Ohad Birk

List of Publications by Year
in descending order

48 papers	1,072 citations	516710 16 h-index	454955 30 g-index
51 all docs	51 docs citations	51 times ranked	2631 citing authors

#	Article	IF	Citations
1	TSHB R75G is a founder variant and prevalent cause of low or undetectable TSH in Indian Jews. European Thyroid Journal, 2022, 11, .	2.4	1
2	The Phenotypic and Mutational Spectrum of the FHONDA Syndrome and Oculocutaneous Albinism: Similarities and Differences. , 2022, 63, 19.		12
3	Looking for the skeleton in the closetâ€"rare genetic diagnoses in patients with diabetes and skeletal manifestations. Acta Diabetologica, 2022, 59, 711.	2.5	2
4	A syndrome of severe intellectual disability, hypotonia, failure to thrive, dysmorphism, and thinning of corpus callosum maps to chromosome 7q21.13â€q21.3. Clinical Genetics, 2022, 102, 123-129.	2.0	2
5	A novel leaky splice variant in centromere protein J (CENPJ)â€associated Seckel syndrome. Annals of Human Genetics, 2022, , .	0.8	0
6	Absence of SCAPER causes male infertility in humans andDrosophilaby modulating microtubule dynamics during meiosis. Journal of Medical Genetics, 2021, 58, 254-263.	3.2	7
7	CDH2 mutation affecting N-cadherin function causes attention-deficit hyperactivity disorder in humans and mice. Nature Communications, 2021, 12, 6187.	12.8	13
8	A nationwide genetic analysis of inherited retinal diseases in Israel as assessed by the Israeli inherited retinal disease consortium (IIRDC). Human Mutation, 2020, 41, 140-149.	2.5	75
9	Pituitary stalk interruption syndrome broadens the clinical spectrum of the <scp>TTC26</scp> ciliopathy. Clinical Genetics, 2020, 98, 303-307.	2.0	8
10	Novel MTMR2 mutation causing severe Charcot-Marie-Tooth type 4B1 disease: a case report. Neurogenetics, 2020, 21, 301-304.	1.4	1
11	B4GALT1â€congenital disorders of glycosylation: Expansion of the phenotypic and molecular spectrum and review of the literature. Clinical Genetics, 2020, 97, 920-926.	2.0	9
12	A homozygous missense variant of SUMF1 in the Bedouin population extends the clinical spectrum in ultrarare neonatal multiple sulfatase deficiency. Molecular Genetics & Genomic Medicine, 2020, 8, e1167.	1.2	4
13	Phenotypic variability and mutation hotspot in COX15 â€related Leigh syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1506-1512.	1.2	6
14	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
15	Mutations in the microtubule-associated protein MAP11 (C7orf43) cause microcephaly in humans and zebrafish. Brain, 2019, 142, 574-585.	7.6	32
16	DEGS1 variant causes neurological disorder. European Journal of Human Genetics, 2019, 27, 1668-1676.	2.8	28
17	TMEM70 deficiency: Novel mutation and hypercitrullinemia during metabolic decompensation. American Journal of Medical Genetics, Part A, 2019, 179, 1293-1298.	1.2	5
18	A novel SLC12A1 mutation in Bedouin kindred with antenatal Bartter syndrome type I. Annals of Human Genetics, 2019, 83, 361-366.	0.8	5

#	Article	IF	Citations
19	SCAPER localizes to primary cilia and its mutation affects cilia length, causing Bardet-Biedl syndrome. European Journal of Human Genetics, 2019, 27, 928-940.	2.8	36
20	SEC31A mutation affects ER homeostasis, causing a neurological syndrome. Journal of Medical Genetics, 2019, 56, 139-148.	3.2	31
21	Hyperuricemia and gout caused by missense mutation in d-lactate dehydrogenase. Journal of Clinical Investigation, 2019, 129, 5163-5168.	8.2	23
22	RSRC1 mutation affects intellect and behaviour through aberrant splicing and transcription, downregulating IGFBP3. Brain, 2018, 141, 961-970.	7.6	20
23	Carrier frequency analysis of mutations causing autosomal-recessive-inherited retinal diseases in the Israeli population. European Journal of Human Genetics, 2018, 26, 1159-1166.	2.8	14
24	A novel homozygous SLC25A1 mutation with impaired mitochondrial complex V: Possible phenotypic expansion. American Journal of Medical Genetics, Part A, 2018, 176, 330-336.	1.2	14
25	Nocturnal Atrial Fibrillation Caused by Mutation in KCND2 , Encoding Pore-Forming (Î±) Subunit of the Cardiac Kv4.2 Potassium Channel. Circulation Genomic and Precision Medicine, 2018, 11, e002293.	3.6	22
26	Combined CNV, haplotyping and whole exome sequencing enable identification of two distinct novel EYS mutations causing RP in a single inbred tribe. American Journal of Medical Genetics, Part A, 2018, 176, 2695-2703.	1.2	3
27	Heterozygous versus homozygous phenotype caused by the same MC4R mutation: novel mutation affecting a large consanguineous kindred. BMC Medical Genetics, 2018, 19, 135.	2.1	18
28	Novel SBF1 spliceâ€site null mutation broadens the clinical spectrum of Charcotâ€Marieâ€Tooth type 4B3 disease. Clinical Genetics, 2018, 94, 473-479.	2.0	9
29	Progressive hereditary spastic paraplegia caused by a homozygous KY mutation. European Journal of Human Genetics, 2017, 25, 966-972.	2.8	18
30	SLC30A9 mutation affecting intracellular zinc homeostasis causes a novel cerebro-renal syndrome. Brain, 2017, 140, 928-939.	7.6	72
31	Mutations in ARMC9, which Encodes a Basal Body Protein, Cause Joubert Syndrome in Humans and Ciliopathy Phenotypes in Zebrafish. American Journal of Human Genetics, 2017, 101, 23-36.	6.2	74
32	PAX7mutation in a syndrome of failure to thrive, hypotonia, and global neurodevelopmental delay. Human Mutation, 2017, 38, 1671-1683.	2.5	12
33	A Rare Variant in PGAP2 Causes Autosomal Recessive Hyperphosphatasia with Mental Retardation Syndrome, with a Mild Phenotype in Heterozygous Carriers. BioMed Research International, 2017, 2017, 1-7.	1.9	9
34	Novel GUCY2D mutation causes phenotypic variability of Leber congenital amaurosis inÂa large kindred. BMC Medical Genetics, 2016, 17, 52.	2.1	18
35	Two novel MYH7 proline substitutions cause Laing Distal Myopathy-like phenotypes with variable expressivity and neck extensor contracture. BMC Medical Genetics, 2016, 17, 57.	2.1	12
36	UNC80mutation causes a syndrome of hypotonia, severe intellectual disability, dyskinesia and dysmorphism, similar to that caused by mutations in its interacting cation channelNALCN. Journal of Medical Genetics, 2016, 53, 397-402.	3.2	40

#	Article	IF	Citations
37	ALFY-Controlled DVL3 Autophagy Regulates Wnt Signaling, Determining Human Brain Size. PLoS Genetics, 2016, 12, e1005919.	3.5	67
38	Novel FAM20A mutation causes autosomal recessive amelogenesis imperfecta. Archives of Oral Biology, 2015, 60, 919-922.	1.8	8
39	CDC174, a novel component of the exon junction complex whose mutation underlies a syndrome of hypotonia and psychomotor developmental delay. Human Molecular Genetics, 2015, 24, 6485-6491.	2.9	13
40	A syndrome of congenital microcephaly, intellectual disability and dysmorphism with a homozygous mutation in FRMD4A. European Journal of Human Genetics, 2015, 23, 1729-1734.	2.8	14
41	A novel GLI3 mutation affecting the zinc finger domain leads to preaxial-postaxial polydactyly-syndactyly complex. BMC Medical Genetics, 2014, 15, 110.	2.1	15
42	Deciphering the fine-structure of tribal admixture in the Bedouin population using genomic data. Heredity, 2014, 112, 182-189.	2.6	21
43	Isolated foveal hypoplasia with secondary nystagmus and low vision is associated with a homozygous SLC38A8 mutation. European Journal of Human Genetics, 2014, 22, 703-706.	2.8	49
44	Autosomal recessive Adamsâ€“Oliver syndrome caused by homozygous mutation in EOGT, encoding an EGF domain-specific O-GlcNAc transferase. European Journal of Human Genetics, 2014, 22, 374-378.	2.8	55
45	Selenocysteinopathies: progressive cerebelloâ€“cerebral atrophy and other diseases of the 21st amino acid, selenocysteine. Future Neurology, 2011, 6, 135-138.	0.5	3
46	High Myopia Caused by a Mutation in LEPREL1, Encoding Prolyl 3-Hydroxylase 2. American Journal of Human Genetics, 2011, 89, 438-445.	6.2	95
47	Polymorphic alleles of the human MEI1 gene are associated with human azoospermia by meiotic arrest. Journal of Human Genetics, 2006, 51, 533-540.	2.3	57
48	Transcript-Based Diagnosis and Expanded Phenotype of an Intronic Mutation in TPM3 Myopathy. Molecular Diagnosis and Therapy, 0, , .	3.8	2