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VIEWPOINT

Surgical Management of Harlequin Ichthyosis

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Sir:

Harlequin ichthyosis (HI) is the most severe congenital ichthyosis and is associated with a high neonatal mortality. It is due to a mutation in the *ABCA12* gene, which codes for an adenosine triphosphate–binding cassette transporter essential to the development and maintenance of the epidermal lipid barrier. As a result of the mutation, the lipid barrier is defective, causing hyperkeratinosis, hyperthermia secondary to ineffective diaphoresis, and dehydration. The hyperkeratinosis can also result in the development of hyperkeratinic bands leading to limb ischemia and autoamputation.[1] Although retinoids have been shown to soften the hyperkeratinic bands,[2] severe limb ischemia necessitates immediate surgical management. Few reports exist that describe the surgical management of HI[3,4]; thus in this article, we present our surgical management of a newborn with HI, who subsequently went on to have a favorable outcome.

The patient was transferred from an outside hospital on the day of birth with HI, which had been undiagnosed prenatally. She had thick white scales over her skin with many open fissures, ectropion, eclabion, and abnormally shortened fingers and toes with shallow web spaces that appeared violaceous with no capillary refill. Further evaluation revealed multiple hyperkeratinic constriction bands on the fingers, wrists, ankles, and toes leading to digital hypoperfusion (Fig. 1). We performed a complete surgical escharotomy release of the hyperkeratinic bands in the neonatal intensive care unit on day of life (DOL) 1 under local anesthesia. Dorsal incisions were made with 15 blade scalpels through the hyperkeratinic dermis across the wrist creases, along individual fingers, and on bilateral legs across the ankles with immediate release of the constriction bands (Fig. 2A). The patient required a 25 cc blood transfusion due to a 20 cc blood loss from sharp dissection, but otherwise she tolerated the procedure extremely well.

Perfusion to the patient's fingers and toes improved within minutes following the release. Within 20 minutes, digital capillary refill was <3 seconds and gentle pressure stopped all bleeding. Over the following weeks, she began to shed the thick scale present at birth according to the natural course of HI. Meticulous sterile nursing with frequent petrolatum application and every other day dilute bleach baths were instituted. She healed the escharotomy incisions without any visible scarring, and with improved digital function and development of her digital web spaces (Fig. 2B). She was transitioned to the nonsterile nursery and was discharged from the hospital on DOL 30. By 6 months of age, she developed very thin scars on her wrists and ankles, but remained scar free at her fingers and toes (Fig. 2C). There was slight scar thickening on her right ankle, which is being treated with silicone sheeting. Overall, she is thriving and has not had significant medical issues or infection.

Given our experience with this patient, we advocate for early, definitive treatment of constriction bands in HI by surgical escharotomy, which avoided ischemic autoamputation without the need for repeated surgical debridement. Children with HI can heal with minimal scarring, and we were able to observe preservation and further development of all digits with no significant morbidity.

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Fig. 1. Initial photograph of the patient with HI on DOL 0 showing thick white scales, ectropion, eclabion, and shortened digits with constriction bands leading to distal digital ischemia.

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Plast Reconstr Surg Glob Open 2019;7:e2161; doi:10.1097/GOX.0000000000002161; Published online 16 May 2019.

Fig. 2. Photograph at 10 months following surgical escharotomy showing further healing of hand incisions with good movement (flexion and extension) of digits.

DISCLOSURE:

None of the authors has a financial interest to declare in relation to the content of this article.

ACKNOWLEDGMENTS

This case report is exempt from institutional review board approval. Completed patient authorization forms for image use have been submitted.

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