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A Rare Case of Hereditary Ichthyosis: Case Report

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Abstract
Hereditary ichthyosis is a group of genetic skin disorders characterized by dry, scaly skin. These conditions vary widely in severity and are often classified based on genetic and clinical features. We report a case of a 17-year-old African male who has had dry and scaly skin since birth, progressively worsening with itching, scaling, and ulcerations despite having no family history of similar skin conditions. Clinical examination revealed extensive hyperkeratotic plaques characterized by significant thickening of the stratum corneum, rough, dry scaly skin with lichenification, indicating chronic rubbing or scratching. There were areas of erythema and fissuring within the hyperkeratotic regions along with secondary bacterial infection and changes in skin tone affecting his entire skin while sparing only the palms, soles, and face. A diagnosis of hereditary ichthyosis was made based on these clinical findings, supported by a skin biopsy showing benign acanthomatous features. Treatment included the use of emollients, keratolytic agents, and retinoids to alleviate symptoms. Regular follow-up appointments were scheduled to monitor progress and adjust treatment whenever necessary, resulting in significant improvement over time. This case highlights the importance of thorough clinical assessments and skin biopsies in confirming diagnoses when genetic testing is not available.

Keywords: Ichthyosis, Hereditary Ichthyosis, X-linked Ichthyosis, Ichthyosis vulgaris, Case report

Introduction

The term "ichthyosis" implies a generalized scaly dermatosis without the component of erythema. Ichthyoses are a group of skin conditions characterized by abnormal skin cornification and are classified as either hereditary or acquired (Takeichi and Akiyama, 2016). Hereditary ichthyosis (HI) is further categorized into syndromic and non-syndromic types. Hereditary ichthyosis is a genetic disorder typically manifesting in early childhood and is inherited through autosomal dominant transmission. Conversely, acquired ichthyosis (AI) may be associated with underlying conditions such as cancer (e.g., Hodgkin's disease, multiple myeloma, cutaneous T cell lymphoma), systemic disorders (e.g., systemic lupus erythematosus, AIDS), or health conditions affecting lipid and vitamin absorption (e.g., celiac disease, Crohn's disease) (Biver-Dalle et al., 2012; Dar et al., 2011). Ichthyosis Vulgaris represents the most prevalent form of ichthyosis, with an estimated prevalence ranging from 1 in 100 to 250 individuals.

Patient and Observation

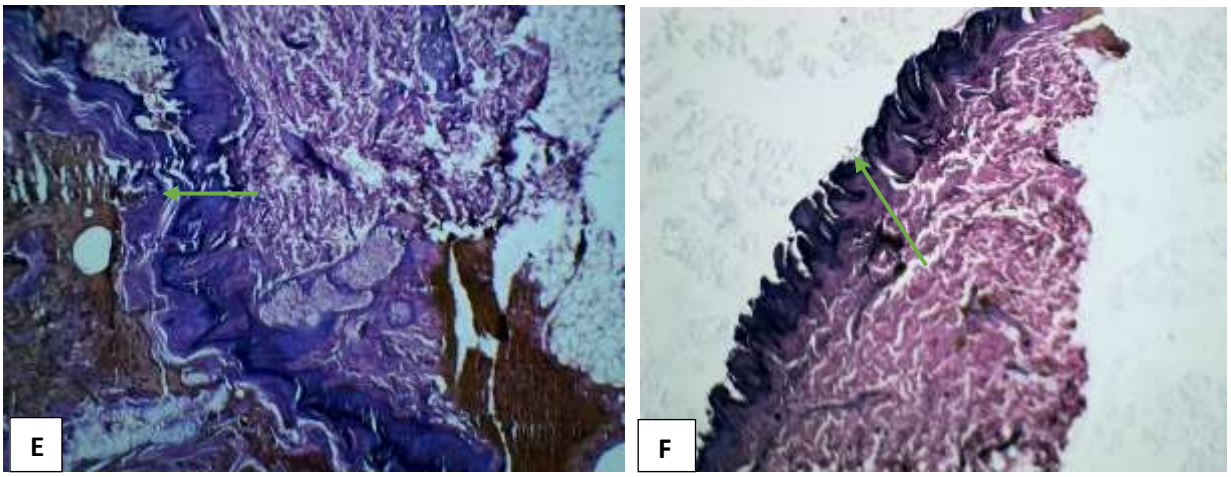
Patient Information: A 17-year-old African male presented to our facility with a chronic history of dry, scaly skin since birth, which has worsened in the past 3 months and is now accompanied by itching, ulcer formation, and skin malodor. In the past 3 months the boy saw a formation of wounds arounds the elbow and creases of upper and lower limbs, which later became infected and started to emit a foul smell. The patient denied having a history of fever. His past medical history was uneventful, and he has not been on any medications throughout his illness. Socially, the patient has experienced isolation due to misconceptions about the condition being contagious, leading to his suspension from school, where the headmaster ordered him to get medical help. There is no reported family history of a similar presentation.

Clinical Findings: The patient was alert and had a normal mental state and cognition; all vital signs were stable. He appeared to have generalized scaly skin with multiple lacerations. Examination of the skin revealed a generalized hyperkeratotic appearance with rough and thick scales covering almost the entire body surface, sparing soles, palms, and face. There were multiple areas of lichenification due to chronic itching, more on the upper limbs. Some areas had old and new wounds where plaques of lesions had been removed, mainly by scratching, leaving behind erythematous skin wounds and some secondary infection areas with pus discharge. Some areas showed fissuring of skin lesions, which the patient reported to be painful. Skin discoloration was noted, characterized by a mixture of hypo- and hyperpigmentation due to chronic inflammation and healing processes (see figures A – D below).

Timeline of the Current Episode: The patient was admitted on January 28, 2024, underwent a course of treatment and was discharged on February 15, 2024.

Diagnostic Assessment: A Complete Blood Count, Renal Function Test, Electrolytes, and Urinalysis were conducted, and all results fell within normal ranges. A skin biopsy was done for pathological diagnosis.

Pathological Findings: The skin section showed epidermis with stratified squamous epithelium with hyperkeratosis and horn pseudocyst formation (see figure E, pointed by green arrow, x 4 HPF, Hematoxylin & Eosin stain). Additionally, an area of reactive squamous atypia with melanin inconsistence along the basal layer was observed, along with a few inflammatory cells infiltrating the dermis (see figure F, pointed by green arrow, X4 HPF, Hematoxylin & Eosin Stain).



Diagnosis: The pathological diagnosis showed features consistent with an acanthomatous benign lesion, specifically Keratosis. Hereditary Ichthyosis with differentials of Ichthyosis vulgaris and X-linked ichthyosis were presumptive diagnoses; however, the molecular genetics study of the biopsy submitted to confirm this was not established due to the lack of a molecular laboratory in our setting.

Therapeutic Interventions: The initial pharmacological treatment regimen consisted of Oral Ampicillin and Cloxacillin 500mg every 8 hours, Metronidazole every 8 hours, Prednisolone 40mg daily with a tapering schedule, and Cetirizine 10mg every 12 hours for 14 days. Topical ointments included Tretinoin every 12 hours, Betamethasone with salicylic acid every 12 hours (while avoiding cracked skin areas), and Miconazole cream every 12 hours. Non-pharmacological interventions involved bathing twice daily with antiseptic soap, followed by the application of Vaseline jelly, and promoting a balanced diet. After the initial treatment and clearing of all infection, follow-up pharmacological treatment included Cetirizine tabs to relieve itching, tretinoin cream, and steroids (oral and systemic).

Follow-up and Outcome of Interventions: Following a two-week course of treatment, significant improvement was observed in the patient's skin condition. These improvements included but were not limited to the clearing of infection, reduction of skin itching, generalized skin softening, and reduction of scales. The patient also noted an improved skin tone and appearance. Most importantly, his confidence and morale improved.

Patient Perspective: "I am incredibly grateful for the comprehensive care and support I received from the entire healthcare team during my treatment. Not only did they effectively address my wounds and skin issues, leading to a significant improvement in my condition with reduced scales, but they also went above and beyond to provide reassurance and counseling. Their efforts helped dispel misconceptions about the disease being contagious, restoring my self-esteem and confidence. Thanks to their holistic approach, I now feel empowered to return to school and resume my normal life without fear of isolation from society. I am truly thankful for the compassionate and professional care I received throughout this journey".

Informed Consent: The parents provided informed consent for the publication.

Discussion

Ichthyosis is a rare genetic skin disorder characterized by dry, scaly skin (Krug et al., 2009). Worldwide, the prevalence of ichthyosis varies, with Ichthyosis vulgaris (IV) standing as the most prevalent form, occurring in approximately 1 in 100 to 250 individuals (Majmundar and Baxi, 2024). In Africa, the exact epidemiology of ichthyosis is not extensively documented, leading to challenges in understanding its

true prevalence in the region. Limited access to healthcare services and genetic testing further complicates the accurate assessment of the disease burden in African populations.

The classification of ichthyosis comprises non-syndromic and syndromic types. Non-syndromic ichthyoses include ichthyosis vulgaris, X-linked recessive ichthyosis, autosomal recessive congenital ichthyosis (ARCI), autosomal dominant ichthyosis vulgaris, lamellar ichthyosis, harlequin ichthyosis, congenital ichthyosiform erythroderma (CIE), and self-improving collodion ichthyosis. Each subtype within this group exhibits specific features and requires tailored management approaches. Syndromic ichthyoses often present with additional systemic manifestations beyond skin involvement, encompassing conditions such as Netherton syndrome, Sjögren-Larsson syndrome, trichothiodystrophy, neutral lipid storage disease with ichthyosis (Chanarin-Dorfman syndrome), Refsum disease, Conradi-Hunermann syndrome, and CHILD syndrome. Furthermore, ichthyosis acquired later in life represents a distinct subgroup characterized by the onset of symptoms in adulthood. Other forms of ichthyosis, such as ichthyosis follicularis with alopecia and photophobia syndrome, ichthyosis hystrix of Curth-Macklin, and ichthyosis prematurity syndrome, add to the diverse spectrum of this skin disorder (Oji et al., 2010).

Diagnosing ichthyosis involves a comprehensive approach that integrates clinical evaluation, family history assessment, and diagnostic tests. The initial step typically includes a thorough physical examination to assess the skin's texture, appearance, and distribution of scales. Detailed documentation of the patient's medical history, including any previous skin conditions or family history of ichthyosis, is essential in establishing a diagnostic framework. Skin biopsies may be performed to examine skin samples under a microscope, aiding in confirming the presence of characteristic changes associated with ichthyosis. Additionally, genetic testing plays a pivotal role in identifying specific mutations responsible for different subtypes of ichthyosis. However, in cases where genetic testing is unavailable, such as the one presented in this report, or inconclusive, a diagnosis can still be established based on clinical presentation and histopathological findings from skin biopsies (Jj and L R-B, 2003). Our patient unfortunately lives in a remote village in Kigoma and comes from a very poor family, so he did not get an opportunity to be seen by a specialist all his life until now. Every time he was taken to a nearby health center he was just given unspecified over-the-counter skin creams which did not do much help, and no test was ever done.

Managing ichthyosis involves a multifaceted approach aimed at alleviating symptoms, improving skin hydration, and enhancing the quality of life for affected individuals. Central to the management of ichthyosis is the regular use of emollients and moisturizers to hydrate the skin and reduce scaling. Topical keratolytics, such as urea or alpha-hydroxy acids, may be prescribed to help shed excess skin cells and promote skin softness. In more severe cases, retinoids or systemic therapy may be considered to address hyperkeratosis and inflammation. Additionally, gentle exfoliation techniques, such as regular bathing with mild cleansers and the use of soft washcloths, can aid in maintaining skin integrity (NIAMS, 2024). Emphasis is also placed on sun protection to prevent exacerbation of symptoms and complications.

Psychological support and counseling play a crucial role in managing the psychosocial impact of ichthyosis, addressing self-esteem issues, and promoting coping strategies. Regular follow-up visits with dermatologists and other healthcare providers are essential to monitor disease progression, adjust treatment regimens as needed, and provide ongoing support to individuals and their families. By incorporating a holistic approach that combines medical treatments, skincare routines, and emotional support, the management of ichthyosis aims to enhance skin health, reduce symptoms, and optimize over

all well-being and prognosis for those living with this chronic skin condition (DermNet, 2024).

Conclusion

This case report underscores the diagnostic intricacies encountered in hereditary ichthyosis, emphasizing the significance of comprehensive clinical assessments and skin biopsies to confirm diagnoses in the absence of genetic analysis. Personalized treatment approaches incorporating emollients, topical therapies, and systemic interventions play a pivotal role in managing skin manifestations effectively. Recognizing the profound psychosocial implications of hereditary ichthyosis underscores the critical need for psychological support and interventions to bolster self-esteem. Sustained educational initiatives are imperative for healthcare professionals to enhance comprehension, facilitate early identification, and optimize outcomes for individuals grappling with the complexities of hereditary ichthyosis.

Competing Interests: The authors declare no competing interests.

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