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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
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37
docs citations

37
times ranked

3765
citing authors

#	ARTICLE	IF	CITATIONS
1	ClinGen Variant Curation Interface: a variant classification platform for the application of evidence criteria from ACMG/AMP guidelines. <i>Genome Medicine</i> , 2022, 14, 6.	8.2	34
2	Specifications of the ACMG/AMP variant interpretation guidelines for germline <i>TP53</i> variants. <i>Human Mutation</i> , 2021, 42, 223-236.	2.5	81
3	International consensus guidelines for constitutional sequence variant interpretation. , 2021, , 29-40.		0
4	Apparently Heterozygous TP53 Pathogenic Variants May Be Blood Limited in Patients Undergoing Hereditary Cancer Panel Testing. <i>Journal of Molecular Diagnostics</i> , 2020, 22, 396-404.	2.8	17
5	Genotype-phenotype correlations among TP53 carriers: Literature review and analysis of probands undergoing multi-gene panel testing and single-gene testing. <i>Cancer Genetics</i> , 2020, 248-249, 11-17.	0.4	8
6	Suggested application of HER2+ breast tumor phenotype for germline <i>TP53</i> variant classification within ACMG/AMP guidelines. <i>Human Mutation</i> , 2020, 41, 1555-1562.	2.5	16
7	The Evolution of Constitutional Sequence Variant Interpretation. <i>Clinics in Laboratory Medicine</i> , 2020, 40, 135-148.	1.4	1
8	The Evolution of Constitutional Sequence Variant Interpretation. <i>Advances in Molecular Pathology</i> , 2019, 2, 1-11.	0.4	0
9	Gene-specific criteria for <i>PTEN</i> variant curation: Recommendations from the ClinGen PTEN Expert Panel. <i>Human Mutation</i> , 2018, 39, 1581-1592.	2.5	123
10	ClinGen Variant Curation Expert Panel experiences and standardized processes for disease and gene-level specification of the ACMG/AMP guidelines for sequence variant interpretation. <i>Human Mutation</i> , 2018, 39, 1614-1622.	2.5	132
11	Germline compound heterozygous poly-glutamine deletion in <i>USF3</i> may be involved in predisposition to heritable and sporadic epithelial thyroid carcinoma. <i>Human Molecular Genetics</i> , 2016, 26, ddw382.	2.9	14
12	Exome sequencing reveals germline gain-of-function <i>EGFR</i> mutation in an adult with Lhermitte-Duclos disease. <i>Journal of Physical Education and Sports Management</i> , 2016, 2, a001230.	1.2	19
13	Assessment of clinical workload for general and specialty genetic counsellors at an academic medical center: a tool for evaluating genetic counselling practices. <i>Npj Genomic Medicine</i> , 2016, 1, 16010.	3.8	28
14	<i>PTEN</i> hamartoma tumor syndrome. <i>Handbook of Clinical Neurology</i> / Edited By P J Vinken and G W Bruyn, 2015, 132, 129-137.	1.8	41
15	<i>KLLN</i> epigenotype-phenotype associations in Cowden syndrome. <i>European Journal of Human Genetics</i> , 2015, 23, 1538-1543.	2.8	19
16	Communicating with Biobank Participants: Preferences for Receiving and Providing Updates to Researchers. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2015, 24, 708-712.	2.5	13
17	Germline Heterozygous Variants in <i>SEC23B</i> Are Associated with Cowden Syndrome and Enriched in Apparently Sporadic Thyroid Cancer. <i>American Journal of Human Genetics</i> , 2015, 97, 661-676.	6.2	76
18	Germline <i>PTEN</i> , <i>SDHB</i> , and <i>KLLN</i> alterations in endometrial cancer patients with Cowden and Cowden-like syndromes: An international, multicenter, prospective study. <i>Cancer</i> , 2015, 121, 688-696.	4.1	46

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19	Pulmonary Arterial Hypertension in a Patient with Cowden Syndrome and the <i>PTEN</i> Mutation. <i>Pulmonary Circulation</i> , 2014, 4, 728-731.	1.7	6
20	Biochemical screening and PTEN mutation analysis in individuals with autism spectrum disorders and macrocephaly. <i>European Journal of Human Genetics</i> , 2014, 22, 273-276.	2.8	72
21	Germline PIK3CA and AKT1 Mutations in Cowden and Cowden-like Syndromes. <i>American Journal of Human Genetics</i> , 2013, 92, 76-80.	6.2	174
22	When Overgrowth Bumps Into Cancer: The PTENopathies. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2013, 163, 114-121.	1.6	112
23	A Novel Germline Mutation in <i>BAP1</i> Predisposes to Familial Clear-Cell Renal Cell Carcinoma. <i>Molecular Cancer Research</i> , 2013, 11, 1061-1071.	3.4	135
24	PTEN Germline Mutations in Patients Initially Tested for Other Hereditary Cancer Syndromes: Would Use of Risk Assessment Tools Reduce Genetic Testing?. <i>Oncologist</i> , 2013, 18, 1083-1090.	3.7	22
25	Second malignant neoplasms (SMN) in Cowden syndrome patients with underlying germline PTEN mutations.. <i>Journal of Clinical Oncology</i> , 2013, 31, 1527-1527.	1.6	0
26	Lifetime Cancer Risks in Individuals with Germline <i>PTEN</i> Mutations. <i>Clinical Cancer Research</i> , 2012, 18, 400-407.	7.0	738
27	Papillary Renal Cell Carcinoma Is Associated With PTEN Hamartoma Tumor Syndrome. <i>Urology</i> , 2012, 79, 1187.e1-1187.e7.	1.0	63
28	Estimate of de novo mutation frequency in probands with PTEN hamartoma tumor syndrome. <i>Genetics in Medicine</i> , 2012, 14, 819-822.	2.4	78
29	Genetic counselors: Your partners in clinical practice. <i>Cleveland Clinic Journal of Medicine</i> , 2012, 79, 560-568.	1.3	9
30	PTEN germline mutations in patients first tested for other hereditary cancer syndromes: Would use of risk assessment tools have reduced health care costs?. <i>Journal of Clinical Oncology</i> , 2012, 30, 1516-1516.	1.6	0
31	Use of PTEN protein dosage to predict for underlying germ-line PTEN mutations among patients presenting with thyroid cancer and Cowden-like phenotypes.. <i>Journal of Clinical Oncology</i> , 2012, 30, 1508-1508.	1.6	0
32	Analysis of prevalence and degree of macrocephaly in patients with germline PTEN mutations and of brain weight in Pten knock-in murine model. <i>European Journal of Human Genetics</i> , 2011, 19, 763-768.	2.8	93
33	A Clinical Scoring System for Selection of Patients for PTEN Mutation Testing Is Proposed on the Basis of a Prospective Study of 3042 Proband. <i>American Journal of Human Genetics</i> , 2011, 88, 42-56.	6.2	332
34	Germline Epigenetic Regulation of <i>KILLIN</i> in Cowden and Cowden-like Syndrome. <i>JAMA - Journal of the American Medical Association</i> , 2010, 304, 2724.	7.4	138
35	Perceptions of Licensure: A Survey of Michigan Genetic Counselors. <i>Journal of Genetic Counseling</i> , 2009, 18, 357-365.	1.6	4