Dorit Lev

List of Publications by Year in Descending Order

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	In-silico phenotype prediction by normal mode variant analysis in TUBB4A-related disease.. *Scientific Reports*, **2022**, 12, 58	4.9	1
71	White matter abnormalities and iron deposition in prenatal mucolipidosis IV- fetal imaging and pathology. *Metabolic Brain Disease*, **2021**, 36, 2155-2167	3.9	0
70	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	
69	NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns. *Genetics in Medicine*, **2021**, 23, 363-373	8.1	4
68	Clinical phenotypes of infantile onset CACNA1A-related disorder. *European Journal of Paediatric Neurology*, **2021**, 30, 144-154	3.8	4
67	Expanding the genotype-phenotype spectrum of ISCA2-related multiple mitochondrial dysfunction syndrome-cavitating leukoencephalopathy and prolonged survival. *Neurogenetics*, **2020**, 21, 243-249	3	2
66	Autosomal dominant TUBB3-related syndrome: Fetal, radiologic, clinical and morphological features. *European Journal of Paediatric Neurology*, **2020**, 26, 46-60	3.8	5
65	Isolated ventricular septal defects demonstrated by fetal echocardiography: prenatal course and postnatal outcome. *Journal of Maternal-Fetal and Neonatal Medicine*, **2020**, 1-5	2	2
64	CHAMP1 Mutations cause Refractory Infantile Myoclonic Epilepsy. *Journal of Pediatric Neurology*, **2020**, 18, 027-032	0.2	2
63	Periventricular pseudocysts of noninfectious origin: Prenatal associated findings and prognostic factors. *Prenatal Diagnosis*, **2020**, 40, 931-941	3.2	2
62	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
61	Brain white matter abnormalities associated with copy number variants. *American Journal of Medical Genetics, Part A*, **2020**, 182, 93-103	2.5	4
60	Genetic and phenotypic spectrum associated with IFIH1 gain-of-function. *Human Mutation*, **2020**, 41, 837-849	4.7	31
59	Myoclonic tremor status as a presenting symptom of adenylosuccinate lyase deficiency. *European Journal of Medical Genetics*, **2020**, 63, 104061	2.6	2
58	Spectrum of genes for inherited hearing loss in the Israeli Jewish population, including the novel human deafness gene ATOH1. *Clinical Genetics*, **2020**, 98, 353-364	4	8
57	Bilateral polymicrogyria associated with dystonia: A new neurogenetic syndrome?. *American Journal of Medical Genetics, Part A*, **2020**, 182, 2207-2213	2.5	
56	Procedure-to-delivery interval after late amniocentesis and the need for routine antenatal corticosteroids. *Journal of Maternal-Fetal and Neonatal Medicine*, **2020**, 1-8	2	1

55	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
54	Familial Intracranial Hypertension in 2 Brothers With Mutation: Expansion of the Phenotypic Spectrum. *Journal of Child Neurology*, **2019**, 34, 506-510	2.5	5
53	Metabolic stroke in a patient with bi-allelic OPA1 mutations. *Metabolic Brain Disease*, **2019**, 34, 1043-1048	3.9	11
52	Ultrasound Nomograms of the Fetal Optic Nerve Sheath Diameter. *Ultraschall in Der Medizin*, **2019**, 40, 476-480	3.8	4
51	Fetal exome sequencing: yield and limitations in a tertiary referral center. *Ultrasound in Obstetrics and Gynecology*, **2019**, 53, 80-86	5.8	31
50	Microarray findings in pregnancies with oligohydramnios - a retrospective cohort study and literature review. *Journal of Perinatal Medicine*, **2019**, 48, 53-58	2.7	3
49	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
48	De novo hotspot variants in CYFIP2 cause early-onset epileptic encephalopathy. *Annals of Neurology*, **2018**, 83, 794-806	9.4	37
47	Isolated fetal horseshoe kidney does not seem to increase the risk for abnormal chromosomal microarray results. *European Journal of Obstetrics, Gynecology and Reproductive Biology*, **2018**, 222, 80-83	2.4	5
46	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
45	Lacosamide for SCN2A-related intractable neonatal and infantile seizures. *Epileptic Disorders*, **2018**, 20, 440-446	1.9	16
44	Expanding the phenotype of IBA57 mutations: related leukodystrophy can remain asymptomatic. *Journal of Human Genetics*, **2018**, 63, 1223-1229	4.3	11
43	Photoreceptor Guanylate Cyclase () Mutations Cause Retinal Dystrophies by Severe Malfunction of Ca-Dependent Cyclic GMP Synthesis. *Frontiers in Molecular Neuroscience*, **2018**, 11, 348	6.1	13
42	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
41	The cerebellar "tilted telephone receiver sign" enables prenatal diagnosis of PHACES syndrome. *European Journal of Paediatric Neurology*, **2018**, 22, 900-909	3.8	9
40	Multiple Causes of Pediatric Early Onset Chorea-Clinical and Genetic Approach. *Neuropediatrics*, **2018**, 49, 246-255	1.6	7
39	Expanding the phenotype of TRAK1 mutations: hyperekplexia and refractory status epilepticus. *Brain*, **2018**, 141, e55	11.2	7
38	Aberrant HRAS transcript processing underlies a distinctive phenotype within the RASopathy clinical spectrum. *Human Mutation*, **2017**, 38, 798-804	4.7	11

37	Severe growth deficiency, microcephaly, intellectual disability, and characteristic facial features are due to a homozygous QARS mutation. *Neurogenetics*, **2017**, 18, 141-146	3	8
36	Rare GABRA3 variants are associated with epileptic seizures, encephalopathy and dysmorphic features. *Brain*, **2017**, 140, 2879-2894	11.2	24
35	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
34	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
33	RARS2 mutations cause early onset epileptic encephalopathy without ponto-cerebellar hypoplasia. *European Journal of Paediatric Neurology*, **2016**, 20, 412-7	3.8	19
32	The molecular and phenotypic spectrum of IQSEC2-related epilepsy. *Epilepsia*, **2016**, 57, 1858-1869	6.4	38
31	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
30	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
29	Copy number variations in cryptogenic cerebral palsy. *Neurology*, **2015**, 84, 1660-8	6.5	62
28	De novo mutations in KIF1A cause progressive encephalopathy and brain atrophy. *Annals of Clinical and Translational Neurology*, **2015**, 2, 623-35	5.3	68
27	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
26	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
25	A possible genotype-phenotype correlation in Ashkenazi-Jewish individuals with Aicardi-Goutiᅠᅠes syndrome associated with SAMHD1 mutation. *Journal of Child Neurology*, **2015**, 30, 490-5	2.5	2
24	GRIN1 mutations cause encephalopathy with infantile-onset epilepsy, and hyperkinetic and stereotyped movement disorders. *Epilepsia*, **2015**, 56, 841-8	6.4	56
23	Familial neonatal seizures in 36 families: Clinical and genetic features correlate with outcome. *Epilepsia*, **2015**, 56, 1071-80	6.4	68
22	Crowdfunding effort identifies the causative mutation in a patient with nystagmus, microcephaly, dystonia and hypomyelination. *Journal of Genetics and Genomics*, **2015**, 42, 79-81	4	9
21	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
20	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

19	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
18	VPS53 mutations cause progressive cerebello-cerebral atrophy type 2 (PCCA2). *Journal of Medical Genetics*, **2014**, 51, 303-8	5.8	53
17	Samaritan myopathy, an ultimately benign congenital myopathy, is caused by a RYR1 mutation. *Acta Neuropathologica*, **2012**, 124, 575-81	14.3	16
16	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
15	Genetic counseling and testing for FSHD (facioscapulohumeral muscular dystrophy) in the Israeli population. *Journal of Genetic Counseling*, **2012**, 21, 557-63	2.5	4
14	Targeted genomic capture and massively parallel sequencing to identify genes for hereditary hearing loss in Middle Eastern families. *Genome Biology*, **2011**, 12, R89	18.3	163
13	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
12	Mutations disrupting selenocysteine formation cause progressive cerebello-cerebral atrophy. *American Journal of Human Genetics*, **2010**, 87, 538-44	11	111
11	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
10	A novel missense mutation in the NDP gene in a child with Norrie disease and severe neurological involvement including infantile spasms. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 921-4	2.5	22
9	. *European Journal of Paediatric Neurology*, **2007**, 11, 55-56	3.8	
8	A benign congenital myopathy in an inbred Samaritan family. *European Journal of Paediatric Neurology*, **2006**, 10, 182-5	3.8	8
7	Automatic scanning of interphase FISH for prenatal diagnosis in uncultured amniocytes. *Genetic Testing and Molecular Biomarkers*, **2005**, 9, 41-7		18
6	Familial optic atrophy with white matter changes. *American Journal of Medical Genetics Part A*, **2003**, 121A, 263-5		6
5	Decreased bone density in carriers and patients of an Israeli family with the osteoporosis-pseudoglioma syndrome. *Israel Medical Association Journal*, **2003**, 5, 419-21	0.9	25
4	Clinical manifestations in a cohort of 41 Rothmund-Thomson syndrome patients. *American Journal of Medical Genetics Part A*, **2001**, 102, 11-7		244
3	Molecular cytogenetic studies in three patients with partial trisomy 2p, including CGH from paraffin-embedded tissue. *American Journal of Medical Genetics Part A*, **2000**, 91, 74-82		28
2	Pulmonary agenesis, microphthalmia, and diaphragmatic defect (PMD): New syndrome or association? **1999**, 86, 6-8		27

1 Spectrum of genes for inherited hearing loss in the Israeli Jewish population, including the novel human deafness gene ATOH1 1