Nathaniel H Robin

List of Publications by Year
in descending order

95 papers	2,441 citations	279798 23 h-index	243625 44 g-index
98 all docs	98 docs citations	98 times ranked	3163 citing authors

#	Article	IF	Citations
1	Mutations in CDMP1 cause autosomal dominant brachydactyly type C. Nature Genetics, 1997, 17, 18-19.	21.4	255
2	Defining the Clinical Spectrum of Deletion 22q11.2. Journal of Pediatrics, 2005, 147, 90-96.	1.8	201
3	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, 155, 2386-2396.	1.2	159
4	Parental Attitudes toward Genetic Testing for Pediatric Deafness. American Journal of Human Genetics, 2000, 67, 1621-1625.	6.2	126
5	Polymicrogyria and deletion 22q11.2 syndrome: Window to the etiology of a common cortical malformation. American Journal of Medical Genetics, Part A, 2006, 140A, 2416-2425.	1.2	125
6	Duty to warn atâ€risk relatives for genetic disease: Genetic counselors' clinical experience. American Journal of Medical Genetics Part A, 2003, 119C, 27-34.	2.4	105
7	Medical geneticists' duty to warn at-risk relatives for genetic disease. American Journal of Medical Genetics Part A, 2003, 120A, 374-380.	2.4	85
8	Opitz G/BBB syndrome: Clinical comparisons of families linked to Xp22 and 22Q and a review of the literature. , 1996, 62, 305-317.		81
9	The Multidisciplinary Evaluation and Management of Cleft Lip and Palate. Southern Medical Journal, 2006, 99, 1111-1120.	0.7	74
10	Amniotic Constriction Band: A Multidisciplinary Assessment of Etiology and Clinical Presentation. Journal of Bone and Joint Surgery - Series A, 2009, 91, 68-75.	3.0	66
11	Genetic Testing in Cardiovascular Disease. Journal of the American College of Cardiology, 2007, 50, 727-737.	2.8	59
12	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.	2.4	49
13	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.	6.2	48
14	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.	1.2	46
15	De Novo Truncating Variants in SON Cause Intellectual Disability, Congenital Malformations, and Failure to Thrive. American Journal of Human Genetics, 2016, 99, 720-727.	6.2	45
16	Child with mosaic variegated aneuploidy and embryonal rhabdomyosarcoma. , 1999, 82, 20-24.		40
17	What Information Do Parents of Newborns With Cleft Lip, Palate, or Both Want to Know?. Cleft Palate-Craniofacial Journal, 2001, 38, 55-58.	0.9	39
18	Stickler Syndrome: A Review of Clinical Manifestations and the Genetics Evaluation. Journal of Personalized Medicine, 2020, 10, 105.	2.5	37

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19	New finding of Schinzel-Giedion syndrome: A case with a malignant sacrococcygeal teratoma. American Journal of Medical Genetics Part A, 1993, 47, 852-856.	2.4	35
20	Disorganization in mice and humans. American Journal of Medical Genetics Part A, 2001, 101, 334-338.	2.4	34
21	Disorganization in mice and humans and its relation to sporadic birth defects. , 1997, 73, 425-436.		33
22	Severe oculocerebrocutaneous (Delleman) syndrome: Overlap with Goldenhar anomaly. , 1998, 78, 282-285.		32
23	Favorable prognosis for children with Pfeiffer syndrome types 2 and 3: Implications for classification. , 1998, 75, 240-244.		31
24	Trisomy 18: A singleâ€center evaluation of management trends and experience with aggressive obstetric or neonatal intervention. American Journal of Medical Genetics, Part A, 2016, 170, 838-846.	1.2	27
25	Unusual craniofacial dysmorphia due to prenatal alcohol and cocaine exposure. Teratology, 1994, 50, 160-164.	1.6	25
26	Syntelencephaly in an infant of a diabetic mother. American Journal of Medical Genetics Part A, 1996, 66, 433-437.	2.4	24
27	Nonpenetrance in FGFR3-associated coronal synostosis syndrome. , 1998, 80, 296-297.		24
28	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, 93-98.	2.4	24
29	Clinical relevance of small copy-number variants in chromosomal microarray clinical testing. Genetics in Medicine, 2017, 19, 377-385.	2.4	24
30	Distal 5q deletion syndrome: Phenotypic correlations. American Journal of Medical Genetics Part A, 2001, 103, 63-68.	2.4	23
31	Microcephaly-lymphedema-chorioretinal dysplasia: A unique genetic syndrome with variable expression and possible characteristic facial appearance. , 1999, 86, 215-218.		22
32	A previously unrecognized 22q13.2 microdeletion syndrome that encompasses TCF20 and TNFRSF13C. American Journal of Medical Genetics, Part A, 2018, 176, 2791-2797.	1.2	22
33	Craniosynostosis, Philadelphia type: A new autosomal dominant syndrome with sagittal craniosynostosis and syndactyly of the fingers and toes. , 1996, 62, 184-191.		20
34	Cardiomyopathy in Coffin-Lowry syndrome. American Journal of Medical Genetics Part A, 2004, 128A, 176-178.	2.4	19
35	It does matter: the importance of making the diagnosis of a genetic syndrome. Current Opinion in Pediatrics, 2006, 18, 595-597.	2.0	19
36	Clinical evaluation and etiologic diagnosis of hearing loss: A clinical practice resource of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2022, 24, 1392-1406.	2.4	18

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37	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.	1.2	16
38	Classical Noonan syndrome is not associated with deletions of 22q11. American Journal of Medical Genetics Part A, 1995, 56, 94-96.	2.4	15
39	Duplication 14(q24.3q31) in a father and daughter: Delineation of a possible imprinted region. , 1997, 71, 361-365.		15
40	Genetic Testing and Genetic Counseling for Deafness: The Future Is Here. Laryngoscope, 2001, 111, 715-718.	2.0	15
41	IRF6mutations in mixed isolated familial clefting. American Journal of Medical Genetics, Part A, 2010, 152A, 3107-3109.	1.2	15
42	Trisomy 18: A survey of opinions, attitudes, and practices of neonatologists. American Journal of Medical Genetics, Part A, 2016, 170, 2638-2643.	1.2	14
43	JARID2 haploinsufficiency is associated with a clinically distinct neurodevelopmental syndrome. Genetics in Medicine, 2021, 23, 374-383.	2.4	13
44	De novo coding variants in the AGO1 gene cause a neurodevelopmental disorder with intellectual disability. Journal of Medical Genetics, 2022, 59, 965-975.	3.2	13
45	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, 115-123.	1.2	12
46	Newborn Craniofacial Malformations. Clinics in Perinatology, 2015, 42, 321-336.	2.1	12
47	Frontonasal malformation and cloacal exstrophy: A previously unreported association. , 1996, 61, 75-78.		11
48	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, 535-538.	1.2	10
49	Non-immune hydrops fetalis associated with impaired fetal movement: A case report and review. American Journal of Medical Genetics Part A, 1994, 53, 251-254.	2.4	9
50	Sibs with cleidocranial dysplasia born to normal parents: Germ line mosaicism?. , 1997, 69, 348-351.		9
51	The primary care physician's approach to congenital anomalies. Primary Care - Clinics in Office Practice, 2004, 31, 605-619.	1.6	9
52	Use of Array Comparative Genome Hybridization in Orofacial Clefting. Journal of Craniofacial Surgery, 2010, 21, 1591-1594.	0.7	9
53	Imperforate anus is a rare associated finding in blepharocheilodontic syndrome. American Journal of Medical Genetics, Part A, 2010, 152A, 438-440.	1.2	9
54	Incidental Detection of Cancer Predisposition Gene Copy Number Variations by Array Comparative Genomic Hybridization. Journal of Pediatrics, 2014, 165, 1057-1059.e4.	1.8	9

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55	The use by Alabama pediatricians of genetics consultation in the evaluation of developmental delay. American Journal of Medical Genetics, Part A, 2008, 146A, 421-425.	1.2	8
56	De novo interstitial deletion of the long arm of chromosome 3:46, XX, del(3) (q25.1q26.1). Clinical Genetics, 1993, 44, 335-337.	2.0	8
57	The awful truth. , 1997, 71, 375-377.		7
58	Classic phenotype of Coffinâ€“lowry syndrome in a female with stimulusâ€induced drop episodes and a genotype with preserved Nâ€terminal kinase domain. American Journal of Medical Genetics, Part A, 2014, 164, 516-521.	1.2	7
59	Array comparative genomic hybridisation testing in CHD. Cardiology in the Young, 2015, 25, 1155-1172.	0.8	7
60	IRF6 Sequencing in Interrupted Clefting. Cleft Palate-Craniofacial Journal, 2016, 53, 373-376.	0.9	7
61	The duty to warn atâ€risk relativesâ€"The experience of genetic counselors and medical geneticists. American Journal of Medical Genetics, Part A, 2020, 182, 314-321.	1.2	7
62	Dysmorphology in the Era of Genomic Diagnosis. Journal of Personalized Medicine, 2020, 10, 18.	2.5	7
63	The natural history of OTOF-related auditory neuropathy spectrum disorders: a multicenter study. Human Genetics, 2022, 141, 853-863.	3.8	7
64	Genetic and Reproductive Knowledge Among Adolescents and Adults With Cystic Fibrosis. Chest, 2008, 133, 1533.	0.8	6
65	Genetic testing hearing loss: The challenge of non syndromic mimics. International Journal of Pediatric Otorhinolaryngology, 2021, 150, 110872.	1.0	6
66	Craniosynostosis and radial ray defect: A rare presentation of 22q11.2 deletion syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 2024-2026.	1.2	5
67	A genetic model for cloacal exstrophy, the extreme cloacal malformation. Journal of Pediatric Urology, 2007, 3, 214-217.	1.1	4
68	Editorial. Current Opinion in Pediatrics, 2018, 30, 699-700.	2.0	4
69	Further delineation of the Kapur-Toriello syndrome. American Journal of Medical Genetics, Part A, 2010, 152A, 1013-1015.	1.2	3
70	The development and implementation of an in-service exam for medical genetics residency programs. Genetics in Medicine, 2012, 14, 552-557.	2.4	3
71	Simultaneous osteosarcoma and renal cell carcinoma with BRCA1 mutation in a young male adult with prior oligodendroglioma. Pediatric Blood and Cancer, 2020, 67, e28116.	1.5	3
72	Editorial: Medical genetics, expert medical testimony, and suspected child abuse cases: a call for evidence-based standards in clinic and the courtroom. Current Opinion in Pediatrics, 2021, 33, 1-2.	2.0	3

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73	Genetic consultations in cases of unexplained fractures and haemorrhage: an evidence-based approach. Current Opinion in Pediatrics, 2021, 33, 3-18.	2.0	3
74	Congenital Muscular Torticollis. Pediatrics in Review, 1996, 17, 374-375.	0.4	3
75	Near complete deletion of <scp>KMT2D</scp> in a college student. American Journal of Medical Genetics, Part A, 2022, 188, 1550-1555.	1.2	3
76	Clinical and molecular studies of brachydactyly type D. , 1999, 85, 413-418.		2
77	Congenital Heart Defects. , 2013, , 1-51.		2
78	Cleft palate in a patient with the nested 22q11.2 LCR C to D deletion. American Journal of Medical Genetics, Part A, 2016, 170, 260-262.	1.2	2
79	Foramen magnum compression in Coffinâ€"Lowry syndrome: A case report. American Journal of Medical Genetics, Part A, 2017, 173, 1087-1089.	1.2	2
80	Congenital Heart Defects. , 2020, , 3-75.		2
81	The good that we do. , 1996, 65, 257-258.		1
82	A smile. American Journal of Medical Genetics Part A, 2003, 118A, 404-406.	2.4	1
83	Treatment for genetic diseases. Current Opinion in Pediatrics, 2008, 20, 625-627.	2.0	1
84	Educating the Adolescent and Young Adult With Cystic Fibrosis About Their Reproductive Risks and Options. Chest, 2013, 143, 580-581.	0.8	1
85	Growth parameters: the cheap and easy genetic test. Current Opinion in Pediatrics, 2016, 28, 679-681.	2.0	1
86	Ethical and Legal Issues. , 2018, , 93-100.		1
87	Inherited cause of in utero digital malformations. BMJ Case Reports, 2020, 13, e232020.	0.5	1
88	The relationship between performance on the medical genetics and genomics in-training and certifying examinations. Genetics in Medicine, 2022, 24, 225-231.	2.4	1
89	Attitudes of deaf individuals towards genetic testing of genes known to cause hearing loss. Clinical Ethics, 2023, 18, 230-235.	0.7	1
90	A few moments. American Journal of Medical Genetics Part A, 2003, 119A, 397-399.	2.4	0

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91	Response to Kessler and Resta's commentary. American Journal of Medical Genetics Part A, 2004, 126A, 439-439.	2.4	0
92	The mall test (or fun with a dysmorphologist). , 2011, 155, 2909-2909.		0
93	Editorial. Current Opinion in Pediatrics, 2015, 27, 657-658.	2.0	0
94	Choose your words carefully. American Journal of Medical Genetics, Part A, 2021, 185, 1953-1953.	1.2	0
95	The first postâ€natal clinical description of true mosaic complete tetrasomy 21: A case report. American Journal of Medical Genetics, Part A, 2021, 185, 3507-3509.	1.2	0