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Case Report

Newborn with Severe Peeling of Skin: Bullous Congenital Ichthyosiform Erythroderma – To Treat or Not to Treat?

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Abstract

A 31-year-old lady with primary infertility conceived underwent in-vitro fertilization (IVF) for bilateral blocked fallopian tubes. She conceived in the first cycle itself with a single pregnancy. Her antenatal course was uneventful. She delivered a female baby of 2.5 kg with APGAR of 9/10. On delivery it was it was noted that baby had excessive peeling of skin all over her body. She was diagnosed with congenital ichthyosiform erythroderma. Unfortunately she succumbed. Her whole exome sequencing revealed pathogenic variant causative of KR10 gene (pathogenic) which is an autosomal dominant inheritance. The couple was advised with whole exome sequencing.

Keywords: Congenital Ichthyosiform Erythroderma; Epidermolysis Bullosa; Collagen; Peeling

Background

Early blisterng or peeling of skin in newborns is seen in a group of rare medical condition known as Epidermolysis bullosa (EB). In this condition there is blistering all over the skin and mucosa due to minor trauma or friction. These can be extremely painful to the newborn. The severity ranges from mild to fatal leading to dead of the baby in many cases. In this condition there is separation of the dermal epidermal component of the basal membrane of skin [1]. Four types of clinical presentation are seen: Epidermolysis bullosa simplex (EBS), Dystrophic epidermolysis bullosa (DEB), Kindler syndrome and Junctional epidermolysis bullosa (JEB) [2,3]. The prognosis is different depending upon the type of lesion, ranging from minor affection of the skin to life threatening condition [4]. EB occurs because of mutation in one of the 16 genes. Certain condition is caused due to autosomal dominant inheritance while some can be autosomal recessive. There is weakness in the structural architecture of the dermal–epidermal junction (DEJ) and mucosal membranes due to reduction in function of type VII collagen. The prevalence of EBS is differs from country to country and ranges from 6:1,000,000 to 28.6:1,000,000 live births. It is difficult to diagnose the exact subtype at birth. Further the management of child is difficult and challenging due to the extensivity of lesion and lack of clear diagnosis at birth. It poses both clinical and ethical dilemma for the clinicians in terms of explaining about the prognosis and quality of life. We hereby share a case of congenital Bullous congenital ichthyosiform erythroderma for the benefit of future patients with discussion on the ethical dilemmas.

Case Report

A 31-year old lady with primary infertility underwent in-vitro fertilization (IVF) due to bilateral tubal block. She conceived in the first cycle itself with a single fetus. Her antenatal course was uneventful. Her nuchal thickness (NT scan), dual marker, anomaly sonography and growth sonographies were normal. Her routine investigations were also normal. She underwent an elective cesarean delivery at 37 weeks of pregnancy for breech presentation. She delivered a female baby of 2.5 kg with APGAR of 9/10 at delivery. However at delivery it was noted that the newborn had extensive peeling of skin all over her body, as seen in figure 1. These lesions occupied almost 70% of her skin surface. The couple was informed about the same and baby shifted to neonatal intensive care unit (NICU) as seen in figure 2. Baby was kept in humidified incubator with continuous cardiorespiratory monitoring. During her stay in NICU, the baby suffered four episodes of convulsion on the second day of life probably due to severe electrolyte imbalance as her majority of skin was peeled off. She was not able to maintain temperature. Investigations revealed electrolyte imbalance specifically hypernatremia. Supportive treatment along with electrolyte correction was continued. A dermatologist opinion was taken. To confirm the diagnosis and with their genetic association, whole exome sequencing of the newborn was done.

During her NICU stay an umbilical catheter was placed in order to provide intravenous saline to prevent dehydration from extended evaporation from the skin. Paracetamol and phenobarbital was

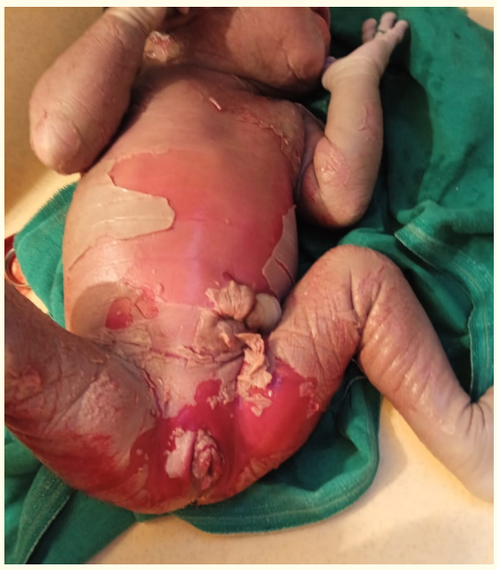

Figure 1

Figure 2

used for pain management (figure 3). The baby was on parenteral nutrition only. As her complete blood count showed elevated infection parameters, neutropenia and raised c-reactive protein (CRP) on day 3 of life, antibiotics were administered through the umbilical catheter. She also required two blood transfusions through a scalp vein catheter. Throughout the stay in NICU the newborn was treated with open exposure in a humid incubator. As and when there was more erosion of skin, she was wrapped with non-adhesive dressings and tubular bandages. Inspite of all supportive measures the baby did not survive and succumbed on the 22nd day of life.

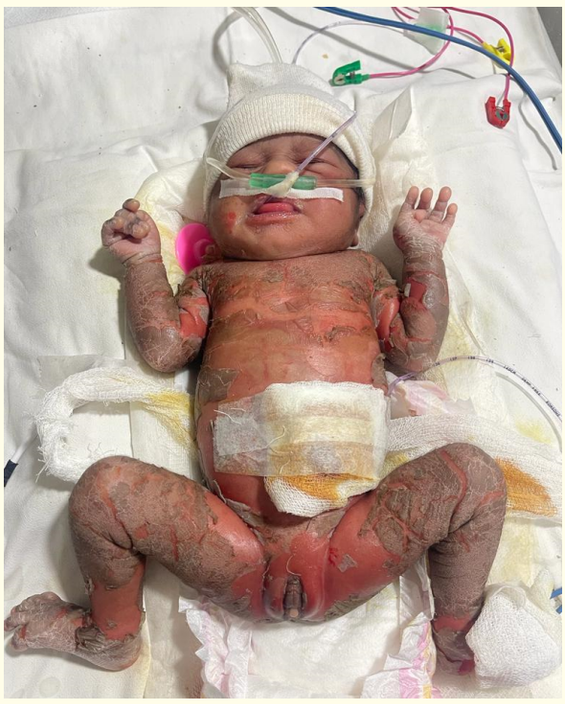

Figure 3

Whole exome sequencing revealed pathogenic variant of Bullous congenital ichthyosiform erythroderma, autosomal dominant inheritance. Her parents were explained about the genetic association of disease and advised with genetic work up. However, the couple refused any further workup or investigations. As this was an IVF conception, the frozen embryos of the couple are still available. If they undergo further testing, we will be able to find out the exact gene defect and the carrier partner. Depending upon the results of these tests, the embryo without the abnormal gene can be identified by PGD (pre implantation genetic diagnosis) and transferred in future cycle. The other option is finding the defective gene in either of the parents and if required using third party reproduction to prevent this occurrence in future.

Discussion

Inherited epidermolysis bullosa (EB) is a rare disease with significantly different clinical presentation and prognosis. It occurs equally commonly in males and females. Establishing an accurate diagnosis is important for genetic counselling and further management. The uncertain outcome and the excessive crying of the newborn due to pain is stressful for both the mother and healthcare provider. These newborns need a collaboration between the dermatologist and NICU specialist as it's a rare occurrence. Diagnosis is of utmost importance as prognosis depends upon the variant of disease. Generally a skin biopsy for immunohistochemistry with transmission electron microscopy and Sanger sequencing of candidate genes clinches the diagnosis.

Prenatal diagnosis is mandatory prior to embryo transfer in such cases. Unfortunately there is no definitive treatment for this condition. There have been a few case reports about such condition in newborns. In a similar case report by Martin Lehmann Boesen et al the baby was diagnosed with EBS, severe generalized (earlier called Dowling Meara) based on the clinical picture and skin biopsy [5]. He was discharged at the age of 6 months of age and at the time of publication of article (2016) he was 2 years old with no need for medical pain relief. However he still has episodes of pain while bathing and playing. Management involves wound care, control of pain, prevention and treatment of infections, nutritional support and management of complications. Supportive treatment can prolong the survival [5,6]. Therefore genetic work up should be considered especially in families with previous history for genetic disorder.

Conclusion

Diagnosis of this condition is challenging. Management largely depends upon the type of lesion. These may require extensive therapy throughout their life. But most babies succumb due to the severity of the illness and the survival rate is quite less. Also the quality of life is severely compromised. If we are able to find out the exact gene defect or mutation in the index case, as well as the parents, therapy can be targeted accordingly. Also with the help of IVF and PGD (pre implantation genetic diagnosis) one can transfer only those embryos which are genetically normal.

Acknowledgements

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