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List of Publications by Year in descending order

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Version: 2024-02-01

34
papers

1,339
citations

471509

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395702

33
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docs citations

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2638
citing authors

#	ARTICLE	IF	CITATIONS
1	Albinism. , 2022, , 393-402.		2
2	The Phenotypic and Mutational Spectrum of the FHONDA Syndrome and Oculocutaneous Albinism: Similarities and Differences. , 2022, 63, 19.		12
3	The Dct ^{+/+} Mouse Model to Unravel Retinogenesis Misregulation in Patients with Albinism. Genes, 2022, 13, 1164.	2.4	3
4	The contribution of common regulatory and protein-coding TYR variants to the genetic architecture of albinism. Nature Communications, 2022, 13, .	12.8	17
5	A new case with Hermansky-Pudlak syndrome type 9, a rare cause of syndromic albinism with severe defect of platelets dense bodies. Platelets, 2021, 32, 420-423.	2.3	5
6	Novel variants in the <i>BLOC1S3</i> gene in patients presenting a mild form of Hermansky-Pudlak syndrome. Pigment Cell and Melanoma Research, 2021, 34, 132-135.	3.3	5
7	Dopachrome tautomerase variants in patients with oculocutaneous albinism. Genetics in Medicine, 2021, 23, 479-487.	2.4	33
8	A recurrent missense variant in EYA3 gene is associated with oculo-auriculo-vertebral spectrum. Human Genetics, 2021, 140, 933-944.	3.8	14
9	Clinical variability and probable founder effect in oculocutaneous albinism type 7. Clinical Genetics, 2020, 97, 527-528.	2.0	1
10	Albinism: An Underdiagnosed Condition. Journal of Investigative Dermatology, 2020, 140, 1449-1451.	0.7	9
11	Hereditary Mucoepithelial Dysplasia Results from Heterozygous Variants at p.Arg557 Mutational Hotspot in SREBF1, Encoding a Transcription Factor Involved in Cholesterol Homeostasis. Journal of Investigative Dermatology, 2020, 140, 1289-1292.e2.	0.7	12
12	Functional and genetic analyses of <i>ZYG11B</i> provide evidences for its involvement in OAVS. Molecular Genetics & Genomic Medicine, 2020, 8, e1375.	1.2	21
13	BLOC1S5 pathogenic variants cause a new type of Hermansky-Pudlak syndrome. Genetics in Medicine, 2020, 22, 1613-1622.	2.4	44
14	Clinical and molecular findings of FRMD7 related congenital nystagmus as a differential diagnosis of ocular albinism. Ophthalmic Genetics, 2019, 40, 161-164.	1.2	2
15	Mild form of oculocutaneous albinism type 1: phenotypic analysis of compound heterozygous patients with the R402Q variant of the <i>TYR</i> gene. British Journal of Ophthalmology, 2019, 103, 1239-1247.	3.9	24
16	Deletion in 2q35 excluding the IHH gene leads to fetal severe limb anomalies and suggests a disruption of chromatin architecture. European Journal of Human Genetics, 2019, 27, 384-388.	2.8	3
17	Molecular characterization of a series of 990 index patients with albinism. Pigment Cell and Melanoma Research, 2018, 31, 466-474.	3.3	92
18	19p13 microduplications encompassing NFIX are responsible for intellectual disability, short stature and small head circumference. European Journal of Human Genetics, 2018, 26, 85-93.	2.8	7

#	ARTICLE	IF	CITATIONS
19	A novel de novo mutation in MYT1, the unique OAVS gene identified so far. European Journal of Human Genetics, 2017, 25, 1083-1086.	2.8	35
20	Clinico-molecular analysis of eleven patients with Hermansky-Pudlak type 5 syndrome, a mild form of HPS. Pigment Cell and Melanoma Research, 2017, 30, 563-570.	3.3	13
21	Complete loss of function of the ubiquitin ligase HERC2 causes a severe neurodevelopmental phenotype. European Journal of Human Genetics, 2017, 25, 52-58.	2.8	28
22	Mutations in MYT1, encoding the myelin transcription factor 1, are a rare cause of OAVS. Journal of Medical Genetics, 2016, 53, 752-760.	3.2	51
23	Identification of a homozygous mutation of SLC24A5 (OCA6) in two patients with oculocutaneous albinism from French Guiana. Pigment Cell and Melanoma Research, 2016, 29, 104-106.	3.3	19
24	Albinism in a patient with mutations at both the OAI and OCA3 loci. Pigment Cell and Melanoma Research, 2016, 29, 107-109.	3.3	3
25	SLC24A5 Mutations Are Associated with Non-Syndromic Oculocutaneous Albinism. Journal of Investigative Dermatology, 2014, 134, 568-571.	0.7	36
26	Increasing the complexity: new genes and new types of albinism. Pigment Cell and Melanoma Research, 2014, 27, 11-18.	3.3	179
27	High-resolution array-CGH in patients with oculocutaneous albinism identifies new deletions of the TYR, OCA2, and SLC45A2 genes and a complex rearrangement of the OCA2 gene. Pigment Cell and Melanoma Research, 2014, 27, 59-71.	3.3	23
28	New candidate loci identified by array-CGH in a cohort of 100 children presenting with syndromic obesity. American Journal of Medical Genetics, Part A, 2014, 164, 1965-1975.	1.2	49
29	Expanding the clinical phenotype at the 3q13.31 locus with a new case of microdeletion and first characterization of the reciprocal duplication. Molecular Genetics and Metabolism, 2013, 110, 90-97.	1.1	13
30	Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. Nature, 2011, 478, 97-102.	27.8	394
31	High resolution mapping of OCA2 intragenic rearrangements and identification of a founder effect associated with a deletion in Polish albino patients. Human Genetics, 2011, 129, 199-208.	3.8	14
32	Array-CGH analysis of a cohort of 86 patients with oculoauriculovertebral spectrum. American Journal of Medical Genetics, Part A, 2010, 152A, 1984-1989.	1.2	53
33	Spectrum of CREBBP gene dosage anomalies in Rubinstein-Taybi Syndrome patients. European Journal of Human Genetics, 2007, 15, 843-847.	2.8	41
34	Oculocutaneous albinism with TYRP1 gene mutations in a Caucasian patient. Pigment Cell & Melanoma Research, 2006, 19, 239-242.	3.6	62