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Case Report

# Harlequin Fetus: A Rare and Unique Ichthyosis. A Case Report at the Sikasso Hospital (Mali)

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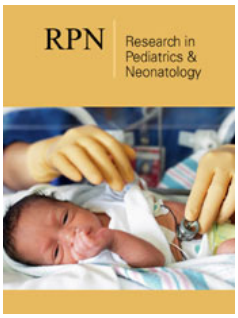


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**Abstract**

Ichthyosis is clinically characterized by the existence of visible flakes on the skin. The four main hereditary ichthyosis are: ichthyosis vulgaris autosomal dominant transmission, X-linked ichthyosis, lamellar ichthyosis, and congenital ichthyosiform necrolysis. At birth, the ichthyosis can be unapparent or realize collodion baby or Harlequin fetus. The Harlequin fetus is the most severe form and usually lethal ichthyosis. It is transmitted as an autosomal recessive. This is clinically a rare form of skin disease with large, thick, and yellowish scales, separated by deep red crevices giving the appearance of a baby frog. We here review a rare form of Harlequin fetus admitted to the neonatal unit at day 1 of life.

**Keywords:** Harlequin fetus; Ichthyosis; Newborn

## Introduction

The term ichthyosis comes from the Greek word ichthyus: fish scale. It brings together a heterogeneous set of congenital or acquired pathologies characterized by cutaneous xerosis with excessive desquamation, skin inflammation [1-3]. They are distinguished by their mode of transmission, age of onset, clinical appearance and causal genetic abnormality [1-3]. The baby Harlequin is an extremely serious and often lethal form of ichthyosis, its incidence is approximately 1 in 300,000 births, it is transmitted in an autosomal recessive manner by mutations in the ABCA12 gene [3,4]. This rare ichthyosis is clinically manifested from birth by an appearance of skin enclosed in large, thick, yellowish scales, separated by deep red crevices [4].

## Observation

This is a newborn female child admitted on the first day of life, third sibling, from the Sikasso region. Third administrative region of Mali (South of Mali), of first-degree consanguineous parents, in the history we find: a pregnancy not followed and carried to term, the infectious anamnesis was negative with an absence of medication or toxic substances. during pregnancy, an absence of similar cases in the family and vaginal delivery. The somatic examination showed a weight of 3.5kg, a height of 50cm, a head circumference of 34cm, a heart rate of 140 beats/minute, a respiratory rate of 32cycles/minute and pulsed oxygen saturation. at 98%. From birth, mucocutaneous examination showed skin with large, thick, yellowish scales separated by deep red crevices (Figure 1), a particular facies with splitting and ectropion, and edematous extremities (Figure 2). The biological assessment including a normal blood count, a normal ionogram and a C reactive protein at 12mg/ml. An x-ray workup including chest x-ray, abdominal ultrasound, cardiac ultrasound and transfontanellar ultrasound were normal. Given the severity of the skin involvement and the resulting dehydration, our patient received symptomatic treatment and broad-spectrum antibiotic therapy (central route

given the difficulty of taking the peripheral route). The evolution was marked by the worsening of the clinical picture despite well-conducted treatment, resulting in death at one week of life in a picture of severe collapse. The genetic study was not carried out due to lack of funds, genetic counseling was offered to parents, and counseling for monitoring future pregnancies.

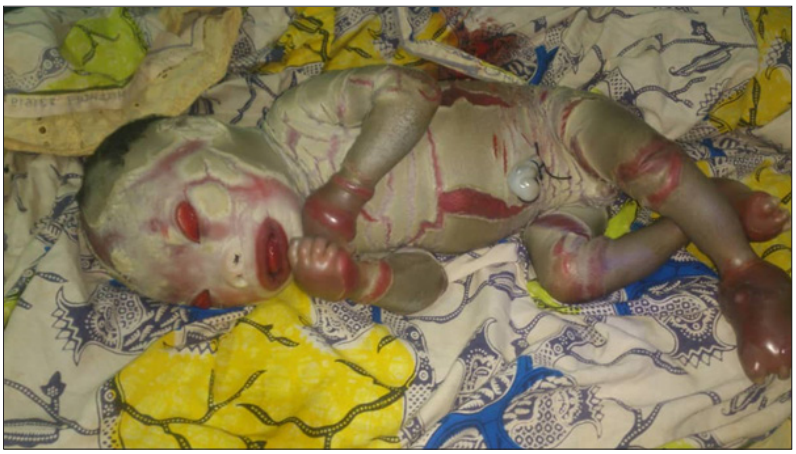

**Figure 1:** The baby harlequin showing skin with large, thick, yellowish scales separated by deep red crevices.

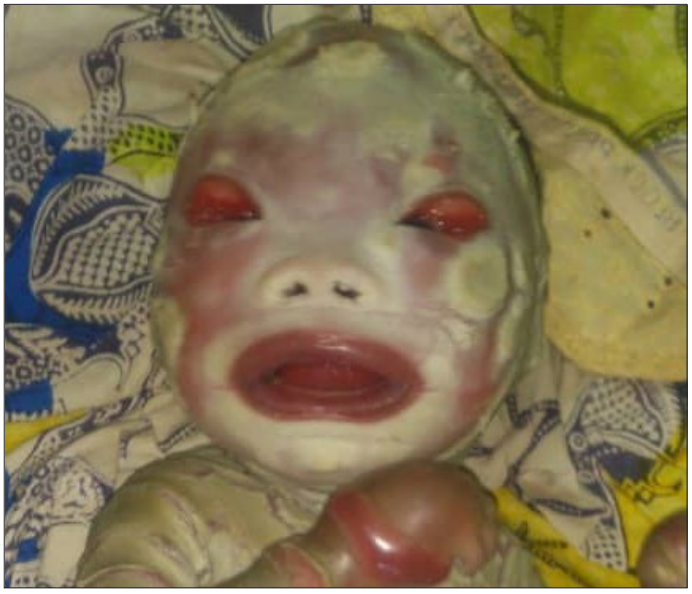

**Figure 2:** The baby harlequin with characteristic eversion of the eyelids (ectropion) and characteristic eversion of the lips (eclabion), ears and nose.

## Discussion

Ichthyosis is clinically characterized by the existence of visible scales on the skin. The positive diagnosis of hereditary ichthyosis was easy given the scaling appearance of our patient's skin. An ichthyosis is first defined on clinical examination by [1,3,5]: the appearance of the scales (size, color, thickness); retention (respect for large folds) or proliferative (non-respect for large folds); the existence of erosion or bubbles. The other elements used to classify the different forms are the age of onset, the type of inheritance. The typing of the different forms still needs to progress with the study of the biological abnormalities involved and the characterization of the genetic abnormalities which are already specified in certain cases [2,3,5].

Characterization of the type of ichthyosis was more difficult, due to the technical and financial difficulties in obtaining pathological examination and genetic tests. According to the consensus meeting of international ichthyosis experts in Sorèze in 2009 [1,2,7], the decision tree remains based on easily accessible clinical and paraclinical data: syndromic or not, semiological characteristics, type of transmission. , age of onset. The involvement of the major folds (proliferative hyperkeratosis) led us to rule out the diagnosis of ichthyosis vulgaris and the female involvement that of X-linked

ichthyosis. The absence of erythroderma led us to rule out the diagnosis of dry congenital ichthyosiform erythroderma (EICS). The appearance of the scales (large adherent scales), and their generalized arrangement with involvement of the large folds and the absence of erythema, led us to classify our patient in the group of lamellar ichthyosis proper. Harlequin ichthyosis is the most serious and often fatal form. It is inherited autosomal recessively [5-7]. This syndrome results clinically at birth by a newborn enclosed in large, thick, yellowish scales, separated by deep red crevices. The extreme skin tension is responsible for the particular frog face with characteristic eversion of the eyelids (ectropion), and also a characteristic eversion of the lips (eclabion), ears and nose. The extremities are swollen as a result of strictures by massive thickening of the skin as in our newborn [6-8]. Children alive at birth quickly die within days of respiratory, infectious or dehydration complications [8]. There is currently no specific curative treatment for ichthyosis. The care given several times a day, without curing, nevertheless allows an acceptable social life. Treatment is most often symptomatic only and consists of daily local care sometimes combined with systemic treatments. Multidisciplinary care, most often hospital, is necessary. Emollient and retinoid treatments are the treatment of choice for severe ichthyosis, their use improves skin condition by reducing hyperkeratosis and increasing heat tolerance, it prevents or improves ectropions [9]. This treatment requires close medical supervision [8,9].

Rajpopat S et al. [5] reported in a study of 45 cases of baby harlequin in 2011 that the overall survival rate was 56% in this study. Death usually occurs within the first three months of life from sepsis or respiratory failure in 75% of cases. The antenatal diagnosis of the baby harlequin is possible two methods are proposed [1,5,9,10]: the study of the amniotic fluid at 17 weeks of pregnancy can show lipid droplets characteristic of the baby harlequin and the examination by electron microscopy of the fetal skin biopsy performed between 19 and 23 weeks of intrauterine life show early hyper keratinization of the stratum corneum. A few authors have successfully diagnosed cases of baby harlequin with a family history of ichthyosis by prenatal 3D and 4D ultrasound; ultrasound signs were skin contracture, flattened ears, nasal hypoplasia, everted eyelids, a fish-like mouth, macroglossia, and a persistent open fetal mouth [11,12]. Given the catastrophic prognosis and the pain suffered by the child, when the diagnosis is made before birth, other teams in France evoke the possibility of a medical termination of pregnancy or else propose the implementation of focused palliative care. on pain management and comfort [13]. Likewise, palliative care is offered from the outset if the diagnosis is made at birth. This team reports the observation of a baby harlequin, diagnosed in utero at 32 weeks; the birth took place at 35 weeks old, the child lived for 12 hours in palliative care with his mother, in a mother-child room in the neonatal unit.

## Conclusion

Baby harlequin is a rare and severe form of congenital ichthyosis, which can be fatal through dehydration and infection. It requires multidisciplinary care from birth. Treatment with emollients and oral retinoids could improve patient survival, however medical termination of pregnancy or palliative care remains possible due to the extreme severity of the prognosis and the pain endured by the child.

## Parental Consent

Informed consent from the parents was obtained for the publication.

## Conflicts of Interest

The authors declare no conflict of interest.

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