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List of Publications by Year in descending order

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Version: 2024-02-01

9
papers

188
citations

1478505

6
h-index

1474206

9
g-index

13
all docs

13
docs citations

13
times ranked

332
citing authors

#	ARTICLE	IF	CITATIONS
1	Establishing the phenotypic spectrum of ZTTK syndrome by analysis of 52 individuals with variants in SON. <i>European Journal of Human Genetics</i> , 2022, 30, 271-281.	2.8	19
2	Phenotype based prediction of exome sequencing outcome using machine learning for neurodevelopmental disorders. <i>Genetics in Medicine</i> , 2022, 24, 645-653.	2.4	6
3	Human disease genes website series: An international, open and dynamic library for up-to-date clinical information. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1039-1046.	1.2	19
4	Comprehensive study of 28 individuals with SIN3A-related disorder underscoring the associated mild cognitive and distinctive facial phenotype. <i>European Journal of Human Genetics</i> , 2021, 29, 625-636.	2.8	17
5	Quantitative facial phenotyping for Koolen-de Vries and 22q11.2 deletion syndrome. <i>European Journal of Human Genetics</i> , 2021, 29, 1418-1423.	2.8	12
6	Mutation-specific pathophysiological mechanisms define different neurodevelopmental disorders associated with SATB1 dysfunction. <i>American Journal of Human Genetics</i> , 2021, 108, 346-356.	6.2	30
7	A Case Series of Familial ARID1B Variants Illustrating Variable Expression and Suggestions to Update the ACMG Criteria. <i>Genes</i> , 2021, 12, 1275.	2.4	5
8	Behavior and cognitive functioning in Witteveen-Kolk syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2384-2390.	1.2	8
9	KAT6A Syndrome: genotype-phenotype correlation in 76 patients with pathogenic KAT6A variants. <i>Genetics in Medicine</i> , 2019, 21, 850-860.	2.4	68