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Colaus	Etude Cohorte Lausannoise	
CROATIA-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. xome 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.
CROATIA-Split	CROATIA-Split	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).
CROATIA-Vis	CROATIA-Vis	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).
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, JeannetteVergeer 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. QLG2-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 (Al) 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).

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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; Neterlands 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).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

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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 (Al) 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.

		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 West Thames Regional Genetics Service, Harrow: Huw Dorkins. Leicestershire Clinical Genetics Service, Leicester: Julian Barwell. 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 Lalloo, Jane Taylor. 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. Northern Clinical Genetics Service, Newcastle: Alex Henderson, Oonagh Claber, Irene Jobson. Oxford Regional Genetics Service, Oxford: Lisa Walker, Diane McLeod, Dorothy Halliday, Sarah Durell, Barbara Stayner. The Institute of Cancer Research and Royal Marsden NHS Foundation Trust: Ros Eeles, Nazneen Rahman, Elizabeth Bancroft, Elizabeth Page, Audrey Ardern-Jones, Kelly Kohut, Jennifer Wiggins, Jenny Pope, Sibel Saya, Natalie Taylor, Zoe Kemp and Angela George. North Trent Clinical Genetics Service, Sheffield: Jackie Cook, Oliver Quarrell, Cathryn Bardsley. South West Thames Regional Genetics Service, London: Shirley Hodgson, Sheila Goff, Glen Brice, Lizzie Winchester, Charlotte Eddy, Vishakha Tripathi, Virginia Attard. Wessex Clinical Genetics Service, Princess Anne Hospital, Southampton: Diana Eccles, Anneke Lucassen, Gillian Crawford, Donna McBride, Sarah Smalley. 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 Lalloo 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.
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. xome 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	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; 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 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/ SHIP-TREND	Study of Health in Pomerania / Study of Health in Pomerania - TREND	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
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.
WHI	Women's Health Initiative	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

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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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

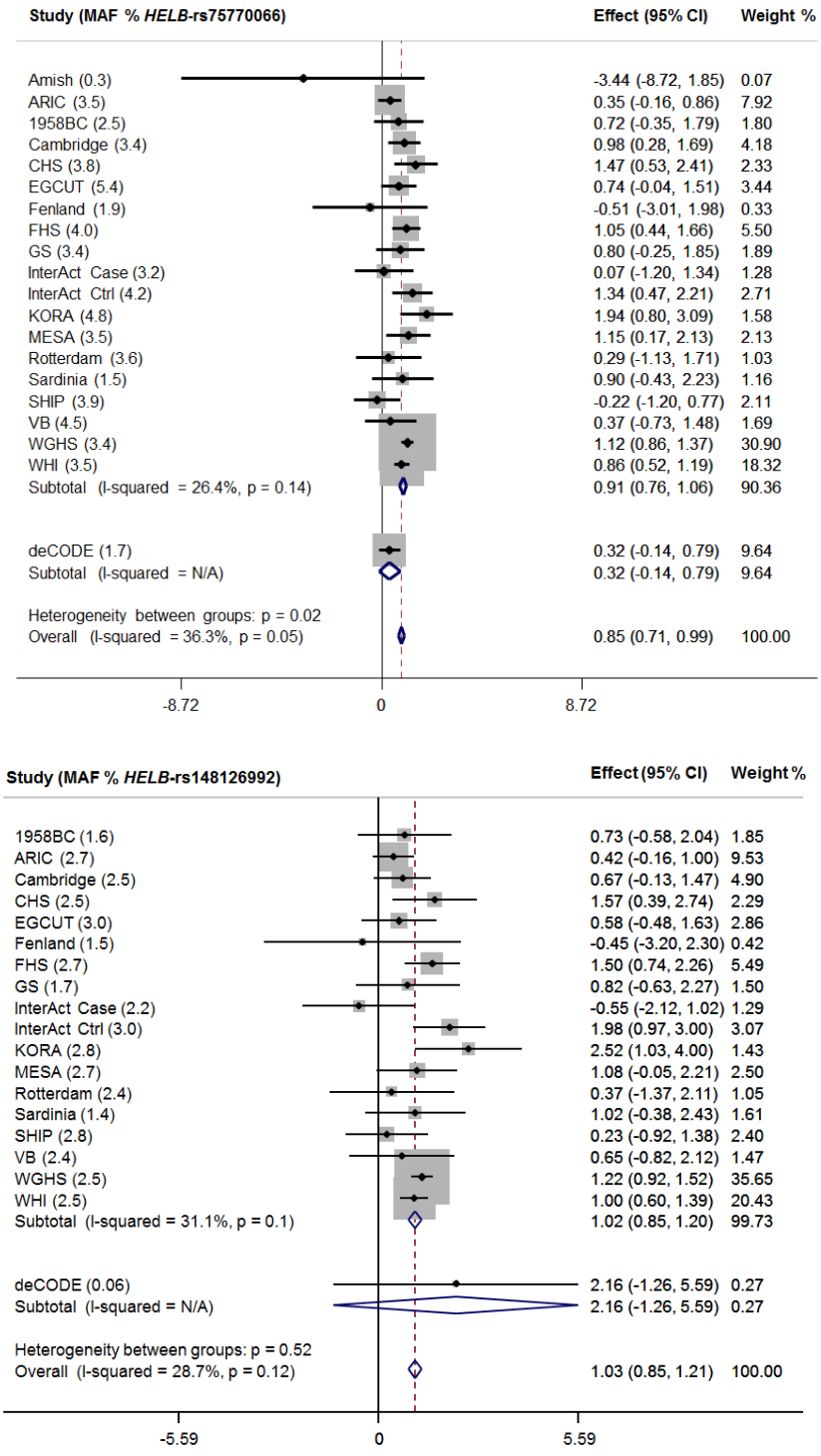
Blood cell related eQTL studies included fresh lymphocytes (17873875), fresh leukocytes (19966804), leukocyte samples in individuals with Celiac disease (19128478), whole blood samples (18344981, 21829388, 22692066, 23818875, 23359819, 23880221, 24013639, 23157493, 23715323, 24092820, 24314549, 24956270, 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), occiptal cortex (25174004), parietal lobe (22212596), pons (20485568), pre-frontal cortex (22031444, 20351726, 22832957, 23622250), putamen (at the level of anterior commussure) (25174004), substantia nigra (25174004), temporal cortex (20485568, 22685416, 22832957, 25174004), thalamus (22832957) and visual cortex (23622250).

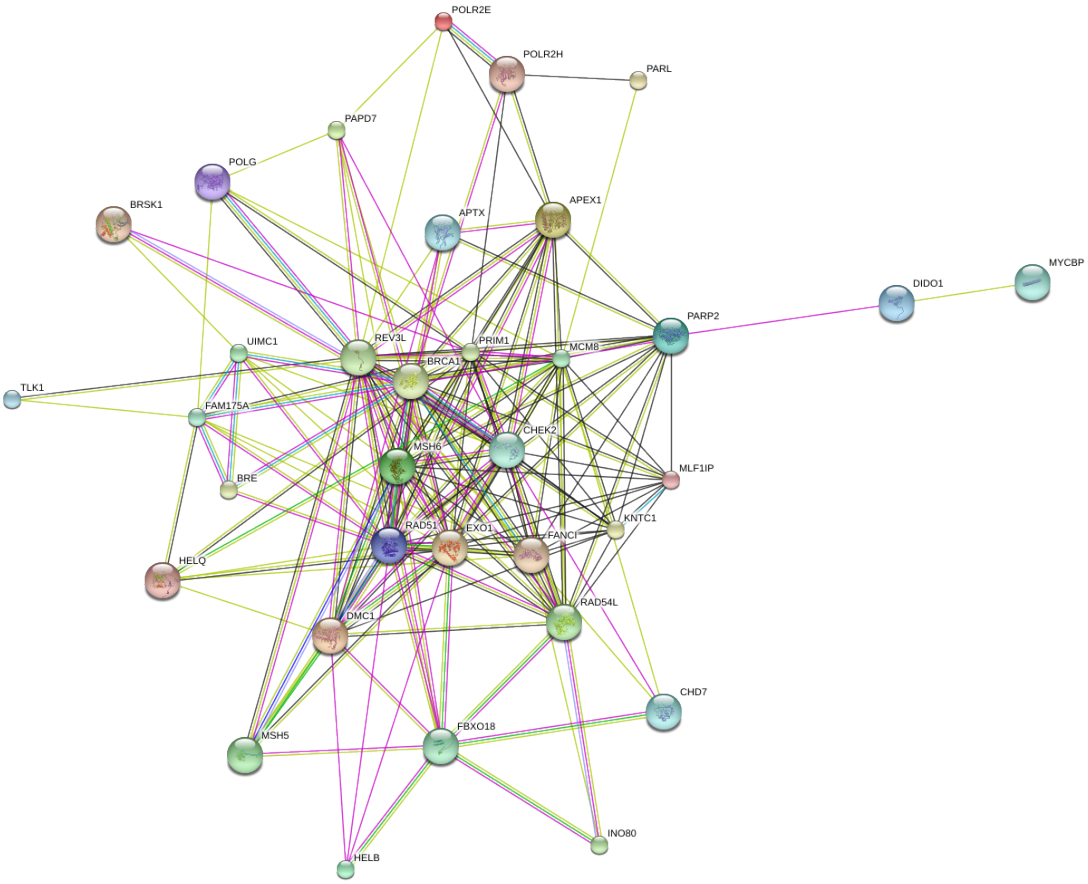
Additional eQTL data was integrated from online sources including ScanDB, the Broad Institute GTex browser, and the Pritchard Lab (eqtl.uchicago.edu). Cerebellum, parietal lobe and liver eQTL data was downloaded from ScanDB and *cis*-eQTLs were limited to those with $P < 1.0E-6$ and trans-eQTLs with $P < 5.0E-8$. 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 $P < 8.4E-07$.

Supplementary Figures

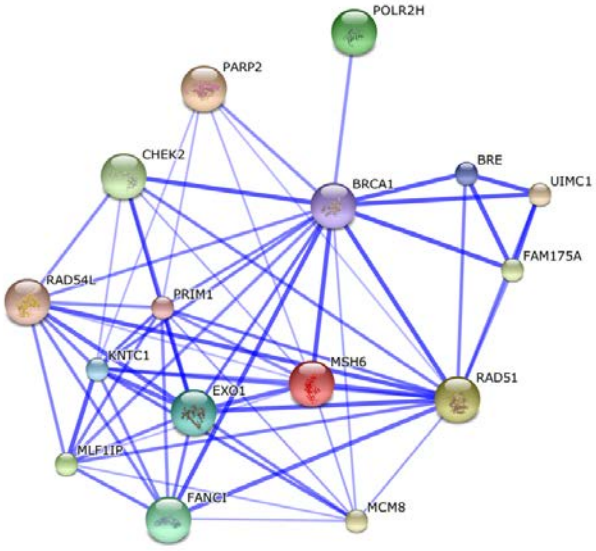


Supplementary Figure 1 Study-specific test statistics and allele frequencies for the exome-chip variants in *HELB*.

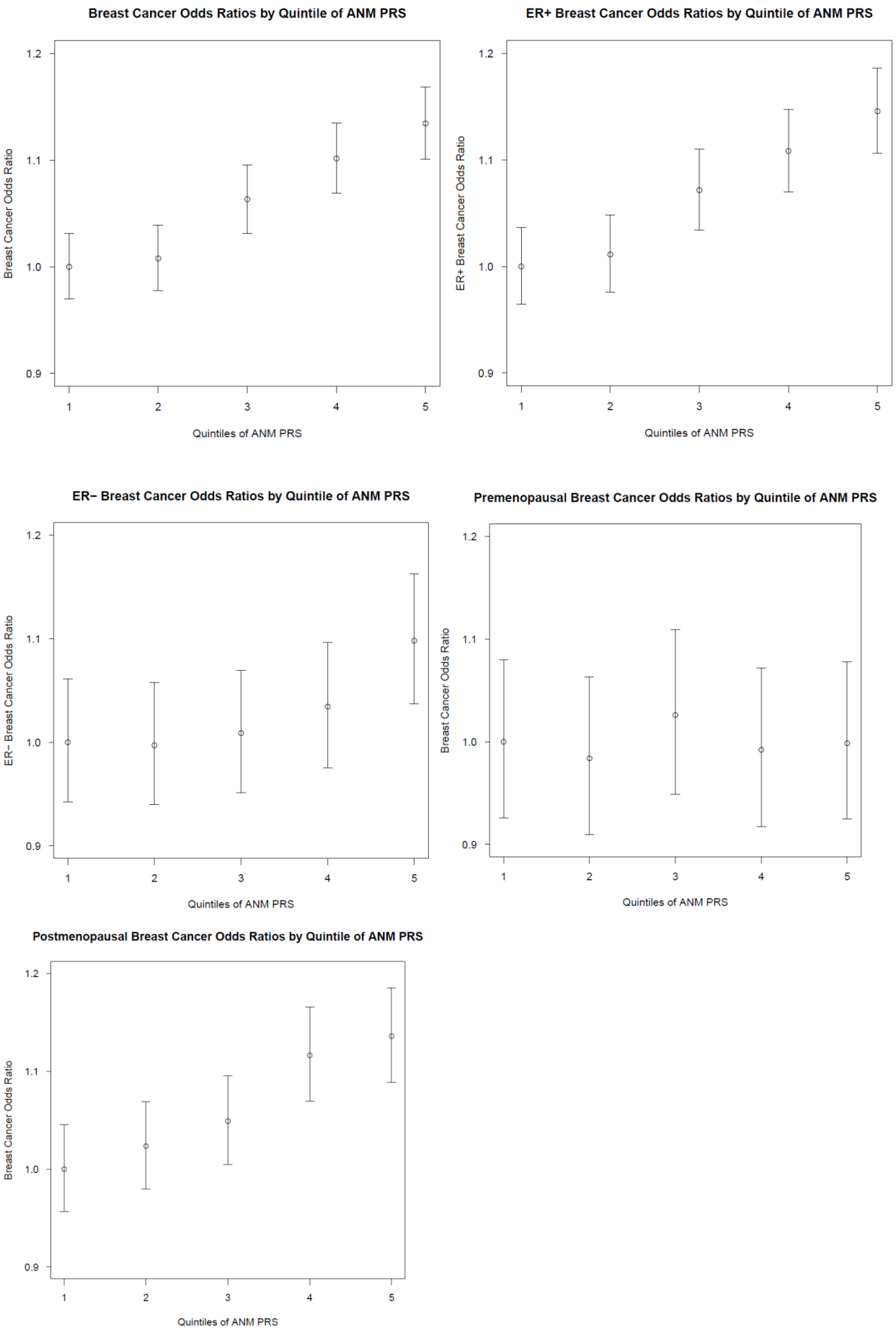
a



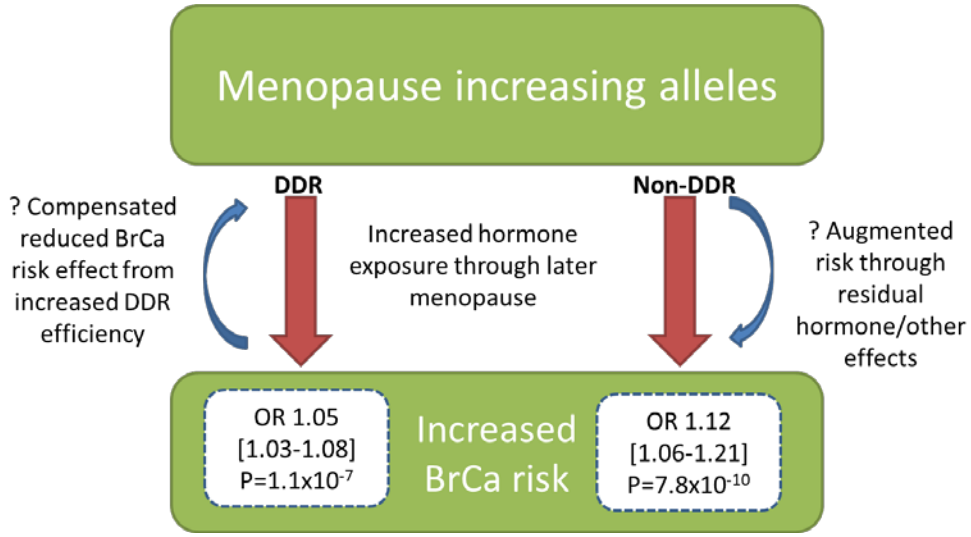
b



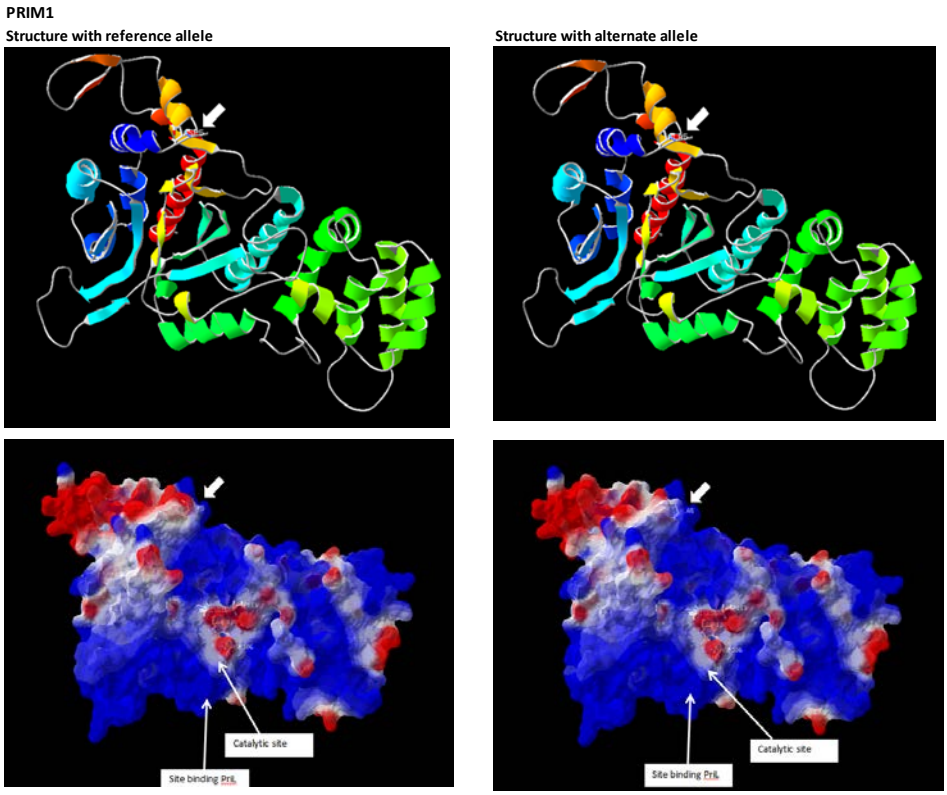
Supplementary Figure 2 STRING analysis of genes highlighted from GWAS. (**a**) Connections for 34 genes highlighted as being involved in DDR at loci associated with age at menopause. (**b**) Genes that are directly linked to *BRCA1* from the list of highlighted genes in **Table 1**. Weight of connecting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

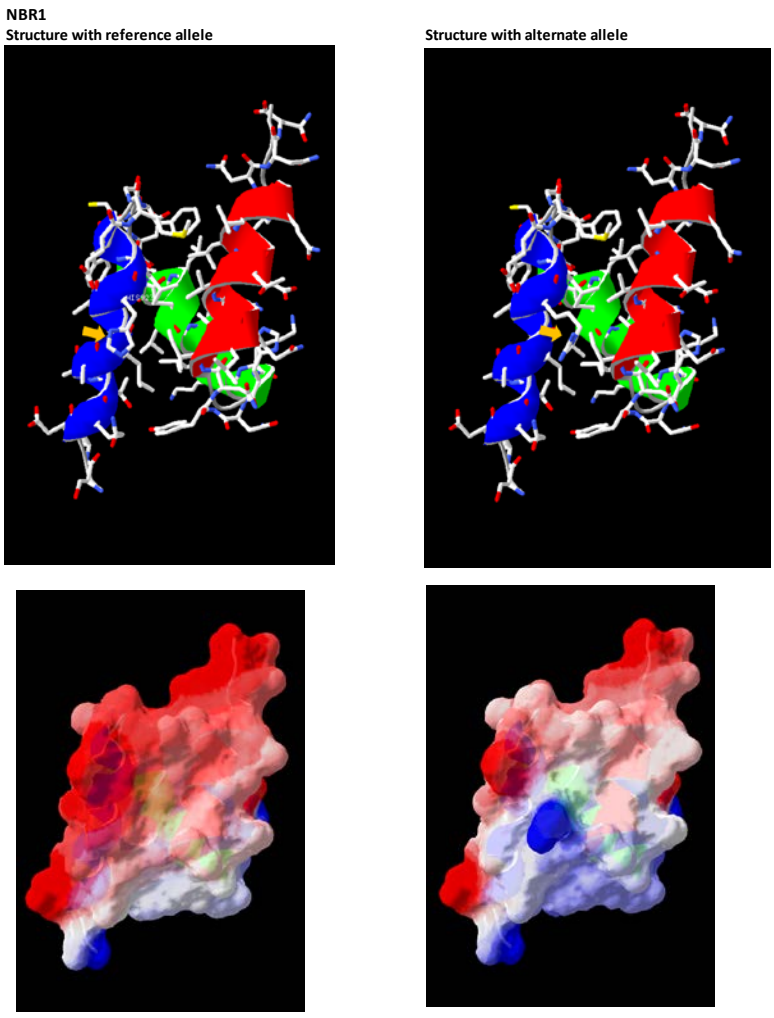


Supplementary Figure 3 Breast cancer ORs by quintile of ANM polygenic risk score (PRS) (quintiles defined in BC controls). The CIs are floating confidence intervals[60].



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.

Table	Title
S1	Study info GWAS
S2	GWAS clumped
S3	GWAS Conditional results
S4	GCTA variance explained
S5	Partitioning heritability
S6	Study info Exome
S7	Exome SV dis+rep
S8	WGHS conditional analysis
S9	GRAIL results
S10	MAGENTA (overall)
S11	eQTLs - full results
S12	STRING results
S13	MAGENTA (custom)
S14	MAGENTA cust. POF
S15	MAGENTA cust. Ovarian.
S16	GWAS catalogue look ups
S17	Genetic correlations
S18	Score BMI > Meno.
S19	Binomial Meno > BMI
S20	Score AAM > Meno.
S21	Score Meno. > AAM
S22	S24 MAGENTA cust. Puberty
S23	Score BC > Meno.
S24	BCAC_stratified
S25	Score Meno. > PC
S26	Non-syn. vars. at r-sq>0.8
S27	Functional_sift_poly_model

Description
Study level information for the contributing GWAS studies
The univariate results for the GWAS analysis, showing the nearest genes and genes within 500kb
The results from GCTA showing secondary signals, and also the highlighted genes from the pathway analysis
Variance explained estimates from GCTA using the InterAct cohort data
Results of 10 tissue categories from Broad analysis
Study level information for the contributing Exome studies
The Exome variants taken forward for replication with the results in both the discovery and replication cohorts
Conditional analysis of the GWAS and Exome chip signals in the WGHS cohort
Results from GRAIL analysis
The results from the default MAGENTA analysis
All eQTLs across all tissues with a significant association for the top SNPs
Details of the protein–protein connections identified from STRING
MAGENTA results from the three custom pathways (POI, ovarian function, monogenic puberty)
Details of the genes inputted for the custom POF MAGENTA pathway
Details of the genes inputted for the custom early menopause MAGENTA pathway
GRASP and NHGRI look up of GWAS associated traits
Genetic correlations across a range of phenotypes using the Broad Group Method
Details of the BMI to age at menopause score analysis
Details of the binomial analysis of directional consistency of age at menopause SNPs on BMI
Details of the age at menarche to age at menopause score analysis
Details of the age at menopause to age at menarche score analysis
MAGENTA analysis of enrichment for puberty timing genes
Details of the breast cancer to age at menopause score analysis
Results from age stratified breast cancer analysis
Details of the menopause score to prostate cancer analysis
Non-synonymous variants in linkage disequilibrium (r2>0.8) with the GWAS signals (from HaploReg v2)
Functional significance of non-synonymous variants, using SIFT, Polyphen and SWISS-MODEL