

Ortal Barel

List of Publications by Year in descending order

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Version: 2024-02-01

54
papers

1,052
citations

516681

16
h-index

501174

28
g-index

55
all docs

55
docs citations

55
times ranked

2071
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutations in <i>STN1</i> cause Coats plus syndrome and are associated with genomic and telomere defects. <i>Journal of Experimental Medicine</i> , 2016, 213, 1429-1440.	8.5	100
2	Impaired complex I repair causes recessive Leber's hereditary optic neuropathy. <i>Journal of Clinical Investigation</i> , 2021, 131, .	8.2	89
3	Disruption of Thrombocyte and T Lymphocyte Development by a Mutation in <i>ARPC1B</i> . <i>Journal of Immunology</i> , 2017, 199, 4036-4045.	0.8	72
4	Somatic NRAS mutation in patient with generalized lymphatic anomaly. <i>Angiogenesis</i> , 2018, 21, 287-298.	7.2	57
5	Deleterious variants in TRAK1 disrupt mitochondrial movement and cause fatal encephalopathy. <i>Brain</i> , 2017, 140, 568-581.	7.6	53
6	Mutations in PPCS, Encoding Phosphopantothenoylecysteine Synthetase, Cause Autosomal-Recessive Dilated Cardiomyopathy. <i>American Journal of Human Genetics</i> , 2018, 102, 1018-1030.	6.2	42
7	Novel MALT1 Mutation Linked to Immunodeficiency, Immune Dysregulation, and an Abnormal T Cell Receptor Repertoire. <i>Journal of Clinical Immunology</i> , 2019, 39, 401-413.	3.8	42
8	De Novo Mutations Affecting the Catalytic C β Subunit of PP2A, PPP2CA, Cause Syndromic Intellectual Disability Resembling Other PP2A-Related Neurodevelopmental Disorders. <i>American Journal of Human Genetics</i> , 2019, 104, 139-156.	6.2	39
9	Congenital protein losing enteropathy: an inborn error of lipid metabolism due to DGAT1 mutations. <i>European Journal of Human Genetics</i> , 2016, 24, 1268-1273.	2.8	37
10	SCUBE3 loss-of-function causes a recognizable recessive developmental disorder due to defective bone morphogenetic protein signaling. <i>American Journal of Human Genetics</i> , 2021, 108, 115-133.	6.2	37
11	Whole exome sequencing in childhood-onset lupus frequently detects single gene etiologies. <i>Pediatric Rheumatology</i> , 2019, 17, 52.	2.1	34
12	Evaluation of Diagnostic Yield in Fetal Whole-Exome Sequencing: A Report on 45 Consecutive Families. <i>Frontiers in Genetics</i> , 2019, 10, 425.	2.3	33
13	Expanding the molecular diversity and phenotypic spectrum of glycerol 3-phosphate dehydrogenase 1 deficiency. <i>Journal of Inherited Metabolic Disease</i> , 2016, 39, 689-695.	3.6	24
14	Whole exome sequencing (WES) approach for diagnosing primary immunodeficiencies (PIDs) in a highly consanguineous community. <i>Clinical Immunology</i> , 2020, 214, 108376.	3.2	22
15	<i>BRPF1</i> associated intellectual disability, ptosis, and facial dysmorphism in a multiplex family. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e665.	1.2	21
16	Inherited SLP76 deficiency in humans causes severe combined immunodeficiency, neutrophil and platelet defects. <i>Journal of Experimental Medicine</i> , 2021, 218, .	8.5	20
17	G23D: Online tool for mapping and visualization of genomic variants on 3D protein structures. <i>BMC Genomics</i> , 2016, 17, 681.	2.8	18
18	Bi-allelic Variants in RALGAP1 Cause Profound Neurodevelopmental Disability, Muscular Hypotonia, Infantile Spasms, and Feeding Abnormalities. <i>American Journal of Human Genetics</i> , 2020, 106, 246-255.	6.2	17

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19	Reduced Function and Diversity of T Cell Repertoire and Distinct Clinical Course in Patients With IL7RA Mutation. <i>Frontiers in Immunology</i> , 2019, 10, 1672.	4.8	16
20	Novel homozygous <i>ENPP1</i> mutation causes generalized arterial calcifications of infancy, thrombocytopenia, and cardiovascular and central nervous system syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 2112-2118.	1.2	16
21	Germline variants in tumor suppressor FBXW7 lead to impaired ubiquitination and a neurodevelopmental syndrome. <i>American Journal of Human Genetics</i> , 2022, 109, 601-617.	6.2	16
22	Clinical significance of E148Q heterozygous variant in paediatric familial Mediterranean fever. <i>Rheumatology</i> , 2021, 60, 5447-5451.	1.9	15
23	<i>LMOD3</i> Associated Nemaline Myopathy: Prenatal Ultrasonographic, Pathologic, and Molecular Findings. <i>Journal of Ultrasound in Medicine</i> , 2018, 37, 1827-1833.	1.7	14
24	Whole-exome sequencing reveals a monogenic cause in 56% of individuals with laterality disorders and associated congenital heart defects. <i>Journal of Medical Genetics</i> , 2022, 59, 691-696.	3.2	14
25	A single center experience with publicly funded clinical exome sequencing for neurodevelopmental disorders or multiple congenital anomalies. <i>Scientific Reports</i> , 2021, 11, 19099.	3.3	13
26	A Novel Mutation in a Critical Region for the Methyl Donor Binding in DNMT3B Causes Immunodeficiency, Centromeric Instability, and Facial Anomalies Syndrome (ICF). <i>Journal of Clinical Immunology</i> , 2016, 36, 801-809.	3.8	12
27	Congenital neutropenia with variable clinical presentation in novel mutation of the SRP54 gene. <i>Pediatric Blood and Cancer</i> , 2020, 67, e28237.	1.5	12
28	A multidisciplinary nephrogenetic referral clinic for children and adultsâ€™ diagnostic achievements and insights. <i>Pediatric Nephrology</i> , 2022, 37, 1623-1646.	1.7	12
29	Genetic and Structural Analysis of a SKIV2L Mutation Causing Tricho-hepato-enteric Syndrome. <i>Digestive Diseases and Sciences</i> , 2018, 63, 1192-1199.	2.3	11
30	Progressive Pseudorheumatoid Dysplasia resolved by whole exome sequencing: a novel mutation in WISP3 and review of the literature. <i>BMC Medical Genetics</i> , 2019, 20, 53.	2.1	11
31	Four patients with D-bifunctional protein (DBP) deficiency: Expanding the phenotypic spectrum of a highly variable disease. <i>Molecular Genetics and Metabolism Reports</i> , 2020, 25, 100631.	1.1	11
32	Sustained Response to Imatinib in a Pediatric Patient with Concurrent Myeloproliferative Disease and Lymphoblastic Lymphoma Associated with a <i>CCDC88C-PDGFRB</i> Fusion Gene. <i>Acta Haematologica</i> , 2019, 141, 119-127.	1.4	10
33	Netrinâ€™2 dysfunction causes a Rettâ€™like phenotype with areflexia. <i>Human Mutation</i> , 2020, 41, 476-486.	2.5	10
34	Identification of a homozygous VRK1 mutation in two patients with adultâ€™onset distal hereditary motor neuropathy. <i>Muscle and Nerve</i> , 2020, 61, 395-400.	2.2	10
35	Broadening the phenotype of LRRK1 mutations - Features of malignant osteopetrosis and optic nerve atrophy with intrafamilial variable expressivity. <i>European Journal of Medical Genetics</i> , 2022, 65, 104383.	1.3	10
36	Diaphanospondylodysostosis: Refining the prenatal diagnosis of a rare skeletal disorder. <i>European Journal of Medical Genetics</i> , 2019, 62, 167-171.	1.3	9

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37	Chronic demodicosis in patients with immune dysregulation: An unexpected infectious manifestation of Signal transducer and activator of transcription (STAT)1 gain-of-function. <i>Clinical and Experimental Immunology</i> , 2021, 206, 56-67.	2.6	8
38	Molecular Mechanisms of Skewed X-Chromosome Inactivation in Female Hemophilia Patients—Lessons from Wide Genome Analyses. <i>International Journal of Molecular Sciences</i> , 2021, 22, 9074.	4.1	8
39	Refining the Phenotypic Spectrum of KMT5B-Associated Developmental Delay. <i>Frontiers in Pediatrics</i> , 2022, 10, 844845.	1.9	8
40	Autosomal dominant non-syndromic hearing loss maps to DFNA33 (13q34) and co-segregates with splice and frameshift variants in ATP11A, a phospholipid flippase gene. <i>Human Genetics</i> , 2022, 141, 431-444.	3.8	7
41	Monogenic Inflammatory Bowel Disease: It's Never Too Late to Make a Diagnosis. <i>Frontiers in Immunology</i> , 2020, 11, 1775.	4.8	6
42	TRMT10A Mutation in a Child with Diabetes, Short Stature, Microcephaly and Hypoplastic Kidneys. <i>JCRPE Journal of Clinical Research in Pediatric Endocrinology</i> , 2022, 14, 227-232.	0.9	6
43	Biallelic variants in ETV2 in a family with congenital heart defects, vertebral abnormalities and preaxial polydactyly. <i>European Journal of Medical Genetics</i> , 2021, 64, 104124.	1.3	6
44	A founder truncating variant in GDF1 causes autosomal-recessive right isomerism and associated congenital heart defects in multiplex Arab kindreds. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 987-993.	1.2	5
45	Abdominal muscle weakness as a presenting symptom in GNE myopathy. <i>Journal of Clinical Neuroscience</i> , 2019, 59, 316-317.	1.5	4
46	Exploring genetic defects in adults who were clinically diagnosed as severe combined immune deficiency during infancy. <i>Immunologic Research</i> , 2021, 69, 145-152.	2.9	3
47	Early and late manifestations of neuropathy due to <i>HSPB1</i> mutation in the Jewish Iranian population. <i>Annals of Clinical and Translational Neurology</i> , 2021, 8, 1260-1268.	3.7	3
48	Deep intronic variant in the ARSB gene as the genetic cause for Maroteaux-Lamy syndrome (MPS VI). <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3804-3809.	1.2	3
49	<i>c.259A>C</i> in the fibrinogen gene of alpha chain (<i>FGA</i>) is a fibrinogen with thrombotic phenotype. <i>The Application of Clinical Genetics</i> , 2019, Volume 12, 27-33.	3.0	2
50	Atypical immune phenotype in severe combined immunodeficiency patients with novel mutations in IL2RG and JAK3. <i>Genes and Immunity</i> , 2020, 21, 326-334.	4.1	2
51	Genetic workup as a complementary tool for the diagnosis of primary complement component deficiencies: a multicenter experience. <i>European Journal of Pediatrics</i> , 2022, 181, 1997-2004.	2.7	1
52	Glycogen Storage Disease type IA refractory to cornstarch: Can next generation sequencing offer a solution?. <i>European Journal of Medical Genetics</i> , 2022, , 104518.	1.3	1
53	FC035: Exome Sequencing of the Israeli Dialysis-Treated Pediatric Population Reveals Monogenic Etiology in 44% of Cases. <i>Nephrology Dialysis Transplantation</i> , 2022, 37, .	0.7	0
54	MO046: Exome sequencing of Israeli Druze individuals on dialysis reveals common as well as population-specific monogenic etiologies in 30%. <i>Nephrology Dialysis Transplantation</i> , 2022, 37, .	0.7	0