# Jacopo Azzollini

List of Publications by Year
in descending order



| 53 | 2,748 | 279798 23 | 206112 48 |
|---|---|---|---|
| papers | citations | h-index | g-index |

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

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Breast and Prostate Cancer Risks for MaleBRCA1andBRCA2Pathogenic Variant Carriers Using Polygenic Risk Scores. Journal of the National Cancer Institute, 2022, 114, 109-122. | 6.3 | 19 |
| 2 | Management of BRCA Tumour Testing in an Integrated Molecular Tumour Board Multidisciplinary Model. Frontiers in Oncology, 2022, 12, 857515. | 2.8 | 1 |
| 3 | A case-only study to identify genetic modifiers of breast cancer risk for BRCA1/BRCA2 mutation carriers. Nature Communications, 2021, 12, 1078. | 12.8 | 19 |
| 4 | 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. | 2.2 | 0 |
| 5 | 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. | 3.7 | 8 |
| 6 | Clinical heterogeneity and reduced penetrance in DICER1 syndrome: a report of three families. Tumori, 2021, 107, NP144-NP148. | 1.1 | 2 |
| 7 | Association of Genomic Domains in BRCA1 and BRCA2 with Prostate Cancer Risk and Aggressiveness. Cancer Research, 2020, 80, 624-638. | 0.9 | 39 |
| 8 | Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. Nature Genetics, 2020, 52, 56-73. | 21.4 | 120 |
| 9 | 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. | 2.4 | 82 |
| 10 | Pre- and Post-Zygotic TP53 De Novo Mutations in SHH-Medulloblastoma. Cancers, 2020, 12, 2503. | 3.7 | 1 |
| 11 | Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses. Nature Genetics, 2020, 52, 572-581. | 21.4 | 265 |
| 12 | Transcriptomeâ€wide association study of breast cancer risk by estrogenâ€receptor status. Genetic Epidemiology, 2020, 44, 442-468. | 1.3 | 32 |
| 13 | The Spectrum of FANCM Protein Truncating Variants in European Breast Cancer Cases. Cancers, 2020, 12, 292. | 3.7 | 11 |
| 14 | Analysis of BRCA1 and RAD51C Promoter Methylation in Italian Families at High-Risk of Breast and Ovarian Cancer. Cancers, 2020, 12, 910. | 3.7 | 13 |
| 15 | Hereditary : BRCA and Other. , 2020, , 23-41. | | 0 |
| 16 | Riskâ€reducing surgery in BRCA1/BRCA2 mutation carriers: Are there factors associated with the choice?. Psycho-Oncology, 2019, 28, 1871-1878. | 2.3 | 9 |
| 17 | The FANCM:p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. Npj Breast Cancer, 2019, 5, 38. | 5.2 | 28 |
| 18 | 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. | 2.5 | 102 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | 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. | 6.4 | 19 |
| 20 | Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. Nature Communications, 2019, 10, 1741. | 12.8 | 90 |
| 21 | 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. | 0.6 | 3 |
| 22 | 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. | 5.1 | 40 |
| 23 | Constitutive BRCA1 Promoter Hypermethylation Can Be a Predisposing Event in Isolated Early-Onset Breast Cancer. Cancers, 2019, 11, 58. | 3.7 | 22 |
| 24 | 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. | 6.3 | 30 |
| 25 | Evaluation of CYP17A1 and CYP1B1 polymorphisms in male breast cancer risk. Endocrine Connections, 2019, 8, 1224-1229. | 1.9 | 6 |
| 26 | Mutational spectrum in a worldwide study of 29,700 families withBRCA1orBRCA2mutations. Human Mutation, 2018, 39, 593-620. | 2.5 | 224 |
| 27 | Survey of gynecological carcinosarcomas in families with breast and ovarian cancer predisposition. Cancer Genetics, 2018, 221, 38-45. | 0.4 | 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. Genetics in Medicine, 2018, 20, 452-457. | 2.4 | 59 |
| 29 | Increased access to TP53 analysis through breast cancer multi-gene panels: clinical considerations. Familial Cancer, 2018, 17, 317-319. | 1.9 | 1 |
| 30 | Contribution of MUTYH Variants to Male Breast Cancer Risk: Results From a Multicenter Study in Italy. Frontiers in Oncology, 2018, 8, 583. | 2.8 | 25 |
| 31 | Two Missense Variants Detected in Breast Cancer Probands Preventing BRCA2-PALB2 Protein Interaction. Frontiers in Oncology, 2018, 8, 480. | 2.8 | 11 |
| 32 | A Dietary Intervention to Lower Serum Levels of IGF-I in BRCA Mutation Carriers. Cancers, 2018, 10, 309. | 3.7 | 18 |
| 33 | Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. Nature Genetics, 2017, 49, 680-691. | 21.4 | 356 |
| 34 | Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. Nature Genetics, 2017, 49, 1767-1778. | 21.4 | 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. Breast Cancer Research and Treatment, 2017, 161, 117-134. | 2.5 | 18 |
| 36 | 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. | 4.1 | 31 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | 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, . | 6.3 | 242 |
| 38 | 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. | 1.6 | 152 |
| 39 | Revertant mosaicism for family mutations is not observed in BRCA1/2 phenocopies. PLoS ONE, 2017, 12, e0171663. | 2.5 | 7 |
| 40 | A Targeted Approach to Genetic Counseling in Breast Cancer Patients: The Experience of an Italian Local Project. Tumori, 2016, 102, 45-50. | 1.1 | 4 |
| 41 | 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. | 2.0 | 38 |
| 42 | 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. | 2.2 | 21 |
| 43 | 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. | 2.0 | 69 |
| 44 | Estimate of the penetrance of BRCA mutation and the COS software for the assessment of BRCA mutation probability. Familial Cancer, 2015, 14, 117-128. | 1.9 | 12 |
| 45 | Overall and allele-specific expression of theSMC1Agene in female Cornelia de Lange syndrome patients and healthy controls. Epigenetics, 2014, 9, 973-979. | 2.7 | 10 |
| 46 | 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. | 2.3 | 11 |
| 47 | Common non-synonymous SNPs associated with breast cancer susceptibility: findings from the Breast Cancer Association Consortium. Human Molecular Genetics, 2014, 23, 6096-6111. | 2.9 | 53 |
| 48 | 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 |
| 49 | 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. | 1.2 | 31 |
| 50 | 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. | 1.3 | 24 |
| 51 | Intragenic and large NIPBL rearrangements revealed by MLPA in Cornelia de Lange patients. European Journal of Human Genetics, 2012, 20, 734-741. | 2.8 | 23 |
| 52 | Novel and recurrent spastin mutations in a large series of SPG4 Italian families. Neuroscience Letters, 2012, 528, 42-45. | 2.1 | 5 |
| 53 | Somatic mosaicism in Cornelia de Lange syndrome: a further contributor to the wide clinical expressivity?. Clinical Genetics, 2010, 78, 560-564. | 2.0 | 30 |