# Ben Pode-Shakked

## List of Publications by Citations



| 58 | 620 | 13 | 22 |
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| papers | citations | h-index | g-index |
| 62 | 904 | 5.2 | 3.28 |
| ext. papers | ext. citations | avg, IF | L-index |

| # | Paper | IF | Citations |
|---|---|---|---|
| 58 | Biallelic Mutations in DNAJC12 Cause Hyperphenylalaninemia, Dystonia, and Intellectual Disability. *American Journal of Human Genetics*, **2017**, 100, 257-266 | 11 | 81 |
| 57 | Deleterious variants in TRAK1 disrupt mitochondrial movement and cause fatal encephalopathy. *Brain*, **2017**, 140, 568-581 | 11.2 | 40 |
| 56 | Dissecting Stages of Human Kidney Development and Tumorigenesis with Surface Markers Affords Simple Prospective Purification of Nephron Stem Cells. *Scientific Reports*, **2016**, 6, 23562 | 4.9 | 38 |
| 55 | A novel inborn error of the coenzyme Q10 biosynthesis pathway: cerebellar ataxia and static encephalomyopathy due to COQ5 C-methyltransferase deficiency. *Human Mutation*, **2018**, 39, 69-79 | 4.7 | 29 |
| 54 | Congenital protein losing enteropathy: an inborn error of lipid metabolism due to DGAT1 mutations. *European Journal of Human Genetics*, **2016**, 24, 1268-73 | 5.3 | 29 |
| 53 | Mutations in PPCS, Encoding Phosphopantothenoylcysteine Synthetase, Cause Autosomal-Recessive Dilated Cardiomyopathy. *American Journal of Human Genetics*, **2018**, 102, 1018-1030 | 11 | 29 |
| 52 | Impaired complex I repair causes recessive Leber's hereditary optic neuropathy. *Journal of Clinical Investigation*, **2021**, 131, | 15.9 | 27 |
| 51 | Acid-Induced Downregulation of ASS1 Contributes to the Maintenance of Intracellular pH in Cancer. *Cancer Research*, **2019**, 79, 518-533 | 10.1 | 23 |
| 50 | Dominant PAX2 mutations may cause steroid-resistant nephrotic syndrome and FSGS in children. *Pediatric Nephrology*, **2019**, 34, 1607-1613 | 3.2 | 21 |
| 49 | Exome sequencing in Jewish and Arab patients with rhabdomyolysis reveals single-gene etiology in 43% of cases. *Pediatric Nephrology*, **2017**, 32, 2273-2282 | 3.2 | 20 |
| 48 | Evaluation of Diagnostic Yield in Fetal Whole-Exome Sequencing: A Report on 45 Consecutive Families. *Frontiers in Genetics*, **2019**, 10, 425 | 4.5 | 19 |
| 47 | Expanding the molecular diversity and phenotypic spectrum of glycerol 3-phosphate dehydrogenase 1 deficiency. *Journal of Inherited Metabolic Disease*, **2016**, 39, 689-695 | 5.4 | 18 |
| 46 | Whole exome sequencing in childhood-onset lupus frequently detects single gene etiologies. *Pediatric Rheumatology*, **2019**, 17, 52 | 3.5 | 16 |
| 45 | Clues and challenges in the diagnosis of intermittent maple syrup urine disease. *European Journal of Medical Genetics*, **2020**, 63, 103901 | 2.6 | 13 |
| 44 | Mutations in TMEM260 Cause a Pediatric Neurodevelopmental, Cardiac, and Renal Syndrome. *American Journal of Human Genetics*, **2017**, 100, 666-675 | 11 | 12 |
| 43 | Noncoding deletions reveal a gene that is critical for intestinal function. *Nature*, **2019**, 571, 107-111 | 50.4 | 12 |
| 42 | Combined Gestational Age- and Birth Weight-Adjusted Cutoffs for Newborn Screening of Congenital Adrenal Hyperplasia. *Journal of Clinical Endocrinology and Metabolism*, **2019**, 104, 3172-3180 | 5.6 | 11 |

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| 41 | BRPF1-associated intellectual disability, ptosis, and facial dysmorphism in a multiplex family. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e665 | 2.3 | 11 |
| 40 | Man made disease: clinical manifestations of low phenylalanine levels in an inadequately treated phenylketonuria patient and mouse study. *Molecular Genetics and Metabolism*, **2013**, 110 Suppl, S66-70 | 3.7 | 11 |
| 39 | Bitterness of glucose/galactose: novel mutations in the SLC5A1 gene. *Journal of Pediatric Gastroenterology and Nutrition*, **2014**, 58, 57-60 | 2.8 | 11 |
| 38 | 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 | 1.8 | 10 |
| 37 | 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 | 2.5 | 10 |
| 36 | Prenatal diagnosis of 17q12 deletion syndrome: from fetal hyperechogenic kidneys to high risk for autism. *Prenatal Diagnosis*, **2016**, 36, 1027-1032 | 3.2 | 9 |
| 35 | 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 | 8 |
| 34 | 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 | 2.6 | 8 |
| 33 | 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 | 5.7 | 8 |
| 32 | 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 | 11 | 8 |
| 31 | 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 | 3.7 | 7 |
| 30 | LMOD3-Associated Nemaline Myopathy: Prenatal Ultrasonographic, Pathologic, and Molecular Findings. *Journal of Ultrasound in Medicine*, **2018**, 37, 1827-1833 | 2.9 | 7 |
| 29 | Phenotype variability in Hajdu-Cheney syndrome. *European Journal of Medical Genetics*, **2019**, 62, 35-38 | 2.6 | 7 |
| 28 | Diaphanospondylodysostosis: Refining the prenatal diagnosis of a rare skeletal disorder. *European Journal of Medical Genetics*, **2019**, 62, 167-171 | 2.6 | 7 |
| 27 | A novel mutation in the C7orf11 gene causes nonphotosensitive trichothiodystrophy in a multiplex highly consanguineous kindred. *European Journal of Medical Genetics*, **2015**, 58, 685-8 | 2.6 | 6 |
| 26 | Glutaric Aciduria type I and acute renal failure - Coincidence or causality?. *Molecular Genetics and Metabolism Reports*, **2014**, 1, 170-175 | 1.8 | 6 |
| 25 | 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 | 11 | 6 |
| 24 | Whole-exome sequencing reveals a monogenic cause in 56% of individuals with laterality disorders and associated congenital heart defects. *Journal of Medical Genetics*, **2021**, | 5.8 | 6 |

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| 23 | Clinical Variability in a Family with an Ectodermal Dysplasia Syndrome and a Nonsense Mutation in the TP63 Gene. *Fetal and Pediatric Pathology*, **2015**, 34, 400-6 | 1.7 | 4 |
| 22 | A founder truncating variant in GDF1 causes autosomal-recessive right isomerism and associated congenital heart defects in multiplex Arab kindreds. *American Journal of Medical Genetics, Part A*, **2020**, 182, 987-993 | 2.5 | 4 |
| 21 | Clinical phenotypes of infantile onset CACNA1A-related disorder. *European Journal of Paediatric Neurology*, **2021**, 30, 144-154 | 3.8 | 4 |
| 20 | 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 | 2.6 | 3 |
| 19 | Severe fetal hydronephrosis: the added value of associated congenital anomalies of the kidneys and urinary tract (CAKUT) in the prediction of postnatal outcome. *Prenatal Diagnosis*, **2018**, 38, 179-183 | 3.2 | 3 |
| 18 | The Effects of a Ketogenic Diet on Patients with Dihydrolipoamide Dehydrogenase Deficiency. *Nutrients*, **2021**, 13, | 6.7 | 3 |
| 17 | The effects of the COVID-19 pandemic on patients with lysosomal storage disorders in Israel. *Orphanet Journal of Rare Diseases*, **2021**, 16, 379 | 4.2 | 3 |
| 16 | Phenotypic expansion of CACNA1C-associated disorders to include isolated neurological manifestations. *Genetics in Medicine*, **2021**, 23, 1922-1932 | 8.1 | 2 |
| 15 | The role of orotic acid measurement in routine newborn screening for urea cycle disorders. *Journal of Inherited Metabolic Disease*, **2021**, 44, 606-617 | 5.4 | 2 |
| 14 | A single center experience with publicly funded clinical exome sequencing for neurodevelopmental disorders or multiple congenital anomalies. *Scientific Reports*, **2021**, 11, 19099 | 4.9 | 2 |
| 13 | Refining the Phenotypic Spectrum of -Associated Developmental Delay.. *Frontiers in Pediatrics*, **2022**, 10, 844845 | 3.4 | 2 |
| 12 | Nonobstructive Diffuse Dilated Bowel Loops: Prenatal Diagnosis, Fetal Characteristics and Neonatal Outcomes. *Journal of Ultrasound in Medicine*, **2017**, 36, 149-154 | 2.9 | 1 |
| 11 | A multidisciplinary nephrogenetic referral clinic for children and adults-diagnostic achievements and insights.. *Pediatric Nephrology*, **2022**, 1 | 3.2 | 1 |
| 10 | 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 | 2.6 | 1 |
| 9 | Deep intronic variant in the ARSB gene as the genetic cause for Maroteaux-Lamy syndrome (MPS VI). *American Journal of Medical Genetics, Part A*, **2021**, 185, 3804-3809 | 2.5 | 1 |
| 8 | Broadening the phenotype of LRRK1 mutations - Features of malignant osteopetrosis and optic nerve atrophy with intrafamilial variable expressivity. *European Journal of Medical Genetics*, **2021**, 65, 104383 | 2.6 | 0 |
| 7 | Enhanced Collagen Deposition in the Duodenum of Patients with Hyaline Fibromatosis Syndrome and Protein Losing Enteropathy. *International Journal of Molecular Sciences*, **2020**, 21, | 6.3 | 0 |
| 6 | Severe Protein C Deficiency due to Novel Biallelic Variants in PROC and Their Phenotype Correlation. *Acta Haematologica*, **2021**, 144, 327-331 | 2.7 | 0 |

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| 5 | DLG4-related synaptopathy: a new rare brain disorder. *Genetics in Medicine*, **2021**, 23, 888-899 | 8.1 | 0 |
| 4 | Kidney and urinary tract findings among patients with Kabuki (make-up) syndrome. *Pediatric Nephrology*, **2020**, 36, 4009-4012 | 3.2 | 0 |
| 3 | Glycogen Storage Disease type IA refractory to cornstarch: Can next generation sequencing offer a solution?. *European Journal of Medical Genetics*, **2022**, 104518 | 2.6 | 0 |
| 2 | Waterhouse Friderichsen syndrome complicating fulminant Enterobacter cloacae sepsis in a preterm infant: the unresolved issue of corticosteroids. *Fetal and Pediatric Pathology*, **2014**, 33, 104-8 | 1.7 | |
| 1 | What Can We Learn from the Parents of Children Affected with Mucopolysaccharidosis Type III-A in Israel?. *Molecular Syndromology*, **2022**, 13, 45-49 | 1.5 | |