Connie R Bezzina

List of Publications by Year
in descending order

206 papers	15,994 citations	14655 66 h-index	18647 119 g-index
215 all docs	215 docs citations	215 times ranked	13765 citing authors

#	Article	IF	Citations
1	A Single Na ⁺ Channel Mutation Causing Both Long-QT and Brugada Syndromes. Circulation Research, 1999, 85, 1206-1213.	4.5	612
2	Mutation in the KCNQ1 Gene Leading to the Short QT-Interval Syndrome. Circulation, 2004, 109, 2394-2397.	1.6	603
3	Common variants at SCN5A-SCN10A and HEY2 are associated with Brugada syndrome, a rare disease with high risk of sudden cardiac death. Nature Genetics, 2013, 45, 1044-1049.	21.4	467
4	Right Ventricular Fibrosis and Conduction Delay in a Patient With Clinical Signs of Brugada Syndrome. Circulation, 2005, 112, 2769-2777.	1.6	401
5	Sodium channel Î²1 subunit mutations associated with Brugada syndrome and cardiac conduction disease in humans. Journal of Clinical Investigation, 2008, 118, 2260-8.	8.2	400
6	A sodium-channel mutation causes isolated cardiac conduction disease. Nature, 2001, 409, 1043-1047.	27.8	377
7	Genotype-phenotype relationship in Brugada syndrome: electrocardiographic features differentiate SCN5A-related patients from nonâ€“SCN5A-related patients. Journal of the American College of Cardiology, 2002, 40, 350-356.	2.8	360
8	Genetic association study of QT interval highlights role for calcium signaling pathways in myocardial repolarization. Nature Genetics, 2014, 46, 826-836.	21.4	281
9	Two Distinct Congenital Arrhythmias Evoked by a Multidysfunctional Na ⁺ Channel. Circulation Research, 2000, 86, E91-7.	4.5	279
10	A Cardiac Sodium Channel Mutation Cosegregates With a Rare Connexin40 Genotype in Familial Atrial Standstill. Circulation Research, 2003, 92, 14-22.	4.5	261
11	Familial Sudden Death Is an Important Risk Factor for Primary Ventricular Fibrillation. Circulation, 2006, 114, 1140-1145.	1.6	258
12	Genetic variation in SCN10A influences cardiac conduction. Nature Genetics, 2010, 42, 149-152.	21.4	248
13	Cardiomyocytes Derived From Pluripotent Stem Cells Recapitulate Electrophysiological Characteristics of an Overlap Syndrome of Cardiac Sodium Channel Disease. Circulation, 2012, 125, 3079-3091.	1.6	245
14	Large-Scale Gene-Centric Meta-analysis across 32 Studies Identifies Multiple Lipid Loci. American Journal of Human Genetics, 2012, 91, 823-838.	6.2	227
15	Type of SCN5A mutation determines clinical severity and degree of conduction slowing in loss-of-function sodium channelopathies. Heart Rhythm, 2009, 6, 341-348.	0.7	224
16	Compound Heterozygosity for Mutations (W156X and R225W) inSCN5AAssociated With Severe Cardiac Conduction Disturbances and Degenerative Changes in the Conduction System. Circulation Research, 2003, 92, 159-168.	4.5	222
17	Utility of Post-Mortem Genetic Testing in Cases of Sudden Arrhythmic Death Syndrome. Journal of the American College of Cardiology, 2017, 69, 2134-2145.	2.8	219
18	Common Sodium Channel Promoter Haplotype in Asian Subjects Underlies Variability in Cardiac Conduction. Circulation, 2006, 113, 338-344.	1.6	215

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19	Genetics of Sudden Cardiac Death. Circulation Research, 2015, 116, 1919-1936.	4.5	211
20	A mutation in the human cardiac sodium channel (E161K) contributes to sick sinus syndrome, conduction disease and Brugada syndrome in two families. Journal of Molecular and Cellular Cardiology, 2005, 38, 969-981.	1.9	184
21	Myocyte necrosis underlies progressive myocardial dystrophy in mouse dsg2-related arrhythmogenic right ventricular cardiomyopathy. Journal of Experimental Medicine, 2009, 206, 1787-1802.	8.5	184
22	Cardiac Sodium Channel Overlap Syndromes: Different Faces of SCN5A Mutations. Trends in Cardiovascular Medicine, 2008, 18, 78-87.	4.9	182
23	Intercalated disc abnormalities, reduced Na+ current density, and conduction slowing in desmoglein-2 mutant mice prior to cardiomyopathic changes. Cardiovascular Research, 2012, 95, 409-418.	3.8	180
24	Overlap Syndrome of Cardiac Sodium Channel Disease in Mice Carrying the Equivalent Mutation of Human SCN5A -1795insD. Circulation, 2006, 114, 2584-2594.	1.6	174
25	HCN4 Mutations in Multiple Families With Bradycardia and Left Ventricular Noncompaction Cardiomyopathy. Journal of the American College of Cardiology, 2014, 64, 745-756.	2.8	173
26	Genome-wide association study identifies a susceptibility locus at 21q21 for ventricular fibrillation in acute myocardial infarction. Nature Genetics, 2010, 42, 688-691.	21.4	170
27	Induced pluripotent stem cell derived cardiomyocytes as models for cardiac arrhythmias. Frontiers in Physiology, 2012, 3, 346.	2.8	168
28	A common genetic variant within SCN10A modulates cardiac SCN5A expression. Journal of Clinical Investigation, 2014, 124, 1844-1852.	8.2	168
29	Genetic variation in T-box binding element functionally affects SCN5A/SCN10A enhancer. Journal of Clinical Investigation, 2012, 122, 2519-2530.	8.2	167
30	Common genetic variants and modifiable risk factors underpin hypertrophic cardiomyopathy susceptibility and expressivity. Nature Genetics, 2021, 53, 135-142.	21.4	165
31	Clinical Aspects of Type 3 Long-QT Syndrome. Circulation, 2016, 134, 872-882.	1.6	162
32	SARS-CoV-2, COVID-19, and inherited arrhythmia syndromes. Heart Rhythm, 2020, 17, 1456-1462.	0.7	162
33	A Mutation in CALM1 Encoding Calmodulin in Familial Idiopathic Ventricular Fibrillation in Childhood and Adolescence. Journal of the American College of Cardiology, 2014, 63, 259-266.	2.8	160
34	Haplotype-Sharing Analysis Implicates Chromosome 7q36 Harboring DPP6 in Familial IdiopathicÂVentricular Fibrillation. American Journal of Human Genetics, 2009, 84, 468-476.	6.2	158
35	Gene-centric Meta-analysis in 87,736 Individuals of European Ancestry Identifies Multiple Blood-Pressure-Related Loci. American Journal of Human Genetics, 2014, 94, 349-360.	6.2	158
36	Genetic control of sodium channel function. Cardiovascular Research, 2003, 57, 961-973.	3.8	157

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37	A common polymorphism in KCNH2 (HERG) hastens cardiac repolarization. Cardiovascular Research, 2003, 59, 27-36.	3.8	156
38	Shared genetic pathways contribute to risk of hypertrophic and dilated cardiomyopathies with opposite directions of effect. Nature Genetics, 2021, 53, 128-134.	21.4	155
39	A Large Candidate Gene Survey Identifies the KCNE1 D85N Polymorphism as a Possible Modulator of Drug-Induced Torsades de Pointes. Circulation: Cardiovascular Genetics, 2012, 5, 91-99.	5.1	150
40	Multilevel analyses of SCN5A mutations in arrhythmogenic right ventricular dysplasia/cardiomyopathy suggest non-canonical mechanisms for disease pathogenesis. Cardiovascular Research, 2017, 113, 102-111.	3.8	148
41	Inherited cardiac arrhythmias. Nature Reviews Disease Primers, 2020, 6, 58.	30.5	146
42	Possible Bradycardic Mode of Death and Successful Pacemaker Treatment in a Large Family with Features of Long QT Syndrome Type 3 and Brugada Syndrome. Journal of Cardiovascular Electrophysiology, 2001, 12, 630-636.	1.7	140
43	Contribution of Sodium Channel Mutations to Bradycardia and Sinus Node Dysfunction in LQT3 Families. Circulation Research, 2003, 92, 976-983.	4.5	140
44	Functional Na _V 1.8 Channels in Intracardiac Neurons. Circulation Research, 2012, 111, 333-343.	4.5	131
45	Identification of FOXP1 deletions in three unrelated patients with mental retardation and significant speech and language deficits. Human Mutation, 2010, 31, E1851-E1860.	2.5	130
46	Variants in the 3â€² untranslated region of the KCNQ1-encoded Kv7.1 potassium channel modify disease severity in patients with type 1 long QT syndrome in an allele-specific manner. European Heart Journal, 2012, 33, 714-723.	2.2	130
47	REVIEW: Sodium Channel (Dys)Function and Cardiac Arrhythmias. Cardiovascular Therapeutics, 2010, 28, 287-294.	2.5	128
48	Genome-wide association of multiple complex traits in outbred mice by ultra-low-coverage sequencing. Nature Genetics, 2016, 48, 912-918.	21.4	124
49	Genome-wide association study of multiple congenital heart disease phenotypes identifies a susceptibility locus for atrial septal defect at chromosome 4p16. Nature Genetics, 2013, 45, 822-824.	21.4	123
50	Genetics of cardiac arrhythmias. Heart, 2005, 91, 1352-1358.	2.9	122
51	Diagnostic Value of Flecainide Testing in Unmasking SCN5A-Related Brugada Syndrome. Journal of Cardiovascular Electrophysiology, 2006, 17, 857-864.	1.7	120
52	Whole Exome Sequencing Reveals the Major Genetic Contributors to Nonsyndromic Tetralogy of Fallot. Circulation Research, 2019, 124, 553-563.	4.5	118
53	Mechanism of right precordial ST-segment elevation in structural heart disease: Excitation failure by current-to-load mismatch. Heart Rhythm, 2010, 7, 238-248.	0.7	117
54	Identification of a Sudden Cardiac Death Susceptibility Locus at 2q24.2 through Genome-Wide Association in European Ancestry Individuals. PLoS Genetics, 2011, 7, e1002158.	3.5	117

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55	Calmodulin mutations and life-threatening cardiac arrhythmias: insights from the International Calmodulinopathy Registry. European Heart Journal, 2019, 40, 2964-2975.	2.2	116
56	PDZ Domainâ€“Binding Motif Regulates Cardiomyocyte Compartment-Specific Na _V 1.5 Channel Expression and Function. Circulation, 2014, 130, 147-160.	1.6	113
57	52 Genetic Loci Influencing MyocardialÂMass. Journal of the American College of Cardiology, 2016, 68, 1435-1448.	2.8	113
58	Role of common and rare variants in SCN10A: results from the Brugada syndrome QRS locus gene discovery collaborative study. Cardiovascular Research, 2015, 106, 520-529.	3.8	108
59	European Heart Rhythm Association (EHRA)/Heart Rhythm Society (HRS)/Asia Pacific Heart Rhythm Society (APHRS)/Latin American Heart Rhythm Society (LAHRS) Expert Consensus Statement on the state of genetic testing for cardiac diseases. Europace, 2022, 24, 1307-1367.	1.7	108
60	Beyond the One Geneâ€“One Disease Paradigm. Circulation, 2019, 140, 595-610.	1.6	101
61	Arrhythmogenic cardiomyopathy: pathology, genetics, and concepts in pathogenesis. Cardiovascular Research, 2017, 113, 1521-1531.	3.8	98
62	Reduced Sodium Channel Function Unmasks Residual Embryonic Slow Conduction in the Adult Right Ventricular Outflow Tract. Circulation Research, 2013, 113, 137-141.	4.5	87
63	Genetically Determined Differences in Sodium Current Characteristics Modulate Conduction Disease Severity in Mice With Cardiac Sodium Channelopathy. Circulation Research, 2009, 104, 1283-1292.	4.5	86
64	Gating-Dependent Mechanisms for Flecainide Action in SCN5A -Linked Arrhythmia Syndromes. Circulation, 2001, 104, 1200-1205.	1.6	85
65	Transethnic Genome-Wide Association Study Provides Insights in the Genetic Architecture and Heritability of Long QT Syndrome. Circulation, 2020, 142, 324-338.	1.6	83
66	European Heart Rhythm Association (EHRA)/Heart Rhythm Society (HRS)/Asia Pacific Heart Rhythm Society (APHRS)/Latin American Heart Rhythm Society (LAHRS) Expert Consensus Statement on the State of Genetic Testing for Cardiac Diseases. Heart Rhythm, 2022, 19, e1-e60.	0.7	78
67	Fever-induced QTc prolongation and ventricular arrhythmias in individuals with type 2 congenital long QT syndrome. Journal of Clinical Investigation, 2008, 118, 2552-61.	8.2	73
68	Natural genetic variation of the cardiac transcriptome in non-diseased donors and patients with dilated cardiomyopathy. Genome Biology, 2017, 18, 170.	8.8	70
69	Tubulin polymerization modifies cardiac sodium channel expression and gating. Cardiovascular Research, 2010, 85, 691-700.	3.8	68
70	Long QT syndrome caused by a large duplication in the KCNH2 (HERG) gene undetectable by current polymerase chain reaction-based exon-scanning methodologies. Heart Rhythm, 2006, 3, 52-55.	0.7	65
71	Polymorphisms in human connexin40 gene promoter are associated with increased risk of hypertension in men. Journal of Hypertension, 2006, 24, 325-330.	0.5	64
72	hiPSC-derived cardiomyocytes from Brugada Syndrome patients without identified mutations do not exhibit clear cellular electrophysiological abnormalities. Scientific Reports, 2016, 6, 30967.	3.3	64

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73	Role of sequence variations in the human ether-a-go-go-related gene (HERG, KCNH2) in the Brugada syndrome. Cardiovascular Research, 2005, 68, 441-453.	3.8	63
74	The Brugada Syndrome Susceptibility Gene HEY2 Modulates Cardiac Transmural Ion Channel Patterning and Electrical Heterogeneity. Circulation Research, 2017, 121, 537-548.	4.5	63
75	Genetics of sudden cardiac death caused by ventricular arrhythmias. Nature Reviews Cardiology, 2014, 11, 96-111.	13.7	59
76	A comprehensive evaluation of the genetic architecture of sudden cardiac arrest. European Heart Journal, 2018, 39, 3961-3969.	2.2	59
77	A Connexin40 Mutation Associated With a Malignant Variant of Progressive Familial Heart Block Type I. Circulation: Arrhythmia and Electrophysiology, 2012, 5, 163-172.	4.8	58
78	Coxsackie and Adenovirus Receptor Is a Modifier of Cardiac Conduction and Arrhythmia Vulnerability in the Setting of Myocardial Ischemia. Journal of the American College of Cardiology, 2014, 63, 549-559.	2.8	58
79	GNB5 Mutations Cause an Autosomal-Recessive Multisystem Syndrome with Sinus Bradycardia and Cognitive Disability. American Journal of Human Genetics, 2016, 99, 704-710.	6.2	58
80	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
81	Genome Wide Analysis of Drug-Induced Torsades de Pointes: Lack of Common Variants with Large Effect Sizes. PLoS ONE, 2013, 8, e78511.	2.5	57
82	Na+ channel mutation leading to loss of function and non-progressive cardiac conduction defects. Journal of Molecular and Cellular Cardiology, 2003, 35, 549-557.	1.9	56
83	Patch-Clamp Recording from Human Induced Pluripotent Stem Cell-Derived Cardiomyocytes: Improving Action Potential Characteristics through Dynamic Clamp. International Journal of Molecular Sciences, 2017, 18, 1873.	4.1	55
84	Predicting cardiac electrical response to sodium-channel blockade and Brugada syndrome using polygenic risk scores. European Heart Journal, 2019, 40, 3097-3107.	2.2	55
85	Genome-wide association analyses identify new Brugada syndrome risk loci and highlight a new mechanism of sodium channel regulation in disease susceptibility. Nature Genetics, 2022, 54, 232-239.	21.4	55
86	Combined reduction of intercellular coupling and membrane excitability differentially affects transverse and longitudinal cardiac conduction. Cardiovascular Research, 2009, 83, 52-60.	3.8	54
87	Switch From Fetal to Adult SCN5A Isoform in Human Induced Pluripotent Stem Cellâ€“Derived Cardiomyocytes Unmasks the Cellular Phenotype of a Conduction Diseaseâ€“Causing Mutation. Journal of the American Heart Association, 2017, 6, .	3.7	54
88	Developmental Aspects of Long QT Syndrome Type 3 and Brugada Syndrome on the Basis of a Single SCN5AMutation in Childhood. Journal of the American College of Cardiology, 2005, 46, 331-337.	2.8	52
89	Genetics of congenital heart disease: the contribution of the noncoding regulatory genome. Journal of Human Genetics, 2016, 61, 13-19.	2.3	52
90	Common genetic variation modulating cardiac ECG parameters and susceptibility to sudden cardiac death. Journal of Molecular and Cellular Cardiology, 2012, 52, 620-629.	1.9	51

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91	Analysis for Genetic Modifiers of Disease Severity in Patients With Long-QT Syndrome Type 2. Circulation: Cardiovascular Genetics, 2015, 8, 447-456.	5.1	51
92	Minor hypertrophic cardiomyopathy genes, major insights into the genetics of cardiomyopathies. Nature Reviews Cardiology, 2022, 19, 151-167.	13.7	50
93	Exclusion of multiple candidate genes and large genomic rearrangements in SCN5A in a Dutch Brugada syndrome cohort. Heart Rhythm, 2007, 4, 752-755.	0.7	48
94	Sudden cardiac arrest associated with use of a non-cardiac drug that reduces cardiac excitability: evidence from bench, bedside, and community. European Heart Journal, 2013, 34, 1506-1516.	2.2	47
95	Dissection of a Quantitative Trait Locus for PR Interval Duration Identifies Tnni3k as a Novel Modulator of Cardiac Conduction. PLoS Genetics, 2012, 8, e1003113.	3.5	45
96	Systematic large-scale assessment of the genetic architecture of left ventricular noncompaction reveals diverse etiologies. Genetics in Medicine, 2021, 23, 856-864.	2.4	45
97	Genome-Wide Identification of Expression Quantitative Trait Loci (eQTLs) in Human Heart. PLoS ONE, 2014, 9, e97380.	2.5	44
98	Genome-Wide Polyadenylation Maps Reveal Dynamic mRNA 3â€²-End Formation in the Failing Human Heart. Circulation Research, 2016, 118, 433-438.	4.5	41
99	Anti-arrhythmic potential of the late sodium current inhibitor GS-458967 in murine Scn5a-1798insD+/â^' and human SCN5A-1795insD+/â^' iPSC-derived cardiomyocytes. Cardiovascular Research, 2017, 113, 829-838.	3.8	41
100	SCN5A Mutation Type and a Genetic Risk Score Associate Variably With Brugada Syndrome Phenotype in SCN5A Families. Circulation Genomic and Precision Medicine, 2020, 13, e002911.	3.6	41
101	Characterization of a novel SCN5A mutation associated with Brugada syndrome reveals involvement of DIIIS4â€“S5 linker in slow inactivation. Cardiovascular Research, 2007, 76, 418-429.	3.8	40
102	When genetic burden reaches threshold. European Heart Journal, 2020, 41, 3849-3855.	2.2	40
103	Dilated Cardiomyopathy due to Sodium Channel Dysfunction. Circulation: Arrhythmia and Electrophysiology, 2008, 1, 80-82.	4.8	38
104	The yield of postmortem genetic testing in sudden death cases with structural findings at autopsy. European Journal of Human Genetics, 2020, 28, 17-22.	2.8	38
105	Epidemiology of inherited arrhythmias. Nature Reviews Cardiology, 2020, 17, 205-215.	13.7	37
106	Arrhythmogenic Cardiomyopathy. Circulation: Cardiovascular Genetics, 2011, 4, 318-326.	5.1	35
107	Yield and Pitfalls of Ajmaline Testing in theÂEvaluation of Unexplained Cardiac Arrest and Sudden Unexplained Death. JACC: Clinical Electrophysiology, 2017, 3, 1400-1408.	3.2	34
108	Voltageâ€gated sodium channels: Action players with many faces. Annals of Medicine, 2006, 38, 472-482.	3.8	33

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109	The Primary Arrhythmia Syndromes: Same Mutation, Different Manifestations. Are We Starting to Understand Why?. Journal of Cardiovascular Electrophysiology, 2008, 19, 445-452.	1.7	33
110	Sodium Channel Remodeling in Subcellular Microdomains of Murine Failing Cardiomyocytes. Journal of the American Heart Association, 2017, 6, .	3.7	31
111	Identification of an INa-dependent and Ito-mediated proarrhythmic mechanism in cardiomyocytes derived from pluripotent stem cells of a Brugada syndrome patient. Scientific Reports, 2018, 8, 11246.	3.3	31
112	Gain-of-function mutation in SCN5A causes ventricular arrhythmias and early onset atrial fibrillation. International Journal of Cardiology, 2017, 236, 187-193.	1.7	30
113	Animal models and animal-free innovations for cardiovascular research: current status and routes to be explored. Consensus document of the ESC Working Group on Myocardial Function and the ESC Working Group on Cellular Biology of the Heart. Cardiovascular Research, 2022, 118, 3016-3051.	3.8	30
114	Sequential Defects in Cardiac Lineage Commitment and Maturation Cause Hypoplastic Left Heart Syndrome. Circulation, 2021, 144, 1409-1428.	1.6	29
115	The role of reninâ€“angiotensinâ€“aldosterone system polymorphisms in phenotypic expression of MYBPC3-related hypertrophic cardiomyopathy. European Journal of Human Genetics, 2012, 20, 1071-1077.	2.8	28
116	Readthrough-Promoting Drugs Gentamicin and PTC124 Fail to Rescue Na _v 1.5 Function of Human-Induced Pluripotent Stem Cellâ€“Derived Cardiomyocytes Carrying Nonsense Mutations in the Sodium Channel Gene SCN5A. Circulation: Arrhythmia and Electrophysiology, 2016, 9, .	4.8	28
117	Impact of Selection Bias on Estimation of Subsequent Event Risk. Circulation: Cardiovascular Genetics, 2017, 10, .	5.1	28
118	Exploring the Relationship Between Schizophrenia and Cardiovascular Disease: A Genetic Correlation and Multivariable Mendelian Randomization Study. Schizophrenia Bulletin, 2022, 48, 463-473.	4.3	28
119	Sudden Cardiac Arrest and Rare Genetic Variants in the Community. Circulation: Cardiovascular Genetics, 2016, 9, 147-153.	5.1	27
120	Homozygous frameshift mutations in FAT1 cause a syndrome characterized by colobomatous-microphthalmia, ptosis, nephropathy and syndactyly. Nature Communications, 2019, 10, 1180.	12.8	27
121	Heart failure following STEMI: a contemporary cohort study of incidence and prognostic factors. Open Heart, 2017, 4, e000551.	2.3	26
122	Developmental aspects of cardiac arrhythmogenesis. Cardiovascular Research, 2011, 91, 243-251.	3.8	25
123	Dilation of the Aorta Ascendens Forms Part of the Clinical Spectrum of HCN4 Mutations. Journal of the American College of Cardiology, 2016, 67, 2313-2315.	2.8	25
124	A Complex Double Deletion in LMNA Underlies Progressive Cardiac Conduction Disease, Atrial Arrhythmias, and Sudden Death. Circulation: Cardiovascular Genetics, 2011, 4, 280-287.	5.1	24
125	Electrophysiological Abnormalities in VLCAD Deficient hiPSC-Cardiomyocytes Can Be Improved by Lowering Accumulation of Fatty Acid Oxidation Intermediates. International Journal of Molecular Sciences, 2020, 21, 2589.	4.1	24
126	Chronically elevated branched chain amino acid levels are pro-arrhythmic. Cardiovascular Research, 2022, 118, 1742-1757.	3.8	24

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127	European Heart Rhythm Association (<scp>EHRA</scp>)/Heart Rhythm Society (<scp>HRS</scp>)/Asia Pacific Heart Rhythm Society (<scp>APHRS</scp>)/Latin American Heart Rhythm Society (<scp>LAHRS</scp>) Expert Consensus Statement on the state of genetic testing for cardiac diseases. Journal of Arrhythmia, 2022, 38, 491-553.	1.2	24
128	A Heterozygous Deletion Mutation in the Cardiac Sodium Channel Gene SCN5A with Loss- and Gain-of-Function Characteristics Manifests as Isolated Conduction Disease, without Signs of Brugada or Long QT Syndrome. PLoS ONE, 2013, 8, e67963.	2.5	23
129	Common and rare susceptibility genetic variants predisposing to Brugada syndrome in Thailand. Heart Rhythm, 2020, 17, 2145-2153.	0.7	23
130	Targeting the Microtubule EB1-CLASP2 Complex Modulates Na _V 1.5 at Intercalated Discs. Circulation Research, 2021, 129, 349-365.	4.5	23
131	Quantitative trait loci for electrocardiographic parameters and arrhythmia in the mouse. Journal of Molecular and Cellular Cardiology, 2011, 50, 380-389.	1.9	22
132	Variants in the SCN5A Promoter Associated With Various Arrhythmia Phenotypes. Journal of the American Heart Association, 2016, 5, .	3.7	22
133	Association of Chromosome 9p21 With Subsequent Coronary Heart Disease Events. Circulation Genomic and Precision Medicine, 2019, 12, e002471.	3.6	22
134	A novel LQT3 mutation implicates the human cardiac sodium channel domain IVS6 in inactivation kinetics. Cardiovascular Research, 2003, 57, 1072-1078.	3.8	21
135	Exome sequencing identifies primary carnitine deficiency in a family with cardiomyopathy and sudden death. European Journal of Human Genetics, 2017, 25, 783-787.	2.8	21
136	Next-Generation Sequencing in Post-mortem Genetic Testing of Young Sudden Cardiac Death Cases. Frontiers in Cardiovascular Medicine, 2016, 3, 13.	2.4	20
137	Variant Intronic Enhancer Controls SCN10A-short Expression and Heart Conduction. Circulation, 2021, 144, 229-242.	1.6	20
138	Genomic organisation and chromosomal localisation of two members of the KCND ion channel family, KCND2 and KCND3. Human Genetics, 2000, 106, 614-619.	3.8	19
139	Genetic variation in GNB5 causes bradycardia by increasing IK,ACh augmenting cholinergic response. DMM Disease Models and Mechanisms, 2019, 12, .	2.4	19
140	Systems Genetics Approaches in Rat Identify Novel Genes and Gene Networks Associated With Cardiac Conduction. Journal of the American Heart Association, 2018, 7, e009243.	3.7	18
141	Supraventricular tachycardias, conduction disease, and cardiomyopathy in 3 families with the same rare variant in TNNI3K (p.Glu768Lys). Heart Rhythm, 2019, 16, 98-105.	0.7	18
142	Genome-wide association studies of cardiac electrical phenotypes. Cardiovascular Research, 2020, 116, 1620-1634.	3.8	18
143	A common co-morbidity modulates disease expression and treatment efficacy in inherited cardiac sodium channelopathy. European Heart Journal, 2018, 39, 2898-2907.	2.2	17
144	Subsequent Event Risk in Individuals With Established Coronary Heart Disease. Circulation Genomic and Precision Medicine, 2019, 12, e002470.	3.6	17

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145	Substitution of a conserved alanine in the domain IIIS4â€“S5 linker of the cardiac sodium channel causes long QT syndrome. Cardiovascular Research, 2005, 67, 459-466.	3.8	16
146	Bioinformatic analysis of a plakophilin-2-dependent transcription network: implications for the mechanisms of arrhythmogenic right ventricular cardiomyopathy in humans and in boxer dogs. Europace, 2018, 20, iii125-iii132.	1.7	16
147	Enhanced late sodium current underlies pro-arrhythmic intracellular sodium and calcium dysregulation in murine sodium channelopathy. International Journal of Cardiology, 2018, 263, 54-62.	1.7	16
148	GATA6 mutations: Characterization of two novel patients and a comprehensive overview of the GATA6 genotypic and phenotypic spectrum. American Journal of Medical Genetics, Part A, 2019, 179, 1836-1845.	1.2	16
149	Biallelic loss-of-function variants in PLD1 cause congenital right-sided cardiac valve defects and neonatal cardiomyopathy. Journal of Clinical Investigation, 2021, 131, .	8.2	16
150	Genetic modulation of cardiac repolarization reserve. Heart Rhythm, 2007, 4, 608-610.	0.7	15
151	Early repolarization pattern: its ECG characteristics, arrhythmogeneity and heritability. Journal of Interventional Cardiac Electrophysiology, 2014, 39, 185-192.	1.3	15
152	TNNI3K in cardiovascular disease and prospects for therapy. Journal of Molecular and Cellular Cardiology, 2015, 82, 167-173.	1.9	15
153	Genetics and genomics of arrhythmic risk: current and future strategies to prevent sudden cardiac death. Nature Reviews Cardiology, 2021, 18, 774-784.	13.7	15
154	Common Genetic Variants Contribute to Risk of Transposition of the Great Arteries. Circulation Research, 2022, 130, 166-180.	4.5	15
155	Maturation of hiPSC-derived cardiomyocytes promotes adult alternative splicing of SCN5A and reveals changes in sodium current associated with cardiac arrhythmia. Cardiovascular Research, 2023, 119, 167-182.	3.8	13
156	A deep learning approach identifies new ECG features in congenital long QT syndrome. BMC Medicine, 2022, 20, 162.	5.5	13
157	Illuminating the path from genetics to clinical outcome in Brugada syndrome. European Heart Journal, 2021, 42, 1091-1093.	2.2	12
158	Pharmacological rescue of mutant ion channels. Cardiovascular Research, 2002, 55, 229-232.	3.8	11
159	Arrhythmogenic Right Ventricular Cardiomyopathy: Growing Evidence for Complex Inheritance. Circulation: Cardiovascular Genetics, 2013, 6, 525-527.	5.1	11
160	Functional Consequences of the SCN5A-p.Y1977N Mutation within the PY Ubiquitylation Motif: Discrepancy between HEK293 Cells and Transgenic Mice. International Journal of Molecular Sciences, 2019, 20, 5033.	4.1	11
161	The molecular genetics of arrhythmias. Cardiovascular Research, 2005, 67, 343-346.	3.8	10
162	Integrative Genomic Approach Identifies Multiple Genes Involved in Cardiac Collagen Deposition. Circulation: Cardiovascular Genetics, 2014, 7, 790-798.	5.1	10

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163	Blood Pressure-Associated Genetic Variants in the Natriuretic Peptide Receptor 1 Gene Modulate Guanylate Cyclase Activity. Circulation Genomic and Precision Medicine, 2019, 12, e002472.	3.6	10
164	SNPs Identified as Modulators of ECG Traits in the General Population Do Not Markedly Affect ECG Traits during Acute Myocardial Infarction nor Ventricular Fibrillation Risk in This Condition. PLoS ONE, 2013, 8, e57216.	2.5	9
165	Functional modulation of atrio-ventricular conduction by enhanced late sodium current and calcium-dependent mechanisms in Scn5a1798insD/+ mice. Europace, 2020, 22, 1579-1589.	1.7	9
166	The Chemical Compound PTC124 Does Not Affect Cellular Electrophysiology of Cardiac Ventricular Myocytes. Cardiovascular Drugs and Therapy, 2012, 26, 41-45.	2.6	8
167	Novel pathogenic role for galectin-3 in early disease stages of arrhythmogenic cardiomyopathy. Heart Rhythm, 2021, 18, 1394-1403.	0.7	8
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