

Yi-Chung Lee

List of Publications by Year in descending order

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127
papers

3,525
citations

159585

30
h-index

168389

53
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130
all docs

130
docs citations

130
times ranked

5934
citing authors

#	ARTICLE	IF	CITATIONS
1	Investigating ZFYVE26 mutations in a Taiwanese cohort with hereditary spastic paraplegia. Journal of the Formosan Medical Association, 2022, 121, 126-133.	1.7	3
2	GGC Repeat Expansion of <i>NOTCH2NLC</i> in Taiwanese Patients With Inherited Neuropathies. Neurology, 2022, 98, .	1.1	24
3	Treatment response, risk of relapse and clinical characteristics of Taiwanese patients with neuromyelitis optica spectrum disorder. Journal of the Formosan Medical Association, 2022, 121, 1647-1656.	1.7	8
4	Gain of C-Ala enables AlaRS to target the L-shaped tRNA ^{Ala} . Nucleic Acids Research, 2022, 50, 2190-2200.	14.5	3
5	Reply to: Adult-onset leukoencephalopathy caused by CSF1R mutations: Is all that glitters gold?. Annals of Clinical and Translational Neurology, 2022, 9, 101-102.	3.7	0
6	Assessing the NOTCH2NLC GGC repeat expansion in Taiwanese patients with hereditary spastic paraplegia. Parkinsonism and Related Disorders, 2022, 96, 43-44.	2.2	2
7	Mutation screening and association analysis of <i>NOTCH3</i> p.R544C in patients with migraine with or without aura. Cephalalgia, 2022, 42, 888-898.	3.9	4
8	Neuronal intranuclear inclusion disease in patients with adult-onset non-vascular leukoencephalopathy. Brain, 2022, 145, 3010-3021.	7.6	28
9	Transient Postictal Hyperglycemia as a Diagnostic Clue of Mitochondrial Encephalomyopathy, Lactic Acidosis, and Stroke-like Episodes.. Acta Neurologica Taiwanica, 2022, 31(2), 79-83.	0.3	0
10	Mitochondrial DNA m.3243A>G mutation rarely causes CADASIL-like phenotype. Neurobiology of Aging, 2021, 97, 145.e5-145.e6.	3.1	2
11	Reply: MELAS can be delineated from CADASIL by genotype and phenotype. Neurobiology of Aging, 2021, 99, 104.	3.1	0
12	Intracerebral Hemorrhage in Cerebral Autosomal Dominant Arteriopathy With Subcortical Infarcts and Leukoencephalopathy. Stroke, 2021, 52, 985-993.	2.0	25
13	Clinical and genetic characterization of hereditary spastic paraplegia type 3A in Taiwan. Parkinsonism and Related Disorders, 2021, 87, 87-91.	2.2	7
14	Analysis of NOTCH2NLC GGC repeat expansion in Taiwanese patients with amyotrophic lateral sclerosis. Neurobiology of Aging, 2021, 108, 210-212.	3.1	9
15	Expanding the phenotype of AFG3L2 mutations: Late-onset autosomal recessive spinocerebellar ataxia. Journal of the Neurological Sciences, 2021, 428, 117600.	0.6	5
16	Clinical and genetic characterization of adult-onset leukoencephalopathy caused by <i>CSF1R</i> mutations. Annals of Clinical and Translational Neurology, 2021, 8, 2121-2131.	3.7	9
17	Investigating ABCD1 mutations in a Taiwanese cohort with hereditary spastic paraplegia phenotype. Parkinsonism and Related Disorders, 2021, 92, 7-12.	2.2	1
18	Investigating TBP CAG/CAA trinucleotide repeat expansions in a Taiwanese cohort with ALS. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2021, 22, 442-447.	1.7	3

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19	Genetic and Functional Analysis of Glycosyltransferase 8 Domain-Containing Protein 1 in Taiwanese Patients With Amyotrophic Lateral Sclerosis. <i>Neurology: Genetics</i> , 2021, 7, e627.	1.9	2
20	A <i>PIAS1</i> Protective Variant <i>S510G</i> Delays <i>polyQ</i> Disease Onset by Modifying Protein Homeostasis. <i>Movement Disorders</i> , 2021, , .	3.9	7
21	<i>NOTCH3</i> cysteine-altering variant is an important risk factor for stroke in the Taiwanese population. <i>Neurology</i> , 2020, 94, e87-e96.	1.1	28
22	Prealbumin Is Associated with the Weaning Outcome for Patients on Prolonged Mechanical Ventilation. , 2020, , .		0
23	A case of GNE myopathy mimicking hereditary motor neuropathy. <i>European Journal of Neurology</i> , 2020, 27, 2389-2391.	3.3	4
24	Rapid progressive ALS in a patient with a <i>DNAJC7</i> loss-of-function mutation. <i>Neurology: Genetics</i> , 2020, 6, e503.	1.9	13
25	Clinical and Genetic Characterization of Autosomal Recessive Spinocerebellar Ataxia Type 16 (SCAR16) in Taiwan. <i>Cerebellum</i> , 2020, 19, 544-549.	2.5	13
26	Hand-onset weakness is a common feature of ALS patients with a <i>NEK1</i> loss-of-function variant. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 965-971.	3.7	8
27	Cerebral Microbleed Burdens in Specific Brain Regions Are Associated With Disease Severity of Cerebral Autosomal Dominant Arteriopathy With Subcortical Infarcts and Leukoencephalopathy. <i>Journal of the American Heart Association</i> , 2020, 9, e016233.	3.7	20
28	Clinical characteristics of Taiwanese patients with Hereditary spastic paraplegia type 5. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 486-496.	3.7	12
29	Investigating <i>PUM1</i> mutations in a Taiwanese cohort with cerebellar ataxia. <i>Parkinsonism and Related Disorders</i> , 2019, 66, 220-223.	2.2	8
30	Reply: A novel <i>WARS</i> mutation causes distal hereditary motor neuropathy in a Chinese family. <i>Brain</i> , 2019, 142, e50-e50.	7.6	0
31	Mutation spectrum of Charcot-Marie-Tooth disease among the Han Chinese in Taiwan. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 1090-1101.	3.7	29
32	Clinical and genetic profiles of hereditary transthyretin amyloidosis in Taiwan. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 913-922.	3.7	29
33	Preimplantation Genetic Diagnosis of Neurodegenerative Diseases: Review of Methodologies and Report of Our Experience as a Regional Reference Laboratory. <i>Diagnostics</i> , 2019, 9, 44.	2.6	5
34	Investigating <i>CYP2C19</i> loss-of-function allele statuses and their association with stroke of different etiologies in a Taiwanese population. <i>Journal of the Chinese Medical Association</i> , 2019, 82, 469-472.	1.4	16
35	Plasma amyloid assay as a pre-screening tool for amyloid positron emission tomography imaging in early-stage Alzheimer's disease. <i>Alzheimer's Research and Therapy</i> , 2019, 11, 111.	6.2	21
36	Comparable progression of spinocerebellar ataxias between Caucasians and Chinese. <i>Parkinsonism and Related Disorders</i> , 2019, 62, 156-162.	2.2	14

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37	Mutations in ATP1A1 Cause Dominant Charcot-Marie-Tooth Type 2. American Journal of Human Genetics, 2018, 102, 505-514.	6.2	59
38	Renal function is associated with 1-month and 1-year mortality in patients with ischemic stroke. Atherosclerosis, 2018, 269, 288-293.	0.8	38
39	Blockade of soluble epoxide hydrolase attenuates post-ischemic neuronal hyperexcitation and confers resilience against stroke with TrkB activation. Scientific Reports, 2018, 8, 118.	3.3	17
40	Investigating CCNF mutations in a Taiwanese cohort with amyotrophic lateral sclerosis. Neurobiology of Aging, 2018, 62, 243.e1-243.e6.	3.1	13
41	Mutational analysis of <i>CCM1</i> , <i>CCM2</i> and <i>CCM3</i> in a Han Chinese cohort with multiple cerebral cavernous malformations in Taiwan. Clinical Genetics, 2018, 94, 389-390.	2.0	3
42	Genetic analysis of ANXA11 variants in a Han Chinese cohort with amyotrophic lateral sclerosis in Taiwan. Neurobiology of Aging, 2018, 72, 188.e1-188.e2.	3.1	12
43	Characterization of Heterozygous <i>HTRA1</i> Mutations in Taiwanese Patients With Cerebral Small Vessel Disease. Stroke, 2018, 49, 1593-1601.	2.0	39
44	ATXN2 trinucleotide repeat length correlates with risk of ALS. Neurobiology of Aging, 2017, 51, 178.e1-178.e9.	3.1	86
45	A novel <i>DNAJB6</i> mutation causes dominantly inherited distal onset myopathy and compromises <i>DNAJB6</i> function. Clinical Genetics, 2017, 92, 150-157.	2.0	16
46	A recurrent WARS mutation is a novel cause of autosomal dominant distal hereditary motor neuropathy. Brain, 2017, 140, 1252-1266.	7.6	75
47	Cholesterol Levels Are Associated with 30-day Mortality from Ischemic Stroke in Dialysis Patients. Journal of Stroke and Cerebrovascular Diseases, 2017, 26, 1349-1356.	1.6	6
48	Clinical and Molecular Characterization of PMP22 point mutations in Taiwanese patients with Inherited Neuropathy. Scientific Reports, 2017, 7, 15363.	3.3	10
49	Investigation for SPTLC1 mutations in a Taiwanese cohort with hereditary neuropathies. Journal of the Neurological Sciences, 2017, 381, 463-464.	0.6	2
50	Unmasking adrenoleukodystrophy in a cohort of cerebellar ataxia. PLoS ONE, 2017, 12, e0177296.	2.5	11
51	Mutational analysis of ITPR1 in a Taiwanese cohort with cerebellar ataxias. PLoS ONE, 2017, 12, e0187503.	2.5	13
52	Clinical and biophysical characterization of 19 <i>GJB1</i> mutations. Annals of Clinical and Translational Neurology, 2016, 3, 854-865.	3.7	16
53	Spinocerebellar ataxia type 36 in the Han Chinese. Neurology: Genetics, 2016, 2, e68.	1.9	24
54	Mutational analysis of TBK1 in Taiwanese patients with amyotrophic lateral sclerosis. Neurobiology of Aging, 2016, 40, 191.e11-191.e16.	3.1	46

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55	Clinical and Molecular Characterization of BSCL2 Mutations in a Taiwanese Cohort with Hereditary Neuropathy. PLoS ONE, 2016, 11, e0147677.	2.5	18
56	PRRT2 mutations lead to neuronal dysfunction and neurodevelopmental defects. Oncotarget, 2016, 7, 39184-39196.	1.8	55
57	The interaction of serum testosterone levels and androgen receptor CAG repeat polymorphism on the risk of erectile dysfunction in aging Taiwanese men. Andrology, 2015, 3, 902-908.	3.5	16
58	Two Novel De Novo GARS Mutations Cause Early-Onset Axonal Charcot-Marie-Tooth Disease. PLoS ONE, 2015, 10, e0133423.	2.5	22
59	Characterization of CADASIL among the Han Chinese in Taiwan: Distinct Genotypic and Phenotypic Profiles. PLoS ONE, 2015, 10, e0136501.	2.5	73
60	What we have learned from the next-generation sequencing: Contributions to the genetic diagnoses and understanding of pathomechanisms of neurodegenerative diseases. Journal of Neurogenetics, 2015, 29, 103-112.	1.4	13
61	Acute simultaneous multiple lacunar infarcts as the initial presentation of cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy. Journal of the Chinese Medical Association, 2015, 78, 424-426.	1.4	8
62	Mutational analysis of MATR3 in Taiwanese patients with amyotrophic lateral sclerosis. Neurobiology of Aging, 2015, 36, 2005.e1-2005.e4.	3.1	64
63	Coexistence of Charcot Marie Tooth disease type 1A and diabetes in Taiwan: A clinicopathological study. Journal of the Neurological Sciences, 2015, 358, 213-220.	0.6	1
64	Connectivity Features for Identifying Cognitive Impairment in Presymptomatic Carotid Stenosis. PLoS ONE, 2014, 9, e85441.	2.5	39
65	A novel <i>TFG</i> mutation causes Charcot-Marie-Tooth disease type 2 and impairs TFG function. Neurology, 2014, 83, 903-912.	1.1	39
66	C9ORF72 repeat expansion is not a significant cause of late onset cerebellar ataxia syndrome. Journal of the Neurological Sciences, 2014, 347, 322-324.	0.6	11
67	Validating a rapid, real-time, PCR-based direct mutation detection assay for preimplantation genetic diagnosis. Gene, 2014, 548, 299-305.	2.2	14
68	Spinocerebellar ataxia 35. Neurology, 2014, 83, 1554-1561.	1.1	33
69	Extensive molecular genetic survey of Taiwanese patients with amyotrophic lateral sclerosis. Neurobiology of Aging, 2014, 35, 2423.e1-2423.e6.	3.1	46
70	Cerebral involvement in spinal and bulbar muscular atrophy (Kennedy's disease): A pilot study of PET. Journal of the Neurological Sciences, 2013, 335, 139-144.	0.6	24
71	Exome Sequencing Identifies GNB4 Mutations as a Cause of Dominant Intermediate Charcot-Marie-Tooth Disease. American Journal of Human Genetics, 2013, 92, 422-430.	6.2	46
72	A homozygous NOTCH3 mutation p.R544C and a heterozygous TREX1 variant p.C99MfsX3 in a family with hereditary small vessel disease of the brain. Journal of the Chinese Medical Association, 2013, 76, 319-324.	1.4	25

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73	Mutational analysis of the 5' non-coding region of GJB1 in a Taiwanese cohort with Charcot-Marie-Tooth neuropathy. <i>Journal of the Neurological Sciences</i> , 2013, 332, 51-55.	0.6	9
74	Allergic rhinitis and risk of erectile dysfunction – a nationwide population-based study. <i>Allergy: European Journal of Allergy and Clinical Immunology</i> , 2013, 68, 440-445.	5.7	11
75	Visual cortex excitability and plasticity associated with remission from chronic to episodic migraine. <i>Cephalalgia</i> , 2012, 32, 537-543.	3.9	48
76	Mutations in <i>KCND3</i> cause spinocerebellar ataxia type 22. <i>Annals of Neurology</i> , 2012, 72, 859-869.	5.3	138
77	FGF21 in ataxia patients with spinocerebellar atrophy and mitochondrial disease. <i>Clinica Chimica Acta</i> , 2012, 414, 225-227.	1.1	13
78	SCA31 is rare in the Chinese population on Taiwan. <i>Neurobiology of Aging</i> , 2012, 33, 426.e23-426.e24.	3.1	11
79	A hexanucleotide repeat expansion in C9ORF72 causes familial and sporadic ALS in Taiwan. <i>Neurobiology of Aging</i> , 2012, 33, 2232.e11-2232.e18.	3.1	52
80	Clinical and cellular characterization of two novel MPZ mutations, p.I135M and p.Q187PfsX63. <i>Clinical Neurology and Neurosurgery</i> , 2012, 114, 124-129.	1.4	5
81	Electrophysiological characterization of Charcot-Marie-Tooth disease type 1A in Taiwan. <i>Journal of the Chinese Medical Association</i> , 2012, 75, 197-202.	1.4	5
82	PRRT2 Mutations in Paroxysmal Kinesigenic Dyskinesia with Infantile Convulsions in a Taiwanese Cohort. <i>PLoS ONE</i> , 2012, 7, e38543.	2.5	46
83	A Recurrent loss-of-function alanyl-tRNA synthetase (AARS) mutation in patients with charcot-marie-tooth disease type 2N (CMT2N). <i>Human Mutation</i> , 2012, 33, 244-253.	2.5	90
84	FUS, TARDBP, and SOD1 mutations in a Taiwanese cohort with familial ALS. <i>Neurobiology of Aging</i> , 2011, 32, 553.e13-553.e21.	3.1	57
85	Gabapentin for complex regional pain syndrome in Machado-Joseph disease: a case report. <i>Journal of Medical Case Reports</i> , 2011, 5, 268.	0.8	4
86	Comparison of cerebellar ataxias: A three-year prospective longitudinal assessment. <i>Movement Disorders</i> , 2011, 26, 2081-2087.	3.9	55
87	Stenting versus Medical Treatment for Severe Symptomatic Intracranial Stenosis. <i>American Journal of Neuroradiology</i> , 2011, 32, 911-916.	2.4	19
88	The Mutational Spectrum in a Cohort of Charcot-Marie-Tooth Disease Type 2 among the Han Chinese in Taiwan. <i>PLoS ONE</i> , 2011, 6, e29393.	2.5	84
89	Transmissible spongiform encephalopathies with P102L mutation of PRNP manifesting different phenotypes: clinical, neuroimaging, and electrophysiological studies in Chinese kindred in Taiwan. <i>Journal of Neurology</i> , 2010, 257, 191-197.	3.6	20
90	Cellular characterization of MPZ mutations presenting with diverse clinical phenotypes. <i>Journal of Neurology</i> , 2010, 257, 1661-1668.	3.6	16

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91	Intronic CArG Box Regulates Cysteine-Rich Protein 2 Expression in the Adult but Not in Developing Vasculature. <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , 2010, 30, 835-842.	2.4	11
92	Mitochondrial DNA damage in spinal and bulbar muscular atrophy patients and carriers. <i>Clinica Chimica Acta</i> , 2010, 411, 626-630.	1.1	14
93	Clinical characterization and genetic analysis of a possible novel type of dominant Charcot-Marie-Tooth disease. <i>Neuromuscular Disorders</i> , 2010, 20, 534-539.	0.6	8
94	Occipital lobe seizures related to marked elevation of hemoglobin A1C: Report of two cases. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2010, 19, 359-362.	2.0	34
95	Fatigue in Colchicine Myopathy: A Study of Transcranial Magnetic Stimulation. <i>Journal of the Chinese Medical Association</i> , 2010, 73, 623-627.	1.4	2
96	Incidental findings on brain magnetic resonance imaging: systematic review and meta-analysis. <i>BMJ: British Medical Journal</i> , 2009, 339, b3016-b3016.	2.3	634
97	Eye of the tiger-like MRI in parkinsonian variant of multiple system atrophy. <i>Journal of Neural Transmission</i> , 2009, 116, 861-866.	2.8	17
98	Population-specific spectrum of NOTCH3 mutations, MRI features and founder effect of CADASIL in Chinese. <i>Journal of Neurology</i> , 2009, 256, 249-255.	3.6	74
99	The remarkably variable expressivity of CADASIL: report of a minimally symptomatic man at an advanced age. <i>Journal of Neurology</i> , 2009, 256, 1026-1027.	3.6	19
100	The "hot cross bun" sign in the patients with spinocerebellar ataxia. <i>European Journal of Neurology</i> , 2009, 16, 513-516.	3.3	90
101	Electrodiagnosis of Carpal Tunnel Syndrome: Which Transcarpal Conduction Technique Is Best?. <i>Journal of Clinical Neurophysiology</i> , 2009, 26, 366-371.	1.7	18
102	Charcot-Marie-Tooth disease. <i>Current Treatment Options in Neurology</i> , 2008, 10, 94-102.	1.8	17
103	Transthyretin Ala97Ser in Chinese "Taiwanese patients with familial amyloid polyneuropathy: Genetic studies and phenotype expression. <i>Journal of the Neurological Sciences</i> , 2008, 267, 91-99.	0.6	26
104	The role of forearm mixed nerve conduction study in the evaluation of proximal conduction slowing in carpal tunnel syndrome. <i>Clinical Neurophysiology</i> , 2008, 119, 2800-2803.	1.5	10
105	<i>MPZ</i> mutation G123S characterization. <i>Neurology</i> , 2008, 70, 273-277.	1.1	12
106	Incidental Findings on Brain MRI. <i>New England Journal of Medicine</i> , 2008, 358, 853-855.	27.0	13
107	Alteration of Proximal Conduction Velocity at Distal Nerve Injury in Carpal Tunnel Syndrome: Demyelinating Versus Axonal Change. <i>Journal of Clinical Neurophysiology</i> , 2008, 25, 161-166.	1.7	16
108	The Real Role of Forearm Mixed Nerve Conduction Velocity in the Assessment of Proximal Forearm Conduction Slowing in Carpal Tunnel Syndrome. <i>Journal of Clinical Neurophysiology</i> , 2008, 25, 373-377.	1.7	8

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109	Motor neuron disease-like syndrome secondary to trapped fourth ventricle and obstruction of cerebrospinal fluid pathway. <i>Clinical Neurology and Neurosurgery</i> , 2007, 109, 383-387.	1.4	6
110	Bromism caused by mix-formulated analgesic injectables. <i>Human and Experimental Toxicology</i> , 2007, 26, 971-973.	2.2	9
111	Common mitochondrial DNA and POLG1 mutations are rare in the Chinese patients with adult-onset ataxia on Taiwan. <i>Journal of the Neurological Sciences</i> , 2007, 254, 65-68.	0.6	16
112	Comparison of sensitivity of transcarpal median motor conduction velocity and conventional conduction techniques in electrodiagnosis of carpal tunnel syndrome. <i>Clinical Neurophysiology</i> , 2006, 117, 984-991.	1.5	55
113	Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy: Two novel mutations in the NOTCH3 gene in Chinese. <i>Journal of the Neurological Sciences</i> , 2006, 246, 111-115.	0.6	19
114	Median nerve motor conduction velocity is concordant with myelin protein zero gene mutation. <i>Journal of Neurology</i> , 2005, 252, 151-155.	3.6	9
115	Spastic paraparesis as a manifestation of metabolic vitamin B12 deficiency. <i>Journal of Neurology</i> , 2005, 252, 1125-1126.	3.6	3
116	An environmental decision support system for the management of water pollution in a tidal river network. <i>International Journal of Geographical Information Science</i> , 2005, 19, 483-500.	4.8	12
117	Cognitive Reserve: A SPECT Study of 132 Alzheimer's Disease Patients with an Education Range of 0-19 Years. <i>Dementia and Geriatric Cognitive Disorders</i> , 2005, 20, 8-14.	1.5	38
118	Gene symbol: NOTCH3. Disease: cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy. <i>Human Genetics</i> , 2005, 116, 242.	3.8	1
119	Longitudinal Cerebral Perfusion Decrease in Mild Alzheimer's Disease Revealed by SPECT with Statistical Parametric Mapping Method. <i>European Neurology</i> , 2004, 52, 42-49.	1.4	5
120	Myelin protein zero gene mutations in Taiwanese patients with Charcot-Marie-Tooth disease type 1. <i>Journal of the Neurological Sciences</i> , 2004, 219, 95-100.	0.6	7
121	Prolonged central motor conduction time of lower limb muscle in spinocerebellar ataxia 6. <i>Journal of Clinical Neuroscience</i> , 2004, 11, 381-383.	1.5	10
122	Prolonged cortical relay time of long latency reflex and central motor conduction in patients with spinocerebellar ataxia type 6. <i>Clinical Neurophysiology</i> , 2003, 114, 458-462.	1.5	20
123	Statistical Parametric Mapping of Brain SPECT Perfusion Abnormalities in Patients with Alzheimer's Disease. <i>European Neurology</i> , 2003, 49, 142-145.	1.4	22
124	Selective Hypoperfusion of Anterior Cingulate Gyrus in Depressed AD Patients: A Brain SPECT Finding by Statistical Parametric Mapping. <i>Dementia and Geriatric Cognitive Disorders</i> , 2003, 16, 238-244.	1.5	41
125	Wernicke's Encephalopathy in a Patient with Multiple Symmetrical Lipomatosis and the A8344G Mutation of Mitochondrial DNA. <i>European Neurology</i> , 2002, 47, 126-128.	1.4	6
126	Cutoff Scores of the Cognitive Abilities Screening Instrument, Chinese Version in Screening of Dementia. <i>Dementia and Geriatric Cognitive Disorders</i> , 2002, 14, 176-182.	1.5	129

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127	Epileptic seizures in a patient by immersing his right hand into hot water. Seizure: the Journal of the British Epilepsy Association, 2000, 9, 605-607.	2.0	13