

Audrey Thurm

List of Publications by Year in descending order

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Version: 2024-02-01

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	Strong evidence for genotypeâ€“phenotype correlations in Phelan-McDermid syndrome: results from the developmental synaptopathies consortium. <i>Human Molecular Genetics</i> , 2022, 31, 625-637.	2.9	32
2	Making Research Possible: Barriers and Solutions For Those With ASD and ID. <i>Journal of Autism and Developmental Disorders</i> , 2022, 52, 4646-4650.	2.7	10
3	eP235: Interim results of the Vigilant observational study: clinical characteristics of creatine transporter deficiency. <i>Genetics in Medicine</i> , 2022, 24, S149.	2.4	0
4	eP259: A phase 1/2 trial of AXO-AAV-GM1 gene therapy for the treatment of infantile- and juvenile-onset GM1 gangliosidosis. <i>Genetics in Medicine</i> , 2022, 24, S164.	2.4	0
5	Insufficient evidence for inclusion of motor deficits in the <scp>ASD</scp> diagnostic criteria: A response to Bhat (2021). <i>Autism Research</i> , 2022, 15, 1374-1375.	3.8	9
6	Sex differences in scores on standardized measures of autism symptoms: a multisite integrative data analysis. <i>Journal of Child Psychology and Psychiatry and Allied Disciplines</i> , 2021, 62, 97-106.	5.2	74
7	Seizure phenotype in CLN3 disease and its relation to other neurologic outcome measures. <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 1013-1020.	3.6	5
8	Prerequisite skills in cognitive testing: Innovations in theory and recommendations for practice. <i>Cognitive Development</i> , 2021, 58, 101038.	1.3	3
9	1-13C-propionate breath testing as a surrogate endpoint to assess efficacy of liver-directed therapies in methylmalonic acidemia (MMA). <i>Genetics in Medicine</i> , 2021, 23, 1522-1533.	2.4	10
10	Severity modeling of propionic acidemia using clinical and laboratory biomarkers. <i>Genetics in Medicine</i> , 2021, 23, 1534-1542.	2.4	13
11	The Early Screening for Autism and Communication Disorders: Field-testing an autism-specific screening tool for children 12 to 36 months of age. <i>Autism</i> , 2021, 25, 2112-2123.	4.1	15
12	X-linked creatine transporter deficiency results in prolonged QTc and increased sudden death risk in humans and disease model. <i>Genetics in Medicine</i> , 2021, 23, 1864-1872.	2.4	8
13	Inclusion of individuals with low IQ in drug development for autism spectrum disorder. <i>European Neuropsychopharmacology</i> , 2021, 48, 37-39.	0.7	2
14	Patterns of delay in early gross motor and expressive language milestone attainment in probands with genetic conditions versus idiopathic ASD from SFARI registries. <i>Journal of Child Psychology and Psychiatry and Allied Disciplines</i> , 2021, 62, 1297-1307.	5.2	13
15	A novel measure of matching categories for early development: Item creation and pilot feasibility study. <i>Research in Developmental Disabilities</i> , 2021, 115, 103993.	2.2	2
16	Systematic Review: Recommendations for Rehabilitation in ASD and ID From Clinical Practice Guidelines. <i>Archives of Rehabilitation Research and Clinical Translation</i> , 2021, 3, 100140.	0.9	9
17	Age of walking and intellectual ability in autism spectrum disorder and other neurodevelopmental disorders: a populationâ€“based study. <i>Journal of Child Psychology and Psychiatry and Allied Disciplines</i> , 2021, 62, 1070-1078.	5.2	6
18	TUBB3 Arg262His causes a recognizable syndrome including CFEOM3, facial palsy, joint contractures, and early-onset peripheral neuropathy. <i>Human Genetics</i> , 2021, 140, 1709-1731.	3.8	13

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19	Parent-reported measure of repetitive behavior in Phelan-McDermid syndrome. <i>Journal of Neurodevelopmental Disorders</i> , 2021, 13, 53.	3.1	6
20	Large-scale analyses of the relationship between sex, age and intelligence quotient heterogeneity and cortical morphometry in autism spectrum disorder. <i>Molecular Psychiatry</i> , 2020, 25, 614-628.	7.9	141
21	Concordance of the Vineland Adaptive Behavior Scales, second and third editions. <i>Journal of Intellectual Disability Research</i> , 2020, 64, 18-26.	2.0	37
22	Neurodevelopmental Characterization of Young Children Diagnosed with Niemann-Pick Disease, Type C1. <i>Journal of Developmental and Behavioral Pediatrics</i> , 2020, 41, 388-396.	1.1	6
23	Dr. Bishop et al. Reply. <i>Journal of the American Academy of Child and Adolescent Psychiatry</i> , 2020, 59, 1200-1202.	0.5	1
24	Psychometric Study of the Social Responsiveness Scale in Phelan-McDermid Syndrome. <i>Autism Research</i> , 2020, 13, 1383-1396.	3.8	14
25	Functional near-infrared spectroscopy in toddlers: Neural differentiation of communicative cues and relation to future language abilities. <i>Developmental Science</i> , 2020, 23, e12948.	2.4	5
26	Practice guideline: Treatment for insomnia and disrupted sleep behavior in children and adolescents with autism spectrum disorder. <i>Neurology</i> , 2020, 94, 392-404.	1.1	119
27	Greater cortical thickness in individuals with ASD. <i>Molecular Psychiatry</i> , 2020, 25, 507-508.	7.9	3
28	Psychiatric illness and regression in individuals with Phelan-McDermid syndrome. <i>Journal of Neurodevelopmental Disorders</i> , 2020, 12, 7.	3.1	51
29	Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. <i>Cell</i> , 2020, 180, 568-584.e23.	28.9	1,422
30	Diffusion Tensor Imaging Abnormalities in the Uncinate Fasciculus and Inferior Longitudinal Fasciculus in Phelan-McDermid Syndrome. <i>Pediatric Neurology</i> , 2020, 106, 24-31.	2.1	9
31	Changes in access to educational and healthcare services for individuals with intellectual and developmental disabilities during COVID-19 restrictions. <i>Journal of Intellectual Disability Research</i> , 2020, 64, 825-833.	2.0	190
32	Outcome Measures for Core Symptoms of Intellectual Disability: State of the Field. <i>American Journal on Intellectual and Developmental Disabilities</i> , 2020, 125, 418-433.	1.6	21
33	State of the Field: Differentiating Intellectual Disability From Autism Spectrum Disorder. <i>Frontiers in Psychiatry</i> , 2019, 10, 526.	2.6	135
34	A framework for the investigation of rare genetic disorders in neuropsychiatry. <i>Nature Medicine</i> , 2019, 25, 1477-1487.	30.7	90
35	The Need for a Developmentally Based Measure of Social Communication Skills. <i>Journal of the American Academy of Child and Adolescent Psychiatry</i> , 2019, 58, 555-560.	0.5	20
36	The gesture-language association over time in toddlers with and without language delays. <i>Autism and Developmental Language Impairments</i> , 2019, 4, 239694151984554.	1.6	13

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37	Long-Term Neuropsychological Outcomes from an Open-Label Phase I/IIa Trial of 2-Hydroxypropyl- β -Cyclodextrins (VTS-270) in Niemann-Pick Disease, Type C1. <i>CNS Drugs</i> , 2019, 33, 677-683.	5.9	28
38	Incontinence in Phelan-McDermid Syndrome. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2019, 69, e39-e42.	1.8	6
39	Early Indicators of Creatine Transporter Deficiency. <i>Journal of Pediatrics</i> , 2019, 206, 283-285.	1.8	10
40	Volumetric Analysis of the Basal Ganglia and Cerebellar Structures in Patients with Phelan-McDermid Syndrome. <i>Pediatric Neurology</i> , 2019, 90, 37-43.	2.1	19
41	Prospective longitudinal overnight video-EEG evaluation in Phelan-McDermid Syndrome. <i>Epilepsy and Behavior</i> , 2018, 80, 312-320.	1.7	12
42	Identification of 22q13 genes most likely to contribute to Phelan McDermid syndrome. <i>European Journal of Human Genetics</i> , 2018, 26, 293-302.	2.8	54
43	Framework for assessing individuals with rare genetic disorders associated with profound intellectual and multiple disabilities (PIMD): the example of Phelan McDermid Syndrome. <i>Clinical Neuropsychologist</i> , 2018, 32, 1226-1255.	2.3	55
44	Trajectories of cognitive development in toddlers with language delays. <i>Research in Developmental Disabilities</i> , 2018, 81, 65-72.	2.2	8
45	Talking About Death or Suicide: Prevalence and Clinical Correlates in Youth with Autism Spectrum Disorder in the Psychiatric Inpatient Setting. <i>Journal of Autism and Developmental Disorders</i> , 2018, 48, 3702-3710.	2.7	55
46	In-depth investigations of adolescents and adults with holoprosencephaly identify unique characteristics. <i>Genetics in Medicine</i> , 2018, 20, 14-23.	2.4	15
47	Loss of skills and onset patterns in neurodevelopmental disorders: Understanding the neurobiological mechanisms. <i>Autism Research</i> , 2018, 11, 212-222.	3.8	25
48	Cerebrospinal fluid vasopressin and symptom severity in children with autism. <i>Annals of Neurology</i> , 2018, 84, 611-615.	5.3	40
49	Classifying and characterizing the development of adaptive behavior in a naturalistic longitudinal study of young children with autism. <i>Journal of Neurodevelopmental Disorders</i> , 2018, 10, 1.	3.1	79
50	SOCIOEMOTIONAL AND BEHAVIORAL PROBLEMS IN TODDLERS WITH LANGUAGE DELAY. <i>Infant Mental Health Journal</i> , 2018, 39, 569-580.	1.8	13
51	Prospective phenotyping of NGLY1-CDDG, the first congenital disorder of deglycosylation. <i>Genetics in Medicine</i> , 2017, 19, 160-168.	2.4	124
52	Idiopathic Autism: Cellular and Molecular Phenotypes in Pluripotent Stem Cell-Derived Neurons. <i>Molecular Neurobiology</i> , 2017, 54, 4507-4523.	4.0	57
53	Modelling gesture use and early language development in autism spectrum disorder. <i>International Journal of Language and Communication Disorders</i> , 2017, 52, 637-651.	1.5	25
54	Identification of Developmental and Behavioral Markers Associated With Genetic Abnormalities in Autism Spectrum Disorder. <i>American Journal of Psychiatry</i> , 2017, 174, 576-585.	7.2	73

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55	Prefrontal Activation During Executive Tasks Emerges Over Early Childhood: Evidence From Functional Near Infrared Spectroscopy. <i>Developmental Neuropsychology</i> , 2017, 42, 253-264.	1.4	23
56	Serum and cerebrospinal fluid immune mediators in children with autistic disorder: a longitudinal study. <i>Molecular Autism</i> , 2017, 8, 1.	4.9	127
57	Intrathecal 2-hydroxypropyl- β -cyclodextrin decreases neurological disease progression in Niemann-Pick disease, type C1: a non-randomised, open-label, phase 1–2 trial. <i>Lancet, The</i> , 2017, 390, 1758-1768.	13.7	275
58	Describing Function in ASD: Using the DSM-5 and Other Methods to Improve Precision. <i>Journal of Autism and Developmental Disorders</i> , 2017, 47, 2938-2941.	2.7	11
59	Effects of Oxytocin and Vasopressin on Preferential Brain Responses to Negative Social Feedback. <i>Neuropsychopharmacology</i> , 2017, 42, 1409-1419.	5.4	40
60	Creatine Transporter Deficiency. <i>Journal of Developmental and Behavioral Pediatrics</i> , 2016, 37, 322-326.	1.1	11
61	Cohort study of neurocognitive functioning and adaptive behaviour in children and adolescents with Niemann-Pick Disease type C1. <i>Developmental Medicine and Child Neurology</i> , 2016, 58, 262-269.	2.1	13
62	CSF concentrations of 5-methyltetrahydrofolate in a cohort of young children with autism. <i>Neurology</i> , 2016, 86, 2258-2263.	1.1	25
63	Development, behavior, and biomarker characterization of Smith-Lemli-Opitz syndrome: an update. <i>Journal of Neurodevelopmental Disorders</i> , 2016, 8, 12.	3.1	45
64	Cortical thickness change in autism during early childhood. <i>Human Brain Mapping</i> , 2016, 37, 2616-2629.	3.6	41
65	Prospective Longitudinal Studies of Infant Siblings of Children With Autism: Lessons Learned and Future Directions. <i>Journal of the American Academy of Child and Adolescent Psychiatry</i> , 2016, 55, 179-187.	0.5	174
66	Autism Spectrum Disorder, Intellectual Disability, and Delayed Walking. <i>Pediatrics</i> , 2016, 137, e20152959.	2.1	32
67	Concurrent validity of the differential ability scales, second edition with the Mullen Scales of Early Learning in young children with and without neurodevelopmental disorders. <i>Child Neuropsychology</i> , 2016, 22, 556-569.	1.3	54
68	Autism screening and diagnosis in low resource settings: Challenges and opportunities to enhance research and services worldwide. <i>Autism Research</i> , 2015, 8, 473-476.	3.8	189
69	State-Dependent Differences in Functional Connectivity in Young Children With Autism Spectrum Disorder. <i>EBioMedicine</i> , 2015, 2, 1905-1915.	6.1	33
70	Convergent and divergent validity of the Mullen Scales of Early Learning in young children with and without autism spectrum disorder.. <i>Psychological Assessment</i> , 2015, 27, 1364-1378.	1.5	53
71	No evidence of antibodies against GAD65 and other specific antigens in children with autism. <i>BBA Clinical</i> , 2015, 4, 81-84.	4.1	6
72	Associated intellectual disability in a non-consanguineous family. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 1374-1380.	1.2	21

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73	Measurement of Nonverbal IQ in Autism Spectrum Disorder: Scores in Young Adulthood Compared to Early Childhood. <i>Journal of Autism and Developmental Disorders</i> , 2015, 45, 966-974.	2.7	67
74	Longitudinal study of symptom severity and language in minimally verbal children with autism. <i>Journal of Child Psychology and Psychiatry and Allied Disciplines</i> , 2015, 56, 97-104.	5.2	71
75	Evaluation of Periodic Limb Movements in Sleep and Iron Status in Children With Autism. <i>Pediatric Neurology</i> , 2015, 53, 343-349.	2.1	28
76	Determination of the allelic frequency in Smith's "Lemli" Opitz syndrome by analysis of massively parallel sequencing data sets. <i>Clinical Genetics</i> , 2015, 87, 570-575.	2.0	43
77	Social (pragmatic) communication disorder: a research review of this new DSM-5 diagnostic category. <i>Journal of Neurodevelopmental Disorders</i> , 2014, 6, 41.	3.1	96
78	Patterns of skill attainment and loss in young children with autism. <i>Development and Psychopathology</i> , 2014, 26, 203-214.	2.3	31
79	Depressive Symptoms in Young, Urban Schoolchildren: Environmental, Social, and Cognitive Risk. <i>Journal of Prevention and Intervention in the Community</i> , 2014, 42, 169-182.	0.7	3
80	Multisite Study of New Autism Diagnostic Interview-Revised (ADI-R) Algorithms for Toddlers and Young Preschoolers. <i>Journal of Autism and Developmental Disorders</i> , 2013, 43, 1527-1538.	2.7	75
81	Pharmacotherapy for the Core Symptoms in Autistic Disorder: Current Status of the Research. <i>Drugs</i> , 2013, 73, 303-314.	10.9	87
82	A pilot open-label trial of minocycline in patients with autism and regressive features. <i>Journal of Neurodevelopmental Disorders</i> , 2013, 5, 9.	3.1	51
83	Association of brain-derived neurotrophic factor (BDNF) haploinsufficiency with lower adaptive behaviour and reduced cognitive functioning in WAGR/11p13 deletion syndrome. <i>Cortex</i> , 2013, 49, 2700-2710.	2.4	61
84	Compared to What? Early Brain Overgrowth in Autism and the Perils of Population Norms. <i>Biological Psychiatry</i> , 2013, 74, 563-575.	1.3	107
85	Lack of Serum Antibodies against <i>Borrelia burgdorferi</i> in Children with Autism. <i>Vaccine Journal</i> , 2013, 20, 1092-1093.	3.1	6
86	Repetitive Behavior and Restricted Interests in Young Children with Autism: Comparisons with Controls and Stability Over 2 Years. <i>Autism Research</i> , 2013, 6, 584-595.	3.8	50
87	First Night Effect Analysis in a Cohort of Young Children with Autism Spectrum Disorder. <i>Journal of Clinical Sleep Medicine</i> , 2013, 09, 67-70.	2.6	15
88	Diffusion Tensor Imaging in Young Children with Autism: Biological Effects and Potential Confounds. <i>Biological Psychiatry</i> , 2012, 72, 1043-1051.	1.3	82
89	The ADOS Calibrated Severity Score: Relationship to Phenotypic Variables and Stability over Time. <i>Autism Research</i> , 2012, 5, 267-276.	3.8	94
90	Brief Report: Symptom Onset Patterns and Functional Outcomes in Young Children with Autism Spectrum Disorders. <i>Journal of Autism and Developmental Disorders</i> , 2011, 41, 1727-1732.	2.7	46

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91	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
92	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
93	Developmental Issues and Milestones. , 2011, , 159-173.		3
94	Testing autism interventions: trials and tribulations. Lancet, The, 2010, 375, 2124-2125.	13.7	24
95	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
96	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
97	Predictors of Language Acquisition in Preschool Children with Autism Spectrum Disorders. Journal of Autism and Developmental Disorders, 2007, 37, 1721-1734.	2.7	210