XERODERMA PIGMENTOSA (WITH REVIEW OF LITERATURE)

By

* D. M. SHAH & M. S. SHAH, ** B. H. SHAH

A pigmented pre-cancerous condition, xeroderma pigmentosa, though an uncommon entity in pigmented race, it is not rare in Indians. Quite a few Indian authors have reported this entity in past (Reddy et al 1958, Khanna et al 1965, Kunwar and Kumar 1967 and Saxena et al in 1968). It has been also reported in Negroes (Kling and Hamilton 1940).

Here with, we add one more family to the above said disease. Patients reported here are in 3rd or 4th decade of their life and have so far been lucky enough not to have any clinical evidence of mitotic transformation of any existing skin lesions.

CASE REPORT

A 35 years old female was first seen in out-patient clinic in March 1968 with the complaints of multiple dark brown spots all over the body since childhood.

Patient is a teetotaler with normal bowel and urinary habits. Her menstrual cycles were regular, 4-5/30 days moderate in amount and painless.

She is married for the last 17 years with the history of four full term normal deliveries. All the four children are healthy and none of them is having any manifestation of this illness. On routine enquiry it was stated that her two brothers are also suffering from similar spots. Later, her entire family was studied and the pedigree study is shown in chart No. 1.

The letters A & B in chart No. 1 personify step brother having same father and different mothers.

On examination, the entire skin was rough and showed mnay ill defined macules and papule of pigmentation. Lesions were more marked on exposed areas, picuture 1. The pigmentation consisted of dark brown maculo-papules of different size and configuration. Picture 2.

Ocular manifestations were not detected. There was no clinical evidence of any malignancy in any of the lesions.

Investigations

Hemogram and urinanalysis were normal. Blood Kahn – negative.

Skin biopsy confirmed the diagnosis of xeroderma pigmentosa revealing, marked hyperkeratiosis and irregular acanthosis with hyperpigmentation in basal layer of epidermis. In areas there was atrophy in epidermis and lymphocytic infiltration of upper dermis. Picture 3.

All the three members were instructed not to expose themselves to sunlight and were advised to use sun protective cream against sunlight. They have also been

* Registrar in Shin & V. D. Department, New Civil Hospital, Ahmedabd

** Hon Assistant (Sr.) skin and V. D. Department, New Civil Hospital, Ahmedabad.

Received for publication on 14-3-1969.

warned to report as soon as they find any rapid change in their existing lesions on skin.

Discussion

Xeroderma pigmentosa is a disease first described by Kaposi in 1870, after whom it was later called "Dermatosis Kaposi". It was also known as 'Angioma pigmentosa' and 'Melanosis lenticulosis progressiva'.

Anderson in 1961 described it as a precancerous familial condition charactrized by areas of atrophy as well as isolated and coalescent scaly patches of keratosis with pigmentation. In our cases, looking at the pedigree study, it is obvious that the disease is partly familial rather than heriditary, as there is no manifestation of the disease in the previous generation.

Boyd in 1953 described this disease in the following clinical stages :

(i) Stage of erythema due to sunburn, giving rise to,
(ii) Stage of pigmentation. These patches then become rough with warty eruptions giving rise to keratosis followed by,
(iii) Stage of atrophy, Some of these eruptions become malignant leading to the,
(iv) Stage of malignancy.

Pillsbury et al (1956) stated about the nature of malignancy in xeroderma pigmentosa. Mostly the basal cell carcinoma is the common sequelae, but some times tumor may turn in to sqaumous-cell carcinoma. Melanoma and sarcoma have also been described in keratosed skin. Lack et al (1960) also reported remote manifestations in lungs, upper lid and musculature of breast also. In present series none of them showed any cancerous manifestation.

The etiology of the disease is not clear. It is believed to be caused by congenital hypersensitiveness to the action of sunlight. It is supposed to be due to incomplete sex-linked recessive gene. Consanguinity is quite frequent. The cases reported here suggest familial nature of disease rather than heriditary as stated by Anderson (1961). Moreover no family history of consanguinity could be obtained in the pedigree.

In literature, it is described that 30% of cases of xeroderma pigmentosa have ocular manifestations. Khanna et al in 1965 also reportetat ocular manifestations in the four cases. In our study only one patient had watering from eyes and erythema of lower eyelids. Other two patients had no ocular manifestation.

It has been also reported that xeroderma pigmentosa patients have AB blood group to the extent of 40%. In our series we could perform blood grouping in only two members of the pedigree. Only one person has blood group 'AB'.

The disease is fatal before the end of second decade (Gougerot 1927), though cases have been reported to survive upto 40 years or even 70 years as reported by Herhxeimer in 1947. In present series all the three members of the family were in third or 4th decades of life with no malignant transformation.

Summary

Three members of a family with xeroderma pigmentosa are reported here. They all were above the age of 20 years with no evidence of malignancy.

Acknowledgement

We are thankful to the Dean, B. J. Medical College and Superintendent, Civil Hospital, Ahmedabad, for permitting us to report this case.

REFERENCES

1. Anderson, W. A. D.: Pathology 4th Ed., 1961. Mosby, St. Louis. P. 1181.
2. Boyd: A text book of pathology. 7th Ed. 1963. P. 409. 1320.
3. Anderson & Begg. Quoted Gougerot. H. (1967) in Text book of pathology 2nd Ed. 1950.
4. Anderson & Begg. Quoted Herhxeimer (1947) in Text book of pathology 2nd Ed. 1950.
5. Khanna, V. N. Garg K. C. and Bisaria K. K.: J. Ind. Med. Ass. 45:550, 1965.
6. K. B. Kunwar: J. I. M. A. 48:273, 1967.
7. King, H. and Hamilton L. M.: Arch. of Dermt. & Syph: 42 570, 1940.
8. K. N. Saxena, P. K. Wahl and V. K. Srivastava: Ind. J. of Derm. & Ven. 34:42, 1968.
9. Reddy, D. J.; Kameswara Rao, Y: Krishna Gupta K. & Sobhanadri, C. J.: J.I.M.A. 31:433, 1958.
10. Sevel, D.: Brit. J. of Opthalmology, 47:637, 1963.
11. K. N. Saxena, P. K. Wahl and V. K. Sriwastawa Quoted LACK. H, and Handerek L. in Ind. Jr. of Dermt & Ven. 34:42, 1968.
12. Behl P. N. and Sachadev. Ind. Jr. of Dermt. & Vene. 23:68, 1957.
13. Kaposi M. Pathology and treatment of disease of skin Balliere, Tindall & Cox, London 1875.
14. Ormbsy and Montegomery Quoted Gains in Diseases of Skin 8th Ed. 1954.
15. Ormbsy and Montegomery Quoted Y. C. White in Deseases of skin 8th Ed. 1954.
16. Loewnthol. British Jr, Dermotology 1.66, 1968.

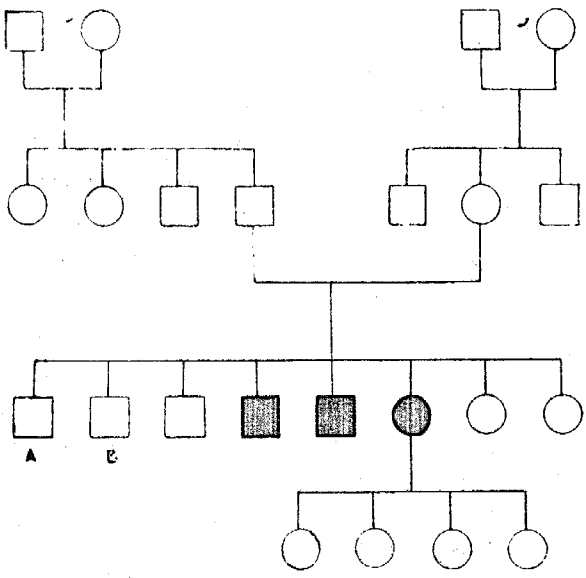

CHART NO. 1