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List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/7721180/publications.pdf

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17 papers

1,674 citations

933447 10 h-index 17 g-index

17 all docs

17 docs citations

17 times ranked

4375 citing authors

#	Article	IF	CITATIONS
1	Breast and Prostate Cancer Risks for Male <i>BRCA1</i> and <i>BRCA2</i> Pathogenic Variant Carriers Using Polygenic Risk Scores. Journal of the National Cancer Institute, 2022, 114, 109-122.	6.3	19
2	Use of whole genome sequencing to determine genetic basis of suspected mitochondrial disorders: cohort study. BMJ, The, 2021, 375, e066288.	6.0	42
3	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
4	Diverse presentations of cutaneous mosaicism occur in CYLD cutaneous syndrome and may result in parent-to-child transmission. Journal of the American Academy of Dermatology, 2019, 81, 1300-1307.	1.2	5
5	Germline selection shapes human mitochondrial DNA diversity. Science, 2019, 364, .	12.6	178
6	A multicentre study of patients with Timothy syndrome. Europace, 2018, 20, 377-385.	1.7	23
7	Association analysis identifies 65 new breast cancer risk loci. Nature, 2017, 551, 92-94.	27.8	1,099
8	Inherited skin tumour syndromes. Clinical Medicine, 2017, 17, 562-567.	1.9	7
9	<i><scp>SOS</scp>1</i> frameshift mutations cause pure mucosal neuroma syndrome, a clinical phenotype distinct from multiple endocrine neoplasia type 2B. Clinical Endocrinology, 2016, 84, 715-719.	2.4	11
10	Breast cancer risk in <scp>MEN</scp> 1 – a cancer genetics perspective. Clinical Endocrinology, 2015, 82, 327-229.	2.4	12
11	Caseâ€finding in Fabry disease: experience from the North of England. Journal of Inherited Metabolic Disease, 2014, 37, 103-107.	3.6	4
12	Evaluation of <scp>SDHB</scp> , <scp> SDHD</scp> and <scp>VHL</scp> gene susceptibility testing in the assessment of individuals with nonâ€syndromic phaeochromocytoma, paraganglioma and head and neck paraganglioma. Clinical Endocrinology, 2013, 78, 898-906.	2.4	62
13	Cancer family history triage: a key step in the decision to offer screening and genetic testing. Familial Cancer, 2013, 12, 497-502.	1.9	4
14	Juvenile idiopathic arthritis, mitral valve prolapse and a familial variant involving the integrinâ€binding fragment of FBN1. American Journal of Medical Genetics, Part A, 2013, 161, 2047-2051.	1.2	5
15	â€~Over-the-counter' genetic testing: what does it really mean for primary care?. British Journal of General Practice, 2009, 59, 283-287.	1.4	41
16	The Teesside Cancer Family History Service: change management and innovation at cancer network level. Familial Cancer, 2007, 6, 181-187.	1.9	8
17	Collecting genetic information in primary care: evaluating a new family history tool. Family Practice, 2005, 22, 663-669.	1.9	72