# Nicola K Poplawski

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/9584743/publications.pdf

Version: 2024-02-01

| 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 | 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 |
| 2 | Familial hypercholesterolaemia: A model of care for Australasia. Atherosclerosis Supplements, 2011, 12, 221-263. | 1.2 | 181 |
| 3 | 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 |
| 4 | Comprehensive Study of the Clinical Phenotype of GermlineBAP1Variant-Carrying Families Worldwide. Journal of the National Cancer Institute, 2018, 110, 1328-1341. | 6.3 | 164 |
| 5 | 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 |
| 6 | 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 |
| 7 | 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 |
| 8 | Association of Genetic Predisposition With Solitary Schwannoma or Meningioma in Children and Young Adults. JAMA Neurology, 2017, 74, 1123. | 9.0 | 63 |
| 9 | Integrated Guidance for Enhancing the Care of Familial Hypercholesterolaemia in Australia. Heart Lung and Circulation, 2021, 30, 324-349. | 0.4 | 51 |
| 10 | 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 |
| 11 | 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 |
| 12 | 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 |
| 13 | 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 |
| 14 | 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 |
| 15 | 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 |
| 16 | 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 |
| 17 | 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 |
| 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 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | A model of care for familial hypercholesterolaemia: key role for clinical biochemistry. Clinical Biochemist Reviews, 2012, 33, 25-31. | 3.3 | 13 |
| 20 | 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 |
| 21 | 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 |
| 22 | Medicareâ€funded cancer genetic tests: a note of caution. Medical Journal of Australia, 2018, 209, 193-196. | 1.7 | 10 |
| 23 | 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 |
| 24 | Aberrant Splicing of SDHC in Families With Unexplained Succinate Dehydrogenase-Deficient Paragangliomas. Journal of the Endocrine Society, 2020, 4, bvaa071. | 0.2 | 9 |
| 25 | 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 |
| 26 | 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 |
| 27 | <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 |
| 28 | 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 |
| 29 | Essentials of a new clinical practice guidance on familial hypercholesterolaemia for physicians. Internal Medicine Journal, 2021, 51, 769-779. | 0.8 | 4 |
| 30 | 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 |
| 31 | 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 |
| 32 | <scp>RNF43</scp> pathogenic Germline variant in a family with colorectal cancer. Clinical Genetics, 2022, 101, 122-126. | 2.0 | 3 |
| 33 | 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 |
| 34 | Familial GATA6 mutation causing variably expressed diabetes mellitus and cardiac and renal abnormalities. Endocrinology, Diabetes and Metabolism Case Reports, 2019, 2019, . | 0.5 | 3 |
| 35 | Germline PALB2 Variants and PARP Inhibitors in Endometrial Cancer. Journal of the National Comprehensive Cancer Network: JNCCN, 2021, 19, 1212-1217. | 4.9 | 3 |
| 36 | An Ethical Issue for Reproductive Technologies*. Asia-Oceania Journal of Obstetrics and Gynaecology, 1990, 16, 291-296. | 0.0 | 2 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | 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 |
| 38 | Australian Familial Haematological Cancer Study - Findings from 15 Years of Aggregated Clinical, Genomic and Transcriptomic Data. Blood, 2019, 134, 1439-1439. | 1.4 | 2 |
| 39 | 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 |
| 40 | 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 |
| 41 | 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 |
| 42 | Familial Clustering of Hematological Malignancies: Harbingers of Wider Germline Cancer Susceptibility. Blood, 2019, 134, 3794-3794. | 1.4 | 0 |