# Burcu Turkgenc

List of Publications by Year
in descending order

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

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|---|---|---|---|
| 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 | Mutations in the interleukin receptor <scp>IL</scp>11<scp>RA</scp> 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.R229G, 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 | STUB1polyadenylation signal variant AACAAA does not affect polyadenylation but decreasesSTUB1translation 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 FBN1 gene variant in an extended family with variable clinical phenotype of Marfan syndrome. Connective Tissue Research, 2019, 60, 146-154. | 2.3 | 2 |
| 8 | â€œHomozygous, and compound heterozygous mutation in 3 Turkish family with Jervell and Lange-Nielsen syndrome: case reportsâ€• BMC Medical Genetics, 2017, 18, 114. | 2.1 | 0 |