Tally Lerman-Sagie

List of Publications by Citations

178 papers	5,578 citations	35 h-index	69 g-index
185 ext. papers	6,471 ext. citations	4.5 avg, IF	5.01 L-index

#	Paper	IF	Citations
178	De novo germline and postzygotic mutations in AKT3, PIK3R2 and PIK3CA cause a spectrum of related megalencephaly syndromes. *Nature Genetics*, **2012**, 44, 934-40	36.3	521
177	Targeted resequencing in epileptic encephalopathies identifies de novo mutations in CHD2 and SYNGAP1. *Nature Genetics*, **2013**, 45, 825-30	36.3	500
176	X-linked protocadherin 19 mutations cause female-limited epilepsy and cognitive impairment. *Nature Genetics*, **2008**, 40, 776-81	36.3	328
175	De Novo Mutations in SLC1A2 and CACNA1A Are Important Causes of Epileptic Encephalopathies. *American Journal of Human Genetics*, **2016**, 99, 287-98	11	180
174	Rare copy number variants are an important cause of epileptic encephalopathies. *Annals of Neurology*, **2011**, 70, 974-85	9.4	176
173	Clinical spectrum of SCN2A mutations expanding to Ohtahara syndrome. *Neurology*, **2013**, 81, 992-8	6.5	158
172	Megalencephaly-capillary malformation (MCAP) and megalencephaly-polydactyly-polymicrogyria-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, 269-91	2.5	154
171	Mortality in Dravet syndrome. *Epilepsy Research*, **2016**, 128, 43-47	3	152
170	Fetal cytomegalovirus infection of the brain: the spectrum of sonographic findings. *American Journal of Neuroradiology*, **2003**, 24, 28-32	4.4	139
169	Early onset epileptic encephalopathy caused by de novo SCN8A mutations. *Epilepsia*, **2014**, 55, 994-1000	6.4	122
168	Effect of early corticosteroid therapy for Landau-Kleffner syndrome. *Developmental Medicine and Child Neurology*, **1991**, 33, 257-60	3.3	118
167	Mutations disrupting selenocysteine formation cause progressive cerebello-cerebral atrophy. *American Journal of Human Genetics*, **2010**, 87, 538-44	11	111
166	Deficiency of asparagine synthetase causes congenital microcephaly and a progressive form of encephalopathy. *Neuron*, **2013**, 80, 429-41	13.9	100
165	The fetal cerebellum. Pitfalls in diagnosis and management. *Prenatal Diagnosis*, **2009**, 29, 372-80	3.2	79
164	White matter involvement in mitochondrial diseases. *Molecular Genetics and Metabolism*, **2005**, 84, 127-35	3.7	74
163	Familial neonatal seizures in 36 families: Clinical and genetic features correlate with outcome. *Epilepsia*, **2015**, 56, 1071-80	6.4	68
162	Imaging of fetal cytomegalovirus infection. *Fetal Diagnosis and Therapy*, **2011**, 29, 117-26	2.4	66

161	Targeted capture and sequencing for detection of mutations causing early onset epileptic encephalopathy. *Epilepsia*, **2013**, 54, 1262-9	6.4	64
160	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, 449-52	2.5	58
159	GRIN1 mutations cause encephalopathy with infantile-onset epilepsy, and hyperkinetic and stereotyped movement disorders. *Epilepsia*, **2015**, 56, 841-8	6.4	56
158	VPS53 mutations cause progressive cerebello-cerebral atrophy type 2 (PCCA2). *Journal of Medical Genetics*, **2014**, 51, 303-8	5.8	53
157	Clinical and radiologic correlates of frontal intermittent rhythmic delta activity. *Journal of Clinical Neurophysiology*, **2002**, 19, 535-9	2.2	52
156	Does normal fetal brain ultrasound predict normal neurodevelopmental outcome in congenital cytomegalovirus infection?. *Prenatal Diagnosis*, **2011**, 31, 360-6	3.2	49
155	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	3	48
154	Acute pediatric rhabdomyolysis. *Journal of Child Neurology*, **2000**, 15, 222-7	2.5	48
153	De novo mutations in GRIN1 cause extensive bilateral polymicrogyria. *Brain*, **2018**, 141, 698-712	11.2	46
152	NDUFS4 mutations cause Leigh syndrome with predominant brainstem involvement. *Molecular Genetics and Metabolism*, **2009**, 97, 185-9	3.7	44
151	Mutation in the AP4B1 gene cause hereditary spastic paraplegia type 47 (SPG47). *Neurogenetics*, **2012**, 13, 73-6	3	43
150	White matter abnormalities and dystonic motor disorder associated with mutations in the SLC16A2 gene. *Developmental Medicine and Child Neurology*, **2010**, 52, 475-82	3.3	43
149	Normal and abnormal fetal brain development during the third trimester as demonstrated by neurosonography. *European Journal of Radiology*, **2006**, 57, 226-32	4.7	42
148	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, 586-92	5.8	39
147	Utility of Whole Exome Sequencing for Genetic Diagnosis of Previously Undiagnosed Pediatric Neurology Patients. *Journal of Child Neurology*, **2016**, 31, 1534-1539	2.5	38
146	Congenital ataxia, mental retardation, and dyskinesia associated with a novel CACNA1A mutation. *Journal of Child Neurology*, **2010**, 25, 892-7	2.5	38
145	The molecular and phenotypic spectrum of IQSEC2-related epilepsy. *Epilepsia*, **2016**, 57, 1858-1869	6.4	38
144	De novo hotspot variants in CYFIP2 cause early-onset epileptic encephalopathy. *Annals of Neurology*, **2018**, 83, 794-806	9.4	37

143	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, 2590-8	5.5	34
142	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, 241-52		34
141	Paroxysmal tonic upward gaze as a presentation of de-novo mutations in CACNA1A. *European Journal of Paediatric Neurology*, **2015**, 19, 292-7	3.8	32
140	Pyrimethamine increases hexosaminidase A activity in patients with Late Onset Tay Sachs. *Molecular Genetics and Metabolism*, **2011**, 102, 356-63	3.7	32
139	Sulthiame in childhood epilepsy. *Pediatrics International*, **2004**, 46, 521-4	1.2	32
138	Adding video recording increases the diagnostic yield of routine electroencephalograms in children with frequent paroxysmal events. *Epilepsia*, **2005**, 46, 716-9	6.4	32
137	Infantile onset of hereditary ascending spastic paralysis with bulbar involvement. *Journal of Child Neurology*, **1996**, 11, 54-7	2.5	32
136	Developmental outcome of children with enlargement of the cisterna magna identified in utero. *Journal of Child Neurology*, **2009**, 24, 1486-92	2.5	31
135	Significant overlap and possible identity of macrocephaly capillary malformation and megalencephaly polymicrogyria-polydactyly hydrocephalus syndromes. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 868-76	2.5	31
134	Non-visualization of the cavum septi pellucidi is not synonymous with agenesis of the corpus callosum. *Ultrasound in Obstetrics and Gynecology*, **2012**, 40, 165-70	5.8	30
133	Prenatal brain disruption in molybdenum cofactor deficiency. *Journal of Child Neurology*, **2011**, 26, 460-4	2.5	30
132	A large homozygous deletion in the SAMHD1 gene causes atypical Aicardi-Goutières syndrome associated with mtDNA deletions. *European Journal of Human Genetics*, **2011**, 19, 287-92	5.3	29
131	Prenatal brain imaging in congenital toxoplasmosis. *Prenatal Diagnosis*, **2011**, 31, 881-6	3.2	29
130	Prenatal exclusion of Leigh syndrome due to T8993C mutation in the mitochondrial DNA. *Prenatal Diagnosis*, **2003**, 23, 31-3	3.2	29
129	Delineating syndrome: From congenital microcephaly to hyperkinetic encephalopathy. *Neurology: Genetics*, **2018**, 4, e281	3.8	29
128	A new locus (SPG47) maps to 1p13.2-1p12 in an Arabic family with complicated autosomal recessive hereditary spastic paraplegia and thin corpus callosum. *Journal of the Neurological Sciences*, **2011**, 305, 67-70	3.2	28
127	Absence seizures aggravated by valproic acid. *Epilepsia*, **2001**, 42, 941-3	6.4	28
126	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	3	26

125	Heterozygous Mutations in the ADCK3 Gene in Siblings with Cerebellar Atrophy and Extreme Phenotypic Variability. *JIMD Reports*, **2014**, 12, 103-7	1.9	26
124	Methylenetetrahydrofolate reductase deficiency: importance of early diagnosis. *Journal of Child Neurology*, **2000**, 15, 539-43	2.5	26
123	Medical Cannabis for Pediatric Moderate to Severe Complex Motor Disorders. *Journal of Child Neurology*, **2018**, 33, 565-571	2.5	26
122	Clinical correlates of occipital intermittent rhythmic delta activity (OIRDA) in children. *Epilepsia*, **2007**, 48, 330-4	6.4	25
121	Should autistic children be evaluated for mitochondrial disorders?. *Journal of Child Neurology*, **2004**, 19, 379-81	2.5	25
120	The spinal muscular atrophy with pontocerebellar hypoplasia gene VRK1 regulates neuronal migration through an amyloid-precursor protein-dependent mechanism. *Journal of Neuroscience*, **2015**, 35, 936-42	6.6	24
119	Rare GABRA3 variants are associated with epileptic seizures, encephalopathy and dysmorphic features. *Brain*, **2017**, 140, 2879-2894	11.2	24
118	Paternal germline mosaicism of a SCN2A mutation results in Ohtahara syndrome in half siblings. *European Journal of Paediatric Neurology*, **2014**, 18, 567-71	3.8	23
117	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	4.2	23
116	Neurologic involvement in a child with systemic capillary leak syndrome. *Pediatrics*, **2010**, 125, e687-92	7.4	23
115	Multiplex families with epilepsy: Success of clinical and molecular genetic characterization. *Neurology*, **2016**, 86, 713-22	6.5	22
114	Fatal outcome following foetal cerebellar haemorrhage associated with placental thrombosis. *European Journal of Paediatric Neurology*, **2006**, 10, 93-6	3.8	22
113	Assessment of fetal intracranial pathologies first demonstrated late in pregnancy: cell proliferation disorders. *Reproductive Biology and Endocrinology*, **2003**, 1, 110	5	21
112	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, 91-6	2.5	21
111	The fetal vermis, pons and brainstem: normal longitudinal development as shown by dedicated neurosonography. *Journal of Maternal-Fetal and Neonatal Medicine*, **2013**, 26, 757-62	2	20
110	Costeff syndrome: clinical features and natural history. *Journal of Neurology*, **2014**, 261, 2275-82	5.5	20
109	Common peroneal neuropathy due to surfing. *Journal of Child Neurology*, **2000**, 15, 420-1	2.5	19
108	Demyelinative brainstem encephalitis responsive to intravenous immunoglobulin therapy. *Pediatrics*, **1999**, 104, 301-3	7.4	19

107	RARS2 mutations cause early onset epileptic encephalopathy without ponto-cerebellar hypoplasia. *European Journal of Paediatric Neurology*, **2016**, 20, 412-7	3.8	19
106	Malformations of Cortical Development: From Postnatal to Fetal Imaging. *Canadian Journal of Neurological Sciences*, **2016**, 43, 611-8	1	18
105	Fetal cerebellar disorders. *Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn*, **2018**, 155, 3-23	3	18
104	Leigh disease presenting in utero due to a novel missense mutation in the mitochondrial DNA-ND3. *Molecular Genetics and Metabolism*, **2010**, 100, 65-70	3.7	18
103	Does a SCN1A gene mutation confer earlier age of onset of febrile seizures in GEFS+?. *Epilepsia*, **2009**, 50, 953-6	6.4	17
102	Israeli children with autism spectrum disorder are not macrocephalic. *Journal of Child Neurology*, **2011**, 26, 580-5	2.5	17
101	Samaritan myopathy, an ultimately benign congenital myopathy, is caused by a RYR1 mutation. *Acta Neuropathologica*, **2012**, 124, 575-81	14.3	16
100	Delineation of the interstitial 6q25 microdeletion syndrome: refinement of the critical causative region. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 1395-9	2.5	16
99	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, 476-81	2.5	16
98	Primary disorders of metabolism and disturbed fetal brain development. *Clinics in Perinatology*, **2009**, 36, 621-38	2.8	16
97	Syndrome of osteopetrosis and muscular degeneration associated with cerebro-oculo-facio-skeletal changes. *American Journal of Medical Genetics Part A*, **1987**, 28, 137-42		16
96	Thick corpus callosum in the second trimester can be transient and is of uncertain significance. *Ultrasound in Obstetrics and Gynecology*, **2016**, 48, 452-457	5.8	16
95	Frequency of CNKSR2 mutation in the X-linked epilepsy-aphasia spectrum. *Epilepsia*, **2017**, 58, e40-e43	6.4	15
94	Primary and maternal 3-methylcrotonyl-CoA carboxylase deficiency: insights from the Israel newborn screening program. *Journal of Inherited Metabolic Disease*, **2016**, 39, 211-7	5.4	15
93	The use of fetal neurosonography and brain MRI in cases of cytomegalovirus infection during pregnancy: A retrospective analysis with outcome correlation. *Prenatal Diagnosis*, **2017**, 37, 1335-1342	3.2	15
92	Sulthiame revisited. *Journal of Child Neurology*, **1995**, 10, 241-2	2.5	15
91	Assessment of fetal midbrain and hindbrain in mid-sagittal cranial plane by three-dimensional multiplanar sonography. Part 1: comparison of new and established nomograms. *Ultrasound in Obstetrics and Gynecology*, **2014**, 44, 575-80	5.8	13
90	Can syndromic macrocephaly be diagnosed in utero?. *Ultrasound in Obstetrics and Gynecology*, **2011**, 37, 72-81	5.8	13

89	Familial partial trisomy 15q11-13 presenting as intractable epilepsy in the child and schizophrenia in the mother. *European Journal of Paediatric Neurology*, **2011**, 15, 230-3	3.8	12
88	Fetal optic nerve sheath measurement as a non-invasive tool for assessment of increased intracranial pressure. *Ultrasound in Obstetrics and Gynecology*, **2011**, 38, 646-51	5.8	12
87	Fetal central nervous system: MR imaging versus dedicated US--need for prospective, blind, comparative studies. *Radiology*, **2004**, 232, 306; author reply 306-7	20.5	12
86	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	3.8	12
85	Metabolic stroke in a patient with bi-allelic OPA1 mutations. *Metabolic Brain Disease*, **2019**, 34, 1043-1048	3.9	11
84	Neonatal seizures associated with a severe neonatal myoclonus like dyskinesia due to a familial KCNQ2 gene mutation. *European Journal of Paediatric Neurology*, **2012**, 16, 356-60	3.8	11
83	Recurrent absence status epilepticus (spike-and-wave stupor) associated with lamotrigine therapy. *Journal of Child Neurology*, **2006**, 21, 807-9	2.5	11
82	Clinical correlates of frontal intermittent rhythmic delta activity in children. *Journal of Child Neurology*, **2003**, 18, 525-9	2.5	11
81	The subarachnoid space: normal fetal development as demonstrated by transvaginal ultrasound. *Prenatal Diagnosis*, **2000**, 20, 890-3	3.2	11
80	Neurological involvement in a child with the wrinkly skin syndrome **1999**, 82, 31-33		11
79	Expanding the phenotype of IBA57 mutations: related leukodystrophy can remain asymptomatic. *Journal of Human Genetics*, **2018**, 63, 1223-1229	4.3	11
78	Assessment of fetal midbrain and hindbrain in mid-sagittal cranial plane by three-dimensional multiplanar sonography. Part 2: application of nomograms to fetuses with posterior fossa malformations. *Ultrasound in Obstetrics and Gynecology*, **2014**, 44, 581-7	5.8	10
77	Infantile onset progressive cerebellar atrophy and anterior horn cell degeneration--a late onset variant of PCH-1?. *European Journal of Paediatric Neurology*, **2008**, 12, 97-101	3.8	10
76	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	3.8	9
75	Walker-Warburg syndrome and tectocerebellar dysraphia: A novel association caused by a homozygous DAG1 mutation. *European Journal of Paediatric Neurology*, **2018**, 22, 525-531	3.8	9
74	The ‘Brain Shadowing Sign’: A Novel Marker of Fetal Craniosynostosis. *Fetal Diagnosis and Therapy*, **2016**, 40, 277-284	2.4	9
73	Phenobarbital still has a role in epilepsy treatment. *Journal of Child Neurology*, **1999**, 14, 820-1	2.5	9
72	New findings in a patient with Dubowitz syndrome: velopharyngeal insufficiency and hypoparathyroidism. *American Journal of Medical Genetics Part A*, **1990**, 37, 241-3		9

71	Low erythrocyte zinc content in acquired aphasia with convulsive disorder (Landau-Kleffner syndrome). *Journal of Child Neurology*, **1987**, 2, 28-30	2.5	9
70	The cerebellar "tilted telephone receiver sign" enables prenatal diagnosis of PHACES syndrome. *European Journal of Paediatric Neurology*, **2018**, 22, 900-909	3.8	9
69	Severe growth deficiency, microcephaly, intellectual disability, and characteristic facial features are due to a homozygous QARS mutation. *Neurogenetics*, **2017**, 18, 141-146	3	8
68	Agenesis of the corpus callosum. An autopsy study in fetuses. *Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin*, **2016**, 468, 219-30	5.1	8
67	Prenatal diagnosis of brainstem anomalies. *European Journal of Paediatric Neurology*, **2018**, 22, 1016-1026	3.8	8
66	Resolution of epileptic encephalopathy following treatment with transdermal nicotine. *Epilepsia*, **2013**, 54, e13-5	6.4	8
65	Diagnostic yield of electroencephalograms in infants and young children with frequent paroxysmal eye movements. *Journal of Child Neurology*, **2008**, 23, 620-3	2.5	8
64	A benign congenital myopathy in an inbred Samaritan family. *European Journal of Paediatric Neurology*, **2006**, 10, 182-5	3.8	8
63	Acute intermittent porphyria, Rasmussen encephalitis, or both?. *Journal of Child Neurology*, **2007**, 22, 99-105	2.5	8
62	Molecular and functional studies of retinal degeneration as a clinical presentation of SACS-related disorder. *European Journal of Paediatric Neurology*, **2015**, 19, 472-6	3.8	7
61	Familial hyperekplexia and refractory status epilepticus: a new autosomal recessive syndrome. *Journal of Child Neurology*, **2004**, 19, 522-5	2.5	7
60	Prenatal and postnatal presentation of PRMT7 related syndrome: Expanding the phenotypic manifestations. *American Journal of Medical Genetics, Part A*, **2019**, 179, 78-84	2.5	7
59	Multiple Causes of Pediatric Early Onset Chorea-Clinical and Genetic Approach. *Neuropediatrics*, **2018**, 49, 246-255	1.6	7
58	Expanding the phenotype of TRAK1 mutations: hyperekplexia and refractory status epilepticus. *Brain*, **2018**, 141, e55	11.2	7
57	Mosaic marker chromosome 16 resulting in 16q11.2-q12.1 gain in a child with intellectual disability, microcephaly, and cerebellar cortical dysplasia. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 2991-6	2.5	6
56	Hepatic coma culminating in severe brain damage in a child with a SCN1A mutation. *European Journal of Paediatric Neurology*, **2010**, 14, 456-9	3.8	6
55	Familial optic atrophy with white matter changes. *American Journal of Medical Genetics Part A*, **2003**, 121A, 263-5		6
54	Periventricular brain heterotopias in a child with adrenocortical insufficiency, achalasia, alacrima, and neurologic abnormalities (Allgrove syndrome). *Journal of Child Neurology*, **1999**, 14, 331-4	2.5	6

53	Carbamazepine versus sulthiame in treating benign childhood epilepsy with centrotemporal spikes. *Journal of Child Neurology*, **2002**, 17, 914-6	2.5	6
52	Familial Intracranial Hypertension in 2 Brothers With Mutation: Expansion of the Phenotypic Spectrum. *Journal of Child Neurology*, **2019**, 34, 506-510	2.5	5
51	Atypical presentation of Costeff syndrome-severe psychomotor involvement and electrical status epilepticus during slow wave sleep. *European Journal of Paediatric Neurology*, **2015**, 19, 733-6	3.8	5
50	Autosomal dominant TUBB3-related syndrome: Fetal, radiologic, clinical and morphological features. *European Journal of Paediatric Neurology*, **2020**, 26, 46-60	3.8	5
49	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	3.8	5
48	Muscle glycogen depletion and increased oxidative phosphorylation following status epilepticus. *Journal of Child Neurology*, **2003**, 18, 876-8	2.5	5
47	Fourth ventricle index: sonographic marker for severe fetal vermian dysgenesis/agenesis. *Ultrasound in Obstetrics and Gynecology*, **2019**, 53, 390-395	5.8	5
46	Diagnostic approach to fetal microcephaly. *European Journal of Paediatric Neurology*, **2018**, 22, 935-943	3.8	5
45	Agenesis of the septum pellucidum: Prenatal diagnosis and outcome. *Prenatal Diagnosis*, **2020**, 40, 674-680	3.2	4
44	Ultrasound Nomograms of the Fetal Optic Nerve Sheath Diameter. *Ultraschall in Der Medizin*, **2019**, 40, 476-480	3.8	4
43	A novel mutation in the TPR6 domain of the RAPSN gene associated with congenital myasthenic syndrome. *Journal of the Neurological Sciences*, **2012**, 316, 112-5	3.2	4
42	Genetic counseling and testing for FSHD (facioscapulohumeral muscular dystrophy) in the Israeli population. *Journal of Genetic Counseling*, **2012**, 21, 557-63	2.5	4
41	The Differential Diagnosis of Fetal Intracranial Cystic Lesions. *Ultrasound Clinics*, **2008**, 3, 553-558		4
40	Brain white matter abnormalities associated with copy number variants. *American Journal of Medical Genetics, Part A*, **2020**, 182, 93-103	2.5	4
39	Application of a novel prenatal vertical cranial biometric measurement can improve accuracy of microcephaly diagnosis in utero. *Ultrasound in Obstetrics and Gynecology*, **2016**, 47, 593-9	5.8	4
38	Clinical phenotypes of infantile onset CACNA1A-related disorder. *European Journal of Paediatric Neurology*, **2021**, 30, 144-154	3.8	4
37	Refining the phenotype of the THG1L (p.Val55Ala mutation)-related mitochondrial autosomal recessive congenital cerebellar ataxia. *American Journal of Medical Genetics, Part A*, **2019**, 179, 1575-1579	2.5	3
36	Fetal pericallosal lipomas - Clues to diagnosis in the second trimester. *European Journal of Paediatric Neurology*, **2018**, 22, 929-934	3.8	3

35	Building Bridges Between the Clinic and the Laboratory: A Meeting Review - Brain Malformations: A Roadmap for Future Research. *Frontiers in Cellular Neuroscience*, **2019**, 13, 434	6.1	3
34	Autistic regression in a child with Silver-Russell syndrome and maternal UPD 7. *European Journal of Paediatric Neurology*, **2012**, 16, 95-8	3.8	3
33	Dominantly inherited nonprogressive cerebellar hypoplasia identified in utero. *Journal of Child Neurology*, **2012**, 27, 1000-3	2.5	3
32	Central apnea in a child with congenital autonomic dysfunction and universal pain loss. *Journal of Child Neurology*, **1996**, 11, 162-4	2.5	3
31	A practical approach to prenatal diagnosis of malformations of cortical development. *European Journal of Paediatric Neurology*, **2021**, 34, 50-61	3.8	3
30	A possible genotype-phenotype correlation in Ashkenazi-Jewish individuals with Aicardi-Goutières syndrome associated with SAMHD1 mutation. *Journal of Child Neurology*, **2015**, 30, 490-5	2.5	2
29	Expanding the genotype-phenotype spectrum of ISCA2-related multiple mitochondrial dysfunction syndrome-cavitating leukoencephalopathy and prolonged survival. *Neurogenetics*, **2020**, 21, 243-249	3	2
28	CHAMP1 Mutations cause Refractory Infantile Myoclonic Epilepsy. *Journal of Pediatric Neurology*, **2020**, 18, 027-032	0.2	2
27	Periventricular pseudocysts of noninfectious origin: Prenatal associated findings and prognostic factors. *Prenatal Diagnosis*, **2020**, 40, 931-941	3.2	2
26	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	2.4	2
25	Familial Brain Periventricular Pseudocysts. *Fetal Diagnosis and Therapy*, **2017**, 42, 42-47	2.4	2
24	Nonprogressive familial leukoencephalopathy with porencephalic cyst and focal seizures. *Journal of Child Neurology*, **2006**, 21, 145-8	2.5	2
23	A new autosomal recessive syndrome with Zellweger-like manifestations. *American Journal of Medical Genetics Part A*, **2003**, 119A, 352-5		2
22	Myoclonic tremor status as a presenting symptom of adenylosuccinate lyase deficiency. *European Journal of Medical Genetics*, **2020**, 63, 104061	2.6	2
21	The role of orotic acid measurement in routine newborn screening for urea cycle disorders. *Journal of Inherited Metabolic Disease*, **2021**, 44, 606-617	5.4	2
20	Congenital Mirror Movements Associated With Brain Malformations. *Journal of Child Neurology*, **2021**, 36, 545-555	2.5	2
19	Fetal Brain Development: Regulating Processes and Related Malformations. *Life*, **2022**, 12, 809	3	2
18	Refractory epilepsy associated with ventriculoperitoneal shunt over-drainage: case report. *Childs Nervous System*, **2019**, 35, 2411-2416	1.7	1

17	A newly recognized syndrome of severe growth deficiency, microcephaly, intellectual disability, and characteristic facial features. *European Journal of Medical Genetics*, **2014**, 57, 288-92	2.6	1
16	Alexander Disease in Israel: Megalencephaly and Leukoencephalopathy and Its Differential Diagnosis. *Journal of Pediatric Neurology*, **2015**, 13, 121-125	0.2	1
15	Fat intolerance in developmentally impaired children with severe feeding intolerance. *Journal of Child Neurology*, **2006**, 21, 167-70	2.5	1
14	Prenatal Diagnosis of Structural Brain Anomalies **2012**, 263-276		1
13	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	2.6	1
12	Delineation of the phenotype of MED17-related disease in Caucasus-Jewish families. *European Journal of Paediatric Neurology*, **2021**, 32, 40-45	3.8	1
11	Why can seizures remain intractable? Clinical vignettes from the life experience of a pediatric epileptologist. *Journal of Child Neurology*, **2011**, 26, 121-5	2.5	0
10	White matter abnormalities and iron deposition in prenatal mucolipidosis IV- fetal imaging and pathology. *Metabolic Brain Disease*, **2021**, 36, 2155-2167	3.9	0
9	Prenatal Diagnosis of Structural Brain Anomalies **2017**, 249-255		
8	Cerebellar Anomalies **2018**, 184-188.e1		
7	Reply to: The many faces of TUBB4A mutations. *Neurogenetics*, **2014**, 15, 83	3	
6	Efficacy of corticosteroid therapy in treating epileptic encephalopathies and refractory epilepsies other than West syndrome. *Journal of Pediatric Neurology*, **2015**, 04, 147-153	0.2	
5	Dominantly inherited nonprogressive cerebellar hypoplasia identified in utero: no doubt. *Journal of Child Neurology*, **2013**, 28, 279-80	2.5	
4	Familial leukoencephalopathy with slowly progressive dystonia and ataxia. *European Journal of Paediatric Neurology*, **2009**, 13, 530-3	3.8	
3	. *European Journal of Paediatric Neurology*, **2007**, 11, 55-56	3.8	
2	Mitochondrial DNA mutations are where to look. *Journal of Child Neurology*, **1999**, 14, 479	2.5	
1	Bilateral polymicrogyria associated with dystonia: A new neurogenetic syndrome?. *American Journal of Medical Genetics, Part A*, **2020**, 182, 2207-2213	2.5	