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List of Publications by Year
in descending order

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35 papers	2,646 citations	394421 19 h-index	454955 30 g-index
37 all docs	37 docs citations	37 times ranked	3765 citing authors

#	Article	IF	Citations
1	Lifetime Cancer Risks in Individuals with Germline PTEN Mutations. Clinical Cancer Research, 2012, 18, 400-407.	7.0	738
2	A Clinical Scoring System for Selection of Patients for PTEN Mutation Testing Is Proposed on the Basis of a Prospective Study of 3042 Probands. American Journal of Human Genetics, 2011, 88, 42-56.	6.2	332
3	Germline PIK3CA and AKT1 Mutations in Cowden and Cowden-like Syndromes. American Journal of Human Genetics, 2013, 92, 76-80.	6.2	174
4	Germline Epigenetic Regulation of <emph type="ital">KILLIN</emph> in Cowden and Cowden-like Syndrome. JAMA - Journal of the American Medical Association, 2010, 304, 2724.	7.4	138
5	A Novel Germline Mutation in BAP1 Predisposes to Familial Clear-Cell Renal Cell Carcinoma. Molecular Cancer Research, 2013, 11, 1061-1071.	3.4	135
6	ClinGen Variant Curation Expert Panel experiences and standardized processes for disease and geneâ€level specification of the ACMG/AMP guidelines for sequence variant interpretation. Human Mutation, 2018, 39, 1614-1622.	2.5	132
7	Geneâ€specific criteria for PTEN variant curation: Recommendations from the ClinGen PTEN Expert Panel. Human Mutation, 2018, 39, 1581-1592.	2.5	123
8	When Overgrowth Bumps Into Cancer: The PTENâ€Opathies. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2013, 163, 114-121.	1.6	112
9	Analysis of prevalence and degree of macrocephaly in patients with germline PTEN mutations and of brain weight in Pten knock-in murine model. European Journal of Human Genetics, 2011, 19, 763-768.	2.8	93
10	Specifications of the ACMG/AMP variant interpretation guidelines for germline TP53 variants. Human Mutation, 2021, 42, 223-236.	2.5	81
11	Estimate of de novo mutation frequency in probands with PTEN hamartoma tumor syndrome. Genetics in Medicine, 2012, 14, 819-822.	2.4	78
12	Germline Heterozygous Variants in SEC23B Are Associated with Cowden Syndrome and Enriched in Apparently Sporadic Thyroid Cancer. American Journal of Human Genetics, 2015, 97, 661-676.	6.2	76
13	Biochemical screening and PTEN mutation analysis in individuals with autism spectrum disorders and macrocephaly. European Journal of Human Genetics, 2014, 22, 273-276.	2.8	72
14	Papillary Renal Cell Carcinoma Is Associated With PTEN Hamartoma Tumor Syndrome. Urology, 2012, 79, 1187.e1-1187.e7.	1.0	63
15	Germline <scp>PTEN</scp>, <scp>SDHBâ€D</scp>, and <scp>KLLN</scp> alterations in endometrial cancer patients with Cowden and Cowdenâ€like syndromes: An international, multicenter, prospective study. Cancer, 2015, 121, 688-696.	4.1	46
16	PTEN hamartoma tumor syndrome. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2015, 132, 129-137.	1.8	41
17	ClinGen Variant Curation Interface: a variant classification platform for the application of evidence criteria from ACMG/AMP guidelines. Genome Medicine, 2022, 14, 6.	8.2	34
18	Assessment of clinical workload for general and specialty genetic counsellors at an academic medical center: a tool for evaluating genetic counselling practices. Npj Genomic Medicine, 2016, 1, 16010.	3.8	28

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19	PTEN Germline Mutations in Patients Initially Tested for Other Hereditary Cancer Syndromes: Would Use of Risk Assessment Tools Reduce Genetic Testing?. Oncologist, 2013, 18, 1083-1090.	3.7	22
20	KLLN epigenotypeâ€“phenotype associations in Cowden syndrome. European Journal of Human Genetics, 2015, 23, 1538-1543.	2.8	19
21	Exome sequencing reveals germline gain-of-function EGFR mutation in an adult with Lhermitteâ€“Duclos disease. Journal of Physical Education and Sports Management, 2016, 2, a001230.	1.2	19
22	Apparently Heterozygous TP53 Pathogenic Variants May Be Blood Limited in Patients Undergoing Hereditary Cancer Panel Testing. Journal of Molecular Diagnostics, 2020, 22, 396-404.	2.8	17
23	Suggested application of HER2+ breast tumor phenotype for germline TP53 variant classification within ACMG/AMP guidelines. Human Mutation, 2020, 41, 1555-1562.	2.5	16
24	Germline compound heterozygous poly-glutamine deletion inUSF3may be involved in predisposition to heritable and sporadic epithelial thyroid carcinoma. Human Molecular Genetics, 2016, 26, ddw382.	2.9	14
25	Communicating with Biobank Participants: Preferences for Receiving and Providing Updates to Researchers. Cancer Epidemiology Biomarkers and Prevention, 2015, 24, 708-712.	2.5	13
26	Genetic counselors: Your partners in clinical practice. Cleveland Clinic Journal of Medicine, 2012, 79, 560-568.	1.3	9
27	Genotype-phenotype correlations among TP53 carriers: Literature review and analysis of probands undergoing multi-gene panel testing and single-gene testing. Cancer Genetics, 2020, 248-249, 11-17.	0.4	8
28	Pulmonary Arterial Hypertension in a Patient with Cowden Syndrome and the PTEN Mutation. Pulmonary Circulation, 2014, 4, 728-731.	1.7	6
29	Perceptions of Licensure: A Survey of Michigan Genetic Counselors. Journal of Genetic Counseling, 2009, 18, 357-365.	1.6	4
30	The Evolution of Constitutional Sequence Variant Interpretation. Clinics in Laboratory Medicine, 2020, 40, 135-148.	1.4	1
31	The Evolution of Constitutional Sequence Variant Interpretation. Advances in Molecular Pathology, 2019, 2, 1-11.	0.4	0
32	International consensus guidelines for constitutional sequence variant interpretation. , 2021, , 29-40.		0
33	PTEN germline mutations in patients first tested for other hereditary cancer syndromes: Would use of risk assessment tools have reduced health care costs?. Journal of Clinical Oncology, 2012, 30, 1516-1516.	1.6	0
34	Use of PTEN protein dosage to predict for underlying germ-line PTEN mutations among patients presenting with thyroid cancer and Cowden-like phenotypes.. Journal of Clinical Oncology, 2012, 30, 1508-1508.	1.6	0
35	Second malignant neoplasms (SMN) in Cowden syndrome patients with underlying germline PTEN mutations.. Journal of Clinical Oncology, 2013, 31, 1527-1527.	1.6	0