Kiyoshi Hayasaka

List of Publications by Year
in descending order

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116	5,732	94433	82547
papers	citations	37 h-index	72 g-index

116	116	116	5945
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	De novo mutations in the gene encoding STXBP1 (MUNC18-1) cause early infantile epileptic encephalopathy. Nature Genetics, 2008, 40, 782-788.	21.4	498
2	Charcotâ€“Marieâ€“Tooth neuropathy type 1B is associated with mutations of the myelin P0 gene. Nature Genetics, 1993, 5, 31-34.	21.4	391
3	Mitochondrial GTPase mitofusin 2 mutation in Charcot?Marie?Tooth neuropathy type 2A. Human Genetics, 2005, 116, 23-27.	3.8	229
4	De novo mutation of the myelin Po gene in Dejerineâ€“Sottas disease (hereditary motor and sensory) Tj ETQq0 0 0 rgBT /Overlock 10 Tf	21.4	227
5	Demyelinating and axonal features of Charcot-Marie-Tooth disease with mutations of myelin-related proteins (PMP22, MPZ and Cx32): a clinicopathological study of 205 Japanese patients. Brain, 2003, 126, 134-151.	7.6	202
6	Clinical spectrum of early onset epileptic encephalopathies caused by <scp>KCNQ2</scp> mutation. Epilepsia, 2013, 54, 1282-1287.	5.1	195
7	Clinical spectrum of SCN2A mutations expanding to Ohtahara syndrome. Neurology, 2013, 81, 992-998.	1.1	188
8	De Novo Mutations in GNAO1, Encoding a GÎ±o Subunit of Heterotrimeric G Proteins, Cause Epileptic Encephalopathy. American Journal of Human Genetics, 2013, 93, 496-505.	6.2	187
9	Molecular analysis of congenital central hypoventilation syndrome. Human Genetics, 2003, 114, 22-26.	3.8	174
10	A Longer Polyalanine Expansion Mutation in the ARX Gene Causes Early Infantile Epileptic Encephalopathy with Suppression-Burst Pattern (Ohtahara Syndrome). American Journal of Human Genetics, 2007, 81, 361-366.	6.2	168
11	Elevated expression of messenger RNA for peripheral myelin protein 22 in biopsied peripheral nerves of patients with Charcot-Marie-Tooth disease type 1A. Annals of Neurology, 1994, 35, 445-450.	5.3	145
12	STXBP1 mutations in early infantile epileptic encephalopathy with suppressionâ€burst pattern. Epilepsia, 2010, 51, 2397-2405.	5.1	133
13	Mutation of the myelin Po gene in Charcot â€" Marie â€" Tooth neuropathy type 1B. Human Molecular Genetics, 1993, 2, 1369-1372.	2.9	101
14	Cardiac Ion Channel Gene Mutations in Sudden Infant Death Syndrome. Pediatric Research, 2008, 64, 482-487.	2.3	95
15	PIGA mutations cause early-onset epileptic encephalopathies and distinctive features. Neurology, 2014, 82, 1587-1596.	1.1	93
16	Whole exome sequencing identifies KCNQ2 mutations in Ohtahara syndrome. Annals of Neurology, 2012, 72, 298-300.	5.3	88
17	Small heat shock protein 27 mutation in a Japanese patient with distal hereditary motor neuropathy. Journal of Human Genetics, 2005, 50, 473-476.	2.3	85
18	De Novo Mutations inSLC35A2Encoding a UDP-Galactose Transporter Cause Early-Onset Epileptic Encephalopathy. Human Mutation, 2013, 34, 1708-1714.	2.5	85

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19	Structure and Chromosomal Localization of the Gene Encoding the Human Myelin Protein Zero (MPZ). Genomics, 1993, 17, 755-758.	2.9	84
20	Neonatal hyperbilirubinemia and mutation of the bilirubin uridine diphosphate-glucuronosyltransferase gene: a common missense mutation among Japanese, Koreans and Chinese. IUBMB Life, 1998, 46, 21-26.	3.4	84
21	Isolation and sequence determination of cDNA encoding the major structural protein of human peripheral myelin. Biochemical and Biophysical Research Communications, 1991, 180, 515-518.	2.1	83
22	Neonatal hyperbilirubinemia and a common mutation of the bilirubin uridine diphosphate-glucuronosyltransferase gene in Japanese. Journal of Human Genetics, 1999, 44, 22-25.	2.3	83
23	Neurofilament light chain polypeptide gene mutations in Charcotâ€“Marieâ€“Tooth disease: nonsense mutation probably causes a recessive phenotype. Journal of Human Genetics, 2009, 54, 94-97.	2.3	83
24	Molecular diagnosis and clinical onset of Charcotâ€“Marieâ€“Tooth disease in Japan. Journal of Human Genetics, 2011, 56, 364-368.	2.3	66
25	CASK aberrations in male patients with Ohtahara syndrome and cerebellar hypoplasia. Epilepsia, 2012, 53, 1441-1449.	5.1	66
26	A Mutation of COX6A1 Causes a Recessive Axonal or Mixed Form of Charcot-Marie-Tooth Disease. American Journal of Human Genetics, 2014, 95, 294-300.	6.2	65
27	Nonketotic hyperglycinemia: Analyses of glycine cleavage system in typical and atypical cases. Journal of Pediatrics, 1987, 110, 873-877.	1.8	59
28	Molecular analysis in Japanese patients with Charcot-Marie-Tooth disease: DGGE analysis for PMP22, MPZ, and Cx32/GJB1 mutations. Human Mutation, 2002, 20, 392-398.	2.5	57
29	A clinical phenotype of distal hereditary motor neuronopathy type II with a novel HSPB1 mutation. Journal of the Neurological Sciences, 2009, 277, 9-12.	0.6	54
30	A Novel Homozygous Mutation of the Myelin Po Gene Producing Dejerineâ€“Sottas Disease (Hereditary) Tj ETQq0 0 0 rgBT /Overlock 10 222, 107-110.	2.1	49
31	Hemizygous mutation of the peripheral myelin protein 22 gene associated with Charcot-Marie-Tooth disease type 1. Annals of Neurology, 2000, 47, 101-103.	5.3	48
32	Mutation of the doublecortin gene in male patients with double cortex syndrome: Somatic mosaicism detected by hair root analysis. Annals of Neurology, 2001, 50, 547-551.	5.3	47
33	Genotypeâ€“phenotype relationship in Japanese patients with congenital central hypoventilation syndrome. Journal of Human Genetics, 2015, 60, 473-477.	2.3	43
34	Isolation and sequence determination of cDNA encoding PMP-22 (PAS-II/SR13/Gas-3) of human peripheral myelin. Biochemical and Biophysical Research Communications, 1992, 186, 827-831.	2.1	41
35	Periaxin mutation causes early-onset but slow-progressive Charcot-Marie-Tooth disease. Journal of Human Genetics, 2004, 49, 376-379.	2.3	41
36	Unusual circadian locomotor activity and pathophysiology in mutant CRY1 transgenic mice. Neuroscience Letters, 2009, 451, 246-251.	2.1	40

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37	Identification of the mutations in the T-protein gene causing typical and atypical nonketotic hyperglycinemia. Human Genetics, 1994, 93, 655-8.	3.8	39
38	Association of breast-fed neonatal hyperbilirubinemia with UGT1A1 polymorphisms: 211G>A (G71R) mutation becomes a risk factor under inadequate feeding. Journal of Human Genetics, 2013, 58, 7-10.	2.3	39
39	Sudden Infant Death Syndrome Is Not Associated with the Mutation of PHOX2B Gene, a Major Causative Gene of Congenital Central Hypoventilation Syndrome. Tohoku Journal of Experimental Medicine, 2004, 203, 65-68.	1.2	37
40	Ankyrin-G Regulates Inactivation Gating of the Neuronal Sodium Channel, Nav1.6. Journal of Neurophysiology, 2006, 96, 1347-1357.	1.8	36
41	Neonatal Hyperbilirubinemia in Japanese Neonates: Analysis of the Heme Oxygenase-1 Gene and Fetal Hemoglobin Composition in Cord Blood. Pediatric Research, 2003, 54, 165-171.	2.3	34
42	Treatment with Lactose (Galactose)-Restricted and Medium-Chain Triglyceride-Supplemented Formula for Neonatal Intrahepatic Cholestasis Caused by Citrin Deficiency. JIMD Reports, 2011, 2, 37-44.	1.5	34
43	Medium-chain triglyceride supplementation under a low-carbohydrate formula is a promising therapy for adult-onset type II citrullinemia. Molecular Genetics and Metabolism Reports, 2014, 1, 42-50.	1.1	33
44	Locations of crossover breakpoints within the CMT1A-REP repeat in Japanese patients with CMT1A and HNPP. Human Genetics, 1997, 99, 151-154.	3.8	32
45	Frameshift mutations of the ARX gene in familial Ohtahara syndrome. Epilepsia, 2010, 51, 1679-1684.	5.1	30
46	Supernumerary impacted teeth in a patient with SOX2 anophthalmia syndrome. American Journal of Medical Genetics, Part A, 2010, 152A, 2355-2359.	1.2	29
47	Phenotype-genotype correlations of PIGO deficiency with variable phenotypes from infantile lethality to mild learning difficulties. Human Mutation, 2017, 38, 805-815.	2.5	29
48	A novel mutation of the doublecortin gene in Japanese patients with X-linked lissencephaly and subcortical band heterotopia. Human Genetics, 1999, 104, 341-344.	3.8	27
49	Mediumâ€chain triglycerides supplement therapy with a lowâ€carbohydrate formula can supply energy and enhance ammonia detoxification in the hepatocytes of patients with adultâ€“onset type II citrullinemia. Journal of Inherited Metabolic Disease, 2018, 41, 777-784.	3.6	27
50	Metabolic basis and treatment of citrin deficiency. Journal of Inherited Metabolic Disease, 2021, 44, 110-117.	3.6	27
51	Structure and Localization of the Gene Encoding Human Peripheral Myelin Protein 2 (PMP2). Genomics, 1993, 18, 244-248.	2.9	26
52	Genetic Analysis of Shwachman-Diamond Syndrome: Phenotypic Heterogeneity in Patients Carrying Identical SBDS Mutations. Tohoku Journal of Experimental Medicine, 2005, 206, 253-259.	1.2	26
53	Improvement of nephrotic syndrome by intensive lipid-lowering therapy in a patient with lipoprotein glomerulopathy. Clinical and Experimental Nephrology, 2009, 13, 659-662.	1.6	26
54	De novo 5q14.3 translocation 121.5â€kb upstream of MEF2C in a patient with severe intellectual disability and earlyâ€onset epileptic encephalopathy. American Journal of Medical Genetics, Part A, 2011, 155, 2879-2884.	1.2	26

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55	Adult-onset type II citrullinemia: Current insights and therapy. The Application of Clinical Genetics, 2018, Volume 11, 163-170.	3.0	25
56	Feasibility of prenatal diagnosis of nonketotic hyperglycinemia: Existence of the glycine cleavage system in placenta. Journal of Pediatrics, 1987, 110, 124-126.	1.8	24
57	Deletion and Nonsense Mutations of the Connexin 32 Gene Associated with Charcot-Marie-Tooth Disease.. Tohoku Journal of Experimental Medicine, 1999, 188, 239-244.	1.2	24
58	Congenital Central Hypoventilation Syndrome: A Novel Mutation of the RET Gene in an Isolated Case.. Tohoku Journal of Experimental Medicine, 2002, 196, 241-246.	1.2	24
59	Screening of the early growth response 2 gene in Japanese patients with Charcotâ€“Marieâ€“Tooth disease type 1. Journal of the Neurological Sciences, 2003, 210, 61-64.	0.6	24
60	Inheritance of polyalanine expansion mutation of PHOX2B in congenital central hypoventilation syndrome. Journal of Human Genetics, 2012, 57, 335-337.	2.3	23
61	Structure and Chromosomal Localization of the Aminomethyltransferase Gene (AMT). Genomics, 1994, 19, 27-30.	2.9	22
62	Association of the uteroglobin gene polymorphism with IgA nephropathy. American Journal of Kidney Diseases, 2002, 39, 36-41.	1.9	22
63	Neonatal hyperbilirubinemia and the bilirubin uridine diphosphate-glucuronosyltransferase gene: The common â^'3263Tâ€ƒ›â€ƒG mutation of phenobarbital response enhancer module is not associated with the neonatal hyperbilirubinemia in Japanese. Pediatrics International, 2005, 47, 137-141.	0.5	22
64	The GARS gene is rarely mutated in Japanese patients with Charcotâ€“Marieâ€“Tooth neuropathy. Journal of Human Genetics, 2009, 54, 310-312.	2.3	22
65	Clustering of CMT1A duplication breakpoints in a 700 bp interval of the CMT1A-REP repeat. Human Mutation, 1998, 11, 109-113.	2.5	21
66	Analysis of the genes responsible for steroid-resistant nephrotic syndrome and/or focal segmental glomerulosclerosis in Japanese patients by whole-exome sequencing analysis. Journal of Human Genetics, 2016, 61, 137-141.	2.3	21
67	Defective Signal Transduction through the Thromboxane A2 Receptor in a Patient with a Miid Bleeding Disorder: Deficiency of the Inositol 1,4,5-Triphosphate Formation despite Normal G-protein Activation. Thrombosis and Haemostasis, 1997, 77, 0991-0995.	3.4	20
68	Isolation and sequence determination of cDNA encoding P2 protein of human peripheral myelin. Biochemical and Biophysical Research Communications, 1991, 181, 204-207.	2.1	19
69	Diffuse pachygyria with cerebellar hypoplasia: A milder form of microlissencephaly or a new genetic syndrome?. Annals of Neurology, 1999, 46, 660-663.	5.3	19
70	Mild phenotype of Charcotâ€“Marieâ€“Tooth disease type 4B1. Journal of the Neurological Sciences, 2013, 334, 176-179.	0.6	19
71	INF2 mutations in Charcotâ€Marieâ€Tooth disease complicated with focal segmental glomerulosclerosis. Journal of the Peripheral Nervous System, 2013, 18, 97-98.	3.1	19
72	GAPO syndrome: Report on the first case in Japan. American Journal of Medical Genetics Part A, 1995, 58, 257-261.	2.4	18

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73	Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes (MELAS) decrease in diastolic left ventricular function assessed by echocardiography. Pediatric Cardiology, 1993, 14, 162-166.	1.3	17
74	Four novel mutations of the connexin 32 gene in four Japanese families with Charcot-Marie-Tooth disease type 1. , 1998, 80, 352-355.		16
75	Molecular analysis of the genes causing recessive demyelinating Charcotâ€“Marieâ€“Tooth disease in Japan. Journal of Human Genetics, 2013, 58, 273-278.	2.3	16
76	Sensitive Detection of Polyalanine Expansions in PHOX2B by Polymerase Chain Reaction Using Bisulfite-Converted DNA. Journal of Molecular Diagnostics, 2005, 7, 638-640.	2.8	15
77	De novo polyalanine expansion of PHOX2B in congenital central hypoventilation syndrome: unequal sister chromatid exchange during paternal gametogenesis. Journal of Human Genetics, 2007, 52, 921-925.	2.3	15
78	Phenotypic variability in a family with Townesâ€“Brocks syndrome. Journal of Human Genetics, 2010, 55, 550-551.	2.3	15
79	Compound heterozygous PMP22 deletion mutations causing severe Charcotâ€“Marieâ€“Tooth disease type 1. Journal of Human Genetics, 2010, 55, 771-773.	2.3	15
80	A founder haplotype of APOE-Sendai mutation associated with lipoprotein glomerulopathy. Journal of Human Genetics, 2013, 58, 254-258.	2.3	15
81	A Case of Congenital Central Hypoventilation Syndrome with a Novel Mutation of the PHOX2B Gene Presenting as Central Sleep Apnea. Journal of Clinical Sleep Medicine, 2014, 10, 327-329.	2.6	15
82	Association of neonatal hyperbilirubinemia in breast-fed infants with UGT1A1 or SLCOs polymorphisms. Journal of Human Genetics, 2015, 60, 35-40.	2.3	15
83	Neural Cell Adhesion Proteins and Neurological Diseases1. Journal of Biochemistry, 1994, 116, 1187-1192.	1.7	14
84	A new mutation of the Po gene in patients with Charcot-Marie-Tooth disease type 1B: screening of the Po gene by heteroduplex analysis. Neuroscience Letters, 1996, 204, 173-176.	2.1	14
85	Periaxin mutation in Japanese patients with Charcot-Marie-Tooth disease. Journal of Human Genetics, 2006, 51, 625-628.	2.3	14
86	Growth impairment in individuals with citrin deficiency. Journal of Inherited Metabolic Disease, 2019, 42, 501-508.	3.6	14
87	Severe Bleeding Tendency in a Patient With Bernard-Soulier Syndrome Associated With a Homozygous Single Base Pair Deletion in the Gene Coding for the Human Platelet Glycoprotein IbÎ±. Journal of Pediatric Hematology/Oncology, 1998, 20, 246-251.	0.6	13
88	Novel mutation of the myelin Po gene in a pedigree with Charcot-Marie-Tooth disease type 1b. , 1997, 71, 246-248.		12
89	Nonâ€obese early onset diabetes mellitus in mutant cryptochrome1 transgenic mice. European Journal of Clinical Investigation, 2010, 40, 1011-1017.	3.4	12
90	Congenital hypomyelination neuropathy: decreased expression of the P2 protein in peripheral nerve with normal DNA sequence of the coding region. Journal of the Neurological Sciences, 1995, 134, 150-159.	0.6	11

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91	Polymorphisms of Heme Oxygenase-1 and Bilirubin UDP-Glucuronosyltransferase Genes are not Associated with Kawasaki Disease Susceptibility. Tohoku Journal of Experimental Medicine, 2003, 200, 155-159.	1.2	11
92	The Effect of Carnitine on the Metabolism of Valproic Acid in Epileptic Patients.. Tohoku Journal of Experimental Medicine, 1992, 167, 89-92.	1.2	10
93	Facilitated diagnosis of CMT1A duplication in chromosome 17p11.2-12: Analysis with a CMT1A-REP repeat probe and photostimulated luminescence imaging. , 1997, 9, 563-566.		10
94	InÂVivo Expression of NUP93 and Its Alteration by NUP93 Mutations Causing Focal Segmental Glomerulosclerosis. Kidney International Reports, 2019, 4, 1312-1322.	0.8	10
95	Adult cases of late-onset congenital central hypoventilation syndrome and paired-like homeobox 2B-mutation carriers: an additional case report and pooled analysis. Journal of Clinical Sleep Medicine, 2020, 16, 1891-1900.	2.6	10
96	Expression of Po protein in sural nerve of a patient with hereditary motor and sensory neuropathy type III. Journal of the Neurological Sciences, 1994, 124, 67-70.	0.6	9
97	Effectiveness of Medium-Chain Triglyceride Oil Therapy in Two Japanese Citrin-Deficient Siblings: Evaluation Using Oral Glucose Tolerance Tests. Tohoku Journal of Experimental Medicine, 2016, 240, 323-328.	1.2	9
98	Localization of PMP-22 gene (candidate gene for the Charcot-Marie-Tooth disease 1a) to band 17p11.2 by direct r-banding fluoreschenceIn situ hybridization. Japanese Journal of Human Genetics, 1992, 37, 303-306.	0.8	7
99	De novo mutation of the myelin Po gene in DÃ©jÃ©rine-Sottas disease (hereditary motor and sensory) Tj ETQq1 1 0.784314 rgBT /O	2.5	7
100	Polyalanine expansion of PHOX2B in congenital central hypoventilation syndrome: rs17884724:A>C is associated with 7-alanine expansion. Journal of Human Genetics, 2010, 55, 4-7.	2.3	7
101	NovelPHOX2Bmutations in congenital central hypoventilation syndrome. Pediatrics International, 2019, 61, 393-396.	0.5	7
102	Two homozygous cases of erythrocyte pyruvate kinase (PK) deficiency in Japan: PK sendai and PK shinshu. American Journal of Hematology, 1988, 28, 186-190.	4.1	6
103	Cross-talk between ?1-adrenoceptors and ETA receptors in modulation of the slow component of delayed rectifier K+ currents. Naunyn-Schmiedeberg's Archives of Pharmacology, 2005, 371, 133-140.	3.0	6
104	A case of progressive familial intrahepatic cholestasis type 1 with compound heterozygous mutations of ATP8B1. Pediatrics International, 2011, 53, 107-110.	0.5	6
105	The first Japanese case of Charcotâ€“Marieâ€“Tooth disease type 4H with a novel FGD4 c.837-1G>A mutation. Neuromuscular Disorders, 2013, 23, 652-655.	0.6	6
106	Characterization of ageâ€associated alterations of islet function and structure in diabetic mutant cryptochromeÂ1 transgenic mice. Journal of Diabetes Investigation, 2013, 4, 428-435.	2.4	6
107	A novel PHOX2B gene mutation in an extremely low birth weight infant with congenital central hypoventilation syndrome and variant Hirschsprung's disease. European Journal of Medical Genetics, 2019, 62, 103541.	1.3	5
108	ALOX12 mutation in a family with dominantly inherited bleeding diathesis. Journal of Human Genetics, 2021, 66, 753-759.	2.3	5

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109	A novel ETFB mutation in a patient with glutaric aciduria type II. Human Genome Variation, 2015, 2, 15016.	0.7	4
110	Diabetes mellitus exacerbates citrin deficiency via glucose toxicity. Diabetes Research and Clinical Practice, 2020, 164, 108159.	2.8	4
111	Unique food-entrained circadian rhythm in cysteine414-alanine mutant mCRY1 transgenic mice. Sleep and Biological Rhythms, 2016, 14, 261-269.	1.0	3
112	Slowly progressive sleep apnea in lateâ€onset central hypoventilation syndrome. Pediatrics International, 2012, 54, 290-292.	0.5	2
113	A T-to-C substitution at nucleotide 12311 in tRNALeu(CUN) gene may be a mtDNA polymorphism. Journal of the Neurological Sciences, 1994, 127, 236.	0.6	1
114	A second pediatric patient with lipoprotein glomerulopathy carrying a heterozygous APOE-Sendai mutation. Japanese Journal of Pediatric Nephrology, 2011, 24, 218-223.	0.0	1
115	Isolation and sequence determination of cDNA encoding mouse rab 4 and candidate approach for the beige mutation in mice. IUBMB Life, 1996, 40, 647-651.	3.4	0
116	Heterozygous calcyclin-binding protein/Siah1-interacting protein (CACYBP/SIP) gene pathogenic variant linked to a dominant family with paucity of interlobular bile duct. Journal of Human Genetics, 2022, , .	2.3	0