Dorien Schepers

List of Publications by Year in Descending Order

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	iPSC-Cardiomyocyte Models of Brugada Syndrome-Achievements, Challenges and Future Perspectives. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	4
15	A human importin-β-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	11	4
14	Clinical characterization of the first Belgian SCN5A founder mutation cohort. *Europace*, **2021**, 23, 918-927	5.9	1
13	Molecular autopsy and subsequent functional analysis reveal de novo DSG2 mutation as cause of sudden death. *European Journal of Medical Genetics*, **2021**, 64, 104322	2.6	
12	Optical Mapping in hiPSC-CM and Zebrafish to Resolve Cardiac Arrhythmias. *Hearts*, **2020**, 1, 181-199	0.6	1
11	Compound Heterozygous Mutations in Severe Sodium Channelopathy With Brugada Syndrome: A Case Report. *Frontiers in Cardiovascular Medicine*, **2020**, 7, 117	5.4	1
10	Novel pathogenic variants in five families with arterial aneurysm and dissection: further delineation of the phenotype. *Journal of Medical Genetics*, **2019**, 56, 220-227	5.8	13
9	Cardiogeneticsbank@UZA: A Collection of DNA, Tissues, and Cell Lines as a Translational Tool. *Frontiers in Medicine*, **2019**, 6, 198	4.9	1
8	A mutation update on the LDS-associated genes TGFB2/3 and SMAD2/3. *Human Mutation*, **2018**, 39, 621-634	4.7	73
7	Severe Phenotype of Cutis Laxa Type 1B with Antenatal Signs due to a Novel Homozygous Nonsense Mutation in. *Molecular Syndromology*, **2018**, 9, 190-196	1.5	4
6	Differences in manifestations of Marfan syndrome, Ehlers-Danlos syndrome, and Loeys-Dietz syndrome. *Annals of Cardiothoracic Surgery*, **2017**, 6, 582-594	4.7	110
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, 174-87	11	87
4	The SMAD-binding domain of SKI: a hotspot for de novo mutations causing Shprintzen-Goldberg syndrome. *European Journal of Human Genetics*, **2015**, 23, 224-8	5.3	42
3	Dominant-negative effects of KCNQ2 mutations are associated with epileptic encephalopathy. *Annals of Neurology*, **2014**, 75, 382-94	9.4	176
2	Mutations in the TGF-β repressor SKI cause Shprintzen-Goldberg syndrome with aortic aneurysm. *Nature Genetics*, **2012**, 44, 1249-54	36.3	199
1	Loss-of-function mutations in TGFB2 cause a syndromic presentation of thoracic aortic aneurysm. *Nature Genetics*, **2012**, 44, 922-7	36.3	323