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Case Report

Malignant Congenital Keratoma: Which Prognosis?

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Abstract

Malignant Congenital Keratoma (MCK) is a rare autosomal recessive disorder with poor prognosis. Affected subjects have hard skin with deep fissures. Survivals rarely exceed one week due to respiratory and infectious complications.

We report the case of an affected still born foetus observed in the Center for Maternity and Neonatology of Tunis, Tunisia in order to give an anatomic description and determine the interest of foetopathology exam of affected foetuses.

A 28 Week Gestational age still born foetus presenting with craniofacial dysmorphic features: dolicocephalia, hyperthelorism, ectropion and aclabium associated with bilateral cataract. The skin was hard and waxy and had multiple fissures most prominent over areas of flexion leading to the limitation of the articulations. The dissection did not reveal other abnormalities.

The diagnosis of kdm was retained. Because of the severness of this pathology, genetic counselling is indicated if there is a history of consanguinity and a previous case in other siblings.

Keywords: *Malignant Congenital Keratoma; Foetus*

Introduction

Malignant Congenital Keratoma (MCK) is a rare autosomal recessive disorder with poor prognosis. Harlequin fetus, with an incidence of about 1 in 300.000 births [1], affected newborn, have hard skin with deep fissures. In the literature, prenatal diagnosis is recommended using a fetal skin biopsy, mainly at approximately 23 weeks of estimated gestational age. Survivals rarely exceed one week due to respiratory and infectious complications [2,3].

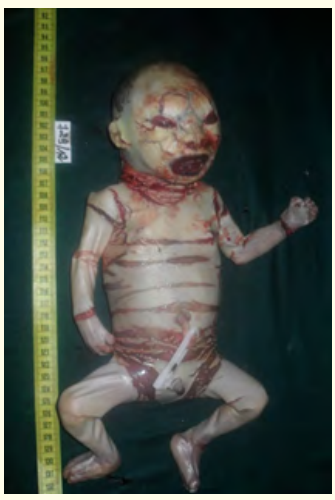
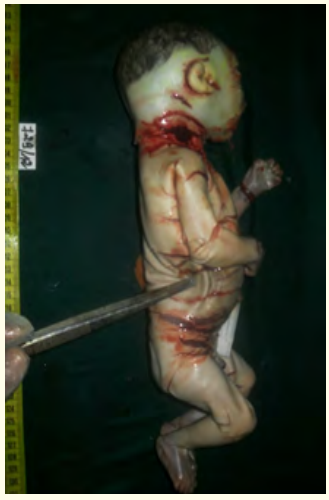
Objective

We report the case of an affected still born foetus observed in the Center for Maternity and Neonatology of Tunis, Tunisia in order to give an anatomic description and determine the interest of foetopathology exam of affected foetuses.

Case Report

We report the case of a male newborn product of a consanguineous marriage.

The mother has a 27 year old, 5th gravida 2nd para. Her first baby died of MCK. A pregnancy very poorly followed, the newborn of a 28 Week Gestational age presented with craniofacial dysmorphic features: dolicocephalia, hyperthelorism, ectropion and aclabium associated with bilateral cataract. The skin was hard and waxy and had multiple fissures most prominent over areas of flexion leading to the limitation of the articulations. The dissection did not reveal other abnormalities. The diagnosis of MCK was retained.

Figures 1 and 2: *Fetopathological examination.*

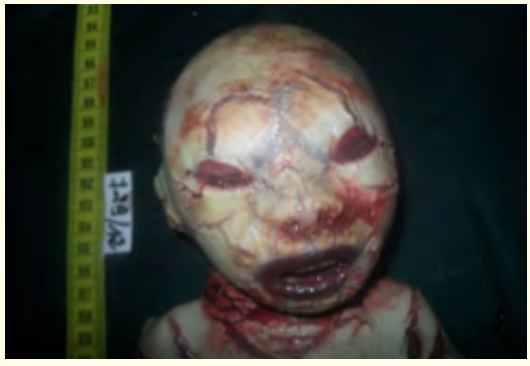
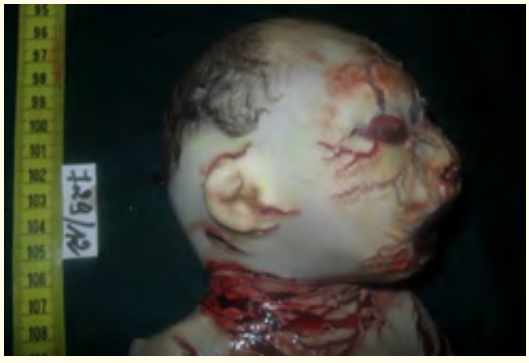



Figure 3 and 4: *Examination of the face: Ectropion, the skin is carded.*

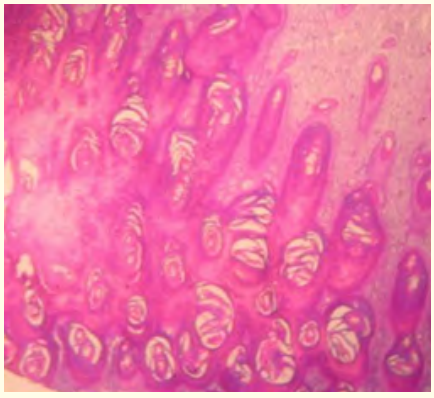
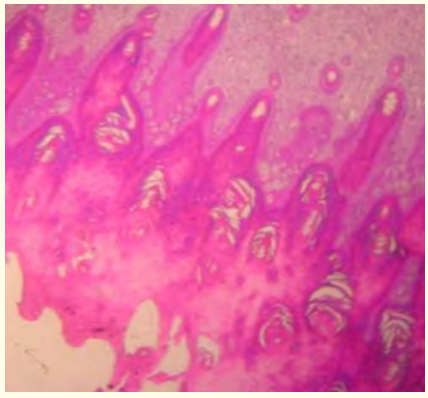



Figure 5 and 6: *Histological examination: Hyperkeratosis, thickening of the stratum corneum.*

Discussion

Collodion baby or Harlequin ichtyosis is a skin disease that reveals at birth by a disseminated hyerkeratosis charecterized by dry scaling skin with deep fissures especially in areas of flexion in limbs that compromise movements, suction is also compromised [4], this limitation of the articulations can be seen in other pathology such as arthrogryposis. Ears and nose are flat and underdevelopped with eclabium. Extremities have poor distal and digital development and fingers can be necrotic. Eversion of eye lids (ectropion), palpebral chemosis, cornea alteration and sometimes cataract can be observed [5]. Respiratory and infectious complications and feeding challenges can lead to death, it occurs most frequently during the first two weeks of life [6].

The disease is caused by the mutation of ABCA2 resulting in the loss of function of the lipid transporters in the lamellar granules of Keratine cells [7,8,9].

The treatment is symptomatic. Some authors report the effectiveness of Eternitate oral treatment in association with neonatal reanimation limiting dehydration [9]. Nevertheless, this evidence is unconclusive due to the fact that the number of treated patients is too little [10]. The prognosis remains very poor.

Conclusion

Because of the severness of this pathology and in the presence of a history of consanguinity and a previous case in other siblings, rigorous follow - up of the pregnancy and a relentless diagnosis must be imposed in order to adapt the most appropriate care.

Knowing that, the prognosis of these infants can be improved with intensive support measures as well as the addition of retinoids, remains that prenatal DNA-based diagnosis [9] for Harlequin ichthyosis in the early stages of pregnancy is the best way to ensure a lower risk for the fetal and a better psychological prognosis for mothers.

Conflict of Interest

We declare that we have no conflict of interest.

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