

# Mitsugu Uematsu

## List of Publications by Year in descending order

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Version: 2024-02-01

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	Two types of early epileptic encephalopathy in a Pitt-Hopkins syndrome patient with a novel TCF4 mutation. <i>Brain and Development</i> , 2022, 44, 148-152.	1.1	2
2	Age-Related Recovery of Daily Living Activity After 1-Stage Complete Corpus Callosotomy: A Retrospective Analysis of 41 Cases. <i>Neurosurgery</i> , 2022, 90, 547-551.	1.1	4
3	The longest reported sibling survivors of a severe form of congenital myasthenic syndrome with the <sc> <i>ALG14</i> </sc> pathogenic variant. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 1293-1298.	1.2	2
4	A patient with a 6q22.1 deletion and a phenotype of non-progressive early-onset generalized epilepsy with tremor. <i>Epilepsy and Behavior Reports</i> , 2021, 15, 100405.	1.0	2
5	A pediatric case of osteosarcoma and tuberous sclerosis complex with a novel germline mutation in the <i>TSC2</i> gene and a somatic mutation in the <i>TP53</i> gene. <i>Pediatric Blood and Cancer</i> , 2021, 68, e28960.	1.5	0
6	The effect of dietary protein restriction in a case of molybdenum cofactor deficiency with MOCS1 mutation. <i>Molecular Genetics and Metabolism Reports</i> , 2021, 26, 100716.	1.1	6
7	Leigh syndrome-like MRI changes in a patient with biallelic HPDL variants treated with ketogenic diet. <i>Molecular Genetics and Metabolism Reports</i> , 2021, 29, 100800.	1.1	4
8	The Onset of Interictal Spike-Related Ripples Facilitates Detection of the Epileptogenic Zone. <i>Frontiers in Neurology</i> , 2021, 12, 724417.	2.4	3
9	Behavioral problems and family distress in tuberous sclerosis complex. <i>Epilepsy and Behavior</i> , 2020, 111, 107321.	1.7	4
10	Long-Term Evaluation of Low-Dose Betamethasone for Ataxia Telangiectasia. <i>Pediatric Neurology</i> , 2019, 100, 60-66.	2.1	6
11	Rett-like features and cortical visual impairment in a Japanese patient with HECW2 mutation. <i>Brain and Development</i> , 2018, 40, 410-414.	1.1	10
12	Attachment Disorder and Early Media Exposure: Neurobehavioral symptoms mimicking autism spectrum disorder. <i>Journal of Medical Investigation</i> , 2018, 65, 280-282.	0.5	17
13	[18F]fluorodeoxyglucose-positron emission tomography study of genetically confirmed patients with Dravet syndrome. <i>Epilepsy Research</i> , 2018, 147, 9-14.	1.6	11
14	Pyridoxal 5â€²-phosphate and related metabolites in hypophosphatasia: Effects of enzyme replacement therapy. <i>Molecular Genetics and Metabolism</i> , 2018, 125, 174-180.	1.1	18
15	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. <i>J Neurol Sci</i> 2015;349:190â€“195. <i>Journal of the Neurological Sciences</i> , 2017, 372, 50.	0.6	0
16	Reversible brain atrophy in glutaric aciduria type 1. <i>Brain and Development</i> , 2017, 39, 532-535.	1.1	7
17	Phenytoin-responsive epileptic encephalopathy with a tandem duplication involving <i>FGF12</i>. <i>Neurology: Genetics</i> , 2017, 3, e133.	1.9	13
18	Mitochondrial Acid 5 (MA-5) Facilitates ATP Synthase Oligomerization and Cell Survival in Various Mitochondrial Diseases. <i>EBioMedicine</i> , 2017, 20, 27-38.	6.1	46

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19	Initial vasodilatation in a child with reversible cerebral vasoconstriction syndrome. <i>Journal of Clinical Neuroscience</i> , 2017, 39, 108-110.	1.5	7
20	Two cases of early-onset myoclonic seizures with continuous parietal delta activity caused by <i>EEF1A2</i> mutations. <i>Brain and Development</i> , 2016, 38, 520-524.	1.1	32
21	Patchy white matter hyperintensity in ring chromosome 18 syndrome. <i>Pediatrics International</i> , 2016, 58, 919-922.	0.5	5
22	Asymptomatic congenital cytomegalovirus infection with neurological sequelae: A retrospective study using umbilical cord. <i>Brain and Development</i> , 2016, 38, 819-826.	1.1	27
23	FDG-PET study of patients with Leigh syndrome. <i>Journal of the Neurological Sciences</i> , 2016, 362, 309-313.	0.6	12
24	Genetic heterogeneity in 26 infants with a hypomyelinating leukodystrophy. <i>Human Genetics</i> , 2016, 135, 89-98.	3.8	26
25	A case of 3p deletion syndrome associated with cerebellar hemangioblastoma. <i>Brain and Development</i> , 2016, 38, 257-260.	1.1	4
26	Novel missense mutation in <i>CLN8</i> in late infantile neuronal ceroid lipofuscinosis: The first report of a <i>CLN8</i> mutation in Japan. <i>Brain and Development</i> , 2016, 38, 341-345.	1.1	7
27	Acute encephalitis with refractory, repetitive partial seizures: Pathological findings and a new therapeutic approach using tacrolimus. <i>Brain and Development</i> , 2016, 38, 772-776.	1.1	22
28	Mucopolipidosis IV: A milder form with novel mutations and serial MRI findings. <i>Brain and Development</i> , 2016, 38, 763-767.	1.1	10
29	Clinical profiles for seizure remission and developmental gains after total corpus callosotomy. <i>Brain and Development</i> , 2016, 38, 47-53.	1.1	24
30	Extremely low-dose vigabatrin for West syndrome with tuberous sclerosis. <i>Journal of Pediatric Epilepsy</i> , 2015, 02, 255-258.	0.2	1
31	<i>GRIN1</i> mutations cause encephalopathy with infantile-onset epilepsy, and hyperkinetic and stereotyped movement disorders. <i>Epilepsia</i> , 2015, 56, 841-848.	5.1	76
32	First Japanese case of Zellweger syndrome with a mutation in <i>PEX14</i> . <i>Pediatrics International</i> , 2015, 57, 1189-1192.	0.5	5
33	Interhemispheric Vertical Hemispherotomy: A Single Center Experience. <i>Pediatric Neurosurgery</i> , 2015, 50, 295-300.	0.7	15
34	Renal function in angiotensinogen gene-mutated renal tubular dysgenesis with glomerular cysts. <i>Pediatric Nephrology</i> , 2015, 30, 357-360.	1.7	4
35	Elevated serum levels of neutrophil elastase in patients with influenza virus-associated encephalopathy. <i>Journal of the Neurological Sciences</i> , 2015, 349, 190-195.	0.6	28
36	Efficacy of long term weekly ACTH therapy for intractable epilepsy. <i>Brain and Development</i> , 2015, 37, 449-454.	1.1	16

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37	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. <i>Brain and Development</i> , 2015, 37, 849-852.	1.1	30
38	A novel PLP1 frameshift mutation causing a milder form of Pelizaeusâ€“Merzbacher disease. <i>Brain and Development</i> , 2015, 37, 455-458.	1.1	5
39	Genomic analysis identifies candidate pathogenic variants in 9 of 18 patients with unexplained West syndrome. <i>Human Genetics</i> , 2015, 134, 649-658.	3.8	51
40	Aquaporin-4 autoimmunity in a child without optic neuritis and myelitis. <i>Brain and Development</i> , 2015, 37, 149-152.	1.1	5
41	Myoclonic axial jerks for diagnosing atypical evolution of ataxia telangiectasia. <i>Brain and Development</i> , 2015, 37, 362-365.	1.1	15
42	Correspondence on â€œClinical Characterization of Gastroenteritis-Related Seizures in Children: Impact of Fever and Serum Sodium Levelsâ€•. <i>Journal of Child Neurology</i> , 2014, 29, 1578-1579.	1.4	0
43	Late vitamin K deficiency bleeding in an infant born at a maternity hospital. <i>Pediatrics International</i> , 2014, 56, 127-128.	0.5	1
44	Pediatric-Onset Extracerebral Stabbing Pain. <i>Journal of Child Neurology</i> , 2014, 29, NP47-NP48.	1.4	1
45	Neonatal â€œSeizureâ€•While Riding in a Car. <i>Clinical Pediatrics</i> , 2014, 53, 605-606.	0.8	1
46	Abdominal and Lower Back Pain in Pediatric Idiopathic Stabbing Headache. <i>Pediatrics</i> , 2014, 133, e245-e247.	2.1	6
47	Early replacement therapy in a first Japanese case with autosomal recessive guanosine triphosphate cyclohydrolase I deficiency with a novel point mutation. <i>Brain and Development</i> , 2014, 36, 268-271.	1.1	7
48	A Case of Acute Cerebellitis With a Unique Sequential Change on Magnetic Resonance Imaging. <i>Pediatric Neurology</i> , 2014, 51, 279-281.	2.1	2
49	RBPJ is disrupted in a case of proximal 4p deletion syndrome with epilepsy. <i>Brain and Development</i> , 2014, 36, 532-536.	1.1	9
50	Effect of a blackout in pediatric patients with home medical devices during the 2011 eastern Japan earthquake. <i>Brain and Development</i> , 2014, 36, 143-147.	1.1	25
51	SLC16A2 mutations in two Japanese patients with Allanâ€“Herndonâ€“Dudley syndrome. <i>Human Genome Variation</i> , 2014, 1, 14010.	0.7	6
52	Parental satisfaction and seizure outcome after corpus callosotomy in patients with infantile or early childhood onset epilepsy. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2013, 22, 303-305.	2.0	37
53	Increased Ki-67 immunoreactivity in the white matter in hemimegalencephaly. <i>Neuroscience Letters</i> , 2013, 548, 244-248.	2.1	4
54	A case of atypical benign partial epilepsy with action myoclonus. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2013, 22, 242-245.	2.0	3

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55	Asymmetric Drug-Induced Tremor: Rare Feature of a Common Event. <i>Pediatric Neurology</i> , 2013, 48, 479-480.	2.1	3
56	The usefulness of subtraction ictal SPECT and ictal near-infrared spectroscopic topography in patients with West syndrome. <i>Brain and Development</i> , 2013, 35, 887-893.	1.1	9
57	Human occipital cortices differentially exert saccadic suppression: Intracranial recording in children. <i>NeuroImage</i> , 2013, 83, 224-236.	4.2	35
58	Periodic Eye Movements and Epileptic Spasms in West Syndrome. <i>Journal of Child Neurology</i> , 2013, 28, 1483-1484.	1.4	0
59	Another Case of Lower Back Pain Associated With Migraine. <i>Journal of Child Neurology</i> , 2013, 28, 680-680.	1.4	5
60	Lower Back Pain as a Symptom of Migrainous Corpalgia. <i>Journal of Child Neurology</i> , 2013, 28, 676-677.	1.4	6
61	Utility of Thallium-201 Scintigraphy in Tolosa-Hunt Syndrome. <i>Tohoku Journal of Experimental Medicine</i> , 2013, 229, 83-86.	1.2	2
62	Brain White Matter Abnormality in a Newborn Infant with Congenital Adrenal Hyperplasia. <i>Clinical Pediatric Endocrinology</i> , 2013, 22, 77-81.	0.8	4
63	Complete remission of seizures after corpus callosotomy. <i>Journal of Neurosurgery: Pediatrics</i> , 2012, 10, 7-13.	1.3	35
64	Clinical Reasoning: A young man with progressive subcortical lesions and optic nerve atrophy. <i>Neurology</i> , 2012, 79, e63-8.	1.1	6
65	Ketotifen overdose in infancy associated with development of epilepsy and mild mental retardation. <i>Pediatrics International</i> , 2012, 54, 963-963.	0.5	2
66	8p deletion and 9p duplication in two children with electrical status epilepticus in sleep syndrome. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2012, 21, 295-299.	2.0	8
67	Hypoperfusion in caudate nuclei in patients with brain "lung" thyroid syndrome. <i>Journal of the Neurological Sciences</i> , 2012, 315, 77-81.	0.6	10
68	Abdominal migraine reviewed from both central and peripheral aspects. <i>World Journal of Experimental Medicine</i> , 2012, 2, 75.	1.7	7
69	Acute encephalopathy in children with Dravet syndrome. <i>Epilepsia</i> , 2012, 53, 79-86.	5.1	53
70	Successful cord blood transplantation with reduced-intensity conditioning for childhood cerebral X-linked adrenoleukodystrophy at advanced and early stages. <i>Pediatric Transplantation</i> , 2012, 16, E63-70.	1.0	13
71	Unusual ribbon-like periventricular heterotopia with congenital cataracts in a Japanese girl. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 674-677.	1.2	1
72	A case with central and peripheral hypomyelination with hypogonadotropic hypogonadism and hypodontia (4H syndrome) plus cataract. <i>Journal of the Neurological Sciences</i> , 2011, 300, 179-181.	0.6	15

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73	Lateralization of interictal spikes after corpus callosotomy. <i>Clinical Neurophysiology</i> , 2011, 122, 2121-2127.	1.5	17
74	A female case of West syndrome with remission of spasms following multiple cerebral hemorrhages. <i>Brain and Development</i> , 2011, 33, 678-682.	1.1	1
75	Abdominal Migraine Associated with Ecchymosis of the Legs and Buttocks: Does the Symptom Imply an Unknown Mechanism of Migraine?. <i>Tohoku Journal of Experimental Medicine</i> , 2010, 221, 49-51.	1.2	10
76	Deletions of SCN1A 5â€² genomic region with promoter activity in Dravet syndrome. <i>Human Mutation</i> , 2010, 31, 820-829.	2.5	47
77	Unique discrepancy between cerebral blood flow and glucose metabolism in hemimegalencephaly. <i>Epilepsy Research</i> , 2010, 92, 201-208.	1.6	17
78	Functional cortical deafferentation from the subcortical structures in a patient with early myoclonic encephalopathy: A functional neuroimaging study. <i>Epilepsia</i> , 2010, 51, 699-702.	5.1	12
79	Efficacy of Sumatriptan in Two Pediatric Cases With Abdominal Pain-Related Functional Gastrointestinal Disorders: Does the Mechanism Overlap That of Migraine?. <i>Journal of Child Neurology</i> , 2010, 25, 234-237.	1.4	33
80	Elevated plasma levels of tissue inhibitors of metalloproteinase-1 and their overexpression in muscle in human and mouse muscular dystrophy. <i>Journal of the Neurological Sciences</i> , 2010, 297, 19-28.	0.6	27
81	Restless legs syndrome: A unique case of relapse after traffic accidents with a long remission. <i>Sleep Medicine</i> , 2010, 11, 596-597.	1.6	0
82	Ictal Vomiting as an Initial Symptom of Severe Myoclonic Epilepsy in Infancy: A Case Report. <i>Journal of Child Neurology</i> , 2009, 24, 228-230.	1.4	1
83	Successful Treatment With Sumatriptan in a Case With Cyclic Vomiting Syndrome Combined With 18qâ€² Syndrome. <i>Journal of Child Neurology</i> , 2009, 24, 1561-1563.	1.4	12
84	A further case of renal tubular dysgenesis surviving the neonatal period. <i>European Journal of Pediatrics</i> , 2009, 168, 207-209.	2.7	28
85	Utility of subtraction ictal SPECT images in detecting focal leading activity and understanding the pathophysiology of spasms in patients with West syndrome. <i>Epilepsy Research</i> , 2009, 83, 177-183.	1.6	18
86	Reduced levels of interleukin-1 receptor antagonist in the cerebrospinal fluid in patients with West syndrome. <i>Epilepsy Research</i> , 2009, 85, 314-317.	1.6	32
87	Neuroepidemiology of West syndrome and early infantile epileptic encephalopathy in Miyagi Prefecture, Japan. <i>Epilepsy Research</i> , 2009, 87, 299-301.	1.6	11
88	Efficacy of idebenone for respiratory failure in a patient with Leigh syndrome: A long-term follow-up study. <i>Journal of the Neurological Sciences</i> , 2009, 278, 112-114.	0.6	28
89	Intramuscular renin-angiotensin system is activated in human muscular dystrophy. <i>Journal of the Neurological Sciences</i> , 2009, 280, 40-48.	0.6	45
90	Neonatal-Onset Brainstem Reticular Reflex Myoclonus Following a Prenatal Brain Insult: Generalized Myoclonic Jerk and a Brainstem Lesion. <i>Tohoku Journal of Experimental Medicine</i> , 2007, 211, 303-308.	1.2	6

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91	The Cerebrospinal Fluid Level of 5-Methyltetrahydrofolate in a Japanese Boy with Hypomyelination with Atrophy of the Basal Ganglia and Cerebellum. <i>Tohoku Journal of Experimental Medicine</i> , 2007, 213, 373.	1.2	3
92	Novel mutations in five Japanese patients with 3-methylcrotonyl-CoA carboxylase deficiency. <i>Journal of Human Genetics</i> , 2007, 52, 1040-1043.	2.3	18
93	A case surviving for over a year of renal tubular dysgenesis with compound heterozygous angiotensinogen gene mutations. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 2355-2360.	1.2	26