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**ORIGINAL RESEARCH**

# Evaluation of cases of Xeroderma pigmentosum

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## ABSTRACT

**Background:** An high sensitivity to ultraviolet (UV) rays from sunlight is a characteristic of Xeroderma pigmentosum (XP), a rare hereditary condition that is inherited. The present study was conducted to evaluate cases of Xeroderma pigmentosum. **Materials & Methods:** 26 patients with Xeroderma pigmentosum (XP) of both genders were recruited for the study. Parameters such as neurological symptomsand ocular symptoms were recorded. **Results:** Out of 26 patients, 15 were males and 11 were females. Neurological symptoms such as deafness was seen in 7, mental retardation in 3, spasticity in 15 and microcephaly in 1 patient. Ocular symptomssuch as lid atrophy was seen in 7, photophobia seen in 11, keratitis in 16 patients. The difference was non- significant (P> 0.05). **Conclusion:** Screening for XPA gene variations should be prompted by neurological problems in Indian individuals with xeroderma pigmentosum. Rapid molecular diagnosis would help with decisive diagnosis, genetic counselling, and prenatal diagnostics.

**Keywords:** keratitis, photophobia, Xeroderma pigmentosum



## INTRODUCTION

An high sensitivity to ultraviolet (UV) rays from sunlight is a characteristic of Xeroderma pigmentosum (XP), a rare hereditary condition that is inherited. The skin and eyes are the main areas affected by this disorder, though in extreme situations, the neurological system may also be affected.[1] Mutations in the genes that repair UV-induced DNA damage are the cause of XP. The nucleotide excision repair (NER) pathway includes these genes. Because XP is autosomal recessive, a kid must inherit two copies of the faulty gene—one from each parent—in order to be affected.[2]

A rare inherited illness called Xeroderma pigmentosum (XP) is characterized by a high sensitivity to ultraviolet (UV) rays from sunshine.[3] Although the neurological system may also be impacted in severe cases, the disorder primarily affects the skin and eyes. XP is caused by mutations in the genes that repair UV-induced DNA damage. These genes are part of the nucleotide excision repair (NER) pathway.[4] A child must inherit two copies of the defective gene—one from each parent—in order to be impacted by XP because it is an autosomal recessive condition.[5,6]Symptoms included extreme sun sensitivity, early and excessive freckling on sun-exposed areas, dryness and scaling, high risk of developing skin cancers (basal cell carcinoma, squamous cell carcinoma, and melanoma) at a young age, sensitivity to light, chronic inflammation of the conjunctiva, inflammation of the cornea, increased risk of cancers on the surface of the eyes, progressive neurological degeneration in some patients, leading to hearing loss, poor coordination, intellectual decline, and seizures.[7]The present study was conducted to evaluate cases of Xeroderma pigmentosum.

## MATERIALS & METHODS

The present study was conducted on 26 patients with Xeroderma pigmentosum (XP) of both genders. All patients were informed regarding the study and their written consent was obtained.

Data such as name, age, gender etc. was recorded. Parameters such as ocular symptoms and neurological symptoms were recorded. Data thus obtained were subjected to statistical analysis. P value < 0.05 was considered significant.

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## RESULTS

**Table I Distribution of patients**

| Total- 26 | | |
|---|---|---|
| **Gender** | **Males** | **Females** |
| Number | 15 | 11 |

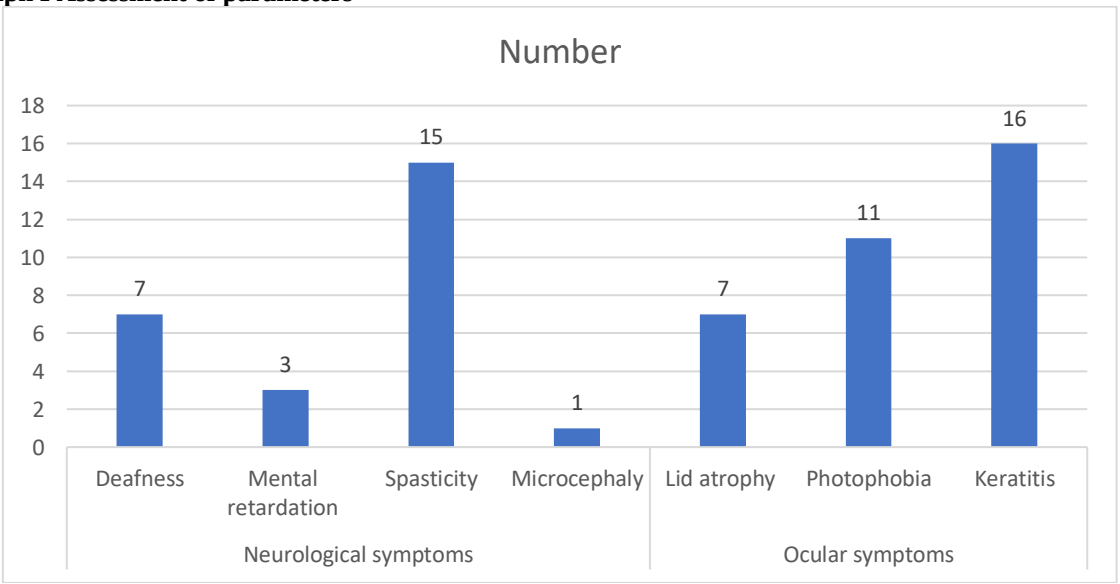
Table I shows that out of 26 patients, 15 were males and 11 were females.

**Table II Assessment of parameters**

| Parameters | Variables | Number | P value |
|---|---|---|---|
| Neurological symptoms | Deafness | 7 | 0.05 |
| | Mental retardation | 3 | |
| | Spasticity | 15 | |
| | Microcephaly | 1 | |
| Ocular symptoms | Lid atrophy | 7 | 0.82 |
| | Photophobia | 11 | |
| | Keratitis | 16 | |

Table II, graph I shows that neurological symptoms such asdeafness was seen in 7, mental retardation in 3, spasticity in 15 andmicrocephalyin 1 patient. Ocular symptoms such as lid atrophywas seen in 7, photophobia seen in 11, keratitis in 16 patients. The difference was non- significant (P> 0.05).

**Graph I Assessment of parameters**




## DISCUSSION

Xeroderma pigmentosum/Cockayne syndrome (XP/CS) can be distinguished from XP by its physical features, such as sunken eyes, thinning skin and hair, and a stooped posture when standing. Ataxia, cataracts, and pigmentary retinopathy are possible conditions in these people. Separating XP from Cerebrooculofacial Syndrome (COFS) is also essential.[8,9] Microcephaly with intracranial calcifications, microcornea, cataracts, optic atrophy, and congenital joint contractures are the hallmarks of COFS. However, XP and COFS share some traits, including telangiectasia, poikiloderma, atrophy, and xerosis. People with XP are significantly more likely to get skin cancer early in life.[10]Early diagnosis and stringent sun protection measures can help manage and reduce this risk.Varies widely depending on the severity of the condition and the effectiveness of preventive measures. Early and continuous protection from UV radiation is crucial for improving quality of life and lifespan.[11]The present study was conducted to evaluate cases of Xeroderma pigmentosum.

We found that out of 26 patients, 15 were males and 11 were females. Kleijer et al[12] in their study laboratory diagnosis for DNA repair diseases was performed in western Europe from the early seventies for xeroderma pigmentosum (XP) and from the mid-eighties for Cockayne syndrome (CS) and trichothiodystrophy (TTD). The combined data from the DNA repair diagnostic centres in France, (West) Germany, Italy, the Netherlands and the United Kingdom have been investigated for three groups of diseases: XP (including XP-variant), CS (including XP/CS complex) and TTD. Incidences in western Europe were for the first time established at 2.3 per million livebirths for XP, 2.7 per million for CS and 1.2 per million for TTD. As immigrant populations were disproportionately represented in the patients'

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groups, incidences were also established for the autochthonic western European population at: 0.9 per million for XP, 1.8 per million for CS and 1.1 per million for TTD. Perhaps contrary to general conceptions, compared to XP the incidence of CS appears to be somewhat higher and the incidence of TTD to be quite similar in the native West-European population.

We observed that neurological symptoms such asdeafness was seen in 7, mental retardation in 3, spasticity in 15 and microcephaly in 1 patient. Ocular symptomssuch as lid atrophy was seen in 7, photophobia seen in 11, keratitis in 16 patients. 86 XP patients from 66 unrelated families—the bulk of whom were consanguineous and from the Maghrebwere studied by Soufir et al[13] for the function of XP genes. 44 probands were subjected to sequencing analysis either directly or following the identification of the XP gene in 22 families by a complementation experiment. XPC and XPA mutations were found in 56/66 and 8/66 probands, respectively. Remarkably, the homozygous frameshift mutation c.1643_1644delTG (p.Val548AlafsX25) was present in 87% of XP-C patients. According to haplotype research, this mutation has a widespread founder impact in the Mediterranean region and is thought to be 50 generations old, or 1,250 years old. In 7/8 XP-A patients, they found the previously reported nonsense homozygous XPA mutation (p.Arg228X). Six mutations were also discovered, five in XPC and one inXPA—that had, as far as we know, never before been reported.

The clinical characteristics of xeroderma pigmentosum patients were examined by Tamhakar et al.[14] Thirteen XP patients in ten households. Patients with moderate to severe mental impairment (6/10 families) had homozygous mutations in the XPA gene, although those without neurological symptoms did not. It was discovered that two unrelated families from the same community in Maharashtra, sharing a shared family name, had the same XPA gene mutation, c.335_338delTTATinsCATAAGAAA (p.F112SfsX2). Following XPC gene testing in two families with four impacted children, the unique mutations c.1243C>T or p.R415X and c.1677C>A or p.Y559X were discovered. Mutations in the XPA, XPB, and XPC genes were not found in two families.

The limitation of the study is small sample size.

## CONCLUSION

Authors found that screening for XPA gene variations should be prompted by neurological problems in Indian individuals with xeroderma pigmentosum. Rapid molecular diagnosis would help with decisive diagnosis, genetic counselling, and prenatal diagnostics.

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