ROTHMUND - THOMSON SYNDROME
(Report of a case in an Indian male)

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Summary

A case of Rothmund-Thomson Syndrome in an 8 year old Indian male is reported. The patient had bilaterally symmetrical superficial pigmentary deposition of thin nebulae in the lower one-third of the cornea and conjunctiva. There were no abnormalities of the lens. Urinary chromatography revealed a dibasic aminoaciduria with increased excretion of arginine, lysine, homocysteine and glycine.

Other features of interest in this patient were onset at the age of 8 days and start of the disease on the hands and feet rather than face. The patient also developed repeated ulcerations on the hands and feet subsequent to minor trauma.

The Rothmund - Thomson syndrome or poikiloderma congenitale, is a rare developmental syndrome probably determined by an autosomal recessive gene. It is twice as common in females as compared to males[1].

The most characteristic feature of the syndrome is the poikilodermatous appearance of the skin which is maximally present on the light exposed areas. Bilateral cataracts are seen in about 40% of cases and they usually develop between the age of 4 to 7 years[1]. More recently, other eye changes have been reported such as keratoconus, prominence of Schwalbe's line, absence of the anterior mesodermal layer of the iris and the presence of tilted optic discs[2]. Other authors have even noted tapeto-retinal degeneration in one patient[3].

We are reporting the case of an 8 year old Indian boy who had Rothmund-Thomson syndrome. He had associated bilaterally symmetrical pigmentary deposits in the cornea and conjunctiva. The patient also had a dibasic aminoaciduria.

Case Report

An 8 year old boy was brought to the Dermatology clinic of our hospital on 18th June 1977. His complaints were dryness and pigmentation of the skin and repeated ulcerations mainly in the hands and feet following minor trauma.

The patient was quite normal at birth. At the age of 8 days lesions first appeared on the hands and feet and then gradually spread to involve other areas of the body. There was no history of aggravation of symptoms on sunexposure, nor was there any history of

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Received for publication on 24—6—1978.

developing bullous lesions at sites of trauma. The patient did not have any systemic or visual complaints. There was no history of consanguinity in the parents. The patient has one brother and one sister both of whom are alive and well. Another brother, who also had a similar disease as the patient, died at the age of 1 year due to some unknown cause. The patient's maternal great uncle developed a similar disease 15 years ago and is alive at present.

General physical and systemic examination of the patient did not reveal any abnormality. The patient was of normal stature and intelligence. Cutaneous examination revealed generalised dryness and atrophy of the skin, mottled hyperpigmentation and depigmentation, and also some telangicetases. These changes were slightly more pronounced on the face (Fig. 1). The patient also had ulcers on the dorsal aspect of the feet. There were no warty hyperkeratoses seen anywhere on the skin.

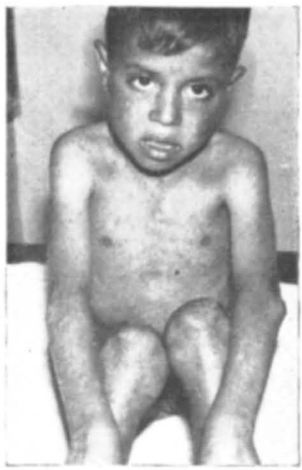

Fig 1 There is generalized dryness and atrophy of the skin. Mottled hyperpigmentation and depigmentation and telangicetases are also seen. These changes are only slightly more pronounced on the sun exposed areas.

There was no abnormality of the nails, scalp hair or oral and genital mucosa. Few of the patient's teeth were carious.

Ophthalmologic Examination

The lower one third of the cornea showed very superficial pigmentary deposition with thin nebulae. The same process was extending medially and laterally on to the conjunctiva and the changes were bilaterally symmetrical. The iris and lens were normal. These findings were interpreted as probably representing a degenerative change of the cornea and conjunctiva.

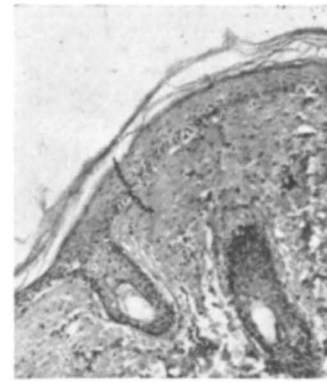

Fig. 2 There is hyperkeratosis and epidermal atrophy and liquefaction degeneration of the basal cell layer (H & E x 100)

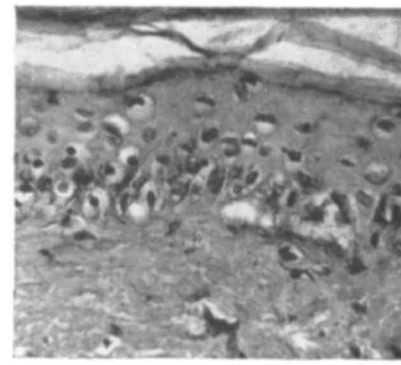

Fig 3 The liquefaction degeneration of the basal cell layer is well shown. There are "sun burn" type of cells with pyknotic nuclei which are present in the stratum spinosum (H & E x 400)

Investigations

1. Skin biopsy from the left forearm showed features compatible with Rothmund - Thomson syndrome. (Figs. 2 & 3)

2. Biochemistry of urine.
   (a) Urinary porphyrins and porphobilinogen were absent.
   (b) Urine test for phenylketonuria was negative.
   (c) Urine chromatography revealed a dibasic aminoaciduria. There was increased excretion of arginine, lysine, homocysteine and glycine.

Discussion

This case of Rothmund - Thomson syndrome presented several interesting features. The patient was an Indian male, 8 years of age. Only one case has been reported so far in a non-white patient[1]. Also, this disease is twice as common in females as compared to males[1].

The onset of the disease in our patient at the age of 8 days was very early compared to the usually reported age of three to six months. The disease usually starts on the face[4]. In our patient the lesions first appeared on the dorsa of hands and feet. He had no history of photosensitivity and on examination there was noticeable involvement of the covered areas also. In our patient, the skin changes were only slightly more pronounced on the sun-exposed areas as compared to the covered areas.

Biochemical examination of the patient's urine revealed a significant aminoaciduria. There was increased excretion of four amino acids, arginine, lysine, homocysteine and glycine. Our patient, however, did not have any increassd excretion of porphyrins in the urine nor was there any evidence of phenylketonuria[1].

The patient had increased vulnerability to trauma, giving rise to ulcers on the hands and feet.

One of the most characteristic features in this patient were the eye changes which were asymptomatic. There was bilaterally symmetrical pigmentary deposition of thin superficial nebulae on the lower one third of the cornea and conjunctiva. These findings were interpreted as probably representing a degenerative change of the cornea and conjunctiva. Corneal dystrophies have occurred in association with Rothmund-Thomson syndrome[2]. However, to our knowledge, there has been no previous report of pigmentary deposition on the cornea and conjunctiva in a case of Rothmund-Thomson syndrome.

More and more cases of various eye changes are being reported in association with Rothmund - Thomson syndrome. We are of the opinion that in our case also, the pigmentary deposition on the cornea and conjunctiva may be more than just a chance occurrence.

References

1. Rook A: Genetics in Dermatology, Textbook of Dermatology, 2nd Ed Edited by Rook A, Wilkinson DS and Ebling FJ Blackwell Scientific Publications, Oxford 1972. P 91.
2. Kirkham TH and Werner EB: The ophthalmic manifestations of Rothmund's syndrome, Canad J Ophthal, 10: 1, 1975.
3. Haneke E and Gutsclmidt E: Warty hyperkeratoses, cellular immune defect and Tapetoretinal degeneration in Poikiloderma Congenitale, Dermatologica 152: 331, 1976.
4. Sri Shanda Rajah Sivayohan et al: Rothmund Thomson syndrome in an oriental patient, Ann Ophthalmol 7 : 417, 1975.