Hope Northrup

List of Publications by Year
in descending order

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59 papers	3,629 citations	331670 21 h-index	138484 58 g-index
64 all docs	64 docs citations	64 times ranked	4780 citing authors

#	Article	IF	Citations
1	Birth Defect Co-Occurrence Patterns Among Infants With Cleft Lip and/or Palate. Cleft Palate-Craniofacial Journal, 2022, 59, 417-426.	0.9	7
2	Birth defect co-occurrence patterns in the Texas Birth Defects Registry. Pediatric Research, 2022, 91, 1278-1285.	2.3	8
3	Discovering a new part of the phenotypic spectrum of Coffin-Siris syndrome in a fetal cohort. Genetics in Medicine, 2022, 24, 1753-1760.	2.4	6
4	Patterns of co-occurring birth defects among infants with hypospadias. Journal of Pediatric Urology, 2021, 17, 64.e1-64.e8.	1.1	4
5	Tuber Locations Associated with Infantile Spasms Map to a Common Brain Network. Annals of Neurology, 2021, 89, 726-739.	5.3	24
6	Patterns of congenital anomalies among individuals with trisomy 13 in Texas. American Journal of Medical Genetics, Part A, 2021, 185, 1787-1793.	1.2	2
7	Frequency, Progression, and Current Management: Report of 16 New Cases of Nonfunctional Pancreatic Neuroendocrine Tumors in Tuberous Sclerosis Complex and Comparison With Previous Reports. Frontiers in Neurology, 2021, 12, 627672.	2.4	7
8	The current state of prenatal detection of genetic conditions in congenital heart defects. Translational Pediatrics, 2021, 10, 2157-2170.	1.2	6
9	The youngest pair of siblings with Mucopolysaccharidosis type IVA to receive enzyme replacement therapy to date: A case report. American Journal of Medical Genetics, Part A, 2021, 185, 3510-3516.	1.2	5
10	Updated International Tuberous Sclerosis Complex Diagnostic Criteria and Surveillance and Management Recommendations. Pediatric Neurology, 2021, 123, 50-66.	2.1	230
11	Epilepsy Is Heterogeneous in Early-Life Tuberous Sclerosis Complex. Pediatric Neurology, 2021, 123, 1-9.	2.1	5
12	Profile of Autism Spectrum Disorder in Tuberous Sclerosis Complex: Results from a Longitudinal, Prospective, Multisite Study. Annals of Neurology, 2021, 90, 874-886.	5.3	13
13	A Comprehensive Assessment of Co-occurring Birth Defects among Infants with Non-Syndromic Anophthalmia or Microphthalmia. Ophthalmic Epidemiology, 2021, 28, 428-435.	1.7	4
14	Language predictors of autism spectrum disorder in young children with tuberous sclerosis complex. Epilepsy and Behavior, 2020, 103, 106844.	1.7	14
15	EEG Spectral Features in Sleep of Autism Spectrum Disorders in Children with Tuberous Sclerosis Complex. Journal of Autism and Developmental Disorders, 2020, 50, 916-923.	2.7	2
16	The Connectivity Fingerprint of the Fusiform Gyrus Captures the Risk of Developing Autism in Infants with Tuberous Sclerosis Complex. Cerebral Cortex, 2020, 30, 2199-2214.	2.9	11
17	Epilepsy Risk Prediction Model for Patients With Tuberous Sclerosis Complex. Pediatric Neurology, 2020, 113, 46-50.	2.1	9
18	TSC1 Variant Associated With Mild or Absent Clinical Features of Tuberous Sclerosis Complex in a Three-Generation Family. Pediatric Neurology, 2020, 110, 89-91.	2.1	3

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19	Snx3 is important for mammalian neural tube closure via its role in canonical and non-canonical WNT signaling. Development (Cambridge), 2020, 147, .	2.5	10
20	Birth defects that coâ€occur with nonâ€syndromic gastroschisis and omphalocele. American Journal of Medical Genetics, Part A, 2020, 182, 2581-2593.	1.2	9
21	Two different genetic etiologies for tuberous sclerosis complex (TSC) in a single family. Molecular Genetics & Genomic Medicine, 2020, 8, e1296.	1.2	3
22	Deep learning in rare disease. Detection of tubers in tuberous sclerosis complex. PLoS ONE, 2020, 15, e0232376.	2.5	23
23	Reproducibility of Structural and Diffusion Tensor Imaging in the TACERN Multi-Center Study. Frontiers in Integrative Neuroscience, 2019, 13, 24.	2.1	32
24	Increased electroencephalography connectivity precedes epileptic spasm onset in infants with tuberous sclerosis complex. Epilepsia, 2019, 60, 1721-1732.	5.1	37
25	Restingâ€State fMRI Networks in Children with Tuberous Sclerosis Complex. Journal of Neuroimaging, 2019, 29, 750-759.	2.0	6
26	Coâ€occurring defect analysis: A platform for analyzing birth defect coâ€occurrence in registries. Birth Defects Research, 2019, 111, 1356-1364.	1.5	12
27	Scalp EEG spikes predict impending epilepsy in TSC infants: A longitudinal observational study. Epilepsia, 2019, 60, 2428-2436.	5.1	45
28	Tuberous Sclerosis Complex Genotypes and Developmental Phenotype. Pediatric Neurology, 2019, 96, 58-63.	2.1	21
29	Early white matter development is abnormal in tuberous sclerosis complex patients who develop autism spectrum disorder. Journal of Neurodevelopmental Disorders, 2019, 11, 36.	3.1	32
30	Evidence- and consensus-based recommendations for the use of pegvaliase in adults with phenylketonuria. Genetics in Medicine, 2019, 21, 1851-1867.	2.4	56
31	The Impact of Psychiatric Symptoms on Tuberous Sclerosis Complex and Utilization of Mental Health Treatment. Pediatric Neurology, 2019, 91, 41-49.	2.1	7
32	Pegvaliase for the treatment of phenylketonuria: Results of a long-term phase 3 clinical trial program (PRISM). Molecular Genetics and Metabolism, 2018, 124, 27-38.	1.1	123
33	Visual and semi-automatic non-invasive detection of interictal fast ripples: A potential biomarker of epilepsy in children with tuberous sclerosis complex. Clinical Neurophysiology, 2018, 129, 1458-1466.	1.5	46
34	Treatment of Disfiguring Cutaneous Lesions in Neurofibromatosis-1 with Everolimus: A Phase II, Open-Label, Single-Arm Trial. Drugs in R and D, 2018, 18, 295-302.	2.2	17
35	Genetics, genomics, and genotypeâ€“phenotype correlations of TSC: Insights for clinical practice. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2018, 178, 281-290.	1.6	65
36	High vigabatrin dosage is associated with lower risk of infantile spasms relapse among children with tuberous sclerosis complex. Epilepsy Research, 2018, 148, 1-7.	1.6	25

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37	Tuberous sclerosis complex. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2018, 178, 274-277.	1.6	25
38	Efficacy and Safety of Topical Rapamycin in Patients With Facial Angiofibromas Secondary to Tuberous Sclerosis Complex. JAMA Dermatology, 2018, 154, 773.	4.1	71
39	The phenotypic spectrum of Schaaf-Yang syndrome: 18 new affected individuals from 14 families. Genetics in Medicine, 2017, 19, 45-52.	2.4	94
40	TSC2 c.1864C>T variant associated with mild cases of tuberous sclerosis complex. American Journal of Medical Genetics, Part A, 2017, 173, 771-775.	1.2	15
41	Wiedemann-Steiner syndrome: Novel pathogenic variant and review of literature. European Journal of Medical Genetics, 2017, 60, 285-288.	1.3	28
42	Mutations in folate transporter genes and risk for human myelomeningocele. American Journal of Medical Genetics, Part A, 2017, 173, 2973-2984.	1.2	17
43	Presentation and Diagnosis of Tuberous Sclerosis Complex in Infants. Pediatrics, 2017, 140, .	2.1	90
44	The genomic landscape of tuberous sclerosis complex. Nature Communications, 2017, 8, 15816.	12.8	154
45	Identification of novel candidate disease genes from de novo exonic copy number variants. Genome Medicine, 2017, 9, 83.	8.2	50
46	A Novel Dominant Mutation in SAG, the Arrestin-1 Gene, Is a Common Cause of Retinitis Pigmentosa in Hispanic Families in the Southwestern United States. , 2017, 58, 2774.		31
47	Catenin delta-1 (CTNND1) phosphorylation controls the mesenchymal to epithelial transition in astrocytic tumors. Human Molecular Genetics, 2016, 25, 4201-4210.	2.9	10
48	KIAA2022 nonsense mutation in a symptomatic female. American Journal of Medical Genetics, Part A, 2016, 170, 703-706.	1.2	20
49	Clinical Electroencephalographic Biomarker for Impending Epilepsy in Asymptomatic Tuberous Sclerosis Complex Infants. Pediatric Neurology, 2016, 54, 29-34.	2.1	93
50	Recurrent Muscle Weakness with Rhabdomyolysis, Metabolic Crises, and Cardiac Arrhythmia Due to Bi-allelic TANGO2 Mutations. American Journal of Human Genetics, 2016, 98, 347-357.	6.2	98
51	Efficacy of a medical genetics rotation during pediatric training. Genetics in Medicine, 2016, 18, 199-202.	2.4	16
52	Association of facilitated glucose transporter 2 gene variants with the myelomeningocele phenotype. Birth Defects Research Part A: Clinical and Molecular Teratology, 2015, 103, 479-487.	1.6	10
53	Identification of HIBCH Gene Mutations Causing Autosomal Recessive Leigh Syndrome: A Gene Involved in Valine Metabolism. Pediatric Neurology, 2015, 52, 361-365.	2.1	28
54	The mTOR inhibitor revolution rolls on. Lancet Oncology, The, 2014, 15, 1418-1419.	10.7	4

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55	Genetic variations in the GLUT3 gene associated with myelomeningocele. American Journal of Obstetrics and Gynecology, 2014, 211, 305.e1-305.e8.	1.3	7
56	Tuberous Sclerosis Complex Diagnostic Criteria Update: Recommendations of the 2012 International Tuberous Sclerosis Complex Consensus Conference. Pediatric Neurology, 2013, 49, 243-254.	2.1	1,185
57	Genotype/phenotype correlation in 325 individuals referred for a diagnosis of tuberous sclerosis complex in the United States. Genetics in Medicine, 2007, 9, 88-100.	2.4	353
58	Genetics of neurocutaneous disorders. , 2004, , 6-23.		0
59	Prevalence of mutations causing retinitis pigmentosa and other inherited retinopathies. Human Mutation, 2001, 17, 42-51.	2.5	292