Dorien Schepers

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/9204540/publications.pdf

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17 papers	1,398 citations	1039406 9 h-index	940134 16 g-index
17 all docs	17 docs citations	17 times ranked	2517 citing authors

#	Article	IF	Citations
1	Loss-of-function mutations in TGFB2 cause a syndromic presentation of thoracic aortic aneurysm. Nature Genetics, 2012, 44, 922-927.	9.4	391
2	Mutations in the TGF-Î² repressor SKI cause Shprintzen-Goldberg syndrome with aortic aneurysm. Nature Genetics, 2012, 44, 1249-1254.	9.4	237
3	Dominantâ€negative effects of KCNQ2 mutations are associated with epileptic encephalopathy. Annals of Neurology, 2014, 75, 382-394.	2.8	225
4	Differences in manifestations of Marfan syndrome, Ehlers-Danlos syndrome, and Loeys-Dietz syndrome. Annals of Cardiothoracic Surgery, 2017, 6, 582-594.	0.6	192
5	Heterozygous Loss-of-Function SEC61A1 Mutations Cause Autosomal-Dominant Tubulo-Interstitial and Glomerulocystic Kidney Disease with Anemia. American Journal of Human Genetics, 2016, 99, 174-187.	2.6	124
6	A mutation update on the LDS-associated genesTGFB2/3andSMAD2/3. Human Mutation, 2018, 39, 621-634.	1.1	116
7	The SMAD-binding domain of SKI: a hotspot for de novo mutations causing Shprintzenâ€“Goldberg syndrome. European Journal of Human Genetics, 2015, 23, 224-228.	1.4	48
8	Novel pathogenicSMAD2variants in five families with arterial aneurysm and dissection: further delineation of the phenotype. Journal of Medical Genetics, 2019, 56, 220-227.	1.5	25
9	iPSC-Cardiomyocyte Models of Brugada Syndromeâ€"Achievements, Challenges and Future Perspectives. International Journal of Molecular Sciences, 2021, 22, 2825.	1.8	13
10	A human importin-Î²-related disorder: Syndromic thoracic aortic aneurysm caused by bi-allelic loss-of-function variants in IPO8. American Journal of Human Genetics, 2021, 108, 1115-1125.	2.6	10
11	Severe Phenotype of Cutis Laxa Type 1B with Antenatal Signs due to a Novel Homozygous Nonsense Mutation in EFEMP2. Molecular Syndromology, 2018, 9, 190-196.	0.3	5
12	Compound Heterozygous SCN5A Mutations in Severe Sodium Channelopathy With Brugada Syndrome: A Case Report. Frontiers in Cardiovascular Medicine, 2020, 7, 117.	1.1	3
13	Clinical characterization of the first Belgian SCN5A founder mutation cohort. Europace, 2021, 23, 918-927.	0.7	3
14	Morpho-functional comparison of differentiation protocols to create iPSC-derived cardiomyocytes. Biology Open, 2022, 11, .	0.6	3
15	Optical Mapping in hiPSC-CM and Zebrafish to Resolve Cardiac Arrhythmias. Hearts, 2020, 1, 181-199.	0.4	2
16	Cardiogeneticsbank@UZA: A Collection of DNA, Tissues, and Cell Lines as a Translational Tool. Frontiers in Medicine, 2019, 6, 198.	1.2	1
17	Molecular autopsy and subsequent functional analysis reveal de novo DSG2 mutation as cause of sudden death. European Journal of Medical Genetics, 2021, 64, 104322.	0.7	0