Management of Harlequin Ichthyosis: A Brief Review of the Recent Literature

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Harlequin ichthyosis (HI) is a life-threatening genetic disorder that largely affects the skin of infants. HI is the most severe form of the autosomal recessive disorder known as ichthyosis. It is caused by mutations in the A12 cassette (lipid-transporter adenosine triphosphate-binding cassette A12). Neonates affected by this disease are born with specific morphological characteristics, the most prominent of which is the appearance of platelet keratotic scales separated by erythematous fissures. The facial features include eclabium, ectropion, a distinct flattened nose, and dysplastic ears. A common finding among those with HI is impaired skin barrier function. The purpose of the present narrative review is to assess the most recent literature regarding the management of HI.

Emphasis is given to surgical management and consultation, to the indications for timing and surgical intervention, to the risks that are presented with surgery, and to the details of the surgical procedure itself. Management of HI requires a multidisciplinary team of experts, and specific guidelines are needed in order for the risks to be minimized and viability to be increased.