Bruno Marino

List of Publications by Year in descending order

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61984 58581 7,834 149 43 82 citations h-index g-index papers 152 152 152 7204 docs citations times ranked citing authors all docs

| # | Article | IF | CITATIONS |
|----|---|------|-----------|
| 1 | 22q11.2 deletion syndrome. Nature Reviews Disease Primers, 2015, 1, 15071. | 30.5 | 954 |
| 2 | Gain-of-function RAF1 mutations cause Noonan and LEOPARD syndromes with hypertrophic cardiomyopathy. Nature Genetics, 2007, 39, 1007-1012. | 21.4 | 624 |
| 3 | Grouping of Multiple-Lentigines/LEOPARD and Noonan Syndromes on the PTPN11 Gene. American Journal of Human Genetics, 2002, 71, 389-394. | 6.2 | 380 |
| 4 | Mutations in a new gene in Ellis-van Creveld syndrome and Weyers acrodental dysostosis. Nature Genetics, 2000, 24, 283-286. | 21.4 | 323 |
| 5 | 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 |
| 6 | 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 |
| 7 | 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 |
| 8 | Prevalence and Clinical Significance of Cardiovascular Abnormalities in Patients With the LEOPARD Syndrome. American Journal of Cardiology, 2007, 100, 736-741. | 1.6 | 150 |
| 9 | NF1 Gene Mutations Represent the Major Molecular Event Underlying Neurofibromatosis-Noonan Syndrome. American Journal of Human Genetics, 2005, 77, 1092-1101. | 6.2 | 139 |
| 10 | Anatomic patterns of conotruncal defects associated with deletion 22q11. Genetics in Medicine, 2001, 3, 45-48. | 2.4 | 135 |
| 11 | LEOPARD syndrome: Clinical diagnosis in the first year of life. American Journal of Medical Genetics, Part A, 2006, 140A, 740-746. | 1.2 | 129 |
| 12 | Atrioventricular Canal in Down Syndrome. American Journal of Diseases of Children, 1990, 144, 1120. | 0.5 | 128 |
| 13 | Mutations of ZFPM2/FOG2 gene in sporadic cases of tetralogy of Fallot. Human Mutation, 2003, 22, 372-377. | 2.5 | 127 |
| 14 | Familial transposition of the great arteries caused by multiple mutations in laterality genes. Heart, 2010, 96, 673-677. | 2.9 | 126 |
| 15 | Clinical Features and Follow-Up in Patients with 22q11.2 Deletion Syndrome. Journal of Pediatrics, 2014, 164, 1475-1480.e2. | 1.8 | 119 |
| 16 | 22q11 deletions in isolated and syndromic patients with tetralogy of Fallot. Human Genetics, 1995, 95, 479-82. | 3.8 | 117 |
| 17 | Complete Transposition of the Great Arteries. Circulation, 2001, 104, 2809-2814. | 1.6 | 113 |
| 18 | Bidirectional cavopulmonary shunts: Clinical applications as staged or definitive palliation. Annals of Thoracic Surgery, 1989, 47, 415-420. | 1.3 | 108 |

| # | Article | IF | Citations |
|----|--|-----|------------|
| 19 | Congenital heart defects in Kabuki syndrome. American Journal of Medical Genetics Part A, 2001, 100, 269-274. | 2.4 | 105 |
| 20 | Genetic Syndromes and Outcome After Surgical Correction of Tetralogy of Fallot. Annals of Thoracic Surgery, 2006, 81, 968-975. | 1.3 | 104 |
| 21 | Cardiac defects and results of cardiac surgery in 22q11.2 deletion syndrome. Developmental Disabilities Research Reviews, 2008, 14, 35-42. | 2.9 | 99 |
| 22 | 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 |
| 23 | 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 |
| 24 | Genetic syndromes and congenital heart defects: how is surgical management affected?. European Journal of Cardio-thoracic Surgery, 2009, 35, 606-614. | 1.4 | 88 |
| 25 | 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 |
| 26 | 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 |
| 27 | Transposition of great arteries: new insights into the pathogenesis. Frontiers in Pediatrics, 2013, 1, 11. | 1.9 | 77 |
| 28 | Familial recurrence of congenital heart disease: an overview and review of the literature. European Journal of Pediatrics, 2006, 166, 111-116. | 2.7 | 76 |
| 29 | Cardiac defects, morbidity and mortality in patients affected by RASopathies. CARNET study results. International Journal of Cardiology, 2017, 245, 92-98. | 1.7 | 7 5 |
| 30 | Associated cardiac anomalies in isolated and syndromic patients with tetralogy of fallot. American Journal of Cardiology, 1996, 77, 505-508. | 1.6 | 73 |
| 31 | Atrioventricular canal defect without Down syndrome: A heterogeneous malformation. American Journal of Medical Genetics Part A, 1999, 85, 140-146. | 2.4 | 66 |
| 32 | 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 |
| 33 | 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 |
| 34 | Copy-Number Variation of the Glucose 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 |
| 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 | Deletion 8p syndrome. , 1998, 75, 534-536. | | 59 |

| # | Article | IF | Citations |
|----|---|-----|-----------|
| 37 | Nonrandom association of atrioventricular canal and del (8p) syndrome. American Journal of Medical Genetics Part A, 1992, 42, 424-427. | 2.4 | 58 |
| 38 | Cardiovascular disease in Down syndrome. Current Opinion in Pediatrics, 2018, 30, 616-622. | 2.0 | 58 |
| 39 | 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 |
| 40 | 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 |
| 41 | Transposition of the great arteries associated with deletion of chromosome 22q11. American Journal of Cardiology, 1995, 75, 95-98. | 1.6 | 54 |
| 42 | Deletion 22q11 in patients with interrupted aortic arch. American Journal of Cardiology, 1999, 84, 360-361. | 1.6 | 53 |
| 43 | 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 |
| 44 | 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 |
| 45 | Clinical Presentation and Natural History of Hypertrophic Cardiomyopathy in RASopathies. Heart Failure Clinics, 2018, 14, 225-235. | 2.1 | 44 |
| 46 | Costello syndrome: clinical diagnosis in the first year of life. European Journal of Pediatrics, 2008, 167, 621-628. | 2.7 | 43 |
| 47 | Rare copy number variants and congenital heart defects in the 22q11.2 deletion syndrome. Human Genetics, 2016, 135, 273-285. | 3.8 | 43 |
| 48 | Noonan syndrome and aortic coarctation. , 1998, 80, 160-162. | | 42 |
| 49 | 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 |
| 50 | Genetic heterogeneity of isolated noncompaction of the left ventricular myocardium., 1999, 85, 90-91. | | 40 |
| 51 | Congenital heart disease and genetic syndromes: new insights into molecular mechanisms. Expert Review of Molecular Diagnostics, 2017, 17, 861-870. | 3.1 | 39 |
| 52 | Atrioventricular canal and 8p- syndrome. American Journal of Medical Genetics Part A, 1993, 47, 437-438. | 2.4 | 38 |
| 53 | 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 |
| 54 | 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 |

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|----|---|-----|-----------|
| 55 | Transposition of the great arteries in asplenia and polysplenia phenotypes. American Journal of Medical Genetics Part A, 2002, 110, 292-294. | 2.4 | 33 |
| 56 | PTPN11 gene mutations: linking the Gln510Glu mutation to the "LEOPARD syndrome phenotype― European Journal of Pediatrics, 2006, 165, 803-805. | 2.7 | 33 |
| 57 | Hypertrophic Cardiomyopathy in RASopathies. Heart Failure Clinics, 2022, 18, 19-29. | 2.1 | 33 |
| 58 | Maternal diabetes causing DiGeorge anomaly and renal agenesis. American Journal of Medical Genetics Part A, 1995, 55, 513-514. | 2.4 | 32 |
| 59 | Single atrium, atrioventricular canal/postaxial hexodactyly indicating Ellis-van Creveld syndrome. Human Genetics, 1995, 96, 251-253. | 3.8 | 32 |
| 60 | What Is New in Genetics of Congenital Heart Defects?. Frontiers in Pediatrics, 2016, 4, 120. | 1.9 | 29 |
| 61 | 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 |
| 62 | Genetic heterogeneity and phenotypic anomalies in children with atrioventricular canal defect and tetralogy of Fallot. Clinical Dysmorphology, 2006, 15, 65-70. | 0.3 | 27 |
| 63 | 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 |
| 64 | Familial deletions of chromosome 22q11., 1997, 73, 95-96. | | 25 |
| 65 | 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 |
| 66 | 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 |
| 67 | Tricuspid atresia and 22q11 deletion. , 1997, 72, 40-42. | | 22 |
| 68 | Atrioventricular canal defect in patients with RASopathies. European Journal of Human Genetics, 2013, 21, 200-204. | 2.8 | 22 |
| 69 | 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 |
| 70 | 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 |
| 71 | Hif1 $\hat{l}\pm$ 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 |
| 72 | Cardiopulmonary Response to Exercise and Cardiac Assessment in Patients With Turner Syndrome. American Journal of Cardiology, 2011, 107, 1076-1082. | 1.6 | 20 |

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|----|--|-----|-----------|
| 73 | ZFPM2/FOG2 andHEY2 genes analysis in nonsyndromic tricuspid atresia. American Journal of Medical Genetics, Part A, 2005, 133A, 68-70. | 1.2 | 19 |
| 74 | 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 |
| 75 | Cardiac Catheterization through the Internal Jugular Vein in Pediatric Patients. Chest, 1992, 101, 1512-1514. | 0.8 | 18 |
| 76 | Atrioventricular canal and 3C (cranio-cerebello-cardiac) syndrome. American Journal of Medical Genetics Part A, 1995, 58, 97-98. | 2.4 | 17 |
| 77 | Atypical cardiac defects in patients with RASopathies: Updated data on CARNET study. Birth Defects Research, 2020, 112, 725-731. | 1.5 | 17 |
| 78 | Common arterial trunk, DiGeorge syndrome and microdeletion 22q11. Progress in Pediatric Cardiology, 2002, 15, 9-17. | 0.4 | 16 |
| 79 | Congenital heart defects in Noonan syndrome and RIT1 mutation. Genetics in Medicine, 2016, 18, 1320. | 2.4 | 16 |
| 80 | Morphology of the atrioventricular valve in asplenia syndrome: A peculiar type of atrioventricular canal defect. Cardiovascular Pathology, 1996, 5, 145-151. | 1.6 | 15 |
| 81 | Nonsyndromic Pulmonary Valve Stenosis and thePTPN11 Gene. American Journal of Medical Genetics Part A, 2003, 116A, 389-390. | 2.4 | 15 |
| 82 | Heterozygous missense mutations in <i>NFATC1</i> are associated with atrioventricular septal defect. Human Mutation, 2018, 39, 1428-1441. | 2.5 | 15 |
| 83 | Some Isolated Cardiac Malformations Can Be Related to Laterality Defects. Journal of Cardiovascular Development and Disease, 2018, 5, 24. | 1.6 | 15 |
| 84 | Clinical Manifestations of 22q11.2 Deletion Syndrome. Heart Failure Clinics, 2021, 18, 155-164. | 2.1 | 15 |
| 85 | Atrioventricular septal defectâ€"anatomic characteristics in patients with and without Down's syndrome. Cardiology in the Young, 1992, 2, 308-310. | 0.8 | 14 |
| 86 | Search for 22q11 deletion in non-syndromic conotruncal cardiac defects. European Journal of Pediatrics, 1996, 155, 619-620. | 2.7 | 14 |
| 87 | 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 |
| 88 | The mendelian basis of congenital heart defects. Cardiology in the Young, 1996, 6, 264-271. | 0.8 | 13 |
| 89 | The atrioventricular canal defect is the congenital heart disease connecting short rib-polydactyly and oral-facial-digital syndromes., 1997, 68, 110-112. | | 13 |
| 90 | Congenital heart defect in sibs with discordant karyotypes. , 1998, 80, 169-172. | | 13 |

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| 91 | Familial recurrence of nonsyndromic interrupted aortic arch and truncus arteriosus with atrioventricular canal., 2000, 61, 329-331. | | 13 |
| 92 | 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 |
| 93 | 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 |
| 94 | 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 |
| 95 | Conotruncal heart defects and chromosome 22q11microdeletion. Journal of Pediatrics, 1997, 130, 675-676. | 1.8 | 12 |
| 96 | 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 |
| 97 | Cardiac Defects and Genetic Syndromes: Old Uncertainties and New Insights. Genes, 2021, 12, 1047. | 2.4 | 12 |
| 98 | Di George Anomaly with Atrioventricular Canal. Chest, 1991, 99, 242-243. | 0.8 | 11 |
| 99 | Diffuse coronary dilation in a young patient with LEOPARD syndrome. International Journal of Cardiology, 2006, 112, E35-E37. | 1.7 | 11 |
| 100 | Familial recurrence of discrete membranous subaortic stenosis. Journal of Thoracic and Cardiovascular Surgery, 2007, 134, 818-819. | 0.8 | 11 |
| 101 | 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 |
| 102 | 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 |
| 103 | Atrioventricular septal defects with severe left ventricular hypoplasia—clinical findings and surgical options. Cardiology in the Young, 1992, 2, 53-55. | 0.8 | 9 |
| 104 | Truncus arteriosus and duplication 8q. American Journal of Medical Genetics Part A, 2003, 121A, 79-81. | 2.4 | 9 |
| 105 | <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 |
| 106 | Genetics of atrioventricular canal defects. Italian Journal of Pediatrics, 2020, 46, 61. | 2.6 | 9 |
| 107 | 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 |
| 108 | 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 |

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|-----|--|-----|-----------|
| 109 | Multiplex Ligation-Dependent Probe Amplification Analysis of (i>GATA4Gene Copy Number Variations in Patients with Isolated Congenital Heart Disease. Disease Markers, 2010, 28, 287-292. | 1.3 | 8 |
| 110 | Progeroid syndrome with characteristic facial appearance and hand anomalies in father and son., 1997, 73, 227-229. | | 7 |
| 111 | Atrioventricular canal defect and hypoplastic left heart syndrome as discordant congenital heart defects in twins. Teratology, 1999, 60, 206-208. | 1.6 | 7 |
| 112 | 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 |
| 113 | 22q11.2 Deletion Syndrome: Impact of Genetics in the Treatment of Conotruncal Heart Defects. Children, 2022, 9, 772. | 1.5 | 7 |
| 114 | Intrafamilial variability of Pfeiffer-type cardiocranial syndrome., 1997, 73, 480-483. | | 6 |
| 115 | Genetic predisposition to ventricular septal defect in Down syndrome. Human Genetics, 2001, 109, 463-463. | 3.8 | 6 |
| 116 | Tetralogy of fallot with aortic valvular stenosis and deletion 22q11. Annals of Thoracic Surgery, 2003, 75, 2010-2011. | 1.3 | 6 |
| 117 | Congenital cardiovascular disease and velo-cardio-facial syndrome. , 2005, , 47-82. | | 6 |
| 118 | Atrioventricular Canal Defect and Associated Genetic Disorders: New Insights into Polydactyly Syndromes. Neurology International, $2011, 1, e7$. | 0.5 | 6 |
| 119 | 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 |
| 120 | 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 |
| 121 | The Secrets of the Frogs Heart. Pediatric Cardiology, 2022, 43, 1471-1480. | 1.3 | 6 |
| 122 | Familial recurrence of transposition of the great arteries and intact ventricular septum., 1997, 73, 93-94. | | 5 |
| 123 | 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 |
| 124 | The heart and shell. Anatomical and genetic similarities. American Heart Journal, 2011, 161, 647-649. | 2.7 | 5 |
| 125 | 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 |
| 126 | Pediatric patients with RASopathy-associated hypertrophic cardiomyopathy: the multifaceted consequences of PTPN11 mutations. Orphanet Journal of Rare Diseases, 2019, 14, 163. | 2.7 | 5 |

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|-----|--|-----|-----------|
| 127 | 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 |
| 128 | Severe, early onset hypertrophic cardiomyopathy in a family with LEOPARD syndrome. Journal of Prenatal Medicine, 2008, 2, 24-6. | 0.2 | 5 |
| 129 | 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 |
| 130 | Inlet ventricular septal defect is not a partial atrioventricular septal defect., 1999, 87, 195-195. | | 4 |
| 131 | Additional evidence that PTPN11 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 |
| 132 | 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 |
| 133 | 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 |
| 134 | Atrioventricular canal defect as partial expression of heterotaxia in patients with Bardet-Biedl syndrome. Journal of Pediatrics, 2020, 218, 263-264. | 1.8 | 4 |
| 135 | 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 |
| 136 | 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 |
| 137 | Atrioventricular canal defect is the classic congenital heart disease in Bardet–Biedl syndrome. Annals of Human Genetics, 2021, 85, 101-102. | 0.8 | 3 |
| 138 | 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 |
| 139 | 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 |
| 140 | Screening for 22q11.2 microdeletion in adults with tetralogy of Fallot. Heart, 2011, 97, 860-860. | 2.9 | 2 |
| 141 | Deletion 8p syndrome. American Journal of Medical Genetics Part A, 1998, 75, 534-536. | 2.4 | 2 |
| 142 | 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 |
| 143 | Morphology of the Atrioventricular Valve in Patients with Right Atrial Isomerism. Pediatric Cardiology, 1998, 19, 501-501. | 1.3 | 0 |
| 144 | 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 |

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|-----|--|-----|-----------|
| 145 | 22q11.2 Deletion (DiGeorge) Syndrome: A Mother's Open Letter. Neurology International, 2011, 1, e11. | 0.5 | 0 |
| 146 | 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 |
| 147 | Anatomical substrate for biventricular repair in patients with left isomerism. Annals of Pediatric Cardiology, 2021, 14, 250. | 0.5 | 0 |
| 148 | Commentary: sVEGFR1 Is Enriched in Hepatic Vein Bloodâ€"Evidence for a Provisional Hepatic Factor Candidate?. Frontiers in Pediatrics, 2021, 9, 782779. | 1.9 | 0 |
| 149 | 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 |