Sikandar G Khan

List of Publications by Year in descending order

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60 3,386 29
papers citations h-index

61 61 61 2649 all docs docs citations times ranked citing authors

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#	Article	IF	Citations
1	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.	1.5	371
2	Phenotypic heterogeneity in the XPB DNA helicase gene (ERCC3): xeroderma pigmentosum without and with Cockayne syndrome. Human Mutation, 2006, 27, 1092-1103.	1.1	365
3	Persistence of repair proteins at unrepaired DNA damage distinguishes diseases with <i>ERCC2 < /i > (<i>XPD < /i>) mutations: cancer-prone xeroderma pigmentosum vs. non-cancer-prone trichothiodystrophy. Human Mutation, 2008, 29, 1194-1208.</i></i>	1.1	317
4	Repair of UV photolesions in xeroderma pigmentosum group C cells induced by translational readthrough of premature termination codons. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 19483-19488.	3.3	310
5	The human XPC DNA repair gene: arrangement, splice site information content and influence of a single nucleotide polymorphism in a splice acceptor site on alternative splicing and function. Nucleic Acids Research, 2002, 30, 3624-3631.	6.5	146
6	A new xeroderma pigmentosum group C poly(AT) insertion/deletion polymorphism. Carcinogenesis, 2000, 21, 1821-1825.	1.3	122
7	Assessment of 3 xeroderma pigmentosum group C gene polymorphisms and risk of cutaneous melanoma: a case–control study. Carcinogenesis, 2005, 26, 1085-1090.	1.3	98
8	Relationship of Neurologic Degeneration to Genotype in Three Xeroderma Pigmentosum Group G Patients. Journal of Investigative Dermatology, 2002, 118, 972-982.	0.3	96
9	An intronic poly (AT) polymorphism of the DNA repair gene XPC and risk of squamous cell carcinoma of the head and neck: a case-control study. Cancer Research, 2001, 61, 3321-5.	0.4	91
10	Evidence of ultraviolet type mutations in xeroderma pigmentosum melanomas. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 6279-6284.	3.3	85
11	The human XPG gene: gene architecture, alternative splicing and single nucleotide polymorphisms. Nucleic Acids Research, 2001, 29, 1443-1452.	6.5	80
12	Reduced XPC DNA repair gene mRNA levels in clinically normal parents of xeroderma pigmentosum patients. Carcinogenesis, 2005, 27, 84-94.	1.3	79
13	The xeroderma pigmentosum group C gene leads to selective repair of cyclobutane pyrimidine dimers rather than 6-4 photoproducts. Proceedings of the National Academy of Sciences of the United States of America, 2000, 97, 2151-2156.	3.3	76
14	Xeroderma Pigmentosum-Variant Patients from America, Europe, and Asia. Journal of Investigative Dermatology, 2008, 128, 2055-2068.	0.3	76
15	Ocular Manifestations of Xeroderma Pigmentosum. Ophthalmology, 2013, 120, 1324-1336.	2.5	74
16	Xeroderma Pigmentosum Group C Splice Mutation Associated with Autism and Hypoglycinemia11An abstract of this manuscript was presented at the annual meeting of the Society for Investigative Dermatology in Washington, DC. Journal of Investigative Dermatology, 1998, 111, 791-796.	0.3	66
17	Two essential splice lariat branchpoint sequences in one intron in a xeroderma pigmentosum DNA repair gene: mutations result in reduced XPC mRNA levels that correlate with cancer risk. Human Molecular Genetics, 2003, 13, 343-352.	1.4	63
18	Auditory analysis of xeroderma pigmentosum 1971–2012: hearing function, sun sensitivity and DNA repair predict neurological degeneration. Brain, 2013, 136, 194-208.	3.7	50

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19	Living with xeroderma pigmentosum: comprehensive photoprotection for highly photosensitive patients. Photodermatology Photoimmunology and Photomedicine, 2014, 30, 146-152.	0.7	50
20	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.	2.6	49
21	A Stop Codon in Xeroderma Pigmentosum Group C Families in Turkey and Italy: Molecular Genetic Evidence for a Common Ancestor. Journal of Investigative Dermatology, 2001, 117, 197-204.	0.3	45
22	Skin cancers, blindness, and anterior tongue mass in African brothers. Journal of the American Academy of Dermatology, 2008, 59, 881-886.	0.6	41
23	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.	2.4	40
24	High frequency of <scp>PTEN</scp> mutations in nevi and melanomas from xeroderma pigmentosum patients. Pigment Cell and Melanoma Research, 2014, 27, 454-464.	1.5	40
25	No association between three xeroderma pigmentosum group C and one group G gene polymorphisms and risk of cutaneous melanoma. European Journal of Human Genetics, 2005, 13, 253-255.	1.4	38
26	XPC initiation codon mutation in xeroderma pigmentosum patients with and without neurological symptoms. DNA Repair, 2009, 8, 114-125.	1.3	35
27	Adverse effects of trichothiodystrophy DNA repair and transcription gene disorder on human fetal development. Clinical Genetics, 2010, 77, 365-373.	1.0	35
28	Mutations in the TTDN1 Gene Are Associated with a Distinct Trichothiodystrophy Phenotype. Journal of Investigative Dermatology, 2015, 135, 734-741.	0.3	32
29	Ocular Manifestations of Trichothiodystrophy. Ophthalmology, 2011, 118, 2335-2342.	2.5	30
30	Xeroderma pigmentosum complementation group G patient with a novel homozygous missense mutation and no neurological abnormalities. Experimental Dermatology, 2012, 21, 304-307.	1.4	30
31	Influence of XPB helicase on recruitment and redistribution of nucleotide excision repair proteins at sites of UV-induced DNA damage. DNA Repair, 2007, 6, 1359-1370.	1.3	27
32	Strict sun protection results in minimal skin changes in a patient with xeroderma pigmentosum and a novel c.2009delG mutation in XPD (ERCC2). Experimental Dermatology, 2009, 18, 64-68.	1.4	27
33	Clinical, Cellular, and Molecular Features of an Israeli Xeroderma Pigmentosum Family with a Frameshift Mutation in the XPC Gene: Sun Protection Prolongs Life. Journal of Investigative Dermatology, 2000, 115, 974-980.	0.3	26
34	Xeroderma Pigmentosum: A Model for Human Premature Aging. Journal of Investigative Dermatology, 2021, 141, 976-984.	0.3	26
35	Highâ€risk pregnancy and neonatal complications in the DNA repair and transcription disorder trichothiodystrophy: report of 27 affected pregnancies. Prenatal Diagnosis, 2011, 31, 1046-1053.	1.1	24
36	Abnormal XPD-induced nuclear receptor transactivation in DNA repair disorders: trichothiodystrophy and xeroderma pigmentosum. European Journal of Human Genetics, 2013, 21, 831-837.	1.4	21

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37	Brittle hair, developmental delay, neurologic abnormalities, and photosensitivity in a 4-year-old girl. Journal of the American Academy of Dermatology, 2010, 63, 323-328.	0.6	18
38	Effect of mutations in XPD(ERCC2) on pregnancy and prenatal development in mothers of patients with trichothiodystrophy or xeroderma pigmentosum. European Journal of Human Genetics, 2012, 20, 1308-1310.	1.4	18
39	Predisposition to hematologic malignancies in patients with xeroderma pigmentosum. Haematologica, 2020, 105, e144-e146.	1.7	18
40	<i>XPC</i> branch-point sequence mutations disrupt U2 snRNP binding, resulting in abnormal pre-mRNA splicing in xeroderma pigmentosum patients. Human Mutation, 2010, 31, 167-175.	1.1	17
41	Readthrough of stop codons by use of aminoglycosides in cells from xeroderma pigmentosum group <scp>C</scp> patients. Experimental Dermatology, 2015, 24, 296-297.	1.4	17
42	Unexpected Occurrence of Xeroderma Pigmentosum in an Uncle and Nephew. Archives of Dermatology, 2009, 145, 1285-91.	1.7	13
43	Chimeric Negative Regulation ofp14ARFandTBX1by a t(9;22) Translocation Associated with Melanoma, Deafness, and DNA Repair Deficiency. Human Mutation, 2013, 34, 1250-1259.	1.1	11
44	Use of Big Data to Estimate Prevalence of Defective DNA Repair Variants in the US Population. JAMA Dermatology, 2019, 155, 72.	2.0	11
45	Farnesyltransferase Activity and mRNA Expression in Human Skin Basal Cell Carcinomas. Biochemical and Biophysical Research Communications, 1996, 220, 795-801.	1.0	10
46	Ancient origin of a Japanese xeroderma pigmentosum founder mutation. Journal of Dermatological Science, 2013, 69, 175-176.	1.0	10
47	Burning issues in the diagnosis of xeroderma pigmentosum. British Journal of Dermatology, 2013, 169, 1176-1176.	1.4	8
48	Pembrolizumab treatment of a patient with xeroderma pigmentosum with disseminated melanoma and multiple nonmelanoma skin cancers. British Journal of Dermatology, 2018, 178, 1009-1009.	1.4	8
49	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.	1.2	8
50	Thyroid nodules in xeroderma pigmentosum patients: a feature of premature aging. Journal of Endocrinological Investigation, 2021, 44, 1475-1482.	1.8	7
51	Variant subtype of xeroderma pigmentosum diagnosed in a 77-year-old woman. JAAD Case Reports, 2018, 4, 1074-1076.	0.4	6
52	Reproductive Health in Xeroderma Pigmentosum. Obstetrics and Gynecology, 2019, 134, 814-819.	1.2	6
53	A Novel Complex Insertion/Deletion Mutation in the XPC DNA Repair Gene Leads to Skin Cancer in an Iraqi Family. Journal of Investigative Dermatology, 2006, 126, 2542-2544.	0.3	5
54	Metronidazole-Induced Hepatitis in a Teenager With Xeroderma Pigmentosum and Trichothiodystrophy Overlap. Pediatrics, 2021, 148, e2021050360.	1.0	4

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55	Differences in peripheral neuropathy in xeroderma pigmentosum complementation groups A and D as evaluated by nerve conduction studies. BMC Neurology, 2021, 21, 393.	0.8	4
56	Ultraviolet light selection assay to optimize oligonucleotide correction of mutations in endogenous xeroderma pigmentosum genes. Gene Therapy, 2004, 11, 1729-1734.	2.3	3
57	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.	1.1	3
58	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.	1.4	0
59	Cockayne syndrome, MEN1, and genomic variants: Exome sequencing is changing our view of the genetic landscape. Pediatric Dermatology, 2021, 38, 913-918.	0.5	0
60	Trichothiodystrophy Hair Shafts Display Distinct Ultrastructural Features. Experimental Dermatology, 0, , .	1.4	0