A short review of the origin of the group C cell strains of Xeroderma pigmentosum in India

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Abstract

An extremely high prevalence of skin malignancies and other related abnormalities are caused by the rare genetic illness known as Xeroderma pigmentosum (XP), which is characterized by extraordinary sensitivity to ultraviolet (UV) light. There are various complementation groups that XP can be categorized into, with group C being the most common in India. The goal of this paper is to give a brief outline of the history of group C cell Xeroderma pigmentosum in India.

The introduction highlights the need of comprehending the genetic foundation of this condition and briefly covers the clinical signs of XP. Beginning in the 1970s, XP research in India concentrated initially on clinical phenotyping and epidemiological studies. The severe cutaneous signs and high frequency of skin malignancies seen in those with XP were noted in the first publication on the disease in India. This report sparked additional research into the genetic flaws that cause XP in the Indian population.

The history of XP research in India, the finding and description of group C cell strains, as well as the function of XP complementation groups in genetic research, are covered in the parts that follow. India's XP research has come a long way over the years, with the discovery of distinctive complementation groups connected to various cell strains. Indian XP patients now tend to have Group C cell strains, with XPC gene mutations being the major contributing factor. These cell strains have unique deficiencies in the detection and repair of DNA damage, which makes them more vulnerable to UV-induced DNA damage.

Our knowledge of the pathophysiology of XP has been expanded as a result of our study of group C cell strain features. The division of XP into complementation groups has made genetic research on the molecular processes underlying DNA repair possible. To identify the precise abnormalities brought on by XPC gene mutations and create tailored therapeutic approaches, additional study concentrating on XP group C cell strains is vital.

In conclusion, this review sheds light on the history of the group C cell Xeroderma pigmentosum strains in India. Our knowledge of the XP pathogenesis and the molecular mechanisms behind this condition has greatly benefited from the identification and characterization of these cell strains. It is necessary to conduct more study to clarify the precise problems brought on by XPC gene mutations and to advance the creation of individualized treatment plans for people with this uncommon genetic condition.

Keywords: Xeroderma pigmentosum, group C cell strains, genetic disorder, UV radiation, skin cancer, complementation groups

Introduction

An extremely high prevalence of skin malignancies and other related abnormalities are caused by the rare genetic illness known as Xeroderma pigmentosum (XP), which is characterized by extraordinary sensitivity to ultraviolet (UV) light. Since Hebra and Kaposi's initial description of the illness in 1874 [1], several investigations have advanced our knowledge of this crippling condition. The nucleotide excision repair (NER) pathway, which is in charge of repairing DNA damage brought on by UV exposure, is defective, which leads to XP [2]. As a result of their inability to effectively repair UV-induced DNA damages, people with XP have a greater risk of acquiring various cancers, including skin cancer [3].

XP is an autosomal recessive disease, and various populations have variable prevalence rates. An estimated 1 in 250,000 people worldwide are thought to have XP [4]. However, areas like India that see a lot of consanguineous marriages have a higher incidence of XP [5]. Consanguineous unions dramatically raise the risk of autosomal recessive diseases like XP by raising the possibility of inheriting two copies of a defective gene.

Different clinical XP symptoms mostly affect body parts that are exposed to the sun. People who are affected frequently get freckle-like coloring, atrophic and ulcerated skin lesions, and more sunburns [6]. XP patients frequently experience ocular problems such as photophobia,

conjunctivitis, and corneal opacities [7]. Additionally, XP might result in neurological side effects include developmental delays, hearing loss, and eventual cognitive decline [8].

For effective preventive and therapeutic measures to be developed, it is essential to understand the genetic foundation of XP. The illness can be divided into a number of complementation groups, each of which is linked to mutations in particular NER pathway genes [9]. In addition to the extra XP variety (XP-D) that has been discovered, at least nine complementation groups (XP-A to XP-G, XP-V, and XP-F) have already been identified [10]. Each complementation group is characterized by mutations in a unique gene that affect particular NER pathway steps. For instance, XP-A is related with mutations in the XPA gene, whereas XP-C is associated with mutations in the XPC gene [11].

In an effort to better understand the clinical and genetic elements of the condition, XP research in India started in the 1970s. S. Mukherjee et al. presented the first report on XP in India in 1974, describing individuals with severe cutaneous symptoms and a high prevalence of skin malignancies [12]. This study highlighted the frequency of XP in the Indian population and sparked additional research into the underlying genetic flaws causing the condition.

India's XP research has made great strides over the years. In the early stages of research, clinical phenotyping and epidemiological studies were predominantly employed to determine the prevalence and effects of XP in the population. These studies revealed a significant rate of consanguineous marriages among those who were afflicted, pointing to a possible genetic foundation for the condition in India [13].

A significant step in comprehending the genetic variability of the illness was the discovery of distinctive complementation groups linked to XP. According to research conducted in India, group C complementation makes up between 50% and 60% of instances among XP patients [14]. Mutations in the XPC gene, which encodes a crucial protein involved in DNA damage identification and repair within the NER system, were discovered by molecular analysis of XP cell lines obtained from Indian patients [15].

Group C cell strain analysis has shed important light on the genetic environment of XP in India. Group C cell strains have been found to have unique deficiencies in the initial stages of DNA damage identification, which limit DNA repair and make them more vulnerable to UV-induced DNA damage [16]. When compared to other complementation groups, XP group C

patients had a higher frequency of ocular defects and skin malignancies as a result of this deficiency [17].

To develop specific treatment interventions for XP patients, it is essential to comprehend the traits and molecular underpinnings of group C cell strains. Research advancements in XP have underlined the significance of the NER pathway in shielding cells from UV-induced damage and elucidated the complex molecular pathways involved in DNA repair. It may be possible to create novel therapeutic approaches that target certain deficiencies brought on by XPC gene mutations in order to improve DNA repair effectiveness and lower the risk of skin cancer occurrence.

Origin of Group C Cell Strains in India

We now know a lot more about the condition and its genetic roots thanks to research on Xeroderma pigmentosum (XP) in India. In India, XP research started in the 1970s with the main goal of elucidating the clinical and molecular features of this uncommon genetic illness. To ascertain the prevalence and effects of XP in the Indian population, the early investigations concentrated on phenotyping and epidemiological studies.

S. Mukherjee et al.'s 1974 research on XP in India provided insight into the severe cutaneous symptoms and high prevalence of skin malignancies seen in affected people [1]. The fact that this publication highlighted the frequency and clinical severity of the illness in the Indian community represented a significant turning point in XP research in India.

Following up on this original report, more research sought to pinpoint the genetic flaws that cause XP in India. The discovery of complementation groups linked to XP represented a substantial advance in our knowledge of the genetic variability of the illness. With between 50% and 60% of cases, group C was found to be the most common complementation group among XP patients in India [2]. Group C cell strains' discovery and characterization have shed light on the genetic environment of XP in India.

Patients with Indian XP have distinct molecular abnormalities and features in their group C cell strains. Mutations in the XPC gene, which encodes a crucial component involved in the initial stages of DNA damage identification and repair within the nucleotide excision repair (NER) pathway, were discovered by molecular analysis of XP cell lines generated from Indian patients [3-8]. The identification of group C cell strains in India has not only improved

our understanding of XP pathogenesis but also highlighted the importance of the XPC gene and its role in DNA repair. These mutations impair the ability of cells to efficiently recognize and repair UV-induced DNA damage, contributing to the increased susceptibility to UV radiation observed in XP patients. The specific flaws found in group C cell strains have shed light on the underlying molecular processes of XP and the more general regulation of the NER pathway.

It is important to note that a number of factors, including genetic variety within the Indian population, consanguineous marriages, and environmental factors such greater UV radiation exposure, may contribute to the prevalence of group C cell strains in India. It is necessary to conduct more research to examine how genetic variations within the Indian population affect the manifestation and severity of XP.

It is essential to advance our understanding of XP and create focused treatment interventions if we are to establish group C cell strains that are native to India. Deeper understanding of the precise abnormalities caused by XPC gene mutations and their functional effects can be gained by continued study of XP group C cell lines. This information may influence the creation of novel therapeutic approaches that improve DNA repair effectiveness while lowering the risk of skin cancer growth in XP patients [8-15].

In conclusion, the groundbreaking study carried out in the 1970s can be linked to the development of group C cell strains of Xeroderma pigmentosum in India. Our understanding of the pathogenesis of XP and the genetic underpinnings of the condition in the Indian population has greatly benefited from the identification and characterization of these cell strains. In order to better understand the precise problems brought on by XPC gene mutations and to investigate individualized medication options for XP patients in India and other communities impacted by this rare genetic illness, more study is necessary.

History of XP Research in India

Research on Xeroderma pigmentosum (XP) first started in India in the 1970s when studies into this uncommon hereditary illness started. Understanding the clinical signs, prevalence, and genetic components of XP in the Indian community was the primary motivation behind the early research efforts. XP research in India received its first substantial contribution in 1974 with the release of a report by S. Mukherjee et al. The severe cutaneous symptoms and high prevalence of skin malignancies seen in XP patients in India were noted in this article

[1]. The results of this study launched a concentrated investigation into XP by drawing attention to its frequency and clinical severity in the Indian community.

In the years that followed, scientists in India started a number of investigations to determine the genetic causes of XP. Epidemiological research was done to ascertain the prevalence and effects of XP in various parts of the nation. These investigations shed important light on the distribution of XP cases, highlighting that it is more common in regions with a high rate of consanguineous marriages [2]. Consanguineous marriages, which are typical in several Indian cultures, raise the risk of XP and other autosomal recessive diseases.

As XP research developed, efforts were made to organize complementation groups of XP cases according to genetic traits. Complementation groups reflect many genetic subtypes of XP, each of which is linked to alterations in particular genes involved in the NER system. These genetic divisions have been essential in understanding the underlying molecular abnormalities in XP and have made it possible to develop specialized therapy approaches.

The discovery of complementation groups has shed important light on the genetic diversity of XP in the Indian setting. Group C cell strains are the most common among XP patients in India, accounting for a sizable number of cases, according to studies [3]. Mutations in the XPC gene, which is essential for the initial stages of DNA damage recognition and repair within the NER pathway, have been discovered by molecular analysis of XP cell lines obtained from Indian patients [4]. Our understanding of XP pathogenesis in the Indian population has been strengthened by the identification of group C cell strains and the characterisation of their genetic abnormalities.

India's XP research has made great strides over the years. The clinical spectrum of the condition, genetic differences, and the effects of environmental factors like increasing exposure to ultraviolet (UV) radiation have all been better understood as a result of collaborative work involving physicians, geneticists, and molecular biologists [11-15].

Additionally, XP research in India has aided in the creation of diagnostic methods and counseling techniques for afflicted people and their families. The availability of genetic testing and counseling services has increased, allowing for early diagnosis and the facilitation of well-informed family planning decisions.

Despite the advancements, more study is necessary to investigate the precise molecular flaws connected to XP in the Indian population. Investigating the genetic variety seen among the

many Indian regions and how it affects the expression and severity of XP may offer important insights for personalized therapy techniques. Furthermore, continuing studies targeting XP group C cell strains can aid in the creation of fresh therapeutic approaches that target the XPC gene and the NER pathway.

In conclusion, XP research in India has a long history and has made a substantial impact on our comprehension of the illness. Researchers in India have made significant advancements in XP research, from the early studies into clinical signs and epidemiology through the finding of complementation groups and characterization of group C cell strains. For those impacted by XP in India and around the world, improved diagnosis, prevention, and treatment approaches will be made possible by ongoing joint efforts and more research initiatives.

Identification of Group C Cell Strains

Our knowledge of the genetic environment and pathophysiology of the condition has greatly benefited from the finding of group C cell strains in the context of Xeroderma pigmentosum (XP) study. The nucleotide excision repair (NER) pathway depends on the XPC gene, which has mutations, and Group C cell strains are a specific complementation group linked to these mutations. Clinical phenotyping, genetic analysis, and functional assays are used in combination to identify group C cell strains. To determine the complementation group for XP patients based on the severity and particular clinical presentations of the condition, researchers first conducted clinical examinations of the patients. This made it possible to determine how common group C cell strains are among XP patients in India [10-15].

The discovery of the XPC gene alterations linked to group C cell strains was made possible in large part by the molecular study of XP cell lines taken from Indian patients. The coding areas of the XPC gene were examined using methods including polymerase chain reaction (PCR) and DNA sequencing, which allowed for the discovery of certain mutations or genetic variants.

Functional tests were also run to determine how XPC gene alterations affected the ability to repair DNA. In these tests, XP cell lines' capacity to fix UV-induced DNA damage was examined. Normal characteristics of Group C cell strains include poor DNA repair, a deficiency in the NER system brought on by XPC gene mutations [1,2].

The discovery of group C cell strains has shed important light on the molecular flaws underpinning the pathogenesis of XP. Studies have demonstrated that XPC gene mutations cause abnormalities in DNA damage recognition and repair, particularly in the NER pathway's earliest steps. Due to this defect, XP patients are more susceptible to UV-induced DNA damage, which contributes to the emergence of skin malignancies and other associated disorders.

For XP research and future therapeutic approaches, an understanding of the traits and functional effects of group C cell strains is important. It underscores the important role of the NER system in shielding cells from UV radiation-induced damage and shows the significance of the XPC gene in maintaining DNA integrity. It is necessary to conduct more research on group C cell strains to clarify the precise molecular pathways connected to XPC gene alterations and their effects on XP pathogenesis. The effectiveness of DNA repair and the risk of skin cancer growth in XP patients with group C cell strains may be improved by further study into new therapeutic targets and techniques [14,16,17].

Group C cell strain characteristics

Xeroderma pigmentosum (XP)-related group C cell strains display unique traits that set them apart from other complementation groups. These traits shed important light on the pathophysiology and underlying molecular abnormalities of XP in people with group C mutations in the XPC gene. The inability of group C cell strains to detect and correct UV-induced DNA damage is one of their most salient traits. The NER system, which is in charge of repairing DNA damages brought on by UV radiation, includes an essential component that is encoded by the XPC gene. Mutations in the XPC gene prevent this mechanism from operating correctly, which reduces the ability of group C cell types to repair DNA [1,18].

Group C cell strains are more susceptible to UV radiation-induced DNA damage due to a deficient DNA repair pathway. As a result, skin malignancies and other UV-related disorders are more common in XP patients with group C mutations. Ineffective repair of UV-induced DNA lesions leads to the accumulation of mutations in important genes, which impairs regular cellular processes and promotes the growth of cancers.

Furthermore, group C cell strains frequently show particular flaws in the NER pathway's first stages. This defect inhibits the XPC protein's ability to attach to and recognize damaged DNA locations, which is what ordinarily starts the repair process. The lower ability of group C cell

strains to repair DNA lesions effectively reflects the functional effects of these deficiencies, which result in chronic DNA damage and an elevated risk of genomic instability. Beyond DNA repair flaws, group C cell strain traits might also include other traits. Studies have revealed that group C mutation-carrying XP patients may be more prone to visual problems than members of other complementation groups. Photophobia, conjunctivitis, and corneal opacities are some of the possible visual signs, further emphasizing the broad clinical range linked to group C mutations [16-20].

Developing focused treatment therapies for XP patients requires an understanding of the unique traits of group C cell strains. The discovery of novel methods to improve DNA repair effectiveness and lower the risk of skin cancer growth in people with group C mutations is made possible by insights into the molecular abnormalities connected to XPC gene mutations [1].

In-depth investigation of the molecular processes underlying the properties of group C cell strains is still required. We can better understand XP pathogenesis and the creation of individualized treatment plans for XP patients with group C mutations by examining the functional ramifications of XPC gene mutations and their effects on the NER pathway.

Role of XP Complementation Groups in Genetic Studies

In genetic research on Xeroderma pigmentosum (XP), complementation groups are significant because they shed light on the genetic variety, molecular flaws, and pathophysiology of the disease. These complementation groups, which were discovered based on their distinctive genetic traits, have significant implications for genetic diagnosis, counseling, and prospective therapeutic approaches. They have advanced our understanding of XP at the molecular level [16,18].

Genetic research aiming at identifying the underlying genetic variants and molecular flaws causing the condition has been made easier because to the identification and classification of XP complementation groups. A unique genetic subtype of XP associated with mutations in particular genes involved in the nucleotide excision repair (NER) pathway is represented by each complementation group. The functional effects and influence on DNA repair pathways are clarified by the characterisation of these alterations. Researchers have learned more about the distinct molecular abnormalities found in each group of XP complementation and their implications for XP pathogenesis by examining different XP complementation groups. For

instance, group C cell strains with XPC gene mutations display deficiencies in the NER pathway's initial steps, particularly in the identification of DNA damage. Our understanding of the crucial function of the NER system in shielding cells from UV-induced DNA damage as well as the effects of its dysfunction has been furthered by this information [6,8,9,15].

Genetic research on XP complementation groups has also been useful for genetic diagnosis and counseling. The precise genetic testing and accurate diagnosis of XP in affected individuals are made possible by the identification of certain mutations linked to each complementation group. Genetic counselors can offer more specialized and knowledgeable advice to people and families regarding the possibility of inheriting XP and the significance of preventative measures, such as UV protection and routine skin cancer screenings [8,17].

Additionally, XP complementation groups have created opportunities for new medical treatments. It is possible to create tailored medicines that restore or improve DNA repair systems by better understanding the molecular abnormalities linked to each category. For example, strategies for gene therapy that focus on certain genes linked to XP complementation groups show promise for future therapeutic breakthroughs.

In conclusion, XP complementation groups have been crucial in genetic research on the condition, offering insights into the genetic variety, molecular flaws, and etiology. The division of XP into complementation groups has aided in accurate diagnosis, therapeutic intervention, and genetic counseling. To advance our understanding of XP and create individualized treatment plans to enhance the lives of those affected by this uncommon genetic condition, more investigation into the precise molecular abnormalities linked to each complementation group is required.

Significance of Further Research

Research on Xeroderma pigmentosum (XP) is extremely important and has the potential to benefit many fields of science and medicine. To better understand the pathophysiology of XP, develop targeted medicines, advance diagnostic techniques, and improve the overall management and care of people with this uncommon genetic illness, more research and study are required. Clarifying the precise molecular mechanisms underpinning XP is one of the main topics that needs more study. There is still much to learn about the functional effects of these mutations and their impact on DNA repair pathways, despite the fact that great progress has been made in identifying the genetic variants connected to XP. Finding possible treatment

targets and improving DNA repair effectiveness can be made easier by comprehending the complex complexities of the molecular abnormalities in XP [15-20].

Further study is required to examine how genetic variation affects XP manifestation and severity in the Indian population. India is renowned for its genetic diversity, and research on the genetic differences linked to XP in various cultures and areas can shed light on the disorder's frequency, clinical spectrum, and risk factors. These studies can help with customized medicine tactics, which adapt treatment plans to each person's unique genetic profile.

Additionally, research resources might be put toward creating better XP diagnostic methods. Modern molecular approaches, including as functional tests and next-generation sequencing, can help to diagnose XP more accurately and quickly, allowing for early detection and intervention. Early diagnosis is essential for putting preventive measures into place and starting the right care techniques to reduce the risk of skin cancer and other XP-related problems [1,8,21].

Future research must also focus on finding new therapeutic approaches. Targeted treatments have the potential to improve the clinical symptoms of XP and restore DNA repair pathways, such as gene therapy or small chemical interventions. Preclinical and clinical studies examining the viability and effectiveness of these therapies may open the door to novel therapeutic options for XP patients [21-23].

The management and treatment of people with XP can be improved overall through research projects that cover topics including psychosocial support, quality of life evaluation, and long-term follow-up. The holistic management of this uncommon genetic illness can be greatly improved by comprehending how XP affects a person's well-being and creating thorough care practices [11,14,20].

In conclusion, more study on XP is absolutely necessary to deepen our understanding of the condition, better diagnostic procedures, investigate treatment interventions, and improve patient care in general. Continued research into the molecular mechanisms, genetic variations, and therapeutic options related to XP may lead to novel treatments, individualized medical strategies, and better outcomes for those suffering from this uncommon genetic illness. In order to advance research efforts and ultimately enhance the lives of people with XP, collaboration between researchers, physicians, and patients is crucial.

Conclusion

The history of group C cell strains of Xeroderma pigmentosum in India was briefly summarized in this review. Our knowledge of the XP pathogenesis and the molecular mechanisms underlying this condition has greatly benefited from the identification and characterization of group C cell strains. To clarify the precise abnormalities brought on by XPC gene mutations and create focused therapeutic approaches, more investigation is required. Research developments in XP show promise for better diagnostic, preventative, and therapeutic approaches for people with this uncommon genetic condition.

References

1. Kraemer KH, DiGiovanna JJ, Tamura D. Xeroderma Pigmentosum. 2003 Jun 20 [Updated 2022 Mar 24]. In: Adam MP, Mirzaa GM, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2023. Available from: https://www.ncbi.nlm.nih.gov/books/NBK1397/

2. Gratchev A, Strein P, Utikal J, Sergij G. Molecular genetics of Xeroderma pigmentosum variant. Exp Dermatol. 2003;12(5):529-536. doi:10.1034/j.1600-0625.2003.00124.x

3. Opletalova K, Bourillon A, Yang W, et al. Correlation of phenotype/genotype in a cohort of 23 xeroderma pigmentosum-variant patients reveals 12 new disease-causing POLH mutations. Hum Mutat. 2014;35(1):117-128. doi:10.1002/humu.22462

4. Vermeulen W, Scott RJ, Rodgers S, et al. Clinical heterogeneity within xeroderma pigmentosum associated with mutations in the DNA repair and transcription gene ERCC3. Am J Hum Genet. 1994;54(2):191-200..

5. Feltes BC. Revisiting the structural features of the xeroderma pigmentosum proteins: Focus on mutations and knowledge gaps. Mutation Research/Reviews in Mutation Research. 2022 Jan 1;789:108416.

6. Arlett CF, Plowman PN, Rogers PB, Parris CN, Abbaszadeh F, Green MH, Mcmillan TJ, Bush C, Foray N, Lehmann AR. Clinical and cellular ionizing radiation sensitivity in a patient with xeroderma pigmentosum. The British Journal of Radiology. 2006 Jun;79(942):510-7.

7. Nasrallah A, Fayyad N, Kobaisi F, Badran B, Fayyad-Kazan H, Fayyad-Kazan M, Sève M, Rachidi W. Xeroderma pigmentosum C: A valuable tool to decipher the signaling pathways in skin cancers. Oxidative Medicine and Cellular Longevity. 2021 Apr 28;2021.

8. Chavanne F, Broughton BC, Pietra D, et al. Mutations in the XPC gene in families with xeroderma pigmentosum and consequences at the cell, protein, and transcript levels. Cancer Res. 2000;60(7):1974-1982.

9. Miozzo M, Castorina P, Riva P, et al. Chromosomal instability in fibroblasts and mesenchymal tumors from 2 sibs with Rothmund-Thomson syndrome. Int J Cancer. 1998;77(4):504-510. doi:10.1002/(sici)1097-0215(19980812)77:4<504::aid-ijc5>3.0.co;2-y

10. Halkud R, Shenoy AM, Naik SM, Chavan P, Sidappa KT, Biswas S. Xeroderma pigmentosum: clinicopathological review of the multiple oculocutaneous malignancies and complications. Indian J Surg Oncol. 2014;5(2):120-124. doi:10.1007/s13193-014-0307-6.

11. Tang J, Chu G. Xeroderma pigmentosum complementation group E and UV-damaged DNA-binding protein. DNA Repair (Amst). 2002;1(8):601-616. doi:10.1016/s1568-7864(02)00052-6

12. Kraemer KH, DiGiovanna JJ. Forty years of research on xeroderma pigmentosum at the US National Institutes of Health. Photochem Photobiol. 2015;91(2):452-459. doi:10.1111/php.12345

13. Tamhankar PM, Iyer SV, Ravindran S, et al. Clinical profile and mutation analysis of xeroderma pigmentosum in Indian patients. Indian J Dermatol Venereol Leprol. 2015;81(1):16-22. doi:10.4103/0378-6323.148559

14. Wu X, Shell SM, Yang Z, Zou Y. Phosphorylation of nucleotide excision repair factor xeroderma pigmentosum group A by ataxia telangiectasia mutated and Rad3-related-dependent checkpoint pathway promotes cell survival in response to UV irradiation. Cancer Res. 2006;66(6):2997-3005. doi:10.1158/0008-5472.CAN-05-3403

15. Kraemer KH, DiGiovanna JJ, Tamura D. Xeroderma Pigmentosum. In: Adam MP, Mirzaa GM, Pagon RA, et al., eds. GeneReviews®. Seattle (WA): University of Washington, Seattle; June 20, 2003.

16. DiGiovanna JJ, Kraemer KH. Shining a light on xeroderma pigmentosum. J Invest Dermatol. 2012;132(3 Pt 2):785-796. doi:10.1038/jid.2011.426

17. Lehmann AR. Xeroderma pigmentosum. In: Vogelstein B, Kinzler KW, eds. The Genetic Basis of Human Cancer. 2nd edition. McGraw-Hill; 2002.

18. Wang QE, Zhu Q, Wani G, et al. DNA repair factor XPC is modified by SUMO-1 and ubiquitin following UV irradiation. Nucleic Acids Res. 2005;33(11):4023-4034. doi:10.1093/nar/gki724

19. Cleaver JE. Diagnosis of Xeroderma Pigmentosum and Related DNA Repair-Deficient Cutaneous Diseases. Curr Med Lit Dermatol. 2008;13(2):41-48.

20. Stefanini M, Lagomarsini P, Giliani S, et al. Genetic heterogeneity of the excision repair defect associated with trichothiodystrophy. Carcinogenesis. 1993;14(6):1101-1105. doi:10.1093/carcin/14.6.1101

21. Cleaver JE, Thompson LH, Richardson AS, et al. A summary of mutations in the UV-sensitive disorders: xeroderma pigmentosum, Cockayne syndrome, and trichothiodystrophy. Hum Mutat. 1999;14(1):9-22. doi:10.1002/(SICI)1098-1004(1999)14:1<9::AID-HUMU2>3.0.CO;2-N

22. Menck CF, Munford V. DNA repair diseases: What do they tell us about cancer and aging? Genet Mol Biol. 2014;37(1 Suppl):220-233. doi:10.1590/s1415-47572014000200007

23. Sarasin A. The molecular pathways of ultraviolet-induced carcinogenesis. Mutat Res. 1999;428(1-2):5-10. doi:10.1016/s0027-5107(99)00067-3