

Eli HersHKovitz

List of Publications by Year in descending order

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Version: 2024-02-01

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	TSHB R75G is a founder variant and prevalent cause of low or undetectable TSH in Indian Jews. <i>European Thyroid Journal</i> , 2022, 11, .	1.2	1
2	Does gender affect the driving performance of young patients with diabetes?. <i>Accident Analysis and Prevention</i> , 2022, 167, 106569.	3.0	1
3	Youthâ€onset type 2 diabetes in Israel: A national cohort. <i>Pediatric Diabetes</i> , 2022, 23, 649-659.	1.2	6
4	Multiple Endocrine Deficiencies are Common in Hypoparathyroidismâ€Retardationâ€Dysmorphism Syndrome. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021, 106, e907-e916.	1.8	4
5	Acute hyperglycaemia can impair driving skill in young type 1 diabetes mellitus patients. <i>Diabetes and Metabolism</i> , 2021, 47, 101176.	1.4	2
6	The role of orotic acid measurement in routine newborn screening for urea cycle disorders. <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 606-617.	1.7	6
7	Thyroid function tests in newborns of mothers with hypothyroidism. <i>European Journal of Pediatrics</i> , 2021, 180, 519-525.	1.3	6
8	The role of autonomy-supportive parenting in the competence, adherence and glycemic control of adolescents with type 1 diabetes. <i>Diabetes Research and Clinical Practice</i> , 2021, 173, 108679.	1.1	6
9	Multiple Acyl-CoA Dehydrogenase Deficiency with Variable Presentation Due to a Homozygous Mutation in a Bedouin Tribe. <i>Genes</i> , 2021, 12, 1140.	1.0	1
10	Effect of a nutritional supplementation on growth and body composition in short and lean preadolescent boys: A randomised, doubleâ€blind, placeboâ€controlled study. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2021, , .	0.7	2
11	The effects of the COVID-19 pandemic on patients with lysosomal storage disorders in Israel. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 379.	1.2	6
12	Air pollution and meteorological conditions during gestation and type 1 diabetes in offspring. <i>Environment International</i> , 2021, 154, 106546.	4.8	7
13	Obstructive sleep apnea and metabolic disorders in morbidly obese adolescents. <i>Pediatric Pulmonology</i> , 2021, 56, 3983-3990.	1.0	5
14	Aldosterone synthase (<scp>CYP11B2</scp>) deficiency among Palestinian infants: Three novel variants and genetic heterogeneity. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1033-1038.	0.7	2
15	High prevalence of hypophosphatasia in Southern Israel. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2020, 109, 851-852.	0.7	1
16	Congenital neuroblastoma in a neonate with hypoparathyroidism-retardation-dysmorphism syndrome. <i>Clinical Dysmorphology</i> , 2020, 29, 46-48.	0.1	0
17	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. <i>Stem Cell Research</i> , 2020, 48, 101975.	0.3	2
18	Primary Ovarian Insufficiency Nationwide Incidence Rate and Etiology Among Israeli Adolescents. <i>Journal of Adolescent Health</i> , 2020, 66, 603-609.	1.2	7

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19	Diagnosis, Management, and Possible Prevention of Hungry Bone Syndrome in an Adolescent with Primary Hyperparathyroidism and Vitamin D Deficiency. <i>Israel Medical Association Journal</i> , 2020, 22, 122-124.	0.1	0
20	A Phase II, Double-Blind, Randomized, Placebo-Controlled, Multicenter Study Evaluating the Efficacy and Safety of Alpha-1 Antitrypsin (AAT) (Glassia [®]) in the Treatment of Recent-Onset Type 1 Diabetes. <i>International Journal of Molecular Sciences</i> , 2019, 20, 6032.	1.8	15
21	Acute Hyperglycemia may impair driving skills of young T1DM patients. <i>Proceedings of the Human Factors and Ergonomics Society</i> , 2019, 63, 1284-1284.	0.2	0
22	Pulmonary involvement in Niemann-Pick C type 1. <i>European Journal of Pediatrics</i> , 2018, 177, 1609-1615.	1.3	21
23	Exertional rhabdomyolysis in carbonic anhydrase 12 deficiency. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2018, 31, 697-699.	0.4	3
24	An 8-year-old Boy With Fever and Abdominal Pain. <i>Pediatric Infectious Disease Journal</i> , 2018, 37, 958-958.	1.1	0
25	<i>PAX7</i> mutation in a syndrome of failure to thrive, hypotonia, and global neurodevelopmental delay. <i>Human Mutation</i> , 2017, 38, 1671-1683.	1.1	12
26	Developing and evaluating rare disease educational materials co-created by expert clinicians and patients: the paradigm of congenital hypogonadotropic hypogonadism. <i>Orphanet Journal of Rare Diseases</i> , 2017, 12, 57.	1.2	26
27	Mitochondrial epileptic encephalopathy, 3-methylglutaconic aciduria and variable complex V deficiency associated with <i>TIMM50</i> mutations. <i>Clinical Genetics</i> , 2017, 91, 690-696.	1.0	28
28	Essential role of carbonic anhydrase XII in secretory gland fluid and HCO ₃ ⁻ secretion revealed by disease causing human mutation. <i>Journal of Physiology</i> , 2015, 593, 5299-5312.	1.3	37
29	Phosphoglucomutase-1 deficiency: Intrafamilial clinical variability and common secondary adrenal insufficiency. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 3139-3143.	0.7	20
30	Long-Term Follow-Up of Pompe Patients in Israel and Gaza: Insights into Therapeutic Effects of Enzyme Replacement Therapy. <i>Journal of Neuromuscular Diseases</i> , 2015, 2, S65-S66.	1.1	0
31	Combined adrenal failure and testicular adrenal rest tumor in a patient with nicotinamide nucleotide transhydrogenase deficiency. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2015, 28, 1187-90.	0.4	14
32	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
33	Calmodulin Methyltransferase Is Required for Growth, Muscle Strength, Somatosensory Development and Brain Function. <i>PLoS Genetics</i> , 2015, 11, e1005388.	1.5	16
34	Long-Term Follow-Up of Pompe Patients in Israel and Gaza: Insights into Therapeutic Effects of Enzyme Replacement Therapy. <i>Journal of Neuromuscular Diseases</i> , 2015, 2, S65-S66.	1.1	0
35	Trends in the incidence of type 1 diabetes among Jews and Arabs in Israel. <i>Pediatric Diabetes</i> , 2014, 15, 422-427.	1.2	27
36	Natural History and Clinical Manifestations of Hyponatremia and Hyperchlorhidrosis due to Carbonic Anhydrase XII Deficiency. <i>Hormone Research in Paediatrics</i> , 2014, 81, 336-342.	0.8	11

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37	Glycogen storage disease type III in Israel: presentation and long-term outcome. <i>Pediatric Endocrinology Reviews</i> , 2014, 11, 318-23.	1.2	8
38	Varied Clinical Presentations of Seven Patients With Mutations in <i>CYP11A1</i> Encoding the Cholesterol Side-Chain Cleavage Enzyme, P450 _{scc} . <i>Journal of Clinical Endocrinology and Metabolism</i> , 2013, 98, 713-720.	1.8	59
39	Ethnic differences in glycemic control and diabetic ketoacidosis rate among children with diabetes mellitus type 1 in the Negev area. <i>Israel Medical Association Journal</i> , 2013, 15, 267-70.	0.1	8
40	The utility of basal serum LH in prediction of central precocious puberty in girls. <i>European Journal of Endocrinology</i> , 2012, 166, 295-299.	1.9	89
41	Successful immune tolerance induction to enzyme replacement therapy in CRIM-negative infantile Pompe disease. <i>Genetics in Medicine</i> , 2012, 14, 135-142.	1.1	183
42	Domain III of the <i>T. thermophilus</i> 23S rRNA folds independently to a near-native state. <i>Rna</i> , 2012, 18, 752-758.	1.6	21
43	Human Calmodulin Methyltransferase: Expression, Activity on Calmodulin, and Hsp90 Dependence. <i>PLoS ONE</i> , 2012, 7, e52425.	1.1	13
44	Increase in the incidence of type 1 diabetes in Israeli children following the Second Lebanon War. <i>Pediatric Diabetes</i> , 2012, 13, 326-333.	1.2	26
45	TMEM70 mutations are a common cause of nuclear encoded ATP synthase assembly defect: further delineation of a new syndrome. <i>Journal of Medical Genetics</i> , 2011, 48, 177-182.	1.5	61
46	Autosomal recessive hyponatremia due to isolated salt wasting in sweat associated with a mutation in the active site of Carbonic Anhydrase 12. <i>Human Genetics</i> , 2011, 129, 397-405.	1.8	35
47	Ethnic and Gender Inequities in the Evaluation of Referred Short Children. <i>Hormone Research in Paediatrics</i> , 2011, 76, 50-55.	0.8	7
48	Woodhouse-Sakati Syndrome in an Israeli-Arab Family Presenting with Youth-Onset Diabetes Mellitus and Delayed Puberty. <i>Hormone Research in Paediatrics</i> , 2011, 75, 362-366.	0.8	20
49	Autosomal-Recessive Hypophosphatemic Rickets Is Associated with an Inactivation Mutation in the ENPP1 Gene. <i>American Journal of Human Genetics</i> , 2010, 86, 273-278.	2.6	262
50	Neonatal hyperthyrotropinemia: population characteristics, diagnosis, management and outcome after cessation of therapy. <i>Clinical Endocrinology</i> , 2010, 72, 264-271.	1.2	36
51	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. <i>Hormone Research</i> , 2009, 72, 287-301.	1.8	69
52	European Multicentre Study in Children Born Small for Gestational Age with Persistent Short Stature: Comparison of Continuous and Discontinuous Growth Hormone Treatment Regimens. <i>Hormone Research in Paediatrics</i> , 2009, 71, 52-59.	0.8	9
53	Total oxidant-scavenging capacities of plasma from glycogen storage disease type Ia patients as measured by cyclic voltammetry, FRAP and luminescence techniques. <i>Journal of Inherited Metabolic Disease</i> , 2009, 32, 651.	1.7	8
54	The influence of diet and/or exercise and parental compliance on health-related quality of life in obese children. <i>Nutrition Research</i> , 2009, 29, 397-404.	1.3	51

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55	Mitochondrial complex I deficiency caused by a deleterious NDUF A11 mutation. <i>Annals of Neurology</i> , 2008, 63, 405-408.	2.8	103
56	Testicular Expressed Genes Are Missing in Familial X-Linked Kallmann Syndrome due to Two Large Different Deletions in Daughters' X Chromosomes. <i>Hormone Research</i> , 2008, 69, 276-283.	1.8	7
57	Homozygous Mutation G539R in the Gene for P450 Oxidoreductase in a Family Previously Diagnosed as Having 17,20-Lyase Deficiency. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2008, 93, 3584-3588.	1.8	82
58	Hypoparathyroidism, Retardation, and Dysmorphism Syndrome: Impaired Early Growth and Increased Susceptibility to Severe Infections Due to Hyposplenism and Impaired Polymorphonuclear Cell Functions. <i>Pediatric Research</i> , 2007, 62, 505-509.	1.1	19
59	Parathyroid Development and the Role of Tubulin Chaperone E. <i>Hormone Research in Paediatrics</i> , 2007, 67, 12-21.	0.8	22
60	Chromosomal Microdeletions and Genes' Functions: A cluster of chromosomal microdeletions and the deleted genes' functions. <i>European Journal of Human Genetics</i> , 2007, 15, 997-998.	1.4	12
61	Incidence of type 1 diabetes mellitus in the 0- to 17-yr-old Israel population, 1997-2003. <i>Pediatric Diabetes</i> , 2007, 8, 60-66.	1.2	24
62	Infant botulism: be aware of this rare disease. <i>Israel Medical Association Journal</i> , 2007, 9, 682-3.	0.1	0
63	Single nucleotide RNA choreography. <i>Nucleic Acids Research</i> , 2006, 34, 1481-1491.	6.5	43
64	A retrospective, multinational, multicenter study on the natural history of infantile-onset Pompe disease. <i>Journal of Pediatrics</i> , 2006, 148, 671-676.e2.	0.9	500
65	Statistical Analysis of RNA Backbone. <i>IEEE/ACM Transactions on Computational Biology and Bioinformatics</i> , 2006, 3, 33-46.	1.9	33
66	Treatment of X-linked childhood cerebral adrenoleukodystrophy by the use of an allogeneic stem cell transplantation with reduced intensity conditioning regimen. <i>Clinical Transplantation</i> , 2005, 19, 840-847.	0.8	21
67	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. <i>Pediatric Research</i> , 2005, 57, 587-590.	1.1	19
68	Decreased First Phase Insulin Response in Children with Congenital Insensitivity to Pain with Anhidrosis. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2005, 18, 873-7.	0.4	10
69	Deficiency of the ADP-Forming Succinyl-CoA Synthase Activity Is Associated with Encephalomyopathy and Mitochondrial DNA Depletion. <i>American Journal of Human Genetics</i> , 2005, 76, 1081-1086.	2.6	284
70	The 2p21 deletion syndrome: Characterization of the transcription content. <i>Genomics</i> , 2005, 86, 195-211.	1.3	34
71	Childhood Obesity. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2005, 90, 1871-1887.	1.8	459
72	Thyroglossal Duct Carcinoma in Children: Case Presentation and Review of the Literature. <i>Thyroid</i> , 2004, 14, 777-785.	2.4	83

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73	The Childhood Component of the ICP Model Is Appropriate for Growth Analysis of Short Israeli Children. <i>Hormone Research in Paediatrics</i> , 2004, 62, 119-123.	0.8	3
74	Hypoparathyroidism-Retardation-Dysmorphism (HRD) Syndrome - A Review. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2004, 17, 1583-90.	0.4	26
75	Carnitine-acylcarnitine translocase deficiency: Identification of a novel molecular defect in a Bedouin patient. <i>Journal of Inherited Metabolic Disease</i> , 2004, 27, 267-273.	1.7	6
76	X-linked spondyloepiphyseal dysplasia tarda: A novel SEDL mutation in a Jewish Ashkenazi family and clinical intervention considerations. <i>American Journal of Medical Genetics Part A</i> , 2004, 125A, 45-48.	2.4	10
77	Defective mitochondrial translation caused by a ribosomal protein (MRPS16) mutation. <i>Annals of Neurology</i> , 2004, 56, 734-738.	2.8	205
78	Retinol concentration in maternal and cord serum: its relation to birth weight in healthy motherâ€“infant pairs. <i>Early Human Development</i> , 2003, 71, 19-28.	0.8	36
79	Prenatal exclusion of Leigh syndrome due to T8993C mutation in the mitochondrial DNA. <i>Prenatal Diagnosis</i> , 2003, 23, 31-33.	1.1	35
80	Automated identification of RNA conformational motifs: theory and application to the HM LSU 23S rRNA. <i>Nucleic Acids Research</i> , 2003, 31, 6249-6257.	6.5	54
81	Cerebral X-linked adrenoleukodystrophy in a girl with Xq27-Ter deletion. <i>Annals of Neurology</i> , 2002, 52, 234-237.	2.8	26
82	Mutation of TBCE causes hypoparathyroidismâ€“retardationâ€“dysmorphism and autosomal recessive Kennyâ€“Caffey syndrome. <i>Nature Genetics</i> , 2002, 32, 448-452.	9.4	248
83	A Recessive Contiguous Gene Deletion of Chromosome 2p16 Associated with Cystinuria and a Mitochondrial Disease. <i>American Journal of Human Genetics</i> , 2001, 69, 869-875.	2.6	69
84	Neurophysiologic studies in congenital insensitivity to pain with anhidrosis. <i>Pediatric Neurology</i> , 2001, 25, 397-400.	1.0	41
85	Continuous glucose monitoring in children with glycogen storage disease type I. <i>Journal of Inherited Metabolic Disease</i> , 2001, 24, 863-869.	1.7	40
86	Micropituitarism and cortical dysplasia: an unknown association of two uncommon CNS disorders. <i>European Radiology</i> , 2001, 11, 1070-1072.	2.3	4
87	Congenital insensitivity to pain with anhidrosis (CIPA) in Israeli-Bedouins: Genetic heterogeneity, novel mutations in the TRKA/NGF receptor gene, clinical findings, and results of nerve conduction studies. <i>American Journal of Medical Genetics Part A</i> , 2000, 92, 353-360.	2.4	129
88	Prenatal diagnosis of hypoparathyroidism retardation and dysmorphism (HRD) syndrome. <i>Prenatal Diagnosis</i> , 2000, 20, 475-477.	1.1	5
89	A Novel Mutation Causing Complete Thyroxine-Binding Globulin Deficiency (TBG-CD-Negev) among the Bedouins in Southern Israel. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2000, 85, 3687-3689.	1.8	15
90	Zinc Supplementation Increases the Level of Serum Insulin-Like Growth Factor-I but Does Not Promote Growth in Infants with Nonorganic Failure to Thrive. <i>Hormone Research in Paediatrics</i> , 1999, 52, 200-204.	0.8	15

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91	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
92	Blood lipids and endothelial function in glycogen storage disease type III. Journal of Inherited Metabolic Disease, 1999, 22, 891-898.	1.7	9
93	Lipoamide Dehydrogenase Deficiency Due to a Novel Mutation in the Interface Domain. Biochemical and Biophysical Research Communications, 1999, 262, 163-166.	1.0	51
94	Two new mutations in the 3' 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
95	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
96	CT findings in neonatal hypothermia. Pediatric Radiology, 1998, 28, 414-417.	1.1	10
97	The Gene for Glycogen-Storage Disease Type 1b Maps to Chromosome 11q23. American Journal of Human Genetics, 1998, 62, 400-405.	2.6	126
98	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
99	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
100	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
101	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
102	Glycogen storage disease type 1a in Israel: Biochemical, clinical, and mutational studies. , 1997, 72, 286-290.		35
103	Adrenal Insufficiency After Achalasia in the Triple-A Syndrome. Clinical Pediatrics, 1996, 35, 99-100.	0.4	20
104	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
105	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
106	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
107	RFLPs for linkage analysis in families with glycogen storage disease type III. Journal of Inherited Metabolic Disease, 1995, 18, 207-210.	1.7	4
108	Differences in cord serum retinol concentrations by ethnic origin in the Negev (Southern Israel). Early Human Development, 1995, 42, 123-130.	0.8	8

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109	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
110	Prenatal ultrasonic diagnosis of nonhypertrophic pyloric stenosis associated with intestinal malrotation. Journal of Clinical Ultrasound, 1994, 22, 52-54.	0.4	2
111	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
112	A Novel Mutation Causing Complete Thyroxine-Binding Globulin Deficiency (TBG-CD-Negev) among the Bedouins in Southern Israel. , 0, .		3