Juan P Kaski

List of Publications by Year
in descending order

109 papers	3,785 citations	172457 29 h-index	138484 58 g-index
116 all docs	116 docs citations	116 times ranked	6079 citing authors

#	Article	IF	Citations
1	De novo mutations in histone-modifying genes in congenital heart disease. Nature, 2013, 498, 220-223.	27.8	798
2	Idiopathic restrictive cardiomyopathy in children is caused by mutations in cardiac sarcomere protein genes. Heart, 2008, 94, 1478-1484.	2.9	188
3	Prevalence of Sarcomere Protein Gene Mutations in Preadolescent Children With Hypertrophic Cardiomyopathy. Circulation: Cardiovascular Genetics, 2009, 2, 436-441.	5.1	176
4	Development of a Novel Risk Prediction Model for Sudden Cardiac Death in Childhood Hypertrophic Cardiomyopathy (HCM Risk-Kids). JAMA Cardiology, 2019, 4, 918.	6.1	147
5	The Congenital Heart Disease Genetic Network Study. Circulation Research, 2013, 112, 698-706.	4.5	142
6	Clinical profile of patients with ATP1A3 mutations in Alternating Hemiplegia of Childhoodâ€"a study of 155 patients. Orphanet Journal of Rare Diseases, 2015, 10, 123.	2.7	117
7	Calmodulin mutations and life-threatening cardiac arrhythmias: insights from the International Calmodulinopathy Registry. European Heart Journal, 2019, 40, 2964-2975.	2.2	116
8	Long-Term Outcomes in Hypertrophic Cardiomyopathy Caused by Mutations in the Cardiac Troponin T Gene. Circulation: Cardiovascular Genetics, 2012, 5, 10-17.	5.1	103
9	Thioredoxin Reductase 2 (TXNRD2) Mutation Associated With Familial Glucocorticoid Deficiency (FGD). Journal of Clinical Endocrinology and Metabolism, 2014, 99, E1556-E1563.	3.6	101
10	The Cardiomyopathy Registry of the EUREObservational Research Programme of the European Society of Cardiology: baseline data and contemporary management of adult patients with cardiomyopathies. European Heart Journal, 2018, 39, 1784-1793.	2.2	94
11	Risk factors for sudden cardiac death in childhood hypertrophic cardiomyopathy: A systematic review and meta-analysis. European Journal of Preventive Cardiology, 2017, 24, 1220-1230.	1.8	89
12	Penetrance of Hypertrophic Cardiomyopathy in Sarcomere Protein Mutation Carriers. Journal of the American College of Cardiology, 2020, 76, 550-559.	2.8	89
13	Irbesartan in Marfan syndrome (AIMS): a double-blind, placebo-controlled randomised trial. Lancet, The, 2019, 394, 2263-2270.	13.7	88
14	Clinical presentation and survival of childhood hypertrophic cardiomyopathy: a retrospective study in United Kingdom. European Heart Journal, 2019, 40, 986-993.	2.2	80
15	Outcomes after implantable cardioverter-defibrillator treatment in children with hypertrophic cardiomyopathy. Heart, 2007, 93, 372-374.	2.9	78
16	Hypertrophic cardiomyopathy in children. Heart, 2012, 98, 1044-1054.	2.9	75
17	Cardiac defects, morbidity and mortality in patients affected by RASopathies. CARNET study results. International Journal of Cardiology, 2017, 245, 92-98.	1.7	75
18	Yield of Clinical Screening for Hypertrophic Cardiomyopathy in Child First-Degree Relatives. Circulation, 2019, 140, 184-192.	1.6	58

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19	Functional Analysis of a Unique Troponin C Mutation, GLY159ASP, that Causes Familial Dilated Cardiomyopathy, Studied in Explanted Heart Muscle. Circulation: Heart Failure, 2009, 2, 456-464.	3.9	46
20	Long-term Safety and Efficacy of Mexiletine for Patients With Skeletal Muscle Channelopathies. JAMA Neurology, 2015, 72, 1531.	9.0	45
21	Clinical Features and Natural History of PRKAG2 Variant Cardiac Glycogenosis. Journal of the American College of Cardiology, 2020, 76, 186-197.	2.8	45
22	Cardiac Disease in Adolescents With Delayed Diagnosis of Vertically Acquired HIV Infection. Clinical Infectious Diseases, 2013, 56, 576-582.	5.8	39
23	An International Multicenter Evaluation of Type 5 Long QT Syndrome. Circulation, 2020, 141, 429-439.	1.6	39
24	Somatic mutations of GNA11 and GNAQ in CTNNB1-mutant aldosterone-producing adenomas presenting in puberty, pregnancy or menopause. Nature Genetics, 2021, 53, 1360-1372.	21.4	37
25	A validation study of the European Society of Cardiology guidelines for risk stratification of sudden cardiac death in childhood hypertrophic cardiomyopathy. Europace, 2019, 21, 1559-1565.	1.7	34
26	SCN5A mutations in 442 neonates and children: genotypeâ€“phenotype correlation and identification of higher-risk subgroups. European Heart Journal, 2018, 39, 2879-2887.	2.2	33
27	Genetic Mosaicism in Calmodulinopathy. Circulation Genomic and Precision Medicine, 2019, 12, 375-385.	3.6	33
28	Epidemiology and Clinical Aspects of Genetic Cardiomyopathies. Heart Failure Clinics, 2018, 14, 119-128.	2.1	32
29	Interpretation and actionability of genetic variants in cardiomyopathies: a position statement from the European Society of Cardiology Council on cardiovascular genomics. European Heart Journal, 2022, 43, 1901-1916.	2.2	32
30	The Classification Concept of the ESC Working Group on Myocardial and Pericardial Diseases for Dilated Cardiomyopathy. Herz, 2007, 32, 446-451.	1.1	31
31	Pigmentary hypertrichosis and non-autoimmune insulin-dependent diabetes mellitus (PHID) syndrome is associated with severe chronic inflammation and cardiomyopathy, and represents a new monogenic autoinflammatory syndrome. Journal of Pediatric Endocrinology and Metabolism, 2013, 26, 877-82.	0.9	31
32	Faulty cardiac repolarization reserve in alternating hemiplegia of childhood broadens the phenotype. Brain, 2015, 138, 2859-2874.	7.6	30
33	External validation of the HCM Risk-Kids model for predicting sudden cardiac death in childhood hypertrophic cardiomyopathy. European Journal of Preventive Cardiology, 2022, 29, 678-686.	1.8	30
34	Multidisciplinary evaluation and management of obstructive hypertrophic cardiomyopathy in 2020: Towards the HCM Heart Team. International Journal of Cardiology, 2020, 304, 86-92.	1.7	29
35	Clinical Profile of Cardiac Involvement in Danon Disease. Circulation Genomic and Precision Medicine, 2020, 13, e003117.	3.6	29
36	B-type natriuretic peptide predicts disease severity in children with hypertrophic cardiomyopathy. Heart, 2007, 94, 1307-1311.	2.9	27

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37	Prevalence of Sequence Variants in the RAS-Mitogen Activated Protein Kinase Signaling Pathway in Pre-Adolescent Children With Hypertrophic Cardiomyopathy. Circulation: Cardiovascular Genetics, 2012, 5, 317-326.	5.1	23
38	Differences between familial and sporadic dilated cardiomyopathy: ESC EORP Cardiomyopathy & Myocarditis registry. ESC Heart Failure, 2021, 8, 95-105.	3.1	23
39	Clinical presentation and longâ€term outcomes of infantile hypertrophic cardiomyopathy: a European multicentre study. ESC Heart Failure, 2021, 8, 5057-5067.	3.1	22
40	Can atrioventricular septal defects exist with intact septal structures?. Heart, 2005, 92, 832-835.	2.9	21
41	Mutations in the cardiac Troponin C gene are a cause of idiopathic dilated cardiomyopathy in childhood. Cardiology in the Young, 2007, 17, 675-7.	0.8	21
42	Normalization of echocardiographically derived paediatric cardiac dimensions to body surface area: time for a standardized approach. European Journal of Echocardiography, 2009, 10, 44-45.	2.3	21
43	Echocardiographic reference ranges in older children and adolescents in sub-Saharan Africa. International Journal of Cardiology, 2017, 248, 409-413.	1.7	20
44	The role of the electrocardiographic phenotype in risk stratification for sudden cardiac death in childhood hypertrophic cardiomyopathy. European Journal of Preventive Cardiology, 2022, 29, 645-653.	1.8	20
45	Clinical Features and Natural History of Preadolescent Nonsyndromic HypertrophicÂCardiomyopathy. Journal of the American College of Cardiology, 2022, 79, 1986-1997.	2.8	20
46	ESC EORP Cardiomyopathy Registry: realâ€life practice of genetic counselling and testing in adult cardiomyopathy patients. ESC Heart Failure, 2020, 7, 3013-3021.	3.1	19
47	Cardiac phenotype in ATP1A3 -related syndromes. Neurology, 2020, 95, e2866-e2879.	1.1	19
48	Atypical cardiac defects in patients with RASopathies: Updated data on CARNET study. Birth Defects Research, 2020, 112, 725-731.	1.5	17
49	Clinical outcomes and programming strategies of implantable cardioverter-defibrillator devices in paediatric hypertrophic cardiomyopathy: a UK National Cohort Study. Europace, 2021, 23, 400-408.	1.7	17
50	Feasibility and outcomes of ajmaline provocation testing for Brugada syndrome in children in a specialist paediatric inherited cardiovascular diseases centre. Open Heart, 2014, 1, e000023.	2.3	16
51	Longâ€Term Followâ€Up of Idiopathic Ventricular Fibrillation in a Pediatric Population: Clinical Characteristics, Management, and Complications. Journal of the American Heart Association, 2019, 8, e011172.	3.7	16
52	Racial Variation in Echocardiographic Reference Ranges for Left Chamber Dimensions in Children and Adolescents: A Systematic Review. Pediatric Cardiology, 2018, 39, 859-868.	1.3	15
53	High prevalence of echocardiographic abnormalities in older HIV-infected children taking antiretroviral therapy. Aids, 2018, 32, 2739-2748.	2.2	14
54	Sudden Arrhythmic Death Syndrome: Diagnostic Yield of Comprehensive Clinical Evaluation of Pediatric Firstâ€Degree Relatives. PACE - Pacing and Clinical Electrophysiology, 2014, 37, 1681-1685.	1.2	13

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55	Nomenclature and systems of classification for cardiomyopathy in children. Cardiology in the Young, 2015, 25, 31-42.	0.8	13
56	Psychosocial adjustment and quality of life in children undergoing screening in a specialist paediatric hypertrophic cardiomyopathy clinic. Cardiology in the Young, 2016, 26, 961-967.	0.8	12
57	Friedreichâ€™s ataxia-associated childhood hypertrophic cardiomyopathy: a national cohort study. Archives of Disease in Childhood, 2022, 107, 450-455.	1.9	12
58	Atrial fibrillation, anticoagulation management and risk of stroke in the Cardiomyopathy/Myocarditis registry of the EURObservational Research Programme of the European Society of Cardiology. ESC Heart Failure, 2020, 7, 3601-3609.	3.1	11
59	Association between common cardiovascular risk factors and clinical phenotype in patients with hypertrophic cardiomyopathy from the European Society of Cardiology (ESC) EurObservational Research Programme (EORP) Cardiomyopathy/Myocarditis registry. European Heart Journal Quality of Care & Clinical Outcomes, 2022, 9, 42-53.	4.0	11
60	Current use of cardiac magnetic resonance in tertiary referral centres for the diagnosis of cardiomyopathy: the ESC EORP Cardiomyopathy/Myocarditis Registry. European Heart Journal Cardiovascular Imaging, 2021, 22, 781-789.	1.2	10
61	Childhood Hypertrophic Cardiomyopathy: A Disease of the Cardiac Sarcomere. Frontiers in Pediatrics, 2021, 9, 708679.	1.9	10
62	A new variety of double-chambered left ventricle. European Heart Journal, 2010, 31, 2676-2676.	2.2	9
63	Viral myocarditis in childhood. Paediatrics and Child Health (United Kingdom), 2007, 17, 11-18.	0.4	8
64	Semi-supine Exercise Stress Echocardiography in Children and Adolescents: Feasibility and Safety. Pediatric Cardiology, 2015, 36, 633-639.	1.3	8
65	High prevalence of early repolarization in the paediatric relatives of sudden arrhythmic death syndrome victims and in normal controls. Europace, 2017, 19, 1385-1391.	1.7	8
66	Relationship Between Maximal Left Ventricular Wall Thickness and Sudden Cardiac Death in Childhood Onset Hypertrophic Cardiomyopathy. Circulation: Arrhythmia and Electrophysiology, 2022, 15, CIRCEP121010075.	4.8	8
67	Cardiac myosin binding protein-C variants in paediatric-onset hypertrophic cardiomyopathy: natural history and clinical outcomes. Journal of Medical Genetics, 2022, 59, 768-775.	3.2	7
68	Cardiovascular safety of growth hormone treatment in Noonan syndrome: real-world evidence. Endocrine Connections, 2022, 11, .	1.9	7
69	Data on cardiac defects, morbidity and mortality in patients affected by RASopathies. CARNET study results. Data in Brief, 2018, 16, 649-654.	1.0	6
70	Outcomes following general anaesthesia in children with hypertrophic cardiomyopathy. Archives of Disease in Childhood, 2019, 104, 471-475.	1.9	6
71	Anxiety in children attending a specialist inherited cardiac arrhythmia clinic: a questionnaire study. BMJ Paediatrics Open, 2018, 2, e000271.	1.4	5
72	Resident inflammatory cells in the myocardium of children: On the way to set histologic reference standards to differentiate normal myocardium from myocarditis. International Journal of Cardiology, 2020, 303, 64-65.	1.7	5

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73	Incidence and Progression of Echocardiographic Abnormalities in Older Children with Human Immunodeficiency Virus and Adolescents Taking Antiretroviral Therapy: A Prospective Cohort Study. Clinical Infectious Diseases, 2020, 70, 1372-1378.	5.8	4
74	Childhood-onset hypertrophic cardiomyopathy research coming of age. European Heart Journal, 2021, 42, 1997-1999.	2.2	4
75	The Risk of Sudden Death in Children with Hypertrophic Cardiomyopathy. Heart Failure Clinics, 2022, 18, 9-18.	2.1	4
76	Hypertrophic cardiomyopathy in children. Paediatrics and Child Health (United Kingdom), 2007, 17, 19-24.	0.4	3
77	Carotid intima media thickness in older children and adolescents with HIV taking antiretroviral therapy. Medicine (United States), 2020, 99, e19554.	1.0	3
78	Prospective follow-up in various subtypes of cardiomyopathies: insights from the ESC EORP Cardiomyopathy Registry. European Heart Journal Quality of Care & Clinical Outcomes, 2021, 7, 134-142.	4.0	3
79	Analysis of buccal mucosa as a prognostic tool in children with arrhythmogenic cardiomyopathy. Progress in Pediatric Cardiology, 2022, 64, 101458.	0.4	3
80	Functional effects of DCM mutation G159D in troponin C from an explanted heart. Journal of Molecular and Cellular Cardiology, 2008, 44, 729-730.	1.9	2
81	Restrictive Cardiomyopathy and Hypertrophic Cardiomyopathy Overlap: The Importance of the Phenotype. Neurology International, 2012, 2, e10.	0.5	2
82	Clinical significance of inferolateral early repolarisation and late potentials in children with Brugada Syndrome. Journal of Electrocardiology, 2021, 66, 79-83.	0.9	2
83	Editorial: Paediatric Cardiomyopathies. Frontiers in Pediatrics, 2021, 9, 696443.	1.9	2
84	Risk stratification in childhood hypertrophic cardiomyopathy. Global Cardiology Science & Practice, 2018, 2018, 24.	0.4	2
85	Noncompaction Cardiomyopathy, SickÂSinus Disease, and Aortic Dilatation. JACC: Case Reports, 2022, 4, 287-293.	0.6	2
86	Obliteration of left superior caval vein draining to the left atrium during spontaneous closure of ventricular septal defect. European Journal of Echocardiography, 2009, 10, 160-162.	2.3	1
87	Echocardiographic Diagnosis of Anomalous Origin of the Left Coronary Artery From The Right Coronary Sinus. Pediatric Cardiology, 2013, 34, 2101-2102.	1.3	1
88	How to useâ€¦ the paediatric ECG. Archives of Disease in Childhood: Education and Practice Edition, 2014, 99, 53-60.	0.5	1
89	Cardiomyopathy in children: importance of aetiology in prognosis. Lancet, The, 2014, 383, 781-782.	13.7	1
90	ECG ABNORMALITIES IN ALTERNATING HEMIPLEGIA: A BROADENED PHENOTYPE. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, e4.191-e4.	1.9	1

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91	Value of Stress Transesophageal Echocardiography in an Asymptomatic Patient With Single Coronary Artery From Noncoronary Sinus, Intramural Course, and Ostial Stenosis. Circulation: Cardiovascular Imaging, 2019, 12, e008560.	2.6	1
92	7â€...Relation between N-terminal pro B-type natriuretic peptide (NT-probnp) and disease severity in paediatric hypertrophic cardiomyopathy. , 2021, , .		1
93	Congenital heart disease: an ageing problem. British Journal of Cardiology, 2018, , .	0.2	1
94	Paediatric cardiology â€“ Not just small hearts in small bodies!. International Journal of Cardiology, 2022, , .	1.7	1
95	077 AJMALINE PROVOCATION TESTING FOR BRUGADA SYNDROME IN CHILDREN: THE GREAT ORMOND STREET EXPERIENCE. Heart, 2013, 99, A48.3-A49.	2.9	0
96	Increased Left Ventricular Posterior Wall End-Diastolic Thickness in Adolescents With Delayed Diagnosis of Vertically Acquired HIV Infection. Journal of Acquired Immune Deficiency Syndromes (1999), 2014, 66, e90-e92.	2.1	0
97	CARDIAC FEATURES IN ADULTS WITH ALTERNATING HEMIPLEGIA. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, e4.214-e4.	1.9	0
98	62 * The response of the QT interval to standing in children with long QT syndrome. Europace, 2014, 16, iii23-iii23.	1.7	0
99	The National Undergraduate Paediatrics Conference, 8â€“9 March 2014, Glasgow. Scottish Medical Journal, 2014, 59, e20-e21.	1.3	0
100	Genetic testing for inheritable cardiac channelopathies. British Journal of Hospital Medicine (London,) Tj ETQq0 0 0 rgBT /Overlock 10 Tf	0.5	0
101	4â€...Clinical features and outcomes of childhood hypertrophic cardiomyopathy: a retrospective study in the united kingdom. , 2018, , .		0
102	Long QT syndrome with a functional 2:1 block and multilevel conduction disease. Progress in Pediatric Cardiology, 2018, 50, 46-49.	0.4	0
103	Becker muscular dystrophy associated with sarcomeric hypertrophic cardiomyopathy in a paediatric patient: a case report. European Heart Journal - Case Reports, 2019, 3, ytz117.	0.6	0
104	Rare diseases hiding in the cardiomyopathy clinic - The importance of seeing and observing. International Journal of Cardiology, 2019, 276, 36-37.	1.7	0
105	Concerns About the HCM Risk-Kids Studyâ€"Reply. JAMA Cardiology, 2020, 5, 363.	6.1	0
106	Prevention of sudden cardiac death in childhood-onset hypertrophic cardiomyopathy. Progress in Pediatric Cardiology, 2021, 62, 101412.	0.4	0
107	Inherited Cardiac Muscle Disorders: Hypertrophic and Restrictive Cardiomyopathies. , 2018, , 259-317.		0
108	Prevalence of Inherited Cardiac Conditions in Pediatric First-Degree Relatives of Patients with Idiopathic Ventricular Fibrillation. Pediatric Cardiology, 2022, , 1.	1.3	0

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109	Lessons from rare cardiomyopathies: The importance of a phenotype-based approach to arrive at a specific diagnosis. International Journal of Cardiology, 2022, , .	1.7	0