# Satoru Takahashi

List of Publications by Year
in descending order



| 43 | 596 | 759233 12 | 677142 22 |
|---|---|---|---|
| papers | citations | h-index | g-index |

| 44 | 44 | 44 | 986 |
|---|---|---|---|
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Severity of low pre-pregnancy body mass index and perinatal outcomes: the Japan Environment and Childrenâ€™s Study. BMC Pregnancy and Childbirth, 2022, 22, 121. | 2.4 | 10 |
| 2 | A Complex Phenotype of a Patient with Spastic Paraplegia Type 4 Caused by a Novel Pathogenic Variant in the <b><i>SPAST</i></b> Gene. Case Reports in Neurology, 2022, 13, 763-771. | 0.7 | 3 |
| 3 | Novel NARS2 variant causing leigh syndrome with normal lactate levels. Human Genome Variation, 2022, 9, 12. | 0.7 | 8 |
| 4 | A generic assay for the identification of splicing variants that induce nonsense-mediated decay in Pompe disease. European Journal of Human Genetics, 2021, 29, 422-433. | 2.8 | 6 |
| 5 | Early diagnosis of MECP2 duplication syndrome: Insights from a nationwide survey in Japan. Journal of the Neurological Sciences, 2021, 422, 117321. | 0.6 | 6 |
| 6 | Profiles of 5Î±-Reduced Androgens in Humans and Eels: 5Î±-Dihydrotestosterone and 11-Ketodihydrotestosterone Are Active Androgens Produced in Eel Gonads. Frontiers in Endocrinology, 2021, 12, 657360. | 3.5 | 9 |
| 7 | Stereotyped Upper Limb Movement in MECP2 Duplication Syndrome. Neurology, 2021, 97, 92-94. | 1.1 | 1 |
| 8 | 11-Ketotestosterone is a major androgen produced in porcine adrenal glands and testes. Journal of Steroid Biochemistry and Molecular Biology, 2021, 210, 105847. | 2.5 | 12 |
| 9 | Analyses of Molecular Characteristics and Enzymatic Activities of Ovine HSD17B3. Animals, 2021, 11, 2876. | 2.3 | 2 |
| 10 | Life-threatening muscle complications of COL4A1-related disorder. Brain and Development, 2020, 42, 93-97. | 1.1 | 3 |
| 11 | Evaluation of 17Î²-hydroxysteroid dehydrogenase activity using androgen receptor-mediated transactivation. Journal of Steroid Biochemistry and Molecular Biology, 2020, 196, 105493. | 2.5 | 20 |
| 12 | MeCP2_e2 partially compensates for lack of MeCP2_e1: A male case of Rett syndrome. Molecular Genetics & Genomic Medicine, 2020, 8, e1088. | 1.2 | 12 |
| 13 | The role of molecular analysis of SLC2A1 in the diagnostic workup of glucose transporter 1 deficiency syndrome. Journal of the Neurological Sciences, 2020, 416, 117041. | 0.6 | 1 |
| 14 | Atypical Rett syndrome in a girl with mosaic triple X and MECP2 variant. Molecular Genetics & Genomic Medicine, 2020, 8, e1122. | 1.2 | 6 |
| 15 | PRRT2 mutations in Japanese patients with benign infantile epilepsy and paroxysmal kinesigenic dyskinesia. Seizure: the Journal of the British Epilepsy Association, 2019, 71, 1-5. | 2.0 | 16 |
| 16 | Cyclooxygenaseâ€2 is acutely induced by CCAAT/enhancerâ€binding protein Î² to produce prostaglandin E 2 and F 2Î± following gonadotropin stimulation in Leydig cells. Molecular Reproduction and Development, 2019, 86, 786-797. | 2.0 | 7 |
| 17 | Genetic landscape of Rett syndrome-like phenotypes revealed by whole exome sequencing. Journal of Medical Genetics, 2019, 56, 396-407. | 3.2 | 30 |
| 18 | A novel mutation in slc2a1 gene causing glut-1 deficiency syndrome in a young adult patient. Turkish Journal of Pediatrics, 2019, 61, 946. | 0.6 | 2 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | MECP2 mutation in a boy with severe apnea and sick sinus syndrome. Brain and Development, 2018, 40, 714-718. | 1.1 | 7 |
| 20 | Hypoplastic hippocampus in atypical Rett syndrome with a novel FOXG1 mutation. Brain and Development, 2018, 40, 49-52. | 1.1 | 6 |
| 21 | Successful treatment of normokalemic periodic paralysis with hydrochlorothiazide. Brain and Development, 2018, 40, 833-836. | 1.1 | 5 |
| 22 | Evolution into moyamoya disease in an infant with internal carotid artery aneurysms. ENeurologicalSci, 2017, 6, 80-82. | 1.3 | 2 |
| 23 | Diethylstilbestrol administration inhibits theca cell androgen and granulosa cell estrogen production in immature rat ovary. Scientific Reports, 2017, 7, 8374. | 3.3 | 15 |
| 24 | Congenital basal meningoceles with different outcomes: a case series. Journal of Medical Case Reports, 2017, 11, 359. | 0.8 | 4 |
| 25 | Outcome of ketogenic diets in GLUT1 deficiency syndrome in Japan: A nationwide survey. Brain and Development, 2016, 38, 628-637. | 1.1 | 31 |
| 26 | Characterization of intragenic tandem duplication in the PAFAH1B1 gene leading to isolated lissencephaly sequence. Molecular Cytogenetics, 2015, 8, 84. | 0.9 | 2 |
| 27 | Electroclinical features of epileptic encephalopathy caused by <scp>SCN8A</scp> mutation. Pediatrics International, 2015, 57, 758-762. | 0.5 | 13 |
| 28 | Human herpesvirus-6 infection-associated acute encephalopathy without skin rash. Brain and Development, 2015, 37, 829-832. | 1.1 | 6 |
| 29 | Focal frontal epileptiform discharges in a patient with eyelid myoclonia and absence seizures. Epilepsy & Behavior Case Reports, 2015, 4, 35-37. | 1.5 | 5 |
| 30 | Nationwide survey of glucose transporter-1 deficiency syndrome (GLUT-1DS) in Japan. Brain and Development, 2015, 37, 780-789. | 1.1 | 35 |
| 31 | Improved prefrontal activity in AD/HD children treated with atomoxetine: A NIRS study. Brain and Development, 2015, 37, 76-87. | 1.1 | 34 |
| 32 | Relation between circulating levels of GH, IGF-1, ghrelin and somatic growth in Rett syndrome. Brain and Development, 2014, 36, 794-800. | 1.1 | 12 |
| 33 | Benign infantile convulsion as a diagnostic clue of paroxysmal kinesigenic dyskinesia: a case series. Journal of Medical Case Reports, 2014, 8, 174. | 0.8 | 4 |
| 34 | Early onset epileptic encephalopathy caused by de novo <scp>SCN</scp>8A mutations. Epilepsia, 2014, 55, 994-1000. | 5.1 | 142 |
| 35 | A haploinsufficiency of FOXG1 identified in a boy with congenital variant of Rett syndrome. Brain and Development, 2014, 36, 725-729. | 1.1 | 14 |
| 36 | Reversible White Matter Lesions During Ketogenic Diet Therapy in Glucose Transporter 1 Deficiency Syndrome. Pediatric Neurology, 2013, 49, 493-496. | 2.1 | 8 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | Oxidative stress markers and phosphorus magnetic resonance spectroscopy in a patient with GLUT1 deficiency treated with modified Atkins diet. Brain and Development, 2012, 34, 372-375. | 1.1 | 8 |
| 38 | Takahashiâ€™s response to Ravnâ€™s correspondence. Clinical Genetics, 2009, 75, 100-100. | 2.0 | 0 |
| 39 | Molecular analysis and anticonvulsant therapy in two patients with glucose transporter 1 deficiency syndrome: A successful use of zonisamide for controlling the seizures. Epilepsy Research, 2008, 80, 18-22. | 1.6 | 11 |
| 40 | Pyruvate dehydrogenase complex deficiency with multiple minor anomalies. Pediatrics International, 1997, 39, 230-232. | 0.5 | 5 |
| 41 | A novel mutation in LICAM gene in a Japanese patient with x-linked hydrocephalus. Japanese Journal of Human Genetics, 1996, 41, 431-437. | 0.8 | 7 |
| 42 | Ictal HMPAO-Single Photon Emission Computed Tomography Findings in Reading Epilepsy in a Japanese Boy. Epilepsia, 1995, 36, 1161-1163. | 5.1 | 17 |
| 43 | Serial neuroimages of acute necrotizing encephalopathy associated with human herpesvirus 6 infection. Brain and Development, 1995, 17, 356-359. | 1.1 | 49 |