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	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
2	Shining a Light on Xeroderma Pigmentosum. Journal of Investigative Dermatology, 2012, 132, 785-796.	0.7	419
3	Xeroderma pigmentosum, trichothiodystrophy and Cockayne syndrome: A complex genotypeâ€“phenotype relationship. Neuroscience, 2007, 145, 1388-1396.	2.3	405
4	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
5	Mutations in the human connexin gene GJB3 cause erythrokeratodermia variabilis. Nature Genetics, 1998, 20, 366-369.	21.4	356
6	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
7	Mutations in the gene for transglutaminase 1 in autosomal recessive lamellar ichthyosis. Nature Genetics, 1995, 9, 279-283.	21.4	336
8	Ichthyosis. American Journal of Clinical Dermatology, 2003, 4, 81-95.	6.7	138
9	Isotretinoin effects on bone. Journal of the American Academy of Dermatology, 2001, 45, S176-S182.	1.2	122
10	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
11	Linkage of epidermolytic hyperkeratosis to the type II keratin gene cluster on chromosome 12q. Nature Genetics, 1992, 1, 301-305.	21.4	103
12	Clinical Heterogeneity in Epidermolytic Hyperkeratosis. Archives of Dermatology, 1994, 130, 1026.	1.4	102
13	Chemoprevention of Skin Cancer in Xeroderma Pigmentosum. Journal of Dermatology, 1992, 19, 715-718.	1.2	82
14	Systemic retinoids in the management of ichthyoses and related skin types. Dermatologic Therapy, 2013, 26, 26-38.	1.7	80
15	Xeroderma Pigmentosum-Variant Patients from America, Europe, and Asia. Journal of Investigative Dermatology, 2008, 128, 2055-2068.	0.7	76
16	Ocular Manifestations of Xeroderma Pigmentosum. Ophthalmology, 2013, 120, 1324-1336.	5.2	74
17	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
18	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

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19	Retinoid chemoprevention in patients at high risk for skin cancer. Medical and Pediatric Oncology, 2001, 36, 564-567.	1.0	55
20	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
21	Living with xeroderma pigmentosum: comprehensive photoprotection for highly photosensitive patients. Photodermatology Photoimmunology and Photomedicine, 2014, 30, 146-152.	1.5	50
22	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
23	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
24	Oral Synthetic Retinoid Treatment in Children. Pediatric Dermatology, 1983, 1, 77-88.	0.9	45
25	Skin cancers, blindness, and anterior tongue mass in African brothers. Journal of the American Academy of Dermatology, 2008, 59, 881-886.	1.2	41
26	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
27	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
28	Etretinate. Persistent serum levels after long-term therapy. Archives of Dermatology, 1989, 125, 246-251.	1.4	39
29	Etretinate. Archives of Dermatology, 1989, 125, 246.	1.4	37
30	Cutaneous Adverse Effects Associated With the Tyrosine-Kinase Inhibitor Cabozantinib. JAMA Dermatology, 2015, 151, 170.	4.1	36
31	Cutaneous adverse events in multiple sclerosis patients treated with daclizumab. Neurology, 2016, 86, 847-855.	1.1	36
32	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
33	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
34	Mutations in the TTDN1 Gene Are Associated with a Distinct Trichothiodystrophy Phenotype. Journal of Investigative Dermatology, 2015, 135, 734-741.	0.7	32
35	Exacerbation of Darier's disease by lithium carbonate. Journal of the American Academy of Dermatology, 1990, 23, 926-928.	1.2	30
36	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

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37	Comedonal and Cystic Fibrofolliculomas in Birt-Hogg-Dube Syndrome. JAMA Dermatology, 2015, 151, 770.	4.1	28
38	Xeroderma Pigmentosum: A Model for Human Premature Aging. Journal of Investigative Dermatology, 2021, 141, 976-984.	0.7	26
39	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
40	Histopathology of the Inner Ear in Patients With Xeroderma Pigmentosum and Neurologic Degeneration. Otology and Neurotology, 2013, 34, 1230-1236.	1.3	20
41	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
42	Predisposition to hematologic malignancies in patients with xeroderma pigmentosum. Haematologica, 2020, 105, e144-e146.	3.5	18
43	Clinical heterogeneity in epidermolytic hyperkeratosis. Archives of Dermatology, 1994, 130, 1026-35.	1.4	15
44	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
45	Use of Big Data to Estimate Prevalence of Defective DNA Repair Variants in the US Population. JAMA Dermatology, 2019, 155, 72.	4.1	11
46	Neurodegeneration as the presenting symptom in 2 adults with xeroderma pigmentosum complementation group F. Neurology: Genetics, 2018, 4, e240.	1.9	9
47	Osteoporosis is a toxic effect of long-term etretinate therapy. Archives of Dermatology, 1995, 131, 1263-7.	1.4	9
48	Ichthyosiform dermatoses: So many discoveries, so little progress. Journal of the American Academy of Dermatology, 2004, 51, 31-34.	1.2	8
49	Burning issues in the diagnosis of xeroderma pigmentosum. British Journal of Dermatology, 2013, 169, 1176-1176.	1.5	8
50	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
51	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
52	Genetic Diversity in Melanoma Metastases from a Patient with Xeroderma Pigmentosum. Journal of Investigative Dermatology, 2010, 130, 1188-1191.	0.7	7
53	Global Contributions to the Understanding of DNA Repair and Skin Cancer. Journal of Investigative Dermatology, 2014, 134, E8-E17.	0.7	7
54	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

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55	Thyroid nodules in xeroderma pigmentosum patients: a feature of premature aging. Journal of Endocrinological Investigation, 2021, 44, 1475-1482.	3.3	7
56	Variant subtype of xeroderma pigmentosum diagnosed in a 77-year-old woman. JAAD Case Reports, 2018, 4, 1074-1076.	0.8	6
57	Reproductive Health in Xeroderma Pigmentosum. Obstetrics and Gynecology, 2019, 134, 814-819.	2.4	6
58	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
59	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
60	Metronidazole-Induced Hepatitis in a Teenager With Xeroderma Pigmentosum and Trichothiodystrophy Overlap. Pediatrics, 2021, 148, e2021050360.	2.1	4
61	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
62	Development and Initial Validation of a Novel System to Assess Ichthyosis Severity. JAMA Dermatology, 2022, 158, 359.	4.1	4
63	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
64	Retinoid Treatment of the Disorders of Cornification. Basic and Clinical Dermatology, 2007, , 153-170.	0.1	2
65	Basic Science Insights into Clinical Puzzles. Dermatologic Clinics, 2017, 35, ix-x.	1.7	0
66	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
67	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
68	Knowledge is power. British Journal of Dermatology, 2022, 186, 607-608.	1.5	0
69	Trichothiodystrophy Hair Shafts Display Distinct Ultrastructural Features. Experimental Dermatology, 0, , .	2.9	0