# Hope Northrup

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/3780398/publications.pdf

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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 | 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 |
| 2 | 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 |
| 3 | Prevalence of mutations causing retinitis pigmentosa and other inherited retinopathies. Human Mutation, 2001, 17, 42-51. | 2.5 | 292 |
| 4 | Updated International Tuberous Sclerosis Complex Diagnostic Criteria and Surveillance and Management Recommendations. Pediatric Neurology, 2021, 123, 50-66. | 2.1 | 230 |
| 5 | The genomic landscape of tuberous sclerosis complex. Nature Communications, 2017, 8, 15816. | 12.8 | 154 |
| 6 | 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 |
| 7 | 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 |
| 8 | The phenotypic spectrum of Schaaf-Yang syndrome: 18 new affected individuals from 14 families. Genetics in Medicine, 2017, 19, 45-52. | 2.4 | 94 |
| 9 | Clinical Electroencephalographic Biomarker for Impending Epilepsy in Asymptomatic Tuberous Sclerosis Complex Infants. Pediatric Neurology, 2016, 54, 29-34. | 2.1 | 93 |
| 10 | Presentation and Diagnosis of Tuberous Sclerosis Complex in Infants. Pediatrics, 2017, 140, . | 2.1 | 90 |
| 11 | Efficacy and Safety of Topical Rapamycin in Patients With Facial Angiofibromas Secondary to Tuberous Sclerosis Complex. JAMA Dermatology, 2018, 154, 773. | 4.1 | 71 |
| 12 | 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 |
| 13 | Evidence- and consensus-based recommendations for the use of pegvaliase in adults with phenylketonuria. Genetics in Medicine, 2019, 21, 1851-1867. | 2.4 | 56 |
| 14 | Identification of novel candidate disease genes from de novo exonic copy number variants. Genome Medicine, 2017, 9, 83. | 8.2 | 50 |
| 15 | 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 |
| 16 | Scalp EEG spikes predict impending epilepsy in TSC infants: A longitudinal observational study. Epilepsia, 2019, 60, 2428-2436. | 5.1 | 45 |
| 17 | Increased electroencephalography connectivity precedes epileptic spasm onset in infants with tuberous sclerosis complex. Epilepsia, 2019, 60, 1721-1732. | 5.1 | 37 |
| 18 | Reproducibility of Structural and Diffusion Tensor Imaging in the TACERN Multi-Center Study. Frontiers in Integrative Neuroscience, 2019, 13, 24. | 2.1 | 32 |

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|---|---|---|---|
| 19 | 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 |
| 20 | 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 |
| 21 | 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 |
| 22 | Wiedemann-Steiner syndrome: Novel pathogenic variant and review of literature. European Journal of Medical Genetics, 2017, 60, 285-288. | 1.3 | 28 |
| 23 | 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 |
| 24 | Tuberous sclerosis complex. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2018, 178, 274-277. | 1.6 | 25 |
| 25 | Tuber Locations Associated with Infantile Spasms Map to a Common Brain Network. Annals of Neurology, 2021, 89, 726-739. | 5.3 | 24 |
| 26 | Deep learning in rare disease. Detection of tubers in tuberous sclerosis complex. PLoS ONE, 2020, 15, e0232376. | 2.5 | 23 |
| 27 | Tuberous Sclerosis Complex Genotypes and Developmental Phenotype. Pediatric Neurology, 2019, 96, 58-63. | 2.1 | 21 |
| 28 | KIAA2022 nonsense mutation in a symptomatic female. American Journal of Medical Genetics, Part A, 2016, 170, 703-706. | 1.2 | 20 |
| 29 | Mutations in folate transporter genes and risk for human myelomeningocele. American Journal of Medical Genetics, Part A, 2017, 173, 2973-2984. | 1.2 | 17 |
| 30 | 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 |
| 31 | Efficacy of a medical genetics rotation during pediatric training. Genetics in Medicine, 2016, 18, 199-202. | 2.4 | 16 |
| 32 | 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 |
| 33 | Language predictors of autism spectrum disorder in young children with tuberous sclerosis complex. Epilepsy and Behavior, 2020, 103, 106844. | 1.7 | 14 |
| 34 | 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 |
| 35 | Coâ€occurring defect analysis: A platform for analyzing birth defect coâ€occurrence in registries. Birth Defects Research, 2019, 111, 1356-1364. | 1.5 | 12 |
| 36 | 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 |

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|---|---|---|---|
| 37 | 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 |
| 38 | Catenin delta-1 (CTNND1) phosphorylation controls the mesenchymal to epithelial transition in astrocytic tumors. Human Molecular Genetics, 2016, 25, 4201-4210. | 2.9 | 10 |
| 39 | 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 |
| 40 | Epilepsy Risk Prediction Model for Patients With Tuberous Sclerosis Complex. Pediatric Neurology, 2020, 113, 46-50. | 2.1 | 9 |
| 41 | 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 |
| 42 | Birth defect co-occurrence patterns in the Texas Birth Defects Registry. Pediatric Research, 2022, 91, 1278-1285. | 2.3 | 8 |
| 43 | Genetic variations in the GLUT3 gene associated with myelomeningocele. American Journal of Obstetrics and Gynecology, 2014, 211, 305.e1-305.e8. | 1.3 | 7 |
| 44 | The Impact of Psychiatric Symptoms on Tuberous Sclerosis Complex and Utilization of Mental Health Treatment. Pediatric Neurology, 2019, 91, 41-49. | 2.1 | 7 |
| 45 | Birth Defect Co-Occurrence Patterns Among Infants With Cleft Lip and/or Palate. Cleft Palate-Craniofacial Journal, 2022, 59, 417-426. | 0.9 | 7 |
| 46 | 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 |
| 47 | Restingâ€State fMRI Networks in Children with Tuberous Sclerosis Complex. Journal of Neuroimaging, 2019, 29, 750-759. | 2.0 | 6 |
| 48 | The current state of prenatal detection of genetic conditions in congenital heart defects. Translational Pediatrics, 2021, 10, 2157-2170. | 1.2 | 6 |
| 49 | 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 |
| 50 | 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 |
| 51 | Epilepsy Is Heterogeneous in Early-Life Tuberous Sclerosis Complex. Pediatric Neurology, 2021, 123, 1-9. | 2.1 | 5 |
| 52 | The mTOR inhibitor revolution rolls on. Lancet Oncology, The, 2014, 15, 1418-1419. | 10.7 | 4 |
| 53 | Patterns of co-occurring birth defects among infants with hypospadias. Journal of Pediatric Urology, 2021, 17, 64.e1-64.e8. | 1.1 | 4 |
| 54 | 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 |

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|---|---|---|---|
| 55 | 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 |
| 56 | Two different genetic etiologies for tuberous sclerosis complex (TSC) in a single family. Molecular Genetics & Genomic Medicine, 2020, 8, e1296. | 1.2 | 3 |
| 57 | 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 |
| 58 | 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 |
| 59 | Genetics of neurocutaneous disorders. , 2004, , 6-23. | | 0 |