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Case report

Recognition and management of congenital ichthyosis in a low-income setting

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Accepted 26 June 2019

SUMMARY

We report the case of a 3-week old girl in The Gambia who presented to hospital with an undiagnosed skin disorder evolving since birth. Using telemedicine to seek specialist dermatology advice abroad, she was diagnosed with and managed for suspected congenital lamellar ichthyosis. Poor early recognition and limited resources, for both acute and chronic care, created significant challenges to optimal management; these were overcome, in part, by adopting a common sense, back-to-basics approach to treatment and by empowering the parents to take ownership of their infant's daily skin and eye care. This case highlights key global health issues associated with managing chronic, often debilitating, paediatric dermatological conditions in a low-income setting; namely, poor access to important diagnostic tools and medications, lack of experience and expertise in the management of severe skin disease and its associated complications, absence of long-term community support, alternative health beliefs and risk of sociocultural stigma.

or postnatal complications or risk factors for sepsis. Abnormality of the skin was noted at birth although no treatment was initiated due to lack of recognition and understanding of the underlying condition. There was no reported family history of dermatological and/or inherited diseases; parents were non-consanguineous.

On examination, her skin showed thick fissured hyperkeratosis (figure 1). There was associated mild eclabium and more pronounced ectropion, with difficulty in eye-closure bilaterally (figure 2). Clinical findings were consistent with a congenital form of ichthyosis, the first such diagnosis to be made at this hospital. Unfortunately, no dermatology and limited ophthalmology expertise was available locally with no resources for genetic testing; this precluded accurate confirmation of the exact subset of the disease on-site, although lamellar ichthyosis (LI) variant was strongly suspected. Basic blood tests excluded electrolyte abnormalities and/or renal failure secondary to poor skin integrity and dehydration.

The baby was admitted to the ward for further management. Following informed consent from the mother, high-quality photographs were taken of the infant; along with a clinical summary, these images were shared electronically with a specialist tertiary paediatric dermatology team in the UK. The suspected diagnosis was discussed, and advice given on further management, specifically treatment aims, optimal nursing care and a recommended daily skin/eye care regime, taking into account the limited resources available.

Given the lack of access to retinoids, the mainstay of treatment in this context was supportive with strict hygienic nursing and application of the very limited topical agents available. Soft white paraffin was applied liberally every hour to reduce fluid loss, enhance skin flexibility and comfort and prevent fissures; staff and family were warned about the potential fire risk. Aqueous cream was used as a soap substitute and to cleanse the skin while zinc-oxide cream was applied to the nappy area. In the absence of an incubator, the baby was placed within a plastic sheet to avoid excessive heat loss. There was no access to a pressure-relieving mattress, therefore, the baby was turned regularly, and pressure areas were monitored to prevent sores and alleviate pain at the fissured sites. On the third day of admission, multiple widespread skin pustules were noted, although the baby remained systemically well. A course of intravenous ofloxacin was successfully completed for suspected superimposed bacterial infection.

BACKGROUND

The recognition and management of congenital paediatric dermatological conditions is difficult in low and middle-income countries (LMICs). This is compounded by alternative health beliefs and increased sociocultural stigma associated with highly visible skin disease, often leading to poor outcomes. From a global health perspective, there is a major disparity in the care patients receive in LMICs compared with higher-income settings. Dermatological conditions may also be overlooked despite the significant global burden of disease and its effects on quality-of-life. Lack of local access to necessary equipment and expertise often necessitates a common sense, back-to-basics approach. Recent advances in telemedicine, however, have enabled specialist opinion to be sought, thereby reducing the gap in knowledge and helping vulnerable groups across different settings. Tackling the poor healthcare of these patients is an important step in promoting health equality and improving the quality-of-life of children worldwide.

CASE PRESENTATION

A 3-week-old Gambian baby was brought by her mother to an urban hospital for review of an undiagnosed skin disorder present since birth. The baby had been born at term by vaginal delivery at a rural health centre with no antenatal, perinatal



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To cite: Saso A, Dowsing B, Forrest K, et al. *BMJ Case Rep* 2019;**12**:e228313. doi:10.1136/bcr-2018-228313



Figure 1 Hyperkeratosis, scaling and fissuring of the skin of a baby with suspected congenital ichthyosis.

In view of the ectropion and limited eye-closure bilaterally, the UK team recommended basic ophthalmic measures to prevent further damage and dryness, including 4 hourly saline washes, artificial tears and bilateral eye patches.

Of note, the neonate still had not regained her birth weight with 12% loss calculated. This was likely multifactorial, including difficulty in latching secondary to eclabium, development of secondary infection and fluid losses through the skin with subsequent dehydration. Hence, feeding support was initiated: breast feeding with top-ups of expressed breast and formula milk, initially by nasogastric tube. Basic mouth care was also applied daily to prevent a dry, cracked and sore mouth (exacerbating feeding difficulties).

The baby was an in-patient for 14 days with improvement in skin appearance, no septic deterioration and adequate weight gain. Prior to discharge, her mother was counselled on the



Figure 2 Facial manifestations of congenital ichthyosis: ectropium and eclabium.

importance of continuing regular emollient use and monitoring for complications. Parental skin and eye care training was also completed on the ward pre-discharge.

GLOBAL HEALTH PROBLEM LIST

1. Congenital ichthyosis (CI) is a chronic condition that occurs worldwide. Lack of early recognition and diagnostic tools can lead to poor outcomes, particularly in low and middle-income countries (LMICs).
2. Genetic testing of affected infants can help to identify the subset of disease and mode of inheritance but is not available in all settings.
3. In LMICs, poor access to resources and/or lack of expert guidance on how and when to perform supportive care may compromise the management of infants with CI.
4. Sociocultural stigma can arise and may exacerbate clinical outcomes in patients with chronic, debilitating skin conditions, particularly in settings with alternative beliefs on health and disease.
5. Absence of community support and long-term follow-up in LMICs further compounds the difficulties in managing chronic dermatological conditions.

GLOBAL HEALTH PROBLEM ANALYSIS

Congenital Ichthyosis (CI) is a chronic condition that occurs worldwide. Lack of early recognition and diagnostic tools can lead to poor outcomes, particularly in low and middle-income countries (LMICs)

CI represents a heterogeneous group of genetic skin diseases, primarily characterised by a defective skin barrier, hyperkeratosis and scaling, often associated with skin inflammation.^{1 2} Autosomal recessive CIs, including LI, are estimated to occur in approximately 1 in 100 000 to 1 in 300 000 births worldwide.³ There is a paucity of data regarding ichthyoses in LMICs and estimates of incidence specifically in Sub-Saharan Africa are not known; a limited number of published case-reports highlight the difficulties in diagnosis and management of more severe subtypes.⁴

CI is present at birth or within the early weeks of life. Diagnosis is based upon the integration of clinical history of the skin

abnormalities, including time-of-onset, pattern of development, natural evolution and family history; examination to identify key cutaneous and associated extra-cutaneous manifestations; and investigations, such as histopathological features on skin biopsy or genetic testing, if available.^{1 2}

In high-income settings, CI is often recognised promptly, particularly in centres with strong dermatological expertise. By contrast, early identification of CI remains a challenge in LMICs, including Sub-Saharan Africa, further exacerbated if a delivery has taken place at home or at a poorly equipped birth centre in a rural or community setting. Rates of home birth vary widely⁵; UNICEF recently estimated that, in Sub-Saharan Africa, up to 44% of women do not give birth in a health facility (2011–2016) and 44% of births are not attended by skilled health personnel (2013–2016).⁶ In these cases, CI may not be recognised nor appropriate action taken. There is also often a preference for and/or easier access to traditional community healers who are either unlikely to have any experience of these conditions, given their rarity, or may recommend measures that exacerbate poor outcomes.^{7 8} Delays in provision of supportive management increase the risks of dehydration, electrolyte imbalance, hypothermia, superimposed infection and further skin and eye damage. Minimal specific data are available on the morbidity and mortality related to LI subtype in LMICs; harlequin ichthyosis, a more severe form of CI, has been associated with a mortality rate of up to 44%, with most deaths occurring in the first 3 months of life primarily due to sepsis and/or respiratory failure.⁹

Skin diseases are the fourth leading cause of disability worldwide, excluding mortality, as estimated by the Global Burden of Disease Study 2013.¹⁰ Despite this, dermatology is not a priority within the global health agenda; there is a distinct lack of access to expertise, diagnostic tools and treatment, with limited WHO guidance on key dermatological conditions.¹¹

Recently, there have been significant international efforts to raise awareness of the burden of skin disease worldwide, expand the role of dermatology in global health discourse and allocate resources appropriately; for example, by encouraging key stakeholders to contribute to international policy-making and by improving evidence-based dermatology research across a variety of settings.¹¹ Education and training of healthcare workers is also of vital importance in tackling this global health problem. Dermatology training and research programmes for doctors and nurses are gradually being established in Sub-Saharan Africa; one successful example is the Regional Dermatology Training Centre in Tanzania which focusses on the ‘prevention, treatment and rehabilitation’ of skin diseases, leprosy and sexually transmitted infections.¹² Further initiatives, such as ‘The Global Baby Skincare initiative’ launched by The Excellence in Paediatrics Institute, aim to raise awareness of optimising skin health worldwide, specifically in early life, recognising the lack of internationally recognised guidelines within this area.¹³ The International Society of Dermatology has also produced a free-to-use, evidence-based dermatological public health syllabus, ‘Skin Care For All’, which aims to engage healthcare practitioners and promote community dermatology, particularly in LMICs.¹⁴ Empowering healthcare workers through training to recognise common and/or severe dermatological conditions and to seek appropriate help, aided by modern technology that may facilitate remote specialist collaboration (see below), has the potential to impact on both mortality and the burden of disability caused by chronic skin conditions.

Genetic testing of affected infants can help to identify the subset of disease and mode of inheritance but is not available in all settings

Genetic testing, if available, is increasingly becoming an important tool in the successful management of CIs. Its benefits are twofold: confirming the diagnosis and guiding parental counselling. Inherited ichthyoses are caused by mutations in genes encoding structural proteins or enzymes involved in epidermal homeostasis, with over 30 mutations currently identified.^{15 16} LI is inherited in an autosomal recessive fashion and is specifically a disorder of keratinisation, secondary to mutations in one (or more) genes, including TGM1, ABCA12, ALOXE3, NIPAL4, ALOX12B and CYP4F22.^{3 15–19} Mutations in the transglutaminase-1 (TGM1) gene are the most common, demonstrated in at least 50%–60% of affected individuals; ABCA12 gene mutations have also been previously reported among LI patients, including those with African backgrounds (although they are more common in harlequin ichthyosis).^{3 17 19}

Identifying the patient’s specific CI subtype and possible causative genetic mutation(s) can help to predict the evolving pathophysiology, severity of cutaneous and associated extra-cutaneous manifestations, suspected prognosis and mode of inheritance.¹⁵ This allows appropriate counselling of parents on clinical outcomes and on the risk of the condition recurring in future pregnancies.^{3 16 19} Carrier testing for at-risk relatives and antenatal testing for potentially affected pregnancies may be appropriate if specific CI-associated mutations have been identified within a family.¹⁸ CIs (including LI) are genetically heterogenous, however, and a clear genotype-phenotype correlation cannot always be established; mutations in the same genes can present with diverging phenotypes, conversely, mutations in multiple genes can give rise to overlapping phenotypes.^{3 20}

With our Gambian infant, confirmation of the underlying genetic cause (if it had been available) may have been informative, but is unlikely to have affected the prognosis, given the management constraints in this setting. Moreover, sophisticated genetic testing is often not feasible in LMICs; as such, a careful clinical assessment and revision of the infant’s family history (including parental consanguinity) is important, with subsequent discussion on the likely mode of inheritance. The risk of recurrence should be addressed with parents in a careful and sensitive manner, enabling access to family planning services if this is desired.

In LMICs, poor access to resources and/or lack of expert guidance on how and when to perform supportive care may compromise the management of infants with CI

As yet, no specific guidelines have been developed on the management of CIs in LMICs. Moreover, there is no gold-standard treatment available worldwide, although retinoids such as acitretin, given by the topical or, in severe cases, oral route, are often routinely used in high-income settings.²¹ Guidance from European countries also primarily focusses on the regular application of emollients, such as petroleum jelly, alongside topical humectant and keratolytic therapy.^{1 2} Supportive care with specialist nursing, often within neonatal high-dependency or intensive care units, and early engagement with a multidisciplinary team is emphasised.²

Many of these services may not be available in LMICs. Therefore, a common sense, back-to-basics approach should be adopted, given that even simple measures without specialist resources can dramatically improve outcomes and reduce common complications. This case emphasises that adhering to

a simple yet consistent skin and eye care regime, as well as an individualised feeding plan, is effective even within this context. Equally, taking the initiative to develop feasible, practical and appropriate alternatives should also be encouraged; where specialist equipment is limited, simple replacements can be developed, such as the use of plastic sheets/bags for neonatal nursing care and thermoregulation, instead of a newborn incubator.

Telemedicine has the potential to provide a further solution to this issue, enabling remote expert diagnosis and guidance. Above all, the use of smartphones with a good-quality camera, email and instant-messaging ensures a simple and cost-effective method of communication.²² This has applications in high-income and low-income settings and to both patient-to-doctor as well as doctor-to-doctor (or other healthcare professionals') consultations. Moreover, telemedicine is particularly relevant to specialities with 'visual' diagnoses, notably dermatology, exploiting the rapid growth in availability and affordability of smartphones (and internet access), even in LMICs.^{23–25} In a cohort of 600 patients presenting with skin lesions in a rural Egyptian setting, diagnosis via 'teledermatology' demonstrated high diagnostic concordance rates with face-to-face on-site patient review.²⁴ Care must be taken, however, to ensure all communication is through a secure server to protect confidentiality, and informed consent has been given by the patient and family prior to any collaborations being initiated.

Beyond this, telemedicine also faces significant technical challenges.²⁶ Although the exchange of images for an individual patient may be straightforward, coordinating multiple partnerships and sustaining a reliable system on a larger, more permanent scale is highly demanding. A 2016 review of teledermatology services set up in South Africa since 1999 found that only four still remained operational. There had been minimal escalation of these services with limited support from regional health departments and poor integration into routine healthcare practices.²⁷ Effective advice also depends on the quality of the information shared, particularly a detailed clinical background and accurate symptom description, which may be suboptimal depending on the experience of those in the field. Furthermore, the remote expert often does not receive feedback on the advice they have given, partly due to the difficulties in confirming the patient's diagnosis and/or poor follow-up of long-term clinical and social outcomes in LMICs; such a feedback loop could improve remote specialist assessment and management of similar cases in the future.²⁵ Finally, financial and legal frameworks may be complex and difficult to establish fairly and robustly, especially when institutions across different settings are involved. These frameworks not only enable the procurement and maintenance of required equipment, but also ensure that both remote and local staff are appropriately reimbursed and held accountable, protecting patient safety.^{26,27}

Sociocultural stigma can arise and may exacerbate clinical outcomes in patients with chronic, debilitating skin conditions, particularly in settings with alternative beliefs on health and disease

Clinicians should be sensitive to the sociocultural stigma of such a rare, highly visible condition; this is often more common within Sub-Saharan Africa, specifically rural or isolated communities, which may have poor health education and propagate alternative (often harmful occult) beliefs on health and disease.⁴ There is very little in the literature describing social attitudes to LI (and attempts to tackle them) in LMICs.^{4,28,29} Affected infants have been called 'snake children', due to the appearance of the

generalised scales on the skin, subsequently leading to social exclusion, even parental abandonment, lack of engagement in familial and educational activities and poor clinical and psychosocial outcomes.⁴ Furthermore, chronic skin conditions generally are associated with significantly reduced quality-of-life and work productivity worldwide.³⁰

Management of CI cases (and other chronic skin disease), therefore, must extend beyond the clinical aspects and provide psychosocial support for the patient and family. Parents should be counselled appropriately to dispel myths and avoid misconceptions, both during acute (in-patient) and long-term (outpatient) healthcare consultations. In the context of other inherited dermatological conditions in Sub-Saharan Africa, namely albinism, non-governmental organisations and charities have played a crucial role in raising awareness and promoting education and advocacy nationwide, both at the grassroots and governmental levels^{31–34}; this may inform future strategies to improve public perceptions and tackle stigma around CI.

Absence of community support and long-term follow-up in LMICs further compounds the difficulties in managing chronic dermatological conditions

Chronic dermatological conditions, including LI, require close monitoring and outpatient follow-up to ensure successful long-term outcomes. Moreover, community support systems, such as patient support groups, play a vital role in promoting emotional and psychological well-being for the patient and family. Examples include the US 'Foundation for Ichthyosis and Related Skin Types' and the UK 'Ichthyosis Support Group,' which provide detailed information and practical advice on CI in high-income settings, promoting contact between affected families and, in some cases, operating telemedicine consultations.^{35,36}

In LMICs, however, outpatient follow-up and community support may not be readily available and/or may be prohibitively expensive. Efforts should, therefore, be made to discuss the rationale for follow-up and to address any barriers to attendance, providing support or solutions, if feasible. Where available, telemedicine again may provide a powerful tool to facilitate regular follow-up remotely.²⁶ Parents should also be empowered to take ownership of their infant's care in the community, with a focus on identifying and preventing potential 'red flags' or complications of CI, most commonly related to development of infection.^{1,2} Appropriate and relevant training on routine skin and eye-care, addressing any parental questions and concerns, can be completed on the ward predischarge.

A successful example of a structured follow-up model in Sub-Saharan Africa is the 'Standing Voice Albinism Skin Cancer Prevention Programme' in Tanzania. Clinics have been set up for individuals with albinism which rotate through local communities at 6 monthly intervals, focussing on equipping and supporting existing healthcare networks. In particular, they co-ordinate skin cancer screening, provision of simple treatments such as cryotherapy and sunscreen, community education and implementation of referral pathways for specialist treatment; in parallel, training and engagement of local healthcare providers is strongly promoted, with the view to sustain the service in the long-term. This model has been commended by the UN and is now being replicated in other Sub-Saharan African countries, with the potential to be applied to further chronic skin diseases in this setting.³⁷

Patient's perspective

I felt sad after my baby was born. Her skin was very dry and others in the village were worried she was cursed. She was small and not feeding well. I did not feel close to her. I felt alone and did not know what to do. The hospital took care of her and told me why this had happened. They took time to show me how to put on the cream and how to look after my baby. I was very happy to know the hospital was getting help from doctors in another place abroad who knew about this problem. With help my baby kept getting better and is now doing well. I'm coming to the hospital regularly to collect medication and to make sure she is doing okay. I could tell others in the village what had happened and show how the creams helped. I'm very happy the hospital helped me and my daughter.

Learning points

- ▶ Despite the high burden of skin disease worldwide, dermatology remains a low priority within the global health agenda. Significant efforts are being made to develop relevant guidelines, set up training for healthcare workers and improve education on the key issues in the diagnosis and management of chronic skin conditions. Nevertheless, further work is required to address important gaps in knowledge and overcome limitations in care worldwide.
- ▶ Early identification of rare skin disorders, for example congenital ichthyoses (CIs), remains a challenge in low and middle-income countries (LMICs) due to delays in presentation to healthcare facilities, lack of provider recognition and difficulty obtaining specialist guidance. This often precludes the timely initiation of effective treatment and increases the risk of further complications.
- ▶ The probability of recurrence of CI in future pregnancies should be discussed with parents, even in settings where genetic testing is not readily available for definitive confirmation. Access to family planning services should also be offered, if feasible.
- ▶ Telemedicine has already demonstrated its potential to transform dermatological services and support both patients and staff worldwide. The ability to access specialist opinions can help bridge the disparity in healthcare faced in LMICs.

Acknowledgements The authors kindly acknowledge the help and support of all the staff at the Clinical Services Department, MRC Unit The Gambia at LSHTM. AS acknowledges funding from the Wellcome Trust Institutional Strategic Support Fund Global Health Fellowship, award number 204834/Z/16/Z.

Contributors AS and BD clinically managed the case, with KF and MG providing expert senior advice. AS collected the photos, performed a literature review and drafted the article. BD, KF and MG contributed to data interpretation and revised the report critically. All authors read and approved the final manuscript.

Funding The authors have not declared a specific grant for this research from any funding agency in the public, commercial or not-for-profit sectors.

Competing interests None declared.

Patient consent for publication Parental/guardian consent obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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