Ortal Barel

List of Publications by Year in descending order

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Version: 2024-02-01

54 papers	1,052 citations	516710 16 h-index	501196 28 g-index
55 all docs	55 docs citations	55 times ranked	2071 citing authors

#	Article	IF	Citations
1	TRMT10A Mutation in a Child with Diabetes, Short Stature, Microcephaly and Hypoplastic Kidneys. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2022, 14, 227-232.	0.9	6
2	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.	3.2	14
3	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.	1.3	10
4	A multidisciplinary nephrogenetic referral clinic for children and adultsâ€"diagnostic achievements and insights. Pediatric Nephrology, 2022, 37, 1623-1646.	1.7	12
5	Genetic workup as a complementary tool for the diagnosis of primary complement component deficiencies: a multicenter experience. European Journal of Pediatrics, 2022, 181, 1997-2004.	2.7	1
6	Refining the Phenotypic Spectrum of KMT5B-Associated Developmental Delay. Frontiers in Pediatrics, 2022, 10, 844845.	1.9	8
7	Autosomal dominant non-syndromic hearing loss maps to DFNA33 (13q34) and co-segregates with splice and frameshift variants in ATP11A, a phospholipid flippase gene. Human Genetics, 2022, 141, 431-444.	3.8	7
8	Germline variants in tumor suppressor FBXW7 lead to impaired ubiquitination and a neurodevelopmental syndrome. American Journal of Human Genetics, 2022, 109, 601-617.	6.2	16
9	FC035: Exome Sequencing of the Israeli Dialysis-Treated Pediatric Population Reveals Monogenic Etiology in â^¼44% of Cases. Nephrology Dialysis Transplantation, 2022, 37, .	0.7	0
10	MO046: Exome sequencing of Israeli Druze individuals on dialysis reveals common as well as population- specific monogenic etiologies in â^¼30%. Nephrology Dialysis Transplantation, 2022, 37, .	0.7	0
11	Glycogen Storage Disease type IA refractory to cornstarch: Can next generation sequencing offer a solution?. European Journal of Medical Genetics, 2022, , 104518.	1.3	1
12	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.	6.2	37
13	Exploring genetic defects in adults who were clinically diagnosed as severe combined immune deficiency during infancy. Immunologic Research, 2021, 69, 145-152.	2.9	3
14	Biallelic variants in ETV2 in a family with congenital heart defects, vertebral abnormalities and preaxial polydactyly. European Journal of Medical Genetics, 2021, 64, 104124.	1.3	6
15	Clinical significance of E148Q heterozygous variant in paediatric familial Mediterranean fever. Rheumatology, 2021, 60, 5447-5451.	1.9	15
16	Impaired complex I repair causes recessive Leberâ€™s hereditary optic neuropathy. Journal of Clinical Investigation, 2021, 131, .	8.2	89
17	Early and late manifestations of neuropathy due to HSPB1 mutation in the Jewish Iranian population. Annals of Clinical and Translational Neurology, 2021, 8, 1260-1268.	3.7	3
18	Chronic demodicosis in patients with immune dysregulation: An unexpected infectious manifestation of Signal transducer and activator of transcription (STAT)1 gainâ€ofâ€function. Clinical and Experimental Immunology, 2021, 206, 56-67.	2.6	8

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19	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.	1.2	3
20	Molecular Mechanisms of Skewed X-Chromosome Inactivation in Female Hemophilia Patientsâ€”Lessons from Wide Genome Analyses. International Journal of Molecular Sciences, 2021, 22, 9074.	4.1	8
21	A single center experience with publicly funded clinical exome sequencing for neurodevelopmental disorders or multiple congenital anomalies. Scientific Reports, 2021, 11, 19099.	3.3	13
22	Inherited SLP76 deficiency in humans causes severe combined immunodeficiency, neutrophil and platelet defects. Journal of Experimental Medicine, 2021, 218, .	8.5	20
23	Netrinâ€G2 dysfunction causes a Rettâ€like phenotype with areflexia. Human Mutation, 2020, 41, 476-486.	2.5	10
24	Identification of a homozygous VRK1 mutation in two patients with adultâ€onset distal hereditary motor neuropathy. Muscle and Nerve, 2020, 61, 395-400.	2.2	10
25	Four patients with D-bifunctional protein (DBP) deficiency: Expanding the phenotypic spectrum of a highly variable disease. Molecular Genetics and Metabolism Reports, 2020, 25, 100631.	1.1	11
26	Atypical immune phenotype in severe combined immunodeficiency patients with novel mutations in IL2RG and JAK3. Genes and Immunity, 2020, 21, 326-334.	4.1	2
27	Monogenic Inflammatory Bowel Disease: It's Never Too Late to Make a Diagnosis. Frontiers in Immunology, 2020, 11, 1775.	4.8	6
28	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.	1.2	5
29	Whole exome sequencing (WES) approach for diagnosing primary immunodeficiencies (PIDs) in a highly consanguineous community. Clinical Immunology, 2020, 214, 108376.	3.2	22
30	Congenital neutropenia with variable clinical presentation in novel mutation of the SRP54 gene. Pediatric Blood and Cancer, 2020, 67, e28237.	1.5	12
31	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.	6.2	17
32	Diaphanospondylodysostosis: Refining the prenatal diagnosis of a rare skeletal disorder. European Journal of Medical Genetics, 2019, 62, 167-171.	1.3	9
33	Evaluation of Diagnostic Yield in Fetal Whole-Exome Sequencing: A Report on 45 Consecutive Families. Frontiers in Genetics, 2019, 10, 425.	2.3	33
34	Reduced Function and Diversity of T Cell Repertoire and Distinct Clinical Course in Patients With IL7RA Mutation. Frontiers in Immunology, 2019, 10, 1672.	4.8	16
35	Whole exome sequencing in childhood-onset lupus frequently detects single gene etiologies. Pediatric Rheumatology, 2019, 17, 52.	2.1	34
36	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.	1.2	16

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37	Novel MALT1 Mutation Linked to Immunodeficiency, Immune Dysregulation, and an Abnormal T Cell Receptor Repertoire. Journal of Clinical Immunology, 2019, 39, 401-413.	3.8	42
38	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
39	BRPF1â€associated intellectual disability, ptosis, and facial dysmorphism in a multiplex family. Molecular Genetics & Genomic Medicine, 2019, 7, e665.	1.2	21
40	c.259A>C in the fibrinogen gene of alpha chain (FGA) is a fibrinogen with thrombotic phenotype . The Application of Clinical Genetics, 2019, Volume 12, 27-33.	3.0	2
41	Sustained Response to Imatinib in a Pediatric Patient with Concurrent Myeloproliferative Disease and Lymphoblastic Lymphoma Associated with a <i>CCDC88C-PDGFRB</i> Fusion Gene. Acta Haematologica, 2019, 141, 119-127.	1.4	10
42	De Novo Mutations Affecting the Catalytic CÎ± Subunit of PP2A, PPP2CA, Cause Syndromic Intellectual Disability Resembling Other PP2A-Related Neurodevelopmental Disorders. American Journal of Human Genetics, 2019, 104, 139-156.	6.2	39
43	Abdominal muscle weakness as a presenting symptom in GNE myopathy. Journal of Clinical Neuroscience, 2019, 59, 316-317.	1.5	4
44	Genetic and Structural Analysis of a SKIV2L Mutation Causing Tricho-hepato-enteric Syndrome. Digestive Diseases and Sciences, 2018, 63, 1192-1199.	2.3	11
45	Somatic NRAS mutation in patient with generalized lymphatic anomaly. Angiogenesis, 2018, 21, 287-298.	7.2	57
46	LMOD3â€Associated Nemaline Myopathy: Prenatal Ultrasonographic, Pathologic, and Molecular Findings. Journal of Ultrasound in Medicine, 2018, 37, 1827-1833.	1.7	14
47	Mutations in PPCS, Encoding Phosphopantothenoylcysteine Synthetase, Cause Autosomal-Recessive Dilated Cardiomyopathy. American Journal of Human Genetics, 2018, 102, 1018-1030.	6.2	42
48	Deleterious variants in TRAK1 disrupt mitochondrial movement and cause fatal encephalopathy. Brain, 2017, 140, 568-581.	7.6	53
49	Disruption of Thrombocyte and T Lymphocyte Development by a Mutation in ARPC1B. Journal of Immunology, 2017, 199, 4036-4045.	0.8	72
50	Expanding the molecular diversity and phenotypic spectrum of glycerol 3â€phosphate dehydrogenase 1 deficiency. Journal of Inherited Metabolic Disease, 2016, 39, 689-695.	3.6	24
51	G23D: Online tool for mapping and visualization of genomic variants on 3D protein structures. BMC Genomics, 2016, 17, 681.	2.8	18
52	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.	3.8	12
53	Mutations in STN1 cause Coats plus syndrome and are associated with genomic and telomere defects. Journal of Experimental Medicine, 2016, 213, 1429-1440.	8.5	100
54	Congenital protein losing enteropathy: an inborn error of lipid metabolism due to DGAT1 mutations. European Journal of Human Genetics, 2016, 24, 1268-1273.	2.8	37