Congenital Ichthyoses in Pediatric Age Group: A Cross sectional Study

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Introduction

Ichthyoses are heterogeneous group of disorders due to defect in keratinization or cornification with abnormal differentiation and desquamation of epidermis which is clinically characterized by dry-rough skin with scaling over much or the entire body surface.¹ The primary function of the stratum corneum is to provide a barrier to water loss without which terrestrial life is not possible. Defective barrier function leads to increased transepidermal water loss, a characteristic feature of ichthyosis.² The terminology and nosology of congenital ichthyosis have continuously evolved and has led to a confusing medley of different terms and classifications.³ A number of well-defined ichthyoses have characteristic features and can be reliably diagnosed. However, a specific diagnosis can be challenging in certain patients due to clinical heterogeneity. In general, determination of whether an ichthyosis is inherited or acquired, presented at birth or later in life, and whether it is limited to the skin or part of multisystem disorder helps in making a diagnosis. Quality and distribution of scales, presence or absence of blistering, erythroderma, and associated abnormalities of skin adnexa are other useful clinical features. A thorough family history is essential for recognizing the inheritance pattern which is necessary for therapeutic decision and offering genetic counseling.

Abstract

Background and Objectives: Ichthyoses comprise a heterogeneous group of disorders due to defect in keratinization. Establishing the correct clinical diagnosis in a patient with ichthyosis is a prerequisite for making prognostic predictions, therapeutic decisions, and offering genetic counseling. However, a specific diagnosis can be challenging in certain patients due to clinical heterogeneity. In this study, we analyzed the clinical presentation of various types of congenital ichthyoses in pediatric age group.

Materials and Methods: A cross-sectional observational study was conducted in dermatology outpatients department. History was elicited with regard to the age of onset, blistering of the skin, seasonal variation, and similar lesions in other family members followed by detailed dermatological and systemic examination and necessary investigations.

Results: A total of 64 patients were included in this study and relative incidence of different types of ichthyoses was noted. 13 cases of collodion babies were followed and 70% of them developed lamellar ichthyosis. A significant proportion of cases with autosomal recessive inheritance had a history of consanguineous marriage in the parents.

Conclusion: A number of well-defined types of ichthyoses have characteristic feature and can be reliably diagnosed. A thorough family history and clinical examination are a prerequisite for making correct diagnosis and for recognizing the inheritance pattern which is necessary for therapeutic decision and offering genetic counseling.

Key words: Congenital ichthyosis, Collodion baby, Ichthyosis vulgaris

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Aim
To study the clinical presentation of various types of congenital ichthyosis in pediatric age group.

MATERIALS AND METHODS

This cross-sectional observational study was conducted at Department of Dermatology at Tirunelveli Medical College. Institutional Ethics Committee approval and informed consent from parents were obtained. In eliciting the history, a set pattern of questionnaire was followed. Enquiries were made with regard to symptoms, age of onset, duration, itching, diminished sweating and heat intolerance, history of collodion baby, blistering of skin, seasonal variation, cyclical shedding of skin, photosensitivity, and photophobia. History regarding involvement of other systems such as central nervous system (CNS), skeletal system was taken. History of any maternal illness and medication during antenatal period, prematurity and prolonged labor was elicited. Patients’ developmental history and family history of similar lesions in the parents and siblings were elicited. History regarding consanguineous marriage of parents was recorded. A detailed general examination was conducted with specific reference to CNS and skeletal system. Measurement of head circumference was performed and evidence of short stature, microcephaly, cataract, and gait were noted. On dermatological examination, skin lesions were examined and nature of scales whether polygonal or lamellar, color of scales, and whether loose or adherent were noted along with distribution of scales with sparing of certain areas. The presence of blisters, erythroderma, lichenification, ectropion, elabion, eczematization, and impetiginization were noted. Hair and nails were examined for alopecia, brittle hair, and nail dystrophy. Palms and soles were examined for hyperlinearity, palmoplantar keratoderma, sclerodactyly, and digital contractures. Apart from routine hematological examination, skin biopsy and microscopic examination of hair were done wherever indicated. Referral to other specialists such as neurology and ophthalmology was done to confirm or rule out associated feature of some syndromes as and when suspected. All the data were compiled and analyzed statistically and inference was drawn.

RESULTS

Table 1 presents the relative incidence of different types of congenital ichthyosis. Out of 64 patients with congenital ichthyosis, ichthyosis vulgaris constituted 72% of cases followed by lamellar ichthyosis 14%. Non-bullous ichthyosiform erythroderma (NBIE) constituted 6% followed by bullous ichthyosiform erythroderma (BIE) and Sjogren–Larsson syndrome each constituted 3%. The incidence of ichthyosis vulgaris was almost equal in both sexes. Incidence of lamellar ichthyosis was more in females. Equal sex distribution was seen in NBIE.

All except two cases of ichthyosis vulgaris had age onset from 3 to 6 months (Table 2). Lamellar ichthyosis, NBIE, BIE, and other ichthyosiform syndromes had age of onset since birth. Table 3 shows that out of 13 collodion babies, 70% of cases evolved into lamellar ichthyosis and 30% evolved with NBIE. In ichthyosis vulgaris, 73% of patients had no history of consanguineous marriage of the patients, and in 24% of patient history of second- and third-degree consanguineous marriage was present. In lamellar ichthyosis, NBIE, Sjogren–Larsson syndrome and Netherton’s syndrome, all the patients had consanguineous parents. In ichthyosis vulgaris 41% of patients had family history of ichthyosis. In lamellar ichthyosis, positive family history was present in 22% of patients.

DISCUSSION

In our study, the incidence of ichthyosis vulgaris was 1 in 200 which complies with that of study by Wells and Kerr which showed that the incidence of ichthyosis vulgaris
CONCLUSION

A number of well-defined types of ichthyoses have characteristic features and can be reliably diagnosed. However, a specific diagnosis can be challenging in certain patients and families due to great clinical heterogeneity. In general, determination of whether an ichthyosis is inherited or acquired, present at birth or later in life, and whether it is limited to the skin or part of a multisystem disorder helps in diagnosis. Quality and distribution of scale, presence of absence of erythroderma, blistering, and associated abnormalities of skin adnexa are other useful clinical features. A thorough family history is essential for recognizing the inheritance pattern. Establishing the correct clinical diagnosis in a patient with ichthyosis is a prerequisite for making prognostic prediction, therapeutic decision, and offering genetic counseling.

REFERENCES