Jan D H Jongbloed

List of Publications by Year in descending order

46 papers	2,914 citations	236612 25 h-index	233125 45 g-index
49 all docs	49 docs citations	49 times ranked	5646 citing authors

#	Article	IF	Citations
1	Impact of genotype on clinical course in arrhythmogenic right ventricular dysplasia/cardiomyopathy-associated mutation carriers. European Heart Journal, 2015, 36, 847-855.	1.0	338
2	Adaptation and validation of the ACMG/AMP variant classification framework for MYH7-associated inherited cardiomyopathies: recommendations by ClinGenâ€™s Inherited Cardiomyopathy Expert Panel. Genetics in Medicine, 2018, 20, 351-359.	1.1	283
3	Genetics, Clinical Features, and Long-TermÂOutcome of NoncompactionÂCardiomyopathy. Journal of the American College of Cardiology, 2018, 71, 711-722.	1.2	242
4	A small gene, designated comS, located within the coding region of the fourth amino acid-activation domain of srfA, is required for competence development in Bacillus subtilis. Molecular Microbiology, 1995, 15, 55-63.	1.2	165
5	Titin gene mutations are common in families with both peripartum cardiomyopathy and dilated cardiomyopathy. European Heart Journal, 2014, 35, 2165-2173.	1.0	159
6	Outcome in Phospholamban R14del Carriers. Circulation: Cardiovascular Genetics, 2014, 7, 455-465.	5.1	146
7	Truncating titin mutations are associated with a mild and treatable form of dilated cardiomyopathy. European Journal of Heart Failure, 2017, 19, 512-521.	2.9	127
8	International Evidence Based Reappraisal of Genes Associated With Arrhythmogenic Right Ventricular Cardiomyopathy Using the Clinical Genome Resource Framework. Circulation Genomic and Precision Medicine, 2021, 14, e003273.	1.6	112
9	Improving the diagnostic yield of exome- sequencing by predicting geneâ€“phenotype associations using large-scale gene expression analysis. Nature Communications, 2019, 10, 2837.	5.8	107
10	Rapid Targeted Genomics in Critically Ill Newborns. Pediatrics, 2017, 140, .	1.0	99
11	Sudden Cardiac Death Prediction in Arrhythmogenic Right Ventricular Cardiomyopathy. Circulation: Arrhythmia and Electrophysiology, 2021, 14, e008509.	2.1	82
12	CoNVaDING: Single Exon Variation Detection in Targeted NGS Data. Human Mutation, 2016, 37, 457-464.	1.1	79
13	Biallelic Truncating Mutations in ALPK3 Cause Severe Pediatric Cardiomyopathy. Journal of the American College of Cardiology, 2016, 67, 515-525.	1.2	70
14	Next-generation sequencing-based genome diagnostics across clinical genetics centers: implementation choices and their effects. European Journal of Human Genetics, 2015, 23, 1142-1150.	1.4	56
15	Isolated Subepicardial Right Ventricular Outflow Tract Scar in Athletes With VentricularÂTachycardia. Journal of the American College of Cardiology, 2017, 69, 497-507.	1.2	56
16	Dynamic loading of human engineered heart tissue enhances contractile function and drives a desmosome-linked disease phenotype. Science Translational Medicine, 2021, 13, .	5.8	48
17	Potential genetic predisposition for anthracycline-associated cardiomyopathy in families with dilated cardiomyopathy. Open Heart, 2014, 1, e000116.	0.9	40
18	Evaluation of CADD Scores in Curated Mismatch Repair Gene Variants Yields a Model for Clinical Validation and Prioritization. Human Mutation, 2015, 36, 712-719.	1.1	39

#	Article	IF	Citations
19	Arrhythmogenic Right Ventricular Cardiomyopathy-Associated Desmosomal Variants Are Rarely De Novo. Circulation Genomic and Precision Medicine, 2019, 12, e002467.	1.6	38
20	Toward an effective exome-based genetic testing strategy in pediatric dilated cardiomyopathy. Genetics in Medicine, 2018, 20, 1374-1386.	1.1	36
21	Large Genomic Rearrangements of Desmosomal Genes in Italian Arrhythmogenic Cardiomyopathy Patients. Circulation: Arrhythmia and Electrophysiology, 2017, 10, .	2.1	35
22	A new prediction model for ventricular arrhythmias in arrhythmogenic right ventricular cardiomyopathy. European Heart Journal, 2022, 43, e1-e9.	1.0	35
23	Plectin Mutations Underlie Epidermolysis Bullosa Simplex in 8% of Patients. Journal of Investigative Dermatology, 2014, 134, 273-276.	0.3	34
24	Relevance of Titin Missense and Non-Frameshifting Insertions/Deletions Variants in Dilated Cardiomyopathy. Scientific Reports, 2019, 9, 4093.	1.6	30
25	Expanding the clinical and genetic spectrum of ALPK3 variants: Phenotypes identified in pediatric cardiomyopathy patients and adults with heterozygous variants. American Heart Journal, 2020, 225, 108-119.	1.2	25
26	Heritability in a SCN5A -mutation founder population with increased female susceptibility to non-nocturnal ventricular tachyarrhythmia and sudden cardiac death. Heart Rhythm, 2017, 14, 1873-1881.	0.3	23
27	Mutations in CYB561 Causing a Novel Orthostatic Hypotension Syndrome. Circulation Research, 2018, 122, 846-854.	2.0	22
28	Phospholamban immunostaining is a highly sensitive and specific method for diagnosing phospholamban p.Arg14del cardiomyopathy. Cardiovascular Pathology, 2017, 30, 23-26.	0.7	17
29	Novel SPEG mutations in congenital myopathies: Genotypeâ€“phenotype correlations. Muscle and Nerve, 2019, 59, 357-362.	1.0	17
30	The first titin (c.59926 + 1G > A) founder mutation associated with dilated cardiomyopathy. European Journal of Heart Failure, 2018, 20, 803-806.	2.9	16
31	Distinct molecular signature of phospholamban p.Arg14del arrhythmogenic cardiomyopathy. Cardiovascular Pathology, 2019, 40, 2-6.	0.7	16
32	Prevalence and Prognostic Impact of Pathogenic Variants in Patients With Dilated Cardiomyopathy Referred for Ventricular Tachycardia Ablation. JACC: Clinical Electrophysiology, 2020, 6, 1103-1114.	1.3	16
33	Homozygous damaging SOD2 variant causes lethal neonatal dilated cardiomyopathy. Journal of Medical Genetics, 2020, 57, 23-30.	1.5	16
34	Identification of sarcomeric variants in probands with a clinical diagnosis of arrhythmogenic right ventricular cardiomyopathy (ARVC). Journal of Cardiovascular Electrophysiology, 2018, 29, 1004-1009.	0.8	15
35	Functional assessment of potential splice site variants in arrhythmogenic right ventricular dysplasia/cardiomyopathy. Heart Rhythm, 2014, 11, 2010-2017.	0.3	13
36	The genetic architecture of Plakophilin 2 cardiomyopathy. Genetics in Medicine, 2021, 23, 1961-1968.	1.1	13

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37	Diagnostic yield of targeted next generation sequencing in 2002 Dutch cardiomyopathy patients. International Journal of Cardiology, 2021, 332, 99-104.	0.8	9
38	The effect of tropomyosin variants on cardiomyocyte function and structure that underlie different clinical cardiomyopathy phenotypes. International Journal of Cardiology, 2021, 323, 251-258.	0.8	8
39	Three female patients with Danon disease presenting with predominant cardiac phenotype: a case series. European Heart Journal - Case Reports, 2019, 3, ytz132.	0.3	6
40	Mortality Risk Associated With Truncating Founder Mutations in Titin. Circulation Genomic and Precision Medicine, 2019, 12, e002436.	1.6	5
41	Lack of evidence for a causal role of CALR3 in monogenic cardiomyopathy. European Journal of Human Genetics, 2018, 26, 1603-1610.	1.4	4
42	Case series, Chemotherapy-induced cardiomyopathy: mind the family history!. European Heart Journal - Case Reports, 2021, 5, ytab333.	0.3	4
43	Functional investigation of two simultaneous or separately segregating DSP variants within a single family supports the theory of a doseâ€dependent disease severity. Experimental Dermatology, 2022, , .	1.4	3
44	No major role for rare plectin variants in arrhythmogenic right ventricular cardiomyopathy. PLoS ONE, 2018, 13, e0203078.	1.1	2
45	Validation of New Gene Variant Classification Methods: a Field-Test in Diagnostic Cardiogenetics. Frontiers in Genetics, 2022, 13, 824510.	1.1	1
46	Dyssynchronopathy Can be a Manifestation of Heritable Cardiomyopathy. Circulation Genomic and Precision Medicine, 2019, 12, e002528.	1.6	0