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Reduced Scaling and Skin-Care Burden Following Topical Silica-Based Treatment in a Child With Harlequin Ichthyosis: A Case Report

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Abstract

Harlequin ichthyosis (HI) is a severe congenital disorder of keratinization caused by pathogenic variants in ABCA12 and characterized by profound epidermal barrier dysfunction, hyperkeratosis, and lifelong skin-care requirements. We describe changes in scaling and caregiver-reported treatment burden following introduction of an amorphous silica-containing topical formulation in a 16-month-old girl with genetically confirmed HI. The patient had a characteristic neonatal phenotype and had been managed with intensive topical emollient and barrier therapy without systemic retinoids. At baseline, she had generalized erythroderma and persistent hyperkeratotic scaling requiring approximately four hours of daily bathing with scale-removal care and frequent emollient application. A developmental topical emollient containing milled amorphous hydrated silica fibers was incorporated into her existing home skin-care regimen. Within days, the caregiver reported reduced scale accumulation and topical product requirements, with bathing intervals extending to approximately 36 hours. Serial photographs and contemporaneous caregiver communications documented sustained reductions in scaling over subsequent months, while erythema remained largely unchanged. An unplanned interruption in product availability was associated with increased scaling, followed by renewed improvement after treatment resumed. No serious treatment-associated adverse events were reported in the available caregiver documentation. In this uncontrolled retrospective case, use of the silica-containing topical formulation was temporally associated with reduced visible scaling and skin-care burden. The withdrawal-rechallenge pattern strengthens this temporal association but does not establish causality. These findings are hypothesis-generating and support prospective investigation of silica-containing topical formulations for HI using objective measures of scaling, epidermal barrier function, safety, and caregiver burden.

Categories: Genetics, Pediatrics, Dermatology
Keywords: abca12, amorphous silica, caregiver burden, harlequin ichthyosis, hyperkeratosis, scaling, skin barrier dysfunction, topical therapy, withdrawal-rechallenge

Introduction

Harlequin ichthyosis (HI), the most severe phenotype of autosomal recessive congenital ichthyosis, results from biallelic pathogenic variants in ABCA12, which disrupt epidermal lipid transport and lamellar granule function [1]. The resulting barrier dysfunction causes increased transepidermal water loss, abnormal cornification, and retention of hyperkeratotic scale [2,3]. Neonates typically present with thick, plate-like scale, deep erythematous fissures, ectropion, and eclabium and are at risk for dehydration, thermoregulatory dysfunction, respiratory compromise, and infection [4]. Although historically associated with high neonatal mortality, survival has improved with intensive supportive care and early systemic retinoid therapy [5]. Survivors develop lifelong ichthyosiform erythroderma, hyperkeratosis, scaling, pruritus, and barrier dysfunction. Management remains symptomatic and includes frequent bathing, scale removal, liberal emollients, keratolytics, and topical or systemic retinoids [6]. No curative therapy exists, and the intensive daily skin-care regimen can impose a substantial burden on patients and caregivers [7]. Because stratum corneum hydration influences desquamation and scale cohesion, topical approaches that support a moist skin-surface environment may be relevant to disorders characterized by abnormal scale retention. The potential effect of amorphous silica-containing topical formulations on scaling in HI has not been established. We describe a child with HI in whom the introduction of an amorphous silica-based topical treatment was temporally associated with reduced visible scale accumulation and a marked reduction in daily skin-care requirements.

Case Presentation

A 16-month-old girl with HI had a history of the characteristic neonatal phenotype of HI, including diffuse, markedly thickened hyperkeratotic skin with large plate-like scales and deep fissuring. The diagnosis was confirmed by genetic testing demonstrating pathogenic variants in ABCA12. During the first 16 months of life, she was treated with intensive topical emollient and barrier therapy. Notably, the patient did not receive systemic retinoid therapy. These measures reduced the severity of hyperkeratosis and scaling but did

not eliminate the need for frequent, labor-intensive skin care. By 16 months of age, she had persistent generalized erythroderma, hyperkeratosis, and continuous scaling requiring intensive daily skin care. Her caregiver reported approximately four hours of bathing with scale-removal care each day to hydrate the skin and facilitate removal of accumulated scale. A petrolatum-based emollient was applied after bathing and reapplied frequently throughout the day. Despite this regimen, substantial scale accumulated and was shed between treatments.

In January 2019, the caregiver contacted SiOxMed and requested access to an amorphous silica-containing topical formulation for her child. The formulation provided at that time was a developmental topical emollient manufactured by SiOxMed, LLC (Winston-Salem, NC), consisting of an over-the-counter emollient base containing milled amorphous hydrated silica fibers [8]. The formulation contained the same concentration of silica fibers as the subsequently cleared SBC1 Cream (tradename SiOxC Cream; SiOxMed, LLC), although the 2019 formulation was a developmental predecessor and was not identical to the later cleared product. At the time of use, the formulation had not received FDA clearance for HI, and its use in this patient was outside any subsequently cleared indication.

The product was incorporated into the child's existing home skin-care regimen and applied after bathing to affected areas of the trunk and extremities, with additional applications as needed according to skin condition and bathing frequency. Because treatment was caregiver-directed rather than administered under a prospective protocol, the amount applied per application, total treated body-surface area, and cumulative dose were not systematically recorded. Lot information was not available in the historical records.

Changes were reported within the first several days of use. After approximately one day, the caregiver described reduced visible scale accumulation and a decreased requirement for topical skin-care products. She subsequently omitted a scheduled bath despite previously reporting approximately four hours of bathing and skin care daily. Within several days, the interval between baths increased to approximately 36 hours, which the caregiver reported had not previously been possible. These changes persisted during subsequent weeks. Contemporaneous caregiver communications documented reduced scale accumulation, decreased topical product use, and longer intervals between intensive skin-care interventions, with corresponding serial photographs (Figure *1*).

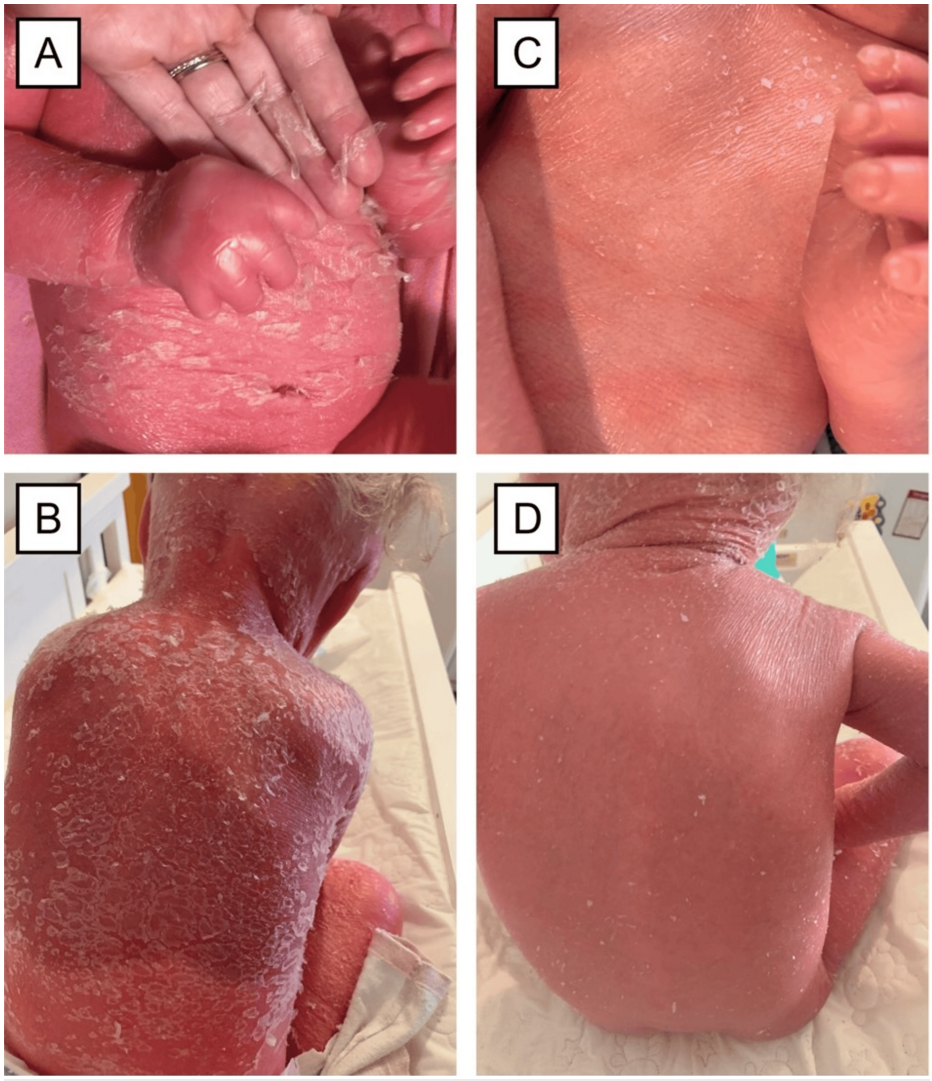


FIGURE 1: Skin appearance before and after initiation of topical amorphous silica-based treatment

Representative caregiver-generated photographs demonstrate extensive hyperkeratotic scaling at baseline (A, B) and markedly reduced scale accumulation following treatment initiation (C, D), with persistent underlying erythema. During this period, the caregiver reported reduced daily skin-care requirements and extension of the interval between baths to approximately 36 hours.

Approximately one month after initiation, the caregiver reported substantially less scale accumulation, while generalized erythema remained largely unchanged. Daily bathing was subsequently resumed in part to facilitate scalp care; however, the caregiver reported that the trunk increasingly tolerated skipped baths. By late February, she described approximately one superficial layer of scale accumulating and shedding between baths, substantially less than the pretreatment pattern. In April, photographs obtained before bathing continued to demonstrate comparatively limited scale accumulation (Figure *2*).

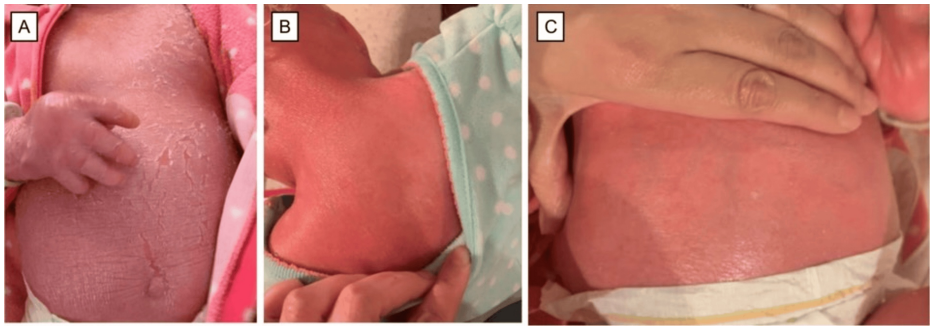


FIGURE 2: Skin appearance during continued topical amorphous silica-based treatment

Representative caregiver-generated photographs (A-C) obtained in April 2019, approximately three months after treatment initiation and before bathing, demonstrate comparatively limited superficial scale accumulation across affected areas of the trunk, with persistent underlying erythema. These findings are consistent with caregiver reports of sustained reductions in scaling during continued treatment.

An unplanned treatment interruption occurred when the caregiver exhausted her supply of the silica-based material. During this period, she reported worsening scaling, with caregiver-generated photographs demonstrating increased scale accumulation involving the face, neck, and trunk (Figure *3*). Following treatment resumption, the caregiver reported recurrence of the previously observed improvement in scaling. No serious treatment-associated adverse events were reported in the available caregiver documentation. A chronological summary of treatment and clinical observations is provided in Table *1*.

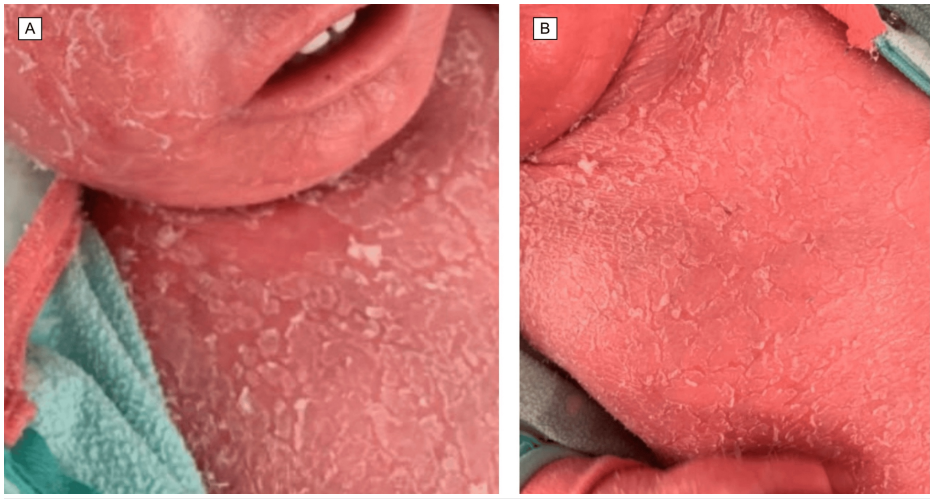


FIGURE 3: Increased scaling following interruption of topical amorphous silica-based treatment

Representative caregiver-generated photographs obtained during an unplanned interruption in treatment availability demonstrate increased scale accumulation involving the face and neck (A) and trunk (B). Following treatment resumption, the caregiver reported renewed improvement in scaling, although post-resumption photographs are not shown.

Written informed consent was obtained from the patient's caregiver to include this image in an open-access article.

Time point	Treatment status	Documented observation	Figure
Baseline, January 2019 (age 16 months)	Before treatment	Approximately 4 hours of daily bathing and skin care, frequent petrolatum application, and extensive generalized scaling	Figure 1A-1B
Approximately Day 1	Treatment initiated	Caregiver reported reduced visible scale accumulation and decreased topical product requirements	Figure 1C-1D (early response)
First several days	Continuing treatment	Interval between baths extended to approximately 36 hours	Caregiver report
Approximately 1 month	Continuing treatment	Substantially reduced scaling; generalized erythema remained largely unchanged. Daily bathing resumed partly for scalp care	Caregiver report
Late February 2019	Continuing treatment	Approximately one superficial layer of scale accumulating and shedding between baths	Caregiver report
April 2019 (~3 months)	Continuing treatment	Photographs obtained before bathing demonstrated comparatively limited scale accumulation	Figure 2A-2C
Date not documented	Treatment interrupted	Product supply exhausted; increased generalized scaling reported and photographed	Figure 3A-3B
Following treatment resumption (date not documented)	Treatment resumed	Caregiver reported renewed reduction in scaling	Caregiver report

TABLE 1: Chronological summary of treatment and clinical observations

Observations were derived from a retrospective review of contemporaneous caregiver communications and caregiver-generated photographs. Treatment was administered at home without a standardized protocol or prospectively defined assessment schedule. Time intervals are approximate. No serious treatment-associated adverse events were reported in the available documentation.

Discussion

HI results from ABCA12 dysfunction, which disrupts epidermal lipid transport and desquamation, leading to severe barrier dysfunction and retention of hyperkeratotic scale [1]. Management remains symptomatic, relying on hydration, scale removal, emollients, and retinoids; no curative therapy exists [5]. The intensive daily care required for autosomal recessive congenital ichthyoses (ARCI) imposes a substantial burden on patients and families. Validated assessments demonstrate impaired patient quality of life and considerable secondary caregiver burden, particularly related to time spent providing care, additional household work, and treatment-related costs; caregiver burden also increases with disease severity [7]. Thus, interventions that reduce scaling and daily treatment requirements may provide clinically meaningful benefits even without altering the underlying disease.

Against this background, the principal observation in this case was a marked reduction in the care required to manage scaling. Before treatment, the child required approximately four hours of bathing daily and frequent whole-body emollient application. Within days of introducing the silica-based topical treatment, intervals between baths increased substantially, and reduced visible scale accumulation persisted over several months. Erythema remained largely unchanged, suggesting that the observed response primarily involved scale accumulation or desquamation rather than generalized improvement in the cutaneous phenotype. This distinction parallels the effects of systemic retinoids, which may reduce hyperkeratosis and scaling without fully resolving erythroderma [9]. This is notable given that the patient did not receive systemic retinoid therapy.

The mechanism underlying the observed reduction in scaling remains uncertain, although a hydration-mediated effect is biologically plausible. Defective epidermal lipid organization and barrier function in HI contribute to abnormal cornification and retention of hyperkeratotic scale [1,5]. Because stratum corneum hydration influences normal desquamation, improved hydration could potentially reduce scale cohesion and facilitate corneocyte shedding. Amorphous silica has a hydrophilic surface capable of interacting with interfacial water, and subsequently developed silica-containing topical formulations are designed to support a moist skin environment. The observed response could therefore reflect changes in hydration, scale adherence, desquamation, or the skin-surface microenvironment rather than correction of the underlying ABCA12-dependent defect or reduced epidermal proliferation. Prospective measurements of transepidermal water loss, stratum corneum hydration, epidermal turnover, and quantitative scale burden will be necessary to distinguish among these possibilities.

The longitudinal course provides additional support for a treatment-associated effect. Improvement was repeatedly described in contemporaneous caregiver communications and supported by serial photographs. During an unplanned interruption in product availability, generalized scaling increased and subsequently improved after treatment resumed. This withdrawal-rechallenge pattern strengthens the temporal association and is consistent with a reversible effect on the skin surface or stratum corneum; however, it cannot establish causality or mechanism in a single uncontrolled case.

Subsequent work with these formulations, SBC1 Cream (trade name SiOxC Cream), has received an FDA 510 (k) and has resulted in FDA clearance for the management of pruritus and dryness associated with atopic dermatitis and allergic contact dermatitis [8]. The regulatory clearance of SiOxC Cream does not establish efficacy for HI, and none of these products is cleared specifically for HI. These products support a moist skin environment but are not cleared for HI. Accordingly, the present case raises the hypothesis that silica-based topical formulations may have broader utility in disorders characterized by epidermal barrier dysfunction and abnormal scaling, but this requires prospective investigation. Additional clinical experience with SiOxC Cream (SBC1) has been reported in other dermatologic and wound-care settings. Published case reports describe its use alongside or following a silica-based fiber wound matrix in post-Mohs surgical wounds, partial-thickness thermal burns, and postoperative wound dehiscence [10-12]. More directly relevant to epidermal barrier dysfunction, a retrospective study of 68 neuropathic patients receiving twice-daily SiOxC Cream as post-closure maintenance therapy reported improvements in xerosis, dermatitis, fissuring, and hyperkeratotic changes over a mean follow-up of six months, with no observed re-ulceration [13]. These findings were presented at the 2026 Symposium on Advanced Wound Care and remain preliminary. Although these clinical observations suggest potential utility in supporting skin integrity, the uncontrolled study designs, concomitant treatments, and differences in underlying conditions limit their interpretation. Importantly, these findings do not establish efficacy in HI, and prospective investigation is needed to determine whether similar benefits can be demonstrated in this population.

This report is limited by its single-patient, uncontrolled design and caregiver-directed home treatment. Disease severity, transepidermal water loss, skin hydration, scale burden, and product consumption were not prospectively quantified. The amount applied per application and total treated body-surface area were not systematically recorded, and treatment requirements were reported by the caregiver. Photographs were obtained under nonstandardized conditions, concomitant skin-care practices continued, and adverse events were not assessed through a prospective safety-monitoring protocol. Additionally, the precise duration of the treatment interruption and timing of subsequent worsening and improvement in scaling were not documented. Systematic clinical follow-up beyond the period documented above was not available for this retrospective report. Natural disease variability further limits attribution of the observed response to the silica-based treatment. Systematic long-term follow-up beyond the available caregiver documentation was not performed. Nevertheless, the magnitude and persistence of the reported reduction in treatment burden, together with the withdrawal-rechallenge observation, warrant prospective evaluation incorporating objective measures of scaling, epidermal barrier function, treatment requirements, caregiver burden, quality of life, and safety. Longer-term outcomes may also be explored in future investigations.

Conclusions

In this child with HI, incorporation of an amorphous silica-based topical treatment into an established skin-care regimen was associated with reduced visible scale accumulation and a substantial reduction in caregiver-reported skin-care burden. Improvements emerged within days and persisted over several months, with increased scaling during an unplanned treatment interruption and renewed improvement following treatment resumption. Although this withdrawal-rechallenge pattern strengthens the temporal association, causality cannot be established from a single uncontrolled case. These findings suggest that silica-based topical formulations warrant further investigation as adjunctive treatments for managing hyperkeratosis and reducing daily care requirements in HI. Prospective studies incorporating objective measures of scaling, epidermal barrier function, safety, and caregiver burden are needed to evaluate these potential benefits.

Additional Information

Author Contributions

All authors have reviewed the final version to be published and agreed to be accountable for all aspects of the work.

Concept and design: Adam M. Jorgensen

Acquisition, analysis, or interpretation of data: Adam M. Jorgensen, Stephen Bearman, Aubrey Montebello

Drafting of the manuscript: Adam M. Jorgensen, Aubrey Montebello

Critical review of the manuscript for important intellectual content: Adam M. Jorgensen, Stephen Bearman, Aubrey Montebello

Disclosures

Human subjects: Informed consent for treatment and open access publication was obtained or waived by all participants in this study. **Conflicts of interest:** In compliance with the ICMJE uniform disclosure form, all authors declare the following: **Payment/services info:** All authors have declared that no financial support was received from any organization for the submitted work. **Financial relationships:** Adam Jorgensen declare(s) employment and stock/stock options from SiOxMed, LLC. Adam M. Jorgensen is an executive and shareholder of SiOxMed, LLC and has a financial interest in the company and its products. SiOxMed provided the topical formulation used by the patient and company resources were used in the retrospective analysis and preparation of this manuscript. SiOxMed may benefit financially from the findings described in this report. **Other relationships:** SiOxMed provided the amorphous silica-based topical material used by the patient for home-based use. The caregiver subsequently shared patient-generated photographs, contemporaneous communications, and observations regarding the patient's experience. SiOxMed obtained written informed consent from the patient's caregiver for publication of the de-identified clinical information and photographs presented in this report. These materials were retrospectively reviewed for preparation of the case report, and SiOxMed resources were used for retrospective analysis and manuscript preparation. SiOxMed may benefit financially from the findings described in this report.

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