**XERODERMA PIGMENTOSA-A RARE CLINICAL ENTITY**

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**ABSTRACT**

A rare case of dermatological Anomaly that came across in outpatient department of skin-.Namely – Xeroderma Pigmentosa has been reported. The case was recorded in outpatient Department of Dermatology at teaching Hospitals of Bangalore during the year 2007 to 2010. About 257 cases were screened in Out Patient Department of Skin in district of Bangalore –A case of Xeroderma pigmentosa was examined in 9 years old boy in detail with proper history of boy’s mother during her antenatal period along with history of drug intake during her first trimester., Exposure to Radiation, similar episodes in the past and Systemic diseases. The case of Xeroderma Pigmentosa in 9 years old boy was examined in the outpatient Department of SKIN at Teaching Hospital of Bangalore district, Bangalore. There was history of bilateral diminution of vision in both eyes. On examination, he was found to have dense pigmentation in upper half of the body and less density of pigmentation in his lower extremities associated with involvement of eye-- conjunctivitis and keratitis, since birth. He was the only son to his consanguineous parents. This is case of a very rare clinically entity, due to genetic disorder causing skin and ocular lesions due to sensitivity of light, resulting in high rate of morbidity. So, complete awareness and knowledge of these diseases should be given to the patients and as well as to the society by the health workers & NGOs in order to prevent further incidences in the family and to avoid consanguineous marriages. Due to its profound genetic importance, it has been studied and reported.

**Key Words:** Xeroderma Pigmentosa, Founder Effect, Global Genome Repair, Transcription Coupled Repair, Nucleotide Excision Repair, Consanguinity, Cutaneous, Ocular Lesions

**INTRODUCTION**

Xeroderma Pigmentosa is a rare genetic disorder of prototypical Nucleotide Excision Repair (NER). This rare genetic disorder occurs in the selected populations. It occurs very commonly in population of five countries of Northern Africa Continent known as MAGHREB. It may be the results of common ancestors in the particular which is referred to as Founder effect. NER normally eliminates lesions of DNA – distorting lesions due to ultraviolet induced photoproducts. There are two types of Nucleotide Excision Repair, namely Global Genome Repair (GGR) & Transcription –coupled repair (TCR) – GGR looks for regions of the genome while transcriptions coupled repairs eliminates lesions caused by the blocking of RNA polymerase (Hoeijmakers-2009).

There are eight subtypes xeroderma pigmentosa. All these subtypes do have defective nucleotide excision repair(NER)The eight type do have normal NER but show synthesis of aberrant translesions Hence these show varying degrees of photosensitivity followed by abnormal pigmentation and carcinoma of skin (Kraemer et al., 1987). There are occurrence of abnormalities in neurological, growth, followed by abnormalities of eyes that differs in the 8 subtypes which are termed Complementation groups (Gregory and Kirsner, 2010) 8th gene defect do not affect NER (Alan et al., 2011).

**CASES**

A 9 years old boy came to outpatient Department of SKIN at Teaching Hospital of Rajarajeswari Medical College, Bangalore with history of diminution of vision in both eyes and pigmentation all over the body, since 6 months. He was only child born to his consanguineous parents. There was family history of similar episode in the past but exact details not known. There was no history of drug intake.
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There was no history of systemic diseases like neither hypertension nor diabetes. Boy was from low socio economic status.

Figure: A boy Showing Signs of Xeroderma Pigmentosa

On General Examination: He was under built and under nourished. No cyanosis, no clubbing, no pedal oedema nor lymphadenopathy. Skin showed generalised pigmentation around his neck and on face deeply pigmented with involvement of eyes. On other parts of the body, there were sparse distribution of pigments seen on his extremities.

On Clinical Examination: His cardiovascular system, Respiratory system, gastrointestinal system and Spine vertebra Extremities were all normal within limits. No other anomalies were observed on his body. Examination of eyes: Both eyes were affected. Both anterior and posterior were normal. Conjunctivitis was congested. He had Photophobia, and conjunctivitis with severe irritation of eyes with watering. He had highly irritable blood shot and clouded eyes but not associated with malignant or non malignant growths. There was Narrow Palpebral fissure, Pigmented conjunctiva, No papilloedema. He had sensitive to light and diminution of vision in both eyes. There was no neurological involvement.

DISCUSSION

It is congenital disorder highly sensitivity to sunlight causing sun burn skin pigmentation changes associated with high incidence of carcinoma of skin (Alan et al., 2011).

INCIDENCE: This rare clinical anomaly is found in all continents and races (Gregory and Robert, 2010). There is high rate of consanguinity in North Africa and middle East, hence the high rate of incidence of
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Xeroderma Pigmentos. Occurs in the regions of North Africa called Maghreb (Hoeijmakers, 2009) 1 in 250,000 in USA (Robbins et al., 1974) 1 IN 20,000 in Japan (Hirai et al., 2006), 2.3 per millions live birth in Western European countries (Kleijer et al., 2008).

Sensitivity to light is the first sign in 60% of cases in cases of Xeroderma Pigmentosa (Bradford et al., 2011). While in 40% they do not show any sunburn reaction (Alan et al., 2011). To start with, there is freckle like pigmentation in sun exposed areas known as lentigines commonly seen on the nose, zygomatic region, forehead, later appear on the side of the cervical region of Photophobia is commonly present Pigmented seborrhoeic warts occur and . skin becomes dry rough and atrophic. Small hypopigmented macules are also present Telangiectasia are also found as late feature. Ocular lesions are also as common as dermatological lesions. They involve lid, cornea, conjunctiva and eye lids which are ultraviolet exposed structures. They cause photophobia, severe keratitis, corneal opacity, and vasculizations. They also cause neoplasms like epithelioma, melanoma, squamous cell carcinoma (Stefanani and Kraemer, 2008 and Ramkumar et al., 2011). Patients with Xeroderma Pigmentosa are more prone to develop carcinoma of oral cavity – Squamous cell carcinoma, especially at the tip of the tongue which is the region of Sun exposed area.

Progress of the Disease: Xeroderma Pigmentosa has 3 stages, namely

First Stage: Develops at very early age of 6 months after birth. To start with it face with reddening of face followed by scaling, freckling, and irregular dark spots begins to appear more on the, neck, lower part of the lower limb and trunk.

Second Stage: This stage is marked by the development of Poikiloderma where there are appearance irregular patches, both light and dark. Associated with spider web like collection of blood spots. There is thinning of skin through which blood vessels can be seen.

Third Stage: This is a stage of development of solar keratoses and development of carcinomas. This development takes place at an early age of 4-5 years. This occurs in exposed part of the body like face. The common malignancies of skin that occurs are Squamous cell carcinoma, or Basal cell carcinoma or melanoma. The other complications that develop are ophthalmological & neurological complications. Ophthalmological complications: In 80% patients develop painful sensitive to SUN –Photophobia, followed by conjunctivitis. Highly irritability blood shot and clouded eyes associated with malignant and non malignant growths (Derm Net NZ).

A rare case of xeroderma pigmentosum was associated with desquamative gingivitis has been reported (Hasan and Khan, 2011) Eight X-P gene defect do not affect Nucleotide Excision Repair (NER) but it is XP variants that creates problems in the replicating DNA that contains ultra violet induced damage (Lehmann et al., 1975) DNA Polymerases will carry out DNA Replications. This DNA polymerase n is used by the cells encoded by gene POLH for U-V damage. Mutation takes place in this gene in XP-V patients. Like XP-C, and XP-E, XP-V (Masutani et al., 1999). Patients rarely suffer from sun burns and neurological complications (Alan et al., 2011). As the frequency of mutation increases, it results in pigmentation of skin and development of skin cancers. In the majority of tumours there is P53 mutations which is characteristic of ultraviolet exposure (Daya-Grosjean and Sarasin, 2005).

PRESENT STUDY: A boy aged 9 years was examined at teaching Hospital of, Bangalore district, Bangalore for pigmentation of skin over the chest and over the limbs associated with involvement of eyes with history of diminution of vision as well as photophobia, conjunctivitis and keratitis. There was high irritation of eyes with blood clouded. There was neither malignant growth nor any growth from the tongue, nor from the oral cavity. Gingiva was normal. There was no papilloedma but narrow palpebral fissure in both eyes. Pigmentation over the other part of the body was less dense. He was only son born to his consangunoueous parents. There was family history of similar episode. He was from low socioeconomic family. There was no drug intake history. Only there was involvement of skin and eyes but no neurological involvement and other systems. So this anomaly may be due to Consanguinity and familial.
CONCLUSION
This case of Xerodema pigmentosa is extremely rare and incurable anomaly so awareness of the disease, followed by early diagnosis is very essential. Well protection from the Sunlight with careful management will improve the quality of life and life span of the individual. Patients with Xeroderma pigmentosa should be shown to multidisciplinary clinics, they are dermatologists, ophthalmologists, neurologists, psychologists, genetists and nursing specialities, hence it has been studied and reported.

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