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A Rare Case Report of a Collodion Baby and Its Sibling with Lamellar Ichthyosis

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Abstract

Ichthyoses are disorders characterised by presence of fish like scales. They are either inherited or acquired. Lamellar ichthyosis (LI) is one among the inherited variants with skin being covered by large, thick, polygonal scales. We report a female neonate who presented to the neonatal clinic with collodion membrane at birth and her sibling a 6 year old male child who presented with large plate like scales, ectropion, and corneal opacity. The older child's skin biopsy demonstrated hyperkeratosis. Because of its rare occurrence, we report this with review of literature.

Keywords: Lamellar ichthyosis, collodion

Introduction

Lamellar ichthyosis (LI) is a rare autosomal recessively inherited disorder with an incidence of 1 per 1,00,000 to 3,00,000. There is mutation of TGM1 gene causing loss of transglutaminase1 activity. Affected babies are born with colloidal membrane. This is shed in about 10-14 days after birth. LI has defective cornification barrier of skin. Collodion baby is associated with very high morbidity and mortality in the neonatal period. Intensive symptomatic care consisting maintenance of oxygen saturation, body temperature, adequate feeding and skin care are essential.

Case Report

A female neonate second born to second degree consanguineous parents through lower segment caesarean section at 36 weeks of gestational age presented with collodion membrane (figure1), with a birth weight of 2500 gm., head circumference of 33 cm, length of 51 cm. Examination revealed presence of abnormal parchment like membrane with broad thick scales all over the body. Apgar score was 6/9/9 at 1,5,10 minutes respectively. There was eclabium due to excessive scaling around the mouth giving the child's mouth a fish like appearance. No other obvious developmental anomalies were seen. Systemic examination was normal. No significant antenatal events. The neonate was admitted in the NICU and managed with adequate humidification in incubator, covered with polythene wraps, intravenous (IV) fluids, IV antibiotics, emollients, lubricant eye drops, oro-gastric tube feeding which was changed to oral feeding after 3 days. Oxygen supplementation with nasal prongs was given for 3 days before slowly weaning off to room air. The infant was closely monitored for sepsis and respiratory distress. She was discharged after 9 days. Detailed family history revealed that her elder sibling, now a 6 year old male child, first born to her parents had

similar history of collodion membrane at birth. Examination of the boy revealed large plate like scales, resembling fish skin (figure 2), ectropion and corneal opacity of right eye. He was treated with emollients, lubricant eye drops. Complete blood count, serum electrolytes, liver and renal function tests were normal for both siblings. Skin biopsy of the elder sibling showed hyperkeratosis (figure 3). The elder sibling was referred to the ophthalmologist for corneal transplant. Both the siblings were advised close follow-up but didn't turn up due to parents' logistic concerns.

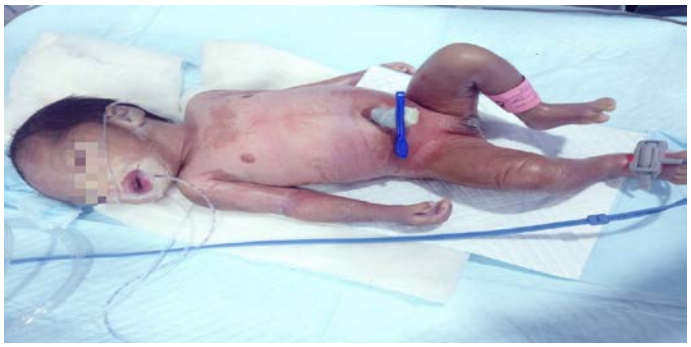

Figure 1

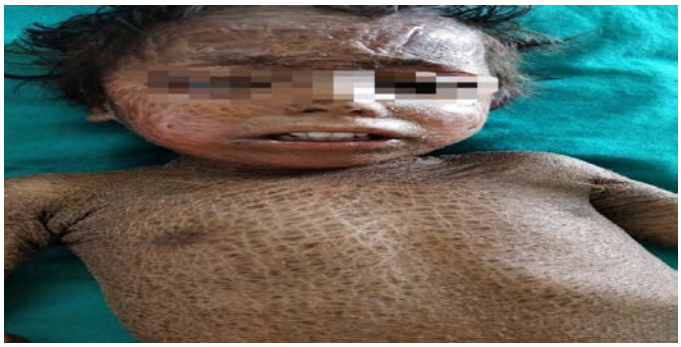

Figure 2

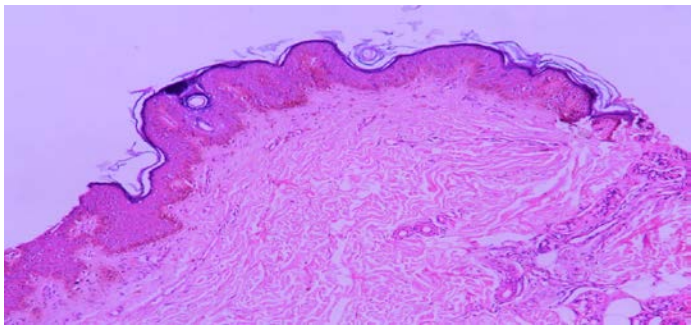

Figure 3

Discussion

Hallopeau and Watelet first used the term collodion baby in 1884(1). LI is an autosomal recessive disorder also known as non-bullous congenital ichthyosis, non-erythrodermic autosomal recessive lamellar ichthyosis, classic LI and ichthyosis lamellaris(2). Infants with lamellar ichthyosis are often born preterm and are covered with a taut membrane resembling a collodion, hence the term "collodion baby".

LI need to be differentiated from a relatively commoner condition Nonbullous Congenital Ichthyosiform Erythroderma (NBCIE). While LI is characteristically has large brown adherent scales, rippled hyperkeratosis around the joints, frequent palmo-plantar keratoderma and minimal erythema, NBCIE has fine diffuse branny scaling and generalised erythema. LI is said to be due to abnormal keratin retention whereas NBCIE is due to excessive proliferation of keratin. Presence of TGM1 mutation on chromosome 14q11.2 can help diagnose LI(3). Pena et.al. studied Filaggrin expression in scales using immunohistochemistry and western blot and determined that Filaggrin expression is variable in patients and higher expression correlated with better prognosis. They suggested its use a prognostic marker(4).

Foetal DNA analysis can be offered to parents who had previous child affected with LI. Foetal genomic DNA can be obtained from amniotic fluid via amniocentesis and chorionic villus sampling.

As a neonate these young patients are susceptible to dehydration, hypothermia, electrolyte imbalance due to insensible fluid loss. Symptomatic management should include humidified incubator, nutrition support, regular monitoring of electrolytes, urine output, liver and renal functions to name a few. Susceptibility to infections persists throughout life and relevant care is to be

discussed with the family. Maintain skin hydration and lubrication by topical petrolatum, urea creams to soften the scales, etc. Topical keratolytic agents remove scales by keratin dehiscence and include 5-10%glycolic acid, 40-60% propylene glycol, and 1-5% salicylic acid. Care should be taken to prevent drug salicylate overdose(5). Topical antimicrobials help reduce infections.

Topical retinoids like tretinoin used as 0.01% gel make keratinocytes less adherent. Oral retinoids if used in long term cause significant adverse effects and are used in refractory patients. Etretinate is a commonly used oral retinoid in a dose of 1-2mg/kg/day.

Hofmann et al used Tazarotene, a receptor selective retinoid, is a topical (0.05% gel) prodrug whose metabolite acts by modulation of proliferation and differentiation of epithelial tissue with possible anti-inflammatory and immunomodulatory effects. It can also be used for treating ectropion(6).

Long term treatment side effects of retinoids include skeletal abnormalities like hyperostosis, slender long bones, ligament & tendon calcification and premature epiphyseal closure. Late effects of LI include eye complications like ectropion, lagophthalmos, exposure keratitis, corneal scarring and ulceration. They are managed with lubricating eye drops, corneal transplant etc. as needed. Ear canal can be blocked by scales and requires regular ENT visits to prevent clogging and conductive hearing loss.

Conclusion

Lamellar Ichthyosis is a rare disease, which requires proper follow up and management of complications. Topical moisturisers and retinoids are common modes of treatment. Family counselling for genetic analysis in prior history of LI is advised. Encouragement and motivation for the family through support groups come a long way in improving the quality of life both for the child and the parents.

Multidisciplinary team including but not limited to Neonatologist, Dermatologist, Ophthalmologist, Otolaryngologist, Orthopaedician, nutritionist and physical therapist are to be involved for a holistic approach in the management of LI.

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