Eli Hershkovitz

List of Publications by Year
in descending order

112 papers	4,598 citations	136740 32 h-index	106150 65 g-index
116 all docs	116 docs citations	116 times ranked	5627 citing authors

#	Article	IF	Citations
1	A retrospective, multinational, multicenter study on the natural history of infantile-onset Pompe disease. Journal of Pediatrics, 2006, 148, 671-676.e2.	0.9	500
2	Childhood Obesity. Journal of Clinical Endocrinology and Metabolism, 2005, 90, 1871-1887.	1.8	459
3	Deficiency of the ADP-Forming Succinyl-CoA Synthase Activity Is Associated with Encephalomyopathy and Mitochondrial DNA Depletion. American Journal of Human Genetics, 2005, 76, 1081-1086.	2.6	284
4	Autosomal-Recessive Hypophosphatemic Rickets Is Associated with an Inactivation Mutation in the ENPP1 Gene. American Journal of Human Genetics, 2010, 86, 273-278.	2.6	262
5	Mutation of TBCE causes hypoparathyroidismâ€“Âretardationâ€“dysmorphism and autosomal recessive Kennyâ€“Caffey syndrome. Nature Genetics, 2002, 32, 448-452.	9.4	248
6	Defective mitochondrial translation caused by a ribosomal protein (MRPS16) mutation. Annals of Neurology, 2004, 56, 734-738.	2.8	205
7	Successful immune tolerance induction to enzyme replacement therapy in CRIM-negative infantile Pompe disease. Genetics in Medicine, 2012, 14, 135-142.	1.1	183
8	Congenital insensitivity to pain with anhidrosis (CIPA) in Israeli-Bedouins: Genetic heterogeneity, novel mutations in theTRKA/NGF receptor gene, clinical findings, and results of nerve conduction studies. American Journal of Medical Genetics Part A, 2000, 92, 353-360.	2.4	129
9	The Gene for Glycogen-Storage Disease Type 1b Maps to Chromosome 11q23. American Journal of Human Genetics, 1998, 62, 400-405.	2.6	126
10	Bone marrow transplantation for Maroteaux-Lamy syndrome (MPS VI): Long-term follow-up. Journal of Inherited Metabolic Disease, 1999, 22, 50-62.	1.7	125
11	Mitochondrial complex I deficiency caused by a deleterious NDUFA11 mutation. Annals of Neurology, 2008, 63, 405-408.	2.8	103
12	The utility of basal serum LH in prediction of central precocious puberty in girls. European Journal of Endocrinology, 2012, 166, 295-299.	1.9	89
13	Thyroglossal Duct Carcinoma in Children: Case Presentation and Review of the Literature. Thyroid, 2004, 14, 777-785.	2.4	83
14	Homozygous Mutation G539R in the Gene for P450 Oxidoreductase in a Family Previously Diagnosed as Having 17,20-Lyase Deficiency. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 3584-3588.	1.8	82
15	Homozygosity and Linkage-Disequilibrium Mapping of the Syndrome of Congenital Hypoparathyroidism, Growth and Mental Retardation, and Dysmorphism to a 1-cM Interval on Chromosome 1q42-43. American Journal of Human Genetics, 1998, 63, 163-169.	2.6	70
16	A Recessive Contiguous Gene Deletion of Chromosome 2p16 Associated with Cystinuria and a Mitochondrial Disease. American Journal of Human Genetics, 2001, 69, 869-875.	2.6	69
17	Effects of a Twelve-Week Randomized Intervention of Exercise and/or Diet on Weight Loss and Weight Maintenance, and Other Metabolic Parameters in Obese Preadolescent Children. Hormone Research, 2009, 72, 287-301.	1.8	69
18	TMEM70 mutations are a common cause of nuclear encoded ATP synthase assembly defect: further delineation of a new syndrome. Journal of Medical Genetics, 2011, 48, 177-182.	1.5	61

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19	Varied Clinical Presentations of Seven Patients With Mutations inCYP11A1Encoding the Cholesterol Side-Chain Cleavage Enzyme, P450scc. Journal of Clinical Endocrinology and Metabolism, 2013, 98, 713-720.	1.8	59
20	Automated identification of RNA conformational motifs: theory and application to the HM LSU 23S rRNA. Nucleic Acids Research, 2003, 31, 6249-6257.	6.5	54
21	Lipoamide Dehydrogenase Deficiency Due to a Novel Mutation in the Interface Domain. Biochemical and Biophysical Research Communications, 1999, 262, 163-166.	1.0	51
22	The influence of diet and/or exercise and parental compliance on health-related quality of life in obese children. Nutrition Research, 2009, 29, 397-404.	1.3	51
23	Single nucleotide RNA choreography. Nucleic Acids Research, 2006, 34, 1481-1491.	6.5	43
24	Neurophysiologic studies in congenital insensitivity to pain with anhidrosis. Pediatric Neurology, 2001, 25, 397-400.	1.0	41
25	Continuous glucose monitoring in children with glycogen storage disease type I. Journal of Inherited Metabolic Disease, 2001, 24, 863-869.	1.7	40
26	Luteinizing hormone-releasing hormone antagonists interfere with autocrine and paracrine growth stimulation of MCF-7 mammary cancer cells by insulin-like growth factors. Journal of Clinical Endocrinology and Metabolism, 1993, 77, 963-968.	1.8	38
27	Essential role of carbonic anhydrase XII in secretory gland fluid and HCO₃^{â^'} secretion revealed by disease causing human mutation. Journal of Physiology, 2015, 593, 5299-5312.	1.3	37
28	Retinol concentration in maternal and cord serum: its relation to birth weight in healthy motherâ€“infant pairs. Early Human Development, 2003, 71, 19-28.	0.8	36
29	Neonatal hyperthyrotropinemia: population characteristics, diagnosis, management and outcome after cessation of therapy. Clinical Endocrinology, 2010, 72, 264-271.	1.2	36
30	Glycogen storage disease type 1a in Israel: Biochemical, clinical, and mutational studies. , 1997, 72, 286-290.		35
31	Prenatal exclusion of Leigh syndrome due to T8993C mutation in the mitochondrial DNA. Prenatal Diagnosis, 2003, 23, 31-33.	1.1	35
32	Autosomal recessive hyponatremia due to isolated salt wasting in sweat associated with a mutation in the active site of Carbonic Anhydrase 12. Human Genetics, 2011, 129, 397-405.	1.8	35
33	The 2p21 deletion syndrome: Characterization of the transcription content. Genomics, 2005, 86, 195-211.	1.3	34
34	Statistical Analysis of RNA Backbone. IEEE/ACM Transactions on Computational Biology and Bioinformatics, 2006, 3, 33-46.	1.9	33
35	Mitochondrial epileptic encephalopathy, 3â€methylglutaconic aciduria and variable complex V deficiency associated with <scp>TIMM50</scp> mutations. Clinical Genetics, 2017, 91, 690-696.	1.0	28
36	Characterization of the mutations in the glucose-6-phosphatase gene in Israeli patients with glycogen storage disease type 1a: R83C in six Jews and a novel V166G mutation in a Muslim Arab. Journal of Inherited Metabolic Disease, 1995, 18, 21-27.	1.7	27

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37	Trends in the incidence of type 1 diabetes among Jews and Arabs in Israel. Pediatric Diabetes, 2014, 15, 422-427.	1.2	27
38	Cerebral X-linked adrenoleukodystrophy in a girl with Xq27-Ter deletion. Annals of Neurology, 2002, 52, 234-237.	2.8	26
39	Hypoparathyroidism-Retardation-Dysmorphism (HRD) Syndrome - A Review. Journal of Pediatric Endocrinology and Metabolism, 2004, 17, 1583-90.	0.4	26
40	Increase in the incidence of type 1 diabetes in Israeli children following the Second Lebanon War. Pediatric Diabetes, 2012, 13, 326-333.	1.2	26
41	Developing and evaluating rare disease educational materials co-created by expert clinicians and patients: the paradigm of congenital hypogonadotropic hypogonadism. Orphanet Journal of Rare Diseases, 2017, 12, 57.	1.2	26
42	Incidence of type 1 diabetes mellitus in the 0- to 17-yr-old Israel population, 1997?2003. Pediatric Diabetes, 2007, 8, 60-66.	1.2	24
43	Parathyroid Development and the Role of Tubulin Chaperone E. Hormone Research in Paediatrics, 2007, 67, 12-21.	0.8	22
44	Treatment of Xâ€linked childhood cerebral adrenoleukodystrophy by the use of an allogeneic stem cell transplantation with reduced intensity conditioning regimen. Clinical Transplantation, 2005, 19, 840-847.	0.8	21
45	Domain III of the T. thermophilus 23S rRNA folds independently to a near-native state. Rna, 2012, 18, 752-758.	1.6	21
46	Pulmonary involvement in Niemann-Pick C type 1. European Journal of Pediatrics, 2018, 177, 1609-1615.	1.3	21
47	Adrenal Insufficiency After Achalasia in the Triple-A Syndrome. Clinical Pediatrics, 1996, 35, 99-100.	0.4	20
48	Woodhouse-Sakati Syndrome in an Israeli-Arab Family Presenting with Youth-Onset Diabetes Mellitus and Delayed Puberty. Hormone Research in Paediatrics, 2011, 75, 362-366.	0.8	20
49	Phosphoglucomutaseâ€1 deficiency: Intrafamilial clinical variability and common secondary adrenal insufficiency. American Journal of Medical Genetics, Part A, 2015, 167, 3139-3143.	0.7	20
50	Nerve Growth Factor-Tyrosine Kinase A Pathway Is Involved in Thermoregulation and Adaptation to Stress: Studies on Patients with Hereditary Sensory and Autonomic Neuropathy Type IV. Pediatric Research, 2005, 57, 587-590.	1.1	19
51	Hypoparathyroidism, Retardation, and Dysmorphism Syndrome: Impaired Early Growth and Increased Susceptibility to Severe Infections Due to Hyposplenism and Impaired Polymorphonuclear Cell Functions. Pediatric Research, 2007, 62, 505-509.	1.1	19
52	An Early Rise in Urine N-Telopeptide Predicts the Growth Response of Normal Prepubertal Short Children to Growth Hormone Therapy. Journal of Pediatric Endocrinology and Metabolism, 1996, 9, 519-21.	0.4	17
53	Calmodulin Methyltransferase Is Required for Growth, Muscle Strength, Somatosensory Development and Brain Function. PLoS Genetics, 2015, 11, e1005388.	1.5	16
54	Two new mutations in the 3â€² coding region of the glycogen debranching enzyme in a glycogen storage disease type IIIa Ashkenazi Jewish patient. Journal of Inherited Metabolic Disease, 1998, 21, 141-148.	1.7	15

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55	Zinc Supplementation Increases the Level of Serum Insulin-Like Growth Factor-I but Does Not Promote Growth in Infants with Nonorganic Failure to Thrive. Hormone Research in Paediatrics, 1999, 52, 200-204.	0.8	15
56	A Novel Mutation Causing Complete Thyroxine-Binding Globulin Deficiency (TBG-CD-Negev) among the Bedouins in Southern Israel. Journal of Clinical Endocrinology and Metabolism, 2000, 85, 3687-3689.	1.8	15
57	A Phase II, Double-Blind, Randomized, Placebo-Controlled, Multicenter Study Evaluating the Efficacy and Safety of Alpha-1 Antitrypsin (AAT) (GlassiaÂ®) in the Treatment of Recent-Onset Type 1 Diabetes. International Journal of Molecular Sciences, 2019, 20, 6032.	1.8	15
58	Short-Term Growth Hormone Therapy Increases Serum Lipoprotein (a) Levels in Normal Short Children without Growth Hormone Deficiency. Hormone Research, 1996, 46, 38-40.	1.8	14
59	Combined adrenal failure and testicular adrenal rest tumor in a patient with nicotinamide nucleotide transhydrogenase deficiency. Journal of Pediatric Endocrinology and Metabolism, 2015, 28, 1187-90.	0.4	14
60	Human Calmodulin Methyltransferase: Expression, Activity on Calmodulin, and Hsp90 Dependence. PLoS ONE, 2012, 7, e52425.	1.1	13
61	Chromosomal Microdeletions and Genes' Functions: A cluster of chromosomal microdeletions and the deleted genes' functions. European Journal of Human Genetics, 2007, 15, 997-998.	1.4	12
62	PAX7mutation in a syndrome of failure to thrive, hypotonia, and global neurodevelopmental delay. Human Mutation, 2017, 38, 1671-1683.	1.1	12
63	Natural History and Clinical Manifestations of Hyponatremia and Hyperchlorhidrosis due to Carbonic Anhydrase XII Deficiency. Hormone Research in Paediatrics, 2014, 81, 336-342.	0.8	11
64	CT findings in neonatal hypothermia. Pediatric Radiology, 1998, 28, 414-417.	1.1	10
65	X-linked spondyloepiphyseal dysplasia tarda: A novelSEDL mutation in a Jewish Ashkenazi family and clinical intervention considerations. American Journal of Medical Genetics Part A, 2004, 125A, 45-48.	2.4	10
66	Decreased First Phase Insulin Response in Children with Congenital Insensitivity to Pain with Anhidrosis. Journal of Pediatric Endocrinology and Metabolism, 2005, 18, 873-7.	0.4	10
67	Thyrotropin Secreting Pituitary Adenoma Associated with Hypopituitarism and Diabetes Insipidus in an Adolescent Boy. Journal of Pediatric Endocrinology and Metabolism, 1995, 8, 47-50.	0.4	9
68	Increase of serum lipoprotein (a) levels during growth hormone therapy in normal short children. European Journal of Pediatrics, 1998, 157, 4-7.	1.3	9
69	Blood lipids and endothelial function in glycogen storage disease type III. Journal of Inherited Metabolic Disease, 1999, 22, 891-898.	1.7	9
70	European Multicentre Study in Children Born Small for Gestational Age with Persistent Short Stature: Comparison of Continuous and Discontinuous Growth Hormone Treatment Regimens. Hormone Research in Paediatrics, 2009, 71, 52-59.	0.8	9
71	Differences in cord serum retinol concentrations by ethnic origin in the Negev (Southern Israel). Early Human Development, 1995, 42, 123-130.	0.8	8
72	Subnormal Cortisol Response to Adrenocorticotropin in Isolated Partial 17,20-Lyase Deficiency. Journal of Pediatric Endocrinology and Metabolism, 1997, 10, 387-90.	0.4	8

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73	Total oxidant-scavenging capacities of plasma from glycogen storage disease type Ia patients as measured by cyclic voltammetry, FRAP and luminescence techniques. Journal of Inherited Metabolic Disease, 2009, 32, 651.	1.7	8
74	Ethnic differences in glycemic control and diabetic ketoacidosis rate among children with diabetes mellitus type 1 in the Negev area. Israel Medical Association Journal, 2013, 15, 267-70.	0.1	8
75	Glycogen storage disease type III in Israel: presentation and long-term outcome. Pediatric Endocrinology Reviews, 2014, 11, 318-23.	1.2	8
76	Serum Insulin-Like Growth Factors I and II Are Not Affected by Undernutrition in Children with Nonorganic Failure to Thrive. Hormone Research in Paediatrics, 1998, 49, 76-79.	0.8	7
77	Testicular Expressed Genes Are Missing in Familial X-Linked Kallmann Syndrome due to Two Large Different Deletions in Daughterâ€™s X Chromosomes. Hormone Research, 2008, 69, 276-283.	1.8	7
78	Ethnic and Gender Inequities in the Evaluation of Referred Short Children. Hormone Research in Paediatrics, 2011, 76, 50-55.	0.8	7
79	Primary Ovarian Insufficiency Nationwide Incidence Rate and Etiology Among Israeli Adolescents. Journal of Adolescent Health, 2020, 66, 603-609.	1.2	7
80	Air pollution and meteorological conditions during gestation and type 1 diabetes in offspring. Environment International, 2021, 154, 106546.	4.8	7
81	Carnitine-acylcarnitine translocase deficiency: Identification of a novel molecular defect in a Bedouin patient. Journal of Inherited Metabolic Disease, 2004, 27, 267-273.	1.7	6
82	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
83	Thyroid function tests in newborns of mothers with hypothyroidism. European Journal of Pediatrics, 2021, 180, 519-525.	1.3	6
84	The role of autonomy-supportive parenting in the competence, adherence and glycemic control of adolescents with type 1 diabetes. Diabetes Research and Clinical Practice, 2021, 173, 108679.	1.1	6
85	The effects of the COVID-19 pandemic on patients with lysosomal storage disorders in Israel. Orphanet Journal of Rare Diseases, 2021, 16, 379.	1.2	6
86	Youthâ€onset type 2 diabetes in Israel: A national cohort. Pediatric Diabetes, 2022, 23, 649-659.	1.2	6
87	Prenatal diagnosis of hypoparathyroidism retardation and dysmorphism (HRD) syndrome. Prenatal Diagnosis, 2000, 20, 475-477.	1.1	5
88	Obstructive sleep apnea and metabolic disorders in morbidly obese adolescents. Pediatric Pulmonology, 2021, 56, 3983-3990.	1.0	5
89	RFLPs for linkage analysis in families with glycogen storage disease type III. Journal of Inherited Metabolic Disease, 1995, 18, 207-210.	1.7	4
90	Micropituitarism and cortical dysplasia: an unknown association of two uncommon CNS disorders. European Radiology, 2001, 11, 1070-1072.	2.3	4

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91	Multiple Endocrine Deficiencies are Common in Hypoparathyroidismâ€“Retardationâ€“Dysmorphism Syndrome. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e907-e916.	1.8	4
92	The Childhood Component of the ICP Model Is Appropriate for Growth Analysis of Short Israeli Children. Hormone Research in Paediatrics, 2004, 62, 119-123.	0.8	3
93	Exertional rhabdomyolysis in carbonic anhydrase 12 deficiency. Journal of Pediatric Endocrinology and Metabolism, 2018, 31, 697-699.	0.4	3
94	A Novel Mutation Causing Complete Thyroxine-Binding Globulin Deficiency (TBG-CD-Negev) among the Bedouins in Southern Israel. , 0, .		3
95	Prenatal ultrasonic diagnosis of nonhypertrophic pyloric stenosis associated with intestinal malrotation. Journal of Clinical Ultrasound, 1994, 22, 52-54.	0.4	2
96	Generation of iPSC lines from two (BGUi002-A and BGUi003-A) homozygous p450 oxidoreductase-deficient patients and from one (BGUi001-A) heterozygous healthy family relative. Stem Cell Research, 2020, 48, 101975.	0.3	2
97	Acute hyperglycaemia can impair driving skill in young type 1 diabetes mellitus patients. Diabetes and Metabolism, 2021, 47, 101176.	1.4	2
98	Effect of a nutritional supplementation on growth and body composition in short and lean preadolescent boys: A randomised, doubleâ€blind, placeboâ€controlled study. Acta Paediatrica, International Journal of Paediatrics, 2021, , .	0.7	2
99	Aldosterone synthase (<scp>CYP11B2</scp>) deficiency among Palestinian infants: Three novel variants and genetic heterogeneity. American Journal of Medical Genetics, Part A, 2021, 185, 1033-1038.	0.7	2
100	Hypoparathyroidism, Dwarfism, Medullary Stenosis of Long Bones, and Eye Abnormalities (Kenny-Caffey Syndrome) and Hypoparathyroidism, Retardation, and Dysmorphism (Sanjad-Sakati) Syndrome. , 2015, , 215-224.		2
101	Once versus twice daily injections of growth hormone in children with idiopathic short stature. Acta Paediatrica, International Journal of Paediatrics, 1998, 87, 518-20.	0.7	2
102	High prevalence of hypophosphatasia in Southern Israel. Acta Paediatrica, International Journal of Paediatrics, 2020, 109, 851-852.	0.7	1
103	Multiple Acyl-CoA Dehydrogenase Deficiency with Variable Presentation Due to a Homozygous Mutation in a Bedouin Tribe. Genes, 2021, 12, 1140.	1.0	1
104	TSHB R75G is a founder variant and prevalent cause of low or undetectable TSH in Indian Jews. European Thyroid Journal, 2022, 11, .	1.2	1
105	Does gender affect the driving performance of young patients with diabetes?. Accident Analysis and Prevention, 2022, 167, 106569.	3.0	1
106	Long-Term Follow-Up of Pompe Patients in Israel and Gaza: Insights into Therapeutic Effects of Enzyme Replacement Therapy. Journal of Neuromuscular Diseases, 2015, 2, S65-S66.	1.1	0
107	An 8-year-old Boy With Fever and Abdominal Pain. Pediatric Infectious Disease Journal, 2018, 37, 958-958.	1.1	0
108	Acute Hyperglycemia may impair driving skills of young T1DM patients. Proceedings of the Human Factors and Ergonomics Society, 2019, 63, 1284-1284.	0.2	0

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109	Congenital neuroblastoma in a neonate with hypoparathyroidism-retardation-dysmorphism syndrome. Clinical Dysmorphology, 2020, 29, 46-48.	0.1	0
110	Infant botulism: be aware of this rare disease. Israel Medical Association Journal, 2007, 9, 682-3.	0.1	0
111	Long-Term Follow-Up of Pompe Patients in Israel and Gaza: Insights into Therapeutic Effects of Enzyme Replacement Therapy. Journal of Neuromuscular Diseases, 2015, 2, S65-S66.	1.1	0
112	Diagnosis, Management, and Possible Prevention of Hungry Bone Syndrome in an Adolescent with Primary Hyperparathyroidism and Vitamin D Deficiency. Israel Medical Association Journal, 2020, 22, 122-124.	0.1	0