Sultan Qaboos University Med J, February 2015, Vol. 15, Iss. 1, pp. e143–145, Epub. 21 Jan 15
Submitted 15 May 14
Revision Req. 3 Jul 14; Revision Recd. 7 Aug 14
Accepted 19 Aug 14

INTERESTING MEDICAL IMAGE

Bart Syndrome with Ear Malformation

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متلازمة بارت مع تشوه الأذن

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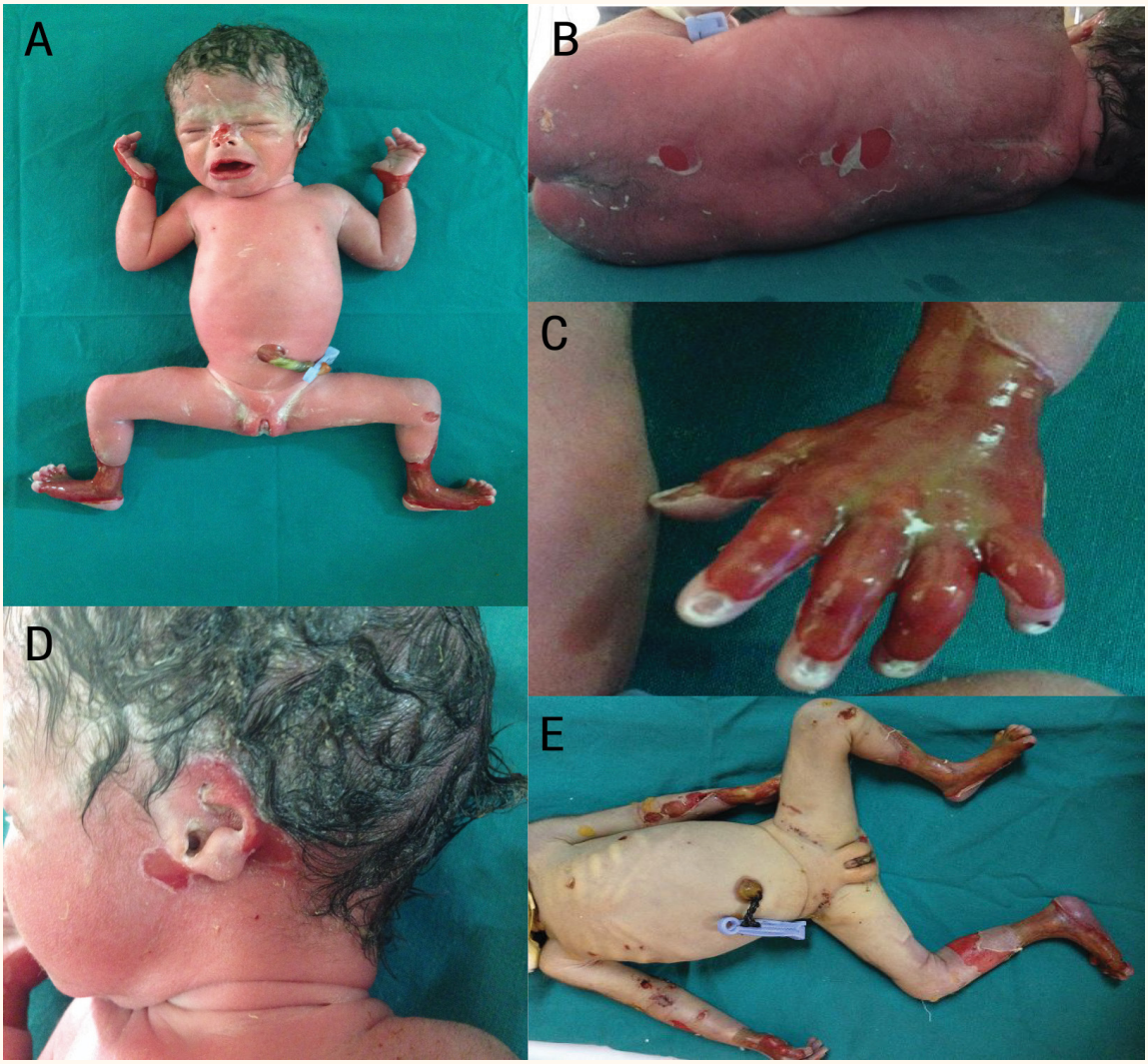

Figure 1A–E: Photographs showing a case of Bart syndrome with ear malformation in a female Egyptian newborn. **A:** Well-demarcated non-inflammatory skin defects covered by a red thin translucent membrane localised to the nose, around both wrist joints, the front of the left knee, *dorsum* of both feet, around both ankle joints and around the lower third of the legs. **B:** Two areas of ulcerations on the back. **C:** Dysplastic nails and absent skin over the *dorsum* of the hand, sparing the fingertips. **D:** Malformed ear with attached helix to the underlying skin and small areas of absent skin around the ear. **E:** Dryness and extension of the old skin lesions and the appearance of multiple bullous lesions was noted over the upper and lower limbs.

Bart syndrome was first described by Bart *et al.* in 1966 with a constellation of congenital localised absence of skin (CLAS), congenital epidermolysis *bullosa* (EB) and associated nail abnormalities.[1] This article reports a case of Bart syndrome with ear malformation in a female Egyptian

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newborn. To the best of the authors' knowledge, this is the first report of Bart's syndrome in an Egyptian family.

A female preterm newborn (with a gestational age of 35 weeks) was referred at birth to the neonatal intensive care unit of Suez Canal University Hospital, Ismailia, Egypt, in February 2014. She presented with multiple areas of congenital CLAS associated with nail dystrophy and malformed ears. The parents were consanguineous and had previously had three daughters with the same condition who had subsequently died at the ages of three hours, four hours and seven days old, respectively. The current neonate was the sixth child born to the couple.

On physical examination, the newborn showed sharply demarcated, bilateral non-inflammatory skin defects covered by a red thin translucent membrane localised to the *dorsum* of both hands and around both wrist joints, the *dorsum* of both feet and around both ankle joints, around the *umbilicus*, anterior aspect of the left knee and nose [Figure 1A] and over the back [Figure 1B]. Dysplastic nails were also observed, as well as bilateral malformed ears in the form of adherent auricles attached to the underlying skin and bilateral *hallux* hypoplasia [Figures 1C & D]. Laboratory investigations revealed a total leukocyte count of 2,300/cmm, C-reactive protein levels of 28 mg/dL, normal blood chemistry and blood gases and a negative skin swab for aerobic bacteria. The newborn was managed with systemic vancomycin and ceftazidime initiated empirically in addition to local topical fusidic acid with atraumatic dressings for the skin lesions.

At day of life (DOL) two, the infant began to develop disseminated bullous skin lesions mainly over the upper and lower limbs and a mucous membrane of the mouth. Oozing, dryness and extension were observed in the old skin lesions [Figure 1E]. At DOL four, the baby developed severe neonatal sepsis with depressed activity as well as extensive lesions in the lips, on the mucous membranes of the mouth and larynx with yellowish endotracheal aspirate. The neonate died on DOL four due to severe neonatal sepsis, septicshock and metabolic acidosis. The rapid progression and fatal course of the disease interrupted the planned ultrastructural, immunohistological and genetic link-age investigations.

Comment

Bart syndrome is one type of dominant dystrophic EB. In some families, a link to the gene for type VII collagen has been demonstrated, suggesting that this is the mutational site.[2] The diagnosis of aplasia *cutis congenita*, including Bart syndrome, is primarily a clinical diagnosis based on the classical cutaneous findings; the extent of involvement depends on the mode of inheritance.[3] In the family of the current case, only female infants were affected. This raises questions regarding whether gender is involved in the phenotypic variation and severity of Bart syndrome. This case may therefore support the hypothesis of Wakasugi *et al*., who suggested that the expression of symptoms might differ depending on the gender of the affected individual.[4]

The similarity of defects and symptoms in all reported cases, as well as in this patient, strongly suggests that CLAS in Bart syndrome is not solely associated with a mechanical aetiology, but may follow Blaschko's lines, particularly with regards to the localisation of lesions to the limbs.[5] Other areas exposed to friction and trauma, such as the skin around the oral cavity, nose and ears, can also be affected. Nail changes include congenital absence in terms of nail dystrophy or progressive loss.

This reported case of Bart syndrome was diagno-sed clinically, postnatally. A skin biopsy is essential in establishing an accurate postnatal classification of inherited EB. The biopsy sample should be examined for a combination of ultra-structural and antigenic features by transmission electron microscopy, immunofluorescence antigenic mapping and EB-related monoclonal antibody investigations.[6] Fetoscopies have been performed successfully to diagnose inherited EB prenatally; fetal skin biopsy specimens for ultrastructural analysis are obtained via transmission electron microscopy in order to identify possible structural defects in the fetal skin.[7] However, fetal skin biopsies have gradually been superseded by DNA-based diagnostic screening using fetal DNA from amniotic fluid cells or chorionic *villi* samples.[8] Despite a strong family history, a prenatal diagnosis in the current patient was not attempted due to the expense of the required tests and procedures.

Septicaemia, electrolyte imbalances, protein loss and failure to thrive complicate severe exudative skin lesions and often lead to death in cases of Bart syndrome.[9] Septicaemia was also the cause of death in this case. Patients with Bart syndrome can be successfully treated with the use of topical antimicrobial drugs and sterile dressing pads for prophylaxis of secondary infections. In infants older than two months, the use of silver sulfadiazine cream and the early debridement of lesions with closed dressings has been shown to improve survival and quality of life for affected patients. Rosmaninho *et al*. reported the complete healing of prominent milia after three months; however, blistering of the lesions recurred.[10] Extra-cutaneous involvement has also

been observed in survivors, commonly reported as mucosal involvement of the gastrointestinal tract, genitourinary tract and eyes. Corrective gene therapy is the ideal therapy for EB, but more research is required prior to its utilisation in clinical practice. Cell-based therapies using fibroblasts and bone marrow cells have recently attracted considerable attention and may lead to effective results.[11]

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