Nathaniel H Robin

List of Publications by Year in Descending Order

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	Near complete deletion of KMT2D in a college student.. *American Journal of Medical Genetics, Part A*, **2022**,	2.5	
85	Genetic consultations in cases of unexplained fractures and haemorrhage: an evidence-based approach. *Current Opinion in Pediatrics*, **2021**, 33, 3-18	3.2	1
84	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females. *American Journal of Human Genetics*, **2021**, 108, 502-516	11	12
83	JARID2 haploinsufficiency is associated with a clinically distinct neurodevelopmental syndrome. *Genetics in Medicine*, **2021**, 23, 374-383	8.1	0
82	The natural history of OTOF-related auditory neuropathy spectrum disorders: a multicenter study. *Human Genetics*, **2021**, 1	6.3	1
81	The first post-natal clinical description of true mosaic complete tetrasomy 21: A case report. *American Journal of Medical Genetics, Part A*, **2021**, 185, 3507-3509	2.5	
80	Genetic testing hearing loss: The challenge of non syndromic mimics. *International Journal of Pediatric Otorhinolaryngology*, **2021**, 150, 110872	1.7	1
79	Inherited cause of in utero digital malformations. *BMJ Case Reports*, **2020**, 13,	0.9	0
78	Congenital Heart Defects **2020**, 3-75		0
77	Simultaneous osteosarcoma and renal cell carcinoma with BRCA1 mutation in a young male adult with prior oligodendroglioma. *Pediatric Blood and Cancer*, **2020**, 67, e28116	3	2
76	The duty to warn at-risk relatives-The experience of genetic counselors and medical geneticists. *American Journal of Medical Genetics, Part A*, **2020**, 182, 314-321	2.5	3
75	Stickler Syndrome: A Review of Clinical Manifestations and the Genetics Evaluation. *Journal of Personalized Medicine*, **2020**, 10,	3.6	11
74	Novel de novo pathogenic variant in the NR2F2 gene in a boy with congenital heart defect and dysmorphic features. *American Journal of Medical Genetics, Part A*, **2018**, 176, 1423-1426	2.5	7
73	A previously unrecognized 22q13.2 microdeletion syndrome that encompasses TCF20 and TNFRSF13C. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2791-2797	2.5	8
72	Ethical and Legal Issues **2018**, 93-100		1
71	Foramen magnum compression in Coffin-Lowry syndrome: A case report. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1087-1089	2.5	2
70	Clinical relevance of small copy-number variants in chromosomal microarray clinical testing. *Genetics in Medicine*, **2017**, 19, 377-385	8.1	13

69	De Novo Truncating Variants in SON Cause Intellectual Disability, Congenital Malformations, and Failure to Thrive. *American Journal of Human Genetics*, **2016**, 99, 720-727	11	28
68	IRF6 Sequencing in Interrupted Clefting. *Cleft Palate-Craniofacial Journal*, **2016**, 53, 373-6	1.9	7
67	Trisomy 18: A survey of opinions, attitudes, and practices of neonatologists. *American Journal of Medical Genetics, Part A*, **2016**, 170, 2638-43	2.5	11
66	Trisomy 18: A single-center evaluation of management trends and experience with aggressive obstetric or neonatal intervention. *American Journal of Medical Genetics, Part A*, **2016**, 170A, 838-46	2.5	14
65	Growth parameters: the cheap and easy genetic test. *Current Opinion in Pediatrics*, **2016**, 28, 679-681	3.2	0
64	Cleft palate in a patient with the nested 22q11.2 LCR C to D deletion. *American Journal of Medical Genetics, Part A*, **2016**, 170A, 260-2	2.5	2
63	Newborn craniofacial malformations: orofacial clefting and craniosynostosis. *Clinics in Perinatology*, **2015**, 42, 321-36, viii	2.8	11
62	Array comparative genomic hybridisation testing in CHD. *Cardiology in the Young*, **2015**, 25, 1155-72	1	7
61	Copy number analysis of NIPBL in a cohort of 510 patients reveals rare copy number variants and a mosaic deletion. *Molecular Genetics & Genomic Medicine*, **2014**, 2, 115-23	2.3	10
60	Incidental detection of cancer predisposition gene copy number variations by array comparative genomic hybridization. *Journal of Pediatrics*, **2014**, 165, 1057-9.e1-4	3.6	6
59	Classic phenotype of Coffin-Lowry syndrome in a female with stimulus-induced drop episodes and a genotype with preserved N-terminal kinase domain. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 516-21	2.5	3
58	The recurrent distal 22q11.2 microdeletions are often de novo and do not represent a single clinical entity: a proposed categorization system. *Genetics in Medicine*, **2014**, 16, 92-100	8.1	37
57	Congenital Heart Defects **2013**, 1-51		1
56	Craniosynostosis and radial ray defect: a rare presentation of 22q11.2 deletion syndrome. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 2024-6	2.5	5
55	Educating the adolescent and young adult with cystic fibrosis about their reproductive risks and options. *Chest*, **2013**, 143, 580-581	5.3	1
54	The development and implementation of an in-service exam for medical genetics residency programs. *Genetics in Medicine*, **2012**, 14, 552-7	8.1	2
53	Clinically relevant single gene or intragenic deletions encompassing critical neurodevelopmental genes in patients with developmental delay, mental retardation, and/or autism spectrum disorders. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 2386-96	2.5	125
52	Genetic drift. The mall test (or fun with a dysmorphologist). *American Journal of Medical Genetics, Part A*, **2011**, 155A, 2909	2.5	

51	Use of array comparative genome hybridization in orofacial clefting. *Journal of Craniofacial Surgery*, **2010**, 21, 1591-4	1.2	9
50	IRF6 mutations in mixed isolated familial clefting. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 3107-9	2.5	14
49	Imperforate anus is a rare associated finding in blepharocheilodontic syndrome. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 438-40	2.5	7
48	Further delineation of the Kapur-Toriello syndrome. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 1013-5	2.5	3
47	Familial transmission of oculoauriculovertebral spectrum (Goldenhar syndrome) is not due to mutations in either EYA1 or SALL1. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 535-8	2.5	9
46	Amniotic constriction band: a multidisciplinary assessment of etiology and clinical presentation. *Journal of Bone and Joint Surgery - Series A*, **2009**, 91 Suppl 4, 68-75	5.6	53
45	Genetic and reproductive knowledge among adolescents and adults with cystic fibrosis. *Chest*, **2008** , 133, 1533	5.3	6
44	Treatment for genetic diseases. *Current Opinion in Pediatrics*, **2008**, 20, 625-7	3.2	1
43	The use by Alabama pediatricians of genetics consultation in the evaluation of developmental delay. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 421-5	2.5	8
42	Genetic testing in cardiovascular disease. *Journal of the American College of Cardiology*, **2007**, 50, 727-37	15.1	51
41	A genetic model for cloacal exstrophy, the extreme cloacal malformation. *Journal of Pediatric Urology*, **2007**, 3, 214-7	1.5	4
40	Polymicrogyria and deletion 22q11.2 syndrome: window to the etiology of a common cortical malformation. *American Journal of Medical Genetics, Part A*, **2006**, 140, 2416-25	2.5	113
39	The multidisciplinary evaluation and management of cleft lip and palate. *Southern Medical Journal*, **2006**, 99, 1111-20	0.6	55
38	Defining the clinical spectrum of deletion 22q11.2. *Journal of Pediatrics*, **2005**, 147, 90-6	3.6	182
37	Clefting, amniotic bands, and polydactyly: a distinct phenotype that supports an intrinsic mechanism for amniotic band sequence. *American Journal of Medical Genetics, Part A*, **2005**, 137A, 298-301	2.5	37
36	Response to Kessler and Restaʦ commentary. *American Journal of Medical Genetics Part A*, **2004**, 126A, 439-439		
35	Cardiomyopathy in Coffin-Lowry syndrome. *American Journal of Medical Genetics Part A*, **2004**, 128A, 176-8		16
34	The primary care physicianʦ approach to congenital anomalies. *Primary Care - Clinics in Office Practice*, **2004**, 31, 605-19, x	2.2	7

33	Genetic drift. A smile. *American Journal of Medical Genetics Part A*, **2003**, 118A, 404-6		1
32	Genetic drift. A few moments. *American Journal of Medical Genetics Part A*, **2003**, 119A, 397-9		
31	Medical GeneticistsTduty to warn at-risk relatives for genetic disease. *American Journal of Medical Genetics Part A*, **2003**, 120A, 374-80		66
30	Duty to warn at-risk relatives for genetic disease: genetic counselorsTclinical experience. *American Journal of Medical Genetics Part A*, **2003**, 119C, 27-34		83
29	Disorganization in mice and humans. *American Journal of Medical Genetics Part A*, **2001**, 101, 334-338		29
28	Distal 5q deletion syndrome: phenotypic correlations. *American Journal of Medical Genetics Part A*, **2001**, 103, 63-8		20
27	Genetic testing and genetic counseling for deafness: the future is here. *Laryngoscope*, **2001**, 111, 715-8	3.6	6
26	What Information Do Parents of Newborns With Cleft Lip, Palate, or Both Want to Know?. *Cleft Palate-Craniofacial Journal*, **2001**, 38, 55-58	1.9	28
25	Adult with an interstitial deletion of chromosome 10 [del(10)(q25. 1q25.3)]: overlap with Coffin-Lowry syndrome. *American Journal of Medical Genetics Part A*, **2000**, 95, 93-8		21
24	Parental attitudes toward genetic testing for pediatric deafness. *American Journal of Human Genetics*, **2000**, 67, 1621-5	11	101
23	Child with mosaic variegated aneuploidy and embryonal rhabdomyosarcoma. *American Journal of Medical Genetics Part A*, **1999**, 82, 20-4		31
22	Clinical and molecular studies of brachydactyly type D. *American Journal of Medical Genetics Part A*, **1999**, 85, 413-8		1
21	Microcephaly-lymphedema-chorioretinal dysplasia: A unique genetic syndrome with variable expression and possible characteristic facial appearance **1999**, 86, 215-218		19
20	Favorable prognosis for children with Pfeiffer syndrome types 2 and 3: Implications for classification **1998**, 75, 240-244		26
19	Severe oculocerebrocutaneous (Delleman) syndrome: overlap with Goldenhar anomaly. *American Journal of Medical Genetics Part A*, **1998**, 78, 282-5		29
18	Nonpenetrance in FGFR3-associated coronal synostosis syndrome **1998**, 80, 296-297		20
17	Mutations in CDMP1 cause autosomal dominant brachydactyly type C. *Nature Genetics*, **1997**, 17, 18-9	36.3	235
16	Sibs with cleidocranial dysplasia born to normal parents: germ line mosaicism?. *American Journal of Medical Genetics Part A*, **1997**, 69, 348-51		6

15	Duplication 14(q24.3q31) in a father and daughter: delineation of a possible imprinted region. *American Journal of Medical Genetics Part A*, **1997**, 71, 361-5		11
14	Genetic drift. The awful truth. *American Journal of Medical Genetics Part A*, **1997**, 71, 375-7		2
13	Disorganization in mice and humans and its relation to sporadic birth defects. *American Journal of Medical Genetics Part A*, **1997**, 73, 425-36		32
12	Frontonasal malformation and cloacal exstrophy: a previously unreported association. *American Journal of Medical Genetics Part A*, **1996**, 61, 75-8		10
11	Craniosynostosis, Philadelphia type: a new autosomal dominant syndrome with sagittal craniosynostosis and syndactyly of the fingers and toes. *American Journal of Medical Genetics Part A* , **1996**, 62, 184-91		19
10	Opitz G/BBB syndrome: clinical comparisons of families linked to Xp22 and 22q, and a review of the literature. *American Journal of Medical Genetics Part A*, **1996**, 62, 305-17		66
9	The good that we do. *American Journal of Medical Genetics Part A*, **1996**, 65, 257-8		
8	Syntelencephaly in an infant of a diabetic mother. *American Journal of Medical Genetics Part A*, **1996** , 66, 433-7		19
7	Congenital Muscular Torticollis. *Pediatrics in Review*, **1996**, 17, 374-375	1.1	1
6	Classical Noonan syndrome is not associated with deletions of 22q11. *American Journal of Medical Genetics Part A*, **1995**, 56, 94-6		14
5	Non-immune hydrops fetalis associated with impaired fetal movement: a case report and review. *American Journal of Medical Genetics Part A*, **1994**, 53, 251-4		8
4	Unusual craniofacial dysmorphia due to prenatal alcohol and cocaine exposure. *Teratology*, **1994**, 50, 160-4		24
3	De novo interstitial deletion of the long arm of chromosome 3: 46,XX,del(3)(q25.1q26.1). *Clinical Genetics*, **1993**, 44, 335-7	4	8
2	New finding of Schinzel-Giedion syndrome: a case with a malignant sacrococcygeal teratoma. *American Journal of Medical Genetics Part A*, **1993**, 47, 852-6		31
1	Attitudes of deaf individuals towards genetic testing of genes known to cause hearing loss. *Clinical Ethics*,147775092110635	1	0