CASE REPORT

Ichthyosis Vulgaris

A Case Report and Review of Literature

CE 1.5 Contact Hours

Sarah E. Mertz, Thea D. Nguyen, Lori A. Spies

Ichthyosis vulgaris (IV) is a hereditary skin condition characterized by an accumulation of cells in the horny layer that manifests as xerotic, plate-like scales. It is most prominent on the extensor surfaces of the extremities, back, abdomen, and legs and exhibits palmar hyperlinearity (Takeichi & Akiyama, 2016). If not properly treated, this build up can cause difficulty in patient care and a decrease in the quality of life of those afflicted with this condition.

There are more than 20 types of ichthyosis to include epidermolytic ichthyosis, congential reticular ichthyosiform erythroderma, and lamellar ichthyosis, with IV being the most common type of hereditary nonsyndromic ichthyosis and characterized as a reduction of keratohyalin granules or a granular layer absence (Takeichi & Akiyama, 2016). This IV is caused by mutations in the filaggrin gene (FLG) associated with this skin barrier formation and has varying phenotypic manifestations. IV is strongly associated with other atopic cutaneous manifestations such as atopic dermatitis and seborrheic dermatitis. Hallmarks of IV include the visible scaling and dryness that spare flexural surfaces, palm and sole hyperlinearity, and a strong disposition for allergic disease comorbidities such as asthma and seasonal allergies (Leight, Zinn, & Jalali, 2015).

A CASE REPORT

A 57-year-old White male who is cognitively disabled presented to the family practice clinic with his social worker for a follow-up visit for IV. The purpose of this visit was to reinforce a skin care plan to improve severely hyperkeratotic skin. The patient was referred to the practice by neurosurgery, who needed enough clear skin to obtain intravenous access on the forearm or dorsal hands for a benign pituitary removal surgery. By patient report, the lesions had been present since childhood but progressively worsened over the past few years because of a lack of skin care routine. As is typical in IV, there was a history of improvement of symptoms in warmer months of the summer. Comorbidities to the long-standing IV and cognitive disability include hypertension and obesity. There was no personal history of skin cancer, and the patient was not able to provide details of his family history. On physical examination, significant and pertinent findings include diffuse symmetric, thick, hyperkeratotic, light-gray, fish-scaled plaques with fissures prominent on the dorsal and ventral surfaces of the bilateral upper extremities including arms, forearms, dorsal hands, and posterior neck and fine, scaly, erythematous areas of seborrheic dermatitis on the occipital scalp.

A plan of care was collaboratively developed by dermatology and family practice providers. Psychosocial considerations, important in all patients with IV, were compounded by his cognitive disability. Care coordination included the involvement of the caregiver and social worker to enhance hygiene and adherence to the treatment plan. Home health was established with five times of weekly visits and reinforcement of skin care instructions. The joint plan of care included the use of Hibiclens daily for bathing, followed by a combination moisturizing regimen of prescription ammonium lactate cream and triamcinolone 0.1% cream applied to affected areas immediately after bathing and at bedtime. In addition, ketoconazole 2% shampoo was to be used daily on the face and scalp, allowing the product to sit on skin for 5–10 minutes before rinsing, for the seborrheic dermatitis.

One month after the treatment plan was reinforced with home health and adherence increased, the patient presented with marked objective improvement of IV. There were few scattered hyperkeratotic papules present on his bilateral upper extremities and abdomen with diffuse areas of thin gray scaling. Mild erythema and a fine waxy scale consistent with seborrheic dermatitis on the occipital scalp had also improved. The improvement enabled successful scheduling of the needed surgical procedure. Subjective satisfaction with the treatment was also noted; the patient reported getting compliments on his skin improvement at church and reports improved quality of life. He continues to be followed in the family practice clinic and dermatology.

Sarah E. Mertz, BSN, RN, *Louise Herrington School of Nursing, Baylor University, Dallas, TX.*
Thea D. Nguyen, BSN, RN, *Louise Herrington School of Nursing, Baylor University, Dallas, TX.*
Lori A. Spies, PhD, RN, NP-C, *Louise Herrington School of Nursing, Baylor University, Dallas, TX.*

The authors declare no conflict of interest.

Correspondence concerning this article should be addressed to Lori A. Spies, PhD, RN, NP-C, 3700 Worth St., Dallas, TX 75246. E-mail: Lori_Spies@Baylor.edu

DOI: 10.1097/JDN.0000000000000423

DISCUSSION

According to Li et al. (2013), IV is an inherited skin disease characterized by xerosis, keratosis pilaris, and palmar hyperlinearity. It is an autosomal semidominant disorder and the most common inherited keratinization disorder with a prevalence of 1:250–1000 (Takeichi & Akiyama, 2016). It is estimated that the prevalence of IV in Europeans range from 4.0% to 7.7% and 2.29%–3.00% in Asians and occurs in Africans, but the FLG mutations are low in darkly pigmented populations (Leight et al., 2015; Takeichi & Akiyama, 2016). Filaggrin is a vital protein that facilitates the differentiation of the epidermis and the development of protective skin barrier (Takeichi & Akiyama, 2016). The term "filaggrin" is a contraction of "filament aggregating protein" (McLean, 2016). Null mutations in the gene coding profilaggrin, a precursor protein of filaggrin, are causative defects leading to IV. Heterozygous patients with a partial FLG deficiency exhibit a milder phenotype, whereas patients with homozygous or compound heterozygous display a more severe phenotype. Histopathological analysis of IV reveals orthohyperkeratosis, reduced or absent keratohyalin granules, and decreased to absent stratum granulosum. In patients with heterozygous mutation for the gene, the keratohyalin granules are present but may appear small (Leight et al., 2015).

Approximately 50%–60% of patients who have IV also have comorbidities of mild atopic dermatitis and allergic rhinoconjunctivitis (Perusquía-Ortiz et al., 2013). Carriers of the FLG mutation are more susceptible to environmental changes (i.e., cats, tobacco smoking) that may lead to the development of other atopic disorders. In addition, there is a correlation between filaggrin deficiency and risk of developing eczema. Patients with two FLG null mutations do not express filaggrin protein in their epidermis and thus have a substantial likelihood of developing eczema (McLean, 2016). It has also been hypothesized that the severity of IV is related to obesity because of the mild chronic inflammation and impaired lymphatic drainage associated with obesity (Leight et al., 2015). In a study conducted by Dreyfus et al. (2014), IV showed to have a severe or very severe effect on quality of life in a third of their study participants.

Recognition of IV should be included in the evaluation for any pediatric or adult patients presenting with dermatitis. A family history of skin disorders and a thorough inspection of the skin including the soles and palms should be obtained (Thyssen, Godoy-Gijon, & Elias, 2013). Choate (2016) suggested that the diagnosis of IV in clinical practice based on the integration of several factors. These factors include the skin phenotype, time of onset and evolution time, associated cutaneous manifestations, family history and apparent inheritance mode, histopathologic findings on skin biopsy, presence or absence of associated extracutaneous manifestations, and genetic testing. Evaluation of skin phenotype should take into consideration the scale pattern, color, and quality and the presence of erythroderma, a collodion membrane at birth, and blistering or erosions. Genetic testing may be beneficial for genetic counseling and for the confirmation of diagnosis (Choate, 2016).

Skin care management for IV is complex and dynamic with primary treatment evidence-based objectives to reduce scaling, support skin barrier function, increase skin hydration, and decrease symptoms (Leight et al., 2015; Thyssen et al., 2013). The primary goal of IV therapy is to remove the excess scales to treat xerosis without causing more irritation. This includes bathing immediately followed by moisturizers, emollients, keratolytic agents, and topical steroids (Leight et al., 2015). Topical creams and ointments that include urea and lactic acid have been used with success to treat IV (Anderson, 2015). Frequent use of moisturizers with glycerol or urea can help prevent further damage of the skin barrier (Hoppe et al., 2015). Avoidance of occupations involving wet work or exposure to contact irritants or excessive metals is recommended to prevent worsening of IV (Thyssen et al., 2013).

Ethical and cultural considerations include disclosing genetic testing results for patients with IV who may have also incidentally screened positive for other genetic abnormalities. Genomic results can be used to discriminate against individuals and their families. Therefore, nurse practitioners must be familiar with the privacy issues, scope, and limitations of the protections as written in the Genetic Information Nondiscrimination Act (Seibert & Darling, 2013). In addition, there are also ethical concerns involving treating genetic skin diseases with gene therapy, which includes replacement of diseased cells, protein therapy, and replacement or repair of missing or fragmented genes (Seibert & Darling, 2013).

Many gaps in the literature exist; exemplars include the direct comparison between moisturizers in treating IV and the effect of dietary changes and environmental exposures that may affect IV. Urea-based and lactic acid moisturizers, when used together, have been shown to be well suited for first-line therapy for IV and associated atopic dermatitis (Hoppe et al., 2015). To establish the efficacy of these different moisturizers, large cohort studies with comparisons between preparations in populations with IV are needed (Lindh & Bradley, 2015). Few studies regarding dietary changes and allergen avoidance improving symptoms and quality of life of patients experiencing IV exist. Additional investigation is needed to show the efficacy of dietary changes and skin improvement with these changes (Anderson, 2015). Furthermore, more research is needed to examine the burden of known environmental exposures, such as low humidity, dust mites, or excessive use of soaps on the exacerbation of barrier dysfunction (Thyssen et al., 2013).

IV is a common hereditary skin disorder that should be considered in patients presenting with difficult-to-treat dermatitis (Choate, 2016; Thyssen et al., 2013). Skin care management is complex for IV, and coordinated treatment plans should be developed with goals to reduce scaling, treat xerosis, and improve skin barrier function (Leight

et al., 2015). IV has proven to have a severe effect on quality of life of patients when left untreated. Because IV is a lifelong skin disorder, patients experiencing this disease often have to address social stigma and isolation related to the noticeable differences in skin color, integrity, and texture (Seibert & Darling, 2013).

CONCLUSION

The case study included an extreme presentation of IV that had been left untreated. This case is a helpful reminder that IV, the most common inherited ichthyosis, can have severe phenotypic presentations that affect the patients' quality of life or interferes with medical treatment. The benefits of a strong multidisciplinary approach were necessary in this complex case to ensure proper skin care and to enable the needed surgery. Dermatology and family practice nurses and nurse practitioners play an essential role in the diagnosis, treatment, and ongoing management of the challenging skin care regimen associated with IV. Nurses and practitioners have the ability to collaboratively diagnose, refer, manage, and educate patients to make an immense difference in health outcomes for patients experiencing IV. ■

REFERENCES

Anderson, B. (2015). The effect of dietary change in a patient with ichthyosis vulgaris: A case report. *Integrative Medicine: A Clinician's Journal*, *14*(3), 55–58.

Choate, K. (2016). *Overview and classification of the inherited ichthyoses*. Retrieved from https://www.uptodate.com/contents/overview-and-classification-of-the-inherited-ichthyoses?search=ichthyosis%20vulgaris§ionRank=1&usage_type=default&anchor=H16647233&source=machineLearning&selectedTitle=1~14&display_rank=1#H16647233

Dreyfus, I., Bourrat, E., Maruani, A., Bessis, D., Chiavérini, C., Vabres, P., … Mazereeuw-Hautier, J. (2014). Factors associated with impaired quality of life in adult patients suffering from ichthyosis. *Acta Dermato-Venereologica*, *94*(3), 344–346. doi:10.2340/00015555-1710

Hoppe, T., Winge, M. C., Bradley, M., Nordenskjöld, M., Vahlquist, A., Törmä, H., & Berne, B. (2015). Moisturizing treatment of patients with atopic dermatitis and ichthyosis vulgaris improves dry skin, but has a modest effect on gene expression regardless of FLG genotype. *Journal of the European Academy of Dermatology and Venereology*, *29*(1), 174–177. doi:10.1111/jdv.12333

Leight, H., Zinn, Z., & Jalali, O. (2015). Bilateral lower extremity hyperkeratotic plaques: A case report of ichthyosis vulgaris. *Clinical, Cosmetic and Investigational Dermatology*, *8*, 485–488. doi:10.2147/CCID.S89871

Li, M., Cheng, R., Shi, M., Liu, J., Zhang, G., Liu, Q., … Yao, Z. (2013). Analyses of FLG mutation frequency and filaggrin expression in isolated ichthyosis vulgaris (IV) and atopic dermatitis-associated IV. *British Journal of Dermatology*, *168*(6), 1335–1338. doi:10.1111/bjd.12206

Lindh, J. D., & Bradley, M. (2015). Clinical effectiveness of moisturizers in atopic dermatitis and related disorders: A systematic review. *American Journal of Clinical Dermatology*, *16*(5), 341–359. doi:10.1007/s40257-015-0146-

McLean, W. H. (2016). Filaggrin failure—From ichthyosis vulgaris to atopic eczema and beyond. *British Journal of Dermatology*, *175*(Suppl. 2), 4–7. doi:10.1111/bjd.14997

Perusquía-Ortiz, A. M., Oji, V., Sauerland, M. C., Tarinski, T., Zaraeva, I., Seller, N., … Traupe, H. (2013). Complete filaggrin deficiency in ichthyosis vulgaris is associated with only moderate changes in epidermal permeability barrier function profile. *Journal of the European Academy of Dermatology and Venereology*, *27*(12), 1552–1558. doi:10.1111/jdv.12079

Seibert, D. C., & Darling, T. N. (2013). Physical, psychological and ethical issues in caring for individuals with genetic skin disease. *Journal of Nursing Scholarship*, *45*(1), 89–95. doi:10.1111/jnu.12004

Takeichi, T., & Akiyama, M. (2016). Inherited ichthyosis: Non-syndromic forms. *The Journal of Dermatology*, *43*(3), 242–251. doi:10.1111/1346-8138.13243

Thyssen, J. P., Godoy-Gijon, E., & Elias, P. M. (2013). Ichthyosis vulgaris: the filaggrin mutation disease. *British Journal of Dermatology*, *168*(6), 1155–1166.

Ichthyosis Vulgaris: A Case Report and Review of Literature

Instructions:

- Read the article on page 235.
- Take the test, recording your answers in the test answers section (Section B) of the CE enrollment form. Each question has only one correct answer.
- Complete registration information (Section A) and course evaluation (Section C).
- Mail completed test with registration fee to: Broad Data Systems, Inc, 74 Brick Blvd, Bldg 4, Suite 206, Brick, NJ 08723.
- Within 46 weeks after your CE enrollment form is received, you will be notified of your test results.
- If you pass, you will receive a certificate of earned contact hours and answer key. If you fail, you have the option of taking the test again at no additional cost.
- A passing score for this test is 7 correct answer.
- Need CE STAT? Visit www.nursingcenter.com for immediate results, other CE activities and your personalized CE planner tool.
- No Internet access? Call 1-800-787-8985, for other rush service options.
- Questions? Contact Lippincott Professional Development: 1-800-787-8985.

Registration Deadline: September 4, 2020

Provider Accreditation:
Lippincott Professional Development, will award 1.0 contact hour for this continuing nursing education activity.

Lippincott Professional Development is accredited as a provider of continuing nursing education by the American Nurses Credentialing Center's Commission on Accreditation. This activity is also provider approved by the California Board of Registered Nursing, Provider Number CEP 11749 for 1.0 contact hour. Lippincott Professional Development is also an approved provider of continuing nursing education by the District of Columbia, Georgia, and Florida #50-1223.

The ANCC's accreditation status of Lippincott Professional Development Department of Continuing Education refers only to its continuing educational activities and does not imply Commission on Accreditation approval or endorsement of any commercial product.

Disclosure Statement:
The authors and planners have disclosed that they have no financial relationships related to this article.

Payment and Discounts:

- The registration fee for this test is $10.00 members; $20.00 nonmembers.
- If you take two or more tests in any nursing journal published by LWW together by mail, and deduct $0.95 from the price of each test.
- We offer special discounts for as few as six tests and institutional bulk discounts for multiple tests. Call 1-800-787-8985, for more information.