List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	Hypertrophic Cardiomyopathy in RASopathies. Heart Failure Clinics, 2022, 18, 19-29.	2.1	33
2	Social Cognition Impairments in 22q11.2DS Individuals With and Without Psychosis: A Comparison Study With a Large Population of Patients With Schizophrenia. Schizophrenia Bulletin Open, 2022, 3, .	1.7	3
3	The Secrets of the Frogs Heart. Pediatric Cardiology, 2022, 43, 1471-1480.	1.3	6
4	Congenital heart defects in molecularly confirmed <scp>KBG</scp> syndrome patients. American Journal of Medical Genetics, Part A, 2022, 188, 1149-1159.	1.2	5
5	Crossed pulmonary arteries: An underestimated cardiovascular variant with a strong association with genetic syndromes—A report of 74 cases with systematic review of the literature. American Journal of Medical Genetics, Part A, 2022, 188, 2351-2359.	1.2	3
6	Anatomically corrected malposition of the great arteries (S,L,D) with mutation of Nodal gene. Cardiology in the Young, 2022, , 1-3.	0.8	0
7	22q11.2 Deletion Syndrome: Impact of Genetics in the Treatment of Conotruncal Heart Defects. Children, 2022, 9, 772.	1.5	7
8	Anatomical substrate for biventricular repair in patients with left isomerism. Annals of Pediatric Cardiology, 2021, 14, 250.	0.5	0
9	Atrioventricular canal defect is the classic congenital heart disease in Bardet–Biedl syndrome. Annals of Human Genetics, 2021, 85, 101-102.	0.8	3
10	Smith–Magenis syndrome: Report of morphological and new functional cardiac findings with review of the literature. American Journal of Medical Genetics, Part A, 2021, 185, 2003-2011.	1.2	5
11	Copy number variation analysis implicates novel pathways in patients with oculoâ€auriculoâ€vertebralâ€spectrum and congenital heart defects. Clinical Genetics, 2021, 100, 268-279.	2.0	9
12	Cardiac Defects and Genetic Syndromes: Old Uncertainties and New Insights. Genes, 2021, 12, 1047.	2.4	12
13	Clinical Manifestations of 22q11.2 Deletion Syndrome. Heart Failure Clinics, 2021, 18, 155-164.	2.1	15
14	Commentary: sVEGFR1 Is Enriched in Hepatic Vein Blood—Evidence for a Provisional Hepatic Factor Candidate?. Frontiers in Pediatrics, 2021, 9, 782779.	1.9	0
15	Complete Sequence of the 22q11.2 Allele in 1,053 Subjects with 22q11.2 Deletion Syndrome Reveals Modifiers of Conotruncal Heart Defects. American Journal of Human Genetics, 2020, 106, 26-40.	6.2	42
16	Atrioventricular canal defect as partial expression of heterotaxia in patients with Bardet-Biedl syndrome. Journal of Pediatrics, 2020, 218, 263-264.	1.8	4
17	Common atrium/atrioventricular canal defect and postaxial polydactyly: A mild clinical subtype of Ellisâ€van Creveld syndrome caused by hypomorphic mutations in the <i>EVC</i> gene. Human Mutation, 2020, 41, 2087-2093.	2.5	7
18	Genetics of atrioventricular canal defects. Italian Journal of Pediatrics, 2020, 46, 61.	2.6	9

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19	Atypical cardiac defects in patients with RASopathies: Updated data on CARNET study. Birth Defects Research, 2020, 112, 725-731.	1.5	17
20	KBG syndrome: Common and uncommon clinical features based on 31 new patients. American Journal of Medical Genetics, Part A, 2020, 182, 1073-1083.	1.2	27
21	Differences in morbidity and mortality in Down syndrome are related to the type of congenital heart defect. American Journal of Medical Genetics, Part A, 2020, 182, 1342-1350.	1.2	19
22	Pediatric patients with RASopathy-associated hypertrophic cardiomyopathy: the multifaceted consequences of PTPN11 mutations. Orphanet Journal of Rare Diseases, 2019, 14, 163.	2.7	5
23	Prevalence, Type, and Molecular Spectrum of NF1 Mutations in Patients with Neurofibromatosis Type 1 and Congenital Heart Disease. Genes, 2019, 10, 675.	2.4	13
24	Familial aggregation of "apple peel―intestinal atresia and cardiac leftâ€sided obstructive lesions: A possible causal relationship with <i>NOTCH1</i> gene mutations. American Journal of Medical Genetics, Part A, 2019, 179, 1570-1574.	1.2	4
25	Left pulmonary artery in 22q11.2 deletion syndrome. Echocardiographic evaluation in patients without cardiac defects and role of Tbx1 in mice. PLoS ONE, 2019, 14, e0211170.	2.5	13
26	Congenital heart diseases and cardiovascular abnormalities in 22q11.2 deletion syndrome: From wellâ€established knowledge to new frontiers. American Journal of Medical Genetics, Part A, 2018, 176, 2087-2098.	1.2	57
27	Late arrhytmias after repair of atrioventricular septal defect: Down's Syndrome is not the culprit. International Journal of Cardiology, 2018, 254, 162-163.	1.7	0
28	Primary lymphedema and other lymphatic anomalies are associated with 22q11.2 deletion syndrome. European Journal of Medical Genetics, 2018, 61, 411-415.	1.3	5
29	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
30	Clinical Presentation and Natural History of Hypertrophic Cardiomyopathy in RASopathies. Heart Failure Clinics, 2018, 14, 225-235.	2.1	44
31	Cardiovascular disease in Down syndrome. Current Opinion in Pediatrics, 2018, 30, 616-622.	2.0	58
32	Long-term survival and phenotypic spectrum in heterotaxy syndrome: A 25-year follow-up experience. International Journal of Cardiology, 2018, 268, 100-105.	1.7	24
33	Heterozygous missense mutations in <i>NFATC1</i> are associated with atrioventricular septal defect. Human Mutation, 2018, 39, 1428-1441.	2.5	15
34	Some Isolated Cardiac Malformations Can Be Related to Laterality Defects. Journal of Cardiovascular Development and Disease, 2018, 5, 24.	1.6	15
35	Congenital heart defects in molecularly proven Kabuki syndrome patients. American Journal of Medical Genetics, Part A, 2017, 173, 2912-2922.	1.2	60
36	Congenital heart disease and genetic syndromes: new insights into molecular mechanisms. Expert Review of Molecular Diagnostics, 2017, 17, 861-870.	3.1	39

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37	Cardiac defects, morbidity and mortality in patients affected by RASopathies. CARNET study results. International Journal of Cardiology, 2017, 245, 92-98.	1.7	75
38	What Is New in Genetics of Congenital Heart Defects?. Frontiers in Pediatrics, 2016, 4, 120.	1.9	29
39	Sprengel anomaly in deletion 22q11.2 (DiGeorge/Velo–Cardio–Facial) syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 661-664.	1.2	3
40	Congenital heart defects in Noonan syndrome and RIT1 mutation. Genetics in Medicine, 2016, 18, 1320.	2.4	16
41	Rare copy number variants and congenital heart defects in the 22q11.2 deletion syndrome. Human Genetics, 2016, 135, 273-285.	3.8	43
42	22q11.2 deletion syndrome. Nature Reviews Disease Primers, 2015, 1, 15071.	30.5	954
43	Left ventricular non compaction with aortic valve anomalies: A recurrent feature of 22q11.2 distal deletion syndrome. European Journal of Medical Genetics, 2015, 58, 406-408.	1.3	1
44	Copy-Number Variation of the Clucose Transporter Gene SLC2A3 and Congenital Heart Defects in the 22q11.2 Deletion Syndrome. American Journal of Human Genetics, 2015, 96, 753-764.	6.2	62
45	Clinical Features and Follow-Up in Patients with 22q11.2 Deletion Syndrome. Journal of Pediatrics, 2014, 164, 1475-1480.e2.	1.8	119
46	<i>JAG1</i> Mutation in a patient with deletion 22q11.2 syndrome and tetralogy of Fallot. American Journal of Medical Genetics, Part A, 2013, 161, 3133-3136.	1.2	9
47	Novel and recurrent EVC and EVC2 mutations in Ellis-van Creveld syndrome and Weyers acrofacial dyostosis. European Journal of Medical Genetics, 2013, 56, 80-87.	1.3	64
48	Atrioventricular canal defect in patients with RASopathies. European Journal of Human Genetics, 2013, 21, 200-204.	2.8	22
49	Association of DiGeorge anomaly and caudal dysplasia sequence in a neonate born to a diabetic mother. Cardiology in the Young, 2013, 23, 14-17.	0.8	6
50	A variant in the carboxyl-terminus of connexin 40 alters GAP junctions and increases risk for tetralogy of Fallot. European Journal of Human Genetics, 2013, 21, 69-75.	2.8	36
51	Transposition of great arteries: new insights into the pathogenesis. Frontiers in Pediatrics, 2013, 1, 11.	1.9	77
52	Spiral shapes in heart and shells: when form and function do matter. European Journal of Cardio-thoracic Surgery, 2012, 41, 473-475.	1.4	4
53	The heart and shell. Anatomical and genetic similarities. American Heart Journal, 2011, 161, 647-649.	2.7	5
54	Atrioventricular Canal Defect and Associated Genetic Disorders: New Insights into Polydactyly Syndromes. Neurology International, 2011, 1, e7.	0.5	6

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55	22q11.2 Deletion (DiGeorge) Syndrome: A Mother's Open Letter. Neurology International, 2011, 1, e11.	0.5	Ο
56	Cardiopulmonary Response to Exercise and Cardiac Assessment in Patients With Turner Syndrome. American Journal of Cardiology, 2011, 107, 1076-1082.	1.6	20
57	Clinical, pathological, and molecular analyses of cardiovascular abnormalities in Costello syndrome: A Ras/MAPK pathway syndrome. American Journal of Medical Genetics, Part A, 2011, 155, 486-507.	1.2	99
58	Genetic dosage compensation in a family with velo ardioâ€facial/DiGeorge/22q11.2 deletion syndrome. American Journal of Medical Genetics, Part A, 2011, 155, 548-554.	1.2	10
59	SOS1 mutations in Noonan syndrome: molecular spectrum, structural insights on pathogenic effects, and genotype-phenotype correlations. Human Mutation, 2011, 32, 760-772.	2.5	97
60	Screening for 22q11.2 microdeletion in adults with tetralogy of Fallot. Heart, 2011, 97, 860-860.	2.9	2
61	Hif1î± down-regulation is associated with transposition of great arteries in mice treated with a retinoic acid antagonist. BMC Genomics, 2010, 11, 497.	2.8	20
62	Shells and heart: Are human laterality and chirality of snails controlled by the same maternal genes?. American Journal of Medical Genetics, Part A, 2010, 152A, 2419-2425.	1.2	28
63	Familial transposition of the great arteries caused by multiple mutations in laterality genes. Heart, 2010, 96, 673-677.	2.9	126
64	Multiplex Ligation-Dependent Probe Amplification Analysis of <i>GATA4</i> Gene Copy Number Variations in Patients with Isolated Congenital Heart Disease. Disease Markers, 2010, 28, 287-292.	1.3	8
65	Impact of DEL22q11, trisomy 21, and other genetic syndromes on surgical outcome of conotruncal heart defects. Journal of Thoracic and Cardiovascular Surgery, 2009, 138, 565-570.e2.	0.8	66
66	Three patients with oculoâ€auriculoâ€vertebral spectrum and microdeletion 22q11.2. American Journal of Medical Genetics, Part A, 2009, 149A, 2860-2864.	1.2	56
67	Germline <i>BRAF</i> mutations in Noonan, LEOPARD, and cardiofaciocutaneous syndromes: Molecular diversity and associated phenotypic spectrum. Human Mutation, 2009, 30, 695-702.	2.5	251
68	Genetic syndromes and congenital heart defects: how is surgical management affected?. European Journal of Cardio-thoracic Surgery, 2009, 35, 606-614.	1.4	88
69	Costello syndrome: clinical diagnosis in the first year of life. European Journal of Pediatrics, 2008, 167, 621-628.	2.7	43
70	Cardiac defects and results of cardiac surgery in 22q11.2 deletion syndrome. Developmental Disabilities Research Reviews, 2008, 14, 35-42.	2.9	99
71	Genotype–phenotype analysis and natural history of left ventricular hypertrophy in LEOPARD syndrome. American Journal of Medical Genetics, Part A, 2008, 146A, 620-628.	1.2	47
72	Severe, early onset hypertrophic cardiomyopathy in a family with LEOPARD syndrome. Journal of Prenatal Medicine, 2008, 2, 24-6.	0.2	5

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73	Genetics of congenital heart diseases in syndromic and non-syndromic patients: new advances and clinical implications. Journal of Cardiovascular Medicine, 2007, 8, 7-11.	1.5	10
74	Gain-of-function RAF1 mutations cause Noonan and LEOPARD syndromes with hypertrophic cardiomyopathy. Nature Genetics, 2007, 39, 1007-1012.	21.4	624
75	Prevalence and Clinical Significance of Cardiovascular Abnormalities in Patients With the LEOPARD Syndrome. American Journal of Cardiology, 2007, 100, 736-741.	1.6	150
76	Familial recurrence of discrete membranous subaortic stenosis. Journal of Thoracic and Cardiovascular Surgery, 2007, 134, 818-819.	0.8	11
77	Genetic Syndromes and Outcome After Surgical Correction of Tetralogy of Fallot. Annals of Thoracic Surgery, 2006, 81, 968-975.	1.3	104
78	Diffuse coronary dilation in a young patient with LEOPARD syndrome. International Journal of Cardiology, 2006, 112, E35-E37.	1.7	11
79	22q11 Deletion syndrome: a review of some developmental biology aspects of the cardiovascular system. Journal of Cardiovascular Medicine, 2006, 7, 77-85.	1.5	3
80	Genetic heterogeneity and phenotypic anomalies in children with atrioventricular canal defect and tetralogy of Fallot. Clinical Dysmorphology, 2006, 15, 65-70.	0.3	27
81	PTPN11 gene mutations: linking the Gln510Glu mutation to the "LEOPARD syndrome phenotype― European Journal of Pediatrics, 2006, 165, 803-805.	2.7	33
82	Familial recurrence of congenital heart disease: an overview and review of the literature. European Journal of Pediatrics, 2006, 166, 111-116.	2.7	76
83	LEOPARD syndrome: Clinical diagnosis in the first year of life. American Journal of Medical Genetics, Part A, 2006, 140A, 740-746.	1.2	129
84	Familial recurrence of anomalous origin of right pulmonary artery from the aorta. American Journal of Medical Genetics, Part A, 2006, 140A, 794-796.	1.2	0
85	Additional evidence thatPTPN11 mutations play only a minor role in the pathogenesis of non-syndromic atrioventricular canal defect. American Journal of Medical Genetics, Part A, 2006, 140A, 1970-1972.	1.2	4
86	Congenital cardiovascular disease and velo-cardio-facial syndrome. , 2005, , 47-82.		6
87	ZFPM2/FOG2 andHEY2 genes analysis in nonsyndromic tricuspid atresia. American Journal of Medical Genetics, Part A, 2005, 133A, 68-70.	1.2	19
88	Congenital intrahepatic portosystemic venous shunt: An unusual feature in LEOPARD syndrome and in neurofibromatosis type 1. American Journal of Medical Genetics, Part A, 2005, 134A, 457-458.	1.2	12
89	Familial recurrence of nonsyndromic congenital heart defects in first degree relatives of patients with deletion 22q11.2. American Journal of Medical Genetics, Part A, 2005, 134A, 158-164.	1.2	13
90	Familial recurrence of heart defects in subjects with congenitally corrected transposition of the great arteries. American Journal of Medical Genetics, Part A, 2005, 137A, 176-180.	1.2	38

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91	NF1 Gene Mutations Represent the Major Molecular Event Underlying Neurofibromatosis-Noonan Syndrome. American Journal of Human Genetics, 2005, 77, 1092-1101.	6.2	139
92	Familial aggregation of genetically heterogeneous hypertrophic cardiomyopathy: A boy with LEOPARD syndrome due to PTPN11 mutation and his nonsyndromic father lacking PTPN11 mutations. Birth Defects Research Part A: Clinical and Molecular Teratology, 2004, 70, 95-98.	1.6	14
93	Ellis-van Creveld Syndrome with hydrometrocolpos is not linked to chromosome arm 4p or 20p. American Journal of Medical Genetics Part A, 2004, 126A, 319-323.	2.4	5
94	Mutations ofZFPM2/FOG2 gene in sporadic cases of tetralogy of Fallot. Human Mutation, 2003, 22, 372-377.	2.5	127
95	Nonsyndromic Pulmonary Valve Stenosis and thePTPN11 Gene. American Journal of Medical Genetics Part A, 2003, 116A, 389-390.	2.4	15
96	Truncus arteriosus and duplication 8q. American Journal of Medical Genetics Part A, 2003, 121A, 79-81.	2.4	9
97	DiGeorge subtypes of nonsyndromic conotruncal defects: evidence against a major role of TBX1 Gene. European Journal of Human Genetics, 2003, 11, 349-351.	2.8	48
98	Tetralogy of fallot with aortic valvular stenosis and deletion 22q11. Annals of Thoracic Surgery, 2003, 75, 2010-2011.	1.3	6
99	Grouping of Multiple-Lentigines/LEOPARD and Noonan Syndromes on the PTPN11 Gene. American Journal of Human Genetics, 2002, 71, 389-394.	6.2	380
100	Common arterial trunk, DiGeorge syndrome and microdeletion 22q11. Progress in Pediatric Cardiology, 2002, 15, 9-17.	0.4	16
101	Transposition of the great arteries in asplenia and polysplenia phenotypes. American Journal of Medical Genetics Part A, 2002, 110, 292-294.	2.4	33
102	Ventricular septal defect and deletion of chromosome 22q11: anatomical types and aortic arch anomalies. European Journal of Pediatrics, 2002, 161, 116-117.	2.7	21
103	Genetic predisposition to ventricular septal defect in Down syndrome. Human Genetics, 2001, 109, 463-463.	3.8	6
104	Congenital heart defects in Kabuki syndrome. American Journal of Medical Genetics Part A, 2001, 100, 269-274.	2.4	105
105	Anatomic patterns of conotruncal defects associated with deletion 22q11. Genetics in Medicine, 2001, 3, 45-48.	2.4	135
106	Complete Transposition of the Great Arteries. Circulation, 2001, 104, 2809-2814.	1.6	113
107	Familial recurrence of nonsyndromic interrupted aortic arch and truncus arteriosus with atrioventricular canal. , 2000, 61, 329-331.		13
108	Heterotaxy with left atrial isomerism in a patient with deletion 18p. American Journal of Medical Genetics Part A, 2000, 94, 198-200.	2.4	25

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109	Mutations in a new gene in Ellis-van Creveld syndrome and Weyers acrodental dysostosis. Nature Genetics, 2000, 24, 283-286.	21.4	323
110	Loss-of-function mutations in the EGF-CFC gene CFC1 are associated with human left-right laterality defects. Nature Genetics, 2000, 26, 365-369.	21.4	319
111	Deletion 22q11 in patients with interrupted aortic arch. American Journal of Cardiology, 1999, 84, 360-361.	1.6	53
112	Cardiac malformations in patients with oral-facial-skeletal syndromes: Clinical similarities with heterotaxia. American Journal of Medical Genetics Part A, 1999, 84, 350-356.	2.4	79
113	Genetic heterogeneity of isolated noncompaction of the left ventricular myocardium. , 1999, 85, 90-91.		40
114	Atrioventricular canal defect without Down syndrome: A heterogeneous malformation. American Journal of Medical Genetics Part A, 1999, 85, 140-146.	2.4	66
115	Inlet ventricular septal defect is not a partial atrioventricular septal defect. , 1999, 87, 195-195.		4
116	Atrioventricular canal defect and hypoplastic left heart syndrome as discordant congenital heart defects in twins. Teratology, 1999, 60, 206-208.	1.6	7
117	Congenital heart diseases in children with Noonan syndrome: An expanded cardiac spectrum with high prevalence of atrioventricular canal. Journal of Pediatrics, 1999, 135, 703-706.	1.8	216
118	Morphology of the Atrioventricular Valve in Patients with Right Atrial Isomerism. Pediatric Cardiology, 1998, 19, 501-501.	1.3	0
119	Deletion 8p syndrome. , 1998, 75, 534-536.		59
120	Noonan syndrome and aortic coarctation. , 1998, 80, 160-162.		42
121	Congenital heart defect in sibs with discordant karyotypes. , 1998, 80, 169-172.		13
122	Deletion 8p syndrome. American Journal of Medical Genetics Part A, 1998, 75, 534-536.	2.4	2
123	Conotruncal heart defects and chromosome 22q11microdeletion. Journal of Pediatrics, 1997, 130, 675-676.	1.8	12
124	Atrioventricular canal defect and postaxial polydactyly indicating phenotypic overlap of Ellis-van Creveld and Kaufman-McKusick syndromes. Pediatric Cardiology, 1997, 18, 74-75.	1.3	21
125	The atrioventricular canal defect is the congenital heart disease connecting short rib-polydactyly and oral-facial-digital syndromes. , 1997, 68, 110-112.		13

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127	Familial recurrence of transposition of the great arteries and intact ventricular septum. , 1997, 73, 93-94.		5
128	Familial deletions of chromosome 22q11. , 1997, 73, 95-96.		25
129	Progeroid syndrome with characteristic facial appearance and hand anomalies in father and son. , 1997, 73, 227-229.		7
130	Intrafamilial variability of Pfeiffer-type cardiocranial syndrome. , 1997, 73, 480-483.		6
131	Morphology of the atrioventricular valve in asplenia syndrome: A peculiar type of atrioventricular canal defect. Cardiovascular Pathology, 1996, 5, 145-151.	1.6	15
132	The mendelian basis of congenital heart defects. Cardiology in the Young, 1996, 6, 264-271.	0.8	13
133	Search for 22q11 deletion in non-syndromic conotruncal cardiac defects. European Journal of Pediatrics, 1996, 155, 619-620.	2.7	14
134	Associated cardiac anomalies in isolated and syndromic patients with tetralogy of fallot. American Journal of Cardiology, 1996, 77, 505-508.	1.6	73
135	Comparison of occurrence of genetic syndromes in ventricular septal defect with pulmonic stenosis (classic tetralogy of Fallot) versus ventricular septal defect with pulmonic atresia. American Journal of Cardiology, 1996, 77, 1375-1376.	1.6	80
136	Exclusion of 22qll deletion in noonan syndrome with tetralogy of fallot. American Journal of Medical Genetics Part A, 1996, 62, 413-414.	2.4	8
137	Transposition of the great arteries associated with deletion of chromosome 22q11. American Journal of Cardiology, 1995, 75, 95-98.	1.6	54
138	Maternal diabetes causing DiGeorge anomaly and renal agenesis. American Journal of Medical Genetics Part A, 1995, 55, 513-514.	2.4	32
139	Atrioventricular canal and 3C (cranio-cerebello-cardiac) syndrome. American Journal of Medical Genetics Part A, 1995, 58, 97-98.	2.4	17
140	Single atrium, atrioventricular canal/postaxial hexodactyly indicating Ellis-van Creveld syndrome. Human Genetics, 1995, 96, 251-253.	3.8	32
141	22q11 deletions in isolated and syndromic patients with tetralogy of Fallot. Human Genetics, 1995, 95, 479-82.	3.8	117
142	Atrioventricular canal and 8p- syndrome. American Journal of Medical Genetics Part A, 1993, 47, 437-438.	2.4	38
143	Cardiac Catheterization through the Internal Jugular Vein in Pediatric Patients. Chest, 1992, 101, 1512-1514.	0.8	18
144	Atrioventricular septal defect—anatomic characteristics in patients with and without Down's syndrome. Cardiology in the Young, 1992, 2, 308-310.	0.8	14

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145	Atrioventricular septal defects with severe left ventricular hypoplasia—clinical findings and surgical options. Cardiology in the Young, 1992, 2, 53-55.	0.8	9
146	Nonrandom association of atrioventricular canal and del (8p) syndrome. American Journal of Medical Genetics Part A, 1992, 42, 424-427.	2.4	58
147	Di George Anomaly with Atrioventricular Canal. Chest, 1991, 99, 242-243.	0.8	11
148	Atrioventricular Canal in Down Syndrome. American Journal of Diseases of Children, 1990, 144, 1120.	0.5	128
149	Bidirectional cavopulmonary shunts: Clinical applications as staged or definitive palliation. Annals of Thoracic Surgery, 1989, 47, 415-420.	1.3	108