Ronit Marom

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/8868170/publications.pdf

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26 papers	1,143 citations	430874 18 h-index	580821 25 g-index
29 all docs	29 docs citations	29 times ranked	2414 citing authors

#	Article	IF	Citations
1	Characterization of adhesion and differentiation markers of osteogenic marrow stromal cells. Journal of Cellular Physiology, 2005, 202, 41-48.	4.1	219
2	Management of Endocrine Disease: Osteogenesis imperfecta: an update on clinical features and therapies. European Journal of Endocrinology, 2020, 183, R95-R106.	3.7	104
3	Loss of tubulin deglutamylase <scp>CCP</scp> 1 causes infantileâ€onset neurodegeneration. EMBO Journal, 2018, 37, .	7.8	86
4	Osteogenesis imperfecta: advancements in genetics and treatment. Current Opinion in Pediatrics, 2019, 31, 708-715.	2.0	84
5	Disrupted nitric oxide signaling due to <scp>GUCY1A3</scp> mutations increases risk for moyamoya disease, achalasia and hypertension. Clinical Genetics, 2016, 90, 351-360.	2.0	62
6	Newborn screening: a review of history, recent advancements, and future perspectives in the era of next generation sequencing. Current Opinion in Pediatrics, 2016, 28, 694-699.	2.0	59
7	Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay. American Journal of Human Genetics, 2019, 104, 164-178.	6.2	59
8	De Novo Missense Variants in TRAF7 Cause Developmental Delay, Congenital Anomalies, and Dysmorphic Features. American Journal of Human Genetics, 2018, 103, 154-162.	6.2	56
9	Pharmacological and biological therapeutic strategies for osteogenesis imperfecta. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2016, 172, 367-383.	1.6	53
10	Identification of novel candidate disease genes from de novo exonic copy number variants. Genome Medicine, 2017, 9, 83.	8.2	50
11	Identification of cultured progenitor cells from human marrow stroma. Journal of Cellular Biochemistry, 2002, 87, 51-57.	2.6	43
12	A Transgenic Mouse Model of OI Type V Supports a Neomorphic Mechanism of the IFITM5 Mutation. Journal of Bone and Mineral Research, 2015, 30, 489-498.	2.8	30
13	Expression and regulation of CReMM, a chromodomain helicase-DNA-binding (CHD), in marrow stroma derived osteoprogenitors. Journal of Cellular Physiology, 2006, 207, 628-635.	4.1	28
14	Variants in MED12L, encoding a subunit of the mediator kinase module, are responsible for intellectual disability associated with transcriptional defect. Genetics in Medicine, 2019, 21, 2713-2722.	2.4	28
15	Heterozygous variants inACTL6A, encoding a component of the BAF complex, are associated with intellectual disability. Human Mutation, 2017, 38, 1365-1371.	2.5	27
16	Recurrent arginine substitutions in the ACTG2 gene are the primary driver of disease burden and severity in visceral myopathy. Human Mutation, 2020, 41, 641-654.	2.5	27
17	Milder clinical and biochemical phenotypes associated with the c.482G > A (p.Arg161Gln) pathogenic variant in cobalamin C disease: Implications for management and screening. Molecular Genetics and Metabolism, 2017, 122, 60-66.	1.1	20
18	Biochemical signatures mimicking multiple carboxylase deficiency in children with mutations in MT-ATP6. Mitochondrion, 2019, 44, 58-64.	3.4	19

#	Article	IF	Citations
19	COPB2 loss of function causes a coatopathy with osteoporosis and developmental delay. American Journal of Human Genetics, 2021, 108, 1710-1724.	6.2	18
20	Neonatal fractures as a presenting feature of LMOD3â€associated congenital myopathy. American Journal of Medical Genetics, Part A, 2017, 173, 2789-2794.	1.2	17
21	A global Slc7a7 knockout mouse model demonstrates characteristic phenotypes of human lysinuric protein intolerance. Human Molecular Genetics, 2020, 29, 2171-2184.	2.9	15
22	New Face for Chromatinâ€Related Mesenchymal Modulator: nâ€CHD9 Localizes to Nucleoli and Interacts With Ribosomal Genes. Journal of Cellular Physiology, 2015, 230, 2270-2280.	4.1	14
23	Retrospective analysis of a clinical exome sequencing cohort reveals the mutational spectrum and identifies candidate diseaseâ€“associated loci for BAFopathies. Genetics in Medicine, 2022, 24, 364-373.	2.4	12
24	Unintentional Oral Beta Agonist Overdose. American Journal of Therapeutics, 2013, 20, 311-314.	0.9	8
25	A novel de novo intronic variant in ITPR1 causes Gillespie syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 2315-2324.	1.2	2
26	A Somnolent Neonate With Hypothermia and Posturing. Clinical Pediatrics, 2020, 59, 841-843.	0.8	0