Agnieszka Anna Koppolu

List of Publications by Year
in descending order

10 papers	99 citations	1684129 5 h-index	1372553 10 g-index

10 all docs	10 docs citations	10 times ranked	282 citing authors

#	Article	IF	Citations
1	Postzygotic mosaicism of a novel PTPN11 mutation in monozygotic twins discordant for metachondromatosis. American Journal of Medical Genetics, Part A, 2022, , .	1.2	3
2	Epithelial Cells of Deep Infiltrating Endometriosis Harbor Mutations in Cancer Driver Genes. Cells, 2021, 10, 749.	4.1	8
3	Brain Tissue Low-Level Mosaicism for MTOR Mutation Causes Smithâ€“Kingsmore Phenotype with Recurrent Hypoglycemiaâ€”A Novel Phenotype and a Further Proof for Testing of an Affected Tissue. Diagnostics, 2021, 11, 1269.	2.6	4
4	Gene Expression Profile of Human Mesenchymal Stromal Cells Exposed to Hypoxic and Pseudohypoxic Preconditioningâ€”An Analysis by RNA Sequencing. International Journal of Molecular Sciences, 2021, 22, 8160.	4.1	4
5	AP4B1-associated hereditary spastic paraplegia: expansion of phenotypic spectrum related to homozygous p.Thr387fs variant. Journal of Applied Genetics, 2020, 61, 213-218.	1.9	6
6	FARSA mutations mimic phenylalanylâ€tRNA synthetase deficiency caused by FARSB defects. Clinical Genetics, 2019, 96, 468-472.	2.0	22
7	A Novel Monoallelic Nonsense Mutation in the NFKB2 Gene Does Not Cause a Clinical Manifestation. Frontiers in Genetics, 2019, 10, 140.	2.3	8
8	A case of severe trichothiodystrophy 3 in a neonate due to mutation in the GTF2H5 gene: Clinical report. European Journal of Medical Genetics, 2019, 62, 103557.	1.3	5
9	Mapping of breakpoints in balanced chromosomal translocations by shallow whole-genome sequencing points toEFNA5,BAHD1andPPP2R5Eas novel candidates for genes causing human Mendelian disorders. Journal of Medical Genetics, 2019, 56, 104-112.	3.2	13
10	Novel calcineurin A (PPP3CA) variant associated with epilepsy, constitutive enzyme activation and downregulation of protein expression. European Journal of Human Genetics, 2019, 27, 61-69.	2.8	26