

Nathaniel H Robin

List of Publications by Citations

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

86
papers

1,895
citations

22
h-index

41
g-index

98
ext. papers

2,180
ext. citations

3.9
avg, IF

4.37
L-index

#	Paper	IF	Citations
86	Mutations in CDMP1 cause autosomal dominant brachydactyly type C. <i>Nature Genetics</i> , 1997 , 17, 18-9	36.3	235
85	Defining the clinical spectrum of deletion 22q11.2. <i>Journal of Pediatrics</i> , 2005 , 147, 90-6	3.6	182
84	Clinically relevant single gene or intragenic deletions encompassing critical neurodevelopmental genes in patients with developmental delay, mental retardation, and/or autism spectrum disorders. <i>American Journal of Medical Genetics, Part A</i> , 2011 , 155A, 2386-96	2.5	125
83	Polymicrogyria and deletion 22q11.2 syndrome: window to the etiology of a common cortical malformation. <i>American Journal of Medical Genetics, Part A</i> , 2006 , 140, 2416-25	2.5	113
82	Parental attitudes toward genetic testing for pediatric deafness. <i>American Journal of Human Genetics</i> , 2000 , 67, 1621-5	11	101
81	Duty to warn at-risk relatives for genetic disease: genetic counselors' clinical experience. <i>American Journal of Medical Genetics Part A</i> , 2003 , 119C, 27-34		83
80	Medical Geneticists' duty to warn at-risk relatives for genetic disease. <i>American Journal of Medical Genetics Part A</i> , 2003 , 120A, 374-80		66
79	Opitz G/BBB syndrome: clinical comparisons of families linked to Xp22 and 22q, and a review of the literature. <i>American Journal of Medical Genetics Part A</i> , 1996 , 62, 305-17		66
78	The multidisciplinary evaluation and management of cleft lip and palate. <i>Southern Medical Journal</i> , 2006 , 99, 1111-20	0.6	55
77	Amniotic constriction band: a multidisciplinary assessment of etiology and clinical presentation. <i>Journal of Bone and Joint Surgery - Series A</i> , 2009 , 91 Suppl 4, 68-75	5.6	53
76	Genetic testing in cardiovascular disease. <i>Journal of the American College of Cardiology</i> , 2007 , 50, 727-37	15.1	51
75	The recurrent distal 22q11.2 microdeletions are often de novo and do not represent a single clinical entity: a proposed categorization system. <i>Genetics in Medicine</i> , 2014 , 16, 92-100	8.1	37
74	Clefting, amniotic bands, and polydactyly: a distinct phenotype that supports an intrinsic mechanism for amniotic band sequence. <i>American Journal of Medical Genetics, Part A</i> , 2005 , 137A, 298-301	5	37
73	Disorganization in mice and humans and its relation to sporadic birth defects. <i>American Journal of Medical Genetics Part A</i> , 1997 , 73, 425-36		32
72	Child with mosaic variegated aneuploidy and embryonal rhabdomyosarcoma. <i>American Journal of Medical Genetics Part A</i> , 1999 , 82, 20-4		31
71	New finding of Schinzel-Giedion syndrome: a case with a malignant sacrococcygeal teratoma. <i>American Journal of Medical Genetics Part A</i> , 1993 , 47, 852-6		31
70	Severe oculocerebrocutaneous (Delleman) syndrome: overlap with Goldenhar anomaly. <i>American Journal of Medical Genetics Part A</i> , 1998 , 78, 282-5		29

69	Disorganization in mice and humans. <i>American Journal of Medical Genetics Part A</i> , 2001 , 101, 334-338		29
68	De Novo Truncating Variants in SON Cause Intellectual Disability, Congenital Malformations, and Failure to Thrive. <i>American Journal of Human Genetics</i> , 2016 , 99, 720-727	11	28
67	What Information Do Parents of Newborns With Cleft Lip, Palate, or Both Want to Know?. <i>Cleft Palate-Craniofacial Journal</i> , 2001 , 38, 55-58	1.9	28
66	Favorable prognosis for children with Pfeiffer syndrome types 2 and 3: Implications for classification 1998 , 75, 240-244		26
65	Unusual craniofacial dysmorphism due to prenatal alcohol and cocaine exposure. <i>Teratology</i> , 1994 , 50, 160-4		24
64	Adult with an interstitial deletion of chromosome 10 [del(10)(q25.1q25.3)]: overlap with Coffin-Lowry syndrome. <i>American Journal of Medical Genetics Part A</i> , 2000 , 95, 93-8		21
63	Nonpenetrance in FGFR3-associated coronal synostosis syndrome 1998 , 80, 296-297		20
62	Distal 5q deletion syndrome: phenotypic correlations. <i>American Journal of Medical Genetics Part A</i> , 2001 , 103, 63-8		20
61	Microcephaly-lymphedema-chorioretinal dysplasia: A unique genetic syndrome with variable expression and possible characteristic facial appearance 1999 , 86, 215-218		19
60	Craniosynostosis, Philadelphia type: a new autosomal dominant syndrome with sagittal craniosynostosis and syndactyly of the fingers and toes. <i>American Journal of Medical Genetics Part A</i> , 1996 , 62, 184-91		19
59	Syntelencephaly in an infant of a diabetic mother. <i>American Journal of Medical Genetics Part A</i> , 1996 , 66, 433-7		19
58	Cardiomyopathy in Coffin-Lowry syndrome. <i>American Journal of Medical Genetics Part A</i> , 2004 , 128A, 176-8		16
57	IRF6 mutations in mixed isolated familial clefting. <i>American Journal of Medical Genetics, Part A</i> , 2010 , 152A, 3107-9	2.5	14
56	Classical Noonan syndrome is not associated with deletions of 22q11. <i>American Journal of Medical Genetics Part A</i> , 1995 , 56, 94-6		14
55	Trisomy 18: A single-center evaluation of management trends and experience with aggressive obstetric or neonatal intervention. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170A, 838-46	2.5	14
54	Clinical relevance of small copy-number variants in chromosomal microarray clinical testing. <i>Genetics in Medicine</i> , 2017 , 19, 377-385	8.1	13
53	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an epismutation of X chromosomes in females. <i>American Journal of Human Genetics</i> , 2021 , 108, 502-516	11	12
52	Newborn craniofacial malformations: orofacial clefting and craniosynostosis. <i>Clinics in Perinatology</i> , 2015 , 42, 321-36, viii	2.8	11

51	Duplication 14(q24.3q31) in a father and daughter: delineation of a possible imprinted region. <i>American Journal of Medical Genetics Part A</i> , 1997 , 71, 361-5		11
50	Stickler Syndrome: A Review of Clinical Manifestations and the Genetics Evaluation. <i>Journal of Personalized Medicine</i> , 2020 , 10,	3.6	11
49	Trisomy 18: A survey of opinions, attitudes, and practices of neonatologists. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170, 2638-43	2.5	11
48	Copy number analysis of NIPBL in a cohort of 510 patients reveals rare copy number variants and a mosaic deletion. <i>Molecular Genetics & Genomic Medicine</i> , 2014 , 2, 115-23	2.3	10
47	Frontonasal malformation and cloacal exstrophy: a previously unreported association. <i>American Journal of Medical Genetics Part A</i> , 1996 , 61, 75-8		10
46	Familial transmission of oculoauriculovertebral spectrum (Goldenhar syndrome) is not due to mutations in either EYA1 or SALL1. <i>American Journal of Medical Genetics, Part A</i> , 2009 , 149A, 535-8	2.5	9
45	Use of array comparative genome hybridization in orofacial clefting. <i>Journal of Craniofacial Surgery</i> , 2010 , 21, 1591-4	1.2	9
44	De novo interstitial deletion of the long arm of chromosome 3: 46,XX,del(3)(q25.1q26.1). <i>Clinical Genetics</i> , 1993 , 44, 335-7	4	8
43	The use by Alabama pediatricians of genetics consultation in the evaluation of developmental delay. <i>American Journal of Medical Genetics, Part A</i> , 2008 , 146A, 421-5	2.5	8
42	Non-immune hydrops fetalis associated with impaired fetal movement: a case report and review. <i>American Journal of Medical Genetics Part A</i> , 1994 , 53, 251-4		8
41	A previously unrecognized 22q13.2 microdeletion syndrome that encompasses TCF20 and TNFRSF13C. <i>American Journal of Medical Genetics, Part A</i> , 2018 , 176, 2791-2797	2.5	8
40	Novel de novo pathogenic variant in the NR2F2 gene in a boy with congenital heart defect and dysmorphic features. <i>American Journal of Medical Genetics, Part A</i> , 2018 , 176, 1423-1426	2.5	7
39	IRF6 Sequencing in Interrupted Clefting. <i>Cleft Palate-Craniofacial Journal</i> , 2016 , 53, 373-6	1.9	7
38	Array comparative genomic hybridisation testing in CHD. <i>Cardiology in the Young</i> , 2015 , 25, 1155-72	1	7
37	Imperforate anus is a rare associated finding in blepharocheilodontic syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2010 , 152A, 438-40	2.5	7
36	The primary care physician's approach to congenital anomalies. <i>Primary Care - Clinics in Office Practice</i> , 2004 , 31, 605-19, x	2.2	7
35	Incidental detection of cancer predisposition gene copy number variations by array comparative genomic hybridization. <i>Journal of Pediatrics</i> , 2014 , 165, 1057-9.e1-4	3.6	6
34	Sibs with cleidocranial dysplasia born to normal parents: germ line mosaicism?. <i>American Journal of Medical Genetics Part A</i> , 1997 , 69, 348-51		6

33	Genetic and reproductive knowledge among adolescents and adults with cystic fibrosis. <i>Chest</i> , 2008 , 133, 1533	5.3	6
32	Genetic testing and genetic counseling for deafness: the future is here. <i>Laryngoscope</i> , 2001 , 111, 715-8	3.6	6
31	Craniosynostosis and radial ray defect: a rare presentation of 22q11.2 deletion syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2013 , 161A, 2024-6	2.5	5
30	A genetic model for cloacal exstrophy, the extreme cloacal malformation. <i>Journal of Pediatric Urology</i> , 2007 , 3, 214-7	1.5	4
29	Classic phenotype of Coffin-Lowry syndrome in a female with stimulus-induced drop episodes and a genotype with preserved N-terminal kinase domain. <i>American Journal of Medical Genetics, Part A</i> , 2014 , 164A, 516-21	2.5	3
28	Further delineation of the Kapur-Toriello syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2010 , 152A, 1013-5	2.5	3
27	The duty to warn at-risk relatives-The experience of genetic counselors and medical geneticists. <i>American Journal of Medical Genetics, Part A</i> , 2020 , 182, 314-321	2.5	3
26	Foramen magnum compression in Coffin-Lowry syndrome: A case report. <i>American Journal of Medical Genetics, Part A</i> , 2017 , 173, 1087-1089	2.5	2
25	The development and implementation of an in-service exam for medical genetics residency programs. <i>Genetics in Medicine</i> , 2012 , 14, 552-7	8.1	2
24	Genetic drift. The awful truth. <i>American Journal of Medical Genetics Part A</i> , 1997 , 71, 375-7		2
23	Simultaneous osteosarcoma and renal cell carcinoma with BRCA1 mutation in a young male adult with prior oligodendroglioma. <i>Pediatric Blood and Cancer</i> , 2020 , 67, e28116	3	2
22	Cleft palate in a patient with the nested 22q11.2 LCR C to D deletion. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170A, 260-2	2.5	2
21	Congenital Heart Defects 2013 , 1-51		1
20	Educating the adolescent and young adult with cystic fibrosis about their reproductive risks and options. <i>Chest</i> , 2013 , 143, 580-581	5.3	1
19	Treatment for genetic diseases. <i>Current Opinion in Pediatrics</i> , 2008 , 20, 625-7	3.2	1
18	Genetic drift. A smile. <i>American Journal of Medical Genetics Part A</i> , 2003 , 118A, 404-6		1
17	Clinical and molecular studies of brachydactyly type D. <i>American Journal of Medical Genetics Part A</i> , 1999 , 85, 413-8		1
16	Congenital Muscular Torticollis. <i>Pediatrics in Review</i> , 1996 , 17, 374-375	1.1	1

15	Genetic consultations in cases of unexplained fractures and haemorrhage: an evidence-based approach. <i>Current Opinion in Pediatrics</i> , 2021 , 33, 3-18	3.2	1
14	Ethical and Legal Issues 2018 , 93-100		1
13	The natural history of OTOF-related auditory neuropathy spectrum disorders: a multicenter study. <i>Human Genetics</i> , 2021 , 1	6.3	1
12	Genetic testing hearing loss: The challenge of non syndromic mimics. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2021 , 150, 110872	1.7	1
11	Inherited cause of in utero digital malformations. <i>BMJ Case Reports</i> , 2020 , 13,	0.9	0
10	Attitudes of deaf individuals towards genetic testing of genes known to cause hearing loss. <i>Clinical Ethics</i> ,147775092110635	1	0
9	Congenital Heart Defects 2020 , 3-75		0
8	Growth parameters: the cheap and easy genetic test. <i>Current Opinion in Pediatrics</i> , 2016 , 28, 679-681	3.2	0
7	JARID2 haploinsufficiency is associated with a clinically distinct neurodevelopmental syndrome. <i>Genetics in Medicine</i> , 2021 , 23, 374-383	8.1	0
6	Genetic drift. The mall test (or fun with a dysmorphologist). <i>American Journal of Medical Genetics, Part A</i> , 2011 , 155A, 2909	2.5	
5	Response to Kessler and Resta's commentary. <i>American Journal of Medical Genetics Part A</i> , 2004 , 126A, 439-439		
4	Genetic drift. A few moments. <i>American Journal of Medical Genetics Part A</i> , 2003 , 119A, 397-9		
3	The good that we do. <i>American Journal of Medical Genetics Part A</i> , 1996 , 65, 257-8		
2	Near complete deletion of KMT2D in a college student.. <i>American Journal of Medical Genetics, Part A</i> , 2022 ,	2.5	
1	The first post-natal clinical description of true mosaic complete tetrasomy 21: A case report. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 3507-3509	2.5	