Bruce D Gelb

List of Publications by Year
in descending order

165 papers	20,085 citations	18887 64 h-index	13274 135 g-index
192 all docs	192 docs citations	192 times ranked	22074 citing authors

#	Article	IF	Citations
1	Hypertrophic Cardiomyopathy in RASopathies. Heart Failure Clinics, 2022, 18, 19-29.	1.0	33
2	US private payersâ€™ perspectives on insurance coverage for genome sequencing versus exome sequencing: A study by the Clinical Sequencing Evidence-Generating Research Consortium (CSER). Genetics in Medicine, 2022, 24, 238-244.	1.1	6
3	Cardiovascular manifestations of hypermobile Ehlersâ€“Danlos syndrome and hypermobility spectrum disorders. Vascular Medicine, 2022, 27, 283-289.	0.8	8
4	Molecular characterization and investigation of the role of genetic variation in phenotypic variability and response to treatment in a large pediatric Marfan syndrome cohort. Genetics in Medicine, 2022, 24, 1045-1053.	1.1	13
5	Genetic determinants of telomere length from 109,122 ancestrally diverse whole-genome sequences in TOPMed. Cell Genomics, 2022, 2, 100084.	3.0	29
6	Genome-Wide De Novo Variants in Congenital Heart Disease Are Not Associated With Maternal Diabetes or Obesity. Circulation Genomic and Precision Medicine, 2022, 15, CIRCGEN121003500.	1.6	8
7	The seventh international <scp>RASopathies</scp> symposium: Pathways to a cureâ€”expanding knowledge, enhancing research, and therapeutic discovery. American Journal of Medical Genetics, Part A, 2022, 188, 1915-1927.	0.7	10
8	Transcription factor protein interactomes reveal genetic determinants in heart disease. Cell, 2022, 185, 794-814.e30.	13.5	39
9	Neither cardiac mitochondrial DNA variation nor copy number contribute to congenital heart disease risk. American Journal of Human Genetics, 2022, 109, 961-966.	2.6	5
10	MEK inhibitors for neurofibromatosis type 1 manifestations: Clinical evidence and consensus. Neuro-Oncology, 2022, 24, 1845-1856.	0.6	30
11	A genotype-first approach to exploring Mendelian cardiovascular traits with clear external manifestations. Genetics in Medicine, 2021, 23, 94-102.	1.1	16
12	Association of Damaging Variants in Genes With Increased Cancer Risk Among Patients With Congenital Heart Disease. JAMA Cardiology, 2021, 6, 457.	3.0	34
13	A de novo pathogenic <scp>BMP2</scp> variantâ€related phenotype with the novel finding of bicuspid aortic valve. American Journal of Medical Genetics, Part A, 2021, 185, 575-578.	0.7	1
14	GUÃA: a digital platform to facilitate result disclosure in genetic counseling. Genetics in Medicine, 2021, 23, 942-949.	1.1	20
15	Drosophila RASopathy models identify disease subtype differences and biomarkers of drug efficacy. IScience, 2021, 24, 102306.	1.9	12
16	Biallelic loss-of-function variants in KCNJ16 presenting with hypokalemic metabolic acidosis. European Journal of Human Genetics, 2021, 29, 1566-1569.	1.4	12
17	â€œIs that something that should concern me?â€: a qualitative exploration of parent understanding of their childâ€™s genomic test results. Human Genetics and Genomics Advances, 2021, 2, 100027.	1.0	8
18	Mechanisms of Congenital Heart Disease Caused by NAA15 Haploinsufficiency. Circulation Research, 2021, 128, 1156-1169.	2.0	27

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19	Novel Variant Findings and Challenges Associated With the Clinical Integration of Genomic Testing. JAMA Pediatrics, 2021, 175, e205906.	3.3	39
20	GenomeDiver: a platform for phenotype-guided medical genomic diagnosis. Genetics in Medicine, 2021, 23, 1998-2002.	1.1	3
21	Dysautonomia in hypermobile <scp>Ehlersâ€“Danlos</scp> syndrome and hypermobility spectrum disorders is associated with exercise intolerance and cardiac atrophy. American Journal of Medical Genetics, Part A, 2021, 185, 3754-3761.	0.7	9
22	Myopathic Cardiac Genotypes Increase Risk for Myocarditis. JACC Basic To Translational Science, 2021, 6, 584-592.	1.9	36
23	NPSV: A simulation-driven approach to genotyping structural variants in whole-genome sequencing data. GigaScience, 2021, 10, .	3.3	4
24	Burden of Cardiomyopathic Genetic Variation in Lethal Pediatric Myocarditis. Circulation Genomic and Precision Medicine, 2021, 14, e003426.	1.6	7
25	Downregulation of exhausted cytotoxic T cells in gene expression networks of multisystem inflammatory syndrome in children. Nature Communications, 2021, 12, 4854.	5.8	42
26	Genetic Testing for Heritable Cardiovascular Diseases in Pediatric Patients: A Scientific Statement From the American Heart Association. Circulation Genomic and Precision Medicine, 2021, 14, e000086.	1.6	43
27	Hope versus reality: Parent expectations of genomic testing. Patient Education and Counseling, 2021, 104, 2073-2079.	1.0	10
28	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.	0.7	21
29	Sequential Defects in Cardiac Lineage Commitment and Maturation Cause Hypoplastic Left Heart Syndrome. Circulation, 2021, 144, 1409-1428.	1.6	29
30	SPRED2 loss-of-function causes a recessive Noonan syndrome-like phenotype. American Journal of Human Genetics, 2021, 108, 2112-2129.	2.6	23
31	DPH1 syndrome: two novel variants and structural and functional analyses of seven missense variants identified in syndromic patients. European Journal of Human Genetics, 2020, 28, 64-75.	1.4	15
32	The CHD4-related syndrome: a comprehensive investigation of the clinical spectrum, genotypeâ€“phenotype correlations, and molecular basis. Genetics in Medicine, 2020, 22, 389-397.	1.1	53
33	Proteomic Analysis of an Induced Pluripotent Stem Cell Model Reveals Strategies to Treat Juvenile Myelomonocytic Leukemia. Journal of Proteome Research, 2020, 19, 194-203.	1.8	8
34	Elucidation of de novo small insertion/deletion biology with parentâ€ofâ€origin phasing. Human Mutation, 2020, 41, 800-806.	1.1	3
35	Advancing <scp>RAS/RASopathy</scp> therapies: An NCIâ€sponsored intramural and extramural collaboration for the study of <scp>RASopathies</scp>. American Journal of Medical Genetics, Part A, 2020, 182, 866-876.	0.7	40
36	Mapping Systemic Inflammation and Antibody Responses in Multisystem Inflammatory Syndrome in Children (MIS-C). Cell, 2020, 183, 982-995.e14.	13.5	440

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37	Sampling the host response to SARS-CoV-2 in hospitals under siege. Nature Medicine, 2020, 26, 1157-1158.	15.2	27
38	Complex Autoinflammatory Syndrome Unveils Fundamental Principles of JAK1 Kinase Transcriptional and Biochemical Function. Immunity, 2020, 53, 672-684.e11.	6.6	66
39	Biallelic MADD variants cause a phenotypic spectrum ranging from developmental delay to a multisystem disorder. Brain, 2020, 143, 2437-2453.	3.7	21
40	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. American Journal of Human Genetics, 2020, 107, 499-513.	2.6	48
41	Headaches in hypermobility syndromes: A pain in the neck?. American Journal of Medical Genetics, Part A, 2020, 182, 2902-2908.	0.7	19
42	Systems Analysis Implicates WAVE2ÂComplex in the Pathogenesis ofÂDevelopmental Left-Sided ObstructiveÂHeart Defects. JACC Basic To Translational Science, 2020, 5, 376-386.	1.9	15
43	Variants in ADRB1 and CYP2C9: Association with Response to Atenolol and Losartan in Marfan Syndrome. Journal of Pediatrics, 2020, 222, 213-220.e5.	0.9	8
44	Genomic analyses implicate noncoding de novo variants in congenital heart disease. Nature Genetics, 2020, 52, 769-777.	9.4	97
45	De Novo Damaging Variants, Clinical Phenotypes, and Post-Operative Outcomes in Congenital Heart Disease. Circulation Genomic and Precision Medicine, 2020, 13, e002836.	1.6	30
46	De novo variants in exomes of congenital heart disease patients identify risk genes and pathways. Genome Medicine, 2020, 12, 9.	3.6	43
47	Enabling Technologies for Personalized and Precision Medicine. Trends in Biotechnology, 2020, 38, 497-518.	4.9	169
48	Congenital heart defects in Noonan syndrome: Diagnosis, management, and treatment. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 73-80.	0.7	68
49	EM-mosaic detects mosaic point mutations that contribute to congenital heart disease. Genome Medicine, 2020, 12, 42.	3.6	17
50	The Phosphatase CSW Controls Life Span by Insulin Signaling and Metabolism Throughout Adult Life in Drosophila. Frontiers in Genetics, 2020, 11, 364.	1.1	8
51	Repeating or spacing learning sessions are strategies for memory improvement with shared molecular and neuronal components. Neurobiology of Learning and Memory, 2020, 172, 107233.	1.0	2
52	Rare genetic variation at transcription factor binding sites modulates local DNA methylation profiles. PLoS Genetics, 2020, 16, e1009189.	1.5	27
53	GATA6 mutations in hiPSCs inform mechanisms for maldevelopment of the heart, pancreas, and diaphragm. ELife, 2020, 9, .	2.8	31
54	De Novo Missense Variants in FBXW11 Cause Diverse Developmental Phenotypes Including Brain, Eye, and Digit Anomalies. American Journal of Human Genetics, 2019, 105, 640-657.	2.6	31

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55	American Pediatric Society 2019 Presidential Address: striving to be a lasting blessing to the community. Pediatric Research, 2019, 86, 428-431.	1.1	0
56	De novo and recessive forms of congenital heart disease have distinct genetic and phenotypic landscapes. Nature Communications, 2019, 10, 4722.	5.8	58
57	Histone H2B monoubiquitination regulates heart development via epigenetic control of cilia motility. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 14049-14054.	3.3	30
58	The Genomic Medicine Integrative Research Framework: A Conceptual Framework for Conducting Genomic Medicine Research. American Journal of Human Genetics, 2019, 104, 1088-1096.	2.6	35
59	Hypertrophic Cardiomyopathy in Noonan Syndrome TreatedÂbyÂMEK-Inhibition. Journal of the American College of Cardiology, 2019, 73, 2237-2239.	1.2	96
60	Heterozygous loss-of-function variants of MEIS2 cause a triad of palatal defects, congenital heart defects, and intellectual disability. European Journal of Human Genetics, 2019, 27, 278-290.	1.4	30
61	ClinGenâ€™s RASopathy Expert Panel consensus methods for variant interpretation. Genetics in Medicine, 2018, 20, 1334-1345.	1.1	126
62	Functional Dysregulation of CDC42 Causes Diverse Developmental Phenotypes. American Journal of Human Genetics, 2018, 102, 309-320.	2.6	138
63	Distinct epigenetic programs regulate cardiac myocyte development and disease in the human heart in vivo. Nature Communications, 2018, 9, 391.	5.8	181
64	Robust identification of mosaic variants in congenital heart disease. Human Genetics, 2018, 137, 183-193.	1.8	43
65	Clinical Presentation and Natural History of Hypertrophic Cardiomyopathy in RASopathies. Heart Failure Clinics, 2018, 14, 225-235.	1.0	44
66	Robust identification of deletions in exome and genome sequence data based on clustering of Mendelian errors. Human Mutation, 2018, 39, 870-881.	1.1	3
67	Genetic Basis for Congenital Heart Disease: Revisited: A Scientific Statement From the American Heart Association. Circulation, 2018, 138, e653-e711.	1.6	387
68	Assessing the geneâ€“disease association of 19 genes with the RASopathies using the ClinGen gene curation framework. Human Mutation, 2018, 39, 1485-1493.	1.1	66
69	The Clinical Sequencing Evidence-Generating Research Consortium: Integrating Genomic Sequencing in Diverse and Medically Underserved Populations. American Journal of Human Genetics, 2018, 103, 319-327.	2.6	122
70	Identification of rare de novo epigenetic variations in congenital disorders. Nature Communications, 2018, 9, 2064.	5.8	82
71	The Congenital Heart Disease Genetic Network Study: Cohort description. PLoS ONE, 2018, 13, e0191319.	1.1	82
72	Structural, Functional, and Clinical Characterization of a NovelPTPN11Mutation Cluster Underlying Noonan Syndrome. Human Mutation, 2017, 38, 451-459.	1.1	39

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73	Cathepsin K Deficiency Ameliorates Systemic Lupus Erythematosus-like Manifestations in Faslpr Mice. Journal of Immunology, 2017, 198, 1846-1854.	0.4	21
74	The Spacing Effect for Structural Synaptic Plasticity Provides Specificity and Precision in Plastic Changes. Journal of Neuroscience, 2017, 37, 4992-5007.	1.7	12
75	Autosomal Recessive Cardiomyopathy Presenting as Acute Myocarditis. Journal of the American College of Cardiology, 2017, 69, 1653-1665.	1.2	94
76	Contribution of rare inherited and de novo variants in 2,871 congenital heart disease probands. Nature Genetics, 2017, 49, 1593-1601.	9.4	624
77	Human Engineered Cardiac Tissues Created Using Induced Pluripotent Stem Cells Reveal Functional Characteristics of BRAF-Mediated Hypertrophic Cardiomyopathy. PLoS ONE, 2016, 11, e0146697.	1.1	72
78	When Participants in Genomic Research Grow Up: Contact and Consent atÂthe Age of Majority. Journal of Pediatrics, 2016, 168, 226-231.e1.	0.9	17
79	Autonomous and Non-autonomous Defects Underlie Hypertrophic Cardiomyopathy in BRAF-Mutant hiPSC-Derived Cardiomyocytes. Stem Cell Reports, 2016, 7, 355-369.	2.3	33
80	SHOC2 subcellular shuttling requires the KEKE motif-rich region andN-terminal leucine-rich repeat domain and impacts on ERK signalling. Human Molecular Genetics, 2016, 25, 3824-3835.	1.4	17
81	Loss of RNA expression and allele-specific expression associated with congenital heart disease. Nature Communications, 2016, 7, 12824.	5.8	51
82	Genetic Discovery for Congenital Heart Defects. , 2016, , 355-360.		2
83	De Novo and Rare Variants at Multiple Loci Support the Oligogenic Origins of Atrioventricular Septal Heart Defects. PLoS Genetics, 2016, 12, e1005963.	1.5	92
84	Construction of Defined Human Engineered Cardiac Tissues to Study Mechanisms of Cardiac Cell Therapy. Journal of Visualized Experiments, 2016, , e53447.	0.2	9
85	The Hole and the Whole: Lessons from Manipulation of Nipbl Deficiency. PLoS Biology, 2016, 14, e2000494.	2.6	0
86	Activating Mutations Affecting the Dbl Homology Domain of SOS2 Cause Noonan Syndrome. Human Mutation, 2015, 36, 1080-1087.	1.1	67
87	Rapidly progressive hypertrophic cardiomyopathy in an infant with Noonan syndrome with multiple lentigines: Palliative treatment with a rapamycin analog. American Journal of Medical Genetics, Part A, 2015, 167, 744-751.	0.7	53
88	De novo mutations in congenital heart disease with neurodevelopmental and other congenital anomalies. Science, 2015, 350, 1262-1266.	6.0	646
89	History of Our Understanding of the Causes of Congenital Heart Disease. Circulation: Cardiovascular Genetics, 2015, 8, 529-536.	5.1	20
90	MATR3 disruption in human and mouse associated with bicuspid aortic valve, aortic coarctation and patent ductus arteriosus. Human Molecular Genetics, 2015, 24, 2375-2389.	1.4	90

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91	Cardiomyopathies in Noonan syndrome and the other RASopathies. Progress in Pediatric Cardiology, 2015, 39, 13-19.	0.2	99
92	Molecular Diversity and Associated Phenotypic Spectrum of GermlineCBLMutations. Human Mutation, 2015, 36, 787-796.	1.1	36
93	Myeloid Dysregulation in a Human Induced Pluripotent Stem Cell Model of PTPN11 -Associated Juvenile Myelomonocytic Leukemia. Cell Reports, 2015, 13, 504-515.	2.9	79
94	Activating mutations in RRAS underlie a phenotype within the RASopathy spectrum and contribute to leukaemogenesis. Human Molecular Genetics, 2014, 23, 4315-4327.	1.4	114
95	Concise Review: Drug Discovery in the Age of the Induced Pluripotent Stem Cell. Stem Cells Translational Medicine, 2014, 3, 500-509.	1.6	65
96	A PTPN11 allele encoding a catalytically impaired SHP2 protein in a patient with a Noonan syndrome phenotype. American Journal of Medical Genetics, Part A, 2014, 164, 2351-2355.	0.7	12
97	RAF1 mutations in childhood-onset dilated cardiomyopathy. Nature Genetics, 2014, 46, 635-639.	9.4	69
98	Frequency of Aortic Dilation in Noonan Syndrome. American Journal of Cardiology, 2014, 113, 368-371.	0.7	22
99	Practical Guidance on Informed Consent for Pediatric Participants in a Biorepository. Mayo Clinic Proceedings, 2014, 89, 1471-1480.	1.4	27
100	Complex Genetics and the Etiology of Human Congenital Heart Disease. Cold Spring Harbor Perspectives in Medicine, 2014, 4, a013953-a013953.	2.9	118
101	Increased Frequency of De Novo Copy Number Variants in Congenital Heart Disease by Integrative Analysis of Single Nucleotide Polymorphism Array and Exome Sequence Data. Circulation Research, 2014, 115, 884-896.	2.0	229
102	Identification and Purification of Human Induced Pluripotent Stem Cell-Derived Atrial-Like Cardiomyocytes Based on Sarcolipin Expression. PLoS ONE, 2014, 9, e101316.	1.1	40
103	Genetics of Congenital Heart Disease. Circulation Research, 2013, 112, 707-720.	2.0	491
104	Noonan syndrome. Lancet, The, 2013, 381, 333-342.	6.3	608
105	De novo mutations in histone-modifying genes in congenital heart disease. Nature, 2013, 498, 220-223.	13.7	798
106	Characteristics of children and young adults with Marfan syndrome and aortic root dilation in a randomized trial comparing atenolol and losartan therapy. American Heart Journal, 2013, 165, 828-835.e3.	1.2	59
107	Recent advances in understanding the genetics of congenital heart defects. Current Opinion in Pediatrics, 2013, 25, 561-566.	1.0	16
108	The Congenital Heart Disease Genetic Network Study. Circulation Research, 2013, 112, 698-706.	2.0	142

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109	Effect of Copy Number Variants on Outcomes for Infants With Single Ventricle Heart Defects. Circulation: Cardiovascular Genetics, 2013, 6, 444-451.	5.1	89
110	Cathepsin K Deficiency Reduces Elastase Perfusionâ€“Induced Abdominal Aortic Aneurysms in Mice. Arteriosclerosis, Thrombosis, and Vascular Biology, 2012, 32, 15-23.	1.1	89
111	The Good SHP2 Association. Circulation: Cardiovascular Genetics, 2012, 5, 271-273.	5.1	4
112	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*. Journal of Biological Chemistry, 2012, 287, 27066-27077.	1.6	35
113	Noonan syndrome and clinically related disorders. Best Practice and Research in Clinical Endocrinology and Metabolism, 2011, 25, 161-179.	2.2	303
114	Cyclosporine attenuates cardiomyocyte hypertrophy induced by RAF1 mutants in Noonan and LEOPARD syndromes. Journal of Molecular and Cellular Cardiology, 2011, 51, 4-15.	0.9	21
115	Induced pluripotent stem cell-derived cardiomyocytes as models for genetic cardiovascular disorders. Current Opinion in Cardiology, 2011, 26, 223-229.	0.8	32
116	SOS1 mutations in Noonan syndrome: molecular spectrum, structural insights on pathogenic effects, and genotype-phenotype correlations. Human Mutation, 2011, 32, 760-772.	1.1	97
117	RAS signaling pathway mutations and hypertrophic cardiomyopathy: getting into and out of the thick of it. Journal of Clinical Investigation, 2011, 121, 844-847.	3.9	49
118	Heterozygous Germline Mutations in the CBL Tumor-Suppressor Gene Cause a Noonan Syndrome-like Phenotype. American Journal of Human Genetics, 2010, 87, 250-257.	2.6	221
119	Protein Tyrosine Phosphatase PTPN14 Is a Regulator of Lymphatic Function and Choanal Development in Humans. American Journal of Human Genetics, 2010, 87, 436-444.	2.6	75
120	Patient-specific induced pluripotent stem-cell-derived models of LEOPARD syndrome. Nature, 2010, 465, 808-812.	13.7	672
121	A restricted spectrum of NRAS mutations causes Noonan syndrome. Nature Genetics, 2010, 42, 27-29.	9.4	271
122	Disorders of dysregulated signal traffic through the RASâ€“MAPK pathway: phenotypic spectrum and molecular mechanisms. Annals of the New York Academy of Sciences, 2010, 1214, 99-121.	1.8	167
123	Fgfr3 Is a Transcriptional Target of Ap2Î´ and Ash2l-Containing Histone Methyltransferase Complexes. PLoS ONE, 2009, 4, e8535.	1.1	16
124	Phosphatase-defective LEOPARD syndrome mutations in PTPN11 gene have gain-of-function effects during Drosophila development. Human Molecular Genetics, 2009, 18, 193-201.	1.4	82
125	GermlineBRAFmutations in Noonan, LEOPARD, and cardiofaciocutaneous syndromes: Molecular diversity and associated phenotypic spectrum. Human Mutation, 2009, 30, 695-702.	1.1	251
126	Mutation of SHOC2 promotes aberrant protein N-myristoylation and causes Noonan-like syndrome with loose anagen hair. Nature Genetics, 2009, 41, 1022-1026.	9.4	358

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127	The Phosphatase SHP2 Regulates the Spacing Effect for Long-Term Memory Induction. Cell, 2009, 139, 186-198.	13.5	139
128	Deficiency and Inhibition of Cathepsin K Reduce Body Weight Gain and Increase Glucose Metabolism in Mice. Arteriosclerosis, Thrombosis, and Vascular Biology, 2008, 28, 2202-2208.	1.1	78
129	Diverse driving forces underlie the invariant occurrence of the T42A, E139D, I282V and T468M SHP2 amino acid substitutions causing Noonan and LEOPARD syndromes. Human Molecular Genetics, 2008, 17, 2018-2029.	1.4	79
130	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.	0.9	28
131	The genetics of congenital heart disease: a review of recent developments. Current Opinion in Cardiology, 2007, 22, 200-206.	0.8	57
132	Genetic Basis for Congenital Heart Defects: Current Knowledge. Circulation, 2007, 115, 3015-3038.	1.6	719
133	Gain-of-function SOS1 mutations cause a distinctive form of Noonan syndrome. Nature Genetics, 2007, 39, 75-79.	9.4	523
134	Gain-of-function RAF1 mutations cause Noonan and LEOPARD syndromes with hypertrophic cardiomyopathy. Nature Genetics, 2007, 39, 1007-1012.	9.4	624
135	Diversity and Functional Consequences of Germline and Somatic PTPN11 Mutations in Human Disease. American Journal of Human Genetics, 2006, 78, 279-290.	2.6	352
136	Germline Missense Mutations Affecting KRAS Isoform B Are Associated with a Severe Noonan Syndrome Phenotype. American Journal of Human Genetics, 2006, 79, 129-135.	2.6	205
137	Noonan syndrome and related disorders: dysregulated RAS-mitogen activated protein kinase signal transduction. Human Molecular Genetics, 2006, 15, R220-R226.	1.4	177
138	Transgenic Drosophila models of Noonan syndrome causing PTPN11 gain-of-function mutations. Human Molecular Genetics, 2006, 15, 543-553.	1.4	66
139	Neurofibromatosis-Noonan syndrome: Molecular evidence of the concurrence of both disorders in a patient. American Journal of Medical Genetics, Part A, 2005, 136A, 242-245.	0.7	74
140	The mutational spectrum of PTPN11 in juvenile myelomonocytic leukemia and Noonan syndrome/myeloproliferative disease. Blood, 2005, 106, 2183-2185.	0.6	247
141	Germ-line and somatic PTPN11 mutations in human disease. European Journal of Medical Genetics, 2005, 48, 81-96.	0.7	128
142	NOONAN SYNDROME AND RELATED DISORDERS: Genetics and Pathogenesis. Annual Review of Genomics and Human Genetics, 2005, 6, 45-68.	2.5	306
143	Noonan syndrome-associated SHP2/PTPN11 mutants cause EGF-dependent prolonged GAB1 binding and sustained ERK2/MAPK1 activation. Human Mutation, 2004, 23, 267-277.	1.1	177
144	Paternal Germline Origin and Sex-Ratio Distortion in Transmission of PTPN11 Mutations in Noonan Syndrome. American Journal of Human Genetics, 2004, 75, 492-497.	2.6	76

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145	Genetic basis of congenital heart disease. Current Opinion in Cardiology, 2004, 19, 110-115.	0.8	68
146	Genetic evidence for lineage-related and differentiation stage-related contribution of somatic PTPN11 mutations to leukemogenesis in childhood acute leukemia. Blood, 2004, 104, 307-313.	0.6	265
147	PTPN11 Mutational Spectrum in Juvenile Myelomonocytic Leukemia and Noonan Syndrome.. Blood, 2004, 104, 3417-3417.	0.6	1
148	Somatic mutations in PTPN11 in juvenile myelomonocytic leukemia, myelodysplastic syndromes and acute myeloid leukemia. Nature Genetics, 2003, 34, 148-150.	9.4	960
149	PTPN11 Mutations in Noonan Syndrome: Molecular Spectrum, Genotype-Phenotype Correlation, and Phenotypic Heterogeneity. American Journal of Human Genetics, 2002, 70, 1555-1563.	2.6	680
150	Absence of PTPN11 mutations in 28 cases of cardiofaciocutaneous (CFC) syndrome. Human Genetics, 2002, 111, 421-427.	1.8	45
151	Mutations in PTPN11, encoding the protein tyrosine phosphatase SHP-2, cause Noonan syndrome. Nature Genetics, 2001, 29, 465-468.	9.4	1,555
152	WHIM syndrome, an autosomal dominant disorder: Clinical, hematological, and molecular studies. American Journal of Medical Genetics Part A, 2000, 91, 368-376.	2.4	193
153	Malignant fibrous histiocytoma: Inherited and sporadic forms have loss of heterozygosity at chromosome bands 9p21-22?evidence for a common genetic defect. Genes Chromosomes and Cancer, 2000, 27, 191-195.	1.5	36
154	Mutations in TFAP2B cause Char syndrome, a familial form of patent ductus arteriosus. Nature Genetics, 2000, 25, 42-46.	9.4	252
155	WHIM syndrome, an autosomal dominant disorder: Clinical, hematological, and molecular studies. , 2000, 91, 368.		2
156	Malignant fibrous histiocytoma: Inherited and sporadic forms have loss of heterozygosity at chromosome bands 9p21â€“22â€”evidence for a common genetic defect. , 2000, 27, 191.		1
157	Mutations in a new gene encoding a thiamine transporter cause thiamine-responsive megaloblastic anaemia syndrome. Nature Genetics, 1999, 22, 309-312.	9.4	201
158	Determination of Bone Markers in Pycnodysostosis: Effects of Cathepsin K Deficiency on Bone Matrix Degradation. Journal of Bone and Mineral Research, 1999, 14, 1902-1908.	3.1	128
159	Recent Advances in the Understanding of Genetic Causes of Congenital Heart Defects. Fetal and Pediatric Pathology, 1998, 18, 501-527.	0.3	0
160	RECENT ADVANCES IN THE UNDERSTANDING OF GENETIC CAUSES OF CONGENITAL HEART DEFECTS. Fetal and Pediatric Pathology, 1998, 18, 501-527.	0.3	0
161	Genetic mapping of the cleidocranial dysplasia (CCD) locus on chromosome band 6p21 to include a microdeletion. American Journal of Medical Genetics Part A, 1995, 58, 200-205.	2.4	39
162	Linkage of pycnodysostosis to chromosome 1q21 by homozygosity mapping. Nature Genetics, 1995, 10, 235-237.	9.4	94

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163	Leigh syndrome and hypertrophic cardiomyopathy in an infant with a mitochondrial DNA point mutation (T8993G). American Journal of Medical Genetics Part A, 1994, 50, 265-271.	2.4	124
164	Noonan Syndrome and Other RAS/MAPK Pathway Syndromes. , 0, , 122-130.		0
165	Prevalence of Genetic Diagnoses in a Cohort With Valvar Pulmonary Stenosis. Circulation Genomic and Precision Medicine, 0, , .	1.6	0