

Ronit Marom

List of Publications by Year in descending order

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Version: 2024-02-01

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	Retrospective analysis of a clinical exome sequencing cohort reveals the mutational spectrum and identifies candidate disease-associated loci for BAFopathies. <i>Genetics in Medicine</i> , 2022, 24, 364-373.	2.4	12
2	A novel de novo intronic variant in <i>ITPR1</i> causes Gillespie syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2315-2324.	1.2	2
3	<i>COPB2</i> loss of function causes a coatopathy with osteoporosis and developmental delay. <i>American Journal of Human Genetics</i> , 2021, 108, 1710-1724.	6.2	18
4	Recurrent arginine substitutions in the <i>ACTG2</i> gene are the primary driver of disease burden and severity in visceral myopathy. <i>Human Mutation</i> , 2020, 41, 641-654.	2.5	27
5	A Somnolent Neonate With Hypothermia and Posturing. <i>Clinical Pediatrics</i> , 2020, 59, 841-843.	0.8	0
6	A global <i>Slc7a7</i> knockout mouse model demonstrates characteristic phenotypes of human lysinuric protein intolerance. <i>Human Molecular Genetics</i> , 2020, 29, 2171-2184.	2.9	15
7	Management of Endocrine Disease: Osteogenesis imperfecta: an update on clinical features and therapies. <i>European Journal of Endocrinology</i> , 2020, 183, R95-R106.	3.7	104
8	Variants in <i>MED12L</i> , encoding a subunit of the mediator kinase module, are responsible for intellectual disability associated with transcriptional defect. <i>Genetics in Medicine</i> , 2019, 21, 2713-2722.	2.4	28
9	Osteogenesis imperfecta: advancements in genetics and treatment. <i>Current Opinion in Pediatrics</i> , 2019, 31, 708-715.	2.0	84
10	Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in <i>SMARCC2</i> Cause a Syndrome with Intellectual Disability and Developmental Delay. <i>American Journal of Human Genetics</i> , 2019, 104, 164-178.	6.2	59
11	Biochemical signatures mimicking multiple carboxylase deficiency in children with mutations in <i>MT-ATP6</i> . <i>Mitochondrion</i> , 2019, 44, 58-64.	3.4	19
12	Loss of tubulin deglutamylase <i>CCP1</i> causes infantile-onset neurodegeneration. <i>EMBO Journal</i> , 2018, 37, .	7.8	86
13	De Novo Missense Variants in <i>TRAF7</i> Cause Developmental Delay, Congenital Anomalies, and Dysmorphic Features. <i>American Journal of Human Genetics</i> , 2018, 103, 154-162.	6.2	56
14	Neonatal fractures as a presenting feature of <i>LMOD3</i> -associated congenital myopathy. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2789-2794.	1.2	17
15	Milder clinical and biochemical phenotypes associated with the c.482G > A (p.Arg161Gln) pathogenic variant in cobalamin C disease: Implications for management and screening. <i>Molecular Genetics and Metabolism</i> , 2017, 122, 60-66.	1.1	20
16	Heterozygous variants in <i>ACTL6A</i> , encoding a component of the BAF complex, are associated with intellectual disability. <i>Human Mutation</i> , 2017, 38, 1365-1371.	2.5	27
17	Identification of novel candidate disease genes from de novo exonic copy number variants. <i>Genome Medicine</i> , 2017, 9, 83.	8.2	50
18	Disrupted nitric oxide signaling due to <i>GUCY1A3</i> mutations increases risk for moyamoya disease, achalasia and hypertension. <i>Clinical Genetics</i> , 2016, 90, 351-360.	2.0	62

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19	Newborn screening: a review of history, recent advancements, and future perspectives in the era of next generation sequencing. <i>Current Opinion in Pediatrics</i> , 2016, 28, 694-699.	2.0	59
20	Pharmacological and biological therapeutic strategies for osteogenesis imperfecta. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2016, 172, 367-383.	1.6	53
21	A Transgenic Mouse Model of OI Type V Supports a Neomorphic Mechanism of the <i>IFITM5</i> Mutation. <i>Journal of Bone and Mineral Research</i> , 2015, 30, 489-498.	2.8	30
22	New Face for Chromatin-Related Mesenchymal Modulator: <i>CHD9</i> Localizes to Nucleoli and Interacts With Ribosomal Genes. <i>Journal of Cellular Physiology</i> , 2015, 230, 2270-2280.	4.1	14
23	Unintentional Oral Beta Agonist Overdose. <i>American Journal of Therapeutics</i> , 2013, 20, 311-314.	0.9	8
24	Expression and regulation of CReMM, a chromodomain helicase-DNA-binding (CHD), in marrow stroma derived osteoprogenitors. <i>Journal of Cellular Physiology</i> , 2006, 207, 628-635.	4.1	28
25	Characterization of adhesion and differentiation markers of osteogenic marrow stromal cells. <i>Journal of Cellular Physiology</i> , 2005, 202, 41-48.	4.1	219
26	Identification of cultured progenitor cells from human marrow stroma. <i>Journal of Cellular Biochemistry</i> , 2002, 87, 51-57.	2.6	43