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CASE REPORT

De Barsy syndrome in an extreme preterm infant

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Abstract:
We report a rare case of an extreme preterm infant with De Barsy syndrome characterized by severe mental retardation, cutis laxa, cloudy corneas, muscular hypotonia, and lax joints. The etiology of this syndrome is unknown but it is probably heterogeneous. Recently a autosomal recessive glycosylation disorder (ATP6V0A2-CDG) has been associated with De Barsy syndrome.

Keywords: De Barsy syndrome, Cutis laxa, Progeroid syndromes
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Introduction

De Barsy syndrome is a rare, autosomal recessive reported by De Barsy et al in 1968 [1]. It consists of cutis laxa, a progeria-like appearance with facial, ophthalmological, neurological and orthopedic abnormalities [2-6].

Some 28 cases have been reported [1]. We describe this syndrome in an extreme preterm infant, the first case to be reported from the United Arab Emirates.

Case Report

This boy was the product of a 27 weeks gestation with a birth weight of 535 g. He was born to consanguineous parents by emergency lower segment cesarean section for severe ante partum hemorrhage; Apgar scores were 6 and 7 at 1 and 5 minutes, respectively. A male sibling with similar features died in the neonatal period at another hospital due to sepsis. He also had a hiatus hernia and cutis laxa. A female sibling is suffering from severe sensory neural hearing impairment. Two other siblings were normal.

Soon after delivery, the baby required admission to the neonatal intensive care unit for respiratory distress. Surfactant was given and ventilation was initiated. His clinical examination showed axial hyotonia, a loose skin over the abdomen, and hypospadias.

A hoarse cry was noticed after extubation to nasal CPAP. His systemic examination revealed a left large

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but reducible inguinal hernia, and an undescended right testicle. The skin over the abdomen continued to appear lax and redundant (Figure I).

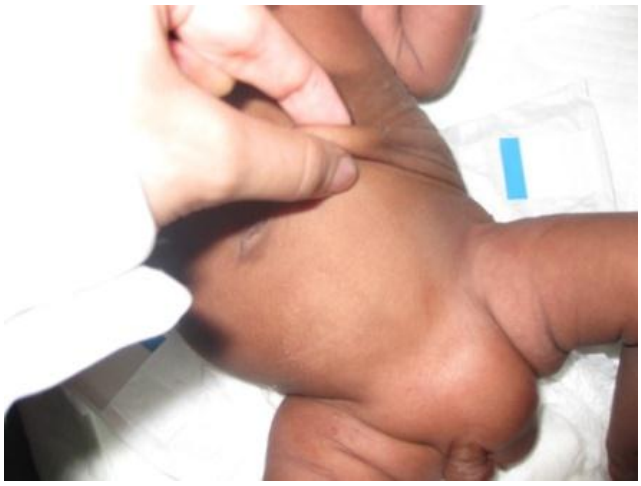

Figure I. Cutis laxa, undescended testicle and left indirect inguinal hernia

Other distinctive features became more prominent as the age of the baby advanced.

He was noted to have an aged appearance due to the lax facial skin, especially over the eyelids (Figure II). His hair was sparse and nostrils were anteverted (Figure II).

Figure II. Note the aged appearance, anteverted nose, lax facial and lower lid skin, sparse hair.

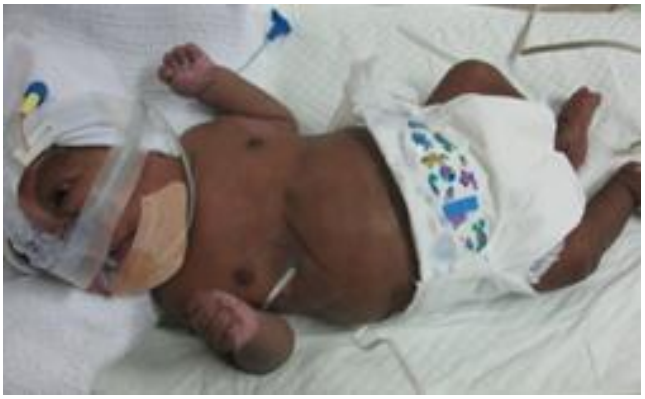

Figure III. Respiratory support with CPAP; also note Hypotonia, marked abdominal distention and clenched hands

His hips were lax and dislocatable bilaterally and he had clenched hands (Figure III).

Hematological, metabolic and endocrinological tests were normal but serum transferrin isoelectrofocusing was not performed.

Fundoscopy revealed cloudy, edematous corneas with bilateral remnant of papillary mucosa and a loculated iris. ***Karyotyping*** in lymphocytes was normal. ***Skin biopsy*** revealed marked paucity of collagen within the whole dermis with hydropic degeneration of the basal layer with an onset of vesicle formation. On elastin staining no intact elastic fibers were identifiable.

Multiple extubations trials failed. Serial chest x-rays showed elements of interstitial emphysema and chronic lung disease. Baby stayed under ventilation in the neonatal intensive care unit until he was declared dead at six months of age.

Discussion

De Barsy syndrome is a rare syndrome with cutis laxa, a progeria-like appearance, and neurological, ophthalmological and orthopedic abnormalities. It is thought to be inherited in an autosomal recessive manner and its cause is mostly unknown [3]. Recently a genetic defect has been identified in one patient namely a glycosylation disorder, APT6V0A2-CDG [7].

Diagnosis of our case took around 2 weeks to be established, as all the clinical manifestations were not present before in this extreme preterm infant. It was

based on family history, clinical features and skin biopsy.

Parental consanguinity was present in this case as well as in 11 other cases [2]. Cutis laxa, progeroid face, small anteverted nostrils, sparse hair, hypotonia, lax hip joints, corneal opacification and inguinal hernia were associated features described in previous case reports as well with this case. Histological findings supported the diagnosis of de Barsy syndrome, as there were abnormalities of the elastic fibers, which were markedly diminished, and paucity of the collagen element. Features such as cutis laxa and cloudy corneas in the De Barsy syndrome might be linked to the abnormal elastogenesis. However, the neurological features and cataracts are not readily explained by abnormalities in elastin [3]. As differential diagnosis, we should mention Menkes syndrome (X-linked recessive ATP7A related copper transport disorder), who also have cutis laxa and have elastin abnormalities due to problems with cross-linking.

The life expectancy of patients with the De Barsy syndrome is not known. The oldest reported case was 25 years old [4], and all other cases were reported during infancy or childhood [2].

Conclusion

In this case, the diagnosis of de Barsy syndrome was primarily based on the unique clinical features and supported by the skin biopsy findings. Clinical features might not appear simultaneously in extreme preterm infants; cutis laxa was present in our case from day one of life. Keep De Barsy syndrome in mind upon receiving an infant or child with cutis laxa as it is the most apparent and striking of features.

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Written consent was taken from the mother for publishing the case and including her baby's photographs.

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