

Samuel J Franklin

List of Publications by Year in descending order

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Version: 2024-02-01

6
papers

28
citations

2258059

3
h-index

2272923

4
g-index

6
all docs

6
docs citations

6
times ranked

16
citing authors

#	ARTICLE	IF	CITATIONS
1	Long-read whole genome sequencing reveals HOXD13 alterations in synpolydactyly. <i>Human Mutation</i> , 2022, 43, 189-199.	2.5	7
2	Biallelic SEPSECS variants in two siblings with pontocerebellar hypoplasia type 2D underscore the relevance of splice-disrupting synonymous variants in disease.. <i>Journal of Physical Education and Sports Management</i> , 2022, , mcs.a006165.	1.2	0
3	Case report and review of the literature: immune dysregulation in a large familial cohort due to a novel pathogenic <i>RELA</i> variant. <i>Rheumatology</i> , 2022, 62, 347-359.	1.9	4
4	Hypomorphic alleles pose challenges in rare disease genomic variant interpretation. <i>Clinical Genetics</i> , 2021, 100, 775-776.	2.0	4
5	Maternal mosaicism for a missense variant in the <i>SMS</i> gene that causes Snyder-Robinson syndrome. <i>Journal of Physical Education and Sports Management</i> , 2021, 7, a006122.	1.2	0
6	Discovery of clinically relevant fusions in pediatric cancer. <i>BMC Genomics</i> , 2021, 22, 872.	2.8	13