Nicola K Poplawski

List of Publications by Year
in descending order

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42	1,501	516710 16	315739 38
papers	citations	h-index	g-index

44	44	44	3049
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	<scp>RNF43</scp> pathogenic Germline variant in a family with colorectal cancer. Clinical Genetics, 2022, 101, 122-126.	2.0	3
2	Survey of germline variants in cancerâ€associated genes in young adults with colorectal cancer. Genes Chromosomes and Cancer, 2022, 61, 105-113.	2.8	7
3	Recurrent pneumothorax in a case of t enascinâ€X deficient Ehlersâ€“Danlos syndrome: Broadening the phenotypic spectrum. American Journal of Medical Genetics, Part A, 2022, , .	1.2	2
4	Integrated guidance to enhance the care of children and adolescents with familial hypercholesterolaemia: Practical advice for the community clinician. Journal of Paediatrics and Child Health, 2022, 58, 1297-1312.	0.8	6
5	Implementing gene curation for hereditary cancer susceptibility in Australia: achieving consensus on genes with clinical utility. Journal of Medical Genetics, 2021, 58, 853-858.	3.2	3
6	Integrated Guidance for Enhancing the Care of Familial Hypercholesterolaemia in Australia. Heart Lung and Circulation, 2021, 30, 324-349.	0.4	51
7	Youngâ€onset colorectal cancer is associated with a personal history of type 2 diabetes. Asia-Pacific Journal of Clinical Oncology, 2021, 17, 131-138.	1.1	19
8	A case of Carney triad complicated by renal cell carcinoma and a germline SDHA pathogenic variant. Endocrinology, Diabetes and Metabolism Case Reports, 2021, 2021, .	0.5	2
9	Targeted gene panels identify a high frequency of pathogenic germline variants in patients diagnosed with a hematological malignancy and at least one other independent cancer. Leukemia, 2021, 35, 3245-3256.	7.2	32
10	Essentials of a new clinical practice guidance on familial hypercholesterolaemia for physicians. Internal Medicine Journal, 2021, 51, 769-779.	0.8	4
11	A Case of Carney Triad Complicated by Renal Cell Carcinoma and a Germline SDHA Pathogenic Variant. Journal of the Endocrine Society, 2021, 5, A985-A985.	0.2	0
12	Synopsis of an integrated guidance for enhancing the care of familial hypercholesterolaemia: an Australian perspective. American Journal of Preventive Cardiology, 2021, 6, 100151.	3.0	3
13	A prospective prostate cancer screening programme for men with pathogenic variants in mismatch repair genes (IMPACT): initial results from an international prospective study. Lancet Oncology, The, 2021, 22, 1618-1631.	10.7	48
14	Germline PALB2 Variants and PARP Inhibitors in Endometrial Cancer. Journal of the National Comprehensive Cancer Network: JNCCN, 2021, 19, 1212-1217.	4.9	3
15	Population-based estimates of breast cancer risk for carriers of pathogenic variants identified by gene-panel testing. Npj Breast Cancer, 2021, 7, 153.	5.2	10
16	Aberrant Splicing of SDHC in Families With Unexplained Succinate Dehydrogenase-Deficient Paragangliomas. Journal of the Endocrine Society, 2020, 4, bvaa071.	0.2	9
17	RUNX1-mutated families show phenotype heterogeneity and a somatic mutation profile unique to germline predisposed AML. Blood Advances, 2020, 4, 1131-1144.	5.2	102
18	A synonymous GATA2 variant underlying familial myeloid malignancy with striking intrafamilial phenotypic variability. British Journal of Haematology, 2020, 190, e297-e301.	2.5	14

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19	<scp>CDH1</scp>â€related blepharocheilodontic syndrome is associated with diffuse gastric cancer risk. American Journal of Medical Genetics, Part A, 2020, 182, 1780-1784.	1.2	5
20	Two monogenic disorders masquerading as one: severe congenital neutropenia with monocytosis and non-syndromic sensorineural hearing loss. BMC Medical Genetics, 2020, 21, 35.	2.1	3
21	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
22	Breast cancer in patients with germline TP53 pathogenic variants have typical tumour characteristics: the Cohort study of TP53 carrier early onset breast cancer (COPE study). Journal of Pathology: Clinical Research, 2019, 5, 189-198.	3.0	18
23	Australian Familial Haematological Cancer Study - Findings from 15 Years of Aggregated Clinical, Genomic and Transcriptomic Data. Blood, 2019, 134, 1439-1439.	1.4	2
24	Familial GATA6 mutation causing variably expressed diabetes mellitus and cardiac and renal abnormalities. Endocrinology, Diabetes and Metabolism Case Reports, 2019, 2019, .	0.5	3
25	An Integrative Genomic Approach to Examine Mutations and Biological Pathways Associated with Hematological Malignancy Development in DDX41 Mutated Families. Blood, 2019, 134, 2686-2686.	1.4	1
26	Familial Clustering of Hematological Malignancies: Harbingers of Wider Germline Cancer Susceptibility. Blood, 2019, 134, 3794-3794.	1.4	0
27	Comprehensive Study of the Clinical Phenotype of GermlineBAP1Variant-Carrying Families Worldwide. Journal of the National Cancer Institute, 2018, 110, 1328-1341.	6.3	164
28	Medicareâ€funded cancer genetic tests: a note of caution. Medical Journal of Australia, 2018, 209, 193-196.	1.7	10
29	Association of Genetic Predisposition With Solitary Schwannoma or Meningioma in Children and Young Adults. JAMA Neurology, 2017, 74, 1123.	9.0	63
30	Point Mutations in Exon 1B of APC Reveal Gastric Adenocarcinoma and Proximal Polyposis of the Stomach as a Familial Adenomatous Polyposis Variant. American Journal of Human Genetics, 2016, 98, 830-842.	6.2	201
31	Novel germ line DDX41 mutations define families with a lower age of MDS/AML onset and lymphoid malignancies. Blood, 2016, 127, 1017-1023.	1.4	179
32	Combined genetic and splicing analysis of BRCA1 c.[594-2A>C; 641A>G] highlights the relevance of naturally occurring in-frame transcripts for developing disease gene variant classification algorithms. Human Molecular Genetics, 2016, 25, 2256-2268.	2.9	106
33	Germline mutations inPMS2andMLH1in individuals with solitary loss of PMS2 expression in colorectal carcinomas from the Colon Cancer Family Registry Cohort. BMJ Open, 2016, 6, e010293.	1.9	33
34	A New Model of Care for Familial Hypercholesterolaemia: What is the Role of Cardiology?. Heart Lung and Circulation, 2012, 21, 543-550.	0.4	16
35	A model of care for familial hypercholesterolaemia: key role for clinical biochemistry. Clinical Biochemist Reviews, 2012, 33, 25-31.	3.3	13
36	Familial hypercholesterolaemia: A model of care for Australasia. Atherosclerosis Supplements, 2011, 12, 221-263.	1.2	181

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37	Characterization of novel SLC6A8 variants with the use of splice-site analysis tools and implementation of a newly developed LOVD database. European Journal of Human Genetics, 2011, 19, 56-63.	2.8	18
38	Poor outcome after liver transplantation for transthyretin amyloid neuropathy in a family with an Ala36Pro transthyretin mutation: Case Report. Liver Transplantation, 2010, 16, NA-NA.	2.4	4
39	Interstitial deletion of 1p22.2p31.1 and medium-chain acyl-CoA dehydrogenase deficiency in a patient with global developmental delay. American Journal of Medical Genetics, Part A, 2008, 146A, 1581-1586.	1.2	11
40	CDG-IL: An infant with a novel mutation in theALG9 gene and additional phenotypic features. American Journal of Medical Genetics, Part A, 2005, 136A, 194-197.	1.2	37
41	Multiple acyl-coenzyme A dehydrogenase deficiency: Diagnosis by acyl-carnitine analysis of a 12-year-old newborn screening card. Journal of Pediatrics, 1999, 134, 764-766.	1.8	11
42	An Ethical Issue for Reproductive Technologies*. Asia-Oceania Journal of Obstetrics and Gynaecology, 1990, 16, 291-296.	0.0	2