Eva Machackova

List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	Risks of Breast, Ovarian, and Contralateral Breast Cancer for <i>BRCA1</i> and <i>BRCA2</i> Mutation Carriers. JAMA - Journal of the American Medical Association, 2017, 317, 2402.	7.4	1,898
2	Mutational spectrum in a worldwide study of 29,700 families with <i>BRCA1</i> or <i>BRCA2</i> mutations. Human Mutation, 2018, 39, 593-620.	2.5	224
3	Male breast cancer in BRCA1 and BRCA2 mutation carriers: pathology data from the Consortium of Investigators of Modifiers of BRCA1/2. Breast Cancer Research, 2016, 18, 15.	5.0	88
4	Differentiating pathogenic mutations from polymorphic alterations in the splice sites of BRCA1 and BRCA2. Genes Chromosomes and Cancer, 2003, 37, 314-320.	2.8	78
5	<i>BRCA2</i> Hypomorphic Missense Variants Confer Moderate Risks of Breast Cancer. Cancer Research, 2017, 77, 2789-2799.	0.9	75
6	Spectrum and characterisation of BRCA1 and BRCA2deleterious mutations in high-risk Czech patients with breast and/or ovarian cancer. BMC Cancer, 2008, 8, 140.	2.6	64
7	Identification of deleterious germline <i>CHEK2</i> mutations and their association with breast and ovarian cancer. International Journal of Cancer, 2019, 145, 1782-1797.	5.1	62
8	Characterization of the Cancer Spectrum in Men With Germline <i>BRCA1</i> and <i>BRCA2</i> Pathogenic Variants. JAMA Oncology, 2020, 6, 1218.	7.1	48
9	High occurrence of BRCA1 intragenic rearrangements in hereditary breast and ovarian cancer syndrome in the Czech Republic. BMC Medical Genetics, 2007, 8, 32.	2.1	45
10	BRCA1 and BRCA2 mutations in women with familial or early-onset breast/ovarian cancer in the Czech Republic. Human Mutation, 2004, 23, 397-398.	2.5	36
11	BRCA1 Circos: a visualisation resource for functional analysis of missense variants. Journal of Medical Genetics, 2015, 52, 224-230.	3.2	32
12	Validation of CZECANCA (CZEch CAncer paNel for Clinical Application) for targeted NGS-based analysis of hereditary cancer syndromes. PLoS ONE, 2018, 13, e0195761.	2.5	31
13	Polygenic risk modeling for prediction of epithelial ovarian cancer risk. European Journal of Human Genetics, 2022, 30, 349-362.	2.8	23
14	Mutation analysis of the BRCA1 and BRCA2 genes results in the identification of novel and recurrent mutations in 6/16 Flemish families with breast and/or ovarian cancer but not in 12 sporadic patients with early-onset disease. Human Mutation, 1999, 13, 256-256.	2.5	22
15	Identification and Functional Testing of ERCC2 Mutations in a Multi-national Cohort of Patients with Familial Breast- and Ovarian Cancer. PLoS Genetics, 2016, 12, e1006248.	3.5	22
16	GAPPS – Gastric Adenocarcinoma and Proximal Polyposis of the Stomach Syndrome in 8 Families Tested at Masaryk Memorial Cancer Institute – Prevention and Prophylactic Gastrectomies. Klinicka Onkologie, 2019, 32, 109-117.	0.3	20
17	Multigene Panel Germline Testing of 1333 Czech Patients with Ovarian Cancer. Cancers, 2020, 12, 956.	3.7	19
18	Fanconi anemia with biallelic FANCD1/BRCA2 mutations – Case report of a family with three affected children. European Journal of Medical Genetics, 2016, 59, 152-157.	1.3	11

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#	ARTICLE	IF	CITATIONS
19	Identification of Germline Mutations in Melanoma Patients with Early Onset, Double Primary Tumors, or Family Cancer History by NGS Analysis of 217 Genes. Biomedicines, 2020, 8, 404.	3.2	10
20	Recommendations for Preventive Care for Women with Rare Genetic Cause of Breast and Ovarian Cancer. Klinicka Onkologie, 2019, 32, 6-13.	0.3	8
21	Novel germline BRCA1 and BRCA2 mutations in breast and breast/ovarian cancer families from the Czech Republic. Human Mutation, 2001, 18, 545-545.	2.5	7
22	BAP1 Syndrome – Predisposition to Malignant Mesothelioma, Skin and Uveal Melanoma, Renal and Other Cancers. Klinicka Onkologie, 2019, 32, 118-122.	0.3	7
23	Genetic and Preventive Services for Hereditary Breast and Ovarian Cancer in the Czech Republic. Hereditary Cancer in Clinical Practice, 2006, 4, 3.	1.5	6
24	The AIB1 gene polyglutamine repeat length polymorphism and the risk of breast cancer development. Journal of Cancer Research and Clinical Oncology, 2011, 137, 331-338.	2.5	5
25	Thorough in silico and in vitro cDNA analysis of 21 putativeBRCA1andBRCA2splice variants and a complex tandem duplication inBRCA2allowing the identification of activated cryptic splice donor sites inBRCA2exon 11. Human Mutation, 2018, 39, 515-526.	2.5	5
26	Twenty Years of BRCA1 and BRCA2 Molecular Analysis at MMCI – Current Developments for the Classifi cation of Variants. Klinicka Onkologie, 2019, 32, 51-71.	0.3	5
27	Germline CHEK2 Gene Mutations in Hereditary Breast Cancer Predisposition – Mutation Types and their Biological and Clinical Relevance. Klinicka Onkologie, 2019, 32, 36-50.	0.3	2
28	Functional evaluation of variants of unknown significance in the <i>BRCA2</i> gene identified in genetic testing. Cancer Biology and Therapy, 2019, 20, 633-641.	3.4	1
29	Re: ERCC3, a new ovarian cancer susceptibility gene?. European Journal of Cancer, 2021, 150, 278-280.	2.8	1
30	Contribution of Massive Parallel Sequencing to Diagnosis of Hereditary Ovarian Cancer in the Czech Republic. Klinicka Onkologie, 2019, 32, 72-78.	0.3	1