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Bart's Syndrome

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Abstract

We present a case of a newborn with congenital absence of skin in both feet, dystrophic nails in both the lower limbs and who shortly after birth developed bullae and erosions in hands, ears, buttocks and mouth. With these findings, the diagnosis of Bart syndrome was made. Bart syndrome is a clinical diagnosis characterized by triad of the localized absence of the skin present at birth, epidermolysis bullosa (EB), lesions of the mouth and mucosa, and disfigured nails. Combination of absence of skin i.e. aplasia cutis congenita (ACC) and with simplex, junctional, or dystrophic types of epidermolysis bullosa (EB) is classified as type six ACC which is a very rare clinical condition and very few cases are available in literature and this case is fifth from India.

Key words: Bart's syndrome, Epidermolysis Bulosa, Aplasia Cutis.

INTRODUCTION

Bart's syndrome was described first by Bart et al in 1966 and it represents the combination of congenital epidermolysis bullosa, congenital localized absence of skin affecting the extremities and shedding or dystrophy of nails.[1] Since, Bart first described this syndrome there has been a debate to consider this syndrome as a separate clinical entity or a variation of dominant dystrophic epidermolysis bullosa. In India, Bart's syndrome has been previously reported by four authors and every reported case had a fatal outcome.[2-5] We present a case of a neonate who was clinically diagnosed as Bart's syndrome and who was managed conservatively and though the wound healed with scarring, the baby survived.

CASE REPORT

A full term female baby born to primigravida at our institute was brought immediately after delivery to NICU (Neonatal intensive care unit) with complains of bilateral lower limb anomaly and blisters over buttock, hand and abdomen. On further history, antenatal period was uneventful, with USG (ultrasonography) done twice which were told to be normal. There was no history of fever, radiation exposure or drug intake apart from iron and folic acid tablets. On examination, birth weight was 2.45 kg, and there was absence of skin in both lower limbs below the ankle, large bullae over legs, thigh, hand, forearm, abdomen and buttock [Figure - 1,2]. Small bullae were present over face but not involving scalp. Dystrophic nail were present in large toe and two fingers of the right lower limb. No other congenital abnormality was present. Further examination revealed erosions of oral mucosa. Baby was irritable but vitals were stable. Systemic examination was within normal limit. Investigations like peripheral smear and CBC revealed no abnormality. USG abdomen was within normal limits, it was done to rule out pyloric obstruction commonly associated with dominant dystrophic epidermolysis bullosa.

Baby was admitted in NICU and intravenous fluids were started, empirical antibiotic therapy with ampicillin and gentamicin was started. Wound dressing was done with sterile gauze, liquid paraffin and povidone iodine. CRP was done on day two of life which was normal and CBC was repeated which was again normal so antibiotics were stopped after 3rd day. Daily wound care was taken in the form of changing the dressing and hypodebridement. Intragastric feeding was initiated on day one of life and was monitored for signs of intolerance, which were not there, so feeds were rapidly increased to full feeds. Breast feeding trial was given daily and baby was shifted to complete breast feeding by day 7 of life. Conservative management was continued. Fresh bullous lesion kept on appearing all over the body except scalp, but the size of lesions was small <1 cm. Baby was discharged on 45th day of life with no fresh lesion since last 7 days and all previous lesion healing and no signs of systemic or local infection. Skin biopsy was not possible as parents were not willing to give consent. Baby is on regular follow up since then and the frequency of appearance of new bullous lesion is now less than one per week and that too less than 1 cm in size.

DISCUSSION

The mode of inheritance of Bart syndrome is that of an autosomal dominant gene, showing full penetrance and variable expressivity.[1] The exact cause of blistering in patient of Bart's syndrome is not known but all cases are associated with mutations of the gene coding for type VII collagen (COL7A1) at the short arm of chromosome 3 in the p21 region. This finding is similar in all the patients having dystrophic epidermolysis bullosa, therefore, Bart's syndrome is mistakenly considered as a intrauterine variant of dystrophic epidermolysis bullosa.[6,7]

Frieden classified Aplasia cutis congenita (ACC) into nine groups according to localization, associated anomalies or syndromes, and the inheritance. Our case with ACC, blistering, oral mucosal involvement and nail dystrophy was classified as type VI aplasia syndrome according to Frieden classification.[8] Other types were ruled out on the basis of clinica presentation as follows - no scalp involvement (type I, II, III, and IV), no fetus papyraceous or placental infarct (type V), presence of epidermolysis bullosa (type VII), no history to any teratogen exposure (type VIII) and finally, no other associated congenital abnormality (type IX).

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Figure1: Absence of Skin in both lower limbs below the ankle

Figure2: large bullae over legs ,thigh, hand, forearm, abdomen and buttock

There is no specific management for Bart's syndrome. The diagnosis is clinical, but skin biopsy will lead to confirmation. Prevention of friction and trauma to the skin will help in preventing new blister formation. Wound care with sterile dressing and topical antibiotics is sufficient, unless patient develops secondary systemic infection. Treatment of lesions is of uttermost importance because this is an extremely painful clinical condition that limits quality of life. Complications arising from a conservative treatment may include sepsis, local infection, hemorrhage, excessive loss of fluids, hypothermia, electrolyte disorders, and later hypertrophic or atrophic scars. Decades ago, when sophisticated products were not readily available and proper techniques to treat wounds were still evolving, infection was the main cause of mortality in any one of the types of congenital epidermolysis bullosa.[6] Currently, with the availability of newer products, prognosis is relatively favourable and depends upon the improvement of lesions as patients get older.

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