

Bruce D Gelb

List of Publications by Year in descending order

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Version: 2024-02-01

165
papers

20,085
citations

16451

64
h-index

11607

135
g-index

192
all docs

192
docs citations

192
times ranked

20373
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutations in PTPN11, encoding the protein tyrosine phosphatase SHP-2, cause Noonan syndrome. <i>Nature Genetics</i> , 2001, 29, 465-468.	21.4	1,555
2	Somatic mutations in PTPN11 in juvenile myelomonocytic leukemia, myelodysplastic syndromes and acute myeloid leukemia. <i>Nature Genetics</i> , 2003, 34, 148-150.	21.4	960
3	De novo mutations in histone-modifying genes in congenital heart disease. <i>Nature</i> , 2013, 498, 220-223.	27.8	798
4	Genetic Basis for Congenital Heart Defects: Current Knowledge. <i>Circulation</i> , 2007, 115, 3015-3038.	1.6	719
5	PTPN11 Mutations in Noonan Syndrome: Molecular Spectrum, Genotype-Phenotype Correlation, and Phenotypic Heterogeneity. <i>American Journal of Human Genetics</i> , 2002, 70, 1555-1563.	6.2	680
6	Patient-specific induced pluripotent stem-cell-derived models of LEOPARD syndrome. <i>Nature</i> , 2010, 465, 808-812.	27.8	672
7	De novo mutations in congenital heart disease with neurodevelopmental and other congenital anomalies. <i>Science</i> , 2015, 350, 1262-1266.	12.6	646
8	Gain-of-function RAF1 mutations cause Noonan and LEOPARD syndromes with hypertrophic cardiomyopathy. <i>Nature Genetics</i> , 2007, 39, 1007-1012.	21.4	624
9	Contribution of rare inherited and de novo variants in 2,871 congenital heart disease probands. <i>Nature Genetics</i> , 2017, 49, 1593-1601.	21.4	624
10	Noonan syndrome. <i>Lancet</i> , The, 2013, 381, 333-342.	18.7	608
11	Gain-of-function SOS1 mutations cause a distinctive form of Noonan syndrome. <i>Nature Genetics</i> , 2007, 39, 75-79.	21.4	523
12	Genetics of Congenital Heart Disease. <i>Circulation Research</i> , 2013, 112, 707-720.	4.5	491
13	Mapping Systemic Inflammation and Antibody Responses in Multisystem Inflammatory Syndrome in Children (MIS-C). <i>Cell</i> , 2020, 183, 982-995.e14.	28.9	440
14	Genetic Basis for Congenital Heart Disease: Revisited: A Scientific Statement From the American Heart Association. <i>Circulation</i> , 2018, 138, e653-e711.	1.6	387
15	Mutation of SHOC2 promotes aberrant protein N-myristoylation and causes Noonan-like syndrome with loose anagen hair. <i>Nature Genetics</i> , 2009, 41, 1022-1026.	21.4	358
16	Diversity and Functional Consequences of Germline and Somatic PTPN11 Mutations in Human Disease. <i>American Journal of Human Genetics</i> , 2006, 78, 279-290.	6.2	352
17	NOONAN SYNDROME AND RELATED DISORDERS: Genetics and Pathogenesis. <i>Annual Review of Genomics and Human Genetics</i> , 2005, 6, 45-68.	6.2	306
18	Noonan syndrome and clinically related disorders. <i>Best Practice and Research in Clinical Endocrinology and Metabolism</i> , 2011, 25, 161-179.	4.7	303

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19	A restricted spectrum of NRAS mutations causes Noonan syndrome. <i>Nature Genetics</i> , 2010, 42, 27-29.	21.4	271
20	Genetic evidence for lineage-related and differentiation stage-related contribution of somatic PTPN11 mutations to leukemogenesis in childhood acute leukemia. <i>Blood</i> , 2004, 104, 307-313.	1.4	265
21	Mutations in TFAP2B cause Char syndrome, a familial form of patent ductus arteriosus. <i>Nature Genetics</i> , 2000, 25, 42-46.	21.4	252
22	Germline <i>BRAF</i> mutations in Noonan, LEOPARD, and cardiofaciocutaneous syndromes: Molecular diversity and associated phenotypic spectrum. <i>Human Mutation</i> , 2009, 30, 695-702.	2.5	251
23	The mutational spectrum of PTPN11 in juvenile myelomonocytic leukemia and Noonan syndrome/myeloproliferative disease. <i>Blood</i> , 2005, 106, 2183-2185.	1.4	247
24	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.	4.5	229
25	Heterozygous Germline Mutations in the CBL Tumor-Suppressor Gene Cause a Noonan Syndrome-like Phenotype. <i>American Journal of Human Genetics</i> , 2010, 87, 250-257.	6.2	221
26	Germline Missense Mutations Affecting KRAS Isoform B Are Associated with a Severe Noonan Syndrome Phenotype. <i>American Journal of Human Genetics</i> , 2006, 79, 129-135.	6.2	205
27	Mutations in a new gene encoding a thiamine transporter cause thiamine-responsive megaloblastic anaemia syndrome. <i>Nature Genetics</i> , 1999, 22, 309-312.	21.4	201
28	WHIM syndrome, an autosomal dominant disorder: Clinical, hematological, and molecular studies. <i>American Journal of Medical Genetics Part A</i> , 2000, 91, 368-376.	2.4	193
29	Distinct epigenetic programs regulate cardiac myocyte development and disease in the human heart in vivo. <i>Nature Communications</i> , 2018, 9, 391.	12.8	181
30	Noonan syndrome-associated SHP2/PTPN11 mutants cause EGF-dependent prolonged GAB1 binding and sustained ERK2/MAPK1 activation. <i>Human Mutation</i> , 2004, 23, 267-277.	2.5	177
31	Noonan syndrome and related disorders: dysregulated RAS-mitogen activated protein kinase signal transduction. <i>Human Molecular Genetics</i> , 2006, 15, R220-R226.	2.9	177
32	Enabling Technologies for Personalized and Precision Medicine. <i>Trends in Biotechnology</i> , 2020, 38, 497-518.	9.3	169
33	Disorders of dysregulated signal traffic through the RAS-MAPK pathway: phenotypic spectrum and molecular mechanisms. <i>Annals of the New York Academy of Sciences</i> , 2010, 1214, 99-121.	3.8	167
34	The Congenital Heart Disease Genetic Network Study. <i>Circulation Research</i> , 2013, 112, 698-706.	4.5	142
35	The Phosphatase SHP2 Regulates the Spacing Effect for Long-Term Memory Induction. <i>Cell</i> , 2009, 139, 186-198.	28.9	139
36	Functional Dysregulation of CDC42 Causes Diverse Developmental Phenotypes. <i>American Journal of Human Genetics</i> , 2018, 102, 309-320.	6.2	138

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37	Determination of Bone Markers in Pycnodysostosis: Effects of Cathepsin K Deficiency on Bone Matrix Degradation. <i>Journal of Bone and Mineral Research</i> , 1999, 14, 1902-1908.	2.8	128
38	Germ-line and somatic PTPN11 mutations in human disease. <i>European Journal of Medical Genetics</i> , 2005, 48, 81-96.	1.3	128
39	ClinGen™s RASopathy Expert Panel consensus methods for variant interpretation. <i>Genetics in Medicine</i> , 2018, 20, 1334-1345.	2.4	126
40	Leigh syndrome and hypertrophic cardiomyopathy in an infant with a mitochondrial DNA point mutation (T8993G). <i>American Journal of Medical Genetics Part A</i> , 1994, 50, 265-271.	2.4	124
41	The Clinical Sequencing Evidence-Generating Research Consortium: Integrating Genomic Sequencing in Diverse and Medically Underserved Populations. <i>American Journal of Human Genetics</i> , 2018, 103, 319-327.	6.2	122
42	Complex Genetics and the Etiology of Human Congenital Heart Disease. <i>Cold Spring Harbor Perspectives in Medicine</i> , 2014, 4, a013953-a013953.	6.2	118
43	Activating mutations in RRAS underlie a phenotype within the RASopathy spectrum and contribute to leukaemogenesis. <i>Human Molecular Genetics</i> , 2014, 23, 4315-4327.	2.9	114
44	Cardiomyopathies in Noonan syndrome and the other RASopathies. <i>Progress in Pediatric Cardiology</i> , 2015, 39, 13-19.	0.4	99
45	SOS1 mutations in Noonan syndrome: molecular spectrum, structural insights on pathogenic effects, and genotype-phenotype correlations. <i>Human Mutation</i> , 2011, 32, 760-772.	2.5	97
46	Genomic analyses implicate noncoding de novo variants in congenital heart disease. <i>Nature Genetics</i> , 2020, 52, 769-777.	21.4	97
47	Hypertrophic Cardiomyopathy in Noonan Syndrome Treated by MEK-Inhibition. <i>Journal of the American College of Cardiology</i> , 2019, 73, 2237-2239.	2.8	96
48	Linkage of pycnodysostosis to chromosome 1q21 by homozygosity mapping. <i>Nature Genetics</i> , 1995, 10, 235-237.	21.4	94
49	Autosomal Recessive Cardiomyopathy Presenting as Acute Myocarditis. <i>Journal of the American College of Cardiology</i> , 2017, 69, 1653-1665.	2.8	94
50	De Novo and Rare Variants at Multiple Loci Support the Oligogenic Origins of Atrioventricular Septal Heart Defects. <i>PLoS Genetics</i> , 2016, 12, e1005963.	3.5	92
51	MATR3 disruption in human and mouse associated with bicuspid aortic valve, aortic coarctation and patent ductus arteriosus. <i>Human Molecular Genetics</i> , 2015, 24, 2375-2389.	2.9	90
52	Cathepsin K Deficiency Reduces Elastase Perfusion-Induced Abdominal Aortic Aneurysms in Mice. <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , 2012, 32, 15-23.	2.4	89
53	Effect of Copy Number Variants on Outcomes for Infants With Single Ventricle Heart Defects. <i>Circulation: Cardiovascular Genetics</i> , 2013, 6, 444-451.	5.1	89
54	Phosphatase-defective LEOPARD syndrome mutations in PTPN11 gene have gain-of-function effects during Drosophila development. <i>Human Molecular Genetics</i> , 2009, 18, 193-201.	2.9	82

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55	Identification of rare de novo epigenetic variations in congenital disorders. <i>Nature Communications</i> , 2018, 9, 2064.	12.8	82
56	The Congenital Heart Disease Genetic Network Study: Cohort description. <i>PLoS ONE</i> , 2018, 13, e0191319.	2.5	82
57	Diverse driving forces underlie the invariant occurrence of the T42A, E139D, I282V and T468M SHP2 amino acid substitutions causing Noonan and LEOPARD syndromes. <i>Human Molecular Genetics</i> , 2008, 17, 2018-2029.	2.9	79
58	Myeloid Dysregulation in a Human Induced Pluripotent Stem Cell Model of PTPN11 -Associated Juvenile Myelomonocytic Leukemia. <i>Cell Reports</i> , 2015, 13, 504-515.	6.4	79
59	Deficiency and Inhibition of Cathepsin K Reduce Body Weight Gain and Increase Glucose Metabolism in Mice. <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , 2008, 28, 2202-2208.	2.4	78
60	Paternal Germline Origin and Sex-Ratio Distortion in Transmission of PTPN11 Mutations in Noonan Syndrome. <i>American Journal of Human Genetics</i> , 2004, 75, 492-497.	6.2	76
61	Protein Tyrosine Phosphatase PTPN14 Is a Regulator of Lymphatic Function and Choanal Development in Humans. <i>American Journal of Human Genetics</i> , 2010, 87, 436-444.	6.2	75
62	Neurofibromatosis-Noonan syndrome: Molecular evidence of the concurrence of both disorders in a patient. <i>American Journal of Medical Genetics, Part A</i> , 2005, 136A, 242-245.	1.2	74
63	Human Engineered Cardiac Tissues Created Using Induced Pluripotent Stem Cells Reveal Functional Characteristics of BRAF-Mediated Hypertrophic Cardiomyopathy. <i>PLoS ONE</i> , 2016, 11, e0146697.	2.5	72
64	RAF1 mutations in childhood-onset dilated cardiomyopathy. <i>Nature Genetics</i> , 2014, 46, 635-639.	21.4	69
65	Genetic basis of congenital heart disease. <i>Current Opinion in Cardiology</i> , 2004, 19, 110-115.	1.8	68
66	Congenital heart defects in Noonan syndrome: Diagnosis, management, and treatment. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2020, 184, 73-80.	1.6	68
67	Activating Mutations Affecting the Dbl Homology Domain of SOS2 Cause Noonan Syndrome. <i>Human Mutation</i> , 2015, 36, 1080-1087.	2.5	67
68	Transgenic <i>Drosophila</i> models of Noonan syndrome causing PTPN11 gain-of-function mutations. <i>Human Molecular Genetics</i> , 2006, 15, 543-553.	2.9	66
69	Assessing the gene-disease association of 19 genes with the RASopathies using the ClinGen gene curation framework. <i>Human Mutation</i> , 2018, 39, 1485-1493.	2.5	66
70	Complex Autoinflammatory Syndrome Unveils Fundamental Principles of JAK1 Kinase Transcriptional and Biochemical Function. <i>Immunity</i> , 2020, 53, 672-684.e11.	14.3	66
71	Concise Review: Drug Discovery in the Age of the Induced Pluripotent Stem Cell. <i>Stem Cells Translational Medicine</i> , 2014, 3, 500-509.	3.3	65
72	Characteristics of children and young adults with Marfan syndrome and aortic root dilation in a randomized trial comparing atenolol and losartan therapy. <i>American Heart Journal</i> , 2013, 165, 828-835.e3.	2.7	59

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73	De novo and recessive forms of congenital heart disease have distinct genetic and phenotypic landscapes. <i>Nature Communications</i> , 2019, 10, 4722.	12.8	58
74	The genetics of congenital heart disease: a review of recent developments. <i>Current Opinion in Cardiology</i> , 2007, 22, 200-206.	1.8	57
75	Rapidly progressive hypertrophic cardiomyopathy in an infant with Noonan syndrome with multiple lentigines: Palliative treatment with a rapamycin analog. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 744-751.	1.2	53
76	The CHD4-related syndrome: a comprehensive investigation of the clinical spectrum, genotype-phenotype correlations, and molecular basis. <i>Genetics in Medicine</i> , 2020, 22, 389-397.	2.4	53
77	Loss of RNA expression and allele-specific expression associated with congenital heart disease. <i>Nature Communications</i> , 2016, 7, 12824.	12.8	51
78	RAS signaling pathway mutations and hypertrophic cardiomyopathy: getting into and out of the thick of it. <i>Journal of Clinical Investigation</i> , 2011, 121, 844-847.	8.2	49
79	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. <i>American Journal of Human Genetics</i> , 2020, 107, 499-513.	6.2	48
80	Absence of PTPN11 mutations in 28 cases of cardiofaciocutaneous (CFC) syndrome. <i>Human Genetics</i> , 2002, 111, 421-427.	3.8	45
81	Clinical Presentation and Natural History of Hypertrophic Cardiomyopathy in RASopathies. <i>Heart Failure Clinics</i> , 2018, 14, 225-235.	2.1	44
82	Robust identification of mosaic variants in congenital heart disease. <i>Human Genetics</i> , 2018, 137, 183-193.	3.8	43
83	De novo variants in exomes of congenital heart disease patients identify risk genes and pathways. <i>Genome Medicine</i> , 2020, 12, 9.	8.2	43
84	Genetic Testing for Heritable Cardiovascular Diseases in Pediatric Patients: A Scientific Statement From the American Heart Association. <i>Circulation Genomic and Precision Medicine</i> , 2021, 14, e000086.	3.6	43
85	Downregulation of exhausted cytotoxic T cells in gene expression networks of multisystem inflammatory syndrome in children. <i>Nature Communications</i> , 2021, 12, 4854.	12.8	42
86	Advancing RAS/RASopathy therapies: An NCI-sponsored intramural and extramural collaboration for the study of RASopathies. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 866-876.	1.2	40
87	Identification and Purification of Human Induced Pluripotent Stem Cell-Derived Atrial-Like Cardiomyocytes Based on Sarcolipin Expression. <i>PLoS ONE</i> , 2014, 9, e101316.	2.5	40
88	Genetic mapping of the cleidocranial dysplasia (CCD) locus on chromosome band 6p21 to include a microdeletion. <i>American Journal of Medical Genetics Part A</i> , 1995, 58, 200-205.	2.4	39
89	Structural, Functional, and Clinical Characterization of a Novel PTPN11 Mutation Cluster Underlying Noonan Syndrome. <i>Human Mutation</i> , 2017, 38, 451-459.	2.5	39
90	Novel Variant Findings and Challenges Associated With the Clinical Integration of Genomic Testing. <i>JAMA Pediatrics</i> , 2021, 175, e205906.	6.2	39

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91	Transcription factor protein interactomes reveal genetic determinants in heart disease. <i>Cell</i> , 2022, 185, 794-814.e30.	28.9	39
92	Malignant fibrous histiocytoma: Inherited and sporadic forms have loss of heterozygosity at chromosome bands 9p21-22?evidence for a common genetic defect. <i>Genes Chromosomes and Cancer</i> , 2000, 27, 191-195.	2.8	36
93	Molecular Diversity and Associated Phenotypic Spectrum of Germline <i>CBL</i> Mutations. <i>Human Mutation</i> , 2015, 36, 787-796.	2.5	36
94	Myopathic Cardiac Genotypes Increase Risk for Myocarditis. <i>JACC Basic To Translational Science</i> , 2021, 6, 584-592.	4.1	36
95	Counteracting Effects Operating on Src Homology 2 Domain-containing Protein-tyrosine Phosphatase 2 (SHP2) Function Drive Selection of the Recurrent Y62D and Y63C Substitutions in Noonan Syndrome*. <i>Journal of Biological Chemistry</i> , 2012, 287, 27066-27077.	3.4	35
96	The Genomic Medicine Integrative Research Framework: A Conceptual Framework for Conducting Genomic Medicine Research. <i>American Journal of Human Genetics</i> , 2019, 104, 1088-1096.	6.2	35
97	Association of Damaging Variants in Genes With Increased Cancer Risk Among Patients With Congenital Heart Disease. <i>JAMA Cardiology</i> , 2021, 6, 457.	6.1	34
98	Autonomous and Non-autonomous Defects Underlie Hypertrophic Cardiomyopathy in BRAF-Mutant hiPSC-Derived Cardiomyocytes. <i>Stem Cell Reports</i> , 2016, 7, 355-369.	4.8	33
99	Hypertrophic Cardiomyopathy in RASopathies. <i>Heart Failure Clinics</i> , 2022, 18, 19-29.	2.1	33
100	Induced pluripotent stem cell-derived cardiomyocytes as models for genetic cardiovascular disorders. <i>Current Opinion in Cardiology</i> , 2011, 26, 223-229.	1.8	32
101	De Novo Missense Variants in FBXW11 Cause Diverse Developmental Phenotypes Including Brain, Eye, and Digit Anomalies. <i>American Journal of Human Genetics</i> , 2019, 105, 640-657.	6.2	31
102	GATA6 mutations in hiPSCs inform mechanisms for maldevelopment of the heart, pancreas, and diaphragm. <i>ELife</i> , 2020, 9, .	6.0	31
103	Histone H2B monoubiquitination regulates heart development via epigenetic control of cilia motility. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2019, 116, 14049-14054.	7.1	30
104	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.	2.8	30
105	De Novo Damaging Variants, Clinical Phenotypes, and Post-Operative Outcomes in Congenital Heart Disease. <i>Circulation Genomic and Precision Medicine</i> , 2020, 13, e002836.	3.6	30
106	MEK inhibitors for neurofibromatosis type 1 manifestations: Clinical evidence and consensus. <i>Neuro-Oncology</i> , 2022, 24, 1845-1856.	1.2	30
107	Sequential Defects in Cardiac Lineage Commitment and Maturation Cause Hypoplastic Left Heart Syndrome. <i>Circulation</i> , 2021, 144, 1409-1428.	1.6	29
108	Genetic determinants of telomere length from 109,122 ancestrally diverse whole-genome sequences in TOPMed. <i>Cell Genomics</i> , 2022, 2, 100084.	6.5	29

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109	Novel functional interaction between Na ⁺ /H ⁺ exchanger 1 and tyrosine phosphatase SHP-2. American Journal of Physiology - Regulatory Integrative and Comparative Physiology, 2007, 292, R2406-R2416.	1.8	28
110	Practical Guidance on Informed Consent for Pediatric Participants in a Biorepository. Mayo Clinic Proceedings, 2014, 89, 1471-1480.	3.0	27
111	Sampling the host response to SARS-CoV-2 in hospitals under siege. Nature Medicine, 2020, 26, 1157-1158.	30.7	27
112	Mechanisms of Congenital Heart Disease Caused by NAA15 Haploinsufficiency. Circulation Research, 2021, 128, 1156-1169.	4.5	27
113	Rare genetic variation at transcription factor binding sites modulates local DNA methylation profiles. PLoS Genetics, 2020, 16, e1009189.	3.5	27
114	SPRED2 loss-of-function causes a recessive Noonan syndrome-like phenotype. American Journal of Human Genetics, 2021, 108, 2112-2129.	6.2	23
115	Frequency of Aortic Dilatation in Noonan Syndrome. American Journal of Cardiology, 2014, 113, 368-371.	1.6	22
116	Cyclosporine attenuates cardiomyocyte hypertrophy induced by RAF1 mutants in Noonan and LEOPARD syndromes. Journal of Molecular and Cellular Cardiology, 2011, 51, 4-15.	1.9	21
117	Cathepsin K Deficiency Ameliorates Systemic Lupus Erythematosus-like Manifestations in FasLpr Mice. Journal of Immunology, 2017, 198, 1846-1854.	0.8	21
118	Biallelic MADD variants cause a phenotypic spectrum ranging from developmental delay to a multisystem disorder. Brain, 2020, 143, 2437-2453.	7.6	21
119	The NYCKidSeq project: study protocol for a randomized controlled trial incorporating genomics into the clinical care of diverse New York City children. Trials, 2021, 22, 56.	1.6	21
120	History of Our Understanding of the Causes of Congenital Heart Disease. Circulation: Cardiovascular Genetics, 2015, 8, 529-536.	5.1	20
121	GUÃA: a digital platform to facilitate result disclosure in genetic counseling. Genetics in Medicine, 2021, 23, 942-949.	2.4	20
122	Headaches in hypermobility syndromes: A pain in the neck?. American Journal of Medical Genetics, Part A, 2020, 182, 2902-2908.	1.2	19
123	When Participants in Genomic Research Grow Up: Contact and Consent at the Age of Majority. Journal of Pediatrics, 2016, 168, 226-231.e1.	1.8	17
124	SHOC2 subcellular shuttling requires the KEKE motif-rich region and N-terminal leucine-rich repeat domain and impacts on ERK signalling. Human Molecular Genetics, 2016, 25, 3824-3835.	2.9	17
125	EM-mosaic detects mosaic point mutations that contribute to congenital heart disease. Genome Medicine, 2020, 12, 42.	8.2	17
126	Fgfr3 Is a Transcriptional Target of Ap2 and Ash2l-Containing Histone Methyltransferase Complexes. PLoS ONE, 2009, 4, e8535.	2.5	16

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127	Recent advances in understanding the genetics of congenital heart defects. <i>Current Opinion in Pediatrics</i> , 2013, 25, 561-566.	2.0	16
128	A genotype-first approach to exploring Mendelian cardiovascular traits with clear external manifestations. <i>Genetics in Medicine</i> , 2021, 23, 94-102.	2.4	16
129	DPH1 syndrome: two novel variants and structural and functional analyses of seven missense variants identified in syndromic patients. <i>European Journal of Human Genetics</i> , 2020, 28, 64-75.	2.8	15
130	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.	4.1	15
131	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.	2.4	13
132	A <i>PTPN11</i> allele encoding a catalytically impaired SHP2 protein in a patient with a Noonan syndrome phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2351-2355.	1.2	12
133	The Spacing Effect for Structural Synaptic Plasticity Provides Specificity and Precision in Plastic Changes. <i>Journal of Neuroscience</i> , 2017, 37, 4992-5007.	3.6	12
134	Drosophila RASopathy models identify disease subtype differences and biomarkers of drug efficacy. <i>IScience</i> , 2021, 24, 102306.	4.1	12
135	Biallelic loss-of-function variants in <i>KCNJ16</i> presenting with hypokalemic metabolic acidosis. <i>European Journal of Human Genetics</i> , 2021, 29, 1566-1569.	2.8	12
136	Hope versus reality: Parent expectations of genomic testing. <i>Patient Education and Counseling</i> , 2021, 104, 2073-2079.	2.2	10
137	The seventh international RASopathies symposium: Pathways to a cure—expanding knowledge, enhancing research, and therapeutic discovery. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 1915-1927.	1.2	10
138	Dysautonomia in hypermobile Ehlers-Danlos syndrome and hypermobility spectrum disorders is associated with exercise intolerance and cardiac atrophy. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3754-3761.	1.2	9
139	Construction of Defined Human Engineered Cardiac Tissues to Study Mechanisms of Cardiac Cell Therapy. <i>Journal of Visualized Experiments</i> , 2016, , e53447.	0.3	9
140	Proteomic Analysis of an Induced Pluripotent Stem Cell Model Reveals Strategies to Treat Juvenile Myelomonocytic Leukemia. <i>Journal of Proteome Research</i> , 2020, 19, 194-203.	3.7	8
141	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.	1.8	8
142	The Phosphatase CSW Controls Life Span by Insulin Signaling and Metabolism Throughout Adult Life in <i>Drosophila</i> . <i>Frontiers in Genetics</i> , 2020, 11, 364.	2.3	8
143	“œls that something that should concern me?” a qualitative exploration of parent understanding of their child’s genomic test results. <i>Human Genetics and Genomics Advances</i> , 2021, 2, 100027.	1.7	8
144	Cardiovascular manifestations of hypermobile Ehlers-Danlos syndrome and hypermobility spectrum disorders. <i>Vascular Medicine</i> , 2022, 27, 283-289.	1.5	8

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145	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.	3.6	8
146	Burden of Cardiomyopathic Genetic Variation in Lethal Pediatric Myocarditis. <i>Circulation Genomic and Precision Medicine</i> , 2021, 14, e003426.	3.6	7
147	US private payers'™ perspectives on insurance coverage for genome sequencing versus exome sequencing: A study by the Clinical Sequencing Evidence-Generating Research Consortium (CSER). <i>Genetics in Medicine</i> , 2022, 24, 238-244.	2.4	6
148	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.	6.2	5
149	The Good SHP2 Association. <i>Circulation: Cardiovascular Genetics</i> , 2012, 5, 271-273.	5.1	4
150	NPSV: A simulation-driven approach to genotyping structural variants in whole-genome sequencing data. <i>GigaScience</i> , 2021, 10, .	6.4	4
151	Robust identification of deletions in exome and genome sequence data based on clustering of Mendelian errors. <i>Human Mutation</i> , 2018, 39, 870-881.	2.5	3
152	Elucidation of de novo small insertion/deletion biology with parent'€of'€origin phasing. <i>Human Mutation</i> , 2020, 41, 800-806.	2.5	3
153	GenomeDiver: a platform for phenotype-guided medical genomic diagnosis. <i>Genetics in Medicine</i> , 2021, 23, 1998-2002.	2.4	3
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