

Payal P Khincha

List of Publications by Year in descending order

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Version: 2024-02-01

38
papers

1,289
citations

567281

15
h-index

377865

34
g-index

40
all docs

40
docs citations

40
times ranked

2227
citing authors

#	ARTICLE	IF	CITATIONS
1	Risks of first and subsequent cancers among <i>TP53</i> mutation carriers in the National Cancer Institute Li-Fraumeni syndrome cohort. <i>Cancer</i> , 2016, 122, 3673-3681.	4.1	346
2	Baseline Surveillance in Li-Fraumeni Syndrome Using Whole-Body Magnetic Resonance Imaging. <i>JAMA Oncology</i> , 2017, 3, 1634.	7.1	148
3	Response to androgen therapy in patients with dyskeratosis congenita. <i>British Journal of Haematology</i> , 2014, 165, 349-357.	2.5	89
4	Novel and known ribosomal causes of Diamond-Blackfan anaemia identified through comprehensive genomic characterisation. <i>Journal of Medical Genetics</i> , 2017, 54, 417-425.	3.2	71
5	Genomic Characterization of the Inherited Bone Marrow Failure Syndromes. <i>Seminars in Hematology</i> , 2013, 50, 333-347.	3.4	69
6	Variable population prevalence estimates of germline <i>TP53</i> variants: A gnomAD-based analysis. <i>Human Mutation</i> , 2019, 40, 97-105.	2.5	66
7	Understanding the evolving phenotype of vascular complications in telomere biology disorders. <i>Angiogenesis</i> , 2019, 22, 95-102.	7.2	45
8	Prevalence of Cancer at Baseline Screening in the National Cancer Institute Li-Fraumeni Syndrome Cohort. <i>JAMA Oncology</i> , 2017, 3, 1640.	7.1	43
9	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.	4.1	42
10	Pulmonary arteriovenous malformations: an uncharacterised phenotype of dyskeratosis congenita and related telomere biology disorders. <i>European Respiratory Journal</i> , 2017, 49, 1601640.	6.7	41
11	Neonatal manifestations of inherited bone marrow failure syndromes. <i>Seminars in Fetal and Neonatal Medicine</i> , 2016, 21, 57-65.	2.3	37
12	Similar telomere attrition rates in androgen-treated and untreated patients with dyskeratosis congenita. <i>Blood Advances</i> , 2018, 2, 1243-1249.	5.2	30
13	Cancer incidence, patterns, and genotype-phenotype associations in individuals with pathogenic or likely pathogenic germline <i>TP53</i> variants: an observational cohort study. <i>Lancet Oncology</i> , The, 2021, 22, 1787-1798.	10.7	29
14	Fundamental immune-oncogenicity trade-offs define driver mutation fitness. <i>Nature</i> , 2022, 606, 172-179.	27.8	23
15	Investigation of chromosome X inactivation and clinical phenotypes in female carriers of <i>DKC1</i> mutations. <i>American Journal of Hematology</i> , 2016, 91, 1215-1220.	4.1	22
16	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.	1.2	21
17	The limitations of qPCR telomere length measurement in diagnosing dyskeratosis congenita. <i>Molecular Genetics & Genomic Medicine</i> , 2016, 4, 475-479.	1.2	20
18	Suggested application of HER2+ breast tumor phenotype for germline <i>TP53</i> variant classification within ACMG/AMP guidelines. <i>Human Mutation</i> , 2020, 41, 1555-1562.	2.5	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. <i>Genome Medicine</i> , 2018, 10, 99.	8.2	15
20	Gastrointestinal Hemorrhage: A Manifestation of the Telomere Biology Disorders. <i>Journal of Pediatrics</i> , 2021, 230, 55-61.e4.	1.8	14
21	Expansion of germline <i>RPS20</i> mutation phenotype to include Diamond-Blackfan anemia. <i>Human Mutation</i> , 2020, 41, 1918-1930.	2.5	13
22	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.	1.9	13
23	Frequency of Pathogenic Germline Variants in Cancer-Susceptibility Genes in the Childhood Cancer Survivor Study. <i>JNCI Cancer Spectrum</i> , 2021, 5, pkab007.	2.9	11
24	Reproductive factors associated with breast cancer risk in Li-Fraumeni syndrome. <i>European Journal of Cancer</i> , 2019, 116, 199-206.	2.8	10
25	Family Health Leaders: Lessons on Living with Li-Fraumeni Syndrome across Generations. <i>Family Process</i> , 2020, 59, 1648-1663.	2.6	10
26	Complex phenotype of dyskeratosis congenita and mood dysregulation with novel homozygous <i>RTEL1</i> and <i>TPH1</i> variants. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1432-1437.	1.2	7
27	Pilot Study Assessing Tolerability and Metabolic Effects of Metformin in Patients With Li-Fraumeni Syndrome. <i>JNCI Cancer Spectrum</i> , 2020, 4, pkaa063.	2.9	6
28	Embodied risk for families with Li-Fraumeni syndrome: Like electricity through my body. <i>Social Science and Medicine</i> , 2022, 301, 114905.	3.8	6
29	Family Identity and Roles in the Context of Li-Fraumeni Syndrome: "No One's Like Us Mutants": Health and Social Work, 2021, 46, 299-307.	1.0	5
30	Pathogenic germline <i>KZF1</i> variant alters hematopoietic gene expression profiles. <i>Journal of Physical Education and Sports Management</i> , 2021, 7, a006015.	1.2	5
31	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.	2.5	5
32	Quantification of Discordant Variant Interpretations in a Large Family-Based Study of Li-Fraumeni Syndrome. <i>JCO Precision Oncology</i> , 2021, 5, 1727-1737.	3.0	3
33	"I can control what I do with my daily life": Occupational experiences of adolescents and young adults with Li-Fraumeni Syndrome. <i>Journal of Occupational Science</i> , 0, , 1-12.	1.3	3
34	Response to: Concern regarding classification of germline TP53 variants as likely pathogenic. <i>Human Mutation</i> , 2019, 40, 832-833.	2.5	1
35	Utility of interim blood tests for cancer screening in Li-Fraumeni syndrome. <i>Familial Cancer</i> , 2022, 21, 333-336.	1.9	1
36	Response to Androgen Therapy and Side Effects in Patients with Dyskeratosis Congenita.. <i>Blood</i> , 2012, 120, 2361-2361.	1.4	1

#	ARTICLE	IF	CITATIONS
37	Lipoprotein particle alterations due to androgen therapy in individuals with dyskeratosis congenita. EBioMedicine, 2022, 75, 103760.	6.1	1
38	Spectrum and Incidence of Skin Cancer among Individuals with Li-Fraumeni Syndrome. Journal of Investigative Dermatology, 2022, 142, 2534-2537.e1.	0.7	1