ORIGINAL RESEARCH PAPER

Dermatology

XERODERMA PIGMENTOSUM GROUP F - A RARE CASE REPORT

KEY WORDS: Xeroderma pigmentosum,Group F

Dr Sunjanaa Dhepa R L*

Assistant Professor,Department of Dermatology venereology & leprosy, Vinayaka Missions Medical College & Hospitals,Karaikal- 609609
*Corresponding Author

Dr Chadha Kirandeepkaur Ajitsingh

Junior Resident,Department of Dermatology venereology & leprosy, Vinayaka Missions Medical College & Hospitals,Karaikal- 609609

ABSTRACT

Xeroderma pigmentosum(XP) is a rare autosomal recessive disorder of DNA repair characterised by progressive pigmentary changes, an increased risk of ultraviolet radiation induced carcinomas, severe photosensitivity and neurodegeneration.It is divided into eight complementation group and the clnical features of each complementation group varies according to the affected DNA repair gene. We hereby report a case of xeroderma pigmentosum group F which was proven by histopathological examination and the group confirmed by analysis of DNA, extracted from blood sample and this case is being reported for its rarity.

INTRODUCTION:

The human genome is made up of billions of DNA base repairs containing around 30000 protein encoding genes. Defects in these DNA repair pathways may result in many disorders which is usually characterised by cutaneous involvement, carcinoma and premature ageing. .In 1972 it was found that the fusion of fibroblasts from different XP patients forms heterokaryons that helps in correcting the defect in DNA repair suggesting that one defect can be corrected by the fusion of cells from a patient with different defect due to the availability of the protein which the other is lacking thus leading to the characterization of XP into different complementation groups.Not many cases belonging to the complementation group F was reported till date.Hence we would like to present this case for its rarity

CASE REPORT:

A 34 year old female presented to our outpatient department with multiple dark coloured skin lesions all over body since 20 year which was insidious in onset and initially these lesions started on the face which then progressed to the trunk followed by bilateral upper limbs and lower limbs and there is history of aggrevation on exposure to sunlight.These lesions are progressive in nature and is also associated with itching and burning sensation on exposure to sunlight thus a positive history of photosensitivity has been elicited. Patient does not give any history of photophobia. Patient does not give any history suggestive of neurological abnormalities. Similar illness present for her family members.Cutaneous examination revealed multiple freckles over face , trunk , upperlimb and minimal freckling present over bilateral lowerlimbs associated with atrophic hypopigmented macules giving rise to a mottled appearance.Hair , nail and mucosa does not reveal any abnormality.Systemic examination did not reveal any abnormality.Routine haematological ,biochemical and radiological investigations were within normal limits.Histopathological examination showed flattening of the epidermis with distinctive irregular proliferation of rete ridges presence of melanin pigments over the basal layer and the group is confirmed by the analysis of DNA extracted from blood sample.The patient was managed in a multidiscipillinary approach, photoprotection was advised with vitamin D supplementation and the nature of the disease explained to the patient and was asked to come for regular followup.

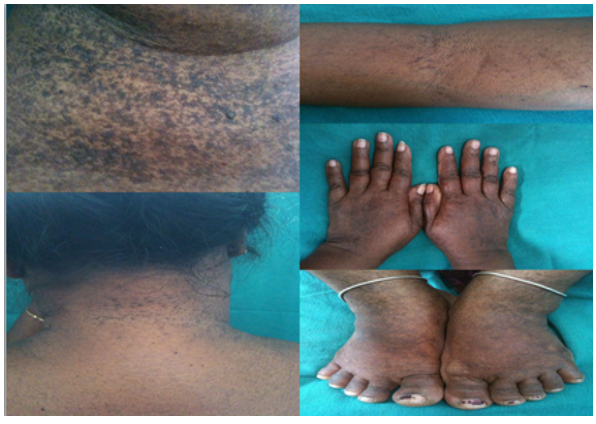

Figure 1. freckles with atrophic hypopigmented macules showing mottled appearance

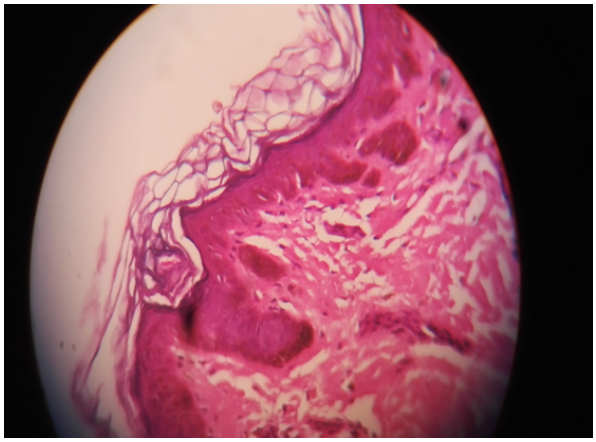

Figure 2: flattening of the epidermis with distinctive irregular proliferation of rete ridges and presence of melanin pigments over the basal layer.

DISCUSSION

Xeroderma pigmentosum is a rare disorder of DNA repair, which presents clinically with progressive pigmentary abnormalities, and an increased incidence of UVR induced carcinomas at sun exposed sites.[1] Xeroderma pigmentosum (XP) was described in Vienna by a Hungarian professor of dermatology Moriz Kaposi in 1870 .The disorder was first called "xeroderma or parchment skin" and in 1882, the term

"pigmentosum" was added to emphasize the striking pigmentary abnormality and is usually found in all continents and racial groups with an incidence of 2.3 per million livebirth. Among the complementation groups, XP A and C is most commonly seen.[2] Mutations in gene ERCC4 results in defect in nucleotide excision repair pathway both glogal genome repair and transcription coupled repair resulting clinical manifestations like photosensitivity, minimal skin changes like freckles, lentigines and hypopigmented macules and no neurological to severe neurological abnormalities[3]Mutations in ERCC4 genes have also been reported in association with Xeroderma pigmentosum/ Cockayne syndrome complex. Confirmation of the disease is mainly by histo pathological examination and by analysis of DNA. There is no cure for XP but if there is no neurological abnormality then with good photo protection, vitamin D supplementation, genetic counselling and with regular followup the prognosis is good.[4]

REFERENCES

1. Fassihi H. Spotlight on 'xeroderma pigmentosum'. Photochem Photobiol Sci 2013;12:78–84.
2. Rooks textbook of Dermatology ninth edition.
3. Lehmann AR, McGibbon D, Stefanini M. Xeroderma pigmentosum. Orphanet J Rare Dis 2011;6:70.
4. Bradford PT, Goldstein AM, Tamura D, et al. Cancer and neurologic degeneration in xeroderma pigmentosum: long term follow up characterises the role of DNA repair. J Med Genet 2010;48:168–76.