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List of Publications by Year
in descending order

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34 papers	1,339 citations	471509 17 h-index	395702 33 g-index
37 all docs	37 docs citations	37 times ranked	2638 citing authors

#	Article	IF	Citations
1	Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. Nature, 2011, 478, 97-102.	27.8	394
2	Increasing the complexity: new genes and new types of albinism. Pigment Cell and Melanoma Research, 2014, 27, 11-18.	3.3	179
3	Molecular characterization of a series of 990 index patients with albinism. Pigment Cell and Melanoma Research, 2018, 31, 466-474.	3.3	92
4	Oculocutaneous albinism with TYRP1 gene mutations in a Caucasian patient. Pigment Cell & Melanoma Research, 2006, 19, 239-242.	3.6	62
5	Arrayâ€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
6	Mutations inMYT1, encoding the myelin transcription factor 1, are a rare cause of OAVS. Journal of Medical Genetics, 2016, 53, 752-760.	3.2	51
7	New candidate loci identified by arrayâ€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
8	BLOC1S5 pathogenic variants cause a new type of Hermanskyâ€“Pudlak syndrome. Genetics in Medicine, 2020, 22, 1613-1622.	2.4	44
9	Spectrum of CREBBP gene dosage anomalies in Rubinsteinâ€“Taybi Syndrome patients. European Journal of Human Genetics, 2007, 15, 843-847.	2.8	41
10	SLC24A5 Mutations Are Associated with Non-Syndromic Oculocutaneous Albinism. Journal of Investigative Dermatology, 2014, 134, 568-571.	0.7	36
11	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
12	Dopachrome tautomerase variants in patients with oculocutaneous albinism. Genetics in Medicine, 2021, 23, 479-487.	2.4	33
13	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
14	Mild form of oculocutaneous albinism type 1: phenotypic analysis of compound heterozygous patients with the R402Q variant of the TYR gene. British Journal of Ophthalmology, 2019, 103, 1239-1247.	3.9	24
15	Highâ€resolution arrayâ€<scp>CGH</scp> in patients with oculocutaneous albinism identifies new deletions of the <scp>TYR</scp>,<scp> OCA</scp>2, and <scp>SLC</scp>45A2 genes and a complex rearrangement of the <scp>OCA</scp>2 gene. Pigment Cell and Melanoma Research, 2014, 27, 59-71.	3.3	23
16	Functional and genetic analyses of ZYG11B provide evidences for its involvement in OAVS. Molecular Genetics & Genomic Medicine, 2020, 8, e1375.	1.2	21
17	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
18	The contribution of common regulatory and protein-coding TYR variants to the genetic architecture of albinism. Nature Communications, 2022, 13, .	12.8	17

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19	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
20	A recurrent missense variant in EYA3 gene is associated with oculo-auriculo-vertebral spectrum. Human Genetics, 2021, 140, 933-944.	3.8	14
21	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
22	Clinicoâ€molecular analysis of eleven patients with Hermanskyâ€“Pudlak type 5 syndrome, a mild form of <scp>HPS</scp>. Pigment Cell and Melanoma Research, 2017, 30, 563-570.	3.3	13
23	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
24	The Phenotypic and Mutational Spectrum of the FHONDA Syndrome and Oculocutaneous Albinism: Similarities and Differences. , 2022, 63, 19.		12
25	Albinism: An Underdiagnosed Condition. Journal of Investigative Dermatology, 2020, 140, 1449-1451.	0.7	9
26	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
27	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
28	Novel variants in the BLOC1S3 gene in patients presenting a mild form of Hermanskyâ€“Pudlak syndrome. Pigment Cell and Melanoma Research, 2021, 34, 132-135.	3.3	5
29	Albinism in a patient with mutations at both the OA1 and OCA3 loci. Pigment Cell and Melanoma Research, 2016, 29, 107-109.	3.3	3
30	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
31	The Dctâ^'/â^' Mouse Model to Unravel Retinogenesis Misregulation in Patients with Albinism. Genes, 2022, 13, 1164.	2.4	3
32	Clinical and molecular findings of FRMD7 related congenital nystagmus as adifferential diagnosis of ocular albinism. Ophthalmic Genetics, 2019, 40, 161-164.	1.2	2
33	Albinism. , 2022, , 393-402.		2
34	Clinical variability and probable founder effect in oculocutaneous albinism type 7. Clinical Genetics, 2020, 97, 527-528.	2.0	1