Saquib A Lakhani

List of Publications by Year
in descending order

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39	3,046	566801	301761
papers	citations	15 h-index	39 g-index

39	39	39	6032
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Caspases 3 and 7: Key Mediators of Mitochondrial Events of Apoptosis. Science, 2006, 311, 847-851.	6.0	1,003
2	Apoptotic Caspases Prevent the Induction of Type I Interferons by Mitochondrial DNA. Cell, 2014, 159, 1563-1577.	13.5	625
3	JNK Expression by Macrophages Promotes Obesity-Induced Insulin Resistance and Inflammation. Science, 2013, 339, 218-222.	6.0	544
4	A crucial role of caspase-3 in osteogenic differentiation of bone marrow stromal stem cells. Journal of Clinical Investigation, 2004, 114, 1704-1713.	3.9	221
5	Role of the Executioner Caspases during Lens Development. Journal of Biological Chemistry, 2005, 280, 30263-30272.	1.6	97
6	Endoplasmic Reticulum Stress-induced Death of Mouse Embryonic Fibroblasts Requires the Intrinsic Pathway of Apoptosis*. Journal of Biological Chemistry, 2007, 282, 14132-14139.	1.6	85
7	Patients with common variable immunodeficiency with autoimmune cytopenias exhibit hyperplastic yet inefficient germinal center responses. Journal of Allergy and Clinical Immunology, 2019, 143, 258-265.	1.5	68
8	Toll-like receptor signaling in sepsis. Current Opinion in Pediatrics, 2003, 15, 278-282.	1.0	50
9	De novo pathogenic variants in neuronal differentiation factor 2 (NEUROD2) cause a form of early infantile epileptic encephalopathy. Journal of Medical Genetics, 2019, 56, 113-122.	1.5	32
10	A novel SAMD9 mutation causing MIRAGE syndrome: An expansion and review of phenotype, dysmorphology, and natural history. American Journal of Medical Genetics, Part A, 2018, 176, 415-420.	0.7	30
11	Human autoinflammatory disease reveals ELF4 as a transcriptional regulator of inflammation. Nature Immunology, 2021, 22, 1118-1126.	7.0	30
12	<scp>DYNC1H1</scp>â€related disorders: A description of four new unrelated patients and a comprehensive review of previously reported variants. American Journal of Medical Genetics, Part A, 2020, 182, 2049-2057.	0.7	27
13	Molar tooth development in caspase-3 deficient mice. International Journal of Developmental Biology, 2006, 50, 491-7.	0.3	24
14	Data Science for Child Health. Journal of Pediatrics, 2019, 208, 12-22.	0.9	22
15	Novel truncating mutations in CTNND1 cause a dominant craniofacial and cardiac syndrome. Human Molecular Genetics, 2020, 29, 1900-1921.	1.4	21
16	Caspases and T lymphocytes: a flip of the coin?. Immunological Reviews, 2003, 193, 22-30.	2.8	18
17	Familial Dilated Cardiomyopathy Associated With a Novel Combination of Compound Heterozygous TNNC1 Variants. Frontiers in Physiology, 2019, 10, 1612.	1.3	15
18	Cytotoxic T-Lymphocyte-Associated Protein 4 Haploinsufficiency-Associated Inflammation Can Occur Independently of T-Cell Hyperproliferation. Frontiers in Immunology, 2018, 9, 1715.	2.2	13

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19	Siblings with lethal primary pulmonary hypoplasia and compound heterozygous variants in the AARS2 gene: further delineation of the phenotypic spectrum. Journal of Physical Education and Sports Management, 2019, 5, a003699.	0.5	12
20	Novel compound heterozygous variants in NHLRC2 in a patient with FINCA syndrome. Journal of Human Genetics, 2020, 65, 911-915.	1.1	11
21	DLG5 variants are associated with multiple congenital anomalies including ciliopathy phenotypes. Journal of Medical Genetics, 2021, 58, 453-464.	1.5	10
22	Whole-Exome Sequencing of Adult and Pediatric Cohorts of the Rare Vascular Disorder Systemic Capillary Leak Syndrome. Shock, 2019, 52, 183-190.	1.0	9
23	A novel variant in MAP3K7 associated with an expanded cardiospondylocarpofacial syndrome phenotype. Journal of Physical Education and Sports Management, 2020, 6, a005207.	0.5	9
24	Financial, Resource Utilization and Mortality Impacts of Teaching Hospital Status on Pediatric Patients Admitted for Sepsis. Pediatric Infectious Disease Journal, 2017, 36, 712-719.	1.1	7
25	Uncontrolled Epstein-Barr Virus as an Atypical Presentation of Deficiency in ADA2 (DADA2). Journal of Clinical Immunology, 2021, 41, 680-683.	2.0	7
26	Expansion of <scp>NEUROD2</scp> phenotypes to include developmental delay without seizures. American Journal of Medical Genetics, Part A, 2021, 185, 1076-1080.	0.7	7
27	Identification of novel mutations and phenotype in the steroid resistant nephrotic syndrome gene NUP93: a case report. BMC Nephrology, 2019, 20, 271.	0.8	6
28	Identification of a novel MYOC variant in a Hispanic family with early-onset primary open-angle glaucoma with elevated intraocular pressure. Journal of Physical Education and Sports Management, 2019, 5, a004374.	0.5	6
29	Two siblings with a novel nonsense variant provide further delineation of the spectrum of recessive KLHL7 diseases. European Journal of Medical Genetics, 2019, 62, 103551.	0.7	6
30	A homozygous variant in RRM2B is associated with severe metabolic acidosis and early neonatal death. European Journal of Medical Genetics, 2019, 62, 103574.	0.7	5
31	The latest FADS : Functional analysis of GLDN patient variants and classification of GLDN â€associated AMC as a type of viable fetal akinesia deformation sequence. American Journal of Medical Genetics, Part A, 2020, 182, 2291-2296.	0.7	5
32	Acid-Base Disorders. Pediatrics in Review, 2016, 37, 361-369.	0.2	4
33	Comparison of Transferred Versus Nontransferred Pediatric Patients Admitted for Sepsis. Air Medical Journal, 2016, 35, 43-45.	0.3	3
34	Rethinking what constitutes a diagnosis in the genomics era: a critical illness perspective. Current Opinion in Pediatrics, 2019, 31, 317-321.	1.0	3
35	A Novel Pathogenic UGT1A1 Variant in a Sudanese Child with Type 1 Crigler-Najjar Syndrome. Drug Metabolism and Disposition, 2019, 47, 45-48.	1.7	3
36	Functional testing for variant prioritization in a family with long QT syndrome. Molecular Genetics and Genomics, 2021, 296, 823-836.	1.0	3

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37	TBX5 variant with the novel phenotype of mixedâ€‘type total anomalous pulmonary venous return in Holtâ€‘Oram Syndrome and variable intrafamilial heart defects. Molecular Medicine Reports, 2022, 25, .	1.1	3
38	Natural sunscreen revealed. Nature Cell Biology, 2001, 3, E272-E272.	4.6	1
39	Healthcare costs and outcomes for pediatric inpatients with bronchiolitis: comparison of urban versus rural hospitals. Rural and Remote Health, 2015, 15, 3380.	0.4	1