Audrey Thurm

List of Publications by Year
in descending order

97 papers	6,320 citations	87888 38 h-index	82547 72 g-index
105 all docs	105 docs citations	105 times ranked	8971 citing authors

#	Article	IF	Citations
1	Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. Cell, 2020, 180, 568-584.e23.	28.9	1,422
2	Patterns of growth in verbal abilities among children with autism spectrum disorder.. Journal of Consulting and Clinical Psychology, 2007, 75, 594-604.	2.0	389
3	Intrathecal 2-hydroxypropyl-Î²-cyclodextrin decreases neurological disease progression in Niemann-Pick disease, type C1: a non-randomised, open-label, phase 1â€“2 trial. Lancet, The, 2017, 390, 1758-1768.	13.7	275
4	Studying the Emergence of Autism Spectrum Disorders in High-risk Infants: Methodological and Practical Issues. Journal of Autism and Developmental Disorders, 2007, 37, 466-480.	2.7	238
5	Predictors of Language Acquisition in Preschool Children with Autism Spectrum Disorders. Journal of Autism and Developmental Disorders, 2007, 37, 1721-1734.	2.7	210
6	Changes in access to educational and healthcare services for individuals with intellectual and developmental disabilities during COVIDâ€19 restrictions. Journal of Intellectual Disability Research, 2020, 64, 825-833.	2.0	190
7	Autism screening and diagnosis in low resource settings: Challenges and opportunities to enhance research and services worldwide. Autism Research, 2015, 8, 473-476.	3.8	189
8	Prospective Longitudinal Studies of Infant Siblings ofÂChildren With Autism: Lessons Learned and FutureÂDirections. Journal of the American Academy of Child and Adolescent Psychiatry, 2016, 55, 179-187.	0.5	174
9	Large-scale analyses of the relationship between sex, age and intelligence quotient heterogeneity and cortical morphometry in autism spectrum disorder. Molecular Psychiatry, 2020, 25, 614-628.	7.9	141
10	State of the Field: Differentiating Intellectual Disability From Autism Spectrum Disorder. Frontiers in Psychiatry, 2019, 10, 526.	2.6	135
11	Serum and cerebrospinal fluid immune mediators in children with autistic disorder: a longitudinal study. Molecular Autism, 2017, 8, 1.	4.9	127
12	Prospective phenotyping of NGLY1-CDDG, the first congenital disorder of deglycosylation. Genetics in Medicine, 2017, 19, 160-168.	2.4	124
13	Practice guideline: Treatment for insomnia and disrupted sleep behavior in children and adolescents with autism spectrum disorder. Neurology, 2020, 94, 392-404.	1.1	119
14	Compared to What? Early Brain Overgrowth in Autism and the Perils of Population Norms. Biological Psychiatry, 2013, 74, 563-575.	1.3	107
15	Social (pragmatic) communication disorder: a research review of this new DSM-5 diagnostic category. Journal of Neurodevelopmental Disorders, 2014, 6, 41.	3.1	96
16	The <scp>ADOS</scp> Calibrated Severity Score: Relationship to Phenotypic Variables and Stability over Time. Autism Research, 2012, 5, 267-276.	3.8	94
17	A framework for the investigation of rare genetic disorders in neuropsychiatry. Nature Medicine, 2019, 25, 1477-1487.	30.7	90
18	Pharmacotherapy for the Core Symptoms in Autistic Disorder: Current Status of the Research. Drugs, 2013, 73, 303-314.	10.9	87

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19	Diffusion Tensor Imaging in Young Children with Autism: Biological Effects and Potential Confounds. Biological Psychiatry, 2012, 72, 1043-1051.	1.3	82
20	Classifying and characterizing the development of adaptive behavior in a naturalistic longitudinal study of young children with autism. Journal of Neurodevelopmental Disorders, 2018, 10, 1.	3.1	79
21	Multisite Study of New Autism Diagnostic Interview-Revised (ADI-R) Algorithms for Toddlers and Young Preschoolers. Journal of Autism and Developmental Disorders, 2013, 43, 1527-1538.	2.7	75
22	Sex differences in scores on standardized measures of autism symptoms: a multisite integrative data analysis. Journal of Child Psychology and Psychiatry and Allied Disciplines, 2021, 62, 97-106.	5.2	74
23	Identification of Developmental and Behavioral Markers Associated With Genetic Abnormalities in Autism Spectrum Disorder. American Journal of Psychiatry, 2017, 174, 576-585.	7.2	73
24	Longitudinal study of symptom severity and language in minimally verbal children with autism. Journal of Child Psychology and Psychiatry and Allied Disciplines, 2015, 56, 97-104.	5.2	71
25	Measurement of Nonverbal IQ in Autism Spectrum Disorder: Scores in Young Adulthood Compared to Early Childhood. Journal of Autism and Developmental Disorders, 2015, 45, 966-974.	2.7	67
26	Association of brain-derived neurotrophic factor (BDNF) haploinsufficiency with lower adaptive behaviour and reduced cognitive functioning in WAGR/11p13 deletion syndrome. Cortex, 2013, 49, 2700-2710.	2.4	61
27	Idiopathic Autism: Cellular and Molecular Phenotypes in Pluripotent Stem Cell-Derived Neurons. Molecular Neurobiology, 2017, 54, 4507-4523.	4.0	57
28	An Open Label Trial of Donepezil for Enhancement of Rapid Eye Movement Sleep in Young Children with Autism Spectrum Disorders. Journal of Child and Adolescent Psychopharmacology, 2011, 21, 353-357.	1.3	56
29	Framework for assessing individuals with rare genetic disorders associated with profound intellectual and multiple disabilities (PIMD): the example of Phelan McDermid Syndrome. Clinical Neuropsychologist, 2018, 32, 1226-1255.	2.3	55
30	Talking About Death or Suicide: Prevalence and Clinical Correlates in Youth with Autism Spectrum Disorder in the Psychiatric Inpatient Setting. Journal of Autism and Developmental Disorders, 2018, 48, 3702-3710.	2.7	55
31	Concurrent validity of the differential ability scales, second edition with the Mullen Scales of Early Learning in young children with and without neurodevelopmental disorders. Child Neuropsychology, 2016, 22, 556-569.	1.3	54
32	Identification of 22q13 genes most likely to contribute to Phelan McDermid syndrome. European Journal of Human Genetics, 2018, 26, 293-302.	2.8	54
33	Convergent and divergent validity of the Mullen Scales of Early Learning in young children with and without autism spectrum disorder.. Psychological Assessment, 2015, 27, 1364-1378.	1.5	53
34	A pilot open-label trial of minocycline in patients with autism and regressive features. Journal of Neurodevelopmental Disorders, 2013, 5, 9.	3.1	51
35	Psychiatric illness and regression in individuals with Phelan-McDermid syndrome. Journal of Neurodevelopmental Disorders, 2020, 12, 7.	3.1	51
36	Repetitive Behavior and Restricted Interests in Young Children with Autism: Comparisons with Controls and Stability Over 2 Years. Autism Research, 2013, 6, 584-595.	3.8	50

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37	Brief Report: Symptom Onset Patterns and Functional Outcomes in Young Children with Autism Spectrum Disorders. Journal of Autism and Developmental Disorders, 2011, 41, 1727-1732.	2.7	46
38	Development, behavior, and biomarker characterization of Smith-Lemli-Opitz syndrome: an update. Journal of Neurodevelopmental Disorders, 2016, 8, 12.	3.1	45
39	Determination of the allelic frequency in Smithâ€“Lemliâ€“Opitz syndrome by analysis of massively parallel sequencing data sets. Clinical Genetics, 2015, 87, 570-575.	2.0	43
40	Cortical thickness change in autism during early childhood. Human Brain Mapping, 2016, 37, 2616-2629.	3.6	41
41	Effects of Oxytocin and Vasopressin on Preferential Brain Responses to Negative Social Feedback. Neuropsychopharmacology, 2017, 42, 1409-1419.	5.4	40
42	Cerebrospinal fluid vasopressin and symptom severity in children with autism. Annals of Neurology, 2018, 84, 611-615.	5.3	40
43	Concordance of the Vineland Adaptive Behavior Scales, second and third editions. Journal of Intellectual Disability Research, 2020, 64, 18-26.	2.0	37
44	State-Dependent Differences in Functional Connectivity in Young Children With Autism Spectrum Disorder. EBioMedicine, 2015, 2, 1905-1915.	6.1	33
45	Autism Spectrum Disorder, Intellectual Disability, and Delayed Walking. Pediatrics, 2016, 137, e20152959.	2.1	32
46	Strong evidence for genotypeâ€“phenotype correlations in Phelan-McDermid syndrome: results from the developmental synaptopathies consortium. Human Molecular Genetics, 2022, 31, 625-637.	2.9	32
47	Patterns of skill attainment and loss in young children with autism. Development and Psychopathology, 2014, 26, 203-214.	2.3	31
48	Evaluation of Periodic Limb Movements in Sleep and Iron Status in Children With Autism. Pediatric Neurology, 2015, 53, 343-349.	2.1	28
49	Long-Term Neuropsychological Outcomes from an Open-Label Phase I/IIa Trial of 2-Hydroxypropyl-Î²-Cyclodextrins (VTS-270) in Niemann-Pick Disease, Type C1. CNS Drugs, 2019, 33, 677-683.	5.9	28
50	CSF concentrations of 5-methyltetrahydrofolate in a cohort of young children with autism. Neurology, 2016, 86, 2258-2263.	1.1	25
51	Modelling gesture use and early language development in autism spectrum disorder. International Journal of Language and Communication Disorders, 2017, 52, 637-651.	1.5	25
52	Loss of skills and onset patterns in neurodevelopmental disorders: Understanding the neurobiological mechanisms. Autism Research, 2018, 11, 212-222.	3.8	25
53	Testing autism interventions: trials and tribulations. Lancet, The, 2010, 375, 2124-2125.	13.7	24
54	Prefrontal Activation During Executive Tasks Emerges Over Early Childhood: Evidence From Functional Near Infrared Spectroscopy. Developmental Neuropsychology, 2017, 42, 253-264.	1.4	23

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55	MED23â€associated intellectual disability in a nonâ€consanguineous family. American Journal of Medical Genetics, Part A, 2015, 167, 1374-1380.	1.2	21
56	Outcome Measures for Core Symptoms of Intellectual Disability: State of the Field. American Journal on Intellectual and Developmental Disabilities, 2020, 125, 418-433.	1.6	21
57	The Need for a Developmentally Based Measure of Social Communication Skills. Journal of the American Academy of Child and Adolescent Psychiatry, 2019, 58, 555-560.	0.5	20
58	Volumetric Analysis of the Basal Ganglia and Cerebellar Structures in Patients with Phelan-McDermid Syndrome. Pediatric Neurology, 2019, 90, 37-43.	2.1	19
59	First Night Effect Analysis in a Cohort of Young Children with Autism Spectrum Disorder. Journal of Clinical Sleep Medicine, 2013, 09, 67-70.	2.6	15
60	In-depth investigations of adolescents and adults with holoprosencephaly identify unique characteristics. Genetics in Medicine, 2018, 20, 14-23.	2.4	15
61	The Early Screening for Autism and Communication Disorders: Field-testing an autism-specific screening tool for children 12 to 36 months of age. Autism, 2021, 25, 2112-2123.	4.1	15
62	Psychometric Study of the Social Responsiveness Scale in Phelanâ€“McDermid Syndrome. Autism Research, 2020, 13, 1383-1396.	3.8	14
63	Cohort study of neurocognitive functioning and adaptive behaviour in children and adolescents with Niemannâ€Pick Disease type C1. Developmental Medicine and Child Neurology, 2016, 58, 262-269.	2.1	13
64	SOCIOEMOTIONAL AND BEHAVIORAL PROBLEMS IN TODDLERS WITH LANGUAGE DELAY. Infant Mental Health Journal, 2018, 39, 569-580.	1.8	13
65	The gestureâ€“language association over time in toddlers with and without language delays. Autism and Developmental Language Impairments, 2019, 4, 239694151984554.	1.6	13
66	Severity modeling of propionic acidemia using clinical and laboratory biomarkers. Genetics in Medicine, 2021, 23, 1534-1542.	2.4	13
67	Patterns of delay in early gross motor and expressive language milestone attainment in probands with genetic conditions versus idiopathic ASD from SFARI registries. Journal of Child Psychology and Psychiatry and Allied Disciplines, 2021, 62, 1297-1307.	5.2	13
68	TUBB3 Arg262His causes a recognizable syndrome including CFEOM3, facial palsy, joint contractures, and early-onset peripheral neuropathy. Human Genetics, 2021, 140, 1709-1731.	3.8	13
69	Prospective longitudinal overnight video-EEG evaluation in Phelanâ€“McDermid Syndrome. Epilepsy and Behavior, 2018, 80, 312-320.	1.7	12
70	Creatine Transporter Deficiency. Journal of Developmental and Behavioral Pediatrics, 2016, 37, 322-326.	1.1	11
71	Describing Function in ASD: Using the DSM-5 and Other Methods to Improve Precision. Journal of Autism and Developmental Disorders, 2017, 47, 2938-2941.	2.7	11
72	Early Indicators of Creatine Transporter Deficiency. Journal of Pediatrics, 2019, 206, 283-285.	1.8	10

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73	1-13C-propionate breath testing as a surrogate endpoint to assess efficacy of liver-directed therapies in methylmalonic acidemia (MMA). Genetics in Medicine, 2021, 23, 1522-1533.	2.4	10
74	Making Research Possible: Barriers and Solutions For Those With ASD and ID. Journal of Autism and Developmental Disorders, 2022, 52, 4646-4650.	2.7	10
75	Diffusion Tensor Imaging Abnormalities in the Uncinate Fasciculus and Inferior Longitudinal Fasciculus in Phelan-McDermid Syndrome. Pediatric Neurology, 2020, 106, 24-31.	2.1	9
76	Systematic Review: Recommendations for Rehabilitation in ASD and ID From Clinical Practice Guidelines. Archives of Rehabilitation Research and Clinical Translation, 2021, 3, 100140.	0.9	9
77	Insufficient evidence for inclusion of motor deficits in the <scp>ASD</scp> diagnostic criteria: A response to Bhat (2021). Autism Research, 2022, 15, 1374-1375.	3.8	9
78	Trajectories of cognitive development in toddlers with language delays. Research in Developmental Disabilities, 2018, 81, 65-72.	2.2	8
79	X-linked creatine transporter deficiency results in prolonged QTc and increased sudden death risk in humans and disease model. Genetics in Medicine, 2021, 23, 1864-1872.	2.4	8
80	Lack of Serum Antibodies against Borrelia burgdorferi in Children with Autism. Vaccine Journal, 2013, 20, 1092-1093.	3.1	6
81	No evidence of antibodies against GAD65 and other specific antigens in children with autism. BBA Clinical, 2015, 4, 81-84.	4.1	6
82	Incontinence in Phelanâ€McDermid Syndrome. Journal of Pediatric Gastroenterology and Nutrition, 2019, 69, e39-e42.	1.8	6
83	Neurodevelopmental Characterization of Young Children Diagnosed with Niemann-Pick Disease, Type C1. Journal of Developmental and Behavioral Pediatrics, 2020, 41, 388-396.	1.1	6
84	Age of walking and intellectual ability in autism spectrum disorder and other neurodevelopmental disorders: a populationâ€based study. Journal of Child Psychology and Psychiatry and Allied Disciplines, 2021, 62, 1070-1078.	5.2	6
85	Parent-reported measure of repetitive behavior in Phelan-McDermid syndrome. Journal of Neurodevelopmental Disorders, 2021, 13, 53.	3.1	6
86	Functional nearâ€infrared spectroscopy in toddlers: Neural differentiation of communicative cues and relation to future language abilities. Developmental Science, 2020, 23, e12948.	2.4	5
87	Seizure phenotype in CLN3 disease and its relation to other neurologic outcome measures. Journal of Inherited Metabolic Disease, 2021, 44, 1013-1020.	3.6	5
88	Depressive Symptoms in Young, Urban Schoolchildren: Environmental, Social, and Cognitive Risk. Journal of Prevention and Intervention in the Community, 2014, 42, 169-182.	0.7	3
89	Greater cortical thickness in individuals with ASD. Molecular Psychiatry, 2020, 25, 507-508.	7.9	3
90	Prerequisite skills in cognitive testing: Innovations in theory and recommendations for practice. Cognitive Development, 2021, 58, 101038.	1.3	3

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91	Developmental Issues and Milestones. , 2011, , 159-173.		3
92	Inclusion of individuals with low IQ in drug development for autism spectrum disorder. European Neuropsychopharmacology, 2021, 48, 37-39.	0.7	2
93	A novel measure of matching categories for early development: Item creation and pilot feasibility study. Research in Developmental Disabilities, 2021, 115, 103993.	2.2	2
94	Dr. Bishop et al. Reply. Journal of the American Academy of Child and Adolescent Psychiatry, 2020, 59, 1200-1202.	0.5	1
95	A Digestible Summary of Research Advances in the Neurochemistry of Autism - The Neurochemical Basis of Autism: From Molecules to Minicolumns.Gene J. Blatt (Ed.). (2009). New York: Springer, 267 pp., $179.00 (HB).. Journal of the International Neuropsychological Society, 2011, 17, 379-380.	1.8	0
96	eP235: Interim results of the Vigilan observational study: clinical characteristics of creatine transporter deficiency. Genetics in Medicine, 2022, 24, S149.	2.4	0
97	eP259: A phase 1/2 trial of AXO-AAV-GM1 gene therapy for the treatment of infantile- and juvenile-onset GM1 gangliosidosis. Genetics in Medicine, 2022, 24, S164.	2.4	0