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List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/1221451/publications.pdf>

Version: 2024-02-01

10
papers

200
citations

1307594

7
h-index

1372567

10
g-index

10
all docs

10
docs citations

10
times ranked

280
citing authors

#	ARTICLE	IF	CITATIONS
1	First evidence of <i>SOX2</i> mutations in Peters' anomaly: Lessons from molecular screening of 95 patients. <i>Clinical Genetics</i> , 2022, 101, 494-506.	2.0	9
2	Re-focusing on Agnathia-Otocephaly complex. <i>Clinical Oral Investigations</i> , 2021, 25, 1353-1362.	3.0	7
3	Confirmation of FZD5 implication in a cohort of 50 patients with ocular coloboma. <i>European Journal of Human Genetics</i> , 2021, 29, 131-140.	2.8	10
4	Parental mosaicism in Marfan and Ehlers-Danlos syndromes and related disorders. <i>European Journal of Human Genetics</i> , 2021, 29, 771-779.	2.8	10
5	Clinical and genetic data of 22 new patients with <i>SMAD3</i> pathogenic variants and review of the literature. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1132.	1.2	11
6	Novel PXDN biallelic variants in patients with microphthalmia and anterior segment dysgenesis. <i>Journal of Human Genetics</i> , 2020, 65, 487-491.	2.3	5
7	Genetics of anophthalmia and microphthalmia. Part 1: Non-syndromic anophthalmia/microphthalmia. <i>Human Genetics</i> , 2019, 138, 799-830.	3.8	64
8	<i>FOXE3</i> mutations: genotype-phenotype correlations. <i>Clinical Genetics</i> , 2018, 93, 837-845.	2.0	27
9	Implication of non-coding PAX6 mutations in aniridia. <i>Human Genetics</i> , 2018, 137, 831-846.	3.8	34
10	Genetic Advances in Microphthalmia. <i>Journal of Pediatric Genetics</i> , 2016, 05, 184-188.	0.7	23