# Payal P Khincha

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/8365392/publications.pdf

Version: 2024-02-01

| 38 | 1,289 | 567144 15 | 377752 34 |
|---|---|---|---|
| papers | citations | h-index | g-index |

| 40 | 40 | 40 | 2227 |
|---|---|---|---|
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Risks of first and subsequent cancers among TP53 mutation carriers in the National Cancer Institute Liâ€Fraumeni syndrome cohort. Cancer, 2016, 122, 3673-3681. | 2.0 | 346 |
| 2 | Baseline Surveillance in Li-Fraumeni Syndrome Using Whole-Body Magnetic Resonance Imaging. JAMA Oncology, 2017, 3, 1634. | 3.4 | 148 |
| 3 | Response to androgen therapy in patients with dyskeratosis congenita. British Journal of Haematology, 2014, 165, 349-357. | 1.2 | 89 |
| 4 | Novel and known ribosomal causes of Diamond-Blackfan anaemia identified through comprehensive genomic characterisation. Journal of Medical Genetics, 2017, 54, 417-425. | 1.5 | 71 |
| 5 | Genomic Characterization of the Inherited Bone Marrow Failure Syndromes. Seminars in Hematology, 2013, 50, 333-347. | 1.8 | 69 |
| 6 | Variable population prevalence estimates of germline TP53 variants: A gnomAD-based analysis. Human Mutation, 2019, 40, 97-105. | 1.1 | 66 |
| 7 | Understanding the evolving phenotype of vascular complications in telomere biology disorders. Angiogenesis, 2019, 22, 95-102. | 3.7 | 45 |
| 8 | Prevalence of Cancer at Baseline Screening in the National Cancer Institute Li-Fraumeni Syndrome Cohort. JAMA Oncology, 2017, 3, 1640. | 3.4 | 43 |
| 9 | Correlation of Leukocyte Telomere Length Measurement Methods in Patients with Dyskeratosis Congenita and in Their Unaffected Relatives. International Journal of Molecular Sciences, 2017, 18, 1765. | 1.8 | 42 |
| 10 | Pulmonary arteriovenous malformations: an uncharacterised phenotype of dyskeratosis congenita and related telomere biology disorders. European Respiratory Journal, 2017, 49, 1601640. | 3.1 | 41 |
| 11 | Neonatal manifestations of inherited bone marrow failure syndromes. Seminars in Fetal and Neonatal Medicine, 2016, 21, 57-65. | 1.1 | 37 |
| 12 | Similar telomere attrition rates in androgen-treated and untreated patients with dyskeratosis congenita. Blood Advances, 2018, 2, 1243-1249. | 2.5 | 30 |
| 13 | Cancer incidence, patterns, and genotypeâ€“phenotype associations in individuals with pathogenic or likely pathogenic germline TP53 variants: an observational cohort study. Lancet Oncology, The, 2021, 22, 1787-1798. | 5.1 | 29 |
| 14 | Fundamental immuneâ€“oncogenicity trade-offs define driver mutationÂfitness. Nature, 2022, 606, 172-179. | 13.7 | 23 |
| 15 | Investigation of chromosome X inactivation and clinical phenotypes in female carriers of DKC1 mutations. American Journal of Hematology, 2016, 91, 1215-1220. | 2.0 | 22 |
| 16 | Couples coping with screening burden and diagnostic uncertainty in Li-Fraumeni syndrome: Connection versus independence. Journal of Psychosocial Oncology, 2019, 37, 178-193. | 0.6 | 21 |
| 17 | The limitations of qPCR telomere length measurement in diagnosing dyskeratosis congenita. Molecular Genetics & Genomic Medicine, 2016, 4, 475-479. | 0.6 | 20 |
| 18 | Suggested application of HER2+ breast tumor phenotype for germline TP53 variant classification within ACMG/AMP guidelines. Human Mutation, 2020, 41, 1555-1562. | 1.1 | 16 |

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|---|---|---|---|
| 19 | Prevalence of pathogenic/likely pathogenic variants in the 24 cancer genes of the ACMG Secondary Findings v2.0 list in a large cancer cohort and ethnicity-matched controls. Genome Medicine, 2018, 10, 99. | 3.6 | 15 |
| 20 | Gastrointestinal Hemorrhage: A Manifestation of the Telomere Biology Disorders. Journal of Pediatrics, 2021, 230, 55-61.e4. | 0.9 | 14 |
| 21 | Expansion of germline RPS20 mutation phenotype to include Diamondâ€“Blackfan anemia. Human Mutation, 2020, 41, 1918-1930. | 1.1 | 13 |
| 22 | Waiting and â€œweighted downâ€: the challenge of anticipatory loss for individuals and families with Li-Fraumeni Syndrome. Familial Cancer, 2020, 19, 259-268. | 0.9 | 13 |
| 23 | Frequency of Pathogenic Germline Variants in Cancer-Susceptibility Genes in the Childhood Cancer Survivor Study. JNCI Cancer Spectrum, 2021, 5, pkab007. | 1.4 | 11 |
| 24 | Reproductive factors associated with breast cancer risk in Liâ€“Fraumeni syndrome. European Journal of Cancer, 2019, 116, 199-206. | 1.3 | 10 |
| 25 | Family Health Leaders: Lessons on Living with Liâ€Fraumeni Syndrome across Generations. Family Process, 2020, 59, 1648-1663. | 1.4 | 10 |
| 26 | Complex phenotype of dyskeratosis congenita and mood dysregulation with novel homozygous RTEL1 and TPH1 variants. American Journal of Medical Genetics, Part A, 2018, 176, 1432-1437. | 0.7 | 7 |
| 27 | Pilot Study Assessing Tolerability and Metabolic Effects of Metformin in Patients With Li-Fraumeni Syndrome. JNCI Cancer Spectrum, 2020, 4, pkaa063. | 1.4 | 6 |
| 28 | Embodied risk for families with Li-Fraumeni syndrome: Like electricity through my body. Social Science and Medicine, 2022, 301, 114905. | 1.8 | 6 |
| 29 | Family Identity and Roles in the Context of Li-Fraumeni Syndrome: â€œNo Oneâ€™s Like Us Mutantsâ€: Health and Social Work, 2021, 46, 299-307. | 0.5 | 5 |
| 30 | Pathogenic germline IKZF1 variant alters hematopoietic gene expression profiles. Journal of Physical Education and Sports Management, 2021, 7, a006015. | 0.5 | 5 |
| 31 | Uptake and timing of bilateral and contralateral risk-reducing mastectomy in women with Liâ€“Fraumeni syndrome. Breast Cancer Research and Treatment, 2022, 191, 159-167. | 1.1 | 5 |
| 32 | Quantification of Discordant Variant Interpretations in a Large Family-Based Study of Li-Fraumeni Syndrome. JCO Precision Oncology, 2021, 5, 1727-1737. | 1.5 | 3 |
| 33 | â€œI can control what I do with my daily lifeâ€: Occupational experiences of adolescents and young adults with Li-Fraumeni Syndrome. Journal of Occupational Science, 0, , 1-12. | 0.7 | 3 |
| 34 | Response to: Concern regarding classification of germline TP53 variants as likely pathogenic. Human Mutation, 2019, 40, 832-833. | 1.1 | 1 |
| 35 | Utility of interim blood tests for cancer screening in Li-Fraumeni syndrome. Familial Cancer, 2022, 21, 333-336. | 0.9 | 1 |
| 36 | Response to Androgen Therapy and Side Effects in Patients with Dyskeratosis Congenita.. Blood, 2012, 120, 2361-2361. | 0.6 | 1 |

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|---|---|---|---|
| 37 | Lipoprotein particle alterations due to androgen therapy in individuals with dyskeratosis congenita. EBioMedicine, 2022, 75, 103760. | 2.7 | 1 |
| 38 | Spectrum and Incidence of Skin Cancer among Individuals with Li-Fraumeni Syndrome. Journal of Investigative Dermatology, 2022, 142, 2534-2537.e1. | 0.3 | 1 |