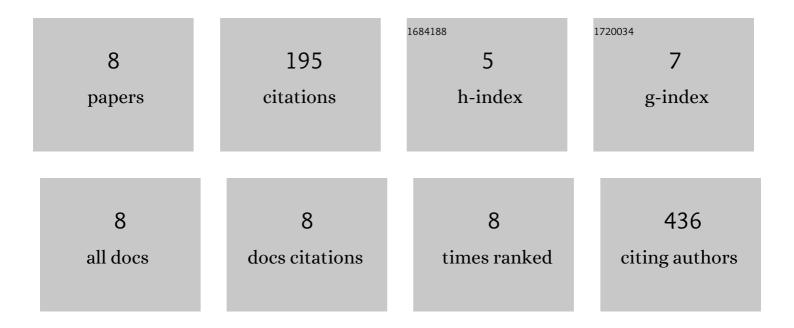
Burcu Turkgenc

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/1103998/publications.pdf Version: 2024-02-01



#	Article	IF	CITATIONS
1	Mutations in the interleukin receptor <i><scp>IL</scp>11<scp>RA</scp></i> cause autosomal recessive Crouzonâ€like craniosynostosis. Molecular Genetics & Genomic Medicine, 2013, 1, 223-237.	1.2	70
2	Bi-allelic Mutations in M1AP Are a Frequent Cause of Meiotic Arrest and Severely Impaired Spermatogenesis Leading to Male Infertility. American Journal of Human Genetics, 2020, 107, 342-351.	6.2	68
3	Skewed X inactivation in an X linked nystagmus family resulted from a novel, p.R229C, missense mutation in the FRMD7 gene. British Journal of Ophthalmology, 2008, 92, 135-141.	3.9	22
4	A Novel TBX19 Gene Mutation in a Case of Congenital Isolated Adrenocorticotropic Hormone Deficiency Presenting with Recurrent Respiratory Tract Infections. Frontiers in Endocrinology, 2017, 8, 64.	3.5	14
5	<i>STUB1</i> polyadenylation signal variant AACAAA does not affect polyadenylation but decreases <i>STUB1</i> translation causing SCAR16. Human Mutation, 2018, 39, 1344-1348.	2.5	11
6	A novel homozygous nonsense mutation in CAST associated with PLACK syndrome. Cell and Tissue Research, 2019, 378, 267-277.	2.9	8
7	Identification and characterization of a novel <i>FBN1</i> gene variant in an extended family with variable clinical phenotype of Marfan syndrome. Connective Tissue Research, 2019, 60, 146-154.	2.3	2
8	"Homozygous, and compound heterozygous mutation in 3 Turkish family with Jervell and Lange-Nielsen syndrome: case reports― BMC Medical Genetics, 2017, 18, 114.	2.1	0