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List of Publications by Year in descending order

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Version: 2024-02-01

17
papers

1,674
citations

933447

10
h-index

888059

17
g-index

17
all docs

17
docs citations

17
times ranked

4375
citing authors

| # | ARTICLE | IF | CITATIONS |
|----|---|------|-----------|
| 1 | Association analysis identifies 65 new breast cancer risk loci. <i>Nature</i> , 2017, 551, 92-94. | 27.8 | 1,099 |
| 2 | Germline selection shapes human mitochondrial DNA diversity. <i>Science</i> , 2019, 364, . | 12.6 | 178 |
| 3 | Polygenic risk scores and breast and epithelial ovarian cancer risks for carriers of BRCA1 and BRCA2 pathogenic variants. <i>Genetics in Medicine</i> , 2020, 22, 1653-1666. | 2.4 | 82 |
| 4 | Collecting genetic information in primary care: evaluating a new family history tool. <i>Family Practice</i> , 2005, 22, 663-669. | 1.9 | 72 |
| 5 | Evaluation of <sc>SDHB</sc>, <sc>SDHD</sc> and <sc>VHL</sc> gene susceptibility testing in the assessment of individuals with nonâ€ syndromic pheochromocytoma, paraganglioma and head and neck paraganglioma. <i>Clinical Endocrinology</i> , 2013, 78, 898-906. | 2.4 | 62 |
| 6 | Use of whole genome sequencing to determine genetic basis of suspected mitochondrial disorders: cohort study. <i>BMJ, The</i> , 2021, 375, e066288. | 6.0 | 42 |
| 7 | â€Over-the-counterâ€™ genetic testing: what does it really mean for primary care?. <i>British Journal of General Practice</i> , 2009, 59, 283-287. | 1.4 | 41 |
| 8 | A multicentre study of patients with Timothy syndrome. <i>Europace</i> , 2018, 20, 377-385. | 1.7 | 23 |
| 9 | Breast and Prostate Cancer Risks for Male <i>BRCA1</i> and <i>BRCA2</i> Pathogenic Variant Carriers Using Polygenic Risk Scores. <i>Journal of the National Cancer Institute</i> , 2022, 114, 109-122. | 6.3 | 19 |
| 10 | Breast cancer risk in <sc>MEN</sc>1 â€ a cancer genetics perspective. <i>Clinical Endocrinology</i> , 2015, 82, 327-229. | 2.4 | 12 |
| 11 | <i>SOS</i>1 frameshift mutations cause pure mucosal neuroma syndrome, a clinical phenotype distinct from multiple endocrine neoplasia type 2B. <i>Clinical Endocrinology</i> , 2016, 84, 715-719. | 2.4 | 11 |
| 12 | The Teesside Cancer Family History Service: change management and innovation at cancer network level. <i>Familial Cancer</i> , 2007, 6, 181-187. | 1.9 | 8 |
| 13 | Inherited skin tumour syndromes. <i>Clinical Medicine</i> , 2017, 17, 562-567. | 1.9 | 7 |
| 14 | Juvenile idiopathic arthritis, mitral valve prolapse and a familial variant involving the integrinâ€ binding fragment of FBN1. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 2047-2051. | 1.2 | 5 |
| 15 | Diverse presentations of cutaneous mosaicism occur in CYLD cutaneous syndrome and may result in parent-to-child transmission. <i>Journal of the American Academy of Dermatology</i> , 2019, 81, 1300-1307. | 1.2 | 5 |
| 16 | Cancer family history triage: a key step in the decision to offer screening and genetic testing. <i>Familial Cancer</i> , 2013, 12, 497-502. | 1.9 | 4 |
| 17 | Caseâ€ finding in Fabry disease: experience from the North of England. <i>Journal of Inherited Metabolic Disease</i> , 2014, 37, 103-107. | 3.6 | 4 |