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Case Report

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BART'S SYNDROME

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ABSTRACT

It is a genetic disorder characterised by congenital localised absence of skin, epidermolysis bullosa, lesions of oral mucosa and dystrophic nails. It may be associated with Epidermolysis bullosa. It is a genetic mechanobullous disorder in which the affected baby is born with areas of denuded skin characterised by focal absence of skin. The lesions tend to occur on those parts of the body which are exposed to friction and trauma. The mode of inheritance is autosomal dominant.

KEYWORDS: Bart's disorder, denuded skin.

INTRODUCTION

It is a genetic disorder characterised by congenital localised absence of skin, epidermolysis bullosa, lesions of oral mucosa and dystrophic nails. It may be associated with Epidermolysis bullosa. We can diagnose it clinically, though for confirmation of diagnosis ultrastructural microscopy is required. This disorder was described in a family in 1966. It affects 1:20,000 children. It is a genetic mechanobullous disorder in which the affected baby is born with areas of denuded skin characterised by focal absence of skin. There are raw, rich red plaques on different parts of the body. The lesions tend to occur on those parts of the body which are exposed to friction and trauma. The mode of inheritance is autosomal dominant.

CASE HISTORY

I am presenting a case of a lady, 31 years of age with bad obstetric history. She was a booked case at Dande Hospital. She came to the emergency department at 4am on April, 2nd 2018 with labour pains at 34 weeks of gestation and vaginal discharge. She was a sixth gravida with one live issue.

Obstetric history

She was married 10 years ago. It was not a consanguineous marriage The first pregnancy was an intrauterine demise at 26 weeks with breech presentation. She delivered spontaneously. The baby was a male child with no obvious congenital malformations.

The second pregnancy was terminated at 21 weeks as the baby had absent kidney, rudimentary ears and a brain tumor.

She was investigated as a case of bad obstetric history. She conceived again and delivered vaginally a male child weighing 2.75kg at term. The child is now 5 years of age. He is physically and mentally normal with normal cognitive development.

The fourth and fifth pregnancy were missed miscarriages which were terminated medically.

This was her sixth pregnancy. There was no medical complication in this pregnancy. There was no family history of similar disorder in any baby.

Investigations

Her haemoglobin, TLC, Platelet count was normal.
Hba1c and TFT was normal
TORCH screen and APLA results were normal.
Ultrasound done at 27 weeks showed polyhydramnios with IUGR.
Cervical length was 27mm.

Management

On examination her blood pressure was 120/70 mm of hg and pulse was 90/minute.

Per abdominal examination revealed a pregnancy of 34 weeks gestation with breech presentation. The fetal heart rate was 144/ min. She had mild uterine contractions.

Per vaginal examination showed that the cervix was 2 cm dilated with bag of membranes bulging in the vagina. She had obvious leaking per vaginum.

An intravenous line was put immediately. She was given ringer lactate and prophylactic antibiotics according to

the hospital protocol. Injection betamethasone 12 mg was given immediately.

Oral tocolytics were started. She was given Nifedipin tablet 10 mg 6 hourly.

She was explained the prognosis regarding prematurity, need for caesarean section, PPH, need for NICU admission. Informed written consent was obtained.

After 3 hours of observation she was 3 to 4 cm dilated. Decision of caesarean section was taken.

Intraoperatively she had excess of amniotic fluid as expected for the gestational age. It was greenish brown coloured with turbid material.

The baby was delivered by breech and handed over to the paediatrician. The baby cried after birth.

On Examination of the Baby

The baby was a female child had absence of skin on both the knees, areas of legs in the dorsal aspect, dorsum of feet, part of wrist and left of the neck.
There were no bullous lesions.
The part of skin which was exposed showed red, beefy areas with the underlying vessels seen clearly.
Nails were dystrophic.
Oral and nasal cavity was normal.
Weight of the baby was 1.5 kg.
She had rudimentary ears, wide set eyes, flattened nose.
The baby was hospitalised in the NICU and needed respiratory support.
Antibiotics and fluid was administered. Sterile dressing was applied over the open skin lesions.
Dermatologists opinion was sought and clinical diagnosis of Bart's syndrome was made. Skin biopsy was advised for confirmation of diagnosis.
After 2 days the baby was diagnosed with pyloric atresia.
The baby was managed with fluids, antibiotics, respiratory support and daily dressing.
The decision to operate on the baby was avoided considering multiple risk factors and sepsis.
The baby succumbed on 7th day post delivery. The cause was fluid and electrolyte imbalance along with associated sepsis.

DISCUSSION

Bart et al in 1966 reported this condition in a large family in which 20 members were affected.

They all had congenital absence or blistering of skin and nail atrophy.

There are three clinical subtypes of the disorder - epidermal, junctional and dermal.

The mode of inheritance is Autosomal dominant.

It has been reported with as a subtype of Epidermolysis Bullosa(EB) i.e Simplex, junctional and dystrophic.

Kanzler et al described a family in which four generations had EB simplex with congenital absence of skin. They did the electron microscopy for the first time. The findings suggested that the areas of congenital absence of skin had the same pathogenic changes as EB.

Zelickson et al found poorly formed anchoring fibrils below the lamina propria.

Christiano et al proved that there was a mutation in the helical domain of type VII collagen in affected individuals.

Mc Kinster suggested that the denuded areas are due to trauma in utero and follows the lines of Blashko.

The Koebner, Weber Cockayne and Dowling Meara suggested that forms of EB are rarely associated with Bart's syndrome.

Involvement of nails suggested that it was possible that this baby had a junctional type of EB. PAS staining or Electron microscopy was not done in this case. Bart's syndrome is often associated with Pyloric stenosis. This was present in this baby. The possible cause of death can be hypothermia, hypoglycemia or sepsis. In this case the cause of death was sepsis and fluid electrolyte imbalance.

Genetic counselling is a must in these patients. Diagnose can be done early by CVS or slightly later by amniocentesis. The defect is in the region of chromosome 3 near the type 7 collagen gene. There is glycine to arginine transition substitution within the triple helical domain of type 7 collagen in affected individuals.

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