

Burcu Turkgenç

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/1103998/publications.pdf>

Version: 2024-02-01

8
papers

195
citations

1684188
5
h-index

1720034
7
g-index

8
all docs

8
docs citations

8
times ranked

436
citing authors

#	ARTICLE	IF	CITATIONS
1	Bi-allelic Mutations in M1AP Are a Frequent Cause of Meiotic Arrest and Severely Impaired Spermatogenesis Leading to Male Infertility. <i>American Journal of Human Genetics</i> , 2020, 107, 342-351.	6.2	68
2	Identification and characterization of a novel <i>FBN1</i> gene variant in an extended family with variable clinical phenotype of Marfan syndrome. <i>Connective Tissue Research</i> , 2019, 60, 146-154.	2.3	2
3	A novel homozygous nonsense mutation in CAST associated with PLACK syndrome. <i>Cell and Tissue Research</i> , 2019, 378, 267-277.	2.9	8
4	<i>STUB1</i> polyadenylation signal variant AACAAA does not affect polyadenylation but decreases <i>STUB1</i> translation causing SCAR16. <i>Human Mutation</i> , 2018, 39, 1344-1348.	2.5	11
5	A Novel TBX19 Gene Mutation in a Case of Congenital Isolated Adrenocorticotrophic Hormone Deficiency Presenting with Recurrent Respiratory Tract Infections. <i>Frontiers in Endocrinology</i> , 2017, 8, 64.	3.5	14
6	Homozygous, and compound heterozygous mutation in 3 Turkish family with Jervell and Lange-Nielsen syndrome: case reports. <i>BMC Medical Genetics</i> , 2017, 18, 114.	2.1	0
7	Mutations in the interleukin receptor <i>IL11RA</i> cause autosomal recessive Crouzon-like craniosynostosis. <i>Molecular Genetics & Genomic Medicine</i> , 2013, 1, 223-237.	1.2	70
8	Skewed X inactivation in an X linked nystagmus family resulted from a novel, p.R229G, missense mutation in the FRMD7 gene. <i>British Journal of Ophthalmology</i> , 2008, 92, 135-141.	3.9	22