Juan P Kaski

List of Publications by Citations

92 papers	2,380 citations	24 h-index	47 g-index
116 ext. papers	3,105 ext. citations	5.9 avg, IF	4.39 L-index

#	Paper	IF	Citations
92	De novo mutations in histone-modifying genes in congenital heart disease. *Nature*, **2013**, 498, 220-3	50.4	591
91	Idiopathic restrictive cardiomyopathy in children is caused by mutations in cardiac sarcomere protein genes. *Heart*, **2008**, 94, 1478-84	5.1	148
90	Prevalence of sarcomere protein gene mutations in preadolescent children with hypertrophic cardiomyopathy. *Circulation: Cardiovascular Genetics*, **2009**, 2, 436-41		129
89	The Congenital Heart Disease Genetic Network Study: rationale, design, and early results. *Circulation Research*, **2013**, 112, 698-706	15.7	104
88	Clinical profile of patients with ATP1A3 mutations in Alternating Hemiplegia of Childhood-a study of 155 patients. *Orphanet Journal of Rare Diseases*, **2015**, 10, 123	4.2	83
87	Thioredoxin Reductase 2 (TXNRD2) mutation associated with familial glucocorticoid deficiency (FGD). *Journal of Clinical Endocrinology and Metabolism*, **2014**, 99, E1556-63	5.6	78
86	Long-term outcomes in hypertrophic cardiomyopathy caused by mutations in the cardiac troponin T gene. *Circulation: Cardiovascular Genetics*, **2012**, 5, 10-7		75
85	Development of a Novel Risk Prediction Model for Sudden Cardiac Death in Childhood Hypertrophic Cardiomyopathy (HCM Risk-Kids). *JAMA Cardiology*, **2019**, 4, 918-927	16.2	67
84	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	3.9	62
83	Calmodulin mutations and life-threatening cardiac arrhythmias: insights from the International Calmodulinopathy Registry. *European Heart Journal*, **2019**, 40, 2964-2975	9.5	61
82	The Cardiomyopathy Registry of the EURObservational 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	9.5	60
81	Outcomes after implantable cardioverter-defibrillator treatment in children with hypertrophic cardiomyopathy. *Heart*, **2007**, 93, 372-4	5.1	59
80	Hypertrophic cardiomyopathy in children. *Heart*, **2012**, 98, 1044-54	5.1	53
79	Cardiac defects, morbidity and mortality in patients affected by RASopathies. CARNET study results. *International Journal of Cardiology*, **2017**, 245, 92-98	3.2	48
78	Irbesartan in Marfan syndrome (AIMS): a double-blind, placebo-controlled randomised trial. *Lancet, The*, **2019**, 394, 2263-2270	40	46
77	Clinical presentation and survival of childhood hypertrophic cardiomyopathy: a retrospective study in United Kingdom. *European Heart Journal*, **2019**, 40, 986-993	9.5	38
76	Functional analysis of a unique troponin c mutation, GLY159ASP, that causes familial dilated cardiomyopathy, studied in explanted heart muscle. *Circulation: Heart Failure*, **2009**, 2, 456-64	7.6	37

75	Long-term Safety and Efficacy of Mexiletine for Patients With Skeletal Muscle Channelopathies. *JAMA Neurology*, **2015**, 72, 1531-3	17.2	32
74	Cardiac disease in adolescents with delayed diagnosis of vertically acquired HIV infection. *Clinical Infectious Diseases*, **2013**, 56, 576-82	11.6	30
73	Penetrance of Hypertrophic Cardiomyopathy in Sarcomere Protein Mutation Carriers. *Journal of the American College of Cardiology*, **2020**, 76, 550-559	15.1	30
72	Yield of Clinical Screening for Hypertrophic Cardiomyopathy in Child First-Degree Relatives. *Circulation*, **2019**, 140, 184-192	16.7	28
71	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	1.6	27
70	Faulty cardiac repolarization reserve in alternating hemiplegia of childhood broadens the phenotype. *Brain*, **2015**, 138, 2859-74	11.2	26
69	The classification concept of the ESC Working Group on myocardial and pericardial diseases for dilated cardiomyopathy. *Herz*, **2007**, 32, 446-51	2.6	24
68	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	3.9	22
67	Epidemiology and Clinical Aspects of Genetic Cardiomyopathies. *Heart Failure Clinics*, **2018**, 14, 119-128	3.3	22
66	B-type natriuretic peptide predicts disease severity in children with hypertrophic cardiomyopathy. *Heart*, **2008**, 94, 1307-11	5.1	22
65	Genetic Mosaicism in Calmodulinopathy. *Circulation Genomic and Precision Medicine*, **2019**, 12, 375-385	5.2	20
64	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, 317-26		19
63	Can atrioventricular septal defects exist with intact septal structures?. *Heart*, **2006**, 92, 832-5	5.1	19
62	SCN5A mutations in 442 neonates and children: genotype-phenotype correlation and identification of higher-risk subgroups. *European Heart Journal*, **2018**, 39, 2879-2887	9.5	18
61	Mutations in the cardiac Troponin C gene are a cause of idiopathic dilated cardiomyopathy in childhood. *Cardiology in the Young*, **2007**, 17, 675-7	1	17
60	Clinical Features and Natural History of PRKAG2 Variant Cardiac Glycogenosis. *Journal of the American College of Cardiology*, **2020**, 76, 186-197	15.1	16
59	An International Multicenter Evaluation of Type 5 Long QT Syndrome: A Low Penetrant Primary Arrhythmic Condition. *Circulation*, **2020**, 141, 429-439	16.7	15
58	Echocardiographic reference ranges in older children and adolescents in sub-Saharan Africa. *International Journal of Cardiology*, **2017**, 248, 409-413	3.2	13

57	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	3	13
56	Sudden arrhythmic death syndrome: diagnostic yield of comprehensive clinical evaluation of pediatric first-degree relatives. *PACE - Pacing and Clinical Electrophysiology*, **2014**, 37, 1681-5	1.6	11
55	Long-Term Follow-Up of Idiopathic Ventricular Fibrillation in a Pediatric Population: Clinical Characteristics, Management, and Complications. *Journal of the American Heart Association*, **2019**, 8, e011172	6	10
54	Clinical Profile of Cardiac Involvement in Danon Disease: A Multicenter European Registry. *Circulation Genomic and Precision Medicine*, **2020**, 13, e003117	5.2	10
53	Nomenclature and systems of classification for cardiomyopathy in children. *Cardiology in the Young*, **2015**, 25 Suppl 2, 31-42	1	10
52	Racial Variation in Echocardiographic Reference Ranges for Left Chamber Dimensions in Children and Adolescents: A Systematic Review. *Pediatric Cardiology*, **2018**, 39, 859-868	2.1	9
51	Multidisciplinary evaluation and management of obstructive hypertrophic cardiomyopathy in 2020: Towards the HCM Heart Team. *International Journal of Cardiology*, **2020**, 304, 86-92	3.2	9
50	The role of the electrocardiographic phenotype in risk stratification for sudden cardiac death in childhood hypertrophic cardiomyopathy. *European Journal of Preventive Cardiology*, **2021**,	3.9	9
49	High prevalence of echocardiographic abnormalities in older HIV-infected children taking antiretroviral therapy. *Aids*, **2018**, 32, 2739-2748	3.5	9
48	Somatic mutations of GNA11 and GNAQ in CTNNB1-mutant aldosterone-producing adenomas presenting in puberty, pregnancy or menopause. *Nature Genetics*, **2021**, 53, 1360-1372	36.3	9
47	Clinical outcomes and programming strategies of implantable cardioverter-defibrillator devices in paediatric hypertrophic cardiomyopathy: a UK National Cohort Study. *Europace*, **2021**, 23, 400-408	3.9	8
46	High prevalence of early repolarization in the paediatric relatives of sudden arrhythmic death syndrome victims and in normal controls. *Europace*, **2017**, 19, 1385-1391	3.9	7
45	Semi-supine exercise stress echocardiography in children and adolescents: feasibility and safety. *Pediatric Cardiology*, **2015**, 36, 633-9	2.1	7
44	Atypical cardiac defects in patients with RASopathies: Updated data on CARNET study. *Birth Defects Research*, **2020**, 112, 725-731	2.9	6
43	A new variety of double-chambered left ventricle. *European Heart Journal*, **2010**, 31, 2676	9.5	6
42	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	3.7	6
41	Cardiac phenotype in -related syndromes: A multicenter cohort study. *Neurology*, **2020**, 95, e2866-e2879	6.5	6
40	Data on cardiac defects, morbidity and mortality in patients affected by RASopathies. CARNET study results. *Data in Brief*, **2018**, 16, 649-654	1.2	5

39	Viral myocarditis in childhood. *Paediatrics and Child Health (United Kingdom)*, **2007**, 17, 11-18	0.6	5
38	External validation of the HCM Risk-Kids model for predicting sudden cardiac death in childhood hypertrophic cardiomyopathy. *European Journal of Preventive Cardiology*, **2021**,	3.9	5
37	Psychosocial adjustment and quality of life in children undergoing screening in a specialist paediatric hypertrophic cardiomyopathy clinic. *Cardiology in the Young*, **2016**, 26, 961-7	1	4
36	Differences between familial and sporadic dilated cardiomyopathy: ESC EORP Cardiomyopathy & Myocarditis registry. *ESC Heart Failure*, **2021**, 8, 95-105	3.7	4
35	Clinical presentation and long-term outcomes of infantile hypertrophic cardiomyopathy: a European multicentre study. *ESC Heart Failure*, **2021**,	3.7	4
34	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**,	9.5	3
33	ESC EORP Cardiomyopathy Registry: real-life practice of genetic counselling and testing in adult cardiomyopathy patients. *ESC Heart Failure*, **2020**, 7, 3013-3021	3.7	3
32	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	11.6	3
31	Restrictive cardiomyopathy and hypertrophic cardiomyopathy overlap: the importance of the phenotype. *Neurology International*, **2012**, 2, 10	0	2
30	Hypertrophic cardiomyopathy in children. *Paediatrics and Child Health (United Kingdom)*, **2007**, 17, 19-24	0.6	2
29	Outcomes following general anaesthesia in children with hypertrophic cardiomyopathy. *Archives of Disease in Childhood*, **2019**, 104, 471-475	2.2	2
28	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	4.1	2
27	Anxiety in children attending a specialist inherited cardiac arrhythmia clinic: a questionnaire study. *BMJ Paediatrics Open*, **2018**, 2, e000271	2.4	2
26	Carotid intima media thickness in older children and adolescents with HIV taking antiretroviral therapy. *Medicine (United States)*, **2020**, 99, e19554	1.8	1
25	Echocardiographic diagnosis of anomalous origin of the left coronary artery from the right coronary sinus. *Pediatric Cardiology*, **2013**, 34, 2101-2	2.1	1
24	ECG ABNORMALITIES IN ALTERNATING HEMIPLEGIA: A BROADENED PHENOTYPE. *Journal of Neurology, Neurosurgery and Psychiatry*, **2015**, 86, e4.191-e4	5.5	1
23	How to use...the paediatric ECG. *Archives of Disease in Childhood: Education and Practice Edition*, **2014**, 99, 53-60	0.5	1
22	Cardiomyopathy in children: importance of aetiology in prognosis. *Lancet, The*, **2014**, 383, 781-2	40	1

21	Obliteration of left superior caval vein draining to the left atrium during spontaneous closure of ventricular septal defect. *European Journal of Echocardiography*, **2009**, 10, 160-2		1
20	Clinical significance of inferolateral early repolarisation and late potentials in children with Brugada Syndrome. *Journal of Electrocardiology*, **2021**, 66, 79-83	1.4	1
19	Clinical Features and Natural History of Preadolescent Nonsyndromic Hypertrophic Cardiomyopathy.. *Journal of the American College of Cardiology*, **2022**, 79, 1986-1997	15.1	1
18	The Risk of Sudden Death in Children with Hypertrophic Cardiomyopathy. *Heart Failure Clinics*, **2022**, 18, 9-18	3.3	0
17	Risk stratification in childhood hypertrophic cardiomyopathy. *Global Cardiology Science & Practice*, **2018**, 2018, 24	0.7	0
16	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	4.6	0
15	Childhood Hypertrophic Cardiomyopathy: A Disease of the Cardiac Sarcomere. *Frontiers in Pediatrics*, **2021**, 9, 708679	3.4	0
14	Relationship Between Maximal Left Ventricular Wall Thickness and Sudden Cardiac Death in Childhood Onset Hypertrophic Cardiomyopathy.. *Circulation: Arrhythmia and Electrophysiology*, **2022**, CIRCEP121010075	6.4	0
13	Concerns About the HCM Risk-Kids Study-Reply. *JAMA Cardiology*, **2020**, 5, 363-364	16.2	
12	Becker muscular dystrophy associated with sarcomeric hypertrophic cardiomyopathy in a paediatric patient: a case report. *European Heart Journal - Case Reports*, **2019**, 3, ytz117	0.9	
11	62 * The response of the QT interval to standing in children with long QT syndrome. *Europace*, **2014**, 16, iii23-iii23	3.9	
10	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, e90-2	3.1	
9	CARDIAC FEATURES IN ADULTS WITH ALTERNATING HEMIPLEGIA. *Journal of Neurology, Neurosurgery and Psychiatry*, **2014**, 85, e4.214-e4	5.5	
8	077 AJMALINE PROVOCATION TESTING FOR BRUGADA SYNDROME IN CHILDREN: THE GREAT ORMOND STREET EXPERIENCE. *Heart*, **2013**, 99, A48.3-A49	5.1	
7	Prevalence of Inherited Cardiac Conditions in Pediatric First-Degree Relatives of Patients with Idiopathic Ventricular Fibrillation.. *Pediatric Cardiology*, **2022**, 1	2.1	
6	Inherited Cardiac Muscle Disorders: Hypertrophic and Restrictive Cardiomyopathies **2018**, 259-317		
5	Genetic testing for inheritable cardiac channelopathies. *British Journal of Hospital Medicine (London, England: 2005)*, **2016**, 77, 294-302	0.8	
4	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	3.9	

List of Publications

3	Long QT syndrome with a functional 2:1 block and multilevel conduction disease. *Progress in Pediatric Cardiology*, **2018**, 50, 46-49	0.4
2	Prevention of sudden cardiac death in childhood-onset hypertrophic cardiomyopathy. *Progress in Pediatric Cardiology*, **2021**, 62, 101412	0.4
1	Noncompaction Cardiomyopathy, Sick Sinus Disease, and Aortic Dilatation: Too Much for a Single Diagnosis?. *JACC: Case Reports*, **2022**, 4, 287-293	1.2