Sandesh C S Nagamani

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/870139/publications.pdf

Version: 2024-02-01

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	Targeting TGF-Î² for treatment of osteogenesis imperfecta. Journal of Clinical Investigation, 2022, 132, .	8.2	26
2	A GMR-based assay for quantification of the human response to influenza. Biosensors and Bioelectronics, 2022, 205, 114086.	10.1	11
3	Widespread disturbance in extracellular matrix collagen biomarker responses to teriparatide therapy in osteogenesis imperfecta. Bone, 2021, 142, 115703.	2.9	4
4	<scp>Healthâ€related</scp> quality of life in adults with osteogenesis imperfecta. Clinical Genetics, 2021, 99, 772-779.	2.0	4
5	Nitric oxide modulates bone anabolism through regulation of osteoblast glycolysis and differentiation. Journal of Clinical Investigation, 2021, 131, .	8.2	39
6	Pregnancy in women with osteogenesis imperfecta: pregnancy characteristics, maternal, and neonatal outcomes. American Journal of Obstetrics & Gynecology MFM, 2021, 3, 100362.	2.6	11
7	Alterations of a serum marker of collagen X in growing children with osteogenesis imperfecta. Bone, 2021, 149, 115990.	2.9	6
8	ASL expression in ALDH1A1+ neurons in the substantia nigra metabolically contributes to neurodegenerative phenotype. Human Genetics, 2021, 140, 1471-1485.	3.8	10
9	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
10	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
11	A Multicenter Study of Intramedullary Rodding in Osteogenesis Imperfecta. JBJS Open Access, 2020, 5, e20.00031-e20.00031.	1.5	7
12	Skeletal disorders. , 2020, , 369-379.		0
13	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
14	Early prediction of phenotypic severity in Citrullinemia Type 1. Annals of Clinical and Translational Neurology, 2019, 6, 1858-1871.	3.7	26
15	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
16	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
17	Mobility in osteogenesis imperfecta: a multicenter North American study. Genetics in Medicine, 2019, 21, 2311-2318.	2.4	15
18	Acid-Induced Downregulation of ASS1 Contributes to the Maintenance of Intracellular pH in Cancer. Cancer Research, 2019, 79, 518-533.	0.9	36

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19	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
20	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
21	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
22	A multicenter study to evaluate pulmonary function in osteogenesis imperfecta. Clinical Genetics, 2018, 94, 502-511.	2.0	33
23	Neuroimaging findings of extensive sphenoethmoidal dysplasia in NF1. Clinical Imaging, 2018, 51, 160-163.	1.5	4
24	Plasma Glutamine Is a Minor Precursor for the Synthesis of Citrulline: A Multispecies Study. Journal of Nutrition, 2017, 147, 549-555.	2.9	16
25	Identification of novel candidate disease genes from de novo exonic copy number variants. Genome Medicine, 2017, 9, 83.	8.2	50
26	A metabolic link between the urea cycle and cancer cell proliferation. Molecular and Cellular Oncology, 2016, 3, e1127314.	0.7	36
27	Adult presentation of Xâ€linked Conradiâ€HÃ¼nermannâ€Happle syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 1309-1314.	1.2	6
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	Diversion of aspartate in ASS1-deficient tumours fosters de novo pyrimidine synthesis. Nature, 2015, 527, 379-383.	27.8	271
30	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
31	Branched-chain amino acid metabolism: from rare Mendelian diseases to more common disorders. Human Molecular Genetics, 2014, 23, R1-R8.	2.9	234
32	Parental Somatic Mosaicism Is Underrecognized and Influences Recurrence Risk of Genomic Disorders. American Journal of Human Genetics, 2014, 95, 173-182.	6.2	219
33	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
34	Evaluation of teriparatide treatment in adults with osteogenesis imperfecta. Journal of Clinical Investigation, 2014, 124, 491-498.	8.2	140
35	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
36	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

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37	Ammonia control and neurocognitive outcome among urea cycle disorder patients treated with glycerol phenylbutyrate. Hepatology, 2013, 57, 2171-2179.	7.3	83
38	Argininosuccinate lyase deficiency. Genetics in Medicine, 2012, 14, 501-507.	2.4	83
39	Response to Srilatha et al.. Genetics in Medicine, 2012, 14, 627-628.	2.4	0
40	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
41	Small genomic rearrangements involving FMR1 support the importance of its gene dosage for normal neurocognitive function. Neurogenetics, 2012, 13, 333-339.	1.4	21
42	Nitric-Oxide Supplementation for Treatment of Long-Term Complications in Argininosuccinic Aciduria. American Journal of Human Genetics, 2012, 90, 836-846.	6.2	73