THE de-SANCTIS CACCHIONE SYNDROME

By

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De-Sanctis and Cacchione in 1932 described three brothers with xeroderma pigmentosum, mental deficiency, microcephaly, retarded skeletal development and testicular hypoplasia. Since then there have been 22 cases on record with clear-cut manifestations of this syndrome (Reed et al, 1965). We have not come across any report since 1965.

In this article we are presenting the report of two Indian Hindu brothers with xeroderma pigmentosum, mental deficiency, microcephaly, retarded growth and testicular hypoplasia seen in the Dermatology Department of the All India Institute of Medical Sciences, New Delhi. (Fig. 1).

CASE I.

S. K. a 15 year old male patient was admitted in the Dermatology Wards on 28th January, 1969 with the complaints of multiple hyperpigmented and depigmented spots on the exposed parts; photophobia, lacrimation and diminution of vision; retarded growth and mental deficiency. The child was born as a full-term normal baby and on the 6th day developed erythema of the face and extremities which, a few days later, developed bullae and left behind, on healing hyperpigmented and depigmented spots. The pigmentary change was progressive in nature despite treatment. The milestones were delayed. There was no history of fits. His awareness of surroundings was seriously compromised and he was not able to look after himself. Assessment of I.Q. revealed that child had severe degree of intellectual retardation. (I. Q. 17-19).

Patient was a mentally defective microcephalic stunted child who was unmindful of his surroundings and could speak only in monosyllables.

Height 100 cms (expected normal 168.75 cm).
Weight 22 lbs (expected normal 103 lbs).
Head circumference 42.5 cms (Normal 62.5 cms).

Teeth normal: Testicles were not palable in the scrotum, inguinal region or abdomen. Penis was small. No secondary sex characters were seen.

Skin: There were multiple freckles and depigmented spots with atrophy and telangiectasia mainly on the face, dorsa of the hands, extensors of the forearm and to a lesser extent on the abdomen and back. Bilateral corneal opacities, conjunctival congestion, photophobia and lacrimation were present. No abnormalities were noted in other systems.

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INVESTIGATIONS

Amino-acids and porphyrins excretory patterns in urine were normal. 17 ketosteroids excretion in urine was 2 mg/24 hour (Normal 5–8 mg/24 hours).

Biopsy: Epidermis was markedly atrophic. Basal cell degeneration was seen at places. There was plenty of melanin in the epidermal basal cells as well as in the dermis. Collagen fibres were degenerated and widely separated. Scattered chronic inflammatory cells were present in the dermis (Fig. 2).

CASE 2.

O. P. 12 year old younger brother of case I presented with similar complaints. The lesions started to appear on the 7th day of birth. The mental and physical development was slightly better than in case I.

Height 86 cms (150 cms normal) weight 24 lbs (Normal 89 lbs). Testicles were not palpable in the scrotum, inguinal region or in the abdomen. Skin and eye conditions were similar to those in case I. He could speak few syllables and his capacity to understand was also slightly better than in the first patient. Examination of the other systems revealed no abnormality.

INVESTIGATIONS

Amino-acid and prophyrin excretion in urine was normal. 17 ketosteroid 1.2 mg/24 hours (normal 3–5 mg/24 hours).

Family history: The parents were unrelated and there was no history of excessive freckling. Two more siblings had similar skin lesions. But one died at the age of 1 year and the other one at the age of 8 months due to unrelated cause.

COMMENT

The two cases described here demonstrated all the characteristics of de-Sanctis Cacchione Syndrome viz;

1. Xeroderma pigmentosum.
2. Microcephaly with progressive mental deterioration.
3. Gonadal under-development.
4. Dwarfism.
5. Increased incidecce of abortions in the mother.

Xeroderma pigmentosum is transmitted as an autosomal recessive hereditary disorder. Most of the cases were thus seen in the products of consanguinous marriages (El Hefnavi, 1962). In the patients presented here, the disease appeared in the offspring from unrelated parents. Yet another unusual feature was that the disease appeared in the first week of life, while in more than 75% of cases, the disease is reported to manifest between the age of 6 months to 3 years (Rook et al 1968).

The exact defect in xeroderma pigmentosum, despite extensive investigations, is not finally settled. El-Hefnavi et al (1962; 1963) postulated multiple amino acid uria as the basic defect. Increased susceptibility to sunlight has been attributed by

Mehregan (1963) to the presenee of porphyrins. Neither porphyrinuria nor abnormal aminoaciduria could be demonstrated in the present cases. Low 17-ketosteroid excretion in urine was an evidence of hypofunction of the gonads. In present state of ignorance, it is difficult to correlate the photosensitivity states with physical, mental and gonadal under development. Whether all these are different expressions of a single metabolic defect is open to speculation.

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