Paul A Van Der Zwaag

List of Publications by Year
in descending order

30	1,481	471509 17	526287 27
papers	citations	h-index	g-index

34	34	34	3212
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Sex-specific aspects of phospholamban cardiomyopathy: The importance and prognostic value of low-voltage electrocardiograms. Heart Rhythm, 2022, 19, 427-434.	0.7	8
2	Validation of New Gene Variant Classification Methods: a Field-Test in Diagnostic Cardiogenetics. Frontiers in Genetics, 2022, 13, 824510.	2.3	1
3	Enhancing rare variant interpretation in inherited arrhythmias through quantitative analysis of consortium disease cohorts and population controls. Genetics in Medicine, 2021, 23, 47-58.	2.4	57
4	The effect of tropomyosin variants on cardiomyocyte function and structure that underlie different clinical cardiomyopathy phenotypes. International Journal of Cardiology, 2021, 323, 251-258.	1.7	8
5	Prediction of ventricular arrhythmia in phospholamban p.Arg14del mutation carriersâ€“reaching the frontiers of individual risk prediction. European Heart Journal, 2021, 42, 2842-2850.	2.2	54
6	Diagnostic yield of targeted next generation sequencing in 2002 Dutch cardiomyopathy patients. International Journal of Cardiology, 2021, 332, 99-104.	1.7	9
7	Coexistence of wild type and hereditary ATTR amyloidosis in one family. Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis, 2020, 27, 71-72.	3.0	0
8	The phospholamban p.(Arg14del) pathogenic variant leads to cardiomyopathy with heart failure and is unresponsive to standard heart failure therapy. Scientific Reports, 2020, 10, 9819.	3.3	38
9	Homozygous damaging SOD2 variant causes lethal neonatal dilated cardiomyopathy. Journal of Medical Genetics, 2020, 57, 23-30.	3.2	16
10	Improving the diagnostic yield of exome- sequencing by predicting geneâ€“phenotype associations using large-scale gene expression analysis. Nature Communications, 2019, 10, 2837.	12.8	107
11	Dyssynchronopathy Can be a Manifestation of Heritable Cardiomyopathy. Circulation Genomic and Precision Medicine, 2019, 12, e002528.	3.6	0
12	Arrhythmogenic Right Ventricular Cardiomyopathy-Associated Desmosomal Variants Are Rarely De Novo. Circulation Genomic and Precision Medicine, 2019, 12, e002467.	3.6	38
13	Relevance of Titin Missense and Non-Frameshifting Insertions/Deletions Variants in Dilated Cardiomyopathy. Scientific Reports, 2019, 9, 4093.	3.3	30
14	Distinct molecular signature of phospholamban p.Arg14del arrhythmogenic cardiomyopathy. Cardiovascular Pathology, 2019, 40, 2-6.	1.6	16
15	Myocardial fibrosis as an early feature in phospholamban p.Arg14del mutation carriers: phenotypic insights from cardiovascular magnetic resonance imaging. European Heart Journal Cardiovascular Imaging, 2019, 20, 92-100.	1.2	48
16	Toward an effective exome-based genetic testing strategy in pediatric dilated cardiomyopathy. Genetics in Medicine, 2018, 20, 1374-1386.	2.4	36
17	A Recurrent De Novo PACS2 Heterozygous Missense Variant Causes Neonatal-Onset Developmental Epileptic Encephalopathy, Facial Dysmorphism, and Cerebellar Dysgenesis. American Journal of Human Genetics, 2018, 102, 995-1007.	6.2	49
18	Mutations in CYB561 Causing a Novel Orthostatic Hypotension Syndrome. Circulation Research, 2018, 122, 846-854.	4.5	22

#	Article	IF	Citations
19	Effect of Ascertainment Bias on Estimates of Patient Mortality in Inherited Cardiac Diseases. Circulation Genomic and Precision Medicine, 2018, 11, e001797.	3.6	10
20	Lack of evidence for a causal role of CALR3 in monogenic cardiomyopathy. European Journal of Human Genetics, 2018, 26, 1603-1610.	2.8	4
21	Missense mutations in the WD40 domain ofAHI1cause non-syndromic retinitis pigmentosa. Journal of Medical Genetics, 2017, 54, 624-632.	3.2	21
22	Phospholamban immunostaining is a highly sensitive and specific method for diagnosing phospholamban p.Arg14del cardiomyopathy. Cardiovascular Pathology, 2017, 30, 23-26.	1.6	17
23	Cardiovascular malformations caused by NOTCH1 mutations do not keep left: data on 428 probands with left-sided CHD and their families. Genetics in Medicine, 2016, 18, 914-923.	2.4	104
24	Biallelic Truncating Mutations in ALPK3 Cause Severe Pediatric Cardiomyopathy. Journal of the American College of Cardiology, 2016, 67, 515-525.	2.8	70
25	The TMEM43 Newfoundland mutation p.S358L causing ARVC-5 was imported from Europe and increases the stiffness of the cell nucleus. European Heart Journal, 2015, 36, 872-881.	2.2	56
26	Outcome in Phospholamban R14del Carriers. Circulation: Cardiovascular Genetics, 2014, 7, 455-465.	5.1	146
27	Phospholamban R14del mutation in patients diagnosed with dilated cardiomyopathy or arrhythmogenic right ventricular cardiomyopathy: evidence supporting the concept of arrhythmogenic cardiomyopathy. European Journal of Heart Failure, 2012, 14, 1199-1207.	7.1	369
28	An interstitial duplication of chromosome 13q31.3q32.1 further delineates the critical region for postaxial polydactyly type A2. European Journal of Medical Genetics, 2010, 53, 45-49.	1.3	27
29	A genetic variants database for arrhythmogenic right ventricular dysplasia/cardiomyopathy. Human Mutation, 2009, 30, 1278-1283.	2.5	105
30	Abstract 2726: Haplotype Sharing Test as a Tool to Map Genes for Familial Cardiomyopathy. Circulation, 2007, 116, .	1.6	0