

# Payal P Khincha

## List of Publications by Year in descending order

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Version: 2024-02-01

38  
papers

1,289  
citations

567144

15  
h-index

377752

34  
g-index

40  
all docs

40  
docs citations

40  
times ranked

2227  
citing authors

#	ARTICLE	IF	CITATIONS
1	Utility of interim blood tests for cancer screening in Li-Fraumeni syndrome. <i>Familial Cancer</i> , 2022, 21, 333-336.	0.9	1
2	Uptake and timing of bilateral and contralateral risk-reducing mastectomy in women with Li-Fraumeni syndrome. <i>Breast Cancer Research and Treatment</i> , 2022, 191, 159-167.	1.1	5
3	Lipoprotein particle alterations due to androgen therapy in individuals with dyskeratosis congenita. <i>EBioMedicine</i> , 2022, 75, 103760.	2.7	1
4	Spectrum and Incidence of Skin Cancer among Individuals with Li-Fraumeni Syndrome. <i>Journal of Investigative Dermatology</i> , 2022, 142, 2534-2537.e1.	0.3	1
5	Embodied risk for families with Li-Fraumeni syndrome: Like electricity through my body. <i>Social Science and Medicine</i> , 2022, 301, 114905.	1.8	6
6	Fundamental immune-oncogenicity trade-offs define driver mutation fitness. <i>Nature</i> , 2022, 606, 172-179.	13.7	23
7	Gastrointestinal Hemorrhage: A Manifestation of the Telomere Biology Disorders. <i>Journal of Pediatrics</i> , 2021, 230, 55-61.e4.	0.9	14
8	Frequency of Pathogenic Germline Variants in Cancer-Susceptibility Genes in the Childhood Cancer Survivor Study. <i>JNCI Cancer Spectrum</i> , 2021, 5, pkab007.	1.4	11
9	Family Identity and Roles in the Context of Li-Fraumeni Syndrome: "No One's Like Us Mutants". <i>Health and Social Work</i> , 2021, 46, 299-307.	0.5	5
10	Pathogenic germline <i>KZF1</i> variant alters hematopoietic gene expression profiles. <i>Journal of Physical Education and Sports Management</i> , 2021, 7, a006015.	0.5	5
11	Quantification of Discordant Variant Interpretations in a Large Family-Based Study of Li-Fraumeni Syndrome. <i>JCO Precision Oncology</i> , 2021, 5, 1727-1737.	1.5	3
12	Cancer incidence, patterns, and genotype-phenotype associations in individuals with pathogenic or likely pathogenic germline TP53 variants: an observational cohort study. <i>Lancet Oncology</i> , The, 2021, 22, 1787-1798.	5.1	29
13	Family Health Leaders: Lessons on Living with Li-Fraumeni Syndrome across Generations. <i>Family Process</i> , 2020, 59, 1648-1663.	1.4	10
14	Pilot Study Assessing Tolerability and Metabolic Effects of Metformin in Patients With Li-Fraumeni Syndrome. <i>JNCI Cancer Spectrum</i> , 2020, 4, pkaa063.	1.4	6
15	Expansion of germline <i>RPS20</i> mutation phenotype to include Diamond-Blackfan anemia. <i>Human Mutation</i> , 2020, 41, 1918-1930.	1.1	13
16	Suggested application of HER2+ breast tumor phenotype for germline TP53 variant classification within ACMG/AMP guidelines. <i>Human Mutation</i> , 2020, 41, 1555-1562.	1.1	16
17	Waiting and "weighted down": the challenge of anticipatory loss for individuals and families with Li-Fraumeni Syndrome. <i>Familial Cancer</i> , 2020, 19, 259-268.	0.9	13
18	Couples coping with screening burden and diagnostic uncertainty in Li-Fraumeni syndrome: Connection versus independence. <i>Journal of Psychosocial Oncology</i> , 2019, 37, 178-193.	0.6	21

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19	Reproductive factors associated with breast cancer risk in Li-Fraumeni syndrome. <i>European Journal of Cancer</i> , 2019, 116, 199-206.	1.3	10
20	Response to: Concern regarding classification of germline TP53 variants as likely pathogenic. <i>Human Mutation</i> , 2019, 40, 832-833.	1.1	1
21	Understanding the evolving phenotype of vascular complications in telomere biology disorders. <i>Angiogenesis</i> , 2019, 22, 95-102.	3.7	45
22	Variable population prevalence estimates of germline TP53 variants: A gnomAD-based analysis. <i>Human Mutation</i> , 2019, 40, 97-105.	1.1	66
23	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. <i>Genome Medicine</i> , 2018, 10, 99.	3.6	15
24	Similar telomere attrition rates in androgen-treated and untreated patients with dyskeratosis congenita. <i>Blood Advances</i> , 2018, 2, 1243-1249.	2.5	30
25	Complex phenotype of dyskeratosis congenita and mood dysregulation with novel homozygous RTEL1 and TPH1 variants. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1432-1437.	0.7	7
26	Pulmonary arteriovenous malformations: an uncharacterised phenotype of dyskeratosis congenita and related telomere biology disorders. <i>European Respiratory Journal</i> , 2017, 49, 1601640.	3.1	41
27	Novel and known ribosomal causes of Diamond-Blackfan anaemia identified through comprehensive genomic characterisation. <i>Journal of Medical Genetics</i> , 2017, 54, 417-425.	1.5	71
28	Prevalence of Cancer at Baseline Screening in the National Cancer Institute Li-Fraumeni Syndrome Cohort. <i>JAMA Oncology</i> , 2017, 3, 1640.	3.4	43
29	Baseline Surveillance in Li-Fraumeni Syndrome Using Whole-Body Magnetic Resonance Imaging. <i>JAMA Oncology</i> , 2017, 3, 1634.	3.4	148
30	Correlation of Leukocyte Telomere Length Measurement Methods in Patients with Dyskeratosis Congenita and in Their Unaffected Relatives. <i>International Journal of Molecular Sciences</i> , 2017, 18, 1765.	1.8	42
31	Risks of first and subsequent cancers among TP53 mutation carriers in the National Cancer Institute Li-Fraumeni syndrome cohort. <i>Cancer</i> , 2016, 122, 3673-3681.	2.0	346
32	The limitations of qPCR telomere length measurement in diagnosing dyskeratosis congenita. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2016, 4, 475-479.	0.6	20
33	Investigation of chromosome X inactivation and clinical phenotypes in female carriers of DKC1 mutations. <i>American Journal of Hematology</i> , 2016, 91, 1215-1220.	2.0	22
34	Neonatal manifestations of inherited bone marrow failure syndromes. <i>Seminars in Fetal and Neonatal Medicine</i> , 2016, 21, 57-65.	1.1	37
35	Response to androgen therapy in patients with dyskeratosis congenita. <i>British Journal of Haematology</i> , 2014, 165, 349-357.	1.2	89
36	Genomic Characterization of the Inherited Bone Marrow Failure Syndromes. <i>Seminars in Hematology</i> , 2013, 50, 333-347.	1.8	69

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37	Response to Androgen Therapy and Side Effects in Patients with Dyskeratosis Congenita.. Blood, 2012, 120, 2361-2361.	0.6	1
38	“I can control what I do with my daily life” Occupational experiences of adolescents and young adults with Li-Fraumeni Syndrome. Journal of Occupational Science, 0, , 1-12.	0.7	3