Supplementary Note

Acknowledged consortia members and affiliations

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


1 The Institute of Cancer Research, 15 Cotswold Road, Sutton, Surrey, SM2 5NG, UK, 2 Royal Marsden NHS Foundation Trust, Fulham and Sutton, London and Surrey, UK, 3 Centre for Cancer Genetic Epidemiology, Department of Public Health and Primary Care, University of Cambridge, Strangeways Laboratory, Worts Causeway, Cambridge, UK, 4 University of Warwick, Coventry, UK, 5 Cancer Epidemiology Centre, Cancer Council Victoria, 615 St Kilda Road, Melbourne Victoria, Australia, 6 Centre for Epidemiology and Biostatistics, Melbourne School of Population and Global Health, The University of Melbourne, Victoria, Australia, 7 Department of Medical Epidemiology and Biostatistics, Karolinska Institute, Stockholm, Sweden, 8 Department of Preventive Medicine, Keck School of Medicine, University of Southern California/Norris Comprehensive Cancer Center, Los Angeles, California, USA, 9 Department of Medical Biochemistry and Genetics, University of Turku, Turku, Finland, 10 Institute of Biomedical Technology/BioMediTech, University of Tampere and FimLab Laboratories, Tampere, Finland, 11 Department of Clinical Biochemistry, Herlev Hospital, Copenhagen University Hospital, Herlev Ringvej 75, DK-2730 Herlev, Denmark, 12 Cancer Epidemiology Unit, Nuffield Department of Clinical Medicine, University of Oxford, Oxford, UK, 13 Surgical Oncology (Uro-Oncology: S4), University of Cambridge, Box 279, Addenbrooke’s Hospital, Hills Road, Cambridge, UK and Cancer Research UK Cambridge Research Institute, 14 Centre for Cancer Genetic Epidemiology, Department of Oncology, University of Cambridge, Strangeways Laboratory, Worts Causeway, Cambridge, UK, 15 Cambridge Institute of Public Health, University of Cambridge, Forvie Site, Robinson Way, Cambridge CB2 0SR, 16 Division of Public Health Sciences, Fred Hutchinson Cancer Research Center, Seattle, Washington, USA, 17 Department of Epidemiology, School of Public Health, University of Washington, Seattle, Washington, USA, 18 International Epidemiology Institute, 1455 Research Blvd., Suite 550, Rockville, MD 20850, 19 Mayo Clinic, Rochester, Minnesota, USA, 20 Department of Urology, University Hospital Ulm, Germany, 21 Institute of Human Genetics University Hospital Ulm, Germany, 22 Brigham and Women's Hospital/Dana-Farber Cancer Institute, 23 Washington University, St Louis, Missouri, 24 International Hereditary Cancer Center, Department of Genetics and Pathology, Pomeranian Medical University, Szczecin, Poland, 25 Division of Genetic Epidemiology, Department of Medicine, University of Utah School of Medicine, 26 Division of Clinical Epidemiology and Aging Research & Division of Preventive Oncology, German Cancer Research Center, Heidelberg Germany, 27 German Cancer Consortium (DKTK), German Cancer Research Center (DKFZ), Heidelberg Germany, 28 Division of Cancer Prevention and Control, H. Lee Moffitt Cancer Center, 29 Magnolia Dr., Tampa, Florida, USA, 30 Molecular Medicine Center and Department of Medical Chemistry and Biochemistry, Medical University - Sofia, 2 Zdrave St, 1431, Sofia, Bulgaria, 31 Australian 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, 32 Department of Genetics, Portuguese Oncology Institute, Porto, Portugal and Biomedical Sciences Institute (ICBAS), Porto University, Porto, Portugal, 33 The 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
Nature Genetics: doi:10.1038/ng.3412
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 Beasley 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 Institute, 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 ONN, 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 Services, 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 Kefferd 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 Pachtser 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 Purser 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
Raghu 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
HK Finucane is supported by R03 CA173785 and the Fannie and John Hertz Foundation
### Study acknowledgments

#### 1) GWAS

<table>
<thead>
<tr>
<th>Study name / acronym</th>
<th>Full study name</th>
<th>Acknowledgments and sources of funding</th>
</tr>
</thead>
<tbody>
<tr>
<td>AGES</td>
<td>Age, Gene/Environment Susceptibility Study</td>
<td>The researchers are indebted to the participants for their willingness to participate in the study. This study has been funded by NIH contract N01-AG-1-2100, the NIA Intramural Research Program, Hjartavernd (the Icelandic Heart Association), and the Althingi (the Icelandic Parliament). The study is approved by the Icelandic National Bioethics Committee, VSN: 00-063.</td>
</tr>
<tr>
<td>ARIC</td>
<td>Atherosclerosis Risk in Communities</td>
<td>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 R01HL08694; 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) (5R2CHL102419).</td>
</tr>
<tr>
<td>CARL</td>
<td>Cancer Aids Registries Linkage</td>
<td>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.</td>
</tr>
<tr>
<td>CHS</td>
<td>Cardiovascular Health Study</td>
<td>Cardiovascular Health Study: This CHS research was supported by NHLBI contracts HHSN268201200036C, HHSN268200800007C, HHSN268200960009C, N01HC55222, N01HC55279, N01HC55080, N01HC55081, N01HC55082, N01HC55083, N01HC55086; 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 <a href="http://chs-nhlbi.org/">http://chs-nhlbi.org/</a>. 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.</td>
</tr>
<tr>
<td>Cilento</td>
<td></td>
<td>We thank the populations of Cilento for their participation in the study. This work was supported by grants from the Italian Ministry of Universities (FIRB-RBNE08NKH7, INTEROMICS Flagship Project), the Assessorato Ricerca Regione Campania, the Fondazione con il SUD (2011-PDR-13) and the Istituto Banco di Napoli - Fondazione to MC.</td>
</tr>
<tr>
<td>COGs</td>
<td>Breast Cancer Association Consortium</td>
<td>This study would not have been possible without the contributions of the following: Per Hall (COGS); Douglas F. Easton, Paul Pharoah, Kyriaki Michailidou, Manjeet K. Bolla, Qin Wang (BCAC), Andrew Berchuck (OCAC), Rosalind A. Heles, Douglas F. Easton, Ali Amin Al Olama, Zsofia Kote-Jarai, Sara Benlloch (PRACTICAL), Georgia Chenevix-Trench, Antonis Antoniou, Lesley McGuffog, Fergus Couch and Ken Offit (CIMBA), Joe Dennis, Alison M. Dunning, Andrew Lee, and Ed Dicks, Craig Luccarini and the staff of the Centre for Genetic Epidemiology Laboratory, Javier Benitez, Anna Gonzalez-Neira and the staff of the CNIO genotyping unit, Jacques Simard and Daniel C. Tessier, Francois Bacot, Daniel Vincent, Sylvie LaBoissière and Frederic Robidoux and the staff of the McGill University and Géomé Québec Innovation Centre, Stig E. Bojesen, Sune F. Nielsen, Borge G. Nordestgaard, and the staff of the Copenhagen DNA laboratory, and Julie M. Cunningham, Sharon A. Windebank, Christopher A. Hilker, Jeffrey Meyer and the staff of Mayo Clinic Genotyping Core Facility. Funding for the iCOGS infrastructure came from: the European Community's Seventh Framework Programme under grant agreement n° 223175 (HEALTH-F2-2009-223175) (COGS), Cancer Research UK (C1287/A10118, C1287/A 10710, C12292/A11174, C1281/A12014, C5047/A8384, C5047/A10507, C5047/A10692), the National Institutes of Health (CA128978) and Post-Cancer GWAS initiative (1U19 CA148537, 1U19 CA148065 and 1U19 CA148112 - the GAME-ON initiative), the Department of Defence (W81XWH-10-1-0341), the Canadian Institutes of Health Research (CIHR) for the CIHR Team in Familial Risks of Breast Cancer, Komen Foundation for the Cure, the Breast Cancer Research Foundation, and the Ovarian Cancer Research Fund. This work was also supported by grant UM1.</td>
</tr>
</tbody>
</table>
CA164920 from the National Cancer Institute. Genotyping was supported by CRUK ref: C8197/A16565.

OBCS thanks Arja Jukkola-Vuorinen, Mervi Grip, Sáila Kauppila, Kari Mononen and Meeri Otsukka for data collection and sample preparation. OBCS was supported by the Academy of Finland (grant number 250083, 122715 and Center of Excellence grant number 251314), the Finnish Cancer Foundation, the Sigrid Juselius Foundation, the University of Oulu, the University of Oulu Support Foundation and the special Governmental EVO funds for Oulu University Hospital-based research activities.

The GENICA Network: Dr. Margarete Fischer-Bosch-Institute of Clinical Pharmacology, Stuttgart, and University of Tübingen, Germany [HB, Wing-Yee Lo, Christina Justenhoven], German Cancer Consortium (DKTK) and German Cancer Research Center (DKFZ) [HB], Department of Internal Medicine, Evangelische Kliniken Bonn gGmbH, Johanniter Krankenhaus, Bonn, Germany [Yon-Dschun Ko, Christian Baisch], Institute of Pathology, University of Bonn, Germany [Hans-Peter Fischer], Molecular Genetics of Breast Cancer, Deutsches Krebsforschungszentrum (DKFZ), Heidelberg, Germany [Ute Hamann], Institute for Prevention and Occupational Medicine of the German Social Accident Insurance, Institute of the Ruhr University Bochum (IPA), Bochum, Germany [TB, Beate Pesch, Sylvia Rabstein, Anne Lotz]; and Institute of Occupational Medicine and Maritime Medicine, University Medical Center Hamburg- Eppendorf, Germany [Volker Harth].

The GENICA was funded by the Federal Ministry of Education and Research (BMBF) Germany grants 01KW9975/5, 01KW9976/8, 01KW9977/0 and 01KW0114, the Robert Bosch Foundation, Stuttgart, Deutsches Krebsforschungszentrum (DKFZ), Heidelberg, the Institute for Prevention and Occupational Medicine of the German Social Accident Insurance, Institute of the Ruhr University Bochum (IPA), Bochum, as well as the Department of Internal Medicine, Evangelische Kliniken Bonn gGmbH, Johanniter Krankenhaus, Bonn, Germany.

We thank Breakthrough Breast Cancer and the Institute of Cancer Research for support and funding of the Breakthrough Generations Study, and the study participants, study staff, and the doctors, nurses and other health care providers and health information sources who have contributed to the study. We acknowledge NHS funding to the Royal Marsden/ICR NIHR Biomedical Research Centre.

MARIE thanks Anja Rudolph, Petra Seibold, Judith Heinz, Nadia Obi, Sabine Behrens, Ursula Elffer, Muhabbet Celik for study management and research, data collection and preparation. MARIE was supported by the Deutsche Krebshilfe e.V. [70-2892-BR I, 106332, 108253, 108419], the Hamburg Cancer Society, the German Cancer Research Center (DKFZ) and the Federal Ministry of Education and Research (BMBF) Germany [01KH0402].

<table>
<thead>
<tr>
<th>Colaus</th>
<th>Etude Cohorte Lausannoise</th>
</tr>
</thead>
<tbody>
<tr>
<td>CROATIA-Korcula</td>
<td>CROATIA-Korcula</td>
</tr>
<tr>
<td>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.</td>
<td></td>
</tr>
<tr>
<td>CROATIA-Split</td>
<td>CROATIA-Split</td>
</tr>
<tr>
<td>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).</td>
<td></td>
</tr>
<tr>
<td>CROATIA-Vis</td>
<td>CROATIA-Vis</td>
</tr>
<tr>
<td>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. Medical Research Council UK and the Ministry of Science, Education and Sport in the Republic of Croatia (number 108-1080315-0302).</td>
<td></td>
</tr>
<tr>
<td>deCODE</td>
<td>Republic of Croatia (number 108-1080315-0302).</td>
</tr>
<tr>
<td>---</td>
<td>---</td>
</tr>
<tr>
<td>EGCUT (370k)</td>
<td>Estonian Genome Center, University of Tartu</td>
</tr>
<tr>
<td>EGCUT OmniX</td>
<td>Estonian Genome Center, University of Tartu</td>
</tr>
<tr>
<td>ERF</td>
<td>Erasmus Rucphen Family study</td>
</tr>
<tr>
<td>FHS</td>
<td>Framingham Heart Study</td>
</tr>
<tr>
<td>Generation Scotland</td>
<td>Generation Scotland: Scottish Family Health Study</td>
</tr>
<tr>
<td>GENOA</td>
<td>Genetic Epidemiology Network of Arteriopathy</td>
</tr>
<tr>
<td>Study</td>
<td>Heredity and Phenotype Intervention (HAPI) Heart Study</td>
</tr>
<tr>
<td>-------------------------------</td>
<td>-------------------------------------------------------</td>
</tr>
<tr>
<td>HAPI Heart Study</td>
<td>The Health, Aging, and Body Composition Study</td>
</tr>
<tr>
<td>HealthABC</td>
<td>Health and Retirement Study</td>
</tr>
<tr>
<td>HRS</td>
<td>Health and Retirement Study</td>
</tr>
<tr>
<td>InChianti</td>
<td>Invecchiare in Chianti</td>
</tr>
<tr>
<td>INGI-FVG</td>
<td>Genetic Park of Friuli Venezia Giulia Project</td>
</tr>
<tr>
<td>INGI-VB</td>
<td>Val Borbera Isolated Population Project</td>
</tr>
<tr>
<td>InterAct cohort/ InterAct cases</td>
<td>European Prospective Investigation into Cancer &amp; Nutrition - InterAct</td>
</tr>
<tr>
<td>KORA F3/ KORA F4</td>
<td>Cooperative Health Research in the Region of Augsburg (follow-up 3) / (follow-up 4)</td>
</tr>
</tbody>
</table>
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_OG, 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, Spinozepremie 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 MH08457-06), 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; Nederlands Exome Chip Project) of the Biobanking and Biomolecular Resource 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). 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 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 01Z20403), 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 03IK012 and 03Z21CN22) 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, 03Z21CN22 and 03IS2061A.
<table>
<thead>
<tr>
<th>TwinsUK/ TwinsUKII/ TwinsUKIII</th>
<th>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.</th>
</tr>
</thead>
<tbody>
<tr>
<td>WGHS</td>
<td>Women’s Genome Health Study</td>
</tr>
<tr>
<td>Study name / acronym</td>
<td>Full study name</td>
</tr>
<tr>
<td>----------------------</td>
<td>----------------</td>
</tr>
<tr>
<td>1958BC</td>
<td>1958 National Child Development Study (also known as the 1958 Birth Cohort Study)</td>
</tr>
<tr>
<td>ARIC</td>
<td>Atherosclerosis Risk in Communities HapMap analysis</td>
</tr>
<tr>
<td>CHS</td>
<td>Cardiovascular Health Study</td>
</tr>
<tr>
<td>Fenland</td>
<td></td>
</tr>
<tr>
<td>FHS</td>
<td>Framingham Heart Study</td>
</tr>
<tr>
<td>INGI-VB</td>
<td>Val Borbera Isolated Population Project</td>
</tr>
<tr>
<td>---------</td>
<td>--------------------------------------</td>
</tr>
<tr>
<td></td>
<td>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 Vigano for technical help, Massimiliano Cocca for building the analysis platform. The research was supported by funds from Compagnia di San Paolo, Torino, Italy; Fondazione Caripio, Italy and Ministry of Health, Ricerca Finalizzata 2008 and CCM 2010, PRIN 2009 and Telethon, Italy to DT.</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>InterAct Cases/InterAct Subcohort</th>
<th>European Prospective Investigation into Cancer &amp; Nutrition - InterAct</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>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).</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>KORA</th>
<th>Cooperative Health Research in the Region of Augsburg (follow-up 4)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>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.</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>MESA</th>
<th>Multi-Ethnic Study of Atherosclerosis</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>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.</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Amish</th>
<th>Old Order Amish Study</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>U01-HL72515, U01-HL84756, R01-088119, P30-DK072488, K01-HL116770</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Cambridge Cancer</th>
<th>The EMBRACE, SEARCH (breast cancer and ovarian cancer) and SIBS studies</th>
</tr>
</thead>
</table>
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.

South East Thames Regional Genetics Service, Guy’s Hospital London: Louise Izatt, Chris Jacobs, Caroline Langman.


North East Thames Regional Genetics Service, NE Thames, London: Lucy Side, Alison Male, Cheryl Berlin.

Nottingham Centre for Medical Genetics, Nottingham: Jacqueline Eason, Rebecca Collier.


CRUK ref: C8197/A16565, CRUK ref: C1287/A8459, CRUK ref: A490/A10124

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 Bankcroft are supported by Cancer Research UK Grant C5047/A8385.

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).
<table>
<thead>
<tr>
<th>Rotterdam Study I</th>
<th>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; Neterlands Exome Chip Project) of the Biobanking and Biomolecular Research Infrastructure Netherlands (BBMRI-NL; <a href="http://www.bbmri.nl">www.bbmri.nl</a>). 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.</th>
</tr>
</thead>
<tbody>
<tr>
<td>SardiNIA</td>
<td>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”.</td>
</tr>
<tr>
<td>SHIP/SHIP-TREND Study of Health in Pomerania / Study of Health in Pomerania - TREND</td>
<td>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</td>
</tr>
<tr>
<td>WGHS Women’s Genome Health Study</td>
<td>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.</td>
</tr>
<tr>
<td>WHI Women’s Health Initiative</td>
<td>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: <a href="http://www.whi.org/researchers/Documents%20%20Write%20a%20Paper/WHI%20Investigator%20Short%20List.pdf">http://www.whi.org/researchers/Documents%20%20Write%20a%20Paper/WHI%20Investigator%20Short%20List.pdf</a></td>
</tr>
</tbody>
</table>
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.

**Val Borbera Isolated Population Project**: The funders had no role in study design, data collection and analysis, decision to publish, or preparation of the manuscript.

**Women’s Genome Health Study**: Support from Amgen to PMR and DIC for the Women’s Genome Health Study.

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.


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) 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 (22031444, 20351726, 22832957, 23622250), 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 (24907047).

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 $P < 1.0E-6$ and trans-eQTLs with $P < 5.0E-8$. 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 $P < 8.4E-07$. 

Nature Genetics: doi:10.1038/ng.3412
**Supplementary Figure 1** Study-specific test statistics and allele frequencies for the exome-chip variants in *HEL*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.
Supplementary Figure 5 SWISS-MODEL predictions for two of the variants, in PRIM1 and NBR1, which may affect protein function.