CUTANEOUS LUPUS ERYTHEMATOSUS IN ASSOCIATION WITH RUSSEL SILVER SYNDROME

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Introduction :

Cutaneous Lupus Erythematosus is generally regarded as uncommon during childhood [1,2].

Damm et al reported 3-4% of all LE to occur in children [3]. Thirteen cases of childhood cutaneous LE were reported in Oman during the period 1991-94 [4], Cases have been reported in association with other abnormalities, such as hereditary complement deficiency [5] and chronic granulomatous disease [6] . Familial cases also do occur [7]. We hereby report cutaneous LE in two brothers who had Russel Silver Syndrome, a syndrome of dysmorphogenesis.

Case History :

Case-1; W.N. a Omani boy aged 9 years, presented with a history of photosensitive erythematous rash on malar area of face, of six months duration. There were no systemic symptoms. Past history revealed that he was born to parents of consanguinous marriage (first cousins), at 36 weeks, of normal delivery. His weight at birth weight was 1.69kg, and height was 46 cms. Developmental milestones were normal. He had four brothers and one sister, of whom one brother of 7 years also had similar features (case-2)

Examination revealed a poorly built boy of weight 15 kg and height 0.8 metres. He had several dysmorphic features such as large triangular face, downturned corners of mouth, dolicocephaly, prominent eyes, frontal bossing, large ears, short stature, high, narrow palate and poor muscular development. (fig 1,2).

Systemic examination was normal. Mental growth was normal.

Cutaneous examination showed erythematous macular rash over cheeks and nose, with mild atrophy, and scarring. There were few telangiectasia on the nose. There was also a crusted erosion on lower lip (fig.2). Mucosae, nails and scalp were normal.

Blood investigations including complete haemogram, blood chemistry, urine analysis, xíray of the chest, hormone assays including growth hormone, serum corisol, thyroxin, T3 and T4 levels were all within normal range. Autoantibody profile including anti nuclear antibody, anti DNA, and Anti Ro antibodies was negative. Skin biopsy of lesional skin, for histopathology, showed typical features of discoid lupus erythematosus, such as hyperkeratosis with keratotic plugging, basal cell

degeneration, mild edema of dermis and superficial patchy perivascular and peri appendageas infiltrate of mononuclear cells. Direct immunofluorescence of lesional skin showed dense homogenous deposit of IgG and C3 along the membrane zone, confirming the diagnosis of discoid lupus erythematosus. Biopsy for DIF from non lesional skin was negative for lupus band.

Case-2; One of his siblings, a boy aged seven years revealed similar dysmorphic features and facial abnormalities (fig.1). He also had developed a malar rash in the last four months, with history of photosensitivity. The erythematous rash distributed over nose and cheeks showed mild atrophy and scarring. Haematological, hormonal, biochemical and serological investigations were all within normal limits. Biopsy of the lesion for H & E and DIF confirmed the diagnosis of DLE.

Both the patients were successfully treated with sunscreens, topical steroid creams and oral chloroquine (3.5 mg/Kg/day), for two months. The patients have been under follow up for six years. The rash recurs in summer and is well controlled with

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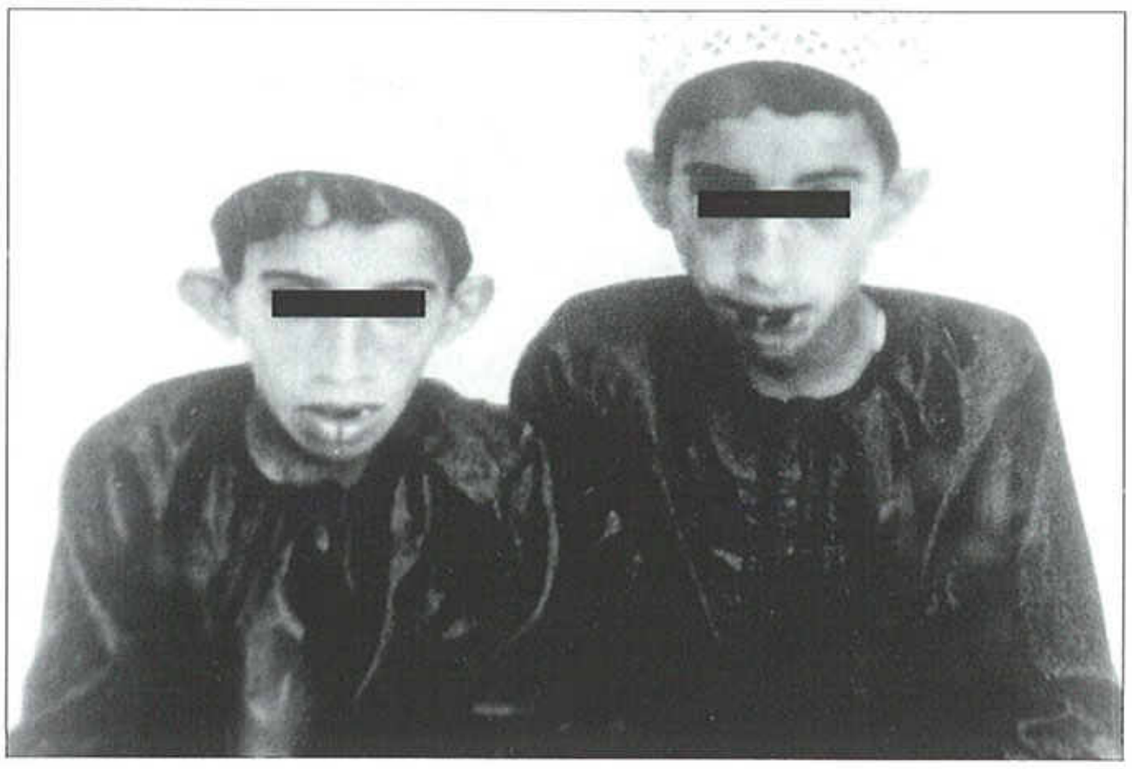

Fig 1. showing the affected siblings with characteristic facies and malar lesion.

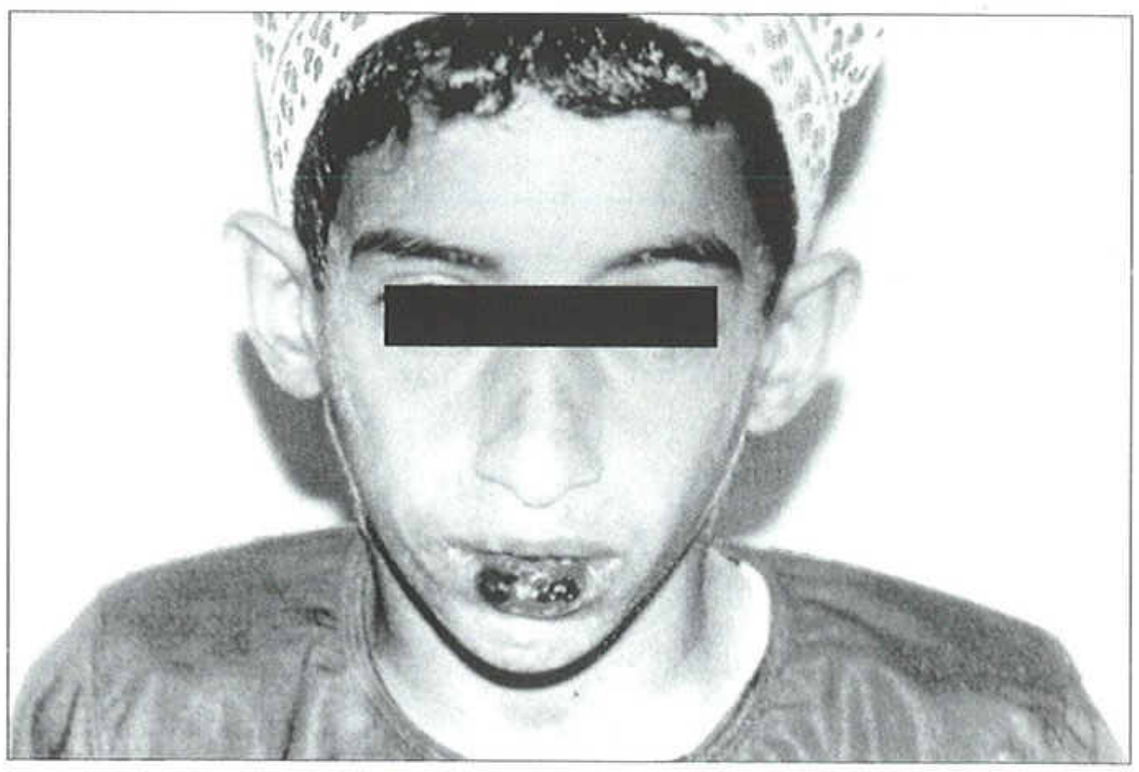

Fig. 2 Showing the patient with characteristic facies, DLE lesion on malar area and crusted lesion on lip.

short course of chloroquine and local steroids. There has been no progression to systemic disease.

Discussion: Cutaneous LE in childhood though uncommon, is an important clinical problem[1]. Association with hereditary complement deficiencies[5] and chronic granulomatous disease[6] are well established. Familial cases have been reported[7] . To the best of our knowledge, this is the first reported association with Russel Silver Syndrome.

Russel Silver Syndrome is a distinct form of intra uterine growth retardation associated with variable asymmetry and other heterogeneous congenital anomalies, first reported by Russel in 1954[8] and Silver in 1964[9] . Of the 200 cases that have been described so far, most cases are sporadic, though familial cases have also been described[10,11]. The aetiology of the syndrome is not known. Various chromosomal abnormalities, such as deletion of short arm 18 have been described, but there is no clear genetic inheritence pattern[10,12]. There is some debate about whether Russel and Silver syndromes are separate entities, the latter associated with asymmetry[10].

Various features of the syndrome are mentioned in table-1[13] . The main features of the syndrome include the dwarfism of prenatal onset, normal mentation, characteristic facies, asymmetry of limbs, normal growth hormone levels. Characteristic facies comprises of small triangular face, decreased facial height, downturned corners of mouth, small mandible and occasional asymmetry. Cutaneous lesions include pigmentary changes such as cafè au lait macules, which however, are not always present[10]. There is a wide variation in manifestations in patients belonging to the same family. The condition improves with age and features are less marked in adult life[13].

Our cases showed typical features of dwarfism of intrauterine onset with characteristic facies and normal mentation, suggesting a diagnosis of Russel-Silver syndrome. Association of Russel-Silver syndrome with LE has not been documented previously. Though the association in our cases is likely to be sporadic, our report is interesting in view of the occurrence of both Russel-Silver syndrome and LE in siblings. The patients have responded well to treatment and have been under follow-up for six years, without developing any signs of systemic lupus erythematosus.

TABLE - 1

Key features :	Other features :
Short stature, prenatal in onset Small, triangular facies with downturning corners of the mouth Clinodactyly, short incurved fifth fingers. Asymmetry most commonly of the limbs Cafè-au-lait spots Head that appears disproportionately large.	Liability to fasting hypoglycemia Prominent eyes Frontal bossing and mandibular hypoplasia on profile Long eyelashes Thin lips Palate high and narrow Crowded teeth Poor muscular development Syndactyly of second and third toes Delayed closure of anterior fontanelle Precocious puberty Genital abnormalities: hypospadias, ambiguous genitalia, small testes, cryptorchidism.

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