Ben Pode-Shakked

List of Publications by Year
in descending order

60	1,142	535685 17	536525 29
papers	citations	h-index	g-index

62	62	62	2399
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Whole-exome sequencing reveals a monogenic cause in 56% of individuals with laterality disorders and associated congenital heart defects. Journal of Medical Genetics, 2022, 59, 691-696.	1.5	14
2	Broadening the phenotype of LRRK1 mutations - Features of malignant osteopetrosis and optic nerve atrophy with intrafamilial variable expressivity. European Journal of Medical Genetics, 2022, 65, 104383.	0.7	10
3	A multidisciplinary nephrogenetic referral clinic for children and adultsâ€"diagnostic achievements and insights. Pediatric Nephrology, 2022, 37, 1623-1646.	0.9	12
4	Refining the Phenotypic Spectrum of KMT5B-Associated Developmental Delay. Frontiers in Pediatrics, 2022, 10, 844845.	0.9	8
5	What Can We Learn from the Parents of Children Affected with Mucopolysaccharidosis Type III-A in Israel?. Molecular Syndromology, 2022, 13, 45-49.	0.3	0
6	Glycogen Storage Disease type IA refractory to cornstarch: Can next generation sequencing offer a solution?. European Journal of Medical Genetics, 2022, , 104518.	0.7	1
7	Hereditary neuropathy with liability to pressure palsies (HNPP): Intrafamilial phenotypic variability and early childhood refusal to walk as the presenting symptom. Italian Journal of Pediatrics, 2022, 48, .	1.0	1
8	Severe Protein C Deficiency due to Novel Biallelic Variants in PROC and Their Phenotype Correlation. Acta Haematologica, 2021, 144, 327-331.	0.7	1
9	Clinical phenotypes of infantile onset CACNA1A-related disorder. European Journal of Paediatric Neurology, 2021, 30, 144-154.	0.7	13
10	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
11	SCUBE3 loss-of-function causes a recognizable recessive developmental disorder due to defective bone morphogenetic protein signaling. American Journal of Human Genetics, 2021, 108, 115-133.	2.6	37
12	DLG4-related synaptopathy: a new rare brain disorder. Genetics in Medicine, 2021, 23, 888-899.	1.1	16
13	Impaired complex I repair causes recessive Leberâ€™s hereditary optic neuropathy. Journal of Clinical Investigation, 2021, 131, .	3.9	89
14	Ophthalmic manifestations in Kabuki (make-up) syndrome: A single-center pediatric cohort and systematic review of the literature. European Journal of Medical Genetics, 2021, 64, 104210.	0.7	2
15	Phenotypic expansion of CACNA1C-associated disorders to include isolated neurological manifestations. Genetics in Medicine, 2021, 23, 1922-1932.	1.1	16
16	Multiple Acyl-CoA Dehydrogenase Deficiency with Variable Presentation Due to a Homozygous Mutation in a Bedouin Tribe. Genes, 2021, 12, 1140.	1.0	1
17	Deep intronic variant in the ARSB gene as the genetic cause for Maroteauxâ€“Lamy syndrome (MPS VI). American Journal of Medical Genetics, Part A, 2021, 185, 3804-3809.	0.7	3
18	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

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19	Kidney and urinary tract findings among patients with Kabuki (make-up) syndrome. Pediatric Nephrology, 2021, 36, 4009-4012.	0.9	3
20	A single center experience with publicly funded clinical exome sequencing for neurodevelopmental disorders or multiple congenital anomalies. Scientific Reports, 2021, 11, 19099.	1.6	13
21	The Effects of a Ketogenic Diet on Patients with Dihydrolipoamide Dehydrogenase Deficiency. Nutrients, 2021, 13, 3523.	1.7	13
22	Enhanced Collagen Deposition in the Duodenum of Patients with Hyaline Fibromatosis Syndrome and Protein Losing Enteropathy. International Journal of Molecular Sciences, 2020, 21, 8200.	1.8	3
23	Clues and challenges in the diagnosis of intermittent maple syrup urine disease. European Journal of Medical Genetics, 2020, 63, 103901.	0.7	26
24	A founder truncating variant in GDF1 causes autosomalâ€recessive right isomerism and associated congenital heart defects in multiplex Arab kindreds. American Journal of Medical Genetics, Part A, 2020, 182, 987-993.	0.7	5
25	Shared facial phenotype of patients with mucolipidosis type IV: A clinical observation reaffirmed by next generation phenotyping. European Journal of Medical Genetics, 2020, 63, 103927.	0.7	10
26	Bi-allelic Variants in RALGAPA1 Cause Profound Neurodevelopmental Disability, Muscular Hypotonia, Infantile Spasms, and Feeding Abnormalities. American Journal of Human Genetics, 2020, 106, 246-255.	2.6	17
27	Phenotype variability in Hajdu-Cheney syndrome. European Journal of Medical Genetics, 2019, 62, 35-38.	0.7	12
28	Diaphanospondylodysostosis: Refining the prenatal diagnosis of a rare skeletal disorder. European Journal of Medical Genetics, 2019, 62, 167-171.	0.7	9
29	Evaluation of Diagnostic Yield in Fetal Whole-Exome Sequencing: A Report on 45 Consecutive Families. Frontiers in Genetics, 2019, 10, 425.	1.1	33
30	Whole exome sequencing in childhood-onset lupus frequently detects single gene etiologies. Pediatric Rheumatology, 2019, 17, 52.	0.9	34
31	Novel homozygous ENPP1 mutation causes generalized arterial calcifications of infancy, thrombocytopenia, and cardiovascular and central nervous system syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 2112-2118.	0.7	16
32	Cerebral and portal vein thrombosis, macrocephaly and atypical absence seizures in Glycosylphosphatidyl inositol deficiency due to a PIGM promoter mutation. Molecular Genetics and Metabolism, 2019, 128, 151-161.	0.5	9
33	Noncoding deletions reveal a gene that is critical for intestinal function. Nature, 2019, 571, 107-111.	13.7	24
34	Combined Gestational Age- and Birth Weightâ€"Adjusted Cutoffs for Newborn Screening of Congenital Adrenal Hyperplasia. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 3172-3180.	1.8	22
35	Dominant PAX2 mutations may cause steroid-resistant nephrotic syndrome and FSGS in children. Pediatric Nephrology, 2019, 34, 1607-1613.	0.9	31
36	Progressive Pseudorheumatoid Dysplasia resolved by whole exome sequencing: a novel mutation in WISP3 and review of the literature. BMC Medical Genetics, 2019, 20, 53.	2.1	11

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37	BRPF1â€associated intellectual disability, ptosis, and facial dysmorphism in a multiplex family. Molecular Genetics & Genomic Medicine, 2019, 7, e665.	0.6	21
38	Acid-Induced Downregulation of ASS1 Contributes to the Maintenance of Intracellular pH in Cancer. Cancer Research, 2019, 79, 518-533.	0.4	36
39	LMOD3â€Associated Nemaline Myopathy: Prenatal Ultrasonographic, Pathologic, and Molecular Findings. Journal of Ultrasound in Medicine, 2018, 37, 1827-1833.	0.8	14
40	Severe fetal hydronephrosis: the added value of associated congenital anomalies of the kidneys and urinary tract (<scp>CAKUT</scp>) in the prediction of postnatal outcome. Prenatal Diagnosis, 2018, 38, 179-183.	1.1	6
41	A novel inborn error of the coenzyme Q10 biosynthesis pathway: cerebellar ataxia and static encephalomyopathy due to COQ5 Câ€methyltransferase deficiency. Human Mutation, 2018, 39, 69-79.	1.1	43
42	Mutations in PPCS, Encoding Phosphopantothenoylcysteine Synthetase, Cause Autosomal-Recessive Dilated Cardiomyopathy. American Journal of Human Genetics, 2018, 102, 1018-1030.	2.6	42
43	Cardiac failure in very long chain acyl-CoA dehydrogenase deficiency requiring extracorporeal membrane oxygenation (ECMO) treatment: A case report and review of the literature. Molecular Genetics and Metabolism Reports, 2017, 10, 5-7.	0.4	10
44	Biallelic Mutations in DNAJC12 Cause Hyperphenylalaninemia, Dystonia, and Intellectual Disability. American Journal of Human Genetics, 2017, 100, 257-266.	2.6	127
45	Nonobstructive Diffuse Dilated Bowel Loops: Prenatal Diagnosis, Fetal Characteristics and Neonatal Outcomes. Journal of Ultrasound in Medicine, 2017, 36, 149-154.	0.8	4
46	Deleterious variants in TRAK1 disrupt mitochondrial movement and cause fatal encephalopathy. Brain, 2017, 140, 568-581.	3.7	53
47	Elucidating the behavioral phenotype of patients affected with mucolipidosis IV: What can we learn from the parents?. European Journal of Medical Genetics, 2017, 60, 340-344.	0.7	3
48	Mutations in TMEM260 Cause a Pediatric Neurodevelopmental, Cardiac, and Renal Syndrome. American Journal of Human Genetics, 2017, 100, 666-675.	2.6	22
49	Exome sequencing in Jewish and Arab patients with rhabdomyolysis reveals single-gene etiology in 43% of cases. Pediatric Nephrology, 2017, 32, 2273-2282.	0.9	40
50	Expanding the molecular diversity and phenotypic spectrum of glycerol 3â€phosphate dehydrogenase 1 deficiency. Journal of Inherited Metabolic Disease, 2016, 39, 689-695.	1.7	24
51	A Novel Mutation in a Critical Region for the Methyl Donor Binding in DNMT3B Causes Immunodeficiency, Centromeric Instability, and Facial Anomalies Syndrome (ICF). Journal of Clinical Immunology, 2016, 36, 801-809.	2.0	12
52	Dissecting Stages of Human Kidney Development and Tumorigenesis with Surface Markers Affords Simple Prospective Purification of Nephron Stem Cells. Scientific Reports, 2016, 6, 23562.	1.6	47
53	Prenatal diagnosis of 17q12 deletion syndrome: from fetal hyperechogenic kidneys to high risk for autism. Prenatal Diagnosis, 2016, 36, 1027-1032.	1.1	22
54	Congenital protein losing enteropathy: an inborn error of lipid metabolism due to DGAT1 mutations. European Journal of Human Genetics, 2016, 24, 1268-1273.	1.4	37

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55	A novel mutation in the C7orf11 gene causes nonphotosensitive trichothiodystrophy in a multiplex highly consanguineous kindred. European Journal of Medical Genetics, 2015, 58, 685-688.	0.7	7
56	Clinical Variability in a Family with an Ectodermal Dysplasia Syndrome and a Nonsense Mutation in theTP63Gene. Fetal and Pediatric Pathology, 2015, 34, 400-406.	0.4	5
57	Bitterness of Glucose/Galactose. Journal of Pediatric Gastroenterology and Nutrition, 2014, 58, 57-60.	0.9	14
58	Waterhouse Friderichsen Syndrome Complicating FulminantEnterobacter CloacaeSepsis in a Preterm Infant: The Unresolved Issue of Corticosteroids. Fetal and Pediatric Pathology, 2014, 33, 104-108.	0.4	2
59	Glutaric Aciduria type I and acute renal failure â€" Coincidence or causality?. Molecular Genetics and Metabolism Reports, 2014, 1, 170-175.	0.4	9
60	Man made disease: Clinical manifestations of low phenylalanine levels in an inadequately treated phenylketonuria patient and mouse study. Molecular Genetics and Metabolism, 2013, 110, S66-S70.	0.5	14