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Response of Oral Acitretin in Congenital Icthyosiform Erythroderma: A Case Report.

Medical Science

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ABSTRACT

The congenital ichthyoses are inherited disorders of keratinisation with distinctive morphological features. We report a case of congenital ichthyosiform erythroderma (CIE), an uncommon autosomal recessive congenital ichthyosis (ARCI) characterised by generalised erythroderma with greyish white scales in a one year old infant and the response to oral acitretin therapy.

Introduction :
The earliest reference to ichthyosis as 'Ekakushtha, an extensive dermatosis that does not sweat and resembles the scale of a fish', in the ancient Indian treatise, 'Charaka Samhita' dates back to the 3rd century BC. [1]

CIE with an incidence of between 1 in 200 000 and 1 in 300 000,2 is an uncommon autosomal recessive ichthyosis often associated with consanguinity that may present at birth with a collodion membrane which is shed to reveal a generalised scaly erythroderma. General supportive measures, topical emollients and systemic retinoid therapy may be associated with an improved outcome.

Case report :
A one year old male, the product of a full term normal delivery of consanguineous parents (second cousins) presented with redness and scaling over the entire body surface since birth. A glistening membrane enveloped the neonate at birth which was shed after a week to reveal persistent redness with scales over the body. The shedding of scales showed waxing and waning. The infant developed fever off and on for which he received treatment.

Dermatological exam revealed a generalised erythroderma with semi-adherent scales over the scalp, face, trunk, extremities.The scales were large, thin and greyish white (Figure 1). Flexural areas as well as palms and soles were involved. Ectropion, with eversion of lid margin of both lower eyelids as well as superciliary madarosis was present (Figure 2). Tinea amiantacea like overlapping adherent scales were observed on the scalp.

A clinical diagnosis of CIE was made and was confirmed by histopathological exam which revealed mild thickening of stratum corneum with foci of parakeratosis, mild acanthosis, normal granular layer and mild upper dermal lymphocytic infiltrate.

The patient was managed with topical emollients as well as supportive measures particularly during febrile episodes. Oral acitretin was instituted in a dose of 0.5 mg/kg/day. There was marked reduction in scaling over the face, scalp, trunk and extremities and ectropion of lower eyelids improved. Gradual decrease in erythema was also observed within three months of initiation of therapy (Figure 3).

Discussion :
ARCI is an umbrella term for non-syndromic congenital forms of ichthyoses without tendency towards blistering. It includes the major subtypes congenital ichthyosiform erythroderma (CIE), lamellar ichthyosis (LI) and harlequin ichthyosis (HI). The minor subtypes of ARCI encompassed by the term pleomophic ichthyoses (PI) include self-improving collodion ichthyosis (SICI), ichthyosis prematurity syndrome (IPS), bathing-suit ichthyosis (BSI) and congenital ichthyosis with fine/mild scaling (CIFS).3

The genotype correlation within the CIE/LI spectrum includes mutations in TGM-1, ABCA12, NIPAL4, CYP4F22, ALOX12B and ALOXE3 genes. Both CIE and LI usually present with a collodion membrane at birth which is gradually shed to reveal the ichthyosis phenotype. CIE is characterized by pronounced erythema with fine white scales while LI presents with coarse brown scales. The histologic features are nonspecific but inflammatory cell infiltration with foci of parakeratosis are seen more frequently in CIE than in LI.4

Neonatal and infantile erythrodermas are potentially life threatening conditions associated with risk of a hypernatremic dehydration, septicaemic infections, hypoalbuminaemia and hyperpyrexia.In a retrospective study of infantile erythrodermas the mortality rate was 16 % and severe dermatoses persisted in 67 % of survivers.5 Lack of functional epidermal barrier leads to increased transepidermal loss of water and temperature imbalance which should be controlled by regular fluid infusion, topical non medicated emmolients (coconut oil/liquid paraffin/petrolatum jelly) and incubators for temperature control. Topical keratolytics such as urea is avoided in first year of life whereas salicylic acid is contraindicated in infants but can be used locally in children and adults.6

Since ARCI is essentially a disorder of keratinization systemic retinoid therapy with acitretin or isotretinoin has been found to be useful. The recommended starting dose of acitretin in CIE is 0.5-0.75 mg/kg/day and maintenance dose is titrated to the lowest effective dose i.e 0.1-0.5 mg/kg/day.7 In view of the potential toxicity of oral retinoids

and the chronicity of the disease intermittent therapy for three months followed by a drug free period of three months may be instituted.7 Though marked improvement in ectropion and scaling was observed in children with CIE the erythrodermic component did not show an equally good response.8

Most patients on systemic retinoids develop mucocutaneous dryness which is dose-dependent and reversible. Minor impairment in liver function tests are recorded but do not necessitate a change of therapy. Though skeletal abnormalities such as premature epiphyseal closure and spinal hyperostosis have been reported in the past it is observed that the physical growth of patients of CIE remains unaltered even after long term systemic retinoid therapy.8

Recently multicentric trials of the retinoic acid metabolism blocking agents (RAMBAs) liarazole and rambazole in the treatment of ARCI have been carried out and have been found to be as effective as acitretin. RAMBAs are inhibitors of cytochrome P450 and increase the intracellular concentration of retinoic acid.9

Neonates with ARCI are prone to infections and restricted pulmonary respiration and require management in a neonatal intensive care unit to prevent hyponatraemic dehydration or hypothermia. Sodium bicarbonate as a bath additive as well as topical emollients without salicylic acid have been found to be helpful.6 Systemic retinoids have been found to reduce skin thickness and scaling in ARCI and have been used even in children and neonates.9 In view of long term therapy and potential adverse effects these patients require close monitoring. The prognosis of patients with ARCI has improved markedly with the use of systemic retinoids.

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Legends to figures

Fig. 1: Erythrodermic child with thin, grayish-white scales.

Fig.2: Ectropion with supracilliary madarosis.

Fig. 3: Decreased erythema and scaling after 3 months of acitretin therapy.