

Elizabeth Goldmuntz

List of Publications by Year in descending order

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Version: 2024-02-01

150
papers

9,998
citations

61687

45
h-index

45040

94
g-index

153
all docs

153
docs citations

153
times ranked

11846
citing authors

#	ARTICLE	IF	CITATIONS
1	De novo mutations in histone-modifying genes in congenital heart disease. <i>Nature</i> , 2013, 498, 220-223.	13.7	798
2	Genetic Basis for Congenital Heart Defects: Current Knowledge. <i>Circulation</i> , 2007, 115, 3015-3038.	1.6	719
3	De novo mutations in congenital heart disease with neurodevelopmental and other congenital anomalies. <i>Science</i> , 2015, 350, 1262-1266.	6.0	646
4	Contribution of rare inherited and de novo variants in 2,871 congenital heart disease probands. <i>Nature Genetics</i> , 2017, 49, 1593-1601.	9.4	624
5	Frequency of 22q11 deletions in patients with conotruncal defects. <i>Journal of the American College of Cardiology</i> , 1998, 32, 492-498.	1.2	527
6	NKX2.5 mutations in patients with congenital heart disease. <i>Journal of the American College of Cardiology</i> , 2003, 42, 1650-1655.	1.2	347
7	NKX2.5 Mutations in Patients With Tetralogy of Fallot. <i>Circulation</i> , 2001, 104, 2565-2568.	1.6	316
8	High-resolution mapping and analysis of copy number variations in the human genome: A data resource for clinical and research applications. <i>Genome Research</i> , 2009, 19, 1682-1690.	2.4	313
9	Analysis of Cardiovascular Phenotype and Genotype-Phenotype Correlation in Individuals With a JAG1 Mutation and/or Alagille Syndrome. <i>Circulation</i> , 2002, 106, 2567-2574.	1.6	273
10	Cognitive and behavior profile of preschool children with chromosome 22q11.2 deletion. <i>American Journal of Medical Genetics Part A</i> , 1999, 85, 127-133.	2.4	263
11	Association of chromosome 22q11 deletion with isolated anomalies of aortic arch laterality and branching. <i>Journal of the American College of Cardiology</i> , 2001, 37, 2114-2119.	1.2	237
12	Increased Frequency of De Novo Copy Number Variants in Congenital Heart Disease by Integrative Analysis of Single Nucleotide Polymorphism Array and Exome Sequence Data. <i>Circulation Research</i> , 2014, 115, 884-896.	2.0	229
13	CFC1 Mutations in Patients with Transposition of the Great Arteries and Double-Outlet Right Ventricle. <i>American Journal of Human Genetics</i> , 2002, 70, 776-780.	2.6	182
14	Missense Mutations and Gene Interruption in PROSIT240 , a Novel TRAP240 -Like Gene, in Patients With Congenital Heart Defect (Transposition of the Great Arteries). <i>Circulation</i> , 2003, 108, 2843-2850.	1.6	154
15	The 22q11.2 Deletion: Screening, Diagnostic Workup, and Outcome of Results; Report on 181 Patients. <i>Genetic Testing and Molecular Biomarkers</i> , 1997, 1, 99-108.	1.7	150
16	The Congenital Heart Disease Genetic Network Study. <i>Circulation Research</i> , 2013, 112, 698-706.	2.0	142
17	Jagged1 mutations in patients ascertained with isolated congenital heart defects. , 1999, 84, 56-60.		137
18	Adults with genetic syndromes and cardiovascular abnormalities: clinical history and management. <i>Genetics in Medicine</i> , 2008, 10, 469-494.	1.1	130

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19	Quantifying Pulmonary Regurgitation and Right Ventricular Function in Surgically Repaired Tetralogy of Fallot. <i>Circulation: Cardiovascular Imaging</i> , 2012, 5, 637-643.	1.3	129
20	CANOES: detecting rare copy number variants from whole exome sequencing data. <i>Nucleic Acids Research</i> , 2014, 42, e97-e97.	6.5	123
21	Deletion of ETS-1, a gene in the Jacobsen syndrome critical region, causes ventricular septal defects and abnormal ventricular morphology in mice. <i>Human Molecular Genetics</i> , 2010, 19, 648-656.	1.4	118
22	DiGeorge Syndrome: New Insights. <i>Clinics in Perinatology</i> , 2005, 32, 963-978.	0.8	109
23	What is new with 22q? An update from the 22q and You Center at the Children's Hospital of Philadelphia. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2058-2069.	0.7	106
24	Genomic analyses implicate noncoding de novo variants in congenital heart disease. <i>Nature Genetics</i> , 2020, 52, 769-777.	9.4	97
25	DNAH6 and Its Interactions with PCD Genes in Heterotaxy and Primary Ciliary Dyskinesia. <i>PLoS Genetics</i> , 2016, 12, e1005821.	1.5	92
26	Recessive DNAH9 Loss-of-Function Mutations Cause Laterality Defects and Subtle Respiratory Ciliary-Beating Defects. <i>American Journal of Human Genetics</i> , 2018, 103, 995-1008.	2.6	92
27	22q11.2 Deletions in Patients with Conotruncal Defects: Data from 1,610 Consecutive Cases. <i>Pediatric Cardiology</i> , 2013, 34, 1687-1694.	0.6	88
28	THE EPIDEMIOLOGY AND GENETICS OF CONGENITAL HEART DISEASE. <i>Clinics in Perinatology</i> , 2001, 28, 1-10.	0.8	85
29	Microdeletions and Microduplications in Patients with Congenital Heart Disease and Multiple Congenital Anomalies. <i>Congenital Heart Disease</i> , 2011, 6, 592-602.	0.0	82
30	Trends in Pulmonary Valve Replacement in Children and Adults With Tetralogy of Fallot. <i>American Journal of Cardiology</i> , 2015, 115, 118-124.	0.7	82
31	The Congenital Heart Disease Genetic Network Study: Cohort description. <i>PLoS ONE</i> , 2018, 13, e0191319.	1.1	82
32	Lean mass deficits, vitamin D status and exercise capacity in children and young adults after Fontan palliation. <i>Heart</i> , 2014, 100, 1702-1707.	1.2	80
33	22q11.2 Deletion syndrome is associated with perioperative outcome in tetralogy of Fallot. <i>Journal of Thoracic and Cardiovascular Surgery</i> , 2013, 146, 868-873.	0.4	71
34	Cumulative ligand activity of NODAL mutations and modifiers are linked to human heart defects and holoprosencephaly. <i>Molecular Genetics and Metabolism</i> , 2009, 98, 225-234.	0.5	67
35	Aortic root dilation in patients with 22q11.2 deletion syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 939-942.	0.7	66
36	22q11.2 deletion syndrome and congenital heart disease. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2020, 184, 64-72.	0.7	66

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37	Chromosome 22q11 Deletion in Patients With Ventricular Septal Defect: Frequency and Associated Cardiovascular Anomalies. <i>Pediatrics</i> , 2003, 112, e472-e476.	1.0	64
38	22q11.2 Deletion syndrome is associated with increased perioperative events and more complicated postoperative course in infants undergoing infant operative correction of truncus arteriosus communis or interrupted aortic arch. <i>Journal of Thoracic and Cardiovascular Surgery</i> , 2014, 148, 1597-1605.	0.4	62
39	Copy-Number Variation of the Glucose Transporter Gene SLC2A3 and Congenital Heart Defects in the 22q11.2 Deletion Syndrome. <i>American Journal of Human Genetics</i> , 2015, 96, 753-764.	2.6	62
40	Variants of Folate Metabolism Genes and the Risk of Conotruncal Cardiac Defects. <i>Circulation: Cardiovascular Genetics</i> , 2008, 1, 126-132.	5.1	61
41	Mouse and Human CRKL Is Dosage Sensitive for Cardiac Outflow Tract Formation. <i>American Journal of Human Genetics</i> , 2015, 96, 235-244.	2.6	58
42	De novo and recessive forms of congenital heart disease have distinct genetic and phenotypic landscapes. <i>Nature Communications</i> , 2019, 10, 4722.	5.8	58
43	Congenital heart diseases and cardiovascular abnormalities in 22q11.2 deletion syndrome: From well-established knowledge to new frontiers. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2087-2098.	0.7	57
44	DiGeorge anomaly with renal agenesis in infants of mothers with diabetes. <i>American Journal of Medical Genetics Part A</i> , 1993, 47, 1078-1082.	2.4	56
45	Loss of RNA expression and allele-specific expression associated with congenital heart disease. <i>Nature Communications</i> , 2016, 7, 12824.	5.8	51
46	22q11.2 Deletion Status and Disease Burden in Children and Adolescents With Tetralogy of Fallot. <i>Circulation: Cardiovascular Genetics</i> , 2015, 8, 74-81.	5.1	50
47	2-Year Outcomes After Complete or Staged Procedure for Tetralogy of Fallot in Neonates. <i>Journal of the American College of Cardiology</i> , 2019, 74, 1570-1579.	1.2	49
48	Rare copy number variants in over 100,000 European ancestry subjects reveal multiple disease associations. <i>Nature Communications</i> , 2020, 11, 255.	5.8	48
49	Genome-Wide Association Studies and Meta-Analyses for Congenital Heart Defects. <i>Circulation: Cardiovascular Genetics</i> , 2017, 10, e001449.	5.1	47
50	Chromosomal and cardiovascular anomalies associated with congenital laryngeal web. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2002, 66, 23-27.	0.4	46
51	Deficits in bone density and structure in children and young adults following Fontan palliation. <i>Bone</i> , 2015, 77, 12-16.	1.4	45
52	Correlation between abnormal cardiac physical examination and echocardiographic findings in neonates with Down syndrome. <i>American Journal of Medical Genetics Part A</i> , 2002, 113, 238-241.	2.4	44
53	Rare copy number variants and congenital heart defects in the 22q11.2 deletion syndrome. <i>Human Genetics</i> , 2016, 135, 273-285.	1.8	43
54	Robust identification of mosaic variants in congenital heart disease. <i>Human Genetics</i> , 2018, 137, 183-193.	1.8	43

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55	Complete Sequence of the 22q11.2 Allele in 1,053 Subjects with 22q11.2 Deletion Syndrome Reveals Modifiers of Conotruncal Heart Defects. <i>American Journal of Human Genetics</i> , 2020, 106, 26-40.	2.6	42
56	Association of Damaging Variants in Genes With Increased Cancer Risk Among Patients With Congenital Heart Disease. <i>JAMA Cardiology</i> , 2021, 6, 457.	3.0	34
57	Habitual Exercise Correlates With Exercise Performance in Patients With Conotruncal Abnormalities. <i>Pediatric Cardiology</i> , 2013, 34, 853-860.	0.6	32
58	The phenotypic spectrum of <i>ZIC3</i> mutations includes isolated dâ€¢transposition of the great arteries and double outlet right ventricle. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 792-802.	0.7	32
59	Morbidity in children and adolescents after surgical correction of truncus arteriosus communis. <i>American Heart Journal</i> , 2013, 166, 512-518.	1.2	31
60	Risk of congenital heart disease in relatives of probands with conotruncal cardiac defects: An evaluation of 1,620 families. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 1490-1495.	0.7	31
61	GATA6 mutations in hiPSCs inform mechanisms for maldevelopment of the heart, pancreas, and diaphragm. <i>ELife</i> , 2020, 9, .	2.8	31
62	Predictors of Length of Hospital Stay After Complete Repair for Tetralogy of Fallot: A Prospective Cohort Study. <i>Journal of the American Heart Association</i> , 2018, 7, .	1.6	30
63	Heterozygous loss-of-function variants of MEIS2 cause a triad of palatal defects, congenital heart defects, and intellectual disability. <i>European Journal of Human Genetics</i> , 2019, 27, 278-290.	1.4	30
64	De Novo Damaging Variants, Clinical Phenotypes, and Post-Operative Outcomes in Congenital Heart Disease. <i>Circulation Genomic and Precision Medicine</i> , 2020, 13, e002836.	1.6	30
65	Gene-Gene Interactions in the Folate Metabolic Pathway and the Risk of Conotruncal Heart Defects. <i>Journal of Biomedicine and Biotechnology</i> , 2010, 2010, 1-7.	3.0	29
66	Genetic counseling for congenital heart disease: New approaches for a new decade. <i>Current Cardiology Reports</i> , 2002, 4, 68-75.	1.3	27
67	Mechanisms of Congenital Heart Disease Caused by NAA15 Haploinsufficiency. <i>Circulation Research</i> , 2021, 128, 1156-1169.	2.0	27
68	Rare genetic variation at transcription factor binding sites modulates local DNA methylation profiles. <i>PLoS Genetics</i> , 2020, 16, e1009189.	1.5	27
69	Genome-Wide Association Study of Maternal and Inherited Loci for Conotruncal Heart Defects. <i>PLoS ONE</i> , 2014, 9, e96057.	1.1	26
70	22q11.2 deletion syndrome as a risk factor for aortic root dilation in tetralogy of Fallot. <i>Cardiology in the Young</i> , 2014, 24, 303-310.	0.4	26
71	MESP1 Mutations in Patients with Congenital Heart Defects. <i>Human Mutation</i> , 2016, 37, 308-314.	1.1	26
72	Utility of genetic evaluation in infants with congenital heart defects admitted to the cardiac intensive care unit. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 3090-3097.	0.7	26

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73	Longitudinal Changes in Right Ventricular Function in Tetralogy of Fallot in the Initial Years after Surgical Repair. <i>Journal of the American Society of Echocardiography</i> , 2018, 31, 816-821.	1.2	25
74	Staged Versus Complete Repair in the Symptomatic Neonate With Tetralogy of Fallot. <i>Annals of Thoracic Surgery</i> , 2020, 109, 802-808.	0.7	25
75	Evaluation of potential modifiers of the cardiac phenotype in the 22q11.2 deletion syndrome. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2009, 85, 125-129.	1.6	24
76	Genome-wide association study of maternal and inherited effects on left-sided cardiac malformations. <i>Human Molecular Genetics</i> , 2015, 24, 265-273.	1.4	24
77	Association of Habitual Activity and Body Mass Index in Survivors of Congenital Heart Surgery: A Study of Children and Adolescents With Tetralogy of Fallot, Transposition of the Great Arteries, and Fontan Palliation. <i>World Journal for Pediatric & Congenital Heart Surgery</i> , 2018, 9, 177-184.	0.3	23
78	Genome-Wide Association Study to Find Modifiers for Tetralogy of Fallot in the 22q11.2 Deletion Syndrome Identifies Variants in the <i>GPR98</i> Locus on 5q14.3. <i>Circulation: Cardiovascular Genetics</i> , 2017, 10, .	5.1	22
79	Diastolic dysfunction in tetralogy of Fallot: Comparison of echocardiography with catheterization. <i>Echocardiography</i> , 2018, 35, 1641-1648.	0.3	21
80	The Impact of Pulmonary Insufficiency on the Right Ventricle: A Comparison of Isolated Valvar Pulmonary Stenosis and Tetralogy of Fallot. <i>Pediatric Cardiology</i> , 2015, 36, 796-801.	0.6	20
81	The genetic contribution to congenital heart disease. <i>Pediatric Clinics of North America</i> , 2004, 51, 1721-1737.	0.9	18
82	Mutations in <i>NTRK3</i> Suggest a Novel Signaling Pathway in Human Congenital Heart Disease. <i>Human Mutation</i> , 2014, 35, 1459-1468.	1.1	17
83	EM-mosaic detects mosaic point mutations that contribute to congenital heart disease. <i>Genome Medicine</i> , 2020, 12, 42.	3.6	17
84	Cardiac Magnetic Resonance Imaging for Accurate Diagnosis of Aortic Arch Anomalies in Patients with 22q11.2 Deletion. <i>American Journal of Cardiology</i> , 2005, 96, 1726-1730.	0.7	16
85	Right Ventricular Contractile Reserve Is Impaired in Children and Adolescents With Repaired Tetralogy of Fallot: An Exercise Strain Imaging Study. <i>Journal of the American Society of Echocardiography</i> , 2019, 32, 135-144.	1.2	16
86	Recent advances in understanding the genetic etiology of congenital heart disease. <i>Current Opinion in Pediatrics</i> , 1999, 11, 437-444.	1.0	15
87	Morbidity in Children and Adolescents After Surgical Correction of Interrupted Aortic Arch. <i>Pediatric Cardiology</i> , 2014, 35, 386-392.	0.6	15
88	Rare copy number variants in patients with congenital conotruncal heart defects. <i>Birth Defects Research</i> , 2017, 109, 271-295.	0.8	15
89	Exercise Performance and 22q11.2 Deletion Status Affect Quality of Life in Tetralogy of Fallot. <i>Journal of Pediatrics</i> , 2017, 189, 162-168.	0.9	15
90	Gene-based genome-wide association studies and meta-analyses of conotruncal heart defects. <i>PLoS ONE</i> , 2019, 14, e0219926.	1.1	15

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91	Systems Analysis Implicates WAVE2-Complex in the Pathogenesis of Developmental Left-Sided Obstructive Heart Defects. <i>JACC Basic To Translational Science</i> , 2020, 5, 376-386.	1.9	15
92	Common Genetic Variants Contribute to Risk of Transposition of the Great Arteries. <i>Circulation Research</i> , 2022, 130, 166-180.	2.0	15
93	Longitudinal changes in adolescents with TOF: implications for care. <i>European Heart Journal Cardiovascular Imaging</i> , 2017, 18, 356-363.	0.5	14
94	Identifying genetic factors that contribute to the increased risk of congenital heart defects in infants with Down syndrome. <i>Scientific Reports</i> , 2020, 10, 18051.	1.6	14
95	Molecular characterization and investigation of the role of genetic variation in phenotypic variability and response to treatment in a large pediatric Marfan syndrome cohort. <i>Genetics in Medicine</i> , 2022, 24, 1045-1053.	1.1	13
96	Variants of folate metabolism genes and risk of left-sided cardiac defects. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2010, 88, 48-53.	1.6	12
97	Analysis of chromosomal structural variation in patients with congenital left-sided cardiac lesions. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2014, 100, 951-964.	1.6	12
98	Relationship Between Habitual Exercise and Performance on Cardiopulmonary Exercise Testing Differs Between Children With Single and Biventricular Circulations. <i>Pediatric Cardiology</i> , 2017, 38, 472-483.	0.6	12
99	22q11.2 Deletion Status and Perioperative Outcomes for Tetralogy of Fallot with Pulmonary Atresia and Multiple Aortopulmonary Collateral Vessels. <i>Pediatric Cardiology</i> , 2018, 39, 906-910.	0.6	12
100	Resource Utilization in the First 2 Years Following Operative Correction for Tetralogy of Fallot: Study Using Data From the Optum's De-identified Clinformatics Data Mart Insurance Claims Database. <i>Journal of the American Heart Association</i> , 2020, 9, e016581.	1.6	12
101	Determinants of Exercise Performance in Children and Adolescents with Repaired Tetralogy of Fallot Using Stress Echocardiography. <i>Pediatric Cardiology</i> , 2019, 40, 71-78.	0.6	11
102	<i>NAT1</i> , <i>NOS3</i> , and <i>TYMS</i> genotypes and the risk of conotruncal cardiac defects. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2011, 91, 61-65.	1.6	10
103	Evaluation of heterogeneity in the association between congenital heart defects and variants of folate metabolism genes: Conotruncal and left-sided cardiac defects. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2011, 91, 879-884.	1.6	10
104	The Cardiovascular Effects of Obesity on Ventricular Function and Mass in Patients after Tetralogy of Fallot Repair. <i>Journal of Pediatrics</i> , 2015, 167, 325-330.e1.	0.9	10
105	The Impact of the Right Ventricular Outflow Tract Patch on Right Ventricular Strain in Tetralogy of Fallot: A Comparison with Valvar Pulmonary Stenosis Utilizing Cardiac Magnetic Resonance. <i>Pediatric Cardiology</i> , 2017, 38, 617-623.	0.6	10
106	Procollagen type III amino-terminal propeptide: a serum biomarker of left ventricular remodelling in paediatric dilated cardiomyopathy. <i>Cardiology in the Young</i> , 2015, 25, 228-236.	0.4	9
107	Adrenergic receptor genotype influences heart failure severity and β -blocker response in children with dilated cardiomyopathy. <i>Pediatric Research</i> , 2015, 77, 363-369.	1.1	8
108	Longitudinal Validation of the Diastolic to Systolic Time-Velocity Integral Ratio as a Doppler-Derived Measure of Pulmonary Regurgitation in Patients with Repaired Tetralogy of Fallot. <i>Pediatric Cardiology</i> , 2017, 38, 240-246.	0.6	8

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109	The association of elevated maternal genetic risk scores for hypertension, type 2 diabetes and obesity and having a child with a congenital heart defect. <i>PLoS ONE</i> , 2019, 14, e0216477.	1.1	8
110	Variants in <i>ADRB1</i> and <i>CYP2C9</i> : Association with Response to Atenolol and Losartan in Marfan Syndrome. <i>Journal of Pediatrics</i> , 2020, 222, 213-220.e5.	0.9	8
111	Gene-based analyses of the maternal genome implicate maternal effect genes as risk factors for conotruncal heart defects. <i>PLoS ONE</i> , 2020, 15, e0234357.	1.1	8
112	Cerebrovascular Malformations in a Pediatric Hereditary Hemorrhagic Telangiectasia Cohort. <i>Pediatric Neurology</i> , 2020, 110, 49-54.	1.0	8
113	Genome-Wide De Novo Variants in Congenital Heart Disease Are Not Associated With Maternal Diabetes or Obesity. <i>Circulation Genomic and Precision Medicine</i> , 2022, 15, CIRCGEN121003500.	1.6	8
114	Late effects in survivors of high-risk neuroblastoma following stem cell transplant with and without total body irradiation. <i>Pediatric Blood and Cancer</i> , 2022, 69, e29537.	0.8	8
115	Identifying Risk Factors for Complicated Post-operative Course in Tetralogy of Fallot Using a Machine Learning Approach. <i>Frontiers in Cardiovascular Medicine</i> , 2021, 8, 685855.	1.1	7
116	Cardiac evaluation of patients with 22q11.2 duplication syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 753-758.	0.7	7
117	Early postoperative remodelling following repair of tetralogy of Fallot utilising unседated cardiac magnetic resonance: a pilot study. <i>Cardiology in the Young</i> , 2018, 28, 697-701.	0.4	6
118	Invited Commentary: The Hunt for Mechanistic Origins of Liver Fibrosis in the Fontan Circulation. <i>World Journal for Pediatric & Congenital Heart Surgery</i> , 2021, 12, 173-175.	0.3	6
119	Aortic Root Dilation in Patients with 22q11.2 Deletion Syndrome Without Intracardiac Anomalies. <i>Pediatric Cardiology</i> , 2021, 42, 1594-1600.	0.6	6
120	Genetic Testing in Congenital Heart Disease. <i>World Journal for Pediatric & Congenital Heart Surgery</i> , 2013, 4, 53-57.	0.3	5
121	Adult CHD: the ongoing need for physician counselling about heredity and contraceptive options. <i>Cardiology in the Young</i> , 2017, 27, 671-676.	0.4	5
122	Quality of Life is Diminished in Patients with Tetralogy of Fallot with Mild Residual Disease: A Comparison of Tetralogy of Fallot and Isolated Valvar Pulmonary Stenosis. <i>Pediatric Cardiology</i> , 2017, 38, 1645-1653.	0.6	5
123	Genetic variants of <i>HIF1α</i> are associated with right ventricular fibrotic load in repaired tetralogy of Fallot patients: a cardiovascular magnetic resonance study. <i>Journal of Cardiovascular Magnetic Resonance</i> , 2019, 21, 51.	1.6	5
124	Neither cardiac mitochondrial DNA variation nor copy number contribute to congenital heart disease risk. <i>American Journal of Human Genetics</i> , 2022, 109, 961-966.	2.6	5
125	The Cardiovascular Manifestations of Alagille Syndrome and <i>JAG1</i> Mutations. , 2006, 126, 217-232.		3
126	Preconceptual Folic Acid Use and Recurrence Risk Counseling for Congenital Heart Disease. <i>Congenital Heart Disease</i> , 2015, 10, 219-225.	0.0	3

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127	Robust identification of deletions in exome and genome sequence data based on clustering of Mendelian errors. <i>Human Mutation</i> , 2018, 39, 870-881.	1.1	3
128	Copy number variations in individuals with conotruncal heart defects reveal some shared developmental pathways irrespective of 22q11.2 deletion status. <i>Birth Defects Research</i> , 2019, 111, 888-905.	0.8	3
129	<i>ANKK2</i> related congenital heart disease: Biallelic homeodomain-disrupting variants and truncus arteriosus. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1454-1459.	0.7	3
130	Marked skeletal muscle deficits are associated with 6-minute walk distance in paediatric pulmonary hypertension. <i>Cardiology in the Young</i> , 2021, 31, 1426-1433.	0.4	3
131	Natural history of the aortic root in Tetralogy of Fallot after repair. <i>International Journal of Cardiology Congenital Heart Disease</i> , 2021, 3, 100092.	0.2	3
132	Exercise is medicine in the Fontan circulation. <i>International Journal of Cardiology</i> , 2021, 343, 50-52.	0.8	3
133	The Prevalence of 16p12.1 Microdeletion in Patients with Left-sided Cardiac Lesions. <i>Congenital Heart Disease</i> , 2014, 9, 83-86.	0.0	2
134	Common Variation in Cytoskeletal Genes Is Associated with Conotruncal Heart Defects. <i>Genes</i> , 2021, 12, 655.	1.0	2
135	Reported practice patterns in the ambulatory care setting for patients with CHD. <i>Cardiology in the Young</i> , 2022, 32, 1421-1426.	0.4	2
136	Gene-Interaction-Sensitive enrichment analysis in congenital heart disease. <i>BioData Mining</i> , 2022, 15, 4.	2.2	2
137	Maternal effect genes as risk factors for congenital heart defects. <i>Human Genetics and Genomics Advances</i> , 2022, 3, 100098.	1.0	2
138	Comparison of serum biomarkers of myocardial fibrosis with cardiac magnetic resonance in patients operated for tetralogy of Fallot. <i>International Journal of Cardiology</i> , 2022, 358, 27-33.	0.8	2
139	The 22q11.2 Deletion Syndrome. , 0, , 100-111.		1
140	X-chromosome association studies of congenital heart defects. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 250-254.	0.7	1
141	Right Ventricular Strain Is Associated with Increased Length of Stay after Tetralogy of Fallot Repair. <i>Journal of Cardiovascular Imaging</i> , 2022, 29, 50-58.	0.2	1
142	Genome-Wide Association Studies of Conotruncal Heart Defects with Normally Related Great Vessels in the United States. <i>Genes</i> , 2021, 12, 1030.	1.0	1
143	Townes-Brocks Syndrome. , 2009, , 2092-2094.		0
144	Cover Image, Volume 176A, Number 10, October 2018. , 2018, 176, i-i.		0

#	ARTICLE	IF	CITATIONS
145	The Molecular Genetics of Conotruncal Defects. <i>Developments in Cardiovascular Medicine</i> , 2000, , 355-374.	0.1	0
146	The Genetics of Congenital Heart Disease. , 2006, , 145-157.		0
147	Title is missing!. , 2020, 15, e0234357.		0
148	Title is missing!. , 2020, 15, e0234357.		0
149	Title is missing!. , 2020, 15, e0234357.		0
150	Title is missing!. , 2020, 15, e0234357.		0