# Mitsugu Uematsu

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/2236565/publications.pdf

Version: 2024-02-01

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|---|---|---|---|
| 93 papers | 1,290 citations | 304743 22 h-index | 454955 30 g-index |
| 93 all docs | 93 docs citations | 93 times ranked | 2435 citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | <scp>GRIN</scp>1 mutations cause encephalopathy with infantileâ€onset epilepsy, and hyperkinetic and stereotyped movement disorders. Epilepsia, 2015, 56, 841-848. | 5.1 | 76 |
| 2 | Acute encephalopathy in children with Dravet syndrome. Epilepsia, 2012, 53, 79-86. | 5.1 | 53 |
| 3 | Genomic analysis identifies candidate pathogenic variants in 9 of 18 patients with unexplained West syndrome. Human Genetics, 2015, 134, 649-658. | 3.8 | 51 |
| 4 | Deletions of SCN1A 5â€² genomic region with promoter activity in Dravet syndrome. Human Mutation, 2010, 31, 820-829. | 2.5 | 47 |
| 5 | Mitochonic Acid 5 (MA-5) Facilitates ATP Synthase Oligomerization and Cell Survival in Various Mitochondrial Diseases. EBioMedicine, 2017, 20, 27-38. | 6.1 | 46 |
| 6 | Intramuscular reninâ€“angiotensin system is activated in human muscular dystrophy. Journal of the Neurological Sciences, 2009, 280, 40-48. | 0.6 | 45 |
| 7 | Parental satisfaction and seizure outcome after corpus callosotomy in patients with infantile or early childhood onset epilepsy. Seizure: the Journal of the British Epilepsy Association, 2013, 22, 303-305. | 2.0 | 37 |
| 8 | Complete remission of seizures after corpus callosotomy. Journal of Neurosurgery: Pediatrics, 2012, 10, 7-13. | 1.3 | 35 |
| 9 | Human occipital cortices differentially exert saccadic suppression: Intracranial recording in children. NeuroImage, 2013, 83, 224-236. | 4.2 | 35 |
| 10 | Efficacy of Sumatriptan in Two Pediatric Cases With Abdominal Pain-Related Functional Gastrointestinal Disorders: Does the Mechanism Overlap That of Migraine?. Journal of Child Neurology, 2010, 25, 234-237. | 1.4 | 33 |
| 11 | Reduced levels of interleukin-1 receptor antagonist in the cerebrospinal fluid in patients with West syndrome. Epilepsy Research, 2009, 85, 314-317. | 1.6 | 32 |
| 12 | Two cases of early-onset myoclonic seizures with continuous parietal delta activity caused by EEF1A2 mutations. Brain and Development, 2016, 38, 520-524. | 1.1 | 32 |
| 13 | Clinical features and long-term outcome of a group of Japanese children with inflammatory central nervous system disorders and seropositivity to myelin-oligodendrocyte glycoprotein antibodies. Brain and Development, 2015, 37, 849-852. | 1.1 | 30 |
| 14 | A further case of renal tubular dysgenesis surviving the neonatal period. European Journal of Pediatrics, 2009, 168, 207-209. | 2.7 | 28 |
| 15 | Efficacy of idebenone for respiratory failure in a patient with Leigh syndrome: A long-term follow-up study. Journal of the Neurological Sciences, 2009, 278, 112-114. | 0.6 | 28 |
| 16 | Elevated serum levels of neutrophil elastase in patients with influenza virus-associated encephalopathy. Journal of the Neurological Sciences, 2015, 349, 190-195. | 0.6 | 28 |
| 17 | Elevated plasma levels of tissue inhibitors of metalloproteinase-1 and their overexpression in muscle in human and mouse muscular dystrophy. Journal of the Neurological Sciences, 2010, 297, 19-28. | 0.6 | 27 |
| 18 | Asymptomatic congenital cytomegalovirus infection with neurological sequelae: A retrospective study using umbilical cord. Brain and Development, 2016, 38, 819-826. | 1.1 | 27 |

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|---|---|---|---|
| 19 | A case surviving for over a year of renal tubular dysgenesis with compound heterozygous angiotensinogen gene mutations. American Journal of Medical Genetics, Part A, 2006, 140A, 2355-2360. | 1.2 | 26 |
| 20 | Genetic heterogeneity in 26 infants with a hypomyelinating leukodystrophy. Human Genetics, 2016, 135, 89-98. | 3.8 | 26 |
| 21 | Effect of a blackout in pediatric patients with home medical devices during the 2011 eastern Japan earthquake. Brain and Development, 2014, 36, 143-147. | 1.1 | 25 |
| 22 | Clinical profiles for seizure remission and developmental gains after total corpus callosotomy. Brain and Development, 2016, 38, 47-53. | 1.1 | 24 |
| 23 | Acute encephalitis with refractory, repetitive partial seizures: Pathological findings and a new therapeutic approach using tacrolimus. Brain and Development, 2016, 38, 772-776. | 1.1 | 22 |
| 24 | Novel mutations in five Japanese patients with 3-methylcrotonyl-CoA carboxylase deficiency. Journal of Human Genetics, 2007, 52, 1040-1043. | 2.3 | 18 |
| 25 | Utility of subtraction ictal SPECT images in detecting focal leading activity and understanding the pathophysiology of spasms in patients with West syndrome. Epilepsy Research, 2009, 83, 177-183. | 1.6 | 18 |
| 26 | Pyridoxal 5â€²-phosphate and related metabolites in hypophosphatasia: Effects of enzyme replacement therapy. Molecular Genetics and Metabolism, 2018, 125, 174-180. | 1.1 | 18 |
| 27 | Unique discrepancy between cerebral blood flow and glucose metabolism in hemimegalencephaly. Epilepsy Research, 2010, 92, 201-208. | 1.6 | 17 |
| 28 | Lateralization of interictal spikes after corpus callosotomy. Clinical Neurophysiology, 2011, 122, 2121-2127. | 1.5 | 17 |
| 29 | Attachment Disorder and Early Media Exposure: Neurobehavioral symptoms mimicking autism spectrum disorder. Journal of Medical Investigation, 2018, 65, 280-282. | 0.5 | 17 |
| 30 | Efficacy of long term weekly ACTH therapy for intractable epilepsy. Brain and Development, 2015, 37, 449-454. | 1.1 | 16 |
| 31 | A case with central and peripheral hypomyelination with hypogonadotropic hypogonadism and hypodontia (4H syndrome) plus cataract. Journal of the Neurological Sciences, 2011, 300, 179-181. | 0.6 | 15 |
| 32 | Interhemispheric Vertical Hemispherotomy: A Single Center Experience. Pediatric Neurosurgery, 2015, 50, 295-300. | 0.7 | 15 |
| 33 | Myoclonic axial jerks for diagnosing atypical evolution of ataxia telangiectasia. Brain and Development, 2015, 37, 362-365. | 1.1 | 15 |
| 34 | Successful cord blood transplantation with reducedâ€intensity conditioning for childhood cerebral Xâ€linked adrenoleukodystrophy at advanced and early stages. Pediatric Transplantation, 2012, 16, E63-70. | 1.0 | 13 |
| 35 | Phenytoin-responsive epileptic encephalopathy with a tandem duplication involving FGF12. Neurology: Genetics, 2017, 3, e133. | 1.9 | 13 |
| 36 | Successful Treatment With Sumatriptan in a Case With Cyclic Vomiting Syndrome Combined With 18qâ€” Syndrome. Journal of Child Neurology, 2009, 24, 1561-1563. | 1.4 | 12 |

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|---|---|---|---|
| 37 | Functional cortical deafferentation from the subcortical structures in a patient with early myoclonic encephalopathy: A functional neuroimaging study. Epilepsia, 2010, 51, 699-702. | 5.1 | 12 |
| 38 | FDG-PET study of patients with Leigh syndrome. Journal of the Neurological Sciences, 2016, 362, 309-313. | 0.6 | 12 |
| 39 | Neuroepidemiology of West syndrome and early infantile epileptic encephalopathy in Miyagi Prefecture, Japan. Epilepsy Research, 2009, 87, 299-301. | 1.6 | 11 |
| 40 | [18F]fluorodeoxyglucose-positron emission tomography study of genetically confirmed patients with Dravet syndrome. Epilepsy Research, 2018, 147, 9-14. | 1.6 | 11 |
| 41 | Abdominal Migraine Associated with Ecchymosis of the Legs and Buttocks: Does the Symptom Imply an Unknown Mechanism of Migraine?. Tohoku Journal of Experimental Medicine, 2010, 221, 49-51. | 1.2 | 10 |
| 42 | Hypoperfusion in caudate nuclei in patients with brainâ€“lungâ€“thyroid syndrome. Journal of the Neurological Sciences, 2012, 315, 77-81. | 0.6 | 10 |
| 43 | Mucolipidosis IV: A milder form with novel mutations and serial MRI findings. Brain and Development, 2016, 38, 763-767. | 1.1 | 10 |
| 44 | Rett-like features and cortical visual impairment in a Japanese patient with HECW2 mutation. Brain and Development, 2018, 40, 410-414. | 1.1 | 10 |
| 45 | The usefulness of subtraction ictal SPECT and ictal near-infrared spectroscopic topography in patients with West syndrome. Brain and Development, 2013, 35, 887-893. | 1.1 | 9 |
| 46 | RBPJ is disrupted in a case of proximal 4p deletion syndrome with epilepsy. Brain and Development, 2014, 36, 532-536. | 1.1 | 9 |
| 47 | 8p deletion and 9p duplication in two children with electrical status epilepticus in sleep syndrome. Seizure: the Journal of the British Epilepsy Association, 2012, 21, 295-299. | 2.0 | 8 |
| 48 | Abdominal migraine reviewed from both central and peripheral aspects. World Journal of Experimental Medicine, 2012, 2, 75. | 1.7 | 7 |
| 49 | Early replacement therapy in a first Japanese case with autosomal recessive guanosine triphosphate cyclohydrolase I deficiency with a novel point mutation. Brain and Development, 2014, 36, 268-271. | 1.1 | 7 |
| 50 | Novel missense mutation in CLN8 in late infantile neuronal ceroid lipofuscinosis: The first report of a CLN8 mutation in Japan. Brain and Development, 2016, 38, 341-345. | 1.1 | 7 |
| 51 | Reversible brain atrophy in glutaric aciduria type 1. Brain and Development, 2017, 39, 532-535. | 1.1 | 7 |
| 52 | Initial vasodilatation in a child with reversible cerebral vasoconstriction syndrome. Journal of Clinical Neuroscience, 2017, 39, 108-110. | 1.5 | 7 |
| 53 | Neonatal-Onset Brainstem Reticular Reflex Myoclonus Following a Prenatal Brain Insult: Generalized Myoclonic Jerk and a Brainstem Lesion. Tohoku Journal of Experimental Medicine, 2007, 211, 303-308. | 1.2 | 6 |
| 54 | Clinical Reasoning: A young man with progressive subcortical lesions and optic nerve atrophy. Neurology, 2012, 79, e63-8. | 1.1 | 6 |

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|---|---|---|---|
| 55 | Lower Back Pain as a Symptom of Migrainous Corpalgia. Journal of Child Neurology, 2013, 28, 676-677. | 1.4 | 6 |
| 56 | Abdominal and Lower Back Pain in Pediatric Idiopathic Stabbing Headache. Pediatrics, 2014, 133, e245-e247. | 2.1 | 6 |
| 57 | SLC16A2 mutations in two Japanese patients with Allanâ€“Herndonâ€“Dudley syndrome. Human Genome Variation, 2014, 1, 14010. | 0.7 | 6 |
| 58 | Long-Term Evaluation of Low-Dose Betamethasone for Ataxia Telangiectasia. Pediatric Neurology, 2019, 100, 60-66. | 2.1 | 6 |
| 59 | The effect of dietary protein restriction in a case of molybdenum cofactor deficiency with MOCS1 mutation. Molecular Genetics and Metabolism Reports, 2021, 26, 100716. | 1.1 | 6 |
| 60 | Another Case of Lower Back Pain Associated With Migraine. Journal of Child Neurology, 2013, 28, 680-680. | 1.4 | 5 |
| 61 | First Japanese case of Zellweger syndrome with a mutation in PEX14. Pediatrics International, 2015, 57, 1189-1192. | 0.5 | 5 |
| 62 | A novel PLP1 frameshift mutation causing a milder form of Pelizaeusâ€“Merzbacher disease. Brain and Development, 2015, 37, 455-458. | 1.1 | 5 |
| 63 | Aquaporin-4 autoimmunity in a child without optic neuritis and myelitis. Brain and Development, 2015, 37, 149-152. | 1.1 | 5 |
| 64 | Patchy white matter hyperintensity in ring chromosome 18 syndrome. Pediatrics International, 2016, 58, 919-922. | 0.5 | 5 |
| 65 | Increased Ki-67 immunoreactivity in the white matter in hemimegalencephaly. Neuroscience Letters, 2013, 548, 244-248. | 2.1 | 4 |
| 66 | Renal function in angiotensinogen gene-mutated renal tubular dysgenesis with glomerular cysts. Pediatric Nephrology, 2015, 30, 357-360. | 1.7 | 4 |
| 67 | A case of 3p deletion syndrome associated with cerebellar hemangioblastoma. Brain and Development, 2016, 38, 257-260. | 1.1 | 4 |
| 68 | Behavioral problems and family distress in tuberous sclerosis complex. Epilepsy and Behavior, 2020, 111, 107321. | 1.7 | 4 |
| 69 | Leigh syndrome-like MRI changes in a patient with biallelic HPDL variants treated with ketogenic diet. Molecular Genetics and Metabolism Reports, 2021, 29, 100800. | 1.1 | 4 |
| 70 | Brain White Matter Abnormality in a Newborn Infant with Congenital Adrenal Hyperplasia. Clinical Pediatric Endocrinology, 2013, 22, 77-81. | 0.8 | 4 |
| 71 | Age-Related Recovery of Daily Living Activity After 1-Stage Complete Corpus Callosotomy: A Retrospective Analysis of 41 Cases. Neurosurgery, 2022, 90, 547-551. | 1.1 | 4 |
| 72 | The Cerebrospinal Fluid Level of 5-Methylterahydrofolate in a Japanese Boy with Hypomyelination with Atrophy of the Basal Ganglia and Cerebellum. Tohoku Journal of Experimental Medicine, 2007, 213, 373. | 1.2 | 3 |

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|---|---|---|---|
| 73 | A case of atypical benign partial epilepsy with action myoclonus. Seizure: the Journal of the British Epilepsy Association, 2013, 22, 242-245. | 2.0 | 3 |
| 74 | Asymmetric Drug-Induced Tremor: Rare Feature of a Common Event. Pediatric Neurology, 2013, 48, 479-480. | 2.1 | 3 |
| 75 | The Onset of Interictal Spike-Related Ripples Facilitates Detection of the Epileptogenic Zone. Frontiers in Neurology, 2021, 12, 724417. | 2.4 | 3 |
| 76 | Ketotifen overdose in infancy associated with development of epilepsy and mild mental retardation. Pediatrics International, 2012, 54, 963-963. | 0.5 | 2 |
| 77 | Utility of Thallium-201 Scintigraphy in Tolosa-Hunt Syndrome. Tohoku Journal of Experimental Medicine, 2013, 229, 83-86. | 1.2 | 2 |
| 78 | A Case of Acute Cerebellitis With a Unique Sequential Change on Magnetic Resonance Imaging. Pediatric Neurology, 2014, 51, 279-281. | 2.1 | 2 |
| 79 | A patient with a 6q22.1 deletion and a phenotype of non-progressive early-onset generalized epilepsy with tremor. Epilepsy and Behavior Reports, 2021, 15, 100405. | 1.0 | 2 |
| 80 | Two types of early epileptic encephalopathy in a Pitt-Hopkins syndrome patient with a novel TCF4 mutation. Brain and Development, 2022, 44, 148-152. | 1.1 | 2 |
| 81 | The longest reported sibling survivors of a severe form of congenital myasthenic syndrome with the <scp> ALG14 </scp> pathogenic variant. American Journal of Medical Genetics, Part A, 2022, 188, 1293-1298. | 1.2 | 2 |
| 82 | Ictal Vomiting as an Initial Symptom of Severe Myoclonic Epilepsy in Infancy: A Case Report. Journal of Child Neurology, 2009, 24, 228-230. | 1.4 | 1 |
| 83 | A female case of West syndrome with remission of spasms following multiple cerebral hemorrhages. Brain and Development, 2011, 33, 678-682. | 1.1 | 1 |
| 84 | Unusual ribbonâ€like periventricular heterotopia with congenital cataracts in a Japanese girl. American Journal of Medical Genetics, Part A, 2012, 158A, 674-677. | 1.2 | 1 |
| 85 | Late vitamin <scp>K</scp> deficiency bleeding in an infant born at a maternity hospital. Pediatrics International, 2014, 56, 127-128. | 0.5 | 1 |
| 86 | Pediatric-Onset Extracephalic Stabbing Pain. Journal of Child Neurology, 2014, 29, NP47-NP48. | 1.4 | 1 |
| 87 | Neonatal â€œSeizureâ€•While Riding in a Car. Clinical Pediatrics, 2014, 53, 605-606. | 0.8 | 1 |
| 88 | Extremely low-dose vigabatrin for West syndrome with tuberous sclerosis. Journal of Pediatric Epilepsy, 2015, 02, 255-258. | 0.2 | 1 |
| 89 | Restless legs syndrome: A unique case of relapse after traffic accidents with a long remission. Sleep Medicine, 2010, 11, 596-597. | 1.6 | 0 |
| 90 | Periodic Eye Movements and Epileptic Spasms in West Syndrome. Journal of Child Neurology, 2013, 28, 1483-1484. | 1.4 | 0 |

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|---|---|---|---|
| 91 | Correspondence on â€œClinical Characterization of Gastroenteritis-Related Seizures in Children: Impact of Fever and Serum Sodium Levelsâ€. Journal of Child Neurology, 2014, 29, 1578-1579. | 1.4 | 0 |
| 92 | In response to letter to Editor by Nosaka et al. on our paper: Elevated serum levels of neutrophil elastase in patients with influenza virus-associated encephalopathy. J Neurol Sci 2015;349:190â€“195. Journal of the Neurological Sciences, 2017, 372, 50. | 0.6 | 0 |
| 93 | A pediatric case of osteosarcoma and tuberous sclerosis complex with a novel germline mutation in the TSC2 gene and a somatic mutation in the TP53 gene. Pediatric Blood and Cancer, 2021, 68, e28960. | 1.5 | 0 |