# John R Ostergaard

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/4265875/publications.pdf

Version: 2024-02-01

| | | | |
|---|---|---|---|
| 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 | Pregnancy outcomes in women with neurofibromatosis 1: a Danish population-based cohort study. Journal of Medical Genetics, 2022, 59, 237-242. | 3.2 | 3 |
| 2 | Pacemaker Implantation in Juvenile Neuronal Ceroid Lipofuscinosis (CLN3)â€“A Long-Term Follow-Up Study. Frontiers in Neurology, 2022, 13, 846240. | 2.4 | 1 |
| 3 | Congenital Heart Defects and the Risk of Spontaneous Preterm Birth. Journal of Pediatrics, 2021, 229, 168-174.e5. | 1.8 | 21 |
| 4 | Spinal manifestations of CLN1 disease start during the early postnatal period. Neuropathology and Applied Neurobiology, 2021, 47, 251-267. | 3.2 | 16 |
| 5 | 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 |
| 6 | Reply. Journal of Pediatrics, 2021, 230, 273-274. | 1.8 | 0 |
| 7 | Exploring associations between constipation, severity of neurofibromatosis type 1 and NF1 mutational spectrum. Scientific Reports, 2021, 11, 9179. | 3.3 | 1 |
| 8 | 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 |
| 9 | Gait phenotype in Batten disease: A marker of disease progression. European Journal of Paediatric Neurology, 2021, 35, 1-7. | 1.6 | 10 |
| 10 | 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 |
| 11 | 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 |
| 12 | 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 |
| 13 | KMT2B-related disorders: expansion of the phenotypic spectrum and long-term efficacy of deep brain stimulation. Brain, 2020, 143, 3242-3261. | 7.6 | 57 |
| 14 | 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 |
| 15 | 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 |
| 16 | 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 |
| 17 | Do individuals with Angelman syndrome have a maladaptive behavior?. American Journal of Medical Genetics, Part A, 2019, 179, 2317-2318. | 1.2 | 0 |
| 18 | Educational delay and attainment in persons with neurofibromatosis 1 in Denmark. European Journal of Human Genetics, 2019, 27, 857-868. | 2.8 | 19 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | 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 |
| 20 | Anxiety and depression in Klinefelter syndrome: The impact of personality and social engagement. PLoS ONE, 2018, 13, e0206932. | 2.5 | 24 |
| 21 | 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 |
| 22 | Paroxysmal sympathetic hyperactivity in Juvenile neuronal ceroid lipofuscinosis (Batten disease). Autonomic Neuroscience: Basic and Clinical, 2018, 214, 15-18. | 2.8 | 18 |
| 23 | 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 |
| 24 | Aortic events in a nationwide Marfan syndrome cohort. Clinical Research in Cardiology, 2017, 106, 105-112. | 3.3 | 54 |
| 25 | Nanceâ€“Horan syndromeâ€”The oral perspective on a rare disease. American Journal of Medical Genetics, Part A, 2017, 173, 88-98. | 1.2 | 21 |
| 26 | Constipation in adults with neurofibromatosis type 1. Orphanet Journal of Rare Diseases, 2017, 12, 139. | 2.7 | 4 |
| 27 | Juvenile neuronal ceroid lipofuscinosis (Batten disease): current insights. Degenerative Neurological and Neuromuscular Disease, 2016, Volume 6, 73-83. | 1.3 | 39 |
| 28 | Epilepsy and cataplexy in Angelman syndrome. Genotype-phenotype correlations. Research in Developmental Disabilities, 2016, 56, 177-182. | 2.2 | 9 |
| 29 | 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 |
| 30 | 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 |
| 31 | Congenital Heart Defects and Risk of Epilepsy. Circulation, 2016, 134, 1689-1691. | 1.6 | 22 |
| 32 | 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 |
| 33 | 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 |
| 34 | 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 |
| 35 | Difficulties in diagnosing Marfan syndrome using current FBN1 databases. Genetics in Medicine, 2016, 18, 98-102. | 2.4 | 17 |
| 36 | AlteredPLP1splicing causes hypomyelination of early myelinating structures. Annals of Clinical and Translational Neurology, 2015, 2, 648-661. | 3.7 | 27 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | Prevalence, incidence, and age at diagnosis in Marfan Syndrome. Orphanet Journal of Rare Diseases, 2015, 10, 153. | 2.7 | 130 |
| 38 | 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 |
| 39 | Impact of child and family characteristics on cerebral palsy treatment. Developmental Medicine and Child Neurology, 2015, 57, 948-954. | 2.1 | 8 |
| 40 | Cataract and Glaucoma Development in Juvenile Neuronal Ceroid Lipofuscinosis (Batten Disease). Ophthalmic Genetics, 2015, 36, 39-42. | 1.2 | 6 |
| 41 | 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 |
| 42 | 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 |
| 43 | 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 |
| 44 | Prenatal Exposure to Antiepileptic Drugs and Dental Agenesis. PLoS ONE, 2014, 9, e84420. | 2.5 | 11 |
| 45 | 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 |
| 46 | Post-Varicella Angiopathy: A Series of 4 Patients With Focus on Virologic and Neuroimaging Findings. Pediatric Neurology, 2014, 50, 581-585. | 2.1 | 11 |
| 47 | Eating behavior, prenatal and postnatal growth in Angelman syndrome. Research in Developmental Disabilities, 2014, 35, 2681-2690. | 2.2 | 18 |
| 48 | Neurodevelopmental outcome in Angelman syndrome: Genotypeâ€“phenotype correlations. Research in Developmental Disabilities, 2014, 35, 1742-1747. | 2.2 | 40 |
| 49 | Valproate-induced hyperammonemia in juvenile ceroid lipofuscinosis (Batten disease). Seizure: the Journal of the British Epilepsy Association, 2014, 23, 429-434. | 2.0 | 6 |
| 50 | 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 |
| 51 | 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 |
| 52 | Developmental Enamel Defects in Children Prenatally Exposed to Anti-Epileptic Drugs. PLoS ONE, 2013, 8, e58213. | 2.5 | 23 |
| 53 | Reliability of GMFCS family report questionnaire. Disability and Rehabilitation, 2012, 34, 721-724. | 1.8 | 15 |
| 54 | 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 |

| # | Article | IF | Citations |
|---|---|---|---|
| 55 | Cardiac involvement in juvenile neuronal ceroid lipofuscinosis (Batten disease). Neurology, 2011, 76, 1245-1251. | 1.1 | 77 |
| 56 | Febrile seizures. Acta Paediatrica, International Journal of Paediatrics, 2009, 98, 771-773. | 1.5 | 25 |
| 57 | Intraneural Perineurioma of the Sciatic Nerve in Early Childhood. Pediatric Neurology, 2009, 41, 68-70. | 2.1 | 9 |
| 58 | Prenatal Exposure to Cigarettes, Alcohol, and Coffee and the Risk for Febrile Seizures. Pediatrics, 2005, 116, 1089-1094. | 2.1 | 51 |
| 59 | Autism in Angelman Syndrome. Autism, 2004, 8, 163-174. | 4.1 | 135 |
| 60 | 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 |
| 61 | Smoking During Pregnancy and Hospitalization of the Child. Pediatrics, 1999, 104, e46-e46. | 2.1 | 30 |