jean-Jacques Schott

List of Publications by Year
in descending order

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107	12,228	44444 50	34195 103
papers	citations	h-index	g-index

113	113	113	11068
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Genome-wide association study reveals novel genetic loci: a new polygenic risk score for mitral valve prolapse. European Heart Journal, 2022, 43, 1668-1680.	1.0	25
2	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.	9.4	55
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.	1.1	57
4	Cadherin 2-Related Arrhythmogenic Cardiomyopathy. Circulation Genomic and Precision Medicine, 2021, 14, e003097.	1.6	21
5	<scp>DZIP1</scp> regulates mammalian cardiac valve development through a Cby1â€Î²â€catenin mechanism. Developmental Dynamics, 2021, 250, 1432-1449.	0.8	6
6	Replacement Myocardial Fibrosis in Patients With Mitral Valve Prolapse. Circulation, 2021, 143, 1763-1774.	1.6	81
7	A consistent arrhythmogenic trait in Brugada syndrome cellular phenotype. Clinical and Translational Medicine, 2021, 11, e413.	1.7	5
8	Functionally validated SCN5A variants allow interpretation of pathogenicity and prediction of lethal events in Brugada syndrome. European Heart Journal, 2021, 42, 2854-2863.	1.0	37
9	Genome-Wide Association Meta-Analysis Supports Genes Involved in Valve and Cardiac Development to Associate With Mitral Valve Prolapse. Circulation Genomic and Precision Medicine, 2021, 14, e003148.	1.6	7
10	A standardised hERG phenotyping pipeline to evaluate KCNH2 genetic variant pathogenicity. Clinical and Translational Medicine, 2021, 11, e609.	1.7	7
11	Cardiac Emerinopathy. Circulation: Arrhythmia and Electrophysiology, 2020, 13, e008712.	2.1	20
12	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.	1.6	41
13	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
14	Genetic Association Analyses Highlight IL6 , ALPL , and NAV1 As 3 New Susceptibility Genes Underlying Calcific Aortic Valve Stenosis. Circulation Genomic and Precision Medicine, 2019, 12, e002617.	1.6	45
15	Genetic Variation in LPA, Calcific Aortic Valve Stenosis in Patients Undergoing Cardiac Surgery, and Familial Risk of Aortic Valve Microcalcification. JAMA Cardiology, 2019, 4, 620.	3.0	32
16	Primary cilia defects causing mitral valve prolapse. Science Translational Medicine, 2019, 11, .	5.8	76
17	RRAD mutation causes electrical and cytoskeletal defects in cardiomyocytes derived from a familial case of Brugada syndrome. European Heart Journal, 2019, 40, 3081-3094.	1.0	48
18	Genome-Wide Association Studyâ€“Driven Gene-Set Analyses, Genetic, and Functional Follow-Up Suggest GLIS1 as a Susceptibility Gene for Mitral Valve Prolapse. Circulation Genomic and Precision Medicine, 2019, 12, e002497.	1.6	31

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19	eDiVAâ€"Classification and prioritization of pathogenic variants for clinical diagnostics. Human Mutation, 2019, 40, 865-878.	1.1	19
20	Mental stress test: a rapid, simple, and efficient test to unmask long QT syndrome. Europace, 2018, 20, 2014-2020.	0.7	15
21	Genetics of syndromic and non-syndromic mitral valve prolapse. Heart, 2018, 104, 978-984.	1.2	44
22	Rare Coding Variants in ANGPTL6 Are Associated with Familial Forms of Intracranial Aneurysm. American Journal of Human Genetics, 2018, 102, 133-141.	2.6	37
23	New insights into mitral valve dystrophy: a Filamin-A genotypeâ€"phenotype and outcome study. European Heart Journal, 2018, 39, 1269-1277.	1.0	44
24	SCN5A mutations in 442 neonates and children: genotypeâ€"phenotype correlation and identification of higher-risk subgroups. European Heart Journal, 2018, 39, 2879-2887.	1.0	33
25	Localized Structural Alterations Underlying a Subset of Unexplained Sudden Cardiac Death. Circulation: Arrhythmia and Electrophysiology, 2018, 11, e006120.	2.1	67
26	Familial Catecholamine-Induced QT Prolongation in Unexplained Sudden Cardiac Death. Journal of the American College of Cardiology, 2017, 69, 1642-1643.	1.2	7
27	The alternatively spliced LRRFIP1 Isoform-1 is a key regulator of the Wnt/Î²-catenin transcription pathway. Biochimica Et Biophysica Acta - Molecular Cell Research, 2017, 1864, 1142-1152.	1.9	13
28	Clinical Yield of Familial Screening After Sudden Death in Young Subjects. Circulation: Arrhythmia and Electrophysiology, 2017, 10, .	2.1	29
29	Progressive Atrial Conduction Defects Associated With Bone Malformation Caused by a Connexin-45 Mutation. Journal of the American College of Cardiology, 2017, 70, 358-370.	1.2	27
30	TRPM4 non-selective cation channel variants in long QT syndrome. BMC Medical Genetics, 2017, 18, 31.	2.1	26
31	DoEstRare: A statistical test to identify local enrichments in rare genomic variants associated with disease. PLoS ONE, 2017, 12, e0179364.	1.1	7
32	The Brugada Syndrome: A Rare Arrhythmia Disorder with Complex Inheritance. Frontiers in Cardiovascular Medicine, 2016, 3, 9.	1.1	48
33	Variants of Transient Receptor Potential Melastatin Member 4 in Childhood Atrioventricular Block. Journal of the American Heart Association, 2016, 5, .	1.6	50
34	Identification of novel APOB mutations by targeted next-generation sequencing for the molecular diagnosis of familial hypobetalipoproteinemia. Atherosclerosis, 2016, 250, 52-56.	0.4	17
35	Variants in the SCN5A Promoter Associated With Various Arrhythmia Phenotypes. Journal of the American Heart Association, 2016, 5, .	1.6	22
36	Dysfunction of the Voltageâ€Gated K ⁺ Channel Î²2 Subunit in a Familial Case of Brugada Syndrome. Journal of the American Heart Association, 2016, 5, .	1.6	20

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37	Search for Rare Copy-Number Variants in Congenital Heart Defects Identifies Novel Candidate Genes and a Potential Role for FOXC1 in Patients With Coarctation of the Aorta. Circulation: Cardiovascular Genetics, 2016, 9, 86-94.	5.1	38
38	Targeted resequencing identifies TRPM4 as a major gene predisposing to progressive familial heart block type I. International Journal of Cardiology, 2016, 207, 349-358.	0.8	62
39	MVP-Associated Filamin A Mutations Affect FlnA-PTPN12 (PTP-PEST) Interactions. Journal of Cardiovascular Development and Disease, 2015, 2, 233-247.	0.8	15
40	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
41	Testing the burden of rare variation in arrhythmia-susceptibility genes provides new insights into molecular diagnosis for Brugada syndrome. Human Molecular Genetics, 2015, 24, 2757-2763.	1.4	130
42	Fine-scale human genetic structure in Western France. European Journal of Human Genetics, 2015, 23, 831-836.	1.4	31
43	Mitral valve diseaseâ€"morphology and mechanisms. Nature Reviews Cardiology, 2015, 12, 689-710.	6.1	281
44	Mutations in DCHS1 cause mitral valve prolapse. Nature, 2015, 525, 109-113.	13.7	150
45	Genetic association analyses highlight biological pathways underlying mitral valve prolapse. Nature Genetics, 2015, 47, 1206-1211.	9.4	103
46	Brugada Syndrome and Nav1.5. Cardiac Electrophysiology Clinics, 2014, 6, 715-721.	0.7	0
47	Complex Brugada syndrome inheritance in a family harbouring compound SCN5A and CACNA1C mutations. Basic Research in Cardiology, 2014, 109, 446.	2.5	20
48	Genetic association study of QT interval highlights role for calcium signaling pathways in myocardial repolarization. Nature Genetics, 2014, 46, 826-836.	9.4	281
49	Valvular dystrophy associated filamin A mutations reveal a new role of its first repeats in small-GTPase regulation. Biochimica Et Biophysica Acta - Molecular Cell Research, 2014, 1843, 234-244.	1.9	30
50	Myotonic dystrophy type 1 mimics and exacerbates Brugada phenotype induced by Nav1.5 sodium channel loss-of-function mutation. Heart Rhythm, 2014, 11, 1393-1400.	0.3	19
51	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.	9.4	467
52	Identification of Large Families in Early Repolarization Syndrome. Journal of the American College of Cardiology, 2013, 61, 164-172.	1.2	81
53	Molecular Genetics and Functional Anomalies in a Series of 248 Brugada Cases with 11 Mutations in the TRPM4 Channel. PLoS ONE, 2013, 8, e54131.	1.1	131
54	Genome Wide Analysis of Drug-Induced Torsades de Pointes: Lack of Common Variants with Large Effect Sizes. PLoS ONE, 2013, 8, e78511.	1.1	57

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55	Progressive Cardiac Conduction Disease. , 2013, , 583-603.		0
56	A Connexin40 Mutation Associated With a Malignant Variant of Progressive Familial Heart Block Type I. Circulation: Arrhythmia and Electrophysiology, 2012, 5, 163-172.	2.1	58
57	Parental Electrocardiographic Screening Identifies a High Degree of Inheritance for Congenital and Childhood Nonimmune Isolated Atrioventricular Block. Circulation, 2012, 126, 1469-1477.	1.6	25
58	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
59	Identification of a strong genetic background for progressive cardiac conduction defect by epidemiological approach. Heart, 2012, 98, 1305-1310.	1.2	13
60	Developmental basis for filamin-A-associated myxomatous mitral valve disease. Cardiovascular Research, 2012, 96, 109-119.	1.8	68
61	Characteristics and long-term outcome of non-immune isolated atrioventricular block diagnosed in utero or early childhood: a multicentre study. European Heart Journal, 2012, 33, 622-629.	1.0	68
62	KLHL3 mutations cause familial hyperkalemic hypertension by impairing ion transport in the distal nephron. Nature Genetics, 2012, 44, 456-460.	9.4	281
63	Multifocal Ectopic Purkinje-Related Premature Contractions. Journal of the American College of Cardiology, 2012, 60, 144-156.	1.2	156
64	Screening for Copy Number Variation in Genes Associated With the Long QT Syndrome. Journal of the American College of Cardiology, 2011, 57, 40-47.	1.2	78
65	Hereditary Cardiac Conduction Diseases. , 2011, , 253-260.		0
66	Filamin-A-Related Myxomatous Mitral Valve Dystrophy: Genetic, Echocardiographic and Functional Aspects. Journal of Cardiovascular Translational Research, 2011, 4, 748-756.	1.1	39
67	Defects in Ankyrin-Based Membrane Protein Targeting Pathways Underlie Atrial Fibrillation. Circulation, 2011, 124, 1212-1222.	1.6	102
68	MOG1. Circulation: Cardiovascular Genetics, 2011, 4, 261-268.	5.1	151
69	Expression of the familial cardiac valvular dystrophy gene, filaminâ€A, during heart morphogenesis. Developmental Dynamics, 2010, 239, 2118-2127.	0.8	46
70	An international compendium of mutations in the SCN5A-encoded cardiac sodium channel in patients referred for Brugada syndrome genetic testing. Heart Rhythm, 2010, 7, 33-46.	0.3	649
71	Variable Nav1.5 Protein Expression from the Wild-Type Allele Correlates with the Penetrance of Cardiac Conduction Disease in the Scn5a+/â^' Mouse Model. PLoS ONE, 2010, 5, e9298.	1.1	67
72	SCN5A Mutations and the Role of Genetic Background in the Pathophysiology of Brugada Syndrome. Circulation: Cardiovascular Genetics, 2009, 2, 552-557.	5.1	262

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73	Ventricular Fibrillation with Prominent Early Repolarization Associated with a Rare Variant of KCNJ8/K_{ATP} Channel. Journal of Cardiovascular Electrophysiology, 2009, 20, 93-98.	0.8	269
74	Type of SCN5A mutation determines clinical severity and degree of conduction slowing in loss-of-function sodium channelopathies. Heart Rhythm, 2009, 6, 341-348.	0.3	224
75	Genetic mechanisms of mitral valve prolapse. Current Cardiovascular Risk Reports, 2008, 2, 463-467.	0.8	5
76	Progressive Cardiac Conduction Disease. , 2008, , 564-576.		6
77	Exon organization and novel alternative splicing of the human ANK2 gene: Implications for cardiac function and human cardiac disease. Journal of Molecular and Cellular Cardiology, 2008, 45, 724-734.	0.9	31
78	Sodium channel blocker tests allow a clear distinction of electrophysiological characteristics and prognosis in patients with a type 2 or 3 Brugada electrocardiogram pattern. Heart Rhythm, 2008, 5, 1561-1564.	0.3	16
79	Dysfunction in ankyrin-B-dependent ion channel and transporter targeting causes human sinus node disease. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 15617-15622.	3.3	163
80	Sodium channel Î²1 subunit mutations associated with Brugada syndrome and cardiac conduction disease in humans. Journal of Clinical Investigation, 2008, 118, 2260-8.	3.9	400
81	Clinical Aspects and Prognosis of Brugada Syndrome in Children. Circulation, 2007, 115, 2042-2048.	1.6	275
82	Mutations in the Gene Encoding Filamin A as a Cause for Familial Cardiac Valvular Dystrophy. Circulation, 2007, 115, 40-49.	1.6	257
83	Defining the Cellular Phenotype of â€œAnkyrin-B Syndromeâ€•Variants. Circulation, 2007, 115, 432-441.	1.6	161
84	Torsades de pointes complicating atrioventricular block: Evidence for a genetic predisposition. Heart Rhythm, 2007, 4, 170-174.	0.3	48
85	Monomorphic Ventricular Tachycardia Due to Brugada Syndrome Successfully Treated by Hydroquinidine Therapy in a 3-Year-Old Child. Journal of Cardiovascular Electrophysiology, 2006, 17, 97-100.	0.8	49
86	Progressive Cardiac Conduction Defect is the Prevailing Phenotype in Carriers of a Brugada Syndrome SCN5A Mutation. Journal of Cardiovascular Electrophysiology, 2006, 17, 270-275.	0.8	90
87	Familial Aggregation of Calcific Aortic Valve Stenosis in the Western Part of France. Circulation, 2006, 113, 856-860.	1.6	74
88	14-3-3 Is a Regulator of the Cardiac Voltage-Gated Sodium Channel Nav1.5. Circulation Research, 2006, 98, 1538-1546.	2.0	77
89	Unusual clinical presentation in a family with catecholaminergic polymorphic ventricular tachycardia due to a G14876A ryanodine receptor gene mutation. American Journal of Cardiology, 2005, 95, 700-702.	0.7	22
90	A Common Antitussive Drug, Clobutinol, Precipitates the Long QT Syndrome 2. Molecular Pharmacology, 2004, 66, 1093-1102.	1.0	53

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91	Novel Brugada SCN5A Mutation Leading to ST Segment Elevation in the Inferior or the Right Precordial Leads. Journal of Cardiovascular Electrophysiology, 2003, 14, 200-203.	0.8	97
92	Ankyrin-B mutation causes type 4 long-QT cardiac arrhythmia and sudden cardiac death. Nature, 2003, 421, 634-639.	13.7	926
93	Haploinsufficiency in combination with aging causes SCN5A-linked hereditary LenÃ¨gre disease. Journal of the American College of Cardiology, 2003, 41, 643-652.	1.2	158
94	Cardiac retention of [¹¹C]HED in genotyped long QT patients: a potential amplifier role for severity of the disease. American Journal of Physiology - Heart and Circulatory Physiology, 2003, 285, H1286-H1293.	1.5	31
95	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.	1.2	360
96	Familial Deafness, Congenital Heart Defects, and Posterior Embryotoxon Caused by Cysteine Substitution in the First Epidermal-Growth-Factorâ€“Like Domain of Jagged 1. American Journal of Human Genetics, 2002, 71, 180-186.	2.6	63
97	Non-invasive testing of acquired long QT syndrome Evidence for multiple arrhythmogenic substrates. Cardiovascular Research, 2001, 50, 386-398.	1.8	53
98	Novel SCN5A Mutation Leading Either to Isolated Cardiac Conduction Defect or Brugada Syndrome in a Large French Family. Circulation, 2001, 104, 3081-3086.	1.6	348
99	Loss of function and inhibitory effects of human CSX/NKX2.5 homeoprotein mutations associated with congenital heart disease. Journal of Clinical Investigation, 2000, 106, 299-308.	3.9	149
100	Cardiac Expression of the Ventricle-Specific Homeobox Gene Irx4 Is Modulated by Nkx2-5 and dHand. Developmental Biology, 2000, 217, 266-277.	0.9	183
101	Clinical characteristics of a familial inherited myxomatous valvular dystrophy mapped to Xq28. Journal of the American College of Cardiology, 2000, 35, 1890-1897.	1.2	48
102	Cardiac conduction defects associate with mutations in SCN5A. Nature Genetics, 1999, 23, 20-21.	9.4	549
103	Mapping of X-Linked Myxomatous Valvular Dystrophy to Chromosome Xq28. American Journal of Human Genetics, 1998, 62, 627-632.	2.6	86
104	Congenital Heart Disease Caused by Mutations in the Transcription Factor NKX2-5. , 1998, 281, 108-111.		1,156
105	A genetic linkage map of the rat derived from recombinant inbred strains. Mammalian Genome, 1996, 7, 117-127.	1.0	108
106	Mapping of quantitative trait loci for blood pressure and cardiac mass in the rat by genome scanning of recombinant inbred strains.. Journal of Clinical Investigation, 1995, 96, 1973-1978.	3.9	146
107	A radiation hybrid map of 506 STS markers spanning human chromosome 11. Nature Genetics, 1994, 8, 70-76.	9.4	157