John R Ostergaard

List of Publications by Year
in descending order

61 papers	1,549 citations	394421 19 h-index	345221 36 g-index
65 all docs	65 docs citations	65 times ranked	2331 citing authors

#	Article	IF	Citations
1	Autism in Angelman Syndrome. Autism, 2004, 8, 163-174.	4.1	135
2	Prevalence, incidence, and age at diagnosis in Marfan Syndrome. Orphanet Journal of Rare Diseases, 2015, 10, 153.	2.7	130
3	Epilepsy and pregnancy: effect of antiepileptic drugs and lifestyle on birthweight. BJOG: an International Journal of Obstetrics and Gynaecology, 2000, 107, 896-902.	2.3	87
4	Congenital Heart Defects and Indices of Placental and Fetal Growth in a Nationwide Study of 924â€‰422 Liveborn Infants. Circulation, 2016, 134, 1546-1556.	1.6	82
5	Cardiac involvement in juvenile neuronal ceroid lipofuscinosis (Batten disease). Neurology, 2011, 76, 1245-1251.	1.1	77
6	Congenital Heart Defects and Indices of Fetal Cerebral Growth in a Nationwide Cohort of 924 422 Liveborn Infants. Circulation, 2016, 133, 566-575.	1.6	71
7	KMT2B-related disorders: expansion of the phenotypic spectrum and long-term efficacy of deep brain stimulation. Brain, 2020, 143, 3242-3261.	7.6	57
8	Early childhoodâ€onset restless legs syndrome: symptoms and effect of oral iron treatment. Acta Paediatrica, International Journal of Paediatrics, 2013, 102, e221-6.	1.5	54
9	Aortic events in a nationwide Marfan syndrome cohort. Clinical Research in Cardiology, 2017, 106, 105-112.	3.3	54
10	Prenatal Exposure to Cigarettes, Alcohol, and Coffee and the Risk for Febrile Seizures. Pediatrics, 2005, 116, 1089-1094.	2.1	51
11	Clinical history and management recommendations of the smooth muscle dysfunction syndrome due to ACTA2 arginine 179 alterations. Genetics in Medicine, 2018, 20, 1206-1215.	2.4	50
12	Neurodevelopmental outcome in Angelman syndrome: Genotypeâ€“phenotype correlations. Research in Developmental Disabilities, 2014, 35, 1742-1747.	2.2	40
13	Juvenile neuronal ceroid lipofuscinosis (Batten disease): current insights. Degenerative Neurological and Neuromuscular Disease, 2016, Volume 6, 73-83.	1.3	39
14	Smoking During Pregnancy and Hospitalization of the Child. Pediatrics, 1999, 104, e46-e46.	2.1	30
15	AlteredPLP1splicing causes hypomyelination of early myelinating structures. Annals of Clinical and Translational Neurology, 2015, 2, 648-661.	3.7	27
16	Endocrine and Growth Abnormalities in 4H Leukodystrophy Caused by Variants in POLR3A, POLR3B, and POLR1C. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e660-e674.	3.6	26
17	Febrile seizures. Acta Paediatrica, International Journal of Paediatrics, 2009, 98, 771-773.	1.5	25
18	Anxiety and depression in Klinefelter syndrome: The impact of personality and social engagement. PLoS ONE, 2018, 13, e0206932.	2.5	24

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19	Further delineation of 17p13.3 microdeletion involving CRK. The effect of growth hormone treatment. European Journal of Medical Genetics, 2012, 55, 22-26.	1.3	23
20	Developmental Enamel Defects in Children Prenatally Exposed to Anti-Epileptic Drugs. PLoS ONE, 2013, 8, e58213.	2.5	23
21	Congenital Heart Defects and Risk of Epilepsy. Circulation, 2016, 134, 1689-1691.	1.6	22
22	Nanceâ€“Horan syndromeâ€”The oral perspective on a rare disease. American Journal of Medical Genetics, Part A, 2017, 173, 88-98.	1.2	21
23	Congenital Heart Defects and the Risk of Spontaneous Preterm Birth. Journal of Pediatrics, 2021, 229, 168-174.e5.	1.8	21
24	Hospital-Diagnosed Pertussis Infection in Children and Long-term Risk of Epilepsy. JAMA - Journal of the American Medical Association, 2015, 314, 1844.	7.4	19
25	Educational delay and attainment in persons with neurofibromatosis 1 in Denmark. European Journal of Human Genetics, 2019, 27, 857-868.	2.8	19
26	Clinical characteristics and quality of life, depression, and anxiety in adults with neurofibromatosis type 1: A nationwide study. American Journal of Medical Genetics, Part A, 2020, 182, 1704-1715.	1.2	19
27	Do females with juvenile ceroid lipofuscinosis (Batten disease) have a more severe disease course? The Danish experience. European Journal of Paediatric Neurology, 2013, 17, 265-268.	1.6	18
28	Eating behavior, prenatal and postnatal growth in Angelman syndrome. Research in Developmental Disabilities, 2014, 35, 2681-2690.	2.2	18
29	Teenagers and young adults with neurofibromatosis type 1 are more likely to experience loneliness than siblings without the illness. Acta Paediatrica, International Journal of Paediatrics, 2015, 104, 604-609.	1.5	18
30	Respiratory distress syndrome in moderately late and late preterm infants and risk of cerebral palsy: a population-based cohort study. BMJ Open, 2016, 6, e011643.	1.9	18
31	Paroxysmal sympathetic hyperactivity in Juvenile neuronal ceroid lipofuscinosis (Batten disease). Autonomic Neuroscience: Basic and Clinical, 2018, 214, 15-18.	2.8	18
32	Cardiac pathology in neuronal ceroid lipofuscinoses (NCL): More than a mere co-morbidity. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2020, 1866, 165643.	3.8	18
33	Difficulties in diagnosing Marfan syndrome using current FBN1 databases. Genetics in Medicine, 2016, 18, 98-102.	2.4	17
34	Spinal manifestations of CLN1 disease start during the early postnatal period. Neuropathology and Applied Neurobiology, 2021, 47, 251-267.	3.2	16
35	Reliability of GMFCS family report questionnaire. Disability and Rehabilitation, 2012, 34, 721-724.	1.8	15
36	Multisystem burden of neurofibromatosis 1 in Denmark: registry- and population-based rates of hospitalizations over the life span. Genetics in Medicine, 2020, 22, 1069-1078.	2.4	15

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37	Klinefelter syndrome has increased brain responses to auditory stimuli and motor output, but not to visual stimuli or Stroop adaptation. NeuroImage: Clinical, 2016, 11, 239-251.	2.7	14
38	Prenatal Exposure to Antiepileptic Drugs and Dental Agenesis. PLoS ONE, 2014, 9, e84420.	2.5	11
39	Post-Varicella Angiopathy: A Series of 4 Patients With Focus on Virologic and Neuroimaging Findings. Pediatric Neurology, 2014, 50, 581-585.	2.1	11
40	A novel single nucleotide splice site mutation in FHL1 confirms an Emery-Dreifuss plus phenotype with pulmonary artery hypoplasia and facial dysmorphology. European Journal of Medical Genetics, 2015, 58, 222-229.	1.3	11
41	Congenital Heart Defects and Measures of Fetal Growth in Newborns with Down Syndrome or 22q11.2 Deletion Syndrome. Journal of Pediatrics, 2016, 175, 116-122.e4.	1.8	10
42	Gait phenotype in Batten disease: A marker of disease progression. European Journal of Paediatric Neurology, 2021, 35, 1-7.	1.6	10
43	Intraneural Perineurioma of the Sciatic Nerve in Early Childhood. Pediatric Neurology, 2009, 41, 68-70.	2.1	9
44	Epilepsy and cataplexy in Angelman syndrome. Genotype-phenotype correlations. Research in Developmental Disabilities, 2016, 56, 177-182.	2.2	9
45	Impact of child and family characteristics on cerebral palsy treatment. Developmental Medicine and Child Neurology, 2015, 57, 948-954.	2.1	8
46	Psychiatric disorders in individuals with neurofibromatosis 1 in Denmark: A nationwide registerâ€based cohort study. American Journal of Medical Genetics, Part A, 2021, 185, 3706-3716.	1.2	8
47	Two Cockayne Syndrome patients with a novel splice site mutation â€“ clinical and metabolic analyses. Mechanisms of Ageing and Development, 2018, 175, 7-16.	4.6	7
48	Valproate-induced hyperammonemia in juvenile ceroid lipofuscinosis (Batten disease). Seizure: the Journal of the British Epilepsy Association, 2014, 23, 429-434.	2.0	6
49	Cataract and Glaucoma Development in Juvenile Neuronal Ceroid Lipofuscinosis (Batten Disease). Ophthalmic Genetics, 2015, 36, 39-42.	1.2	6
50	Bone Geometry, Density, and Microarchitecture in the Distal Radius and Tibia in Adults With Marfan Syndrome Assessed by <scp>HRâ€pQCT</scp>. Journal of Bone and Mineral Research, 2020, 35, 2335-2344.	2.8	6
51	Development of hypomelanotic macules is associated with constitutive activated mTORC1 in tuberous sclerosis complex. Molecular Genetics and Metabolism, 2017, 120, 384-391.	1.1	5
52	Prevalence of mental disorders in children and adolescents with cerebral palsy: Danish nationwide follow-up study. European Journal of Paediatric Neurology, 2020, 27, 98-103.	1.6	5
53	Forming and ending marital or cohabiting relationships in a Danish population-based cohort of individuals with neurofibromatosis 1. European Journal of Human Genetics, 2020, 28, 1028-1033.	2.8	5
54	Constipation in adults with neurofibromatosis type 1. Orphanet Journal of Rare Diseases, 2017, 12, 139.	2.7	4

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55	Pregnancy outcomes in women with neurofibromatosis 1: a Danish population-based cohort study. Journal of Medical Genetics, 2022, 59, 237-242.	3.2	3
56	Phenotype of a child with Angelman syndrome born to a woman with Praderâ€“Willi syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 2138-2144.	1.2	2
57	Exploring associations between constipation, severity of neurofibromatosis type 1 and NF1 mutational spectrum. Scientific Reports, 2021, 11, 9179.	3.3	1
58	Pacemaker Implantation in Juvenile Neuronal Ceroid Lipofuscinosis (CLN3)â€“A Long-Term Follow-Up Study. Frontiers in Neurology, 2022, 13, 846240.	2.4	1
59	Evidence for an association between prematurity and enamel defects in permanent teeth is still relatively sparse. European Journal of Oral Sciences, 2014, 122, 361-361.	1.5	0
60	Do individuals with Angelman syndrome have a maladaptive behavior?. American Journal of Medical Genetics, Part A, 2019, 179, 2317-2318.	1.2	0
61	Reply. Journal of Pediatrics, 2021, 230, 273-274.	1.8	0