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List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/4506195/publications.pdf>

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6
papers

73
citations

1937685
4
h-index

1872680
6
g-index

8
all docs

8
docs citations

8
times ranked

110
citing authors

#	ARTICLE	IF	CITATIONS
1	The RUNX1 database (RUNX1db): establishment of an expert curated RUNX1 registry and genomics database as a public resource for familial platelet disorder with myeloid malignancy. <i>Haematologica</i> , 2021, 106, 3004-3007.	3.5	29
2	Paternal mosaicism for a novel <i>PBX1</i> mutation associated with recurrent perinatal death: Phenotypic expansion of the <i>PBX1</i> -related syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1273-1277.	1.2	12
3	A non-coding variant in the 5' UTR of <i>DLC3</i> attenuates protein translation to cause non-syndromic intellectual disability. <i>European Journal of Human Genetics</i> , 2016, 24, 1612-1616.	2.8	10
4	Familial epilepsy with anterior polymicrogyria as a presentation of <i>COL18A1</i> mutations. <i>European Journal of Medical Genetics</i> , 2017, 60, 437-443.	1.3	10
5	OUP accepted manuscript. <i>Human Molecular Genetics</i> , 2021, 30, 2068-2081.	2.9	7
6	Compound heterozygous variants in <i>LAMC3</i> in association with posterior periventricular nodular heterotopia. <i>BMC Medical Genomics</i> , 2021, 14, 64.	1.5	5