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	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
2	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
3	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
4	SLC30A9 mutation affecting intracellular zinc homeostasis causes a novel cerebro-renal syndrome. Brain, 2017, 140, 928-939.	7.6	72
5	ALFY-Controlled DVL3 Autophagy Regulates Wnt Signaling, Determining Human Brain Size. PLoS Genetics, 2016, 12, e1005919.	3.5	67
6	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
7	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
8	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
9	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
10	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
11	Mutations in the microtubule-associated protein MAP11 (C7orf43) cause microcephaly in humans and zebrafish. Brain, 2019, 142, 574-585.	7.6	32
12	SEC31A mutation affects ER homeostasis, causing a neurological syndrome. Journal of Medical Genetics, 2019, 56, 139-148.	3.2	31
13	DEGS1 variant causes neurological disorder. European Journal of Human Genetics, 2019, 27, 1668-1676.	2.8	28
14	Hyperuricemia and gout caused by missense mutation in d-lactate dehydrogenase. Journal of Clinical Investigation, 2019, 129, 5163-5168.	8.2	23
15	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
16	Deciphering the fine-structure of tribal admixture in the Bedouin population using genomic data. Heredity, 2014, 112, 182-189.	2.6	21
17	RSRC1 mutation affects intellect and behaviour through aberrant splicing and transcription, downregulating IGFBP3. Brain, 2018, 141, 961-970.	7.6	20
18	Novel GUCY2D mutation causes phenotypic variability of Leber congenital amaurosis inÂa large kindred. BMC Medical Genetics, 2016, 17, 52.	2.1	18

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19	Progressive hereditary spastic paraplegia caused by a homozygous KY mutation. European Journal of Human Genetics, 2017, 25, 966-972.	2.8	18
20	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
21	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
22	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
23	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
24	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
25	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
26	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
27	CDH2 mutation affecting N-cadherin function causes attention-deficit hyperactivity disorder in humans and mice. Nature Communications, 2021, 12, 6187.	12.8	13
28	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
29	PAX7mutation in a syndrome of failure to thrive, hypotonia, and global neurodevelopmental delay. Human Mutation, 2017, 38, 1671-1683.	2.5	12
30	The Phenotypic and Mutational Spectrum of the FHONDA Syndrome and Oculocutaneous Albinism: Similarities and Differences. , 2022, 63, 19.		12
31	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
32	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
33	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
34	Novel FAM20A mutation causes autosomal recessive amelogenesis imperfecta. Archives of Oral Biology, 2015, 60, 919-922.	1.8	8
35	Pituitary stalk interruption syndrome broadens the clinical spectrum of the <scp>TTC26</scp> ciliopathy. Clinical Genetics, 2020, 98, 303-307.	2.0	8
36	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

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37	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
38	TMEM70 deficiency: Novel mutation and hypercitrullinemia during metabolic decompensation. American Journal of Medical Genetics, Part A, 2019, 179, 1293-1298.	1.2	5
39	A novel SLC12A1 mutation in Bedouin kindred with antenatal Bartter syndrome type I. Annals of Human Genetics, 2019, 83, 361-366.	0.8	5
40	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
41	Selenocysteinopathies: progressive cerebelloâ€“cerebral atrophy and other diseases of the 21st amino acid, selenocysteine. Future Neurology, 2011, 6, 135-138.	0.5	3
42	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
43	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
44	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
45	Transcript-Based Diagnosis and Expanded Phenotype of an Intronic Mutation in TPM3 Myopathy. Molecular Diagnosis and Therapy, 0, , .	3.8	2
46	Novel MTMR2 mutation causing severe Charcot-Marie-Tooth type 4B1 disease: a case report. Neurogenetics, 2020, 21, 301-304.	1.4	1
47	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
48	A novel leaky splice variant in centromere protein J (CENPJ)â€associated Seckel syndrome. Annals of Human Genetics, 2022, , .	0.8	0