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	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
2	DLG5 variants are associated with multiple congenital anomalies including ciliopathy phenotypes. Journal of Medical Genetics, 2021, 58, 453-464.	1.5	10
3	Uncontrolled Epstein-Barr Virus as an Atypical Presentation of Deficiency in ADA2 (DADA2). Journal of Clinical Immunology, 2021, 41, 680-683.	2.0	7
4	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
5	Functional testing for variant prioritization in a family with long QT syndrome. Molecular Genetics and Genomics, 2021, 296, 823-836.	1.0	3
6	Human autoinflammatory disease reveals ELF4 as a transcriptional regulator of inflammation. Nature Immunology, 2021, 22, 1118-1126.	7.0	30
7	<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
8	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
9	Novel compound heterozygous variants in NHLRC2 in a patient with FINCA syndrome. Journal of Human Genetics, 2020, 65, 911-915.	1.1	11
10	Novel truncating mutations in CTNND1 cause a dominant craniofacial and cardiac syndrome. Human Molecular Genetics, 2020, 29, 1900-1921.	1.4	21
11	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
12	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
13	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
14	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
15	Data Science for Child Health. Journal of Pediatrics, 2019, 208, 12-22.	0.9	22
16	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
17	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
18	Rethinking what constitutes a diagnosis in the genomics era: a critical illness perspective. Current Opinion in Pediatrics, 2019, 31, 317-321.	1.0	3

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19	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
20	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
21	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
22	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
23	Familial Dilated Cardiomyopathy Associated With a Novel Combination of Compound Heterozygous TNNC1 Variants. Frontiers in Physiology, 2019, 10, 1612.	1.3	15
24	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
25	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
26	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
27	Acid-Base Disorders. Pediatrics in Review, 2016, 37, 361-369.	0.2	4
28	Comparison of Transferred Versus Nontransferred Pediatric Patients Admitted for Sepsis. Air Medical Journal, 2016, 35, 43-45.	0.3	3
29	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
30	Apoptotic Caspases Prevent the Induction of Type I Interferons by Mitochondrial DNA. Cell, 2014, 159, 1563-1577.	13.5	625
31	JNK Expression by Macrophages Promotes Obesity-Induced Insulin Resistance and Inflammation. Science, 2013, 339, 218-222.	6.0	544
32	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
33	Caspases 3 and 7: Key Mediators of Mitochondrial Events of Apoptosis. Science, 2006, 311, 847-851.	6.0	1,003
34	Molar tooth development in caspase-3 deficient mice. International Journal of Developmental Biology, 2006, 50, 491-7.	0.3	24
35	Role of the Executioner Caspases during Lens Development. Journal of Biological Chemistry, 2005, 280, 30263-30272.	1.6	97
36	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

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37	Caspases and T lymphocytes: a flip of the coin?. Immunological Reviews, 2003, 193, 22-30.	2.8	18
38	Toll-like receptor signaling in sepsis. Current Opinion in Pediatrics, 2003, 15, 278-282.	1.0	50
39	Natural sunscreen revealed. Nature Cell Biology, 2001, 3, E272-E272.	4.6	1