

Robert Meyer

List of Publications by Year in descending order

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Version: 2024-02-01

12
papers

123
citations

1163117

8
h-index

1281871

11
g-index

13
all docs

13
docs citations

13
times ranked

275
citing authors

#	ARTICLE	IF	CITATIONS
1	C2orf69 mutations disrupt mitochondrial function and cause a multisystem human disorder with recurring autoinflammation. <i>Journal of Clinical Investigation</i> , 2021, 131, .	8.2	13
2	Aggressive infantile myofibromatosis with intestinal involvement. <i>Molecular and Cellular Pediatrics</i> , 2021, 8, 7.	1.8	2
3	Germline variants in DNA repair genes, including <i>BRCA1</i> and <i>BRCA2</i> , may cause familial myeloproliferative neoplasms. <i>Blood Advances</i> , 2021, 5, 3373-3376.	5.2	7
4	One test for all: whole exome sequencing significantly improves the diagnostic yield in growth retarded patients referred for molecular testing for Silver-Russell syndrome. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 42.	2.7	12
5	Loss of supervillin causes myopathy with myofibrillar disorganization and autophagic vacuoles. <i>Brain</i> , 2020, 143, 2406-2420.	7.6	15
6	Inherited cases of <i>CNOT3</i> associated intellectual developmental disorder with speech delay, autism, and dysmorphic facies. <i>Clinical Genetics</i> , 2020, 98, 408-412.	2.0	9
7	Patient with an autosomal recessive <i>MBTPS1</i> linked phenotype and clinical features of Silver-Russell syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2727-2730.	1.2	10
8	<i>HMGA2</i> Variants in Silver-Russell Syndrome: Homozygous and Heterozygous Occurrence. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2020, 105, 2401-2407.	3.6	11
9	Die GfH-JuniorAkademie 2020 – virtueller Vorreiter in der GfH. <i>Medizinische Genetik</i> , 2020, 32, 347-348.	0.2	0
10	Next generation sequencing and imprinting disorders: Current applications and future perspectives: Lessons from Silver-Russell syndrome. <i>Molecular and Cellular Probes</i> , 2019, 44, 1-7.	2.1	11
11	NLRP genes and their role in preeclampsia and multi-locus imprinting disorders. <i>Journal of Perinatal Medicine</i> , 2018, 46, 169-173.	1.4	11
12	Targeted Next Generation Sequencing Approach in Patients Referred for Silver-Russell Syndrome Testing Increases the Mutation Detection Rate and Provides Decisive Information for Clinical Management. <i>Journal of Pediatrics</i> , 2017, 187, 206-212.e1.	1.8	22