John Digiovanna

List of Publications by Year in descending order

69 papers	4,875 citations	136950 32 h-index	110387 64 g-index
70 all docs	70 docs citations	70 times ranked	4072 citing authors

#	Article	IF	Citations
1	Development and Initial Validation of a Novel System to Assess Ichthyosis Severity. JAMA Dermatology, 2022, 158, 359.	4.1	4
2	Knowledge is power. British Journal of Dermatology, 2022, 186, 607-608.	1.5	0
3	Thyroid nodules in xeroderma pigmentosum patients: a feature of premature aging. Journal of Endocrinological Investigation, 2021, 44, 1475-1482.	3.3	7
4	Consensus recommendations for the use of retinoids in ichthyosis and other disorders of cornification in children and adolescents. Pediatric Dermatology, 2021, 38, 164-180.	0.9	34
5	Xeroderma Pigmentosum: A Model for Human Premature Aging. Journal of Investigative Dermatology, 2021, 141, 976-984.	0.7	26
6	Cockayne syndrome, MEN1, and genomic variants: Exome sequencing is changing our view of the genetic landscape. Pediatric Dermatology, 2021, 38, 913-918.	0.9	0
7	Metronidazole-Induced Hepatitis in a Teenager With Xeroderma Pigmentosum and Trichothiodystrophy Overlap. Pediatrics, 2021, 148, e2021050360.	2.1	4
8	Differences in peripheral neuropathy in xeroderma pigmentosum complementation groups A and D as evaluated by nerve conduction studies. BMC Neurology, 2021, 21, 393.	1.8	4
9	A novel frameshift mutation in SOX10 causes Waardenburg syndrome with peripheral demyelinating neuropathy, visual impairment and the absence of Hirschsprung disease. American Journal of Medical Genetics, Part A, 2020, 182, 1278-1283.	1.2	7
10	Predisposition to hematologic malignancies in patients with xeroderma pigmentosum. Haematologica, 2020, 105, e144-e146.	3.5	18
11	Mortalityâ€associated immunological abnormalities in trichothiodystrophy: correlation of reduced levels of immunoglobulin and neutrophils with poor patient survival. British Journal of Haematology, 2019, 185, 752-754.	2.5	8
12	Hydroa vacciniformeâ€“like lymphoproliferative disorder: an EBV disease with a low risk of systemic illness in whites. Blood, 2019, 133, 2753-2764.	1.4	46
13	Reproductive Health in Xeroderma Pigmentosum. Obstetrics and Gynecology, 2019, 134, 814-819.	2.4	6
14	Use of Big Data to Estimate Prevalence of Defective DNA Repair Variants in the US Population. JAMA Dermatology, 2019, 155, 72.	4.1	11
15	Recurrent scarring papulovesicular lesions on sun-exposed skin in a 22-year-old man. Journal of the American Academy of Dermatology, 2018, 78, 637-642.	1.2	5
16	Variant subtype of xeroderma pigmentosum diagnosed in a 77-year-old woman. JAAD Case Reports, 2018, 4, 1074-1076.	0.8	6
17	Neurodegeneration as the presenting symptom in 2 adults with xeroderma pigmentosum complementation group F. Neurology: Genetics, 2018, 4, e240.	1.9	9
18	Four-dimensional, dynamic mosaicism is a hallmark of normal human skin that permits mapping of the organization and patterning of human epidermis during terminal differentiation. PLoS ONE, 2018, 13, e0198011.	2.5	3

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19	Basic Science Insights into Clinical Puzzles. Dermatologic Clinics, 2017, 35, ix-x.	1.7	0
20	Molecular diagnosis of xeroderma pigmentosum variant in an isolated population: the interface between precision medicine and public health. British Journal of Dermatology, 2017, 176, 1125-1126.	1.5	0
21	GTF2E2 Mutations Destabilize the General Transcription Factor Complex TFIIE in Individuals with DNA Repair-Proficient Trichothiodystrophy. American Journal of Human Genetics, 2016, 98, 627-642.	6.2	49
22	Comparing histopathology from patients with X-linked recessive ichthyosis and autosomal recessive congenital ichthyosis with transglutaminase 1 mutation: A report from the National Registry for Ichthyosis and Related Skin Disorders. Journal of the American Academy of Dermatology, 2016, 74, 1008-1010.e2.	1.2	5
23	Cutaneous adverse events in multiple sclerosis patients treated with daclizumab. Neurology, 2016, 86, 847-855.	1.1	36
24	Comedonal and Cystic Fibrofolliculomas in Birt-Hogg-Dube Syndrome. JAMA Dermatology, 2015, 151, 770.	4.1	28
25	Mutations in the TTDN1 Gene Are Associated with a Distinct Trichothiodystrophy Phenotype. Journal of Investigative Dermatology, 2015, 135, 734-741.	0.7	32
26	Cutaneous Adverse Effects Associated With the Tyrosine-Kinase Inhibitor Cabozantinib. JAMA Dermatology, 2015, 151, 170.	4.1	36
27	Global Contributions to the Understanding of DNA Repair and Skin Cancer. Journal of Investigative Dermatology, 2014, 134, E8-E17.	0.7	7
28	High frequency of <scp>PTEN</scp> mutations in nevi and melanomas from xeroderma pigmentosum patients. Pigment Cell and Melanoma Research, 2014, 27, 454-464.	3.3	40
29	Living with xeroderma pigmentosum: comprehensive photoprotection for highly photosensitive patients. Photodermatology Photoimmunology and Photomedicine, 2014, 30, 146-152.	1.5	50
30	Rapid development of migratory, linear, and serpiginous lesions in association with immunosuppression. Journal of the American Academy of Dermatology, 2014, 70, 1130-1134.	1.2	18
31	Fluorouracil and Other Predictors of Morpheaform Basal Cell Carcinoma Among High-Risk Patients: The Veterans Affairs Topical Tretinoin Chemoprevention Trial. JAMA Dermatology, 2014, 150, 332.	4.1	8
32	The influence of DNA repair on neurological degeneration, cachexia, skin cancer and internal neoplasms: autopsy report of four xeroderma pigmentosum patients (XP-A, XP-C and XP-D). Acta Neuropathologica Communications, 2013, 1, 4.	5.2	40
33	Ocular Manifestations of Xeroderma Pigmentosum. Ophthalmology, 2013, 120, 1324-1336.	5.2	74
34	Auditory analysis of xeroderma pigmentosum 1971â€“2012: hearing function, sun sensitivity and DNA repair predict neurological degeneration. Brain, 2013, 136, 194-208.	7.6	50
35	Histopathology of the Inner Ear in Patients With Xeroderma Pigmentosum and Neurologic Degeneration. Otology and Neurotology, 2013, 34, 1230-1236.	1.3	20
36	Systemic retinoids in the management of ichthyoses and related skin types. Dermatologic Therapy, 2013, 26, 26-38.	1.7	80

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37	Burning issues in the diagnosis of xeroderma pigmentosum. British Journal of Dermatology, 2013, 169, 1176-1176.	1.5	8
38	Shining a Light on Xeroderma Pigmentosum. Journal of Investigative Dermatology, 2012, 132, 785-796.	0.7	419
39	Cancer and neurologic degeneration in xeroderma pigmentosum: long term follow-up characterises the role of DNA repair. Journal of Medical Genetics, 2011, 48, 168-176.	3.2	371
40	Nucleotide Excision Repair Proteins Rapidly Accumulate but Fail to Persist in Human XPâ€E (DDB2) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50	2.5	14
41	Multiple Skin Cancers in Adults with Mutations in the XP-E (DDB2) DNA Repair Gene. Journal of Investigative Dermatology, 2011, 131, 785-788.	0.7	23
42	Genetic Diversity in Melanoma Metastases from a Patient with Xeroderma Pigmentosum. Journal of Investigative Dermatology, 2010, 130, 1188-1191.	0.7	7
43	Revised nomenclature and classification of inherited ichthyoses: Results of the First Ichthyosis Consensus Conference in SorÃ¨ze 2009. Journal of the American Academy of Dermatology, 2010, 63, 607-641.	1.2	610
44	Xeroderma Pigmentosum-Variant Patients from America, Europe, and Asia. Journal of Investigative Dermatology, 2008, 128, 2055-2068.	0.7	76
45	Histopathologic characterization of epidermolytic hyperkeratosis: A systematic review of histology from the National Registry for Ichthyosis and Related Skin Disorders. Journal of the American Academy of Dermatology, 2008, 59, 86-90.	1.2	68
46	Skin cancers, blindness, and anterior tongue mass in African brothers. Journal of the American Academy of Dermatology, 2008, 59, 881-886.	1.2	41
47	Xeroderma pigmentosum, trichothiodystrophy and Cockayne syndrome: A complex genotypeâ€“phenotype relationship. Neuroscience, 2007, 145, 1388-1396.	2.3	405
48	Retinoid Treatment of the Disorders of Cornification. Basic and Clinical Dermatology, 2007, , 153-170.	0.1	2
49	Ichthyosiform dermatoses: So many discoveries, so little progress. Journal of the American Academy of Dermatology, 2004, 51, 31-34.	1.2	8
50	Genetic Heterogeneity in Erythrokeratodermia Variabilis: Novel Mutations in the Connexin Gene GJB4 (Cx30.3) and Genotype-Phenotype Correlations. Journal of Investigative Dermatology, 2003, 120, 601-609.	0.7	112
51	Ichthyosis. American Journal of Clinical Dermatology, 2003, 4, 81-95.	6.7	138
52	Missense Mutations in GJB2 Encoding Connexin-26 Cause the Ectodermal Dysplasia Keratitis-Ichthyosis-Deafness Syndrome. American Journal of Human Genetics, 2002, 70, 1341-1348.	6.2	345
53	Isotretinoin effects on bone. Journal of the American Academy of Dermatology, 2001, 45, S176-S182.	1.2	122
54	Retinoid chemoprevention in patients at high risk for skin cancer. Medical and Pediatric Oncology, 2001, 36, 564-567.	1.0	55

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55	Splice-site mutation in TGM1 in congenital recessive ichthyosis in American families: molecular, genetic, genealogic, and clinical studies. Human Genetics, 2000, 106, 492-499.	3.8	32
56	Mutations in the human connexin gene GJB3 cause erythrokeratodermia variabilis. Nature Genetics, 1998, 20, 366-369.	21.4	356
57	Nevoid basal cell carcinoma syndrome with medulloblastoma in an African-American boy: A rare case illustrating gene-environment interaction. American Journal of Medical Genetics Part A, 1997, 69, 309-314.	2.4	30
58	Mutations in the gene for transglutaminase 1 in autosomal recessive lamellar ichthyosis. Nature Genetics, 1995, 9, 279-283.	21.4	336
59	Osteoporosis is a toxic effect of long-term etretinate therapy. Archives of Dermatology, 1995, 131, 1263-7.	1.4	9
60	Clinical Heterogeneity in Epidermolytic Hyperkeratosis. Archives of Dermatology, 1994, 130, 1026.	1.4	102
61	Clinical findings in two Africanâ€American families with the nevoid basal cell carcinoma syndrome (NBCC). American Journal of Medical Genetics Part A, 1994, 50, 272-281.	2.4	64
62	Clinical heterogeneity in epidermolytic hyperkeratosis. Archives of Dermatology, 1994, 130, 1026-35.	1.4	15
63	Chemoprevention of Skin Cancer in Xeroderma Pigmentosum. Journal of Dermatology, 1992, 19, 715-718.	1.2	82
64	Linkage of epidermolytic hyperkeratosis to the type II keratin gene cluster on chromosome 12q. Nature Genetics, 1992, 1, 301-305.	21.4	103
65	Exacerbation of Darier's disease by lithium carbonate. Journal of the American Academy of Dermatology, 1990, 23, 926-928.	1.2	30
66	Etretinate. Archives of Dermatology, 1989, 125, 246.	1.4	37
67	Etretinate. Persistent serum levels after long-term therapy. Archives of Dermatology, 1989, 125, 246-251.	1.4	39
68	Oral Synthetic Retinoid Treatment in Children. Pediatric Dermatology, 1983, 1, 77-88.	0.9	45
69	Trichothiodystrophy Hair Shafts Display Distinct Ultrastructural Features. Experimental Dermatology, 0, , .	2.9	0