

# Sikandar G Khan

## List of Publications by Year in descending order

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Version: 2024-02-01

60  
papers

3,386  
citations

172207

29  
h-index

143772

57  
g-index

61  
all docs

61  
docs citations

61  
times ranked

2649  
citing authors

| #  | ARTICLE  | IF  | CITATIONS |
|----|--|-----|-----------|
| 1  | Thyroid nodules in xeroderma pigmentosum patients: a feature of premature aging. <i>Journal of Endocrinological Investigation</i> , 2021, 44, 1475-1482.   | 1.8 | 7         |
| 2  | Xeroderma Pigmentosum: A Model for Human Premature Aging. <i>Journal of Investigative Dermatology</i> , 2021, 141, 976-984.  | 0.3 | 26        |
| 3  | Cockayne syndrome, MEN1, and genomic variants: Exome sequencing is changing our view of the genetic landscape. <i>Pediatric Dermatology</i> , 2021, 38, 913-918.   | 0.5 | 0         |
| 4  | Metronidazole-Induced Hepatitis in a Teenager With Xeroderma Pigmentosum and Trichothiodystrophy Overlap. <i>Pediatrics</i> , 2021, 148, e2021050360.  | 1.0 | 4         |
| 5  | Differences in peripheral neuropathy in xeroderma pigmentosum complementation groups A and D as evaluated by nerve conduction studies. <i>BMC Neurology</i> , 2021, 21, 393.   | 0.8 | 4         |
| 6  | Predisposition to hematologic malignancies in patients with xeroderma pigmentosum. <i>Haematologica</i> , 2020, 105, e144-e146.  | 1.7 | 18        |
| 7  | Mortality-associated immunological abnormalities in trichothiodystrophy: correlation of reduced levels of immunoglobulin and neutrophils with poor patient survival. <i>British Journal of Haematology</i> , 2019, 185, 752-754. | 1.2 | 8         |
| 8  | Reproductive Health in Xeroderma Pigmentosum. <i>Obstetrics and Gynecology</i> , 2019, 134, 814-819.   | 1.2 | 6         |
| 9  | Use of Big Data to Estimate Prevalence of Defective DNA Repair Variants in the US Population. <i>JAMA Dermatology</i> , 2019, 155, 72.   | 2.0 | 11        |
| 10 | Variant subtype of xeroderma pigmentosum diagnosed in a 77-year-old woman. <i>JAAD Case Reports</i> , 2018, 4, 1074-1076.  | 0.4 | 6         |
| 11 | Pembrolizumab treatment of a patient with xeroderma pigmentosum with disseminated melanoma and multiple nonmelanoma skin cancers. <i>British Journal of Dermatology</i> , 2018, 178, 1009-1009.                                  | 1.4 | 8         |
| 12 | 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. <i>PLoS ONE</i> , 2018, 13, e0198011.         | 1.1 | 3         |
| 13 | Molecular diagnosis of xeroderma pigmentosum variant in an isolated population: the interface between precision medicine and public health. <i>British Journal of Dermatology</i> , 2017, 176, 1125-1126.                        | 1.4 | 0         |
| 14 | GTF2E2 Mutations Destabilize the General Transcription Factor Complex TFIIE in Individuals with DNA Repair-Proficient Trichothiodystrophy. <i>American Journal of Human Genetics</i> , 2016, 98, 627-642.                        | 2.6 | 49        |
| 15 | Readthrough of stop codons by use of aminoglycosides in cells from xeroderma pigmentosum group C patients. <i>Experimental Dermatology</i> , 2015, 24, 296-297.  | 1.4 | 17        |
| 16 | Mutations in the TTDN1 Gene Are Associated with a Distinct Trichothiodystrophy Phenotype. <i>Journal of Investigative Dermatology</i> , 2015, 135, 734-741.  | 0.3 | 32        |
| 17 | High frequency of PTEN mutations in nevi and melanomas from xeroderma pigmentosum patients. <i>Pigment Cell and Melanoma Research</i> , 2014, 27, 454-464.   | 1.5 | 40        |
| 18 | Living with xeroderma pigmentosum: comprehensive photoprotection for highly photosensitive patients. <i>Photodermatology Photoimmunology and Photomedicine</i> , 2014, 30, 146-152.  | 0.7 | 50        |

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|----|---|-----|-----------|
| 19 | 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). <i>Acta Neuropathologica Communications</i> , 2013, 1, 4.          | 2.4 | 40        |
| 20 | Chimeric Negative Regulation of p14ARF and TBX1 by a t(9;22) Translocation Associated with Melanoma, Deafness, and DNA Repair Deficiency. <i>Human Mutation</i> , 2013, 34, 1250-1259.  | 1.1 | 11        |
| 21 | Ocular Manifestations of Xeroderma Pigmentosum. <i>Ophthalmology</i> , 2013, 120, 1324-1336.  | 2.5 | 74        |
| 22 | Ancient origin of a Japanese xeroderma pigmentosum founder mutation. <i>Journal of Dermatological Science</i> , 2013, 69, 175-176.  | 1.0 | 10        |
| 23 | Auditory analysis of xeroderma pigmentosum 1971-2012: hearing function, sun sensitivity and DNA repair predict neurological degeneration. <i>Brain</i> , 2013, 136, 194-208.  | 3.7 | 50        |
| 24 | Repair of UV photolesions in xeroderma pigmentosum group C cells induced by translational readthrough of premature termination codons. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2013, 110, 19483-19488. | 3.3 | 310       |
| 25 | Burning issues in the diagnosis of xeroderma pigmentosum. <i>British Journal of Dermatology</i> , 2013, 169, 1176-1176.   | 1.4 | 8         |
| 26 | Abnormal XPD-induced nuclear receptor transactivation in DNA repair disorders: trichothiodystrophy and xeroderma pigmentosum. <i>European Journal of Human Genetics</i> , 2013, 21, 831-837.  | 1.4 | 21        |
| 27 | Effect of mutations in XPD(ERCC2) on pregnancy and prenatal development in mothers of patients with trichothiodystrophy or xeroderma pigmentosum. <i>European Journal of Human Genetics</i> , 2012, 20, 1308-1310.                                      | 1.4 | 18        |
| 28 | Xeroderma pigmentosum complementation group G patient with a novel homozygous missense mutation and no neurological abnormalities. <i>Experimental Dermatology</i> , 2012, 21, 304-307.   | 1.4 | 30        |
| 29 | Cancer and neurologic degeneration in xeroderma pigmentosum: long term follow-up characterises the role of DNA repair. <i>Journal of Medical Genetics</i> , 2011, 48, 168-176.  | 1.5 | 371       |
| 30 | Ocular Manifestations of Trichothiodystrophy. <i>Ophthalmology</i> , 2011, 118, 2335-2342.  | 2.5 | 30        |
| 31 | High-risk pregnancy and neonatal complications in the DNA repair and transcription disorder trichothiodystrophy: report of 27 affected pregnancies. <i>Prenatal Diagnosis</i> , 2011, 31, 1046-1053.  | 1.1 | 24        |
| 32 | XPC branch-point sequence mutations disrupt U2 snRNP binding, resulting in abnormal pre-mRNA splicing in xeroderma pigmentosum patients. <i>Human Mutation</i> , 2010, 31, 167-175.   | 1.1 | 17        |
| 33 | Adverse effects of trichothiodystrophy DNA repair and transcription gene disorder on human fetal development. <i>Clinical Genetics</i> , 2010, 77, 365-373.   | 1.0 | 35        |
| 34 | Brittle hair, developmental delay, neurologic abnormalities, and photosensitivity in a 4-year-old girl. <i>Journal of the American Academy of Dermatology</i> , 2010, 63, 323-328.  | 0.6 | 18        |
| 35 | Unexpected Occurrence of Xeroderma Pigmentosum in an Uncle and Nephew. <i>Archives of Dermatology</i> , 2009, 145, 1285-91.   | 1.7 | 13        |
| 36 | XPC initiation codon mutation in xeroderma pigmentosum patients with and without neurological symptoms. <i>DNA Repair</i> , 2009, 8, 114-125.   | 1.3 | 35        |

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|----|--|-----|-----------|
| 37 | Strict sun protection results in minimal skin changes in a patient with xeroderma pigmentosum and a novel c.2009delG mutation in XPD (ERCC2). <i>Experimental Dermatology</i> , 2009, 18, 64-68.   | 1.4 | 27        |
| 38 | Evidence of ultraviolet type mutations in xeroderma pigmentosum melanomas. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2009, 106, 6279-6284.  | 3.3 | 85        |
| 39 | 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. <i>Human Mutation</i> , 2008, 29, 1194-1208. | 1.1 | 317       |
| 40 | Xeroderma Pigmentosum-Variant Patients from America, Europe, and Asia. <i>Journal of Investigative Dermatology</i> , 2008, 128, 2055-2068.   | 0.3 | 76        |
| 41 | Skin cancers, blindness, and anterior tongue mass in African brothers. <i>Journal of the American Academy of Dermatology</i> , 2008, 59, 881-886.  | 0.6 | 41        |
| 42 | Influence of XPB helicase on recruitment and redistribution of nucleotide excision repair proteins at sites of UV-induced DNA damage. <i>DNA Repair</i> , 2007, 6, 1359-1370.  | 1.3 | 27        |
| 43 | A Novel Complex Insertion/Deletion Mutation in the XPC DNA Repair Gene Leads to Skin Cancer in an Iraqi Family. <i>Journal of Investigative Dermatology</i> , 2006, 126, 2542-2544.  | 0.3 | 5         |
| 44 | Phenotypic heterogeneity in the XPB DNA helicase gene (ERCC3): xeroderma pigmentosum without and with Cockayne syndrome. <i>Human Mutation</i> , 2006, 27, 1092-1103.  | 1.1 | 365       |
| 45 | No association between three xeroderma pigmentosum group C and one group G gene polymorphisms and risk of cutaneous melanoma. <i>European Journal of Human Genetics</i> , 2005, 13, 253-255.   | 1.4 | 38        |
| 46 | Reduced XPC DNA repair gene mRNA levels in clinically normal parents of xeroderma pigmentosum patients. <i>Carcinogenesis</i> , 2005, 27, 84-94.   | 1.3 | 79        |
| 47 | Assessment of 3 xeroderma pigmentosum group C gene polymorphisms and risk of cutaneous melanoma: a case-control study. <i>Carcinogenesis</i> , 2005, 26, 1085-1090.  | 1.3 | 98        |
| 48 | Ultraviolet light selection assay to optimize oligonucleotide correction of mutations in endogenous xeroderma pigmentosum genes. <i>Gene Therapy</i> , 2004, 11, 1729-1734.  | 2.3 | 3         |
| 49 | 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. <i>Human Molecular Genetics</i> , 2003, 13, 343-352.        | 1.4 | 63        |
| 50 | 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. <i>Nucleic Acids Research</i> , 2002, 30, 3624-3631.   | 6.5 | 146       |
| 51 | Relationship of Neurologic Degeneration to Genotype in Three Xeroderma Pigmentosum Group C Patients. <i>Journal of Investigative Dermatology</i> , 2002, 118, 972-982.   | 0.3 | 96        |
| 52 | A Stop Codon in Xeroderma Pigmentosum Group C Families in Turkey and Italy: Molecular Genetic Evidence for a Common Ancestor. <i>Journal of Investigative Dermatology</i> , 2001, 117, 197-204.  | 0.3 | 45        |
| 53 | The human XPG gene: gene architecture, alternative splicing and single nucleotide polymorphisms. <i>Nucleic Acids Research</i> , 2001, 29, 1443-1452.  | 6.5 | 80        |
| 54 | 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. <i>Cancer Research</i> , 2001, 61, 3321-5.   | 0.4 | 91        |

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|----|---|-----|-----------|
| 55 | Clinical, Cellular, and Molecular Features of an Israeli Xeroderma Pigmentosum Family with a Frameshift Mutation in the XPC Gene: Sun Protection Prolongs Life. <i>Journal of Investigative Dermatology</i> , 2000, 115, 974-980.   | 0.3 | 26        |
| 56 | The xeroderma pigmentosum group C gene leads to selective repair of cyclobutane pyrimidine dimers rather than 6-4 photoproducts. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2000, 97, 2151-2156.  | 3.3 | 76        |
| 57 | A new xeroderma pigmentosum group C poly(AT) insertion/deletion polymorphism. <i>Carcinogenesis</i> , 2000, 21, 1821-1825.  | 1.3 | 122       |
| 58 | 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. <i>Journal of Investigative Dermatology</i> , 1998, 111, 791-796. | 0.3 | 66        |
| 59 | Farnesyltransferase Activity and mRNA Expression in Human Skin Basal Cell Carcinomas. <i>Biochemical and Biophysical Research Communications</i> , 1996, 220, 795-801.  | 1.0 | 10        |
| 60 | Trichothiodystrophy Hair Shafts Display Distinct Ultrastructural Features. <i>Experimental Dermatology</i> , 0, , .   | 1.4 | 0         |