

Autosomal Recessive Congenital Ichthyosis with Cataract – A Case Report from Pakistan

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Abstract

The ichthyoses are a diverse group of disorders with variable clinical presentations. There are syndromic and non-syndromic forms with multiple associated features. The association of autosomal recessive congenital ichthyosis (ARCI) with cataract is a rare phenomenon and no such case has been reported in recent literature. We hereby report the case of a 5-year-old female patient, a product of consanguineous marriage, who presented to the pediatric department of Dr. Ruth K.M Pfau Civil Hospital Karachi, Pakistan, with the complaint of rough skin with scaling and peeling over the entire body since birth due to congenital ichthyosis. She further developed complaints of reduced vision which later was diagnosed as cataract. Her previous two siblings had similar illness and they expired. Widespread scaling is the defining feature of ichthyosis specifically when it comes to ARCI it manifests at birth and can later progress to any spectra of illness. A multidisciplinary approach is required for the management of such patient.

Key words: Ichthyosis, Genetic diseases, Cataract, Genetic skin disease

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Introduction:

The ichthyosis are a heterogeneous group of conditions depicted by generalized dry skin, evident scaling and in multiple cases by inflammation, with both inherited and acquired forms.¹ Inherited ichthyosis can be classified into non-syndromic and syndromic forms being sure upon the clinical indications. Non-syndromic ichthyosis are characterized by the phenotypic expression of the syndrome being seen only in the skin, while in syndromic ichthyoses multiple organ systems may be involved.^{1,2} Non-syndromic ichthyosis can be further categorized into various forms based on clinical presentation and pattern of inheritance which entails ichthyosis vulgaris, recessive X-linked ichthyosis, autosomal recessive congenital ichthyosis (ARCI), keratopathic ichthyosis and other forms.³ The

general ocular complication or association of ichthyosis is ectropion which may direct to corneal dryness, scarring, infection and perforation.⁴ The coalition of cataract with ichthyosis is not well described in literature. There had been two very old papers documenting association of ichthyosis with cataract,^{5,6} however no such case reports are established in modern literature. We hereby, report a case of a 5 year old female with autosomal recessive congenital ichthyosis with cataract

Case Presentation

A 5-year-old female patient presented to the pediatric outpatient department of Dr Ruth K.M Pfau Civil Hospital Karachi, Pakistan, with the complaint of rough skin with scaling and peeling over the entire body since birth. The mother complained that she has noticed decreased vision in the child for the last 4 months. The baby was born preterm at 8 months of gestation. The mother reported a history of leaking since the 6th month of gestation leading to oligohydramnios during her third trimester. At birth, the child had tight dry skin overall which later turned to scales and peeled off with oiling at home. The skin condition waxes and wanes with the use of topical medicines from the doctor, only to recur. She had been hospitalized four times at the ages of 1 year, 3 years, 3.5 years, and 4 years with a diagnosis of sepsis during the first admission and for the management

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of protein-calorie malnutrition in the other admissions. She is the third child of her parents with first-degree consanguinity. Two of her male siblings expired at the ages of 2.5 months and 4 months of age and they had similar skin conditions. 1 younger sister is alive and healthy, and 1 child was aborted at 5 months gestation.

On examination, she was an emaciated child with generalized dry and rough skin. She was vitally stable. Her height and weight were below the 3rd percentile for her age. There was scaling overall with peeling of scales at various areas on both lower and upper limbs, palmoplantar keratoderma [figure 1], grade 3 acanthosis in the diaper area with raw skin specifically in the groin [figure 2], and axillary area. She had patchy alopecia with scaling of scalp



Figure 1



Figure 2

and easily pluckable hair. She had taut shiny skin around her eyes and mouth, her tongue was smooth with angular cheilosis and there was poor oral hygiene with dental caries. Associated findings include bilateral leukocoria due to cataracts. There was no other dysmorphology. Systemic examination revealed firm hepatomegaly with a span of 8 cm (about 3.15 in).

Investigations revealed haemoglobin of 8.2 gm/dl, a total leucocyte count of 6.2/UL, and platelets of 245,000 /UL. A peripheral smear was negative for Jordan's anomaly which ruled out lipid storage disease associated with ichthyosis. The zinc levels were normal 87.35 (50-150).

A dermatology opinion was taken, and they made a final diagnosis of ichthyosis based on a typical clinical presentation. White soft paraffin (WSP) was advised to be applied topically daily after bathing, which yielded significant improvement in the scaling. Fusidic acid was applied topically to raw areas and fissures. She was counselled that she has to protect the skin from drying which can be done by applying any emollients including WSP or any oil. The ophthalmology department planned surgery for a cataract after a month. Mother was counselled regarding the genetic nature of the condition and its recurrence risk of 25% in every next pregnancy. Nutritional rehabilitation of the child was done and counseling regarding diet and hygiene was given.

Discussion

ARCI typically manifests at birth, frequently as a collodion baby, and can later proceed into any of the spectrum of illnesses.⁷ 8 percent of these instances provide a history of consanguinity, and 51 percent have siblings who also have the same condition. Premature birth rates for these children are around 25%.⁸ In a similar vein, our patient was born of a consanguineous union and had a family history of two siblings passing away with the same skin ailment.

Ramer et al. found scarring alopecia and dark brown scaly lesions that were prevalent in the palmoplantar region as clinical symptoms in our patient.⁹

Additionally, there was scaling all over with scales peeling in different places on both the lower and upper limbs, palmoplantar keratoderma, grade 3 acanthosis in the diaper area, and raw skin in particular in the groin and axillary area. Gingivitis, periodontitis, enamel hypoplasia, high caries incidence, delayed primary and secondary eruption, bruxism, alveolar ridging, bifid teeth, abnormal tooth morphology, and hyperkeratotic plaques on the tongue have all been observed in people with ichthyosis.¹⁰ Our patient had a smooth tongue with angular cheilosis, poor oral hygiene,

and dental caries. These kids¹¹ who previously had 4 hospital admissions for sepsis and protein calorie deficiency, are more prone to systemic infection. Time of skin scale shedding was also reported.¹² Ocular manifestations of ARCI comprise exposure keratitis secondary to ectropion, unilateral megalocornea, enlarged corneal nerve, blepharitis, absence of the meibomian gland, trichiasis, madarosis, and absence of lacrimal puncta. Ectropion of both upper and lower lids have been documented.¹³ One-third of the children develop bilateral cicatricial ectropion due to excessive dryness of the skin and subsequent contracture. Secondary corneal ulceration may occur due to exposure. The X-linked type has been noted to have congenital cataract¹⁴ whereas in our patient, only ocular finding was bilateral cataract which is very rare with Lamellar ichthyosis.^{5,6}

An expertise of dermatologist, pediatrician, genetist, ophthalmologist and physiotherapist play an important role in managing such cases. The management is aimed at decreasing symptoms and include emollients (petrolatum, coconut oil, alpha hydroxyl acetic acid), keratolytic containing salicylates with propylene glycol and local and systemic retinoids.¹⁵ Our dermatologist advised to use White soft paraffin (WSP) which was applied topically daily after bathing, which yielded significant improvement in the scaling. Fusidic acid was applied topically to raw areas and fissures. Furthermore, She was also counselled that she has to protect the skin from drying which can be done by applying any emollients including WSP or any oil.

Conclusion

Autosomal recessive congenital ichthyosis is a rare entity with even rarer association with bilateral cataract. Multidisciplinary approach is needed for the management of such patients.

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Authors contribution: All authors contributed equally to this work.

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