

# Jacopo Azzollini

## List of Publications by Year in descending order

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Version: 2024-02-01

53  
papers

2,748  
citations

279701

23  
h-index

206029

48  
g-index

57  
all docs

57  
docs citations

57  
times ranked

5833  
citing authors

#	ARTICLE	IF	CITATIONS
1	Breast and Prostate Cancer Risks for Male <i>BRCA1</i> and <i>BRCA2</i> Pathogenic Variant Carriers Using Polygenic Risk Scores. <i>Journal of the National Cancer Institute</i> , 2022, 114, 109-122.	3.0	19
2	Management of BRCA Tumour Testing in an Integrated Molecular Tumour Board Multidisciplinary Model. <i>Frontiers in Oncology</i> , 2022, 12, 857515.	1.3	1
3	A case-only study to identify genetic modifiers of breast cancer risk for <i>BRCA1/BRCA2</i> mutation carriers. <i>Nature Communications</i> , 2021, 12, 1078.	5.8	19
4	Correlation between oncological family history and clinical outcome in a large monocentric cohort of pediatric patients with rhabdomyosarcoma. <i>International Journal of Clinical Oncology</i> , 2021, 26, 1561-1568.	1.0	0
5	Analysis of Italian <i>BRCA1/2</i> Pathogenic Variants Identifies a Private Spectrum in the Population from the Bergamo Province in Northern Italy. <i>Cancers</i> , 2021, 13, 532.	1.7	8
6	Clinical heterogeneity and reduced penetrance in <i>DICER1</i> syndrome: a report of three families. <i>Tumori</i> , 2021, 107, NP144-NP148.	0.6	2
7	Association of Genomic Domains in <i>BRCA1</i> and <i>BRCA2</i> with Prostate Cancer Risk and Aggressiveness. <i>Cancer Research</i> , 2020, 80, 624-638.	0.4	39
8	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. <i>Nature Genetics</i> , 2020, 52, 56-73.	9.4	120
9	Polygenic risk scores and breast and epithelial ovarian cancer risks for carriers of <i>BRCA1</i> and <i>BRCA2</i> pathogenic variants. <i>Genetics in Medicine</i> , 2020, 22, 1653-1666.	1.1	82
10	Pre- and Post-Zygotic <i>TP53</i> De Novo Mutations in <i>SHH</i> -Medulloblastoma. <i>Cancers</i> , 2020, 12, 2503.	1.7	1
11	Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses. <i>Nature Genetics</i> , 2020, 52, 572-581.	9.4	265
12	Transcriptome-wide association study of breast cancer risk by estrogen receptor status. <i>Genetic Epidemiology</i> , 2020, 44, 442-468.	0.6	32
13	The Spectrum of <i>FANCM</i> Protein Truncating Variants in European Breast Cancer Cases. <i>Cancers</i> , 2020, 12, 292.	1.7	11
14	Analysis of <i>BRCA1</i> and <i>RAD51C</i> Promoter Methylation in Italian Families at High-Risk of Breast and Ovarian Cancer. <i>Cancers</i> , 2020, 12, 910.	1.7	13
15	Hereditary : <i>BRCA</i> and Other. , 2020, , 23-41.		0
16	Risk-reducing surgery in <i>BRCA1/BRCA2</i> mutation carriers: Are there factors associated with the choice?. <i>Psycho-Oncology</i> , 2019, 28, 1871-1878.	1.0	9
17	The <i>FANCM:p.Arg658*</i> truncating variant is associated with risk of triple-negative breast cancer. <i>Npj Breast Cancer</i> , 2019, 5, 38.	2.3	28
18	Large scale multifactorial likelihood quantitative analysis of <i>BRCA1</i> and <i>BRCA2</i> variants: An ENIGMA resource to support clinical variant classification. <i>Human Mutation</i> , 2019, 40, 1557-1578.	1.1	102

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19	Mendelian randomisation study of height and body mass index as modifiers of ovarian cancer risk in 22,588 BRCA1 and BRCA2 mutation carriers. <i>British Journal of Cancer</i> , 2019, 121, 180-192.	2.9	19
20	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. <i>Nature Communications</i> , 2019, 10, 1741.	5.8	90
21	Co-occurrence of Mayer-Rokitansky-Küster-Hauser syndrome and ovarian cancer: A case report and review of the literature. <i>Gynecologic Oncology Reports</i> , 2019, 28, 68-70.	0.3	3
22	Insight into genetic susceptibility to male breast cancer by multigene panel testing: Results from a multicenter study in Italy. <i>International Journal of Cancer</i> , 2019, 145, 390-400.	2.3	40
23	Constitutive BRCA1 Promoter Hypermethylation Can Be a Predisposing Event in Isolated Early-Onset Breast Cancer. <i>Cancers</i> , 2019, 11, 58.	1.7	22
24	Height and Body Mass Index as Modifiers of Breast Cancer Risk in BRCA1/2 Mutation Carriers: A Mendelian Randomization Study. <i>Journal of the National Cancer Institute</i> , 2019, 111, 350-364.	3.0	30
25	Evaluation of CYP17A1 and CYP1B1 polymorphisms in male breast cancer risk. <i>Endocrine Connections</i> , 2019, 8, 1224-1229.	0.8	6
26	Mutational spectrum in a worldwide study of 29,700 families with BRCA1 or BRCA2 mutations. <i>Human Mutation</i> , 2018, 39, 593-620.	1.1	224
27	Survey of gynecological carcinosarcomas in families with breast and ovarian cancer predisposition. <i>Cancer Genetics</i> , 2018, 221, 38-45.	0.2	4
28	Individuals with FANCM biallelic mutations do not develop Fanconi anemia, but show risk for breast cancer, chemotherapy toxicity and may display chromosome fragility. <i>Genetics in Medicine</i> , 2018, 20, 452-457.	1.1	59
29	Increased access to TP53 analysis through breast cancer multi-gene panels: clinical considerations. <i>Familial Cancer</i> , 2018, 17, 317-319.	0.9	1
30	Contribution of MUTYH Variants to Male Breast Cancer Risk: Results From a Multicenter Study in Italy. <i>Frontiers in Oncology</i> , 2018, 8, 583.	1.3	25
31	Two Missense Variants Detected in Breast Cancer Probands Preventing BRCA2-PALB2 Protein Interaction. <i>Frontiers in Oncology</i> , 2018, 8, 480.	1.3	11
32	A Dietary Intervention to Lower Serum Levels of IGF-I in BRCA Mutation Carriers. <i>Cancers</i> , 2018, 10, 309.	1.7	18
33	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. <i>Nature Genetics</i> , 2017, 49, 680-691.	9.4	356
34	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. <i>Nature Genetics</i> , 2017, 49, 1767-1778.	9.4	289
35	Association of breast cancer risk in BRCA1 and BRCA2 mutation carriers with genetic variants showing differential allelic expression: identification of a modifier of breast cancer risk at locus 11q22.3. <i>Breast Cancer Research and Treatment</i> , 2017, 161, 117-134.	1.1	18
36	Whole-exome sequencing and targeted gene sequencing provide insights into the role of PALB2 as a male breast cancer susceptibility gene. <i>Cancer</i> , 2017, 123, 210-218.	2.0	31

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37	Evaluation of Polygenic Risk Scores for Breast and Ovarian Cancer Risk Prediction in BRCA1 and BRCA2 Mutation Carriers. <i>Journal of the National Cancer Institute</i> , 2017, 109, .	3.0	242
38	Prediction of Breast and Prostate Cancer Risks in Male <i>BRCA1</i> and <i>BRCA2</i> Mutation Carriers Using Polygenic Risk Scores. <i>Journal of Clinical Oncology</i> , 2017, 35, 2240-2250.	0.8	152
39	Revertant mosaicism for family mutations is not observed in BRCA1/2 phenocopies. <i>PLoS ONE</i> , 2017, 12, e0171663.	1.1	7
40	A Targeted Approach to Genetic Counseling in Breast Cancer Patients: The Experience of an Italian Local Project. <i>Tumori</i> , 2016, 102, 45-50.	0.6	4
41	Expanding the clinical spectrum of the <i>HDAC8</i> phenotype™ implications for molecular diagnostics, counseling and risk prediction. <i>Clinical Genetics</i> , 2016, 89, 564-573.	1.0	38
42	Mutation detection rates associated with specific selection criteria for BRCA1/2 testing in 1854 high-risk families: A monocentric Italian study. <i>European Journal of Internal Medicine</i> , 2016, 32, 65-71.	1.0	21
43	Broadening of cohesinopathies: exome sequencing identifies mutations in <i>ANKRD11</i> in two patients with Cornelia de Lange overlapping phenotype. <i>Clinical Genetics</i> , 2016, 89, 74-81.	1.0	69
44	Estimate of the penetrance of BRCA mutation and the COS software for the assessment of BRCA mutation probability. <i>Familial Cancer</i> , 2015, 14, 117-128.	0.9	12
45	Overall and allele-specific expression of the <i>SMC1A</i> gene in female Cornelia de Lange syndrome patients and healthy controls. <i>Epigenetics</i> , 2014, 9, 973-979.	1.3	10
46	Functional characterisation of a novel mutation affecting the catalytic domain of MMP2 in siblings with multicentric osteolysis, nodulosis and arthropathy. <i>Journal of Human Genetics</i> , 2014, 59, 631-637.	1.1	11
47	Common non-synonymous SNPs associated with breast cancer susceptibility: findings from the Breast Cancer Association Consortium. <i>Human Molecular Genetics</i> , 2014, 23, 6096-6111.	1.4	53
48	Genomic imbalances in patients with a clinical presentation in the spectrum of Cornelia de Lange syndrome. <i>BMC Medical Genetics</i> , 2013, 14, 41.	2.1	15
49	Cornelia de Lange individuals with new and recurrent <i>SMC1A</i> mutations enhance delineation of mutation repertoire and phenotypic spectrum. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 2909-2919.	0.7	31
50	Molecular characterization of a mosaic NIPBL deletion in a Cornelia de Lange patient with severe phenotype. <i>European Journal of Medical Genetics</i> , 2013, 56, 138-143.	0.7	24
51	Intragenic and large NIPBL rearrangements revealed by MLPA in Cornelia de Lange patients. <i>European Journal of Human Genetics</i> , 2012, 20, 734-741.	1.4	23
52	Novel and recurrent spastin mutations in a large series of SPG4 Italian families. <i>Neuroscience Letters</i> , 2012, 528, 42-45.	1.0	5
53	Somatic mosaicism in Cornelia de Lange syndrome: a further contributor to the wide clinical expressivity?. <i>Clinical Genetics</i> , 2010, 78, 560-564.	1.0	30