

Jorge Frank

List of Publications by Year in descending order

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Version: 2024-02-01

56
papers

1,551
citations

471371

17
h-index

315616

38
g-index

70
all docs

70
docs citations

70
times ranked

1929
citing authors

#	ARTICLE	IF	CITATIONS
1	Exposing the human nude phenotype. <i>Nature</i> , 1999, 398, 473-474.	13.7	247
2	Afamelanotide for Erythropoietic Protoporphria. <i>New England Journal of Medicine</i> , 2015, 373, 48-59.	13.9	206
3	Hereditary leiomyomatosis and renal cell cancer in families referred for fumarate hydratase germline mutation analysis. <i>Clinical Genetics</i> , 2011, 79, 49-59.	1.0	154
4	Treatment Of Acute Edema Attacks In Hereditary Angioedema With A Bradykinin Receptor-2 Antagonist (Icatibant). <i>Journal of Allergy and Clinical Immunology</i> , 2007, 119, S278.	1.5	127
5	Mutations in the Gene Encoding the Wnt-Signaling Component R-Spondin 4 (RSPO4) Cause Autosomal Recessive Anonychia. <i>American Journal of Human Genetics</i> , 2006, 79, 1105-1109.	2.6	94
6	Phenotype Diversity in Familial Cyndromatosis: A Frameshift Mutation in the Tumor Suppressor Gene CYLD Underlies Different Tumors of Skin Appendages. <i>Journal of Investigative Dermatology</i> , 2002, 119, 527-531.	0.3	87
7	Mutations in Î³-secretase subunit encoding PSENEN underlie Dowling-Degos disease associated with acne inversa. <i>Journal of Clinical Investigation</i> , 2017, 127, 1485-1490.	3.9	73
8	Porphyria cutanea tarda " When skin meets liver. <i>Bailliere's Best Practice and Research in Clinical Gastroenterology</i> , 2010, 24, 735-745.	1.0	67
9	Bi-allelic Mutations in LSS, Encoding Lanosterol Synthase, Cause Autosomal-Recessive Hypotrichosis Simplex. <i>American Journal of Human Genetics</i> , 2018, 103, 777-785.	2.6	55
10	Clinical and molecular genetic aspects of hereditary multiple cutaneous leiomyomatosis. <i>European Journal of Dermatology</i> , 2009, 19, 545-551.	0.3	49
11	Pathogenicity of POFUT1 in Dowling-Degos Disease: Additional Mutations and Clinical Overlap with Reticulate Acropigmentation of Kitamura. <i>Journal of Investigative Dermatology</i> , 2015, 135, 615-618.	0.3	25
12	C73R is a hotspot mutation in the uroporphyrinogen III synthase gene in congenital erythropoietic porphyria. <i>Annals of Human Genetics</i> , 1998, 62, 225-230.	0.3	24
13	Hepatocellular Carcinoma in Variegated Porphyria: A Serious Complication. <i>Acta Dermato-Venereologica</i> , 2010, 90, 512-515.	0.6	24
14	Erythropoietic protoporphyria without skin symptoms-you do not always see what they feel. <i>European Journal of Pediatrics</i> , 2008, 167, 703-706.	1.3	23
15	Molecular pathways involved in hair follicle tumor formation: all about mammalian target of rapamycin?. <i>Experimental Dermatology</i> , 2009, 18, 185-191.	1.4	22
16	Biallelic inactivation of protoporphyrinogen oxidase and hydroxymethylbilane synthase is associated with liver cancer in acute porphyrias. <i>Journal of Hepatology</i> , 2015, 62, 734-738.	1.8	22
17	Linkage refinement of Bazex-Dupr�-Christol syndrome to an 11.4-Mb interval on chromosome Xq25-27.1. <i>British Journal of Dermatology</i> , 2011, 165, 201-203.	1.4	18
18	Delayed diagnosis and diminished quality of life in erythropoietic protoporphyria: results of a cross-sectional study in Sweden. <i>Journal of Internal Medicine</i> , 2011, 269, 270-274.	2.7	18

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19	Mutations in the translation initiation codon of the protoporphyrinogen oxidase gene underlie variegate porphyria. <i>Clinical and Experimental Dermatology</i> , 1999, 24, 296-301.	0.6	17
20	Characterization of the desmosomal cadherin gene family: Genomic organization of two desmoglein genes on human chromosome 18q12. <i>Experimental Dermatology</i> , 2001, 10, 90-94.	1.4	17
21	Altered Notch Signaling in Dowling-Degos Disease: Additional Mutations in POGUT1 and Further Insights into Disease Pathogenesis. <i>Journal of Investigative Dermatology</i> , 2019, 139, 960-964.	0.3	15
22	The genetic basis of "Scarsdale Gourmet Diet" variegate porphyria: a missense mutation in the protoporphyrinogen oxidase gene. <i>Archives of Dermatological Research</i> , 1998, 290, 441-445.	1.1	14
23	A homozygous mutation in the ferrochelatase gene underlies erythropoietic protoporphyria associated with palmar keratoderma. <i>British Journal of Dermatology</i> , 2009, 160, 1330-1334.	1.4	14
24	A novel therapeutic strategy for turban tumor: scalp excision and combined reconstruction with artificial dermis and split skin graft. <i>International Journal of Dermatology</i> , 2014, 53, 246-249.	0.5	14
25	Recurrent missense mutation in the protoporphyrinogen oxidase gene underlies variegate porphyria. , 1998, 79, 22-26.		13
26	Continuing medical and student education in dermatology during the coronavirus pandemic – a major challenge. <i>JDDG - Journal of the German Society of Dermatology</i> , 2020, 18, 835-840.	0.4	12
27	Molecular basis of variegate porphyria: a missense mutation in the protoporphyrinogen oxidase gene.. <i>Journal of Medical Genetics</i> , 1998, 35, 244-247.	1.5	11
28	The Wnt signalling ligand RSPO4, causing inherited onychia, is not mutated in a patient with congenital nail hypoplasia/aplasia with underlying skeletal defects. <i>British Journal of Dermatology</i> , 2007, 157, 801-802.	1.4	8
29	Phenotype variability in tumor disorders of the skin appendages associated with mutations in the CYLD gene. <i>Archives of Dermatological Research</i> , 2018, 310, 599-606.	1.1	7
30	Photosensitivity in the Elderly – Think of Late-Onset Protoporphyria. <i>Journal of Investigative Dermatology</i> , 2013, 133, 1467-1471.	0.3	6
31	Hereditäre Hauterkrankungen – Klinisch und genetisch heterogen. <i>JDDG - Journal of the German Society of Dermatology</i> , 2017, 15, 881-882.	0.4	5
32	Intra- and Interfamilial Phenotype Variability Associated with Mutations in Î³-Secretase Subunit-Encoding PSENEN. <i>Journal of Investigative Dermatology</i> , 2018, 138, 1215-1218.	0.3	4
33	Comorbid acne inversa and Dowling – Degos disease due to a single NCSTN mutation: is there enough evidence?. <i>British Journal of Dermatology</i> , 2021, 184, 374-374.	1.4	3
34	A woman with hyperpigmented macules and papules. <i>JDDG - Journal of the German Society of Dermatology</i> , 2021, 19, 1656-1660.	0.4	3
35	Erythropoietic protoporphyria: identification of novel mutations in the ferrochelatase gene and comparison of biochemical markers versus molecular analysis as diagnostic strategies. <i>Journal of Investigative Medicine</i> , 1999, 47, 278-84.	0.7	3
36	C73R is a hotspot mutation in the uroporphyrinogen III synthase gene in congenital erythropoietic porphyria. , 0, .		2

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37	Evolutionary distinct roles of β -secretase subunit nicastrin in zebrafish and humans. Journal of Dermatological Science, 2022, , .	1.0	2
38	Recurrent missense mutation in the protoporphyrinogen oxidase gene underlies variegate porphyria. American Journal of Medical Genetics Part A, 1998, 79, 22-6.	2.4	2
39	Identification of a recurrent mutation in the protoporphyrinogen oxidase gene in Swiss patients with variegate porphyria: clinical and genetic implications. Cellular and Molecular Biology, 2009, 55, 96-101.	0.3	2
40	The Porphyrrias. JDDG - Journal of the German Society of Dermatology, 2022, 20, 316-331.	0.4	2
41	Valuable insights from molecular genetic studies in the porphyrias. British Journal of Dermatology, 2010, 162, 470-471.	1.4	1
42	Functional implications of novel ADAM10 mutations in reticulate acropigmentation of Kitamura. British Journal of Dermatology, 2017, 177, e340-e343.	1.4	1
43	<i>cyld</i> mutations differentially affect splicing and mRNA decay in Brooke-Spiegler syndrome. Journal of the European Academy of Dermatology and Venereology, 2018, 32, e331-e333.	1.3	1
44	NCSTN Deficiency and Depigmentation: All About Tyrosinase?. Journal of Investigative Dermatology, 2021, 141, 1331-1334.	0.3	1
45	The Porphyrrias. , 2010, , 469-486.		1
46	Eine Frau mit hyperpigmentierten Maculae und Papeln. JDDG - Journal of the German Society of Dermatology, 2021, 19, 1655-1659.	0.4	1
47	Basal Cell Carcinoma in Erythropoietic Protoporphyrria: All About Ultraviolet Light?. Journal of Cutaneous Medicine and Surgery, 2017, 21, 270-271.	0.6	0
48	Hereditäre Hautadnextumorsyndrome – Eine interdisziplinäre Herausforderung. JDDG - Journal of the German Society of Dermatology, 2018, 16, 259-261.	0.4	0
49	Two females with hair loss. JDDG - Journal of the German Society of Dermatology, 2019, 17, 845-847.	0.4	0
50	Painful skin swelling after water contact. JDDG - Journal of the German Society of Dermatology, 2020, 18, 764-768.	0.4	0
51	Porphyrias. , 2021, , 1-15.		0
52	Porphyrien. , 2018, , 1-16.		0
53	A woman with blistering and photosensitivity. JDDG - Journal of the German Society of Dermatology, 2022, 20, 358-360.	0.4	0
54	Eine Frau mit Blasenbildung und Photosensitivität. JDDG - Journal of the German Society of Dermatology, 2022, 20, 358-361.	0.4	0

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55	Die Porphyrien. JDDG - Journal of the German Society of Dermatology, 2022, 20, 316-333.	0.4	0
56	Porphyrias. , 2022, , 1691-1705.		0