AG-1-2111); the InCHIANTI Follow-ups 2 and 3 studies (2004-2010) were financed by the U.S. National Institute on Aging (Contract: N01-AG-5-0002); supported in part by the Intramural research program of the National Institute on Aging, National Institutes of Health, Baltimore, Maryland.
INGI-FVG	Genetic Park of Friuli Venezia Giulia Project	We are very grateful to the municipal administrators for their collaboration on the project and for logistic support. We would like to thank all participants to this study. We thank Anna Morgan and Angela D’Eustacchio for technical support. Fondo Trieste (2008) and Regione FVG (L.26.2008)
INGI-VB	Val Borbera Isolated Population Project	We thank the inhabitants of the VB that made this study possible, the local administrations, the Tortona and Genova archdiocese and the ASL-22, Novi Ligure (AI) for support. We also thank Clara Camaschella for data collection supervision and organization of the clinical data collection, Fiammetta Viganò for technical help, Massimiliano Cocca for building the analysis platform. The research was supported by funds from Compagnia di San Paolo, Torino, Italy; Fondazione Cariplo, Italy and Ministry of Health, Ricerca Finalizzata 2008 and CCM 2010, PRIN 2009 and Telethon, Italy to DT.
InterAct cohort/ InterAct cases	European Prospective Investigation into Cancer & Nutrition - InterAct	We thank all EPIC participants and staff for their contribution to the study. We thank staff from the Technical, Field Epidemiology and Data Functional Group Teams of the MRC Epidemiology Unit in Cambridge, UK, for carrying out sample preparation, DNA provision and quality control, genotyping and data-handling work. The EPIC-InterAct study received funding from the European Union (Integrated Project LSHM-CT-2006-037197 in the Framework Programme 6 of the European Community).
KORA F3/ KORA F4	Cooperative Health Research in the Region of Augsburg (follow-up 3) / (follow-up 4)	We thank all the study participants, all members of staff of the Institutes of Epidemiology and the field staff in Augsburg who planned and conducted the study. The KORA study group consists of A. Peters (speaker), R. Holle, K. Strauch, J. Heinrich, R. Leidl, C. Meisinger, and their co-workers, who are responsible for the design and conduct of the KORA studies. The KORA research platform (KORA, Cooperative Health Research in the Region of Augsburg) was initiated and financed by the Helmholtz Zentrum München - German Research Center for Environmental Health, which is funded by the German Federal Ministry of Education and Research and by the State of Bavaria. Furthermore, KORA research was supported within the Munich Center of Health Sciences (MC Health), Ludwig-Maximilians-Universität, as part of LMUinnovativ. The funders had no role in study design, data collection and analysis, decision to publish, or preparation of the manuscript. Elisabeth Altmaier - European Union’s Seventh Framework Programme (FP7-Health-F5-2012) under Grant agreement No 305280 (MIMOmics). Christian Gieger is supported by Russian Foundation for Basic Research (RFBR)-Helmholtz research group program.

Lifelines	The LifeLines Cohort Study and Biobank	Lifelines (LifeLines) - We thank Behrooz Z. Alizadeh, Annemieke Boesjes, Marcel Bruinenberg, Noortje Festen, Pim van der Harst, Ilja Nolte, Lude Franke, Mitra Valimohammadi for their help in creating the GWAS database, and Rob Bieringa, Joost Keers, René Oostergo, Rosalie Visser, Judith Vonk for their work related to data-collection and validation. The authors are grateful to the study participants, the staff from the Lifelines Cohort Study and Medical Biobank Northern Netherlands, and the participating general practitioners and pharmacists. Researchers interested in using the Lifelines data must obtain approval for a specific analysis plan from the scientific board of Lifelines to obtain access to the data. Researchers using the data are required to follow the terms of a signed agreement containing a number of clauses designed to ensure protection of privacy and compliance with relevant laws. For further information, contact Harold Snieder (h.snieder@umcg.nl). Elisabeth Altmaier - European Union's Seventh Framework Programme (FP7-Health-F5-2012) under Grant agreement No 305280 (MIMOMics).
MESA	Multi-Ethnic Study of Atherosclerosis	MESA and the MESA SHARe project are conducted and supported by the National Heart, Lung, and Blood Institute (NHLBI) in collaboration with MESA investigators. Support for MESA is provided by contracts N01-HC-95159, N01-HC-95160, N01-HC-95161, N01-HC-95162, N01-HC-95163, N01-HC-95164, N01-HC-95165, N01-HC-95166, N01-HC-95167, N01-HC-95168, N01-HC-95169, UL1-TR-001079, and UL1-TR-000040. MESA Family is conducted and supported by the National Heart, Lung, and Blood Institute (NHLBI) in collaboration with MESA investigators. Genotyping and analysis support was provided by NHLBI grant R01HL071205. Support is provided by grants and contracts R01HL071051, R01HL071205, R01HL071250, R01HL071251, R01HL071258, R01HL071259, by the National Center for Research Resources, Grant UL1RR033176, and the National Center for Advancing Translational Sciences, Grant UL1TR000124. Funding for SHARe genotyping was provided by NHLBI Contract N02-HL-64278. Genotyping was performed at Affymetrix (Santa Clara, California, USA) and the Broad Institute of Harvard and MIT (Boston, Massachusetts, USA) using the Affymetrix Genome-Wide Human SNP Array 6.0.
NHS Illumina Chip / NHS Omni Chip / NHS Affy Chip	Nurses' Health Study	Nurses' Health Study (NHS_BRCA, NHS_T2D, NHS_CHD, NHS_KS, NHS_GA, NHS_CC, NHS_EC, NHS_GO, NHS_MD, NHS2_BRCA, and NHS2_KS). We would like to thank the participants and staff of the NHS and NHSII for their valuable contributions as well as the following state cancer registries for their help: AL, AZ, AR, CA, CO, CT, DE, FL, GA, ID, IL, IN, IA, KY, LA, ME, MD, MA, MI, NE, NH, NJ, NY, NC, ND, OH, OK, OR, PA, RI, SC, TN, TX, VA, WA, WY. The authors assume full responsibility for analyses and interpretation of these data. The NHS GWAS were supported by grants from the National Institutes of Health [NCI (CA40356, CA087969, CA055075, CA98233, U01 CA137088, R01 CA059045, R01 CA137178, R01 CA082838, R01 CA131332), NIDDK (DK058845, DK070756), NHGRI (HG004399, HG004728), NHLBI (HL35464), NIAMS (R01 AR056291)].
NTR	Netherlands Twin Register	We like to acknowledge and thank families who take part in the Netherlands Twin Register and the NTR team, which includes academic researchers, IT staff, laboratory technicians, statisticians and research managers. Support for the Netherlands Twin Register studies and research was obtained from the Netherlands Organization for Scientific Research (NWO) and The Netherlands Organisation for Health Research and Development (ZonMW) grants, 904-61-193,480-04-004,400-05-717, Addiction-31160008, 911-09-032, Spinozapremie 56-464-14192, Biobanking and Biomolecular Resources Research Infrastructure (BBMRI -NL, 184.021.007); the European Research Council (ERC-230374); Rutgers University Cell and DNA Repository (NIMH U24 MH068457-06), the Avera Institute, Sioux Falls, South Dakota (USA) and the National Institutes of Health (NIH R01 HD042157-01A1). Part of the genotyping was funded by the Genetic Association Information Network (GAIN) of the Foundation for the National Institutes of Health and Grand Opportunity grants 1RC2 MH089951). We acknowledge support from VU University's Institute for Health and Care Research (EMGO+), the Neuroscience Campus Amsterdam (NCA) and the faculty of Psychology and Education of VU University.
ORCADES	Orkney Complex Disease Study	We would like to acknowledge the invaluable contributions of the research nurses in Orkney, the administrative team in Edinburgh and the people of Orkney. ORCADES was supported by the Chief Scientist Office of the Scottish Government, the Royal Society, the MRC Human Genetics Unit, Arthritis Research UK and the European Union framework program 6 EUROSPAN project (contract no. LSHG-CT-2006-018947).

QIMR	QIMR Berghofer	IMR: We thank the twins and their families for their participation. We also thank Enda Byrne, Anjali Henders, Dixie Statham, Ann Eldridge, Marlene Grace, Kerrie McAloney, and Lisa Bowdler. A portion of the genotyping on which this study was based (Illumina 370K scans on 4300 individuals) was carried out at the Center for Inherited Disease Research, Baltimore (CIDR), through an access award to our late colleague Dr. Richard Todd (Psychiatry, Washington University School of Medicine, St Louis). Funding was provided by the Australian National Health and Medical Research Council (241944, 339462, 89927, 389875, 389891, 389892, 389938, 442915, 442981, 496739, 552485, 552498), the Australian Research Council (A7960034, A79906588, A79801419, DP0770096, DP0212016, DP0343921), the FP-5 GenomEUtwin Project (QLG2-CT-2002-01254), and the U.S. National Institutes of Health (NIH grants AA07535, AA10248, AA13320, AA13321, AA13326, AA14041, MH66206). G.W.M. is supported by the National Health and Medical Research Council (NHMRC) Fellowship Scheme. Statistical analyses were carried out on the Genetic Cluster Computer, which is financially supported by the Netherlands Scientific Organization (NWO 480-05-003).
RSI / RSII / RSIII	Rotterdam Study I, II, III	The generation and management of the Illumina exome chip v1.0 array data for the Rotterdam Study (RS-I) was executed by the Human Genotyping Facility of the Genetic Laboratory of the Department of Internal Medicine, Erasmus MC, Rotterdam, The Netherlands. The Exome chip array data set was funded by the Genetic Laboratory of the Department of Internal Medicine, Erasmus MC, from the Netherlands Genomics Initiative (NGI)/Netherlands Organisation for Scientific Research (NWO)-sponsored Netherlands Consortium for Healthy Aging (NCHA; project nr. 050-060-810); the Netherlands Organization for Scientific Research (NWO; project number 184021007) and by the Rainbow Project (RP10; Netherlands Exome Chip Project) of the Biobanking and Biomolecular Research Infrastructure Netherlands (BBMRI-NL; www.bbmr.nl). We thank Ms. Mila Jhamai, Ms. Sarah Higgins, and Mr. Marijn Verkerk for their help in creating the exome chip database, and Carolina Medina-Gomez, BSc, Lennard Karsten, BSc, and Dr. Linda Broer for QC and variant calling. Variants were called using the best practice protocol developed by Grove et al. as part of the CHARGE consortium exome chip central calling effort (Grove et al., PLoS One, 2014). The Rotterdam Study is funded by Erasmus Medical Center and Erasmus University, Rotterdam, Netherlands Organization for the Health Research and Development (ZonMw), the Research Institute for Diseases in the Elderly (RIDE), the Ministry of Education, Culture and Science, the Ministry for Health, Welfare and Sports, the European Commission (DG XII), and the Municipality of Rotterdam. The authors are grateful to the study participants, the staff from the Rotterdam Study and the participating general practitioners and pharmacists.
SardinIA	SardinIA	We thank all the volunteers and all the staff for their contribution to the study. This study was funded in part by the National Institutes of Health (National Institute on Aging, National Heart Lung and Blood Institute, and National Human Genome Research Institute). This research was supported by National Human Genome Research Institute grants HG005581, HG005552, HG006513, HG007089, HG007022, and HG007089; by National Heart Lung and Blood Institute grant HL117626; by the Intramural Research Program of the NIH, National Institute on Aging, with contracts N01-AG-1-2109 and HHSN271201100005C; by Sardinian Autonomous Region (L.R. no. 7/2009) grant cRP3-154; by grant FaReBio2011 "Farmaci e Reti Biotecnologiche di Qualità".
SHIP	Study of Health in Pomerania	SHIP is part of the Community Medicine Research net of the University of Greifswald, Germany, which is funded by the Federal Ministry of Education and Research (grants no. 01ZZ9603, 01ZZ0103, and 01ZZ0403), the Ministry of Cultural Affairs as well as the Social Ministry of the Federal State of Mecklenburg-West Pomerania, and the network 'Greifswald Approach to Individualized Medicine (GANI_MED)' funded by the Federal Ministry of Education and Research (grant 03IS2061A). Genome-wide and ExomeChip data have been supported by the Federal Ministry of Education and Research (grants no. 03ZIK012 and 03Z1CN22) and a joint grant from Siemens Healthcare, Erlangen, Germany and the Federal State of Mecklenburg- West Pomerania. The University of Greifswald is a member of the 'Center of Knowledge Interchange' program of the Siemens AG and the Caché Campus program of the InterSystems GmbH. grants no. 01ZZ9603, 01ZZ0103, 01ZZ0403, 03ZIK012, 03Z1CN22 and 03IS2061A

TwinsUKI/ TwinsUKII/ TwinsUKIII		TwinsUK. The study was funded by the Wellcome Trust; European Community's Seventh Framework Programme (FP7/2007-2013). The study also receives support from the National Institute for Health Research (NIHR) BioResource Clinical Research Facility and Biomedical Research Centre based at Guy's and St Thomas' NHS Foundation Trust and King's College London. SNP Genotyping was performed by The Wellcome Trust Sanger Institute and National Eye Institute via NIH/CIDR.
WGHS	Women's Genome Health Study	The WGHS is supported by HL043851 and HL080467 from the National Heart, Lung, and Blood Institute and CA047988 from the National Cancer Institute, and the Donald W. Reynolds Foundation, with collaborative scientific support and funding for genotyping provided by Amgen.

2) Exome chip

Study name / acronym	Full study name	Acknowledgments and source of funding
1958BC	1958 National Child Development Study (also known as the 1958 Birth Cohort Study)	This work made use of data and samples generated by the 1958 Birth Cohort (NCDS). Access to these resources was enabled via the 58READIE Project funded by Wellcome Trust and Medical Research Council (grant numbers WT095219MA and G1001799). A full list of the financial, institutional and personal contributions to the development of the 1958 Birth Cohort Biomedical resource is available at http://www2.le.ac.uk/projects/birthcohort . Genotyping was undertaken as part of the Wellcome Trust Case-Control Consortium (WTCCC) under Wellcome Trust award 076113, and a full list of the investigators who contributed to the generation of the data is available at www.wtccc.org.uk . Wellcome Trust grant WT095219MA, Medical Research Council grant G1001799, Wellcome Trust award 076113.
ARIC	Atherosclerosis Risk in Communities HapMap analysis	The Atherosclerosis Risk in Communities Study is carried out as a collaborative study supported by National Heart, Lung, and Blood Institute contracts (HHSN268201100005C, HHSN268201100006C, HHSN268201100007C, HHSN268201100008C, HHSN268201100009C, HHSN268201100010C, HHSN268201100011C, and HHSN268201100012C), R01HL087641, R01HL59367 and R01HL086694; National Human Genome Research Institute contract U01HG004402; and National Institutes of Health contract HHSN268200625226C. The authors thank the staff and participants of the ARIC study for their important contributions. Infrastructure was partly supported by Grant Number UL1RR025005, a component of the National Institutes of Health and NIH Roadmap for Medical Research. Funding support for "Building on GWAS for NHLBI-diseases: the U.S. CHARGE consortium" was provided by the NIH through the American Recovery and Reinvestment Act of 2009 (ARRA) (5RC2HL102419).
CHS	Cardiovascular Health Study	This CHS research was supported by NHLBI contracts HHSN268201200036C, HHSN268200800007C, HHSN268200960009C, N01HC55222, N01HC85079, N01HC85080, N01HC85081, N01HC85082, N01HC85083, N01HC85086; and NHLBI grants HL080295, HL087652, HL105756 with additional contribution from the National Institute of Neurological Disorders and Stroke (NINDS). Additional support was provided through AG023629 from the National Institute on Aging (NIA). A full list of CHS investigators and institutions can be found at http://chs-nhlbi.org/ . The provision of genotyping data was supported in part by the National Center for Advancing Translational Sciences, CTSI grant UL1TR000124, and the National Institute of Diabetes and Digestive and Kidney Disease Diabetes Research Center (DRC) grant DK063491 to the Southern California Diabetes Endocrinology Research Center.
Fenland		The Fenland Study is funded by the Wellcome Trust and the Medical Research Council, as well as by the Support for Science Funding programme and CamStrad. We are grateful to all the volunteers for their time and help, and to the General Practitioners and practice staff for help with recruitment. We thank the Fenland Study co-ordination team and the Field Epidemiology team of the MRC Epidemiology Unit for recruitment and clinical testing
FHS	Framingham Heart Study	The authors thank the Framingham Heart Study participants and staff. The Framingham Heart Study phenotype-genotype analyses were supported by the National Institute of Aging (Genetics of Reproductive Life Period and Health Outcomes, R21AG032598; JMM, KL and R01AG29451 JMM, KL). The Framingham Heart Study of the National Heart Lung and Blood Institute of the National Institutes of Health and Boston University School of Medicine was supported by the National Heart, Lung and Blood Institute's Framingham Heart Study Contract No. N01-HC-25195 and its contract with Affymetrix, Inc for genotyping services (Contract No. N02-HL-6-4278). Analyses reflect intellectual input and resource development from the Framingham Heart Study investigators participating in the SNP Health Association Resource (SHARe) project. A portion of this research was conducted using the Linux Cluster for Genetic Analysis (LinGA-II) funded by the Robert Dawson Evans Endowment of the Department of Medicine at Boston University School of Medicine and Boston Medical Center. Genotyping, quality control and calling of the Illumina HumanExome BeadChip in the Framingham Heart Study was supported by funding from the National Heart, Lung and Blood Institute Division of Intramural Research (Daniel Levy and Christopher J. O'Donnell, Principal Investigators).

INGI-VB	Val Borbera Isolated Population Project	We thank the inhabitants of the VB that made this study possible, the local administrations, the Tortona and Genova archdiocese and the ASL-22, Novi Ligure (AI) for support. We also thank Clara Camaschella for data collection supervision and organization of the clinical data collection, Fiammetta Viganò for technical help, Massimiliano Cocca for building the analysis platform. The research was supported by funds from Compagnia di San Paolo, Torino, Italy; Fondazione Cariplo, Italy and Ministry of Health, Ricerca Finalizzata 2008 and CCM 2010, PRIN 2009 and Telethon, Italy to DT.
InterAct Cases/ InterAct Subcohort	European Prospective Investigation into Cancer & Nutrition - InterAct	We thank all EPIC participants and staff for their contribution to the study. We thank staff from the Technical, Field Epidemiology and Data Functional Group Teams of the MRC Epidemiology Unit in Cambridge, UK, for carrying out sample preparation, DNA provision and quality control, genotyping and data-handling work. The EPIC-InterAct study received funding from the European Union (Integrated Project LSHM-CT-2006-037197 in the Framework Programme 6 of the European Community).
KORA	Cooperative Health Research in the Region of Augsburg (follow-up 4)	We thank all the study participants, all members of staff of the Institutes of Epidemiology and the field staff in Augsburg who planned and conducted the study. The KORA study group consists of A. Peters (speaker), R. Holle, K. Strauch, J. Heinrich, R. Leidl, C. Meisinger, and their co-workers, who are responsible for the design and conduct of the KORA studies. The KORA research platform (KORA, Cooperative Health Research in the Region of Augsburg) was initiated and financed by the Helmholtz Zentrum München - German Research Center for Environmental Health, which is funded by the German Federal Ministry of Education and Research and by the State of Bavaria. Furthermore, KORA research was supported within the Munich Center of Health Sciences (MC Health), Ludwig-Maximilians-Universität, as part of LMUinnovativ. The funders had no role in study design, data collection and analysis, decision to publish, or preparation of the manuscript. Elisabeth Altmaier - European Union's Seventh Framework Programme (FP7-Health-F5-2012) under Grant agreement No 305280 (MIMOMics). Christian Gieger is supported by Russian Foundation for Basic Research (RFBR)-Helmholtz research group program.
MESA	Multi-Ethnic Study of Atherosclerosis	MESA and the MESA SHARe project are conducted and supported by the National Heart, Lung, and Blood Institute (NHLBI) in collaboration with MESA investigators. Support for MESA is provided by contracts N01-HC-95159, N01-HC-95160, N01-HC-95161, N01-HC-95162, N01-HC-95163, N01-HC-95164, N01-HC-95165, N01-HC-95166, N01-HC-95167, N01-HC-95168, N01-HC-95169, UL1-TR-001079, and UL1-TR-000040. MESA Family is conducted and supported by the National Heart, Lung, and Blood Institute (NHLBI) in collaboration with MESA investigators. Support is provided by grants and contracts R01HL071051, R01HL071205, R01HL071250, R01HL071251, R01HL071258, R01HL071259, by the National Center for Research Resources, Grant UL1RR033176, and the National Center for Advancing Translational Sciences, Grant UL1TR000124. Funding for SHARe genotyping was provided by NHLBI Contract N02-HL-64278. Genotyping was performed at Affymetrix (Santa Clara, California, USA) and the Broad Institute of Harvard and MIT (Boston, Massachusetts, USA) using the Affymetrix Genome-Wide Human SNP Array 6.0.
Amish	Old Order Amish Study	U01-HL72515, U01-HL84756, R01-088119, P30-DK072488, K01-HL116770
Cambridge Cancer	The EMBRACE, SEARCH (breast cancer and ovarian cancer) and SIBS studies	Douglas F. Easton is the PI of the study. EMBRACE Collaborating Centres are: Coordinating Centre, Cambridge: Debra Frost, Steve Ellis, Radka Platte, Jo Perkins. North of Scotland Regional Genetics Service, Aberdeen: Zosia Miedzybrodzka, Helen Gregory. Northern Ireland Regional Genetics Service, Belfast: Patrick Morrison, Lisa Jeffers. West Midlands Regional Clinical Genetics Service, Birmingham: Kai-ren Ong, Jonathan Hoffman. South West Regional Genetics Service, Bristol: Alan Donaldson, Margaret James. East Anglian Regional Genetics Service, Cambridge: Joan Paterson, Marc Tischkowitz, Sarah Downing, Amy Taylor. Medical Genetics Services for Wales, Cardiff: Alexandra Murray, Mark T. Rogers, Emma McCann. St James's Hospital, Dublin & National Centre for Medical Genetics, Dublin: M. John Kennedy, David Barton. South East of Scotland Regional Genetics Service, Edinburgh: Mary Porteous, Sarah Drummond. Peninsula Clinical Genetics Service, Exeter: Carole Brewer, Emma Kivuva, Anne Searle, Selina Goodman, Kathryn Hill.

		<p>West of Scotland Regional Genetics Service, Glasgow: Rosemarie Davidson, Victoria Murday, Nicola Bradshaw, Lesley Snadden, Mark Longmuir, Catherine Watt, Sarah Gibson, Eshika Haque, Ed Tobias, Alexis Duncan.</p> <p>South East Thames Regional Genetics Service, Guy's Hospital London: Louise Izatt, Chris Jacobs, Caroline Langman.</p> <p>North West Thames Regional Genetics Service, Harrow: Huw Dorkins. Leicestershire Clinical Genetics Service, Leicester: Julian Barwell.</p> <p>Yorkshire Regional Genetics Service, Leeds: Julian Adlard, Gemma Serra-Feliu. Cheshire & Merseyside Clinical Genetics Service, Liverpool: Ian Ellis, Claire Foo. Manchester Regional Genetics Service, Manchester: D Gareth Evans, Fiona Laloo, Jane Taylor.</p> <p>North East Thames Regional Genetics Service, NE Thames, London: Lucy Side, Alison Male, Cheryl Berlin.</p> <p>Nottingham Centre for Medical Genetics, Nottingham: Jacqueline Eason, Rebecca Collier.</p> <p>Northern Clinical Genetics Service, Newcastle: Alex Henderson, Oonagh Claber, Irene Jobson.</p> <p>Oxford Regional Genetics Service, Oxford: Lisa Walker, Diane McLeod, Dorothy Halliday, Sarah Durell, Barbara Stayner.</p> <p>The Institute of Cancer Research and Royal Marsden NHS Foundation Trust: Ros Eeles, Nazneen Rahman, Elizabeth Bancroft, Elizabeth Page, Audrey Arden-Jones, Kelly Kohut, Jennifer Wiggins, Jenny Pope, Sibel Saya, Natalie Taylor, Zoe Kemp and Angela George.</p> <p>North Trent Clinical Genetics Service, Sheffield: Jackie Cook, Oliver Quarrell, Cathryn Bardsley.</p> <p>South West Thames Regional Genetics Service, London: Shirley Hodgson, Sheila Goff, Glen Brice, Lizzie Winchester, Charlotte Eddy, Vishakha Tripathi, Virginia Attard.</p> <p>Wessex Clinical Genetics Service, Princess Anne Hospital, Southampton: Diana Eccles, Anneke Lucassen, Gillian Crawford, Donna McBride, Sarah Smalley.</p> <p>CRUK ref: C8197/A16565, CRUK ref: C1287/A8459, CRUK ref: A490/A10124</p> <p>EMBRACE is supported by Cancer Research UK Grants C1287/A10118, C1287/A16563 and C1287/A17523. Genotyping was supported by Cancer Research–UK grant C12292/A11174D. Gareth Evans and Fiona Laloo are supported by an NIHR grant to the Biomedical Research Centre, Manchester. The Investigators at The Institute of Cancer Research and The Royal Marsden NHS Foundation Trust are supported by an NIHR grant to the Biomedical Research Centre at The Institute of Cancer Research and The Royal Marsden NHS Foundation Trust. Ros Eeles and Elizabeth Bancroft are supported by Cancer Research UK Grant C5047/A8385.</p>
deCODE		
EGCUT	Estonian Genome Center, University of Tartu	EGCUT work was supported by the Targeted Financing from the Estonian Ministry of Science and Education [SF0180142s08]; the US National Institute of Health [R01DK075787]; the Development Fund of the University of Tartu (grant SP1GVARENG); the European Regional Development Fund to the Centre of Excellence in Genomics (EXCEGEN; grant 3.2.0304.11-0312); and through FP7 grant 313010.
Generation Scotland	Generation Scotland: Scottish Family Health Study	We would like to acknowledge the contributions of the families who took part in the Generation Scotland: Scottish Family Health Study, the general practitioners and Scottish School of Primary Care for their help in recruiting them, and the whole Generation Scotland team, which includes academic researchers, IT staff, laboratory technicians, statisticians and research managers. Genotyping was performed at the Wellcome Trust Clinical Research Facility Genetics Core at Western General Hospital, Edinburgh, UK. Scottish Executive Health Department, Chief Scientist Office, grant number CZD/16/6. Exome array genotyping for GS:SFHS was funded by the Medical Research Council UK
Korcula	CROATIA_Korcula	We would like to acknowledge the contributions of the recruitment team in Korcula, the administrative teams in Croatia and Edinburgh and the people of Korcula. The SNP genotyping for the KORCULA cohort was performed in Helmholtz Zentrum München, Neuherberg, Germany. Exome array genotyping was performed at the Wellcome Trust Clinical Research Facility Genetics Core at Western General Hospital, Edinburgh, UK. Medical Research Council UK and the Ministry of Science, Education and Sport in the Republic of Croatia (number 108-1080315-0302).

Rotterdam	Rotterdam Study I	<p>The generation and management of the Illumina exome chip v1.0 array data for the Rotterdam Study (RS-I) was executed by the Human Genotyping Facility of the Genetic Laboratory of the Department of Internal Medicine, Erasmus MC, Rotterdam, The Netherlands. The Exome chip array data set was funded by the Genetic Laboratory of the Department of Internal Medicine, Erasmus MC, from the Netherlands Genomics Initiative (NGI)/Netherlands Organisation for Scientific Research (NWO)-sponsored Netherlands Consortium for Healthy Aging (NCHA; project nr. 050-060-810); the Netherlands Organization for Scientific Research (NWO; project number 184021007) and by the Rainbow Project (RP10; Netherlands Exome Chip Project) of the Biobanking and Biomolecular Research Infrastructure Netherlands (BBMRI-NL; www.bbMRI.nl). We thank Ms. Mila Jhamai, Ms. Sarah Higgins, and Mr. Marijn Verkerk for their help in creating the exome chip database, and Carolina Medina-Gomez, BSc, Lennard Karsten, BSc, and Dr. Linda Broer for QC and variant calling. Variants were called using the best practice protocol developed by Grove et al. as part of the CHARGE consortium exome chip central calling effort (Grove et al., PLoS One, 2014).</p> <p>The Rotterdam Study is funded by Erasmus Medical Center and Erasmus University, Rotterdam, Netherlands Organization for the Health Research and Development (ZonMw), the Research Institute for Diseases in the Elderly (RIDE), the Ministry of Education, Culture and Science, the Ministry for Health, Welfare and Sports, the European Commission (DG XII), and the Municipality of Rotterdam. The authors are grateful to the study participants, the staff from the Rotterdam Study and the participating general practitioners and pharmacists.</p>
Sardinia	SardiNIA	<p>We thank all the volunteers and all the staff for their contribution to the study. This study was funded in part by the National Institutes of Health (National Institute on Aging, National Heart Lung and Blood Institute, and National Human Genome Research Institute). This research was supported by National Human Genome Research Institute grants HG005581, HG005552, HG006513, HG007089, HG007022, and HG007089; by National Heart Lung and Blood Institute grant HL117626; by the Intramural Research Program of the NIH, National Institute on Aging, with contracts N01-AG-1-2109 and HHSN271201100005C; by Sardinian Autonomous Region (L.R. no. 7/2009) grant cRP3-154; by grant FaReBio2011 "Farmaci e Reti Biotechnologiche di Qualità".</p>
SHIP/ SHIP-TREND	Study of Health in Pomerania / Study of Health in Pomerania - TREND	<p>SHIP is part of the Community Medicine Research net of the University of Greifswald, Germany, which is funded by the Federal Ministry of Education and Research (grants no. 01ZZ9603, 01ZZ0103, and 01ZZ0403), the Ministry of Cultural Affairs as well as the Social Ministry of the Federal State of Mecklenburg-West Pomerania, and the network 'Greifswald Approach to Individualized Medicine (GANI_MED)' funded by the Federal Ministry of Education and Research (grant 03IS2061A). Genome-wide and ExomeChip data have been supported by the Federal Ministry of Education and Research (grants no. 03ZIK012 and 03Z1CN22) and a joint grant from Siemens Healthcare, Erlangen, Germany and the Federal State of Mecklenburg- West Pomerania. The University of Greifswald is a member of the 'Center of Knowledge Interchange' program of the Siemens AG and the Caché Campus program of the InterSystems GmbH. grants no. 01ZZ9603, 01ZZ0103, 01ZZ0403, 03ZIK012, 03Z1CN22 and 03IS2061A</p>
WGHS	Women's Genome Health Study	<p>The WGHS is supported by HL043851 and HL080467 from the National Heart, Lung, and Blood Institute and CA047988 from the National Cancer Institute, and the Donald W. Reynolds Foundation, with collaborative scientific support and funding for genotyping provided by Amgen.</p>
WHI	Women's Health Initiative	<p>The WHI program is funded by the National Heart, Lung, and Blood Institute, National Institutes of Health, U.S. Department of Health and Human Services through contracts HHSN268201100046C, HHSN268201100001C, HHSN268201100002C, HHSN268201100003C, HHSN268201100004C, and HHSN271201100004C." The authors thank the WHI investigators and staff for their dedication, and the study participants for making the program possible. A full listing of WHI investigators can be found at: http://www.whi.org/researchers/Documents%20%20Write%20a%20Paper/WHI%20Investigator%20Short%20List.pdf</p>

3) Individual Study disclosures

The National Cancer Institute: The content of this manuscript does not necessarily reflect the views or policies of the National Cancer Institute or any of the collaborating centers in the Breast Cancer Family Registry (BCFR), nor does mention of trade names, commercial products, or organizations imply endorsement by the US Government or the BCFR.

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All other studies declared no conflict of interest.

4) Additional acknowledgments

The development of methods for LD score regression[1] and genetic correlation[2] were funded by NIH grant R03 CA173785.

1. Gusev, A. *et al.* Partitioning heritability of regulatory and cell-type-specific variants across 11 common diseases. *Am. J. Hum. Genet.* **95**, 535-552 (2014).
2. Bulik-Sullivan, B. *et al.* An atlas of genetic correlations across human diseases and traits. *Nat. Genet.*, in the press.

Additional Methods

Expression quantitative trait loci (eQTL) analysis

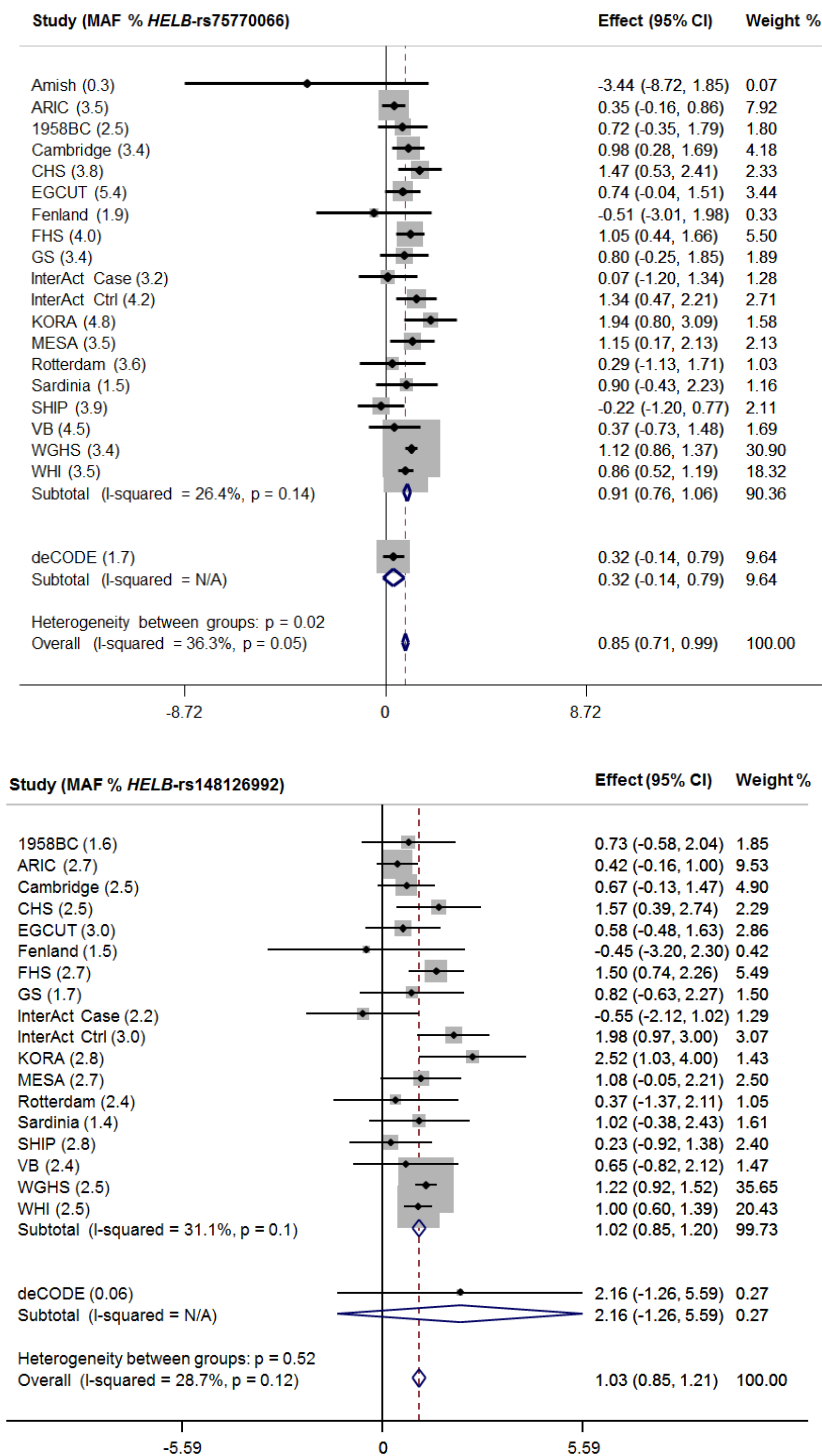
Blood cell related eQTL studies included fresh lymphocytes (17873875), fresh leukocytes (19966804), leukocyte samples in individuals with Celiac disease (19128478), whole blood samples (18344981, 21829388, 22692066, 23818875, 23359819, 23880221, 24013639, 23157493, 23715323, 24092820, 24314549, 24956270, 24592274, 24728292, 24740359), lymphoblastoid cell lines (LCL) derived from asthmatic children (17873877, 23345460), HapMap LCL from 3 populations (17873874), a separate study on HapMap CEU LCL (18193047), additional LCL population samples (19644074, 22286170, 22941192, 23755361, 23995691, 25010687), CD19+ B cells (22446964), primary PHA-stimulated T cells (19644074, 23755361), CD4+ T cells (20833654), peripheral blood monocytes (19222302, 20502693, 22446964) and CD14+ monocytes before and after stimulation with LPS or interferon-gamma (24604202), CD11+ dendritic cells before and after *Mycobacterium tuberculosis* infection (22233810) and a separate study of dendritic cells before or after stimulation with LPS, influenza or interferon-beta (24604203). Micro-RNA QTLs (21691150) and DNase-I QTLs (22307276) were also queried for LCL.

Non-blood cell tissue eQTLs searched included omental and subcutaneous adipose (18344981, 21602305, 22941192, 23715323), stomach (21602305), endometrial carcinomas (21226949), ER+ and ER- breast cancer tumor cells (23374354), liver (18462017, 21602305, 21637794, 22006096, 24665059), osteoblasts (19654370), intestine (23474282) and normal and cancerous colon (25079323), skeletal muscle (24306210), breast tissue (normal and cancer) (24388359, 22522925), lung (23209423, 23715323, 24307700), skin (21129726, 22941192, 23715323), primary fibroblasts (19644074, 23755361, 24555846), sputum (21949713), pancreatic islet cells (25201977) and heart tissue from left ventricles (23715323, 24846176) and left and right atria (24177373). Micro-RNA QTLs were also queried for gluteal and abdominal adipose (22102887) and liver (23758991). Further mRNA and micro-RNA QTLs were queried from ER+ invasive breast cancer samples, colon-, kidney renal clear-, lung- and prostate-adenocarcinoma samples (24907074).

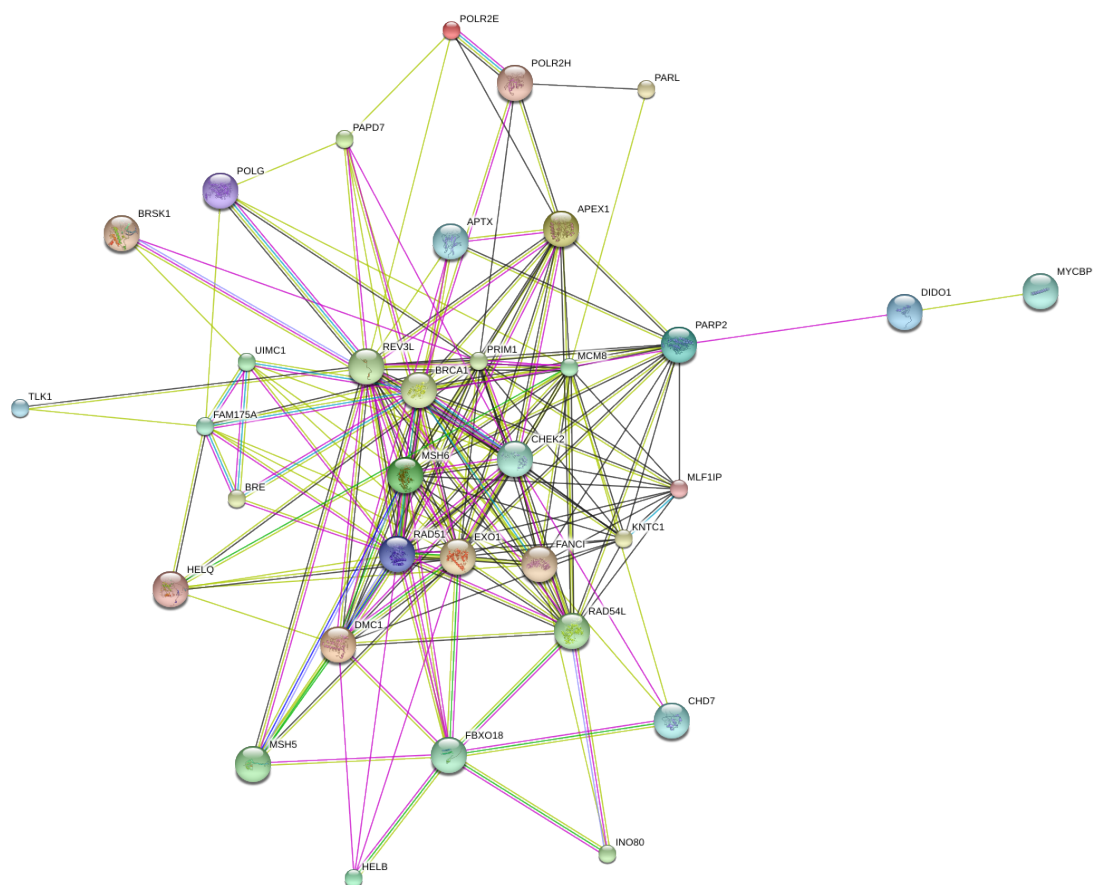
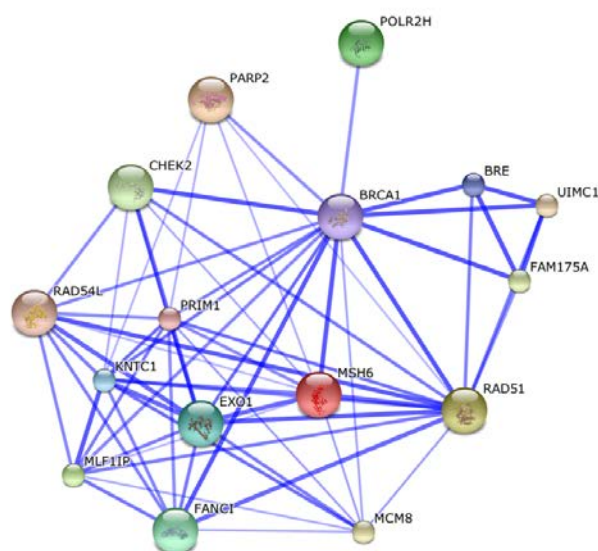
Brain eQTL studies included brain cortex (19222302, 19361613, 22685416), cerebellar cortex (25174004), cerebellum (20485568, 22685416, 22212596, 22832957, 23622250), frontal cortex (20485568, 22832957, 25174004), gliomas (24607568), hippocampus (22832957, 25174004), inferior olivary nucleus (from medulla) (25174004), intralobular white matter (25174004), occipital cortex (25174004), parietal lobe (22212596), pons (20485568), pre-frontal cortex (22031444, 20351726, 22832957, 23622250), putamen (at the level of anterior commissure) (25174004), substantia nigra (25174004), temporal cortex (20485568, 22685416, 22832957, 25174004), thalamus (22832957) and visual cortex (23622250).

Additional eQTL data was integrated from online sources including ScanDB, the Broad Institute GTex browser, and the Pritchard Lab (eqtl.uchicago.edu). Cerebellum, parietal lobe and liver eQTL data was downloaded from ScanDB and *cis*-eQTLs were limited to those with $P < 1.0\text{E-}6$ and trans-eQTLs with $P < 5.0\text{E-}8$. The top 1000 eQTL results were downloaded from the GTex Browser at the Broad Institute for 9 tissues on 11/26/2013: thyroid, leg skin (sun exposed), tibial nerve, tibial artery, skeletal muscle, lung, heart (left ventricle), whole blood, and subcutaneous adipose (23715323). All GTex results had associations with $P < 8.4\text{E-}07$.

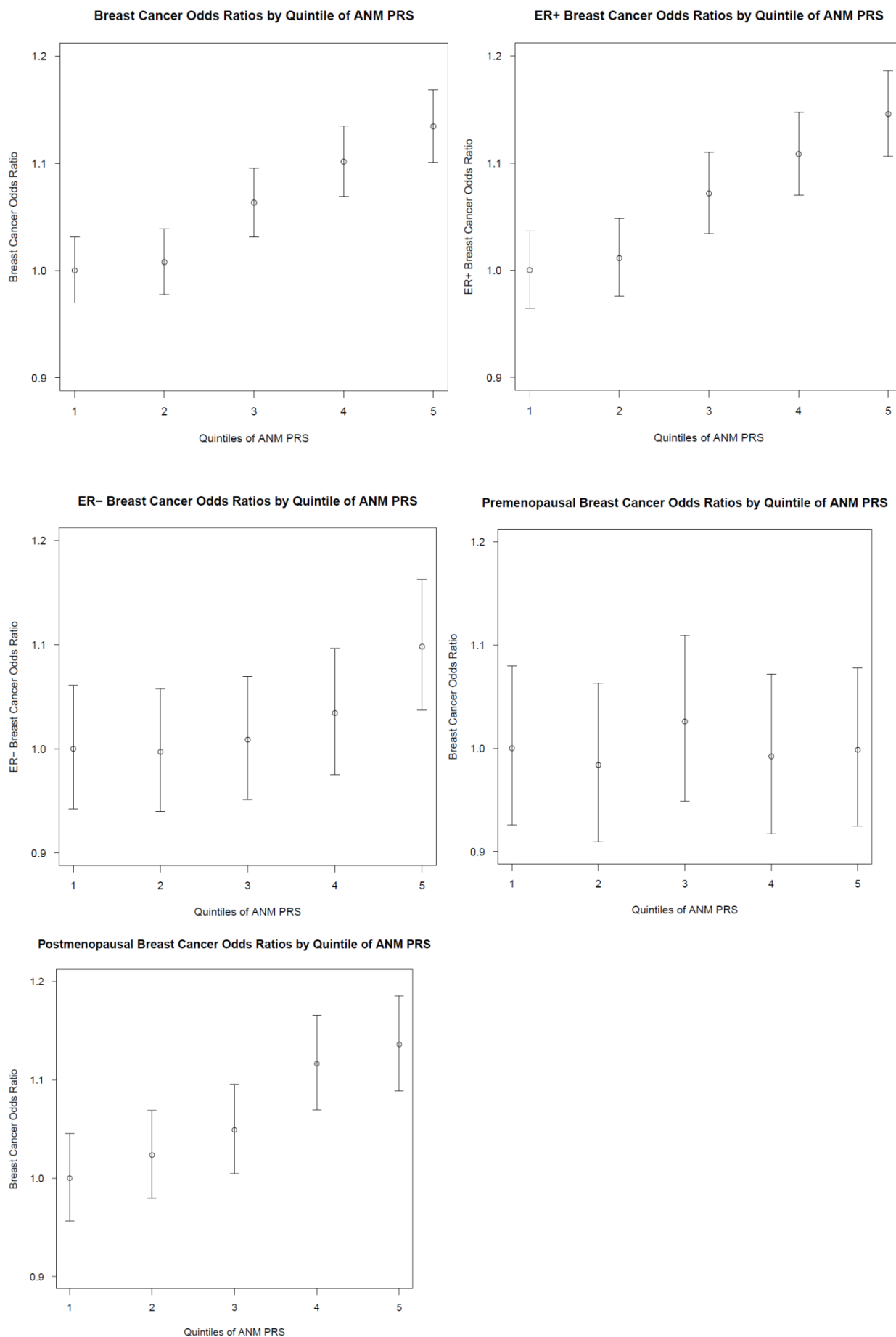
Supplementary Figures



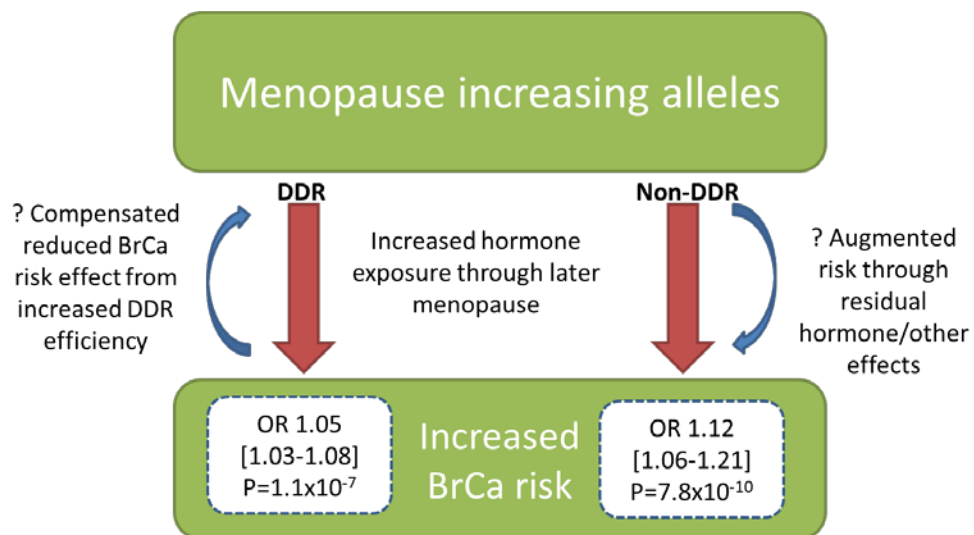
Supplementary Figure 1 Study-specific test statistics and allele frequencies for the exome-chip variants in *HELB*.

a**b**

Supplementary Figure 2 STRING analysis of genes highlighted from GWAS. **(a)** Connections for 34 genes highlighted as being involved in DDR at loci associated with age at menopause. **(b)** Genes that are directly linked to *BRCA1* from the list of highlighted genes in **Table 1**. Weight of connecting line indicates the strength of the evidence for the connection.



Supplementary Figure 3 Breast cancer ORs by quintile of ANM polygenic risk score (PRS) (quintiles defined in BC controls). The CIs are floating confidence intervals⁶⁰.



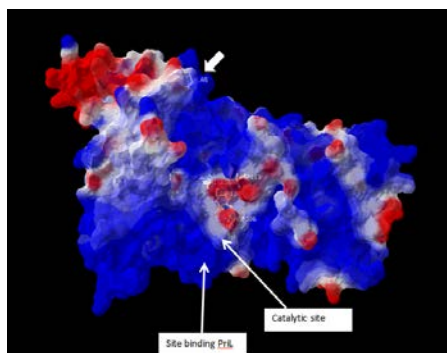
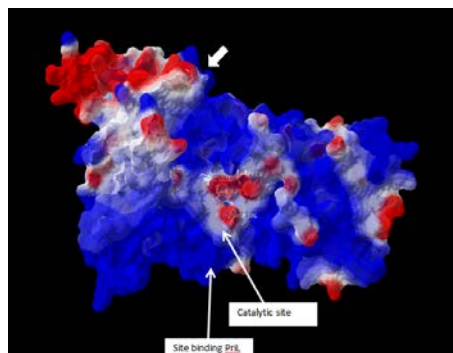
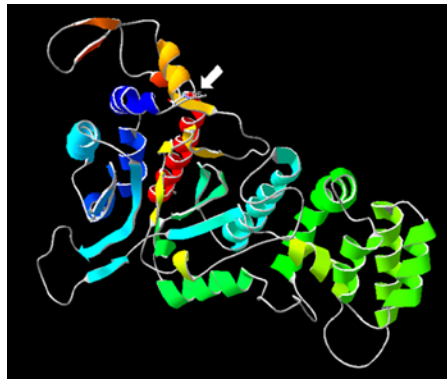
Supplementary Figure 4 Proposed mechanism of effect of SNPs on breast cancer risk.

PRIM1

Structure with reference allele



Structure with alternate allele

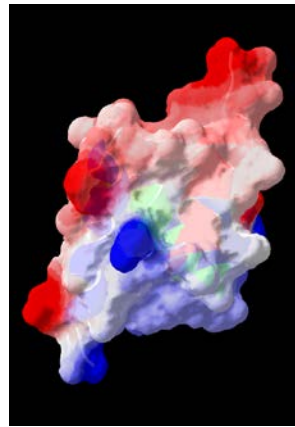
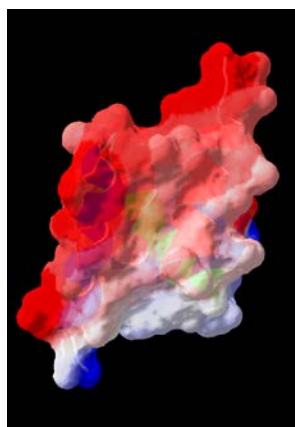


NBR1

Structure with reference allele



Structure with alternate allele



Supplementary Figure 5 SWISS-MODEL predictions for two of the variants, in *PRIM1* and *NBR1*, which may affect protein function.