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The role of targeted BRCA1/BRCA2 mutation analysis in hereditary breast/ovarian cancer families of Portuguese ancestry

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#	Paper	IF	Citations
22	Development and analytical validation of a 25-gene next generation sequencing panel that includes the BRCA1 and BRCA2 genes to assess hereditary cancer risk. <i>BMC Cancer</i> , 2015 , 15, 215	4.8	79
21	Prevalence of BRCA1/BRCA2 mutations in a Brazilian population sample at-risk for hereditary breast cancer and characterization of its genetic ancestry. <i>Oncotarget</i> , 2016 , 7, 80465-80481	3.3	39
20	Analysis of Founder Mutations in Rare Tumors Associated With Hereditary Breast/Ovarian Cancer Reveals a Novel Association of BRCA2 Mutations with Ampulla of Vater Carcinomas. <i>PLoS ONE</i> , 2016 , 11, e0161438	3.7	10
19	Implementation of next-generation sequencing for molecular diagnosis of hereditary breast and ovarian cancer highlights its genetic heterogeneity. <i>Breast Cancer Research and Treatment</i> , 2016 , 159, 245-56	4.4	20
18	The role of germline mutations in the BRCA1/2 and mismatch repair genes in men ascertained for early-onset and/or familial prostate cancer. <i>Familial Cancer</i> , 2016 , 15, 111-21	3	18
17	Validation of a Next-Generation Sequencing Pipeline for the Molecular Diagnosis of Multiple Inherited Cancer Predisposing Syndromes. <i>Journal of Molecular Diagnostics</i> , 2017 , 19, 502-513	5.1	7
16	Impact of germline and somatic BRCA1/2 mutations: tumor spectrum and detection platforms. <i>Gene Therapy</i> , 2017 , 24, 601-609	4	3
15	Identification of pathogenic retrotransposon insertions in cancer predisposition genes. <i>Cancer Genetics</i> , 2017 , 216-217, 159-169	2.3	19
14	First-degree family history of breast cancer is associated with prostate cancer risk: a systematic review and meta-analysis. <i>BMC Cancer</i> , 2019 , 19, 871	4.8	8
13	Non- Variants Detected in a High-Risk Chilean Cohort With a History of Breast and/or Ovarian Cancer. <i>Journal of Global Oncology</i> , 2019 , 5, 1-14	2.6	1
12	BRCA gene mutations: A population based review. <i>Gene Reports</i> , 2019 , 15, 100380	1.4	
11	The nonsense mutation MSH2 c.2152C>T shows a founder effect in Portuguese Lynch syndrome families. <i>Genes Chromosomes and Cancer</i> , 2019 , 58, 657-664	5	1
10	Tumor Testing for Somatic and Germline / Variants in Ovarian Cancer Patients in the Context of Strong Founder Effects. <i>Frontiers in Oncology</i> , 2020 , 10, 1318	5.3	3
9	Early-onset breast cancer in a woman with a germline mobile element insertion resulting in disruption: a case report. <i>Human Genome Variation</i> , 2020 , 7, 24	1.8	2
8	The Variant C.349A>G Is Associated with Prostate Cancer Risk and Carriers Share a Common Ancestor. <i>Cancers</i> , 2020 , 12,	6.6	4
7	Generation and characterization of induced pluripotent stem cells heterozygous for the Portuguese BRCA2 founder mutation. <i>Stem Cell Research</i> , 2021 , 53, 102364	1.6	1
6	Hereditary breast cancer and ancestry in the Madeira archipelago: an exploratory study. <i>Ecancermedicalscience</i> , 2021 , 15, 1261	2.7	O

CITATION REPORT

5	Development and validation of a 36-gene sequencing assay for hereditary cancer risk assessment. <i>PeerJ</i> , 2017 , 5, e3046	3.1	15
4	Development and validation of a 36-gene sequencing assay for hereditary cancer risk assessment.		
3	Expression Profiling in Ovarian Cancer Reveals Coordinated Regulation of and Homologous Recombination Genes <i>Biomedicines</i> , 2022 , 10,	4.8	O
2	Mutation Patterns in Portuguese Families with Hereditary Breast and Ovarian Cancer Syndrome. 2022 , 14, 4717		O
1	Exome sequencing of affected duos and trios uncovers PRUNE2 as a novel prostate cancer predisposition gene.		2