

# Elizabeth Goldmuntz

## List of Publications by Citations

**Source:** <https://exaly.com/author-pdf/8587168/elizabeth-goldmuntz-publications-by-citations.pdf>

**Version:** 2024-04-18

This document has been generated based on the publications and citations recorded by exaly.com. For the latest version of this publication list, visit the link given above.

The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

146  
papers

7,366  
citations

41  
h-index

84  
g-index

153  
ext. papers

8,932  
ext. citations

6.3  
avg, IF

5.4  
L-index

#	Paper	IF	Citations
146	Genetic basis for congenital heart defects: current knowledge: a scientific statement from the American Heart Association Congenital Cardiac Defects Committee, Council on Cardiovascular Disease in the Young: endorsed by the American Academy of Pediatrics. <i>Circulation</i> , <b>2007</b> , 115, 3015-38	16.7	613
145	De novo mutations in histone-modifying genes in congenital heart disease. <i>Nature</i> , <b>2013</b> , 498, 220-3	50.4	591
144	Frequency of 22q11 deletions in patients with conotruncal defects. <i>Journal of the American College of Cardiology</i> , <b>1998</b> , 32, 492-8	15.1	457
143	De novo mutations in congenital heart disease with neurodevelopmental and other congenital anomalies. <i>Science</i> , <b>2015</b> , 350, 1262-6	33.3	406
142	Contribution of rare inherited and de novo variants in 2,871 congenital heart disease probands. <i>Nature Genetics</i> , <b>2017</b> , 49, 1593-1601	36.3	348
141	NKX2.5 mutations in patients with congenital heart disease. <i>Journal of the American College of Cardiology</i> , <b>2003</b> , 42, 1650-5	15.1	295
140	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> , <b>2009</b> , 19, 1682-90	9.7	293
139	NKX2.5 mutations in patients with tetralogy of fallot. <i>Circulation</i> , <b>2001</b> , 104, 2565-8	16.7	275
138	Cognitive and behavior profile of preschool children with chromosome 22q11.2 deletion. <i>American Journal of Medical Genetics Part A</i> , <b>1999</b> , 85, 127-133		235
137	Analysis of cardiovascular phenotype and genotype-phenotype correlation in individuals with a JAG1 mutation and/or Alagille syndrome. <i>Circulation</i> , <b>2002</b> , 106, 2567-74	16.7	227
136	Association of chromosome 22q11 deletion with isolated anomalies of aortic arch laterality and branching. <i>Journal of the American College of Cardiology</i> , <b>2001</b> , 37, 2114-9	15.1	204
135	CFC1 mutations in patients with transposition of the great arteries and double-outlet right ventricle. <i>American Journal of Human Genetics</i> , <b>2002</b> , 70, 776-80	11	164
134	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> , <b>2014</b> , 115, 884-896	15.7	158
133	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> , <b>2003</b> , 108, 2843-50	16.7	141
132	The 22q11.2 deletion: screening, diagnostic workup, and outcome of results; report on 181 patients. <i>Genetic Testing and Molecular Biomarkers</i> , <b>1997</b> , 1, 99-108		124
131	Jagged1 mutations in patients ascertained with isolated congenital heart defects <b>1999</b> , 84, 56-60		113
130	Adults with genetic syndromes and cardiovascular abnormalities: clinical history and management. <i>Genetics in Medicine</i> , <b>2008</b> , 10, 469-94	8.1	106

129	The Congenital Heart Disease Genetic Network Study: rationale, design, and early results. <i>Circulation Research</i> , <b>2013</b> , 112, 698-706	15.7	104
128	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> , <b>2010</b> , 19, 648-56	5.6	93
127	Quantifying pulmonary regurgitation and right ventricular function in surgically repaired tetralogy of Fallot: a comparative analysis of echocardiography and magnetic resonance imaging. <i>Circulation: Cardiovascular Imaging</i> , <b>2012</b> , 5, 637-43	3.9	89
126	DiGeorge syndrome: new insights. <i>Clinics in Perinatology</i> , <b>2005</b> , 32, 963-78, ix-x	2.8	88
125	CANOES: detecting rare copy number variants from whole exome sequencing data. <i>Nucleic Acids Research</i> , <b>2014</b> , 42, e97	20.1	75
124	22q11.2 deletions in patients with conotruncal defects: data from 1,610 consecutive cases. <i>Pediatric Cardiology</i> , <b>2013</b> , 34, 1687-94	2.1	72
123	Trends in pulmonary valve replacement in children and adults with tetralogy of fallot. <i>American Journal of Cardiology</i> , <b>2015</b> , 115, 118-24	3	68
122	The epidemiology and genetics of congenital heart disease. <i>Clinics in Perinatology</i> , <b>2001</b> , 28, 1-10	2.8	67
121	Microdeletions and microduplications in patients with congenital heart disease and multiple congenital anomalies. <i>Congenital Heart Disease</i> , <b>2011</b> , 6, 592-602	3.1	63
120	22q11.2 Deletion syndrome is associated with perioperative outcome in tetralogy of Fallot. <i>Journal of Thoracic and Cardiovascular Surgery</i> , <b>2013</b> , 146, 868-73	1.5	61
119	Cumulative ligand activity of NODAL mutations and modifiers are linked to human heart defects and holoprosencephaly. <i>Molecular Genetics and Metabolism</i> , <b>2009</b> , 98, 225-34	3.7	61
118	Recessive DNAH9 Loss-of-Function Mutations Cause Laterality Defects and Subtle Respiratory Ciliary-Beating Defects. <i>American Journal of Human Genetics</i> , <b>2018</b> , 103, 995-1008	11	61
117	Lean mass deficits, vitamin D status and exercise capacity in children and young adults after Fontan palliation. <i>Heart</i> , <b>2014</b> , 100, 1702-7	5.1	60
116	DNAH6 and Its Interactions with PCD Genes in Heterotaxy and Primary Ciliary Dyskinesia. <i>PLoS Genetics</i> , <b>2016</b> , 12, e1005821	6	58
115	Variants of folate metabolism genes and the risk of conotruncal cardiac defects. <i>Circulation: Cardiovascular Genetics</i> , <b>2008</b> , 1, 126-32		57
114	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> , <b>2015</b> , 96, 753-64	11	54
113	Aortic root dilation in patients with 22q11.2 deletion syndrome. <i>American Journal of Medical Genetics, Part A</i> , <b>2009</b> , 149A, 939-42	2.5	54
112	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> , <b>2018</b> , 176, 2058-2069	2.5	54

111	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> , <b>2014</b> , 148, 1597-605	1.5	51
110	Chromosome 22q11 deletion in patients with ventricular septal defect: frequency and associated cardiovascular anomalies. <i>Pediatrics</i> , <b>2003</b> , 112, e472	7.4	51
109	DiGeorge anomaly with renal agenesis in infants of mothers with diabetes. <i>American Journal of Medical Genetics Part A</i> , <b>1993</b> , 47, 1078-82		48
108	Mouse and human CRKL is dosage sensitive for cardiac outflow tract formation. <i>American Journal of Human Genetics</i> , <b>2015</b> , 96, 235-44	11	47
107	The Congenital Heart Disease Genetic Network Study: Cohort description. <i>PLoS ONE</i> , <b>2018</b> , 13, e0191319	3.7	43
106	Chromosomal and cardiovascular anomalies associated with congenital laryngeal web. <i>International Journal of Pediatric Otorhinolaryngology</i> , <b>2002</b> , 66, 23-27	1.7	41
105	22q11.2 deletion status and disease burden in children and adolescents with tetralogy of Fallot. <i>Circulation: Cardiovascular Genetics</i> , <b>2015</b> , 8, 74-81		37
104	Correlation between abnormal cardiac physical examination and echocardiographic findings in neonates with Down syndrome. <i>American Journal of Medical Genetics Part A</i> , <b>2002</b> , 113, 238-41		36
103	Deficits in bone density and structure in children and young adults following Fontan palliation. <i>Bone</i> , <b>2015</b> , 77, 12-6	4.7	35
102	Genomic analyses implicate noncoding de novo variants in congenital heart disease. <i>Nature Genetics</i> , <b>2020</b> , 52, 769-777	36.3	33
101	Loss of RNA expression and allele-specific expression associated with congenital heart disease. <i>Nature Communications</i> , <b>2016</b> , 7, 12824	17.4	33
100	Rare copy number variants and congenital heart defects in the 22q11.2 deletion syndrome. <i>Human Genetics</i> , <b>2016</b> , 135, 273-85	6.3	31
99	The phenotypic spectrum of ZIC3 mutations includes isolated d-transposition of the great arteries and double outlet right ventricle. <i>American Journal of Medical Genetics, Part A</i> , <b>2013</b> , 161A, 792-802	2.5	30
98	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> , <b>2018</b> , 176, 2087-2098	2.5	29
97	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> , <b>2014</b> , 164A, 1490-5	2.5	29
96	Robust identification of mosaic variants in congenital heart disease. <i>Human Genetics</i> , <b>2018</b> , 137, 183-193	6.3	26
95	Morbidity in children and adolescents after surgical correction of truncus arteriosus communis. <i>American Heart Journal</i> , <b>2013</b> , 166, 512-8	4.9	26
94	De novo and recessive forms of congenital heart disease have distinct genetic and phenotypic landscapes. <i>Nature Communications</i> , <b>2019</b> , 10, 4722	17.4	25

93	Habitual exercise correlates with exercise performance in patients with conotruncal abnormalities. <i>Pediatric Cardiology</i> , <b>2013</b> , 34, 853-60	2.1	25
92	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> , <b>2020</b> , 106, 26-40	11	24
91	MESP1 Mutations in Patients with Congenital Heart Defects. <i>Human Mutation</i> , <b>2016</b> , 37, 308-14	4.7	24
90	2-Year Outcomes After Complete or Staged Procedure for Tetralogy of Fallot in Neonates. <i>Journal of the American College of Cardiology</i> , <b>2019</b> , 74, 1570-1579	15.1	23
89	Gene-gene interactions in the folate metabolic pathway and the risk of conotruncal heart defects. <i>Journal of Biomedicine and Biotechnology</i> , <b>2010</b> , 2010, 630940		23
88	Genome-Wide Association Studies and Meta-Analyses for Congenital Heart Defects. <i>Circulation: Cardiovascular Genetics</i> , <b>2017</b> , 10, e001449		22
87	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> , <b>2009</b> , 85, 125-9		21
86	Genetic counseling for congenital heart disease: new approaches for a new decade. <i>Current Cardiology Reports</i> , <b>2002</b> , 4, 68-75	4.2	21
85	Genome-wide association study of maternal and inherited loci for conotruncal heart defects. <i>PLoS ONE</i> , <b>2014</b> , 9, e96057	3.7	20
84	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> , <b>2018</b> , 7,	6	19
83	22q11.2 deletion syndrome and congenital heart disease. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , <b>2020</b> , 184, 64-72	3.1	19
82	Genome-wide association study of maternal and inherited effects on left-sided cardiac malformations. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 265-73	5.6	18
81	22q11.2 deletion syndrome as a risk factor for aortic root dilation in tetralogy of Fallot. <i>Cardiology in the Young</i> , <b>2014</b> , 24, 303-10	1	18
80	Rare copy number variants in over 100,000 European ancestry subjects reveal multiple disease associations. <i>Nature Communications</i> , <b>2020</b> , 11, 255	17.4	17
79	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> , <b>2016</b> , 170, 3090-3097	2.5	17
78	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> , <b>2018</b> , 31, 816-821	5.8	16
77	De Novo Damaging Variants, Clinical Phenotypes, and Post-Operative Outcomes in Congenital Heart Disease. <i>Circulation Genomic and Precision Medicine</i> , <b>2020</b> , 13, e002836	5.2	15
76	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> , <b>2019</b> , 27, 278-290	5.3	15

75	Genome-Wide Association Study to Find Modifiers for Tetralogy of Fallot in the 22q11.2 Deletion Syndrome Identifies Variants in the Locus on 5q14.3. <i>Circulation: Cardiovascular Genetics</i> , <b>2017</b> , 10,		14
74	Diastolic dysfunction in tetralogy of Fallot: Comparison of echocardiography with catheterization. <i>Echocardiography</i> , <b>2018</b> , 35, 1641-1648	1.5	14
73	The impact of pulmonary insufficiency on the right ventricle: a comparison of isolated valvar pulmonary stenosis and tetralogy of fallot. <i>Pediatric Cardiology</i> , <b>2015</b> , 36, 796-801	2.1	14
72	The genetic contribution to congenital heart disease. <i>Pediatric Clinics of North America</i> , <b>2004</b> , 51, 1721-37, x	3.6	14
71	Rare copy number variants in patients with congenital conotruncal heart defects. <i>Birth Defects Research</i> , <b>2017</b> , 109, 271-295	2.9	13
70	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 &amp; Congenital Heart Surgery</i> , <b>2018</b> , 9, 177-184	1.1	12
69	Analysis of chromosomal structural variation in patients with congenital left-sided cardiac lesions. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , <b>2014</b> , 100, 951-64		12
68	Cardiac magnetic resonance imaging for accurate diagnosis of aortic arch anomalies in patients with 22q11.2 deletion. <i>American Journal of Cardiology</i> , <b>2005</b> , 96, 1726-30	3	12
67	Recent advances in understanding the genetic etiology of congenital heart disease. <i>Current Opinion in Pediatrics</i> , <b>1999</b> , 11, 437-43	3.2	12
66	Association of Damaging Variants in Genes With Increased Cancer Risk Among Patients With Congenital Heart Disease. <i>JAMA Cardiology</i> , <b>2021</b> , 6, 457-462	16.2	12
65	Morbidity in children and adolescents after surgical correction of interrupted aortic arch. <i>Pediatric Cardiology</i> , <b>2014</b> , 35, 386-92	2.1	11
64	Staged Versus Complete Repair in the Symptomatic Neonate With Tetralogy of Fallot. <i>Annals of Thoracic Surgery</i> , <b>2020</b> , 109, 802-808	2.7	11
63	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> , <b>2019</b> , 32, 135-144	5.8	11
62	Mutations in NTRK3 suggest a novel signaling pathway in human congenital heart disease. <i>Human Mutation</i> , <b>2014</b> , 35, 1459-68	4.7	10
61	NAT1, NOS3, and TYMS genotypes and the risk of conotruncal cardiac defects. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , <b>2011</b> , 91, 61-5		10
60	Variants of folate metabolism genes and risk of left-sided cardiac defects. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , <b>2010</b> , 88, 48-53		10
59	22q11.2 Deletion Status and Perioperative Outcomes for Tetralogy of Fallot with Pulmonary Atresia and Multiple Aortopulmonary Collateral Vessels. <i>Pediatric Cardiology</i> , <b>2018</b> , 39, 906-910	2.1	9
58	Relationship Between Habitual Exercise and Performance on Cardiopulmonary Exercise Testing Differs Between Children With Single and Biventricular Circulations. <i>Pediatric Cardiology</i> , <b>2017</b> , 38, 472-483	2.1	9

57	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> , <b>2011</b> , 91, 879-84		9
56	mutations in hiPSCs inform mechanisms for maldevelopment of the heart, pancreas, and diaphragm. <i>ELife</i> , <b>2020</b> , 9,	8.9	9
55	EM-mosaic detects mosaic point mutations that contribute to congenital heart disease. <i>Genome Medicine</i> , <b>2020</b> , 12, 42	14.4	8
54	Exercise Performance and 22q11.2 Deletion Status Affect Quality of Life in Tetralogy of Fallot. <i>Journal of Pediatrics</i> , <b>2017</b> , 189, 162-168	3.6	8
53	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> , <b>2017</b> , 38, 240-246	2.1	8
52	The Cardiovascular Effects of Obesity on Ventricular Function and Mass in Patients after Tetralogy of Fallot Repair. <i>Journal of Pediatrics</i> , <b>2015</b> , 167, 325-30.e1	3.6	8
51	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> , <b>2017</b> , 38, 617-623	2.1	7
50	Adrenergic receptor genotype influences heart failure severity and $\beta$ -blocker response in children with dilated cardiomyopathy. <i>Pediatric Research</i> , <b>2015</b> , 77, 363-9	3.2	7
49	Rare genetic variation at transcription factor binding sites modulates local DNA methylation profiles. <i>PLoS Genetics</i> , <b>2020</b> , 16, e1009189	6	7
48	Longitudinal changes in adolescents with TOF: implications for care. <i>European Heart Journal Cardiovascular Imaging</i> , <b>2017</b> , 18, 356-363	4.1	6
47	Procollagen type III amino-terminal propeptide: a serum biomarker of left ventricular remodelling in paediatric dilated cardiomyopathy. <i>Cardiology in the Young</i> , <b>2015</b> , 25, 228-36	1	6
46	Gene-based analyses of the maternal genome implicate maternal effect genes as risk factors for conotruncal heart defects. <i>PLoS ONE</i> , <b>2020</b> , 15, e0234357	3.7	5
45	Early postoperative remodelling following repair of tetralogy of Fallot utilising unседated cardiac magnetic resonance: a pilot study. <i>Cardiology in the Young</i> , <b>2018</b> , 28, 697-701	1	5
44	Gene-based genome-wide association studies and meta-analyses of conotruncal heart defects. <i>PLoS ONE</i> , <b>2019</b> , 14, e0219926	3.7	5
43	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> , <b>2019</b> , 14, e0216477	3.7	4
42	Determinants of Exercise Performance in Children and Adolescents with Repaired Tetralogy of Fallot Using Stress Echocardiography. <i>Pediatric Cardiology</i> , <b>2019</b> , 40, 71-78	2.1	4
41	Genetic testing in congenital heart disease: ethical considerations. <i>World Journal for Pediatric &amp; Congenital Heart Surgery</i> , <b>2013</b> , 4, 53-7	1.1	4
40	Cerebrovascular Malformations in a Pediatric Hereditary Hemorrhagic Telangiectasia Cohort. <i>Pediatric Neurology</i> , <b>2020</b> , 110, 49-54	2.9	4

39	Resource Utilization in the First 2 Years Following Operative Correction for Tetralogy of Fallot: Study Using Data From the Optum <sup>®</sup> De-identified Clinformatics Data Mart Insurance Claims Database. <i>Journal of the American Heart Association</i> , <b>2020</b> , 9, e016581	6	4
38	Identifying genetic factors that contribute to the increased risk of congenital heart defects in infants with Down syndrome. <i>Scientific Reports</i> , <b>2020</b> , 10, 18051	4.9	4
37	Adult CHD: the ongoing need for physician counselling about heredity and contraceptive options. <i>Cardiology in the Young</i> , <b>2017</b> , 27, 671-676	1	3
36	Variants in ADRB1 and CYP2C9: Association with Response to Atenolol and Losartan in Marfan Syndrome. <i>Journal of Pediatrics</i> , <b>2020</b> , 222, 213-220.e5	3.6	3
35	Genetic variants of HIF1 $\alpha$ 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> , <b>2019</b> , 21, 51	6.9	3
34	The cardiovascular manifestations of Alagille syndrome and JAG1 mutations. <i>Methods in Molecular Medicine</i> , <b>2006</b> , 126, 217-31		3
33	Late effects in survivors of high-risk neuroblastoma following stem cell transplant with and without total body irradiation.. <i>Pediatric Blood and Cancer</i> , <b>2021</b> , e29537	3	3
32	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> , <b>2019</b> , 111, 888-905	2.05	2
31	Systems Analysis Implicates WAVE2 Complex in the Pathogenesis of Developmental Left-Sided Obstructive Heart Defects. <i>JACC Basic To Translational Science</i> , <b>2020</b> , 5, 376-386	8.7	2
30	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> , <b>2017</b> , 38, 1645-1653	2.1	2
29	Preconceptual Folic Acid Use and Recurrence Risk Counseling for Congenital Heart Disease. <i>Congenital Heart Disease</i> , <b>2015</b> , 10, 219-25	3.1	2
28	Common Genetic Variants Contribute to Risk of Transposition of the Great Arteries. <i>Circulation Research</i> , <b>2021</b> ,	15.7	2
27	Mechanisms of Congenital Heart Disease Caused by NAA15 Haploinsufficiency. <i>Circulation Research</i> , <b>2021</b> , 128, 1156-1169	15.7	2
26	Robust identification of deletions in exome and genome sequence data based on clustering of Mendelian errors. <i>Human Mutation</i> , <b>2018</b> , 39, 870-881	4.7	1
25	The 22q11.2 Deletion Syndrome100-111		1
24	The prevalence of 16p12.1 microdeletion in patients with left-sided cardiac lesions. <i>Congenital Heart Disease</i> , <b>2014</b> , 9, 83-6	3.1	1
23	Invited Commentary: The Hunt for Mechanistic Origins of Liver Fibrosis in the Fontan Circulation. <i>World Journal for Pediatric &amp; Congenital Heart Surgery</i> , <b>2021</b> , 12, 173-175	1.1	1
22	Common Variation in Cytoskeletal Genes is Associated with Conotruncal Heart Defects. <i>Genes</i> , <b>2021</b> , 12,	4.2	1



21	Aortic Root Dilation in Patients with 22q11.2 Deletion Syndrome Without Intracardiac Anomalies. <i>Pediatric Cardiology</i> , <b>2021</b> , 42, 1594-1600	2.1	1
20	Identifying Risk Factors for Complicated Post-operative Course in Tetralogy of Fallot Using a Machine Learning Approach. <i>Frontiers in Cardiovascular Medicine</i> , <b>2021</b> , 8, 685855	5.4	1
19	Cardiac evaluation of patients with 22q11.2 duplication syndrome. <i>American Journal of Medical Genetics, Part A</i> , <b>2021</b> , 185, 753-758	2.5	1
18	Neither cardiac mitochondrial DNA variation or copy number contribute to congenital heart disease risk.. <i>American Journal of Human Genetics</i> , <b>2022</b> ,	11	1
17	NKX2-6 related congenital heart disease: Biallelic homeodomain-disrupting variants and truncus arteriosus. <i>American Journal of Medical Genetics, Part A</i> , <b>2020</b> , 182, 1454-1459	2.5	0
16	Genome-Wide De Novo Variants in Congenital Heart Disease Are Not Associated With Maternal Diabetes or Obesity.. <i>Circulation Genomic and Precision Medicine</i> , <b>2022</b> , CIRCGEN121003500	5.2	0
15	X-chromosome association studies of congenital heart defects. <i>American Journal of Medical Genetics, Part A</i> , <b>2020</b> , 182, 250-254	2.5	0
14	Natural history of the aortic root in Tetralogy of Fallot after repair. <i>International Journal of Cardiology Congenital Heart Disease</i> , <b>2021</b> , 3, 100092	0.7	0
13	Right Ventricular Strain Is Associated With Increased Length of Stay After Tetralogy of Fallot Repair.. <i>Journal of Cardiovascular Imaging</i> , <b>2022</b> , 30, 50-58	1.3	0
12	Marked skeletal muscle deficits are associated with 6-minute walk distance in paediatric pulmonary hypertension. <i>Cardiology in the Young</i> , <b>2021</b> , 31, 1426-1433	1	0
11	Maternal effect genes as risk factors for congenital heart defects.. <i>Human Genetics and Genomics Advances</i> , <b>2022</b> , 3, 100098	0.8	0
10	Deciphering the Genetic Etiology of Conotruncal Defects <b>2007</b> , 238-241		
9	Gene-Interaction-Sensitive enrichment analysis in congenital heart disease.. <i>BioData Mining</i> , <b>2022</b> , 15, 4	4.3	
8	Reported practice patterns in the ambulatory care setting for patients with CHD. <i>Cardiology in the Young</i> , <b>2021</b> , 1-6	1	
7	The Molecular Genetics of Conotruncal Defects. <i>Developments in Cardiovascular Medicine</i> , <b>2000</b> , 355-374		
6	The Genetics of Congenital Heart Disease <b>2006</b> , 145-157		
5	Gene-based analyses of the maternal genome implicate maternal effect genes as risk factors for conotruncal heart defects <b>2020</b> , 15, e0234357		
4	Gene-based analyses of the maternal genome implicate maternal effect genes as risk factors for conotruncal heart defects <b>2020</b> , 15, e0234357		

- 3 Gene-based analyses of the maternal genome implicate maternal effect genes as risk factors for conotruncal heart defects **2020**, 15, e0234357
- 2 Gene-based analyses of the maternal genome implicate maternal effect genes as risk factors for conotruncal heart defects **2020**, 15, e0234357
- 1 Comparison of serum biomarkers of myocardial fibrosis with cardiac magnetic resonance in patients operated for tetralogy of Fallot.. *International Journal of Cardiology*, **2022**, 358, 27-33 3-2