

Dorit Lev

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

72
papers

1,527
citations

21
h-index

37
g-index

78
ext. papers

1,840
ext. citations

4.3
avg, IF

3.8
L-index

#	Paper	IF	Citations
72	Clinical manifestations in a cohort of 41 Rothmund-Thomson syndrome patients. <i>American Journal of Medical Genetics Part A</i> , 2001 , 102, 11-7		244
71	Targeted genomic capture and massively parallel sequencing to identify genes for hereditary hearing loss in Middle Eastern families. <i>Genome Biology</i> , 2011 , 12, R89	18.3	163
70	Mutations disrupting selenocysteine formation cause progressive cerebello-cerebral atrophy. <i>American Journal of Human Genetics</i> , 2010 , 87, 538-44	11	111
69	De novo mutations in KIF1A cause progressive encephalopathy and brain atrophy. <i>Annals of Clinical and Translational Neurology</i> , 2015 , 2, 623-35	5.3	68
68	Familial neonatal seizures in 36 families: Clinical and genetic features correlate with outcome. <i>Epilepsia</i> , 2015 , 56, 1071-80	6.4	68
67	Copy number variations in cryptogenic cerebral palsy. <i>Neurology</i> , 2015 , 84, 1660-8	6.5	62
66	GRIN1 mutations cause encephalopathy with infantile-onset epilepsy, and hyperkinetic and stereotyped movement disorders. <i>Epilepsia</i> , 2015 , 56, 841-8	6.4	56
65	VPS53 mutations cause progressive cerebello-cerebral atrophy type 2 (PCCA2). <i>Journal of Medical Genetics</i> , 2014 , 51, 303-8	5.8	53
64	Utility of Whole Exome Sequencing for Genetic Diagnosis of Previously Undiagnosed Pediatric Neurology Patients. <i>Journal of Child Neurology</i> , 2016 , 31, 1534-1539	2.5	38
63	The molecular and phenotypic spectrum of IQSEC2-related epilepsy. <i>Epilepsia</i> , 2016 , 57, 1858-1869	6.4	38
62	De novo hotspot variants in CYFIP2 cause early-onset epileptic encephalopathy. <i>Annals of Neurology</i> , 2018 , 83, 794-806	9.4	37
61	Paroxysmal tonic upward gaze as a presentation of de-novo mutations in CACNA1A. <i>European Journal of Paediatric Neurology</i> , 2015 , 19, 292-7	3.8	32
60	Fetal exome sequencing: yield and limitations in a tertiary referral center. <i>Ultrasound in Obstetrics and Gynecology</i> , 2019 , 53, 80-86	5.8	31
59	Genetic and phenotypic spectrum associated with IFIH1 gain-of-function. <i>Human Mutation</i> , 2020 , 41, 837-849	4.7	31
58	Molecular cytogenetic studies in three patients with partial trisomy 2p, including CGH from paraffin-embedded tissue. <i>American Journal of Medical Genetics Part A</i> , 2000 , 91, 74-82		28
57	Pulmonary agenesis, microphthalmia, and diaphragmatic defect (PMD): New syndrome or association? 1999 , 86, 6-8		27
56	Decreased bone density in carriers and patients of an Israeli family with the osteoporosis-pseudoglioma syndrome. <i>Israel Medical Association Journal</i> , 2003 , 5, 419-21	0.9	25

55	The spinal muscular atrophy with pontocerebellar hypoplasia gene VRK1 regulates neuronal migration through an amyloid- β -precursor protein-dependent mechanism. <i>Journal of Neuroscience</i> , 2015 , 35, 936-42	6.6	24
54	Rare GABRA3 variants are associated with epileptic seizures, encephalopathy and dysmorphic features. <i>Brain</i> , 2017 , 140, 2879-2894	11.2	24
53	Paternal germline mosaicism of a SCN2A mutation results in Ohtahara syndrome in half siblings. <i>European Journal of Paediatric Neurology</i> , 2014 , 18, 567-71	3.8	23
52	A novel missense mutation in the NDP gene in a child with Norrie disease and severe neurological involvement including infantile spasms. <i>American Journal of Medical Genetics, Part A</i> , 2007 , 143A, 921-4	2.5	22
51	RARS2 mutations cause early onset epileptic encephalopathy without ponto-cerebellar hypoplasia. <i>European Journal of Paediatric Neurology</i> , 2016 , 20, 412-7	3.8	19
50	Automatic scanning of interphase FISH for prenatal diagnosis in uncultured amniocytes. <i>Genetic Testing and Molecular Biomarkers</i> , 2005 , 9, 41-7		18
49	Samaritan myopathy, an ultimately benign congenital myopathy, is caused by a RYR1 mutation. <i>Acta Neuropathologica</i> , 2012 , 124, 575-81	14.3	16
48	Delineation of the interstitial 6q25 microdeletion syndrome: refinement of the critical causative region. <i>American Journal of Medical Genetics, Part A</i> , 2012 , 158A, 1395-9	2.5	16
47	Lacosamide for SCN2A-related intractable neonatal and infantile seizures. <i>Epileptic Disorders</i> , 2018 , 20, 440-446	1.9	16
46	Photoreceptor Guanylate Cyclase () Mutations Cause Retinal Dystrophies by Severe Malfunction of Ca-Dependent Cyclic GMP Synthesis. <i>Frontiers in Molecular Neuroscience</i> , 2018 , 11, 348	6.1	13
45	Progressive cerebello-cerebral atrophy and progressive encephalopathy with edema, hypsarrhythmia and optic atrophy may be allelic syndromes. <i>European Journal of Paediatric Neurology</i> , 2018 , 22, 1133-1138	3.8	12
44	Aberrant HRAS transcript processing underlies a distinctive phenotype within the RASopathy clinical spectrum. <i>Human Mutation</i> , 2017 , 38, 798-804	4.7	11
43	Metabolic stroke in a patient with bi-allelic OPA1 mutations. <i>Metabolic Brain Disease</i> , 2019 , 34, 1043-1048	3.9	11
42	Expanding the phenotype of IBA57 mutations: related leukodystrophy can remain asymptomatic. <i>Journal of Human Genetics</i> , 2018 , 63, 1223-1229	4.3	11
41	Infantile onset progressive cerebellar atrophy and anterior horn cell degeneration--a late onset variant of PCH-1?. <i>European Journal of Paediatric Neurology</i> , 2008 , 12, 97-101	3.8	10
40	Walker-Warburg syndrome and tectocerebellar dysraphia: A novel association caused by a homozygous DAG1 mutation. <i>European Journal of Paediatric Neurology</i> , 2018 , 22, 525-531	3.8	9
39	Crowdfunding effort identifies the causative mutation in a patient with nystagmus, microcephaly, dystonia and hypomyelination. <i>Journal of Genetics and Genomics</i> , 2015 , 42, 79-81	4	9
38	The cerebellar "tilted telephone receiver sign" enables prenatal diagnosis of PHACES syndrome. <i>European Journal of Paediatric Neurology</i> , 2018 , 22, 900-909	3.8	9

37	Severe growth deficiency, microcephaly, intellectual disability, and characteristic facial features are due to a homozygous QARS mutation. <i>Neurogenetics</i> , 2017 , 18, 141-146	3	8
36	Agensis of the corpus callosum. An autopsy study in fetuses. <i>Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin</i> , 2016 , 468, 219-30	5.1	8
35	A benign congenital myopathy in an inbred Samaritan family. <i>European Journal of Paediatric Neurology</i> , 2006 , 10, 182-5	3.8	8
34	Spectrum of genes for inherited hearing loss in the Israeli Jewish population, including the novel human deafness gene ATOH1. <i>Clinical Genetics</i> , 2020 , 98, 353-364	4	8
33	Molecular and functional studies of retinal degeneration as a clinical presentation of SACS-related disorder. <i>European Journal of Paediatric Neurology</i> , 2015 , 19, 472-6	3.8	7
32	Prenatal and postnatal presentation of PRMT7 related syndrome: Expanding the phenotypic manifestations. <i>American Journal of Medical Genetics, Part A</i> , 2019 , 179, 78-84	2.5	7
31	Multiple Causes of Pediatric Early Onset Chorea-Clinical and Genetic Approach. <i>Neuropediatrics</i> , 2018 , 49, 246-255	1.6	7
30	Expanding the phenotype of TRAK1 mutations: hyperekplexia and refractory status epilepticus. <i>Brain</i> , 2018 , 141, e55	11.2	7
29	Mosaic marker chromosome 16 resulting in 16q11.2-q12.1 gain in a child with intellectual disability, microcephaly, and cerebellar cortical dysplasia. <i>American Journal of Medical Genetics, Part A</i> , 2011 , 155A, 2991-6	2.5	6
28	Familial optic atrophy with white matter changes. <i>American Journal of Medical Genetics Part A</i> , 2003 , 121A, 263-5		6
27	Familial Intracranial Hypertension in 2 Brothers With Mutation: Expansion of the Phenotypic Spectrum. <i>Journal of Child Neurology</i> , 2019 , 34, 506-510	2.5	5
26	Atypical presentation of Costeff syndrome-severe psychomotor involvement and electrical status epilepticus during slow wave sleep. <i>European Journal of Paediatric Neurology</i> , 2015 , 19, 733-6	3.8	5
25	Autosomal dominant TUBB3-related syndrome: Fetal, radiologic, clinical and morphological features. <i>European Journal of Paediatric Neurology</i> , 2020 , 26, 46-60	3.8	5
24	Isolated fetal horseshoe kidney does not seem to increase the risk for abnormal chromosomal microarray results. <i>European Journal of Obstetrics, Gynecology and Reproductive Biology</i> , 2018 , 222, 80-83 ^{2,4}		5
23	Diagnosis by whole exome sequencing of atypical infantile onset Alexander disease masquerading as a mitochondrial disorder. <i>European Journal of Paediatric Neurology</i> , 2014 , 18, 495-501	3.8	5
22	Ultrasound Nomograms of the Fetal Optic Nerve Sheath Diameter. <i>Ultraschall in Der Medizin</i> , 2019 , 40, 476-480	3.8	4
21	Genetic counseling and testing for FSHD (facioscapulohumeral muscular dystrophy) in the Israeli population. <i>Journal of Genetic Counseling</i> , 2012 , 21, 557-63	2.5	4
20	Brain white matter abnormalities associated with copy number variants. <i>American Journal of Medical Genetics, Part A</i> , 2020 , 182, 93-103	2.5	4

19	NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns. <i>Genetics in Medicine</i> , 2021 , 23, 363-373	8.1	4
18	Clinical phenotypes of infantile onset CACNA1A-related disorder. <i>European Journal of Paediatric Neurology</i> , 2021 , 30, 144-154	3.8	4
17	Refining the phenotype of the THG1L (p.Val55Ala mutation)-related mitochondrial autosomal recessive congenital cerebellar ataxia. <i>American Journal of Medical Genetics, Part A</i> , 2019 , 179, 1575-1579	3.5	3
16	Microarray findings in pregnancies with oligohydramnios - a retrospective cohort study and literature review. <i>Journal of Perinatal Medicine</i> , 2019 , 48, 53-58	2.7	3
15	A possible genotype-phenotype correlation in Ashkenazi-Jewish individuals with Aicardi-Goutières syndrome associated with SAMHD1 mutation. <i>Journal of Child Neurology</i> , 2015 , 30, 490-5	2.5	2
14	Expanding the genotype-phenotype spectrum of ISCA2-related multiple mitochondrial dysfunction syndrome-cavitating leukoencephalopathy and prolonged survival. <i>Neurogenetics</i> , 2020 , 21, 243-249	3	2
13	Isolated ventricular septal defects demonstrated by fetal echocardiography: prenatal course and postnatal outcome. <i>Journal of Maternal-Fetal and Neonatal Medicine</i> , 2020 , 1-5	2	2
12	CHAMP1 Mutations cause Refractory Infantile Myoclonic Epilepsy. <i>Journal of Pediatric Neurology</i> , 2020 , 18, 027-032	0.2	2
11	Periventricular pseudocysts of noninfectious origin: Prenatal associated findings and prognostic factors. <i>Prenatal Diagnosis</i> , 2020 , 40, 931-941	3.2	2
10	Myoclonic tremor status as a presenting symptom of adenylosuccinate lyase deficiency. <i>European Journal of Medical Genetics</i> , 2020 , 63, 104061	2.6	2
9	A newly recognized syndrome of severe growth deficiency, microcephaly, intellectual disability, and characteristic facial features. <i>European Journal of Medical Genetics</i> , 2014 , 57, 288-92	2.6	1
8	In-silico phenotype prediction by normal mode variant analysis in TUBB4A-related disease.. <i>Scientific Reports</i> , 2022 , 12, 58	4.9	1
7	Spectrum of genes for inherited hearing loss in the Israeli Jewish population, including the novel human deafness gene ATOH1		1
6	Infantile onset progressive cerebellar atrophy and anterior horn cell Degeneration-A novel phenotype associated with mutations in the PLA2G6 gene. <i>European Journal of Medical Genetics</i> , 2020 , 63, 103801	2.6	1
5	Procedure-to-delivery interval after late amniocentesis and the need for routine antenatal corticosteroids. <i>Journal of Maternal-Fetal and Neonatal Medicine</i> , 2020 , 1-8	2	1
4	White matter abnormalities and iron deposition in prenatal mucopolipidosis IV- fetal imaging and pathology. <i>Metabolic Brain Disease</i> , 2021 , 36, 2155-2167	3.9	0
3	. <i>European Journal of Paediatric Neurology</i> , 2007 , 11, 55-56	3.8	
2	Bilateral polymicrogyria associated with dystonia: A new neurogenetic syndrome?. <i>American Journal of Medical Genetics, Part A</i> , 2020 , 182, 2207-2213	2.5	

- 1 X-linked myopathy with excessive autophagy: First report of an Israeli family presenting with late onset lower limb girdle weakness. *Neuromuscular Disorders*, **2021**, 31, 854-858

2.9