

Dorien Schepers

List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

16
papers

1,039
citations

8
h-index

17
g-index

17
ext. papers

1,248
ext. citations

8.6
avg. IF

3.18
L-index

#	Paper	IF	Citations
16	Loss-of-function mutations in TGF β 2 cause a syndromic presentation of thoracic aortic aneurysm. <i>Nature Genetics</i> , 2012 , 44, 922-7	36.3	323
15	Mutations in the TGF β -repressor SKI cause Shprintzen-Goldberg syndrome with aortic aneurysm. <i>Nature Genetics</i> , 2012 , 44, 1249-54	36.3	199
14	Dominant-negative effects of KCNQ2 mutations are associated with epileptic encephalopathy. <i>Annals of Neurology</i> , 2014 , 75, 382-94	9.4	176
13	Differences in manifestations of Marfan syndrome, Ehlers-Danlos syndrome, and Loeys-Dietz syndrome. <i>Annals of Cardiothoracic Surgery</i> , 2017 , 6, 582-594	4.7	110
12	Heterozygous Loss-of-Function SEC61A1 Mutations Cause Autosomal-Dominant Tubulo-Interstitial and Glomerulocystic Kidney Disease with Anemia. <i>American Journal of Human Genetics</i> , 2016 , 99, 174-87 ¹¹	11	87
11	A mutation update on the LDS-associated genes TGF β 2/3 and SMAD2/3. <i>Human Mutation</i> , 2018 , 39, 621-634	11	73
10	The SMAD-binding domain of SKI: a hotspot for de novo mutations causing Shprintzen-Goldberg syndrome. <i>European Journal of Human Genetics</i> , 2015 , 23, 224-8	5.3	42
9	Novel pathogenic variants in five families with arterial aneurysm and dissection: further delineation of the phenotype. <i>Journal of Medical Genetics</i> , 2019 , 56, 220-227	5.8	13
8	Severe Phenotype of Cutis Laxa Type 1B with Antenatal Signs due to a Novel Homozygous Nonsense Mutation in. <i>Molecular Syndromology</i> , 2018 , 9, 190-196	1.5	4
7	iPSC-Cardiomyocyte Models of Brugada Syndrome-Achievements, Challenges and Future Perspectives. <i>International Journal of Molecular Sciences</i> , 2021 , 22,	6.3	4
6	A human importin β -related disorder: Syndromic thoracic aortic aneurysm caused by bi-allelic loss-of-function variants in IPO8. <i>American Journal of Human Genetics</i> , 2021 , 108, 1115-1125	11	4
5	Optical Mapping in hiPSC-CM and Zebrafish to Resolve Cardiac Arrhythmias. <i>Hearts</i> , 2020 , 1, 181-199	0.6	1
4	Compound Heterozygous Mutations in Severe Sodium Channelopathy With Brugada Syndrome: A Case Report. <i>Frontiers in Cardiovascular Medicine</i> , 2020 , 7, 117	5.4	1
3	Cardiogeneticsbank@UZA: A Collection of DNA, Tissues, and Cell Lines as a Translational Tool. <i>Frontiers in Medicine</i> , 2019 , 6, 198	4.9	1
2	Clinical characterization of the first Belgian SCN5A founder mutation cohort. <i>Europace</i> , 2021 , 23, 918-923.	3.9	1
1	Molecular autopsy and subsequent functional analysis reveal de novo DSG2 mutation as cause of sudden death. <i>European Journal of Medical Genetics</i> , 2021 , 64, 104322	2.6	