Sandesh C S Nagamani

List of Publications by Year
in descending order

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42	1,915	394421	289244
papers	citations	19 h-index	40 g-index

43	43	43	3903
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Diversion of aspartate in ASS1-deficient tumours fosters de novo pyrimidine synthesis. Nature, 2015, 527, 379-383.	27.8	271
2	Branched-chain amino acid metabolism: from rare Mendelian diseases to more common disorders. Human Molecular Genetics, 2014, 23, R1-R8.	2.9	234
3	Parental Somatic Mosaicism Is Underrecognized and Influences Recurrence Risk of Genomic Disorders. American Journal of Human Genetics, 2014, 95, 173-182.	6.2	219
4	Evaluation of teriparatide treatment in adults with osteogenesis imperfecta. Journal of Clinical Investigation, 2014, 124, 491-498.	8.2	140
5	Argininosuccinate lyase deficiency. Genetics in Medicine, 2012, 14, 501-507.	2.4	83
6	Ammonia control and neurocognitive outcome among urea cycle disorder patients treated with glycerol phenylbutyrate. Hepatology, 2013, 57, 2171-2179.	7.3	83
7	BAFopathiesâ€™ DNA methylation epi-signatures demonstrate diagnostic utility and functional continuum of Coffinâ€“Siris and Nicolaidesâ€“Baraitser syndromes. Nature Communications, 2018, 9, 4885.	12.8	83
8	Nitric-Oxide Supplementation for Treatment of Long-Term Complications in Argininosuccinic Aciduria. American Journal of Human Genetics, 2012, 90, 836-846.	6.2	73
9	Sodium phenylbutyrate decreases plasma branched-chain amino acids in patients with urea cycle disorders. Molecular Genetics and Metabolism, 2014, 113, 131-135.	1.1	58
10	Detection of copy-number variation in AUTS2 gene by targeted exonic array CGH in patients with developmental delay and autistic spectrum disorders. European Journal of Human Genetics, 2013, 21, 343-346.	2.8	56
11	Identification of novel candidate disease genes from de novo exonic copy number variants. Genome Medicine, 2017, 9, 83.	8.2	50
12	Untargeted metabolomic profiling reveals multiple pathway perturbations and new clinical biomarkers in urea cycle disorders. Genetics in Medicine, 2019, 21, 1977-1986.	2.4	47
13	Delineation of candidate genes responsible for structural brain abnormalities in patients with terminal deletions of chromosome 6q27. European Journal of Human Genetics, 2015, 23, 54-60.	2.8	45
14	Nitric oxide modulates bone anabolism through regulation of osteoblast glycolysis and differentiation. Journal of Clinical Investigation, 2021, 131, .	8.2	39
15	A metabolic link between the urea cycle and cancer cell proliferation. Molecular and Cellular Oncology, 2016, 3, e1127314.	0.7	36
16	Acid-Induced Downregulation of ASS1 Contributes to the Maintenance of Intracellular pH in Cancer. Cancer Research, 2019, 79, 518-533.	0.9	36
17	Growth characteristics in individuals with osteogenesis imperfecta in North America: results from a multicenter study. Genetics in Medicine, 2019, 21, 275-283.	2.4	34
18	A multicenter study to evaluate pulmonary function in osteogenesis imperfecta. Clinical Genetics, 2018, 94, 502-511.	2.0	33

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19	A randomized controlled trial to evaluate the effects of high-dose versus low-dose of arginine therapy on hepatic function tests in argininosuccinic aciduria. Molecular Genetics and Metabolism, 2012, 107, 315-321.	1.1	32
20	Early prediction of phenotypic severity in Citrullinemia Type 1. Annals of Clinical and Translational Neurology, 2019, 6, 1858-1871.	3.7	26
21	Targeting TGF-Î² for treatment of osteogenesis imperfecta. Journal of Clinical Investigation, 2022, 132, .	8.2	26
22	A Multicenter Observational Cohort Study to Evaluate the Effects of Bisphosphonate Exposure on Bone Mineral Density and Other Health Outcomes in Osteogenesis Imperfecta. JBMR Plus, 2019, 3, e10118.	2.7	22
23	Small genomic rearrangements involving FMR1 support the importance of its gene dosage for normal neurocognitive function. Neurogenetics, 2012, 13, 333-339.	1.4	21
24	Hearing loss in individuals with osteogenesis imperfecta in North America: Results from a multicenter study. American Journal of Medical Genetics, Part A, 2020, 182, 697-704.	1.2	17
25	Plasma Glutamine Is a Minor Precursor for the Synthesis of Citrulline: A Multispecies Study. Journal of Nutrition, 2017, 147, 549-555.	2.9	16
26	Mobility in osteogenesis imperfecta: a multicenter North American study. Genetics in Medicine, 2019, 21, 2311-2318.	2.4	15
27	Pediatric Outcomes Data Collection Instrument is a Useful Patient-Reported Outcome Measure for Physical Function in Children with Osteogenesis Imperfecta. Genetics in Medicine, 2020, 22, 581-589.	2.4	14
28	Self-reported treatment-associated symptoms among patients with urea cycle disorders participating in glycerol phenylbutyrate clinical trials. Molecular Genetics and Metabolism, 2015, 116, 29-34.	1.1	12
29	Serum Sclerostin Levels in Adults With Osteogenesis Imperfecta: Comparison With Normal Individuals and Response to Teriparatide Therapy. Journal of Bone and Mineral Research, 2018, 33, 307-315.	2.8	12
30	Pregnancy in women with osteogenesis imperfecta: pregnancy characteristics, maternal, and neonatal outcomes. American Journal of Obstetrics & Gynecology MFM, 2021, 3, 100362.	2.6	11
31	A GMR-based assay for quantification of the human response to influenza. Biosensors and Bioelectronics, 2022, 205, 114086.	10.1	11
32	The practice of adult genetics: A 7â€year experience from a single center. American Journal of Medical Genetics, Part A, 2013, 161, 89-93.	1.2	10
33	ASL expression in ALDH1A1+ neurons in the substantia nigra metabolically contributes to neurodegenerative phenotype. Human Genetics, 2021, 140, 1471-1485.	3.8	10
34	A randomized trial to study the comparative efficacy of phenylbutyrate and benzoate on nitrogen excretion and ureagenesis in healthy volunteers. Genetics in Medicine, 2018, 20, 708-716.	2.4	8
35	A Multicenter Study of Intramedullary Rodding in Osteogenesis Imperfecta. JBJS Open Access, 2020, 5, e20.00031-e20.00031.	1.5	7
36	Adult presentation of Xâ€linked Conradiâ€HÃ¼nermannâ€Happle syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 1309-1314.	1.2	6

#	Article	IF	Citations
37	Alterations of a serum marker of collagen X in growing children with osteogenesis imperfecta. Bone, 2021, 149, 115990.	2.9	6
38	Neuroimaging findings of extensive sphenoethmoidal dysplasia in NF1. Clinical Imaging, 2018, 51, 160-163.	1.5	4
39	Widespread disturbance in extracellular matrix collagen biomarker responses to teriparatide therapy in osteogenesis imperfecta. Bone, 2021, 142, 115703.	2.9	4
40	<scp>Healthâ€related</scp> quality of life in adults with osteogenesis imperfecta. Clinical Genetics, 2021, 99, 772-779.	2.0	4
41	Response to Srilatha et al.. Genetics in Medicine, 2012, 14, 627-628.	2.4	0
42	Skeletal disorders. , 2020, , 369-379.		0