

Ana Maria Fortuna

List of Publications by Year in descending order

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Version: 2024-02-01

9
papers

371
citations

1163117
8
h-index

1474206
9
g-index

10
all docs

10
docs citations

10
times ranked

725
citing authors

#	ARTICLE	IF	CITATIONS
1	Usher syndrome and Nebulin-associated myopathy in a single patient due to variants in MYO7A and NEB. <i>Clinical Case Reports (discontinued)</i> , 2020, 8, 2476-2482.	0.5	0
2	A Novel Domain-Specific Mutation in a Sclerosteosis Patient Suggests a Role of LRP4 as an Anchor for Sclerostin in Human Bone. <i>Journal of Bone and Mineral Research</i> , 2016, 31, 874-881.	2.8	65
3	The Koolen-de Vries syndrome: a phenotypic comparison of patients with a 17q21.31 microdeletion versus a KANSL1 sequence variant. <i>European Journal of Human Genetics</i> , 2016, 24, 652-659.	2.8	108
4	New splicing mutation in the choline kinase beta (CHKB) gene causing a muscular dystrophy detected by whole-exome sequencing. <i>Journal of Human Genetics</i> , 2015, 60, 305-312.	2.3	33
5	Genotype-phenotype correlations in L1 syndrome: a guide for genetic counselling and mutation analysis. <i>Journal of Medical Genetics</i> , 2010, 47, 169-175.	3.2	82
6	Private dysferlin exon skipping mutation (c.5492G>A) with a founder effect reveals further alternative splicing involving exons 49–51. <i>Journal of Human Genetics</i> , 2010, 55, 546-549.	2.3	12
7	A NONSENSE<i>PORCN</i> MUTATION IN SEVERE FOCAL DERMAL HYPOPLASIA WITH NATAL TEETH. <i>Fetal and Pediatric Pathology</i> , 2010, 29, 305-313.	0.7	11
8	Galactosialidosis presenting as nonimmune fetal hydrops: a case report. <i>Prenatal Diagnosis</i> , 2009, 29, 895-896.	2.3	10
9	T Cell Numbers Relate to Bone Involvement in Gaucher Disease. <i>Blood Cells, Molecules, and Diseases</i> , 1999, 25, 130-138.	1.4	47