## Diana M Eccles

## List of Publications by Year

 in descending order[^0]

UK recommendations for < i > SDHA < /i> germline genetic testing and surveillance in clinical practice.
Journal of Medical Genetics, 2023, 60, 107-111.

High likelihood of actionable pathogenic variant detection in breast cancer genes in women with very early onset breast cancer. Journal of Medical Genetics, 2022, 59, 115-121.
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Risk-Adjusted Cancer Screening and Prevention (RiskAP): Complementing Screening for Early Disease Detection by a Learning Screening Based on Risk Factors. Breast Care, 2022, 17, 208-223.

Quantifying prediction of pathogenicity for within-codon concordance (PM5) using 7541 functional classifications of BRCA1 and MSH2 missense variants. Genetics in Medicine, 2022, 24, 552-563.

Risks of breast and ovarian cancer for women harboring pathogenic missense variants in BRCA1 and
5 BRCA2 compared with those harboring protein truncating variants. Genetics in Medicine, 2022, 24, 119-129.

6 Quantifying evidence toward pathogenicity for rare phenotypes: The case of succinate dehydrogenase genes, SDHB and SDHD. Genetics in Medicine, 2022, 24, 41-50.

Polygenic risk modeling for prediction of epithelial ovarian cancer risk. European Journal of Human
$7 \quad \begin{aligned} & \text { Polygenic risk modeling for predin } \\ & \text { Genetics, 2022, 30, 349-362. }\end{aligned}$

Common variants in breast cancer risk loci predispose to distinct tumor subtypes. Breast Cancer
Research, 2022, $24,2$.
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Reclassification of clinically-detected sequence variants: Framework for genetic clinicians and
9 clinical scientists by CanVIG-UK (Cancer Variant Interpretation Group UK). Genetics in Medicine, 2022, 24, 1867-1877.

Talking about Risk, UncertaintieS of Testing IN Genetics (TRUSTING): development and evaluation of an
10 educational programme for healthcare professionals about BRCA1 \& BRCA2 testing. British
Journal of Cancer, 2022, 127, 1116-1122.
Associations of a breast cancer polygenic risk score with tumor characteristics and survival..
11 Journal of Clinical Oncology, 2022, 40, 563-563.

Combining evidence for and against pathogenicity for variants in cancer susceptibility genes:
12 CanVIG-UK consensus recommendations. Journal of Medical Genetics, 2021, 58, 297-304.
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A case-only study to identify genetic modifiers of breast cancer risk for BRCA1/BRCA2 mutation
carriers. Nature Communications, 2021, 12, 1078.
The predictive ability of the 313 variantâ€"based polygenic risk score for contralateral breast cancer
14 risk prediction in women of European ancestry with a heterozygous BRCA1 or BRCA2 pathogenic
variant. Genetics in Medicine, 2021, 23, 1726-1737.
Functional annotation of the $2 q 35$ breast cancer risk locus implicates a structural variant in
15 influencing activity of a long-range enhancer element. American Journal of Human Genetics, 2021, 108,
$2.6 \quad 6$
1190-1203.
Association of germline genetic variants with breast cancer-specific survival in patient subgroups
16 defined by clinic-pathological variables related to tumor biology and type of systemic treatment. Breast Cancer Research, 2021, 23, 86.

Mendelian randomisation study of smoking exposure in relation to breast cancer risk. British Journal
of Cancer, 2021, 125, 1135-1145.

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Association of Genomic Domains in <i>BRCA1</i> and <i>BRCA2</i> with Prostate Cancer Risk and Aggressiveness. Cancer Research, 2020, 80, 624-638.
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Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. Nature Genetics, 2020, 52, 56-73.

Breast Cancer Polygenic Risk Score and Contralateral Breast Cancer Risk. American Journal of Human Genetics, 2020, 107, 837-848.
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Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses. Nature Genetics, 2020, 52, 572-581.

Psychosocial effects of whole-body MRI screening in adult high-risk pathogenic <i>TP53</i> mutation carriers: a case-controlled study (SIGNIFY). Journal of Medical Genetics, 2020, 57, 226-236.
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Pathogenic Variants in <i>CHEK2</i> Are Associated With an Adverse Prognosis in Symptomatic
Early-Onset Breast Cancer. JCO Precision Oncology, 2020, 4, 472-485.
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Cancer Variant Interpretation Group UK (CanVIG-UK): an exemplar national subspecialty multidisciplinary network. Journal of Medical Genetics, 2020, 57, 829-834.

Germline HOXB13 mutations p.G84E and p.R217C do not confer an increased breast cancer risk.
$27 \quad \begin{aligned} & \text { Germline HOXB13 mutations p.G84 } \\ & \text { Scientific Reports, } 2020,10,9688 .\end{aligned}$
Cancer prevention with aspirin in hereditary colorectal cancer (Lynch syndrome), 10-year follow-up
28 and registry-based 20-year data in the CAPP2 study: a double-blind, randomised, placebo-controlled
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220 trial. Lancet, The, 2020, 395, 1855-1863.

> Variants of uncertain clinical significance in hereditary breast and ovarian cancer genes: best
> practices in functional analysis for clinical annotation. Journal of Medical Genetics, 2020, 57, 509-518.
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30 Survival and disease characteristics of de novo versus recurrent metastatic breast cancer in a cohort of young patients. British Journal of Cancer, 2020, 122, 1618-1629.
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Obesity Alters Endoxifen Plasma Levels in Young Breast Cancer Patients: A Pharmacometric Simulation
Approach. Clinical Pharmacology and Therapeutics, 2020, 108, 661-670.

Transcriptomeâ€wide association study of breast cancer risk by estrogenâ€eceptor status. Genetic Epidemiology, 2020, 44, 442-468.
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$33 \quad \begin{aligned} & \text { A network analysis to identify mediators } \\ & \text { Nature Communications, 2020, 11, } 312 .\end{aligned}$
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prospective cohort of BRCA1 and BRCA2 mutation carriers. Breast Cancer Research, 2020, $22,8$.
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Multifocal breast cancers are more prevalent in $\langle i\rangle B R C A 2<|i\rangle$ versus <i>BRCA1</i> mutation carriers.
Journal of Pathology: Clinical Research, 2020, 6, 146-153.
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Genetic Data from Nearly 63,000 Women of European Descent Predicts DNA Methylation Biomarkers
and Epithelial Ovarian Cancer Risk. Cancer Research, 2019, 79, 505-517.

Psychosocial impact of undergoing prostate cancer screening for men with <i> <scp>BRCA</scp>1 or <scp>BRCA</scp>2</i> mutations. BJU International, 2019, 123, 284-292.

Development of Breast Cancer Choices: a decision support tool for young women with breast cancer
39 deciding whether to have genetic testing for BRCA1/2 mutations. Supportive Care in Cancer, 2019, 27,
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297-309.

40 Hereditary Breast and Ovarian Cancer Testing in the Genomic Era. JAMA Oncology, 2019, 5, 58.
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| 41 | The FANCM:p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. Npj Breast Cancer, 2019, 5, 38. | 2.3 | 28 |
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| 42 | Two truncating variants in FANCC and breast cancer risk. Scientific Reports, 2019, 9, 12524. | 1.6 | 5 |
| 43 | Interim Results from the IMPACT Study: Evidence for Prostate-specific Antigen Screening in BRCA2 Mutation Carriers. European Urology, 2019, 76, 831-842. | 0.9 | 148 |
| 44 | A Cost-effectiveness Analysis of Multigene Testing for All Patients With Breast Cancer. JAMA Oncology, 2019, 5, 1718. | 3.4 | 91 |
| 45 | Shared heritability and functional enrichment across six solid cancers. Nature Communications, 2019, 10, 431. | 5.8 | 88 |
| 46 | Large scale multifactorial likelihood quantitative analysis of <i>BRCA1</i> and <i>BRCA2</i> variants: An ENIGMA resource to support clinical variant classification. Human Mutation, 2019, 40, 1557-1578. | 1.1 | 102 |
| 47 | Evaluation of vitamin D biosynthesis and pathway target genes reveals UCT2A1/2 and EGFR polymorphisms associated with epithelial ovarian cancer in African American Women. Cancer Medicine, 2019, 8, 2503-2513. | 1.3 | 6 |
| 48 | Towards controlled terminology for reporting germline cancer susceptibility variants: an ENIGMA report. Journal of Medical Genetics, 2019, 56, 347-357. | 1.5 | 32 |
| 49 | Breast cancer in patients with germline TP53 pathogenic variants have typical tumour characteristics: the Cohort study of TP53 carrier early onset breast cancer (COPE study). Journal of Pathology: Clinical Research, 2019, 5, 189-198. | 1.3 | 18 |

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Renewed: Protocol for a randomised controlled trial of a digital intervention to support quality of life in cancer survivors. BMJ Open, 2019, 9, e024862.

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75 The Influence of Number and Timing of Pregnancies on Breast Cancer Risk for Women With BRCA1 or
BRCA2 Mutations. JNCI Cancer Spectrum, 2018, 2, pky078.

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Triple-Negative Breast Cancer Risk Genes Identified by Multigene Hereditary Cancer Panel Testing.
Journal of the National Cancer Institute, 2018, 110, 855-862.
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A Transcriptome-Wide Association Study Among 97,898 Women to Identify Candidate Susceptibility
Genes for Epithelial Ovarian Cancer Risk. Cancer Research, 2018, 78, 5419-5430.
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Variants in genes encoding small GTPases and association with epithelial ovarian cancer
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80 correlate with lymph node negative involvement and longer disease free survival: a multi-center POSH
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A transcriptome-wide association study of 229,000 women identifies new candidate susceptibility
82 genes for breast cancer. Nature Genetics, 2018, 50, 968-978.
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Surgery, 2017, 266, 165-172.

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105 Adult body mass index and risk of ovarian cancer by subtype: a Mendelian randomization study.
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> A Method to Exploit the Structure of Genetic Ancestry Space to Enhance Case-Control Studies.
> American Journal of Human Genetics, 2016, 98, 857-868.
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Genes associated with histopathologic features of triple negative breast tumors predict molecular
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$110 \quad$ Genes associated with histopathologic features of triple negative breast
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Association of vitamin D levels and risk of ovarian cancer: a Mendelian randomization study.
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112 Multiple New Susceptibility Loci Shared by at Least Two Cancer Types. Cancer Discovery, 2016, 6,
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Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. Nature
113 Communications, $2016,7,11375$.
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Prognostic value of automated KI67 scoring in breast cancer: a centralised evaluation of 8088 patients
from 10 study groups. Breast Cancer Research, 2016, 18, 104.
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## 115 Classification and Clinical Management of Variants of Uncertain Significance in High Penetrance

 Cancer Predisposition Genes. Human Mutation, 2016, 37, 331-336.1.1

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116 Selecting Patients with Ovarian Cancer for Germline BRCA Mutation Testing: Findings from Guidelines and a Systematic Literature Review. Advances in Therapy, 2016, 33, 129-150.

117 Information requirements of young women with breast cancer treated with mastectomy or breast conserving surgery: A systematic review. Breast, 2016, 25, 1-13.

Highâ€throughput automated scoring of Ki67 in breast cancer tissue microarrays from the Breast
118 Cancer Association Consortium. Journal of Pathology: Clinical Research, 2016, 2, 138-153.
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121 Evidence of a genetic link between endometriosis and ovarian cancer. Fertility and Sterility, 2016, 105, 35-43.e10.
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| 146 | Evaluating the ovarian cancer gonadotropin hypothesis: A candidate gene study. Gynecologic Oncology, 2015, 136, 542-548. | 0.6 | 15 |
| 147 | Association of Type and Location of <i>BRCA1</i> and<i>BRCA2</i>Mutations With Risk of Breast and Ovarian Cancer. JAMA - Journal of the American Medical Association, 2015, 313, 1347. | 3.8 | 390 |
| 148 | Assessment of variation in immunosuppressive pathway genes reveals TGFBR2 to be associated with prognosis of estrogen receptor-negative breast cancer after chemotherapy. Breast Cancer Research, 2015, 17, 18. | 2.2 | 20 |
| 149 | Cis-eQTL analysis and functional validation of candidate susceptibility genes for high-grade serous ovarian cancer. Nature Communications, 2015, 6, 8234. | 5.8 | 63 |
| 150 | Common variants at the<i>CHEK2</i>gene locus and risk of epithelial ovarian cancer. Carcinogenesis, 2015, 36, 1341-1353. | 1.3 | 24 |
| 151 | An evaluation of the prognostic model PREDICT using the POSH cohort of women aged â@ $1 / 240$ years at breast cancer diagnosis. British Journal of Cancer, 2015, 112, 983-991. | 2.9 | 27 |
| 152 | Shared genetics underlying epidemiological association between endometriosis and ovarian cancer. Human Molecular Genetics, 2015, 24, 5955-5964. | 1.4 | 68 |
| 153 | Obesity, Aspirin, and Risk of Colorectal Cancer in Carriers of Hereditary Colorectal Cancer: A Prospective Investigation in the CAPP2 Study. Journal of Clinical Oncology, 2015, 33, 3591-3597. | 0.8 | 91 |
| 154 | Obesity and the outcome of young breast cancer patients in the UK: the POSH study. Annals of Oncology, 2015, 26, 101-112. | 0.6 | 72 |
| 155 | Effect of BRCA Mutations on Metastatic Relapse and Cause-specific Survival After Radical Treatment for Localised Prostate Cancer. European Urology, 2015, 68, 186-193. | 0.9 | 279 |
| 156 | Candidate Genetic Modifiers for Breast and Ovarian Cancer Risk in <i>BRCA1</i> and <i>BRCA2</i> Mutation Carriers. Cancer Epidemiology Biomarkers and Prevention, 2015, 24, 308-316. | 1.1 | 22 |
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171 Cancer-Affected Status. Cancer Epidemiology Biomarkers and Prevention, 2014, 23, 1018-1024.

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$175 \quad$ Targeted Prostate Cancer Screening in BRCA1 and BRCA2 Mutation Carriers: R95Genome-wide association study identifies 25 known breast cancer susceptibility loci as risk factors1.3145for triple-negative breast cancer. Carcinogenesis, 2014, 35, 1012-1019.Consortium analysis of gene and geneâ€"folate interactions in purine and pyrimidine metabolism1.516pathways with ovarian carcinoma risk. Molecular Nutrition and Food Research, 2014, 58, 2023-2035.Genetic variation in mitotic regulatory pathway genes is associated with breast tumor grade. Human

| 181 | CWAS meta-analysis and replication identifies three new susceptibility loci for ovarian cancer. Nature Genetics, 2013, 45, 362-370. | 9.4 | 326 |
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| 182 | Loss-of-function mutations in SMARCE1 cause an inherited disorder of multiple spinal meningiomas. Nature Genetics, 2013, 45, 295-298. | 9.4 | 208 |
| 183 | Germline <i>BRCA</i> Mutations Are Associated With Higher Risk of Nodal Involvement, Distant Metastasis, and Poor Survival Outcomes in Prostate Cancer. Journal of Clinical Oncology, 2013, 31, 1748-1757. | 0.8 | 641 |
| 184 | Cancer Risks for BRCA1 and BRCA2 Mutation Carriers: Results From Prospective Analysis of EMBRACE. Journal of the National Cancer Institute, 2013, 105, 812-822. | 3.0 | 753 |
| 185 | Machine learning approaches for the discovery of gene-gene interactions in disease data. Briefings in Bioinformatics, 2013, 14, 251-260. | 3.2 | 81 |
| 186 | Multiple independent variants at the TERT locus are associated with telomere length and risks of breast and ovarian cancer. Nature Genetics, 2013, 45, 371-384. | 9.4 | 493 |
| 187 | Critical research gaps and translational priorities for the successful prevention and treatment of breast cancer. Breast Cancer Research, 2013, 15, R92. | 2.2 | 320 |
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| 192 | Identification of Inherited Genetic Variations Influencing Prognosis in Early-Onset Breast Cancer. Cancer Research, 2013, 73, 1883-1891. | 0.4 | 42 |
| 193 | Prospective Observational Study of Breast Cancer Treatment Outcomes for UK Women Aged 18ấ"40 Years at Diagnosis: The POSH Study. Journal of the National Cancer Institute, 2013, 105, 978-988. | 3.0 | 156 |199 Support Vector Machine Classifier for Estrogen Receptor Positive and Negative Early-Onset Breast

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A Nonsynonymous Polymorphism in <i>IRS1<|i〉 Modifies Risk of Developing Breast and Ovarian

| 204 | Cancers in <i>BRCA1<\|i> and Ovarian Cancer in <i>BRCA2</i> Mutation Carriers. Cancer Epidemiology | 1.1 |
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210 Cancer and Ovarian Cancer Risk in <i>BRCA1<|i> and <i>BRCA2</i〉Mutation Carriers. Cancer
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Pathology of Breast and Ovarian Cancers among <i>BRCA1<|i> and <i>BRCA2 < $|i\rangle$ Mutation Carriers:
Results from the Consortium of Investigators of Modifiers of $\langle i\rangle$ BRCA1 $\langle\mid i\rangle|<i\rangle 2<|i\rangle$ (CIMBA). Cancer
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Long-term effect of resistant starch on cancer risk in carriers of hereditary colorectal cancer: an
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Treatment, 2012, 134, 543-547.
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