OPEN ACCESS

Saudi Journal of Medicine
Abbreviated Key Title: Saudi J Med
ISSN 2518-3389 (Print) | ISSN 2518-3397 (Online)
Scholars Middle East Publishers, Dubai, United Arab Emirates
Journal homepage: https://saudijournals.com

Case Report

Recovery of an Orphanage Disease at the Markala Reference Health Center: The Harlequin Baby

Fomba, D[1], Kanthé, D[1], Koné, O[1], Samaké, Y[2], Dembélé, B. T[3], Kéita, M[1], Ongoiba, S[5], Théra, A[4], Bagayoko, T. B[6], O. Koné[7]

[1]Markala Reference Health Center, Mali
[2]Fana Reference Health Center, Mali
[3]University Hospital Center of Gabriel Touré and National Center for Scientific Research and Technology (CNRST), Bamako, Mali
[4]Department of Obstetrics and Gynecology, University Hospital Center of Point.G, Mali
[5]Gavardo Hospital of Bamako, Mali
[6]Forensic and Occupational Medicine Department of Nianankoro Fomba Ségou Hospital (Mali)
[7]National Institute of Public Health-Bamako, Mali

DOI: 10.36348/sjm.2023.v08i05.001 | **Received:** 08.03.2023 | **Accepted:** 17.04.2023 | **Published:** 05.05.2023

***Corresponding Author:** Fomba, D
Markala Reference Health Center, Mali

Abstract

The name ichthyosis comes from the Greek word for "fish" because sufferers can have dry, scaly skin. The HARLEQUIN Baby disease or congenital ichthyosis of the Harlequin fetus type is an orphan genetic disease that mainly affects babies and young children Harlequin Ichthyosis is a very rare genetic disease with an estimated prevalence of less than 1/1000,000 associated with significant morbidity (<50%) and mortality shortly after birth. Diagnosis is based on clinical examination (brown, scaly and very dry skin making it difficult for the baby to move), skin biopsy and analysis.

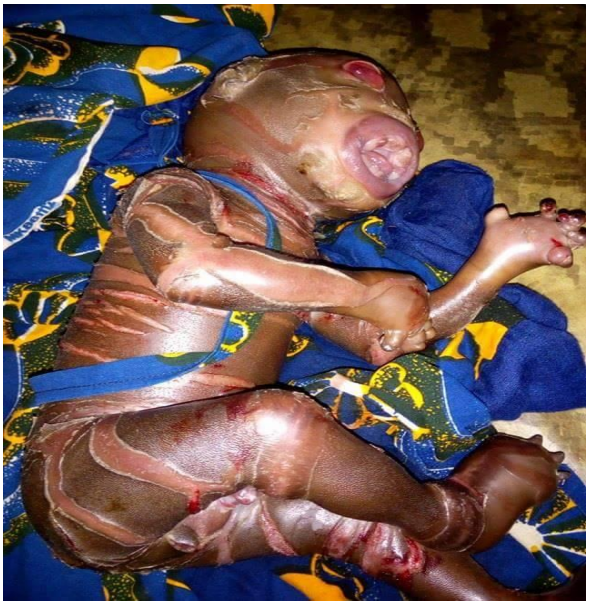
Keywords: orphan disease, rare genetic aberation, Markala-Mali.

INTRODUCTION

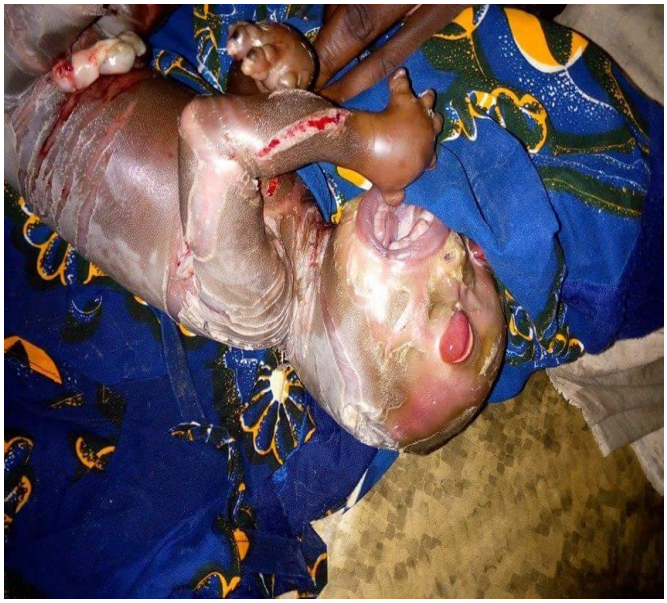
The HARLEQUIN Baby disease or congenital ichthyosis of the Harlequin fetus type is an orphan genetic disease that mainly affects babies and young children. The name ichthyosis comes from the Greek word for "fish" because sufferers can have dry, scaly skin. The disease causes hyperkeratinisation of the epidermis, so it becomes very thick and hard and cracks may appear, reminiscent of a harlequin suit, hence the name. There are many varieties of ichthyosis, but the congenital Harlequin fetus type is the most dangerous.

CLINICAL OBSERVATION

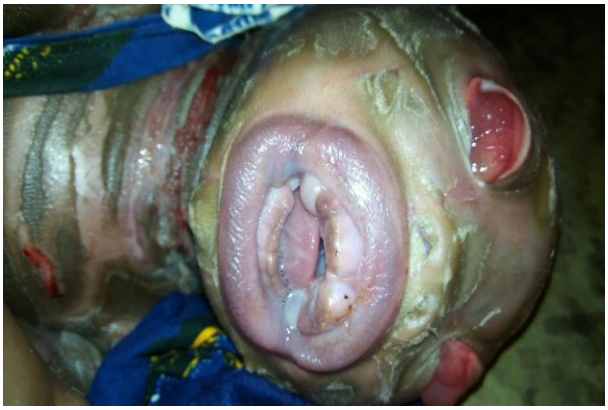
We report the medical file of a 30 year old woman, housewife G7P6V4D2, IIG 2 years old with no notable medical or surgical history referred by a 1st level structure for haemorrhage from an unattended pregnancy, oedema of the lower limbs and excessive HU at 38cm.On entry examination, the patient was in good general condition, good mucosal and phanerial colouration, good consciousness, BP: 130/80mmhg, T°: 35.8C, presence of oedema of the lower limbs, no jaundice or cyanosis, supple, anodular breasts with gravid appearance. Uterus with longitudinal development, HU: 38cm BDCF: 146 bats/min, 1CUD/10mn.TV: effaced cervix dilated to 2cm, water sac intact, breech presentation not engaged on a clinically normal pelvis.



Scaly rigidity of the labial mucous membranes preventing any closure of the mouth.



Significant inflammation of the conjunctivae also preventing the closure of the eyes



Complementary Examinations

Haemoglobin level: 10.2g/dl Rhesus grouping: O positive Proteinuria: None HIV: Negative Obstetric ultrasound: Monofetal pregnancy with evolving intrauterine malformation of 37SA +2 days with significant hydramnios.

Six hours after admission, our parturient delivered a male neonate, weighing 2500 gr, measuring 44cm with a PT at 30cm, a PC at 31cm; Apgar at the 1st minute 8/10 and at the 5th minute 10/10.The neonate presented the following characteristic : Fissured and bleeding scaly patches, covering the whole body and considerably limiting limb movement. Scaly rigidity of the labial mucosa preventing any closure of the mouth.Severe inflammation of the conjunctiva also preventing closure of the eyes.Limbs are shortened with malformed fingers and toes, seats of a generalized onycodystrophy. Malformation of the ears with retracted and very thickened auricles.

DISCUSSION

We found only one case of harlequin baby in the African literature, a case reported by the team of Pr A Es SEDDIKI from Morocco who described for the first time in February 2016 a case of neonatal ichthyosis in the Berkane region. Ichthyosis characterized clinically by the existence of visible scales on the skin [1]. Main hereditary ichthyosis: Ichthyosis vulgaris with autosomal dominant transmission,Lamellar ichthyosis and congenital bullous ichthyosiform erythroderma.Other ichthyosis remain localized: Pityriasis rotunda, cocoon genodermatosis, erythrokeratoderma variable, chondrodysplasia punctata and CHILD syndrome [1-3]. Recently, a new classification of ichthyosis was developed at the First Ichthyosis Consensus Conference in SORENEZIA [7], based on clinical data, and distinguishes between non-syndromic ichthyosis (affecting only the skin) and syndromic ichthyosis (also affecting other organs) [7]. Baby Harlequin is the severe and often lethal form. It is transmitted in an autosomal recessive manner [5-7]. The skin of the newborn is characterised by the presence of

large, thick, yellowish scales separated by deep red crevices. The extreme skin tension is responsible for the distinctive Harlequin Baby facies with characteristic eversion of the eyelids (ectropion), and also characteristic eversion of the lips (eclabion), ears and nose. The extremities are oedematous due to strictures from massive skin thickening as in our newborn [6, 7]. Live births die rapidly within days from respiratory complications, infection or dehydration. Recently, mutations in the adenosine triphosphate binding cassette A12 gene (already implicated in some forms of lamellar ichthyosis) have been reported. It is a rare form of ichthyosis, clinically manifested by skin covered in large, thick, yellowish scales, separated by deep red cracks.It is a very rare genetic disease associated with a very high morbidity (<50%) and mortality soon after birth.Prenatal screening offered to parents whose family has a history of the disease allows for its effective prevention.Treatment is based on the management of infections, dehydration, or respiratory distress.

CONCLUSION

Harlequin Ichthyosis is a very rare genetic disease with an estimated prevalence of less than 1/1000,000 associated with significant morbidity (<50%) and mortality shortly after birth. Diagnosis is based on clinical examination (brown, scaly and very dry skin making it difficult for the baby to move), skin biopsy and analysis.

Conflict of Interest: None

REFERENCES

1. Seddiki, A. E., Messaoudi, S., & Amrani, R. (2016). Bébé Arlequin: une ichtyose rare et unique. À propos d'un cas. *Revue de médecine périnatale*, *8*(2), 118-121.
2. Rodríguez-Pazos, L., Ginarte, M., Vega, A., & Toribio, J. (2013). Autosomal recessive congenital ichthyosis. *Actas Dermo-Sifiliográficas (English Edition)*, *104*(4), 270-284.
3. Chiavérini, C. (2009). La Société française de dermatologie pédiatrique. Ichtyoses génétiques. *Ann Dermatol Venereol*, 136, 923-934.
4. Harvey, H. B., Shaw, M. G., & Morrell, D. S. (2010). Perinatal management of harlequin ichthyosis: a case report and literature review. *Journal of Perinatology*, *30*(1), 66-72.
5. Rajpopat, S., Moss, C., Mellerio, J., Vahlquist, A., Gånemo, A., Hellstrom-Pigg, M., ... & O'Toole, E. (2011). Harlequin ichthyosis: a review of clinical and molecular findings in 45 cases. *Archives of dermatology*, *147*(6), 681-686.
6. Martínez-García, S., Vera, A., & Romero, J. (2003). Feto Arlequin. *Actas Dermosifiliogr*, 94, 392-394.
7. Oji, V., Tadini, G., Akiyama, M., Bardon, C. B., Bodemer, C., Bourrat, E., ... & Traupe, H. (2010). Revised nomenclature and classification of inherited ichthyoses: results of the First Ichthyosis Consensus Conference in Sorèze 2009. *Journal of the American Academy of Dermatology*, *63*(4), 607-641.