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Ectropion in a collodion baby: A case report and literature review

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Abstract

Background: Collodion baby is a severe form of congenital ichthyosis with neonatal onset. The clinical picture is often characteristic. When it is not fatal, the disease usually progresses to ichthyosis sicca. Thanks to molecular biology techniques, prenatal diagnosis is now possible from the 10-12th week of amenorrhoea, enabling genetic counselling. The prognosis for the disease depends on a number of parameters, including the degree of initial involvement, the duration of desquamation and the underlying ichthyosis.

Case: We report a case of a 7-day-old baby who was having severe bilateral ectropion of the upper lids without corneal involvement. There was peeling of the skin, more over the chest around the neck region and over the flexor aspect of limbs.

Conclusion: Management of collodion baby requires a multi-disciplinary approach.

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Keywords: Ichthyosis; Collodion baby; Congenital dermatosis; Ectropion.

Introduction

The "collodion baby" phenotype is a rare clinical entity in a heterogeneous group of cornification skin disorders, first described by Hallopeau and Watelet in 1844, with an estimated incidence of 1 in 50,000 to 100,000 live births. The collodion phenotype is common to several different forms of autosomal recessive congenital ichthyosis, characterised by xerosis of the skin, either in isolation or in association with other extra-cutaneous signs, particularly ophthalmological, which may affect the functional prognosis as a result of blinding corneal sequelae. We report a rare case of congenital ichthyosis with ophthalmological involvement.

Case description

A male neonate was born at term by spontaneous vaginal delivery to a healthy 31-year-old woman, G2P2. The mother received routine antenatal care and all antenatal laboratory and fetal ultrasound results were within normal limits. There was no consanguinity or family history of skin disorders. Birth weight was 2300 g, and birth length could not be measured correctly due to limited extension of the lower limbs.

On examination, there was white scale present all over body with peeling of skin more over the chest just below neck. On ocular examination, severe ectropion of both upper lid was present as well as abundant hyaline type ocular secretion (Figure 1). Cornea was not involved and pupil was of normal size and normally reacting to light. Lens was clear and red glow was present. Fundus examination of both eyes was within normal limits. The child was advised a lubricating eye drop along with antibiotic eye drop. We wanted to evaluate the child for ectropion but the patient lost to follow up.

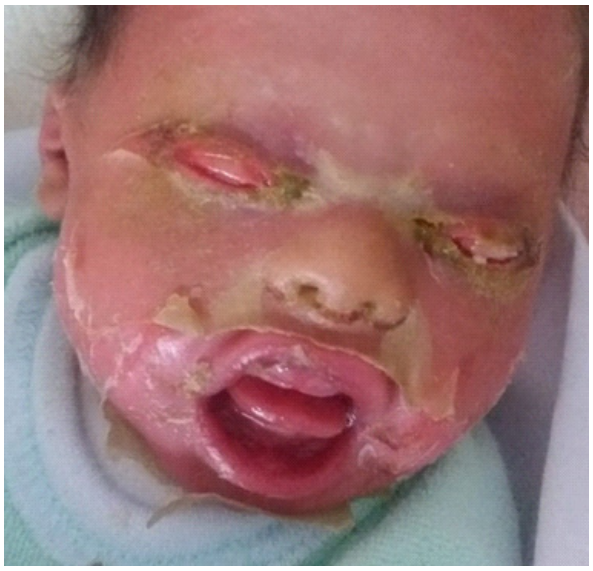

Figure 1: Severe ectropion in a collodion baby.

Discussion

Collodion baby is an infrequent clinical presentation of numerous genetic conditions, particularly disorders of cornification [1]. Most commonly these babies have autosomal reces¬sive inheritance owing to mutations in genes TGM 1, ALOXE3, or ALOX12B [2]. The positive diagnosis of collodion baby, as well as differential diagnoses, is primarily clinical. If there is any doubt, a skin biopsy will confirm orthokeratotic hyperkeratosis. The recurrence of this syndrome in a previously unaffected family points to an autosomal recessive genetic disorder. The pathogenesis of the syndrome is poorly understood. Certain socio-cultural specificities are at the root of a high rate of consanguinity and a higher frequency of these hereditary disorders of keratinisation. At birth, the collodion baby presents with a parchment like shiny tight skin covering the whole body. A minority of baby collodions associated with a keratinocyte transglutaminase deficiency due to a mutation in transglutaminase 1 are self-limiting [3]. This variant is inherited in an autosomal recessive manner, and one case caused by a particular mutation with minimal keratinocyte transglutaminase deficiency has been reported [4]. The course of the disease is highly variable and unpredictable. Congenital collodion baby has been reported in 53% of dry congenital erythroderma and in 12% of lamellar ichthyosis. In 10% of cases, collodion baby may be the first sign of ichthyosis vulgaris. In 10% of cases, it is a self healing collodion baby, while in 20% of cases, it may be Gaucher disease or Trichothiodystrophy showing a collodion baby phenotype at birth [5,6].

The collodion baby is the congenital and initial expression of many forms of ichthyosis, but it does not prejudge severity. The newborn presents with skin resembling desiccated collodion covering the entire epidermal surface. Exfoliation begins early with cracks in the collodion membrane. The cracks may remain superficial, or deepen and affect the superficial dermis, creating fissures [3]. The tight skin causes mechanical compression leading to alteration of facial features and extremities. Ocular manifestations of ichthyosis vary according to the type of ichthyosis (Ena, Pinna, 2003). Scales on eyelashes and eyelids may be seen in all varieties. However, the tight collodion membrane covering the newborn and producing ectropion of lids is characteristically found in lamellar ichthyosis. Ocular manifestation include bilateral ectropion of lower eyelid, chronic blepharitis and rarely cataract. Other signs like eclabium, pseudo contractures, absence of eyebrows, sparse hair and hypoplasia of nasal and auricular cartilage encountered in these babies are a result of the distortion [4]. The impaired skin barrier function puts these babies at a risk for various complications, including dehydration, hypothermia, infections and sepsis [3,4]. Advances in neonatal care have significantly improved the prognosis of these babies. Hence, prompt initial diagnosis and multidisciplinary involvement can optimize the outcomes.

In our case, the baby was referred for ophthalmologist opinion one week after birth, when ectropion became very evident. Cicatricial ectropion is the most common ocular abnormality of collodion baby. Ectropion of the eyelid, upper eyelid restriction, lack of Bell's phenomenon, and eyelash retraction are assumed to cause exposure keratopathy in severe cases. In our patient no signs of exposure keratopathy were seen despite the presence of ectropion. A good Bell's phenomenon along with upper eyelid movement could have attributed to this. Hence, conservative management was considered to deal with ectropion. Early initiation of oral acitretin, a second generation oral retinoid is also highly beneficial. Acitretin induces keratinocyte differentiation in epidermal layers and accelerates shedding of hyperkeratotic skin [7,8]. It also has additional anti-inflammatory, anti-neoplastic and wound-healing effects, Artificial tears if there is severe ectropion. The optimum results obtained with conservative management, highlights the importance of early diagnosis and prompt ophthalmology care in these babies. Sight threatening complications like exposure keratopathy, corneal perforation and the need for surgical interventions could thus be averted.

Conclusion

In conclusion, a clinical diagnosis of collodion baby based on phenotype alone is sufficient instead of a definitive diagnosis. Management requires the expertise of a dermatologist and the pediatric team. Other specialists that may need to be involved include ophthalmologist, geneticist and physiotherapist.

Declarations

Conflicts of interest: The authors have no conflicts of interest to declare.

Sources of funding: We have any financial sources.

Ethical approval: Not required.

Consent: Written informed consent was obtained from the patient's parents for publication of this case report and accompanying image. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

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