Jacopo Azzollini

List of Publications by Year
in descending order

53	2,748	279487	205818
papers	citations	23 h-index	48 g-index

57	57	57	5833
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. Nature Genetics, 2017, 49, 680-691.	9.4	356
2	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. Nature Genetics, 2017, 49, 1767-1778.	9.4	289
3	Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses. Nature Genetics, 2020, 52, 572-581.	9.4	265
4	Evaluation of Polygenic Risk Scores for Breast and Ovarian Cancer Risk Prediction in BRCA1 and BRCA2 Mutation Carriers. Journal of the National Cancer Institute, 2017, 109, .	3.0	242
5	Mutational spectrum in a worldwide study of 29,700 families withBRCA1orBRCA2mutations. Human Mutation, 2018, 39, 593-620.	1.1	224
6	Prediction of Breast and Prostate Cancer Risks in Male BRCA1 and BRCA2 Mutation Carriers Using Polygenic Risk Scores. Journal of Clinical Oncology, 2017, 35, 2240-2250.	0.8	152
7	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. Nature Genetics, 2020, 52, 56-73.	9.4	120
8	Large scale multifactorial likelihood quantitative analysis of BRCA1 and BRCA2 variants: An ENIGMA resource to support clinical variant classification. Human Mutation, 2019, 40, 1557-1578.	1.1	102
9	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. Nature Communications, 2019, 10, 1741.	5.8	90
10	Polygenic risk scores and breast and epithelial ovarian cancer risks for carriers of BRCA1 and BRCA2 pathogenic variants. Genetics in Medicine, 2020, 22, 1653-1666.	1.1	82
11	Broadening of cohesinopathies: exome sequencing identifies mutations in <scp>ANKRD11</scp> in two patients with Cornelia de Langeâ€overlapping phenotype. Clinical Genetics, 2016, 89, 74-81.	1.0	69
12	Individuals with FANCM biallelic mutations do not develop Fanconi anemia, but show risk for breast cancer, chemotherapy toxicity and may display chromosome fragility. Genetics in Medicine, 2018, 20, 452-457.	1.1	59
13	Common non-synonymous SNPs associated with breast cancer susceptibility: findings from the Breast Cancer Association Consortium. Human Molecular Genetics, 2014, 23, 6096-6111.	1.4	53
14	Insight into genetic susceptibility to male breast cancer by multigene panel testing: Results from a multicenter study in Italy. International Journal of Cancer, 2019, 145, 390-400.	2.3	40
15	Association of Genomic Domains in BRCA1 and BRCA2 with Prostate Cancer Risk and Aggressiveness. Cancer Research, 2020, 80, 624-638.	0.4	39
16	Expanding the clinical spectrum of the â€˜<scp>HDAC8</scp>â€phenotypeâ€™ â€“ implications for molecular diagnostics, counseling and risk prediction. Clinical Genetics, 2016, 89, 564-573.	1.0	38
17	Transcriptomeâ€wide association study of breast cancer risk by estrogenâ€receptor status. Genetic Epidemiology, 2020, 44, 442-468.	0.6	32
18	Cornelia de Lange individuals with new and recurrent SMC1A mutations enhance delineation of mutation repertoire and phenotypic spectrum. American Journal of Medical Genetics, Part A, 2013, 161, 2909-2919.	0.7	31

#	Article	IF	Citations
19	Wholeâ€exome sequencing and targeted gene sequencing provide insights into the role of PALB2 as a male breast cancer susceptibility gene. Cancer, 2017, 123, 210-218.	2.0	31
20	Somatic mosaicism in Cornelia de Lange syndrome: a further contributor to the wide clinical expressivity?. Clinical Genetics, 2010, 78, 560-564.	1.0	30
21	Height and Body Mass Index as Modifiers of Breast Cancer Risk in BRCA1/2 Mutation Carriers: A Mendelian Randomization Study. Journal of the National Cancer Institute, 2019, 111, 350-364.	3.0	30
22	The FANCM:p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. Npj Breast Cancer, 2019, 5, 38.	2.3	28
23	Contribution of MUTYH Variants to Male Breast Cancer Risk: Results From a Multicenter Study in Italy. Frontiers in Oncology, 2018, 8, 583.	1.3	25
24	Molecular characterization of a mosaic NIPBL deletion in a Cornelia de Lange patient with severe phenotype. European Journal of Medical Genetics, 2013, 56, 138-143.	0.7	24
25	Intragenic and large NIPBL rearrangements revealed by MLPA in Cornelia de Lange patients. European Journal of Human Genetics, 2012, 20, 734-741.	1.4	23
26	Constitutive BRCA1 Promoter Hypermethylation Can Be a Predisposing Event in Isolated Early-Onset Breast Cancer. Cancers, 2019, 11, 58.	1.7	22
27	Mutation detection rates associated with specific selection criteria for BRCA1/2 testing in 1854 high-risk families: A monocentric Italian study. European Journal of Internal Medicine, 2016, 32, 65-71.	1.0	21
28	Mendelian randomisation study of height and body mass index as modifiers of ovarian cancer risk in 22,588 BRCA1 and BRCA2 mutation carriers. British Journal of Cancer, 2019, 121, 180-192.	2.9	19
29	A case-only study to identify genetic modifiers of breast cancer risk for BRCA1/BRCA2 mutation carriers. Nature Communications, 2021, 12, 1078.	5.8	19
30	Breast and Prostate Cancer Risks for MaleBRCA1andBRCA2Pathogenic Variant Carriers Using Polygenic Risk Scores. Journal of the National Cancer Institute, 2022, 114, 109-122.	3.0	19
31	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. Breast Cancer Research and Treatment, 2017, 161, 117-134.	1.1	18
32	A Dietary Intervention to Lower Serum Levels of IGF-I in BRCA Mutation Carriers. Cancers, 2018, 10, 309.	1.7	18
33	Genomic imbalances in patients with a clinical presentation in the spectrum of Cornelia de Lange syndrome. BMC Medical Genetics, 2013, 14, 41.	2.1	15
34	Analysis of BRCA1 and RAD51C Promoter Methylation in Italian Families at High-Risk of Breast and Ovarian Cancer. Cancers, 2020, 12, 910.	1.7	13
35	Estimate of the penetrance of BRCA mutation and the COS software for the assessment of BRCA mutation probability. Familial Cancer, 2015, 14, 117-128.	0.9	12
36	Functional characterisation of a novel mutation affecting the catalytic domain of MMP2 in siblings with multicentric osteolysis, nodulosis and arthropathy. Journal of Human Genetics, 2014, 59, 631-637.	1.1	11

#	Article	IF	Citations
37	Two Missense Variants Detected in Breast Cancer Probands Preventing BRCA2-PALB2 Protein Interaction. Frontiers in Oncology, 2018, 8, 480.	1.3	11
38	The Spectrum of FANCM Protein Truncating Variants in European Breast Cancer Cases. Cancers, 2020, 12, 292.	1.7	11
39	Overall and allele-specific expression of theSMC1Agene in female Cornelia de Lange syndrome patients and healthy controls. Epigenetics, 2014, 9, 973-979.	1.3	10
40	Riskâ€reducing surgery in BRCA1/BRCA2 mutation carriers: Are there factors associated with the choice?. Psycho-Oncology, 2019, 28, 1871-1878.	1.0	9
41	Analysis of Italian BRCA1/2 Pathogenic Variants Identifies a Private Spectrum in the Population from the Bergamo Province in Northern Italy. Cancers, 2021, 13, 532.	1.7	8
42	Revertant mosaicism for family mutations is not observed in BRCA1/2 phenocopies. PLoS ONE, 2017, 12, e0171663.	1.1	7
43	Evaluation of CYP17A1 and CYP1B1 polymorphisms in male breast cancer risk. Endocrine Connections, 2019, 8, 1224-1229.	0.8	6
44	Novel and recurrent spastin mutations in a large series of SPG4 Italian families. Neuroscience Letters, 2012, 528, 42-45.	1.0	5
45	A Targeted Approach to Genetic Counseling in Breast Cancer Patients: The Experience of an Italian Local Project. Tumori, 2016, 102, 45-50.	0.6	4
46	Survey of gynecological carcinosarcomas in families with breast and ovarian cancer predisposition. Cancer Genetics, 2018, 221, 38-45.	0.2	4
47	Co-occurrence of Mayer-Rokitansky-KÃ¼ster-Hauser syndrome and ovarian cancer: A case report and review of the literature. Gynecologic Oncology Reports, 2019, 28, 68-70.	0.3	3
48	Clinical heterogeneity and reduced penetrance in DICER1 syndrome: a report of three families. Tumori, 2021, 107, NP144-NP148.	0.6	2
49	Increased access to TP53 analysis through breast cancer multi-gene panels: clinical considerations. Familial Cancer, 2018, 17, 317-319.	0.9	1
50	Pre- and Post-Zygotic TP53 De Novo Mutations in SHH-Medulloblastoma. Cancers, 2020, 12, 2503.	1.7	1
51	Management of BRCA Tumour Testing in an Integrated Molecular Tumour Board Multidisciplinary Model. Frontiers in Oncology, 2022, 12, 857515.	1.3	1
52	Correlation between oncological family history and clinical outcome in a large monocentric cohort of pediatric patients with rhabdomyosarcoma. International Journal of Clinical Oncology, 2021, 26, 1561-1568.	1.0	0
53	Hereditary : BRCA and Other. , 2020, , 23-41.		0