

# Kiyoshi Hayasaka

## List of Publications by Year in descending order

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116  
papers

5,732  
citations

94269

37  
h-index

82410

72  
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116  
all docs

116  
docs citations

116  
times ranked

5945  
citing authors

#	ARTICLE	IF	CITATIONS
1	Heterozygous calcyclin-binding protein/Siah1-interacting protein (CACYPB/SIP) gene pathogenic variant linked to a dominant family with paucity of interlobular bile duct. <i>Journal of Human Genetics</i> , 2022, , .	1.1	0
2	Metabolic basis and treatment of citrin deficiency. <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 110-117.	1.7	27
3	ALOX12 mutation in a family with dominantly inherited bleeding diathesis. <i>Journal of Human Genetics</i> , 2021, 66, 753-759.	1.1	5
4	Diabetes mellitus exacerbates citrin deficiency via glucose toxicity. <i>Diabetes Research and Clinical Practice</i> , 2020, 164, 108159.	1.1	4
5	Adult cases of late-onset congenital central hypoventilation syndrome and paired-like homeobox 2B-mutation carriers: an additional case report and pooled analysis. <i>Journal of Clinical Sleep Medicine</i> , 2020, 16, 1891-1900.	1.4	10
6	InÂVivo Expression of NUP93 and Its Alteration by NUP93 Mutations Causing Focal Segmental Glomerulosclerosis. <i>Kidney International Reports</i> , 2019, 4, 1312-1322.	0.4	10
7	Growth impairment in individuals with citrin deficiency. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 501-508.	1.7	14
8	NovelPHOX2Bmutations in congenital central hypoventilation syndrome. <i>Pediatrics International</i> , 2019, 61, 393-396.	0.2	7
9	A novel PHOX2B gene mutation in an extremely low birth weight infant with congenital central hypoventilation syndrome and variant Hirschsprung's disease. <i>European Journal of Medical Genetics</i> , 2019, 62, 103541.	0.7	5
10	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. <i>Journal of Inherited Metabolic Disease</i> , 2018, 41, 777-784.	1.7	27
11	Adult-onset type II citrullinemia: Current insights and therapy. <i>The Application of Clinical Genetics</i> , 2018, Volume 11, 163-170.	1.4	25
12	Phenotype-genotype correlations of PIGO deficiency with variable phenotypes from infantile lethality to mild learning difficulties. <i>Human Mutation</i> , 2017, 38, 805-815.	1.1	29
13	Effectiveness of Medium-Chain Triglyceride Oil Therapy in Two Japanese Citrin-Deficient Siblings: Evaluation Using Oral Glucose Tolerance Tests. <i>Tohoku Journal of Experimental Medicine</i> , 2016, 240, 323-328.	0.5	9
14	Unique food-entrained circadian rhythm in cysteine414-alanine mutant mCRY1 transgenic mice. <i>Sleep and Biological Rhythms</i> , 2016, 14, 261-269.	0.5	3
15	Analysis of the genes responsible for steroid-resistant nephrotic syndrome and/or focal segmental glomerulosclerosis in Japanese patients by whole-exome sequencing analysis. <i>Journal of Human Genetics</i> , 2016, 61, 137-141.	1.1	21
16	A novel ETFB mutation in a patient with glutaric aciduria type II. <i>Human Genome Variation</i> , 2015, 2, 15016.	0.4	4
17	Genotypeâ€phenotype relationship in Japanese patients with congenital central hypoventilation syndrome. <i>Journal of Human Genetics</i> , 2015, 60, 473-477.	1.1	43
18	Association of neonatal hyperbilirubinemia in breast-fed infants with UGT1A1 or SLCOs polymorphisms. <i>Journal of Human Genetics</i> , 2015, 60, 35-40.	1.1	15

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19	A Case of Congenital Central Hypoventilation Syndrome with a Novel Mutation of the <i>PHOX2B</i> Gene Presenting as Central Sleep Apnea. <i>Journal of Clinical Sleep Medicine</i> , 2014, 10, 327-329.	1.4	15
20	<i>PIGA</i> mutations cause early-onset epileptic encephalopathies and distinctive features. <i>Neurology</i> , 2014, 82, 1587-1596.	1.5	93
21	A Mutation of <i>COX6A1</i> Causes a Recessive Axonal or Mixed Form of Charcot-Marie-Tooth Disease. <i>American Journal of Human Genetics</i> , 2014, 95, 294-300.	2.6	65
22	Medium-chain triglyceride supplementation under a low-carbohydrate formula is a promising therapy for adult-onset type II citrullinemia. <i>Molecular Genetics and Metabolism Reports</i> , 2014, 1, 42-50.	0.4	33
23	De Novo Mutations in <i>GNAO1</i> , Encoding a G $\beta$ o Subunit of Heterotrimeric G Proteins, Cause Epileptic Encephalopathy. <i>American Journal of Human Genetics</i> , 2013, 93, 496-505.	2.6	187
24	The first Japanese case of Charcot-Marie-Tooth disease type 4H with a novel <i>FGD4</i> c.837-1G>A mutation. <i>Neuromuscular Disorders</i> , 2013, 23, 652-655.	0.3	6
25	Mild phenotype of Charcot-Marie-Tooth disease type 4B1. <i>Journal of the Neurological Sciences</i> , 2013, 334, 176-179.	0.3	19
26	De Novo Mutations in <i>SLC35A2</i> Encoding a UDP-Galactose Transporter Cause Early-Onset Epileptic Encephalopathy. <i>Human Mutation</i> , 2013, 34, 1708-1714.	1.1	85
27	Clinical spectrum of early onset epileptic encephalopathies caused by <i>KCNQ2</i> mutation. <i>Epilepsia</i> , 2013, 54, 1282-1287.	2.6	195
28	A founder haplotype of <i>APOE</i> -Sendai mutation associated with lipoprotein glomerulopathy. <i>Journal of Human Genetics</i> , 2013, 58, 254-258.	1.1	15
29	Clinical spectrum of <i>SCN2A</i> mutations expanding to Ohtahara syndrome. <i>Neurology</i> , 2013, 81, 992-998.	1.5	188
30	Characterization of age-associated alterations of islet function and structure in diabetic mutant <i>cryptochrome1</i> transgenic mice. <i>Journal of Diabetes Investigation</i> , 2013, 4, 428-435.	1.1	6
31	<i>INF2</i> mutations in Charcot-Marie-Tooth disease complicated with focal segmental glomerulosclerosis. <i>Journal of the Peripheral Nervous System</i> , 2013, 18, 97-98.	1.4	19
32	Molecular analysis of the genes causing recessive demyelinating Charcot-Marie-Tooth disease in Japan. <i>Journal of Human Genetics</i> , 2013, 58, 273-278.	1.1	16
33	Association of breast-fed neonatal hyperbilirubinemia with <i>UGT1A1</i> polymorphisms: 211G>A (G71R) mutation becomes a risk factor under inadequate feeding. <i>Journal of Human Genetics</i> , 2013, 58, 7-10.	1.1	39
34	Inheritance of polyalanine expansion mutation of <i>PHOX2B</i> in congenital central hypoventilation syndrome. <i>Journal of Human Genetics</i> , 2012, 57, 335-337.	1.1	23
35	<i>CASK</i> aberrations in male patients with Ohtahara syndrome and cerebellar hypoplasia. <i>Epilepsia</i> , 2012, 53, 1441-1449.	2.6	66
36	Whole exome sequencing identifies <i>KCNQ2</i> mutations in Ohtahara syndrome. <i>Annals of Neurology</i> , 2012, 72, 298-300.	2.8	88

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37	Slowly progressive sleep apnea in late-onset central hypoventilation syndrome. <i>Pediatrics International</i> , 2012, 54, 290-292.	0.2	2
38	Molecular diagnosis and clinical onset of Charcot-Marie-Tooth disease in Japan. <i>Journal of Human Genetics</i> , 2011, 56, 364-368.	1.1	66
39	A case of progressive familial intrahepatic cholestasis type 1 with compound heterozygous mutations of <i>ATP8B1</i> . <i>Pediatrics International</i> , 2011, 53, 107-110.	0.2	6
40	De novo 5q14.3 translocation 121.5 kb upstream of <i>MEF2C</i> in a patient with severe intellectual disability and early-onset epileptic encephalopathy. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 2879-2884.	0.7	26
41	Treatment with Lactose (Galactose)-Restricted and Medium-Chain Triglyceride-Supplemented Formula for Neonatal Intrahepatic Cholestasis Caused by Citrin Deficiency. <i>JIMD Reports</i> , 2011, 2, 37-44.	0.7	34
42	A second pediatric patient with lipoprotein glomerulopathy carrying a heterozygous APOE-Sendai mutation. <i>Japanese Journal of Pediatric Nephrology</i> , 2011, 24, 218-223.	0.0	1
43	Supernumerary impacted teeth in a patient with <i>SOX2</i> anophthalmia syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 2355-2359.	0.7	29
44	Frameshift mutations of the <i>ARX</i> gene in familial Ohtahara syndrome. <i>Epilepsia</i> , 2010, 51, 1679-1684.	2.6	30
45	<i>STXBP1</i> mutations in early infantile epileptic encephalopathy with suppression-burst pattern. <i>Epilepsia</i> , 2010, 51, 2397-2405.	2.6	133
46	Non-obese early onset diabetes mellitus in mutant cryptochrome1 transgenic mice. <i>European Journal of Clinical Investigation</i> , 2010, 40, 1011-1017.	1.7	12
47	Phenotypic variability in a family with Townes-Brocks syndrome. <i>Journal of Human Genetics</i> , 2010, 55, 550-551.	1.1	15
48	Polyalanine expansion of PHOX2B in congenital central hypoventilation syndrome: rs17884724:A>C is associated with 7-alanine expansion. <i>Journal of Human Genetics</i> , 2010, 55, 4-7.	1.1	7
49	Compound heterozygous PMP22 deletion mutations causing severe Charcot-Marie-Tooth disease type 1. <i>Journal of Human Genetics</i> , 2010, 55, 771-773.	1.1	15
50	The GARS gene is rarely mutated in Japanese patients with Charcot-Marie-Tooth neuropathy. <i>Journal of Human Genetics</i> , 2009, 54, 310-312.	1.1	22
51	Neurofilament light chain polypeptide gene mutations in Charcot-Marie-Tooth disease: nonsense mutation probably causes a recessive phenotype. <i>Journal of Human Genetics</i> , 2009, 54, 94-97.	1.1	83
52	Improvement of nephrotic syndrome by intensive lipid-lowering therapy in a patient with lipoprotein glomerulopathy. <i>Clinical and Experimental Nephrology</i> , 2009, 13, 659-662.	0.7	26
53	Unusual circadian locomotor activity and pathophysiology in mutant CRY1 transgenic mice. <i>Neuroscience Letters</i> , 2009, 451, 246-251.	1.0	40
54	A clinical phenotype of distal hereditary motor neuronopathy type II with a novel HSPB1 mutation. <i>Journal of the Neurological Sciences</i> , 2009, 277, 9-12.	0.3	54

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55	De novo mutations in the gene encoding STXBP1 (MUNC18-1) cause early infantile epileptic encephalopathy. <i>Nature Genetics</i> , 2008, 40, 782-788.	9.4	498
56	Cardiac Ion Channel Gene Mutations in Sudden Infant Death Syndrome. <i>Pediatric Research</i> , 2008, 64, 482-487.	1.1	95
57	A Longer Polyalanine Expansion Mutation in the ARX Gene Causes Early Infantile Epileptic Encephalopathy with Suppression-Burst Pattern (Ohtahara Syndrome). <i>American Journal of Human Genetics</i> , 2007, 81, 361-366.	2.6	168
58	De novo polyalanine expansion of PHOX2B in congenital central hypoventilation syndrome: unequal sister chromatid exchange during paternal gametogenesis. <i>Journal of Human Genetics</i> , 2007, 52, 921-925.	1.1	15
59	Ankyrin-G Regulates Inactivation Gating of the Neuronal Sodium Channel, Nav1.6. <i>Journal of Neurophysiology</i> , 2006, 96, 1347-1357.	0.9	36
60	Periaxin mutation in Japanese patients with Charcot-Marie-Tooth disease. <i>Journal of Human Genetics</i> , 2006, 51, 625-628.	1.1	14
61	Genetic Analysis of Shwachman-Diamond Syndrome: Phenotypic Heterogeneity in Patients Carrying Identical SBDS Mutations. <i>Tohoku Journal of Experimental Medicine</i> , 2005, 206, 253-259.	0.5	26
62	Neonatal hyperbilirubinemia and the bilirubin uridine diphosphate-glucuronosyltransferase gene: The common $\Delta 3263T \rightarrow \Delta fG$ mutation of phenobarbital response enhancer module is not associated with the neonatal hyperbilirubinemia in Japanese. <i>Pediatrics International</i> , 2005, 47, 137-141.	0.2	22
63	Cross-talk between $\beta 1$ -adrenoceptors and ETA receptors in modulation of the slow component of delayed rectifier K <sup>+</sup> currents. <i>Naunyn-Schmiedeberg's Archives of Pharmacology</i> , 2005, 371, 133-140.	1.4	6
64	Mitochondrial GTPase mitofusin 2 mutation in Charcot-Marie-Tooth neuropathy type 2A. <i>Human Genetics</i> , 2005, 116, 23-27.	1.8	229
65	Small heat shock protein 27 mutation in a Japanese patient with distal hereditary motor neuropathy. <i>Journal of Human Genetics</i> , 2005, 50, 473-476.	1.1	85
66	Sensitive Detection of Polyalanine Expansions in PHOX2B by Polymerase Chain Reaction Using Bisulfite-Converted DNA. <i>Journal of Molecular Diagnostics</i> , 2005, 7, 638-640.	1.2	15
67	Periaxin mutation causes early-onset but slow-progressive Charcot-Marie-Tooth disease. <i>Journal of Human Genetics</i> , 2004, 49, 376-379.	1.1	41
68	Sudden Infant Death Syndrome Is Not Associated with the Mutation of PHOX2B Gene, a Major Causative Gene of Congenital Central Hypoventilation Syndrome. <i>Tohoku Journal of Experimental Medicine</i> , 2004, 203, 65-68.	0.5	37
69	Molecular analysis of congenital central hypoventilation syndrome. <i>Human Genetics</i> , 2003, 114, 22-26.	1.8	174
70	Screening of the early growth response 2 gene in Japanese patients with Charcot-Marie-Tooth disease type 1. <i>Journal of the Neurological Sciences</i> , 2003, 210, 61-64.	0.3	24
71	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. <i>Brain</i> , 2003, 126, 134-151.	3.7	202
72	Neonatal Hyperbilirubinemia in Japanese Neonates: Analysis of the Heme Oxygenase-1 Gene and Fetal Hemoglobin Composition in Cord Blood. <i>Pediatric Research</i> , 2003, 54, 165-171.	1.1	34

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73	Polymorphisms of Heme Oxygenase-1 and Bilirubin UDP-Glucuronosyltransferase Genes are not Associated with Kawasaki Disease Susceptibility. <i>Tohoku Journal of Experimental Medicine</i> , 2003, 200, 155-159.	0.5	11
74	Congenital Central Hypoventilation Syndrome: A Novel Mutation of the RET Gene in an Isolated Case.. <i>Tohoku Journal of Experimental Medicine</i> , 2002, 196, 241-246.	0.5	24
75	Association of the uteroglobin gene polymorphism with IgA nephropathy. <i>American Journal of Kidney Diseases</i> , 2002, 39, 36-41.	2.1	22
76	Molecular analysis in Japanese patients with Charcot-Marie-Tooth disease: DGGE analysis for PMP22, MPZ, and Cx32/GJB1 mutations. <i>Human Mutation</i> , 2002, 20, 392-398.	1.1	57
77	Mutation of the doublecortin gene in male patients with double cortex syndrome: Somatic mosaicism detected by hair root analysis. <i>Annals of Neurology</i> , 2001, 50, 547-551.	2.8	47
78	Hemizygous mutation of the peripheral myelin protein 22 gene associated with Charcot-Marie-Tooth disease type 1. <i>Annals of Neurology</i> , 2000, 47, 101-103.	2.8	48
79	Neonatal hyperbilirubinemia and a common mutation of the bilirubin uridine diphosphate-glucuronosyltransferase gene in Japanese. <i>Journal of Human Genetics</i> , 1999, 44, 22-25.	1.1	83
80	A novel mutation of the doublecortin gene in Japanese patients with X-linked lissencephaly and subcortical band heterotopia. <i>Human Genetics</i> , 1999, 104, 341-344.	1.8	27
81	Diffuse pachygyria with cerebellar hypoplasia: A milder form of microlissencephaly or a new genetic syndrome?. <i>Annals of Neurology</i> , 1999, 46, 660-663.	2.8	19
82	Deletion and Nonsense Mutations of the Connexin 32 Gene Associated with Charcot-Marie-Tooth Disease.. <i>Tohoku Journal of Experimental Medicine</i> , 1999, 188, 239-244.	0.5	24
83	Neonatal hyperbilirubinemia and mutation of the bilirubin uridine diphosphate-glucuronosyltransferase gene: a common missense mutation among Japanese, Koreans and Chinese. <i>IUBMB Life</i> , 1998, 46, 21-26.	1.5	84
84	De novo mutation of the myelin Po gene in DÃ©jÃ©rine-Sottas disease (hereditary motor and sensory) Tj ETQqO 0,0 rgBT /Oyerlock 10	1.1	16
85	Four novel mutations of the connexin 32 gene in four Japanese families with Charcot-Marie-Tooth disease type 1. , 1998, 80, 352-355.		16
86	Clustering of CMT1A duplication breakpoints in a 700 bp interval of the CMT1A-REP repeat. , 1998, 11, 109-113.		21
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 Ibl±. <i>Journal of Pediatric Hematology/Oncology</i> , 1998, 20, 246-251.	0.3	13
88	Locations of crossover breakpoints within the CMT1A-REP repeat in Japanese patients with CMT1A and HNPP. <i>Human Genetics</i> , 1997, 99, 151-154.	1.8	32
89	Novel mutation of the myelin Po gene in a pedigree with Charcot-Marie-Tooth disease type 1b. , 1997, 71, 246-248.		12
90	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

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91	Defective Signal Transduction through the Thromboxane A2 Receptor in a Patient with a Mild Bleeding Disorder: Deficiency of the Inositol 1,4,5-Triphosphate Formation despite Normal G-protein Activation. <i>Thrombosis and Haemostasis</i> , 1997, 77, 0991-0995.	1.8	20
92	A new mutation of the Po gene in patients with Charcot-Marie-Tooth disease type 1B: screening of the Po gene by heteroduplex analysis. <i>Neuroscience Letters</i> , 1996, 204, 173-176.	1.0	14
93	A Novel Homozygous Mutation of the Myelin Po Gene Producing Dejerine-Sottas Disease (Hereditary) <i>Tj ETQq1 1 0.784314 rgBT / O</i> 222, 107-110.	1.0	49
94	Isolation and sequence determination of cDNA encoding mouse rab 4 and candidate approach for the beige mutation in mice. <i>IUBMB Life</i> , 1996, 40, 647-651.	1.5	0
95	GAPO syndrome: Report on the first case in Japan. <i>American Journal of Medical Genetics Part A</i> , 1995, 58, 257-261.	2.4	18
96	Congenital hypomyelination neuropathy: decreased expression of the P2 protein in peripheral nerve with normal DNA sequence of the coding region. <i>Journal of the Neurological Sciences</i> , 1995, 134, 150-159.	0.3	11
97	Identification of the mutations in the T-protein gene causing typical and atypical nonketotic hyperglycinemia. <i>Human Genetics</i> , 1994, 93, 655-8.	1.8	39
98	Elevated expression of messenger RNA for peripheral myelin protein 22 in biopsied peripheral nerves of patients with Charcot-Marie-Tooth disease type 1A. <i>Annals of Neurology</i> , 1994, 35, 445-450.	2.8	145
99	Expression of Po protein in sural nerve of a patient with hereditary motor and sensory neuropathy type III. <i>Journal of the Neurological Sciences</i> , 1994, 124, 67-70.	0.3	9
100	A T-to-C substitution at nucleotide 12311 in tRNA <sup>Leu</sup> (CUN) gene may be a mtDNA polymorphism. <i>Journal of the Neurological Sciences</i> , 1994, 127, 236.	0.3	1
101	Structure and Chromosomal Localization of the Aminomethyltransferase Gene (AMT). <i>Genomics</i> , 1994, 19, 27-30.	1.3	22
102	Neural Cell Adhesion Proteins and Neurological Diseases1. <i>Journal of Biochemistry</i> , 1994, 116, 1187-1192.	0.9	14
103	Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes (MELAS) decrease in diastolic left ventricular function assessed by echocardiography. <i>Pediatric Cardiology</i> , 1993, 14, 162-166.	0.6	17
104	Charcot-Marie-Tooth neuropathy type 1B is associated with mutations of the myelin P0 gene. <i>Nature Genetics</i> , 1993, 5, 31-34.	9.4	391
105	De novo mutation of the myelin Po gene in Dejerine-Sottas disease (hereditary motor and sensory) <i>Tj ETQq1 1 0.784314 rgBT / O</i> 227	9.4	227
106	Structure and Chromosomal Localization of the Gene Encoding the Human Myelin Protein Zero (MPZ). <i>Genomics</i> , 1993, 17, 755-758.	1.3	84
107	Structure and Localization of the Gene Encoding Human Peripheral Myelin Protein 2 (PMP2). <i>Genomics</i> , 1993, 18, 244-248.	1.3	26
108	Mutation of the myelin Po gene in Charcot-Marie-Tooth neuropathy type 1B. <i>Human Molecular Genetics</i> , 1993, 2, 1369-1372.	1.4	101

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109	The Effect of Carnitine on the Metabolism of Valproic Acid in Epileptic Patients.. Tohoku Journal of Experimental Medicine, 1992, 167, 89-92.	0.5	10
110	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.	1.0	41
111	Localization of PMP-22 gene (candidate gene for the Charcot-Marie-Tooth disease 1a) to band 17p11.2 by direct r-banding fluorescenceln situ hybridization. Japanese Journal of Human Genetics, 1992, 37, 303-306.	0.8	7
112	Isolation and sequence determination of cDNA encoding the major structural protein of human peripheral myelin. Biochemical and Biophysical Research Communications, 1991, 180, 515-518.	1.0	83
113	Isolation and sequence determination of cDNA encoding P2 protein of human peripheral myelin. Biochemical and Biophysical Research Communications, 1991, 181, 204-207.	1.0	19
114	Two homozygous cases of erythrocyte pyruvate kinase (PK) deficiency in Japan: PK sendai and PK shinshu. American Journal of Hematology, 1988, 28, 186-190.	2.0	6
115	Feasibility of prenatal diagnosis of nonketotic hyperglycinemia: Existence of the glycine cleavage system in placenta. Journal of Pediatrics, 1987, 110, 124-126.	0.9	24
116	Nonketotic hyperglycinemia: Analyses of glycine cleavage system in typical and atypical cases. Journal of Pediatrics, 1987, 110, 873-877.	0.9	59