Print ISSN: 2321-6379
Online ISSN: 2321-595X
DOI: 10.17354/ijss/2017/114

Original Article

Congenital Ichthyoses in Pediatric Age Group: A Cross sectional Study

P Sivayadevi[1], R Karthikeyan[2], Heber Anandan[3]

[1]Associate Professor, Department of Dermato Venereoleprology, Government Medical College, Thanjavur, Tamil Nadu, India, [2]Assistant Professor, Department of Dermato Venereoleprology, Government Medical College, Tirunelveli, Tamil Nadu, India, [3]Senior Clinical Scientist, Department of Clinical Research, Dr. Agarwal's Healthcare Limited, Tirunelveli, Tamil Nadu, India

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

INTRODUCTION

Icthyoses 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 and families 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. Establishing a correct clinical diagnosis in a patient with ichthyosis is a prerequisite for making prognostic prediction and therapeutic decision.[4] Recent advances in the molecular genetics have provided tools to categorize ichthyosis on the basis of their underlying genetic defect which helps in offering genetic counselling.[5]

Access this article online

www.ijss-sn.com

Month of Submission : 01-2017
Month of Peer Review : 02-2017
Month of Acceptance : 02-2017
Month of Publishing : 03-2017

Corresponding Author: Dr. P Sivayadevi, Department of Dermato Venereoleprology, Government Medical College, Thanjavur, Tamil Nadu, India. E-mail: drkrishidev@yahoo.co.in

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, eclabion, 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 babes, 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

Table 1: Relative incidence of different types of congenital ichthyoses

Clinical types	Number of cases (%)
Ichthyosis vulgaris	46 (71.8)
Lamellar ichthyosis	9 (14.1)
NBIE	4 (6.3)
BIE	1 (3.1)
Netherton's syndrome	1 (1.5)
Sjogren–Larsson syndrome	2 (3.1)

BIE: Bullous ichthyosiform erythroderma, NBIE: Non-bullous ichthyosiform erythroderma

Table 2: Age of onset of congenital ichthyoses

Types of ichthyosis	Birth	3 months	6 months	1 year
Ichthyosis vulgaris	-	22	22	2
Lamellar ichtyosis	9	-	-	-
NBIE	4	-	-	-
BIE	1	-	-	-
Sjogren–Larsson syndrome	2	-	-	-
Netherton's syndrome	1	-	-	-

BIE: Bullous ichthyosiform erythroderma, NBIE: Non-bullous ichthyosiform erythroderma

Table 3: Evolution of collodion babies

Collodion babies	Number of babies (%)
Lamellar ichthyosis	9 (70)
NBIE	4 (30)

NBIE: Non-bullous ichthyosiform erythroderma

maybe as common as 1 in 250.[6] The age of onset of ichthyosis vulgaris was around 3-6 months in 98% of patients. In lamellar ichthyosis, NBIE and BIE the age of onset of the disease were from birth. This complies with that of the description about the age of onset of the disease given by Traupe *et al.*, in the guide to clinical diagnosis of ichthyosis.[7] In the Van Gysel *et al.* studied of follow-up to 17 cases of collodion baby, 60-80% of the infants developed NBIE, and lamellar ichthyosis.[8] In our study of follow-up of 13 cases of collodion babies, 70% of the patients developed lamellar ichthyosis and 30% of the patients developed NBIE with the ratio 2.3:1. The most common presenting complaints of patients were dryness, roughness, and disfigurement. Itching was present in patients with associated atopy. Winter exacerbation of the disease was present in 46% of ichthyosis vulgaris patients. A study by Kuokanen showed an association of atopy in 37-50% of patients which was 6.5% in our study.[9] In ichthyosis vulgaris, 76% of patients had no history of consanguineous marriage of the parents and in 24% patient history of second-degree and third-degree consanguineous marriage was present. In lamellar ichthyosis second-degree, consanguineous marriage was present in 44% of patients and third-degree consanguineous marriage was present in 55% of patients. This complies with that of autosomal recessive inheritance. In BIE 50% had consanguinity of parents. In Netherton's syndrome second-degree consanguineous marriage was present in the parents and it complies with that of autosomal recessive inheritance. In ichthyosis vulgaris, family history of ichthyosis was present in 41% of patients. In lamellar ichthyosis, positive family history was present in one family whose two siblings were affected. As it is an autosomal dominantly inherited disorder, the risk of having a further affected child is 25% which is seen in this case. In BIE, no family history of ichthyosis was present. Because it is an autosomal dominantly inherited disorder, it can be presumed that the patient suffered a new keratin gene mutation.

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 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

1. Moeschler JB, Shevell M; American Academy of Pediatrics Committee on Genetics. Clinical genetic evaluation of the child with mental retardation or developmental delays. Pediatrics 2006;117:2304-16.
2. Elias PM, Menon GK. Structural and lipid biochemical correlates of the epidermal permeability barrier. Adv Lipid Res 1991;24:1-26.
3. Oji V, Tadini G, Akiyama M, Blanchet Bardon C, Bodemer C, Bourrat E, *et al.* Revised nomenclature and classification of inherited ichthyoses: Results of the First Ichthyosis Consensus Conference in Sorèze 2009. J Am Acad Dermatol 2010;63:607-41.
4. Pietrusinski M, Stanczyk-Przyluska A, Chlebna-Sokól D, Borkowska E, Kaluzewski B, Borowiec M, *et al.* Identification and clinical consequences of a novel mutation in the gene for transglutaminase 1 in a patient with lamellar ichthyosis. Clin Exp Dermatol 2015;40:921-3.
5. Richard G. Molecular genetics of the ichthyoses. Am J Med Genet C Semin Med Genet 2004;131C:32-44.
6. Wells RS, Kerr CB. Clinical features of autosomal dominant and sex-linked ichthyosis in an English population. Br Med J 1966;1:947-50.
7. Traupe H, Fischer J, Oji V. Nonsyndromic types of ichthyoses - An update. J Dtsch Dermatol Ges 2014;12:109-21.
8. Van Gysel D, Lijnen RL, Moekti SS, de Laat PC, Oranje AP. Collodion baby: A follow-up study of 17 cases. J Eur Acad Dermatol Venereol 2002;16:472-5.
9. Kuokkanen K. Ichthyosis vulgaris. A clinical and histopathological study of patients and their close relatives in the autosomal dominant and sex-linked forms of the disease. Acta Derm Venereol Suppl (Stockh) 1969;62:1-72.

How to cite this article: Sivayadevi P, Karthikeyan R, Anandan H. Congenital Ichthyoses in Pediatric Age Group: A Prospective Study. Int J Sci Stud 2017;4(12):143-145.

Source of Support: Nil, **Conflict of Interest:** None declared.