Tally Lerman-Sagie

List of Publications by Year
in descending order

182	7,155	76294 40	69214 77
papers	citations	h-index	g-index

185	185	185	9964
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	De novo germline and postzygotic mutations in AKT3, PIK3R2 and PIK3CA cause a spectrum of related megalencephaly syndromes. Nature Genetics, 2012, 44, 934-940.	9.4	621
2	Targeted resequencing in epileptic encephalopathies identifies de novo mutations in CHD2 and SYNGAP1. Nature Genetics, 2013, 45, 825-830.	9.4	589
3	X-linked protocadherin 19 mutations cause female-limited epilepsy and cognitive impairment. Nature Genetics, 2008, 40, 776-781.	9.4	397
4	De Novo Mutations in SLC1A2 and CACNA1A Are Important Causes of Epileptic Encephalopathies. American Journal of Human Genetics, 2016, 99, 287-298.	2.6	247
5	Rare copy number variants are an important cause of epileptic encephalopathies. Annals of Neurology, 2011, 70, 974-985.	2.8	222
6	Mortality in Dravet syndrome. Epilepsy Research, 2016, 128, 43-47.	0.8	218
7	Megalencephalyâ€capillary malformation (MCAP) and megalencephalyâ€polydactylyâ€polymicrogyriaâ€hydrocephalus (MPPH) syndromes: Two closely related disorders of brain overgrowth and abnormal brain and body morphogenesis. American Journal of Medical Genetics, Part A, 2012, 158A, 269-291.	0.7	188
8	Clinical spectrum of SCN2A mutations expanding to Ohtahara syndrome. Neurology, 2013, 81, 992-998.	1.5	188
9	Fetal cytomegalovirus infection of the brain: the spectrum of sonographic findings. American Journal of Neuroradiology, 2003, 24, 28-32.	1.2	169
10	Effect of Early Corticosteroid Therapy for Landauâ€Kleffner Syndrome. Developmental Medicine and Child Neurology, 1991, 33, 257-260.	1.1	144
11	Early onset epileptic encephalopathy caused by de novo <scp>SCN</scp>8A mutations. Epilepsia, 2014, 55, 994-1000.	2.6	142
12	Deficiency of Asparagine Synthetase Causes Congenital Microcephaly and a Progressive Form of Encephalopathy. Neuron, 2013, 80, 429-441.	3.8	137
13	Mutations Disrupting Selenocysteine Formation Cause Progressive Cerebello-Cerebral Atrophy. American Journal of Human Genetics, 2010, 87, 538-544.	2.6	131
14	The fetal cerebellum. Pitfalls in diagnosis and management. Prenatal Diagnosis, 2009, 29, 372-380.	1.1	98
15	Familial neonatal seizures in 36 families: Clinical and genetic features correlate with outcome. Epilepsia, 2015, 56, 1071-1080.	2.6	94
16	White matter involvement in mitochondrial diseases. Molecular Genetics and Metabolism, 2005, 84, 127-136.	0.5	84
17	VPS53mutations cause progressive cerebello-cerebral atrophy type 2 (PCCA2). Journal of Medical Genetics, 2014, 51, 303-308.	1.5	82
18	Imaging of Fetal Cytomegalovirus Infection. Fetal Diagnosis and Therapy, 2011, 29, 117-126.	0.6	79

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19	Targeted capture and sequencing for detection of mutations causing early onset epileptic encephalopathy. Epilepsia, 2013, 54, 1262-1269.	2.6	76
20	<scp>GRIN</scp>1 mutations cause encephalopathy with infantileâ€onset epilepsy, and hyperkinetic and stereotyped movement disorders. Epilepsia, 2015, 56, 841-848.	2.6	76
21	De novo mutations in GRIN1 cause extensive bilateral polymicrogyria. Brain, 2018, 141, 698-712.	3.7	72
22	Mitochondrial Myopathy, Sideroblastic Anemia, and Lactic Acidosis: An Autosomal Recessive Syndrome in Persian Jews Caused by a Mutation in the PUS1 Gene. Journal of Child Neurology, 2005, 20, 449-452.	0.7	67
23	Does normal fetal brain ultrasound predict normal neurodevelopmental outcome in congenital cytomegalovirus infection?. Prenatal Diagnosis, 2011, 31, 360-366.	1.1	67
24	Clinical and Radiologic Correlates of Frontal Intermittent Rhythmic Delta Activity. Journal of Clinical Neurophysiology, 2002, 19, 535-539.	0.9	66
25	De novo hotspot variants in CYFIP2 cause earlyâ€onset epileptic encephalopathy. Annals of Neurology, 2018, 83, 794-806.	2.8	60
26	White matter abnormalities and dystonic motor disorder associated with mutations in the SLC16A2 gene. Developmental Medicine and Child Neurology, 2010, 52, 475-482.	1.1	58
27	Acute Pediatric Rhabdomyolysis. Journal of Child Neurology, 2000, 15, 222-227.	0.7	57
28	Prediction of microcephaly at birth using three reference ranges for fetal head circumference: can we improve prenatal diagnosis?. Ultrasound in Obstetrics and Gynecology, 2016, 47, 586-592.	0.9	55
29	NDUFS4 mutations cause Leigh syndrome with predominant brainstem involvement. Molecular Genetics and Metabolism, 2009, 97, 185-189.	0.5	54
30	Autoimmune epilepsy: Distinct subpopulations of epilepsy patients harbor serum autoantibodies to either glutamate/AMPA receptor GluR3, glutamate/NMDA receptor subunit NR2A or double-stranded DNA. Epilepsy Research, 2005, 65, 11-22.	0.8	53
31	Mutation in the AP4B1 gene cause hereditary spastic paraplegia type 47 (SPG47). Neurogenetics, 2012, 13, 73-76.	0.7	52
32	Prenatal brain imaging in congenital toxoplasmosis. Prenatal Diagnosis, 2011, 31, 881-886.	1.1	51
33	Utility of Whole Exome Sequencing for Genetic Diagnosis of Previously Undiagnosed Pediatric Neurology Patients. Journal of Child Neurology, 2016, 31, 1534-1539.	0.7	51
34	Delineating FOXG1 syndrome. Neurology: Genetics, 2018, 4, e281.	0.9	51
35	Medical Cannabis for Pediatric Moderate to Severe Complex Motor Disorders. Journal of Child Neurology, 2018, 33, 565-571.	0.7	49
36	Normal and abnormal fetal brain development during the third trimester as demonstrated by neurosonography. European Journal of Radiology, 2006, 57, 226-232.	1.2	47

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37	The molecular and phenotypic spectrum of <scp>IQSEC</scp>2â€related epilepsy. Epilepsia, 2016, 57, 1858-1869.	2.6	46
38	Paroxysmal tonic upward gaze as a presentation of de-novo mutations in CACNA1A. European Journal of Paediatric Neurology, 2015, 19, 292-297.	0.7	44
39	Congenital Ataxia, Mental Retardation, and Dyskinesia Associated With a Novel CACNA1A Mutation. Journal of Child Neurology, 2010, 25, 892-897.	0.7	43
40	Sulthiame in childhood epilepsy. Pediatrics International, 2004, 46, 521-524.	0.2	42
41	A compound heterozygous missense mutation and a large deletion in the KCTD7 gene presenting as an opsoclonus-myoclonus ataxia-like syndrome. Journal of Neurology, 2012, 259, 2590-2598.	1.8	42
42	Absence Seizures Aggravated by Valproic Acid. Epilepsia, 2001, 42, 941-943.	2.6	41
43	Infantile Onset of Hereditary Ascending Spastic Paralysis With Bulbar Involvement. Journal of Child Neurology, 1996, 11, 54-57.	0.7	40
44	Adding Video Recording Increases the Diagnostic Yield of Routine Electroencephalograms in Children with Frequent Paroxysmal Events. Epilepsia, 2005, 46, 716-719.	2.6	40
45	Developmental Outcome of Children With Enlargement of the Cisterna Magna Identified in Utero. Journal of Child Neurology, 2009, 24, 1486-1492.	0.7	40
46	Nonâ€visualization of the cavum septi pellucidi is not synonymous with agenesis of the corpus callosum. Ultrasound in Obstetrics and Gynecology, 2012, 40, 165-170.	0.9	39
47	Prenatal Brain Disruption in Molybdenum Cofactor Deficiency. Journal of Child Neurology, 2011, 26, 460-464.	0.7	37
48	The Spinal Muscular Atrophy with Pontocerebellar Hypoplasia GeneVRK1Regulates Neuronal Migration through an Amyloid-Î² Precursor Protein-Dependent Mechanism. Journal of Neuroscience, 2015, 35, 936-942.	1.7	36
49	Prenatal exclusion of Leigh syndrome due to T8993C mutation in the mitochondrial DNA. Prenatal Diagnosis, 2003, 23, 31-33.	1.1	35
50	Autoimmune Epilepsy: Some Epilepsy Patients Harbor Autoantibodies to Glutamate Receptors and dsDNA on both Sides of the Blood-brain Barrier, which may Kill Neurons and Decrease in Brain Fluids after Hemispherotomy. Clinical and Developmental Immunology, 2004, 11, 241-252.	3.3	35
51	A large homozygous deletion in the SAMHD1 gene causes atypical Aicardiâ€“GoutiÃ©res syndrome associated with mtDNA deletions. European Journal of Human Genetics, 2011, 19, 287-292.	1.4	35
52	Heterozygous Mutations in the ADCK3 Gene in Siblings with Cerebellar Atrophy and Extreme Phenotypic Variability. JIMD Reports, 2013, 12, 103-107.	0.7	35
53	Clinical Correlates of Occipital Intermittent Rhythmic Delta Activity (OIRDA) in Children. Epilepsia, 2007, 48, 330-334.	2.6	34
54	Significant overlap and possible identity of macrocephaly capillary malformation and megalencephaly polymicrogyriaâ€polydactyly hydrocephalus syndromes. American Journal of Medical Genetics, Part A, 2009, 149A, 868-876.	0.7	34

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55	Expansion of the spectrum of TUBB4A-related disorders: a new phenotype associated with a novel mutation in the TUBB4A gene. Neurogenetics, 2014, 15, 107-13.	0.7	34
56	Methylenetetrahydrofolate Reductase Deficiency: Importance of Early Diagnosis. Journal of Child Neurology, 2000, 15, 539-543.	0.7	33
57	Pyrimethamine increases Î²-hexosaminidase A activity in patients with Late Onset Tay Sachs. Molecular Genetics and Metabolism, 2011, 102, 356-363.	0.5	33
58	Effect of cyclic, low dose pyrimethamine treatment in patients with Late Onset Tay Sachs: an open label, extended pilot study. Orphanet Journal of Rare Diseases, 2015, 10, 45.	1.2	33
59	Rare GABRA3 variants are associated with epileptic seizures, encephalopathy and dysmorphic features. Brain, 2017, 140, 2879-2894.	3.7	33
60	A new locus (SPG47) maps to 1p13.2â€“1p12 in an Arabic family with complicated autosomal recessive hereditary spastic paraplegia and thin corpus callosum. Journal of the Neurological Sciences, 2011, 305, 67-70.	0.3	31
61	Fetal cerebellar disorders. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 155, 3-23.	1.0	31
62	Should Autistic Children Be Evaluated for Mitochondrial Disorders?. Journal of Child Neurology, 2004, 19, 379-381.	0.7	30
63	Technetium 99m Ethylcysteinate Dimer Single-Photon Emission Computed Tomography (SPECT) During Intellectual Stress Test in Children and Adolescents With Pure Versus Comorbid Attention-Deficit Hyperactivity Disorder (ADHD). Journal of Child Neurology, 2004, 19, 91-96.	0.7	30
64	Neurologic Involvement in a Child With Systemic Capillary Leak Syndrome. Pediatrics, 2010, 125, e687-e692.	1.0	29
65	Assessment of fetal midbrain and hindbrain in midâ€sagittal cranial plane by threeâ€dimensional multiplanar sonography. Part 1: comparison of new and established nomograms. Ultrasound in Obstetrics and Gynecology, 2014, 44, 575-580.	0.9	28
66	Paternal germline mosaicism of a SCN2A mutation results in Ohtahara syndrome in half siblings. European Journal of Paediatric Neurology, 2014, 18, 567-571.	0.7	28
67	Costeff syndrome: clinical features and natural history. Journal of Neurology, 2014, 261, 2275-2282.	1.8	26
68	Malformations of Cortical Development: From Postnatal to Fetal Imaging. Canadian Journal of Neurological Sciences, 2016, 43, 611-618.	0.3	26
69	Common Peroneal Neuropathy due to Surfing. Journal of Child Neurology, 2000, 15, 420-421.	0.7	25
70	RARS2 mutations cause early onset epileptic encephalopathy without ponto-cerebellar hypoplasia. European Journal of Paediatric Neurology, 2016, 20, 412-417.	0.7	25
71	The use of fetal neurosonography and brain <scp>MRI</scp> in cases of cytomegalovirus infection during pregnancy: A retrospective analysis with outcome correlation. Prenatal Diagnosis, 2017, 37, 1335-1342.	1.1	25
72	Assessment of fetal intracranial pathologies first demonstrated late in pregnancy: cell proliferation disorders. Reproductive Biology and Endocrinology, 2003, 1, 110.	1.4	24

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73	The Fetal vermis, pons and brainstem: normal longitudinal development as shown by dedicated neurosonography. Journal of Maternal-Fetal and Neonatal Medicine, 2013, 26, 757-762.	0.7	24
74	Multiplex families with epilepsy. Neurology, 2016, 86, 713-722.	1.5	23
75	Frequency of <scp>CNKSR</scp>2 mutation in the Xâ€linked epilepsyâ€aphasia spectrum. Epilepsia, 2017, 58, e40-e43.	2.6	23
76	Demyelinative Brainstem Encephalitis Responsive to Intravenous Immunoglobulin Therapy. Pediatrics, 1999, 104, 301-303.	1.0	22
77	Fatal outcome following foetal cerebellar haemorrhage associated with placental thrombosis. European Journal of Paediatric Neurology, 2006, 10, 93-96.	0.7	22
78	Does a SCN1A gene mutation confer earlier age of onset of febrile seizures in GEFS+?. Epilepsia, 2009, 50, 953-956.	2.6	22
79	Leigh disease presenting in utero due to a novel missense mutation in the mitochondrial DNAâ€“ND3. Molecular Genetics and Metabolism, 2010, 100, 65-70.	0.5	22
80	Samaritan myopathy, an ultimately benign congenital myopathy, is caused by a RYR1 mutation. Acta Neuropathologica, 2012, 124, 575-581.	3.9	22
81	Thick corpus callosum in the second trimester can be transient and is of uncertain significance. Ultrasound in Obstetrics and Gynecology, 2016, 48, 452-457.	0.9	22
82	Juvenile Leigh Syndrome, Optic Atrophy, Ataxia, Dystonia, and Epilepsy due to T14487C Mutation in the mtDNA-ND6 Gene: A Mitochondrial Syndrome Presenting From Birth to Adolescence. Journal of Child Neurology, 2011, 26, 476-481.	0.7	21
83	Primary and maternal 3â€methylcrotonylâ€CoA carboxylase deficiency: insights from the Israel newborn screening program. Journal of Inherited Metabolic Disease, 2016, 39, 211-217.	1.7	21
84	Can syndromic macrocephaly be diagnosed in utero?. Ultrasound in Obstetrics and Gynecology, 2011, 37, 72-81.	0.9	20
85	Israeli Children With Autism Spectrum Disorder Are not Macrocephalic. Journal of Child Neurology, 2011, 26, 580-585.	0.7	20
86	Neonatal seizures associated with a severe neonatal myoclonus like dyskinesia due to a familial KCNQ2 gene mutation. European Journal of Paediatric Neurology, 2012, 16, 356-360.	0.7	20
87	Progressive cerebello-cerebral atrophy and progressive encephalopathy with edema, hypsarrhythmia and optic atrophy may be allelic syndromes. European Journal of Paediatric Neurology, 2018, 22, 1133-1138.	0.7	20
88	Syndrome of osteopetrosis and muscular degeneration associated with cerebro-oculo-facio-skeletal changes. American Journal of Medical Genetics Part A, 1987, 28, 137-142.	2.4	19
89	Primary Disorders of Metabolism and Disturbed Fetal Brain Development. Clinics in Perinatology, 2009, 36, 621-638.	0.8	19
90	Delineation of the interstitial 6q25 microdeletion syndrome: Refinement of the critical causative region. American Journal of Medical Genetics, Part A, 2012, 158A, 1395-1399.	0.7	19

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91	Fetal Central Nervous System: MR Imaging versus Dedicated USâ€"Need for Prospective, Blind, Comparative Studies [letter]. Radiology, 2004, 232, 306-307.	3.6	16
92	Familial partial trisomy 15q11-13 presenting as intractable epilepsy in the child and schizophrenia in the mother. European Journal of Paediatric Neurology, 2011, 15, 230-233.	0.7	16
93	Expanding the phenotype of IBA57 mutations: related leukodystrophy can remain asymptomatic. Journal of Human Genetics, 2018, 63, 1223-1229.	1.1	16
94	A practical approach to prenatal diagnosis of malformations of cortical development. European Journal of Paediatric Neurology, 2021, 34, 50-61.	0.7	16
95	Fetal Brain Development: Regulating Processes and Related Malformations. Life, 2022, 12, 809.	1.1	16
96	Sulthiame Revisited. Journal of Child Neurology, 1995, 10, 241-242.	0.7	15
97	Assessment of fetal midbrain and hindbrain in midâ€sagittal cranial plane by threeâ€dimensional multiplanar sonography. Part 2: application of nomograms to fetuses with posterior fossa malformations. Ultrasound in Obstetrics and Gynecology, 2014, 44, 581-587.	0.9	15
98	Walker-Warburg syndrome and tectocerebellar dysraphia: A novel association caused by a homozygous DAG1 mutation. European Journal of Paediatric Neurology, 2018, 22, 525-531.	0.7	15
99	The cerebellar â€œtilted telephone receiver signâ€•enables prenatal diagnosis of PHACES syndrome. European Journal of Paediatric Neurology, 2018, 22, 900-909.	0.7	15
100	Metabolic stroke in a patient with bi-allelic OPA1 mutations. Metabolic Brain Disease, 2019, 34, 1043-1048.	1.4	15
101	Fetal optic nerve sheath measurement as a nonâ€invasive tool for assessment of increased intracranial pressure. Ultrasound in Obstetrics and Gynecology, 2011, 38, 646-651.	0.9	14
102	Novel WWOX deleterious variants cause early infantile epileptic encephalopathy, severe developmental delay and dysmorphism among Yemenite Jews. European Journal of Paediatric Neurology, 2019, 23, 418-426.	0.7	14
103	Clinical Correlates of Frontal Intermittent Rhythmic Delta Activity in Children. Journal of Child Neurology, 2003, 18, 525-529.	0.7	13
104	Recurrent Absence Status Epilepticus (Spike-and-Wave Stupor) Associated With Lamotrigine Therapy. Journal of Child Neurology, 2006, 21, 807-809.	0.7	13
105	Agenesis of the corpus callosum. An autopsy study in fetuses. Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin, 2016, 468, 219-230.	1.4	13
106	Diagnostic approach to fetal microcephaly. European Journal of Paediatric Neurology, 2018, 22, 935-943.	0.7	13
107	Prenatal and postnatal presentation of PRMT7 related syndrome: Expanding the phenotypic manifestations. American Journal of Medical Genetics, Part A, 2019, 179, 78-84.	0.7	13
108	Clinical phenotypes of infantile onset CACNA1A-related disorder. European Journal of Paediatric Neurology, 2021, 30, 144-154.	0.7	13

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109	New findings in a patient with dubowitz syndrome: Velopharyngeal insufficiency and hypoparathyroidism. American Journal of Medical Genetics Part A, 1990, 37, 241-243.	2.4	12
110	Phenobarbital Still Has a Role in Epilepsy Treatment. Journal of Child Neurology, 1999, 14, 820-821.	0.7	12
111	The subarachnoid space: normal fetal development as demonstrated by transvaginal ultrasound. Prenatal Diagnosis, 2000, 20, 890-893.	1.1	12
112	Neurological involvement in a child with the wrinkly skin syndrome. , 1999, 82, 31-33.		11
113	Infantile onset progressive cerebellar atrophy and anterior horn cell degenerationâ€"A late onset variant of PCH-1?. European Journal of Paediatric Neurology, 2008, 12, 97-101.	0.7	11
114	Expanding the phenotype of TRAK1 mutations: hyperekplexia and refractory status epilepticus. Brain, 2018, 141, e55-e55.	3.7	11
115	Prenatal diagnosis of brainstem anomalies. European Journal of Paediatric Neurology, 2018, 22, 1016-1026.	0.7	11
116	Autosomal dominant TUBB3-related syndrome: Fetal, radiologic, clinical and morphological features. European Journal of Paediatric Neurology, 2020, 26, 46-60.	0.7	11
117	Low Erythrocyte Zinc Content in Acquired Aphasia With Convulsive Disorder (Landau-Kleffner) Tj ETQq1 1 0.784314 rgBT /Overlock 10	0.7	10
118	Molecular and functional studies of retinal degeneration as a clinical presentation of SACS-related disorder. European Journal of Paediatric Neurology, 2015, 19, 472-476.	0.7	10
119	The â€˜Brain Shadowing Sign': A Novel Marker of Fetal Craniosynostosis. Fetal Diagnosis and Therapy, 2016, 40, 277-284.	0.6	10
120	Multiple Causes of Pediatric Early Onset Choreaâ€"Clinical and Genetic Approach. Neuropediatrics, 2018, 49, 246-255.	0.3	10
121	Periventricular Brain Heterotopias in a Child With Adrenocortical Insufficiency, Achalasia, Alacrima, and Neurologic Abnormalities (Allgrove Syndrome). Journal of Child Neurology, 1999, 14, 331-334.	0.7	9
122	Familial Hyperekplexia and Refractory Status Epilepticus: A New Autosomal Recessive Syndrome. Journal of Child Neurology, 2004, 19, 522-525.	0.7	9
123	A benign congenital myopathy in an inbred Samaritan family. European Journal of Paediatric Neurology, 2006, 10, 182-185.	0.7	9
124	Hepatic coma culminating in severe brain damage in a child with a SCN1A mutation. European Journal of Paediatric Neurology, 2010, 14, 456-459.	0.7	9
125	Severe growth deficiency, microcephaly, intellectual disability, and characteristic facial features are due to a homozygous QARS mutation. Neurogenetics, 2017, 18, 141-146.	0.7	9
126	Familial optic atrophy with white matter changes. American Journal of Medical Genetics Part A, 2003, 121A, 263-265.	2.4	8

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127	Acute Intermittent Porphyria, Rasmussen Encephalitis, or Both?. Journal of Child Neurology, 2007, 22, 99-105.	0.7	8
128	Diagnostic Yield of Electroencephalograms in Infants and Young Children With Frequent Paroxysmal Eye Movements. Journal of Child Neurology, 2008, 23, 620-623.	0.7	8
129	Resolution of epileptic encephalopathy following treatment with transdermal nicotine. Epilepsia, 2013, 54, e13-5.	2.6	8
130	Atypical presentation of Costeff syndrome-severe psychomotor involvement and electrical status epilepticus during slow wave sleep. European Journal of Paediatric Neurology, 2015, 19, 733-736.	0.7	8
131	Application of a novel prenatal vertical cranial biometric measurement can improve accuracy of microcephaly diagnosisin utero. Ultrasound in Obstetrics and Gynecology, 2016, 47, 593-599.	0.9	8
132	Fourth ventricle index: sonographic marker for severe fetal vermian dysgenesis/agenesis. Ultrasound in Obstetrics and Gynecology, 2019, 53, 390-395.	0.9	8
133	Carbamazepine versus sulthiame in treating benign childhood epilepsy with centrotemporal spikes. Journal of Child Neurology, 2002, 17, 914-6.	0.7	8
134	Genetic Counseling and Testing for FSHD (Facioscapulohumeral Muscular Dystrophy) in the Israeli Population. Journal of Genetic Counseling, 2012, 21, 557-563.	0.9	7
135	Diagnosis by whole exome sequencing of atypical infantile onset Alexander disease masquerading as a mitochondrial disorder. European Journal of Paediatric Neurology, 2014, 18, 495-501.	0.7	7
136	Familial Intracranial Hypertension in 2 Brothers WithPTENMutation: Expansion of the Phenotypic Spectrum. Journal of Child Neurology, 2019, 34, 506-510.	0.7	7
137	Brain white matter abnormalities associated with copy number variants. American Journal of Medical Genetics, Part A, 2020, 182, 93-103.	0.7	7
138	Expanding the genotype-phenotype spectrum of ISCA2-related multiple mitochondrial dysfunction syndrome-cavitating leukoencephalopathy and prolonged survival. Neurogenetics, 2020, 21, 243-249.	0.7	7
139	Agenesis of the septum pellucidum: Prenatal diagnosis and outcome. Prenatal Diagnosis, 2020, 40, 674-680.	1.1	7
140	Congenital Mirror Movements Associated With Brain Malformations. Journal of Child Neurology, 2021, 36, 545-555.	0.7	7
141	Mosaic marker chromosome 16 resulting in 16q11.2â€“q12.1 gain in a child with intellectual disability, microcephaly, and cerebellar cortical dysplasia. American Journal of Medical Genetics, Part A, 2011, 155, 2991-2996.	0.7	6
142	Fetal pericallosal lipomas â€“ Clues to diagnosis in the second trimester. European Journal of Paediatric Neurology, 2018, 22, 929-934.	0.7	6
143	The role of orotic acid measurement in routine newborn screening for urea cycle disorders. Journal of Inherited Metabolic Disease, 2021, 44, 606-617.	1.7	6
144	White matter abnormalities and iron deposition in prenatal mucolipidosis IV- fetal imaging and pathology. Metabolic Brain Disease, 2021, 36, 2155-2167.	1.4	6

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145	Muscle Glycogen Depletion and Increased Oxidative Phosphorylation Following Status Epilepticus. Journal of Child Neurology, 2003, 18, 876-878.	0.7	5
146	The Differential Diagnosis of Fetal Intracranial Cystic Lesions. Ultrasound Clinics, 2008, 3, 553-558.	0.2	5
147	A novel mutation in the TPR6 domain of the RAPSN gene associated with congenital myasthenic syndrome. Journal of the Neurological Sciences, 2012, 316, 112-115.	0.3	5
148	Refractory epilepsy associated with ventriculoperitoneal shunt over-drainage: case report. Child's Nervous System, 2019, 35, 2411-2416.	0.6	5
149	Periventricular pseudocysts of noninfectious origin: Prenatal associated findings and prognostic factors. Prenatal Diagnosis, 2020, 40, 931-941.	1.1	5
150	Familial Brain Periventricular Pseudocysts. Fetal Diagnosis and Therapy, 2017, 42, 42-47.	0.6	4
151	Unique Imaging Features Enabling the Prenatal Diagnosis of Developmental Venous Anomalies: A Persistent Echogenic Brain Lesion Drained by a Collecting Vein in Contrast with Normal Brain Parenchyma on MRI. Fetal Diagnosis and Therapy, 2018, 43, 53-60.	0.6	4
152	Ultrasound Nomograms of the Fetal Optic Nerve Sheath Diameter. Ultraschall in Der Medizin, 2019, 40, 476-480.	0.8	4
153	Refining the phenotype of the THG1L (p.Val55Ala mutation)â€related mitochondrial autosomal recessive congenital cerebellar ataxia. American Journal of Medical Genetics, Part A, 2019, 179, 1575-1579.	0.7	4
154	Myoclonic tremor status as a presenting symptom of adenylosuccinate lyase deficiency. European Journal of Medical Genetics, 2020, 63, 104061.	0.7	4
155	Central Apnea in a Child With Congenital Autonomic Dysfunction and Universal Pain Loss. Journal of Child Neurology, 1996, 11, 162-164.	0.7	3
156	A new autosomal recessive syndrome with Zellweger-like manifestations. American Journal of Medical Genetics Part A, 2003, 119A, 352-355.	2.4	3
157	Dominantly Inherited Nonprogressive Cerebellar Hypoplasia Identified in Utero. Journal of Child Neurology, 2012, 27, 1000-1003.	0.7	3
158	Autistic regression in a child with Silverâ€“Russell Syndrome and maternal UPD 7. European Journal of Paediatric Neurology, 2012, 16, 95-98.	0.7	3
159	Fetal Neurology. European Journal of Paediatric Neurology, 2018, 22, 895-897.	0.7	3
160	Building Bridges Between the Clinic and the Laboratory: A Meeting Review â€“ Brain Malformations: A Roadmap for Future Research. Frontiers in Cellular Neuroscience, 2019, 13, 434.	1.8	3
161	CHAMP1 Mutations cause Refractory Infantile Myoclonic Epilepsy. Journal of Pediatric Neurology, 2020, 18, 027-032.	0.0	3
162	Delineation of the phenotype of MED17-related disease in Caucasus-Jewish families. European Journal of Paediatric Neurology, 2021, 32, 40-45.	0.7	3

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163	Nonprogressive Familial Leukoencephalopathy With Porencephalic Cyst and Focal Seizures. Journal of Child Neurology, 2006, 21, 145-148.	0.7	2
164	A Possible Genotype-Phenotype Correlation in Ashkenazi-Jewish Individuals With Aicardi-GoutiÃ¨res Syndrome Associated With SAMHD1 Mutation. Journal of Child Neurology, 2015, 30, 490-495.	0.7	2
165	Infantile onset progressive cerebellar atrophy and anterior horn cell Degeneration-A novel phenotype associated with mutations in the PLA2G6 gene. European Journal of Medical Genetics, 2020, 63, 103801.	0.7	2
166	Major brain malformations: corpus callosum dysgenesis, agenesis of septum pellucidum and polymicrogyria in patients with BCORL1-related disorders. Journal of Human Genetics, 2022, 67, 95-101.	1.1	2
167	Fat Intolerance in Developmentally Impaired Children With Severe Feeding Intolerance. Journal of Child Neurology, 2006, 21, 167-170.	0.7	1
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