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De Sanctis - Cacchione Syndrome

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Abstract

De Sanctis - Cacchione syndrome is an uncommon cause of mental retardation, associated with skin manifestations of xeroderma pigmentosum, physical retardation and neurological abnormalities. Three cases illustrating classical features of the syndrome are described and discussed.

Key words -
De Sanctis - Cacchione Syndrome,
Xerodermic idiocy

In 1932, De Sanctis and Cacchione described a family with 3 siblings in whom the skin lesions of xeroderma pigmentosum (XP) were associated with mental and physical retardation, testicular hypoplasia and neurological abnormalities [1].

The patients, who may be normal at birth, develop skin manifestations similar to those in XP, in early childhood. These include acute sun-sensitivity, freckles, xerosis and scaling, hypopigmentation, telangiectasia, atrophy, actinic keratoses, basal and squamous cell carcinomas, malignant melanoma and other tumours [2].

De Sanctis Cacchione syndrome (DSCS) is rare, with only about 60 cases having been reported in literature since 1932, and is thought to occur more often in Jews and Muslims [3]. We report on 3 Indian cases of DSCS, who came to attention because of their mental retardation and illustrate the typical clinical features of the syndrome.

Case Reports

Case No. I

V., a 2 ½ years old girl, presented with the complaint of a general delay in development. She was the younger of 2 children born of a union between first cousins (Figure 1). Her sister aged 5 years, was healthy. The birth history was uneventful. V. could not walk unsupported till the age of 2 years, and achieved monosyllabic speech only at 2 ½ years. Her parents reported that she was extremely sensitive to sunlight, and had developed redness and peeling of facial skin at l-1 ½ years, which later developed

into dark patches.

On examination at age 2 ½ years, the head circumference was 40.8 cm (microcephaly). There were black pigmented areas over the cheeks, nose and forehead, with hypopigmented, pin-head sized, spots in between. Conjunctiva of the left eye was inflamed. Similar skin lesions as on the face were also present on the exposed parts of the forearms. Mild spasticity was present in all 4 limbs. The dermatologist opined that V. suffered from XP, and skin biopsy from the face confirmed this.

The nature of the problem was explained to the parents, and measures for protection from the sun were suggested. On Vineland Social Maturity Scale, at the age of 6 years 3 months, V. had a social age of 3 years 2 months. When followed up at the age of 7 years 6 months, the pigmentation had increased marginally and the eyelid borders were thinned out and atrophic.

Case No.2

P., a 2 ½ years old girl, presented with a delay in all milestones. She was the youngest of 3 children born of an uncle-niece marriage (Figure 2). There were no perinatal problems. She attained neck control at 9 months, sat with support at 1 year 3 months, and spoke 2 words at 2 ½ years. The skin changes appeared first over the cheeks at 9 months and gradually increased.

On examination, she had microcephaly (head circumference 40 cm). There were black areas mottled with brown and white over the cheeks, nose, forehead and forearms. The eyelid margins were atrophic and the left cornea was hazy. Her developmental age was 9 months on Gessell's Developmental Schedule. Urine for amino acid was negative. Skull x-ray and EEG were normal. Skin biopsy indicated XP.

Protection from sun-rays, use of a sun-screening ointment, protection of the eyes, periodic checks by the dermatologist, and training at home in motor and self-help skills and speech, were advised. Two years later, there was no noticeable increase in the skin lesions. She could Sit un-supported, speak a few more words and recognize many people (Figure 3).

Case No.3

M., a boy aged 10 years, was brought with complaints of muttering to himself, not understanding questions, and generally slow development. At the age of 2 years, he had developed hyperpigmentation over the face, which had gradually increased. His parents were consanguineous (uncle-niece), and he was the first of 7 children (Figure 4). The second sib, a boy, had died soon after birth due to an uncertain cause. The fourth sib, a girl, had developed skin lesions, similar to those of the patient, at age 1 ½ years, gradually covering the whole face. She was also slow in her development and was considered to have been "a very dull child". She had died in an accident when she was 4 years old. The other sibs were normal.

The patient's milestones were all delayed: He could walk unsupported at 2 ½ years, spoke 2 words at 3 years, and was toilet-trained at 7 years. At the time of examination there was dark pigmentation over the nose, forehead, below the eyes, cheeks and temples, dotted with hypopigmented spots. There was another dark area on the chest, corresponding to the neck opening of the shirt. The child had angular stomatitis, anaemia and minimal incoordination of the upper limbs. Head circumference was 55 cm.

Investigations showed normocytic anaemia, and normal serum proteins, albumin/globulin ratio, blood sugar, urea, urine chromatography for amino acids and skull x-ray.

The dermatologist confirmed the diagnosis of XP. Parents were counselled and protection from sun

was advised. The child was seen again at the age of 12 years. Psychometry done at this time showed him to be below 3 ½ years mental age on Seguin Form Board, and 3 years on Binet Kamat. On Vineland Social Maturity Scale, the social age was 2 years 6 months. The EEG was abnormal with bursts of sharp waves and spike discharges.

Discussion

The diagnosis of DSCS in these 3 cases is based on the clinical features of mental retardation, neurological abnormalities and the skin biopsy reports confirming the lesions as those of XP. The classical features of the syndrome noted in their patients by De Sanctis and Cacchione included skin manifestations of XP, microcephaly with progressive mental retardation, retarded growth and sexual development, hearing loss, choreoathetosis, ataxia and eventual quadri-paresis [2]. However, not all patients display the complete syndrome.

All the 3 patients were born of consanguineous parents, and 1 had, in all probability, an affected sister. This is consistent with the proposed autosomal recessive inheritance of DSCS [4].

Cleaver [5] presented evidence suggesting defective excision repair of ultra-violet (UV) light induced damage to DNA in XP. The neurological damage in DSCS may be caused by chemical damage to DNA, since the nervous system is not reached by UV light [6].

Management of DSCS generally involves protection of the skin and eyes from UV radiation - natural and artificial, and frequent examination by self and dermatologists to detect malignancies early [4]. Although mental retardation is thought to be progressive, some gains may be achieved with appropriate training, as in case no.2.

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1.Adams R D, Neurocutaneous disease. In: Fitzpatrick T B, Eisen A Z, Wolff K, Freedberg I M & Austen K F. (Eds)
Dermotology in General Medicine. New York, McGraw Hill Page: 1206 - 1246, 1979

2.Robbins J H, Kraemer K H, Lutzner M A, Festoff B W & Coon H G, Xeroderma pigmentosum; an inherited disease with sun-sensitivity, multiple cutaneous neoplasms and abnormal DNA repair
Annals of Internal Medicine Page: 80: 221 - 248, 1974

3.Frias J L, De Sanctis - Cacchione syndrome (xerodermic idiocy). In: Vinken P J & Bruyn G W. (Eds)
Handbook of Clinical Neurology, Vol. 43, Amsterdam: North-Holland Page: 12-13, 1972

4.Koch G, Genetic aspects of the phakomatoses. In: Vinken P J & Bruyn G W. (Eds)
Handbook of Clinical Neurology, Vol. 14. Amsterdam: North-Holland Page: 488-561, 1972

5.Cleaver I E, Defective repair replication of DNA in xeroderma pigmentosum
Nature Page: 215: 652-656, 1968

6.Andrews A D, Barrett S F & Robbins J H, Relation of DNA repair process to pathological ageing of the nervous system in xeroderma pigmentosum
Lancet Page: 1318-1320, 1976