

Ohad Birk

List of Publications by Year in descending order

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48
papers

1,072
citations

516215

16
h-index

454577

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.	2.6	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.	1.1	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.	2.6	74
4	SLC30A9 mutation affecting intracellular zinc homeostasis causes a novel cerebro-renal syndrome. Brain, 2017, 140, 928-939.	3.7	72
5	ALFY-Controlled DVL3 Autophagy Regulates Wnt Signaling, Determining Human Brain Size. PLoS Genetics, 2016, 12, e1005919.	1.5	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.	1.1	57
7	Autosomal recessive Adams-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.	1.4	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.	1.4	49
9	UNC80 mutation causes a syndrome of hypotonia, severe intellectual disability, dyskinesia and dysmorphism, similar to that caused by mutations in its interacting cation channel NALCN. Journal of Medical Genetics, 2016, 53, 397-402.	1.5	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.	1.4	36
11	Mutations in the microtubule-associated protein MAP11 (C7orf43) cause microcephaly in humans and zebrafish. Brain, 2019, 142, 574-585.	3.7	32
12	SEC31A mutation affects ER homeostasis, causing a neurological syndrome. Journal of Medical Genetics, 2019, 56, 139-148.	1.5	31
13	DEGS1 variant causes neurological disorder. European Journal of Human Genetics, 2019, 27, 1668-1676.	1.4	28
14	Hyperuricemia and gout caused by missense mutation in d-lactate dehydrogenase. Journal of Clinical Investigation, 2019, 129, 5163-5168.	3.9	23
15	Nocturnal Atrial Fibrillation Caused by Mutation in KCND2, Encoding Pore-Forming ($\hat{\pm}$) Subunit of the Cardiac Kv4.2 Potassium Channel. Circulation Genomic and Precision Medicine, 2018, 11, e002293.	1.6	22
16	Deciphering the fine-structure of tribal admixture in the Bedouin population using genomic data. Heredity, 2014, 112, 182-189.	1.2	21
17	RSRC1 mutation affects intellect and behaviour through aberrant splicing and transcription, downregulating IGFBP3. Brain, 2018, 141, 961-970.	3.7	20
18	Novel GUCY2D mutation causes phenotypic variability of Leber congenital amaurosis in a 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. <i>European Journal of Human Genetics</i> , 2017, 25, 966-972.	1.4	18
20	Heterozygous versus homozygous phenotype caused by the same MC4R mutation: novel mutation affecting a large consanguineous kindred. <i>BMC Medical Genetics</i> , 2018, 19, 135.	2.1	18
21	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.	2.2	16
22	A novel GLI3 mutation affecting the zinc finger domain leads to preaxial-postaxial polydactyly-syndactyly complex. <i>BMC Medical Genetics</i> , 2014, 15, 110.	2.1	15
23	A syndrome of congenital microcephaly, intellectual disability and dysmorphism with a homozygous mutation in FRMD4A. <i>European Journal of Human Genetics</i> , 2015, 23, 1729-1734.	1.4	14
24	Carrier frequency analysis of mutations causing autosomal-recessive-inherited retinal diseases in the Israeli population. <i>European Journal of Human Genetics</i> , 2018, 26, 1159-1166.	1.4	14
25	A novel homozygous <i>SLC25A1</i> mutation with impaired mitochondrial complex V: Possible phenotypic expansion. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 330-336.	0.7	14
26	<i>CDC174</i> , a novel component of the exon junction complex whose mutation underlies a syndrome of hypotonia and psychomotor developmental delay. <i>Human Molecular Genetics</i> , 2015, 24, 6485-6491.	1.4	13
27	CDH2 mutation affecting N-cadherin function causes attention-deficit hyperactivity disorder in humans and mice. <i>Nature Communications</i> , 2021, 12, 6187.	5.8	13
28	Two novel MYH7 proline substitutions cause Laing Distal Myopathy-like phenotypes with variable expressivity and neck extensor contracture. <i>BMC Medical Genetics</i> , 2016, 17, 57.	2.1	12
29	<i>PAX7</i> mutation in a syndrome of failure to thrive, hypotonia, and global neurodevelopmental delay. <i>Human Mutation</i> , 2017, 38, 1671-1683.	1.1	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 <i>PGAP2</i> Causes Autosomal Recessive Hyperphosphatasia with Mental Retardation Syndrome, with a Mild Phenotype in Heterozygous Carriers. <i>BioMed Research International</i> , 2017, 2017, 1-7.	0.9	9
32	Novel <i>SBF1</i> splice-site null mutation broadens the clinical spectrum of Charcot-Marie-Tooth type 4B3 disease. <i>Clinical Genetics</i> , 2018, 94, 473-479.	1.0	9
33	B4GALT1 congenital disorders of glycosylation: Expansion of the phenotypic and molecular spectrum and review of the literature. <i>Clinical Genetics</i> , 2020, 97, 920-926.	1.0	9
34	Novel FAM20A mutation causes autosomal recessive amelogenesis imperfecta. <i>Archives of Oral Biology</i> , 2015, 60, 919-922.	0.8	8
35	Pituitary stalk interruption syndrome broadens the clinical spectrum of the <i>TTC26</i> ciliopathy. <i>Clinical Genetics</i> , 2020, 98, 303-307.	1.0	8
36	Absence of SCAPER causes male infertility in humans and <i>Drosophila</i> by modulating microtubule dynamics during meiosis. <i>Journal of Medical Genetics</i> , 2021, 58, 254-263.	1.5	7

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37	Phenotypic variability and mutation hotspot in COX15 -related Leigh syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1506-1512.	0.7	6
38	TMEM70 deficiency: Novel mutation and hypercitrullinemia during metabolic decompensation. American Journal of Medical Genetics, Part A, 2019, 179, 1293-1298.	0.7	5
39	A novel <i>SLC12A1</i> mutation in Bedouin kindred with antenatal Bartter syndrome type I. Annals of Human Genetics, 2019, 83, 361-366.	0.3	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.	0.6	4
41	Selenocysteinopathies: progressive cerebello-cerebral atrophy and other diseases of the 21st amino acid, selenocysteine. Future Neurology, 2011, 6, 135-138.	0.9	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.	0.7	3
43	Looking for the skeleton in the closet - rare genetic diagnoses in patients with diabetes and skeletal manifestations. Acta Diabetologica, 2022, 59, 711.	1.2	2
44	A syndrome of severe intellectual disability, hypotonia, failure to thrive, dysmorphism, and thinning of corpus callosum maps to chromosome 7q21.13-q21.3. Clinical Genetics, 2022, 102, 123-129.	1.0	2
45	Transcript-Based Diagnosis and Expanded Phenotype of an Intronic Mutation in TPM3 Myopathy. Molecular Diagnosis and Therapy, 0, , .	1.6	2
46	Novel MTMR2 mutation causing severe Charcot-Marie-Tooth type 4B1 disease: a case report. Neurogenetics, 2020, 21, 301-304.	0.7	1
47	TSHB R75G is a founder variant and prevalent cause of low or undetectable TSH in Indian Jews. European Thyroid Journal, 2022, 11, .	1.2	1
48	A novel leaky splice variant in centromere protein J (<i>CENPJ</i>) - associated Seckel syndrome. Annals of Human Genetics, 2022, , .	0.3	0