Jorge Frank

List of Publications by Year
in descending order

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56	1,551	471371 17	315616 38
papers	citations	h-index	g-index

70	70	70	1929
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A woman with blistering and photosensitivity. JDDG - Journal of the German Society of Dermatology, 2022, 20, 358-360.	0.4	0
2	Evolutionary distinct roles of Î³-secretase subunit nicastrin in zebrafish and humans. Journal of Dermatological Science, 2022, , .	1.0	2
3	The Porphyrias. JDDG - Journal of the German Society of Dermatology, 2022, 20, 316-331.	0.4	2
4	Eine Frau mit Blasenbildung und PhotosensitivitÃt. JDDG - Journal of the German Society of Dermatology, 2022, 20, 358-361.	0.4	0
5	Die Porphyrien. JDDG - Journal of the German Society of Dermatology, 2022, 20, 316-333.	0.4	0
6	Porphyrias. , 2022, , 1691-1705.		0
7	Comorbid acne inversa and Dowlingâ€“Degos disease due to a single NCSTN mutation: is there enough evidence?. British Journal of Dermatology, 2021, 184, 374-374.	1.4	3
8	NCSTN Deficiency and Depigmentation: All About Tyrosinase?. Journal of Investigative Dermatology, 2021, 141, 1331-1334.	0.3	1
9	Porphyrias. , 2021, , 1-15.		0
10	A woman with hyperpigmented macules and papules. JDDG - Journal of the German Society of Dermatology, 2021, 19, 1656-1660.	0.4	3
11	Eine Frau mit hyperpigmentierten Maculae und Papeln. JDDG - Journal of the German Society of Dermatology, 2021, 19, 1655-1659.	0.4	1
12	Painful skin swelling after water contact. JDDG - Journal of the German Society of Dermatology, 2020, 18, 764-768.	0.4	0
13	Continuing medical and student education in dermatology during the coronavirus pandemic â€“ a major challenge. JDDG - Journal of the German Society of Dermatology, 2020, 18, 835-840.	0.4	12
14	Two females with hair loss. JDDG - Journal of the German Society of Dermatology, 2019, 17, 845-847.	0.4	0
15	Altered Notch Signaling in Dowling-Degos Disease: Additional Mutations in POGLUT1 and Further Insights into Disease Pathogenesis. Journal of Investigative Dermatology, 2019, 139, 960-964.	0.3	15
16	<scp>CYLD</scp> mutations differentially affect splicing and <scp>mRNA</scp> decay in Brookeâ€“Spiegler syndrome. Journal of the European Academy of Dermatology and Venereology, 2018, 32, e331-e333.	1.3	1
17	Intra- and Interfamilial Phenotype Variability Associated with Mutations in Î³-Secretase Subunit-Encoding PSENEN. Journal of Investigative Dermatology, 2018, 138, 1215-1218.	0.3	4
18	HereditÃre Hautadnextumorâ€Syndrome â€“ Eine interdisziplinÃre Herausforderung. JDDG - Journal of the German Society of Dermatology, 2018, 16, 259-261.	0.4	0

#	Article	IF	Citations
19	Bi-allelic Mutations in LSS, Encoding Lanosterol Synthase, Cause Autosomal-Recessive Hypotrichosis Simplex. American Journal of Human Genetics, 2018, 103, 777-785.	2.6	55
20	Phenotype variability in tumor disorders of the skin appendages associated with mutations in the CYLD gene. Archives of Dermatological Research, 2018, 310, 599-606.	1.1	7
21	Porphyrien. , 2018, , 1-16.		0
22	Basal Cell Carcinoma in Erythropoietic Protoporphyria: All About Ultraviolet Light?. Journal of Cutaneous Medicine and Surgery, 2017, 21, 270-271.	0.6	0
23	Functional implications of novel ADAM10 mutations in reticulate acropigmentation of Kitamura. British Journal of Dermatology, 2017, 177, e340-e343.	1.4	1
24	HereditÃre Hauterkrankungen â€•Klinisch und genetisch heterogen. JDDG - Journal of the German Society of Dermatology, 2017, 15, 881-882.	0.4	5
25	Mutations in Î³-secretase subunitâ€“encoding PSENEN underlie Dowling-Degos disease associated with acne inversa. Journal of Clinical Investigation, 2017, 127, 1485-1490.	3.9	73
26	Biallelic inactivation of protoporphyrinogen oxidase and hydroxymethylbilane synthase is associated with liver cancer in acute porphyrias. Journal of Hepatology, 2015, 62, 734-738.	1.8	22
27	Pathogenicity of POFUT1 in Dowling-Degos Disease: Additional Mutations and Clinical Overlap with Reticulate Acropigmentation of Kitamura. Journal of Investigative Dermatology, 2015, 135, 615-618.	0.3	25
28	Afamelanotide for Erythropoietic Protoporphyria. New England Journal of Medicine, 2015, 373, 48-59.	13.9	206
29	A novel therapeutic strategy for turban tumor: scalp excision and combined reconstruction with artificial dermis and split skin graft. International Journal of Dermatology, 2014, 53, 246-249.	0.5	14
30	Photosensitivity in the Elderlyâ€”Think of Late-Onset Protoporphyria. Journal of Investigative Dermatology, 2013, 133, 1467-1471.	0.3	6
31	Hereditary leiomyomatosis and renal cell cancer in families referred for fumarate hydratase germline mutation analysis. Clinical Genetics, 2011, 79, 49-59.	1.0	154
32	Linkage refinement of Bazex-DuprÃ©-Christol syndrome to an 11Â·4-Mb interval on chromosome Xq25-27.1. British Journal of Dermatology, 2011, 165, 201-203.	1.4	18
33	Delayed diagnosis and diminished quality of life in erythropoietic protoporphyria: results of a cross-sectional study in Sweden. Journal of Internal Medicine, 2011, 269, 270-274.	2.7	18
34	Porphyria cutanea tarda â€“ When skin meets liver. Bailliere's Best Practice and Research in Clinical Gastroenterology, 2010, 24, 735-745.	1.0	67
35	Valuable insights from molecular genetic studies in the porphyrias. British Journal of Dermatology, 2010, 162, 470-471.	1.4	1
36	Hepatocellular Carcinoma in Variegate Porphyria: A Serious Complication. Acta Dermato-Venereologica, 2010, 90, 512-515.	0.6	24

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37	The Porphyrias. , 2010, , 469-486.		1
38	A homozygous mutation in the ferrochelatase gene underlies erythropoietic protoporphyria associated with palmar keratoderma. British Journal of Dermatology, 2009, 160, 1330-1334.	1.4	14
39	Molecular pathways involved in hair follicle tumor formation: all about mammalian target of rapamycin?. Experimental Dermatology, 2009, 18, 185-191.	1.4	22
40	Clinical andÂmolecular genetic aspects ofÂhereditary multiple cutaneous leiomyomatosis. European Journal of Dermatology, 2009, 19, 545-551.	0.3	49
41	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
42	Erythropoietic protoporphyria without skin symptoms-you do not always see what they feel. European Journal of Pediatrics, 2008, 167, 703-706.	1.3	23
43	Treatment Of Acute Edema Attacks In Hereditary Angioedema With A Bradykinin Receptor-2 Antagonist (Icatibant). Journal of Allergy and Clinical Immunology, 2007, 119, S278.	1.5	127
44	The Wnt signalling ligand RSPO4, causing inherited anonychia, is not mutated in a patient with congenital nail hypoplasia/aplasia with underlying skeletal defects. British Journal of Dermatology, 2007, 157, 801-802.	1.4	8
45	Mutations in the Gene Encoding the Wnt-Signaling Component R-Spondin 4 (RSPO4) Cause Autosomal Recessive Anonychia. American Journal of Human Genetics, 2006, 79, 1105-1109.	2.6	94
46	Phenotype Diversity in Familial Cylindromatosis: A Frameshift Mutation in the Tumor Suppressor Gene CYLD Underlies Different Tumors of Skin Appendages. Journal of Investigative Dermatology, 2002, 119, 527-531.	0.3	87
47	Characterization of the desmosomal cadherin gene family: Genomic organization of two desmoglein genes on human chromosome 18q12. Experimental Dermatology, 2001, 10, 90-94.	1.4	17
48	Mutations in the translation initiation codon of the protoporphyrinogen oxidase gene underlie variegate porphyria. Clinical and Experimental Dermatology, 1999, 24, 296-301.	0.6	17
49	Exposing the human nude phenotype. Nature, 1999, 398, 473-474.	13.7	247
50	Erythropoietic protoporphyria: identification of novel mutations in the ferrochelatase gene and comparison of biochemical markers versus molecular analysis as diagnostic strategies. Journal of Investigative Medicine, 1999, 47, 278-84.	0.7	3
51	Recurrent missense mutation in the protoporphyrinogen oxidase gene underlies variegate porphyria. , 1998, 79, 22-26.		13
52	The genetic basis of "Scarsdale Gourmet Diet" variegate porphyria: a missense mutation in the protoporphyrinogen oxidase gene. Archives of Dermatological Research, 1998, 290, 441-445.	1.1	14
53	C73R is a hotspot mutation in the uroporphyrinogen III synthase gene in congenital erythropoietic porphyria. Annals of Human Genetics, 1998, 62, 225-230.	0.3	24
54	Molecular basis of variegate porphyria: a missense mutation in the protoporphyrinogen oxidase gene.. Journal of Medical Genetics, 1998, 35, 244-247.	1.5	11

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55	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
56	C73R is a hotspot mutation in the uroporphyrinogen III synthase gene in congenital erythropoietic porphyria. , 0, .		2