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Case Report

Case report of adolescent male with non-bullous congenital ichthyosiform erythroderma visiting dermatology clinic of King Fahad Military Hospital, Jeddah, Saudi Arabia, 2017

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ABSTRACT

Non-bullous congenital ichthyosiform erythroderma (NBCIE) is an inherited disease with autosomal recessive form. It is considered as an extremely rare skin disorder that estimated to occur in 1/300,000 births, characterized by abnormal scaling of the skin with underlying redness. This is a case report study design with detailed history, examination and genetic analysis of 17 years old male with Non-bullous congenital ichthyosiform erythroderma (NBCIE).

Keywords: ABCA12, Non-bullous congenital ichthysiform, Autosomal recessive, Keratoderma

INTRODUCTION

The ABCA12 gene encodes instructions that allow a cell to make a protein known as an ATP-binding cassette (ABC) transporter. ABC transporter proteins carry many types of molecules across cell membranes. In particular, the ABCA12 protein plays a major role in transporting fats (lipids) in cells that make up the outermost layer of skin (the epidermis).[1]

Genetic mutation in ABCA12 gene can lead to inherited skin abnormalities.[2] About 65 mutations in this gene can lead to harlequin ichthyosis that is characterized by hard, thick scales that are present at birth; excessive dehydration; and increased risk of infections.[3,4] At least 20 mutations in this gene have been found to cause non bullous congenital ichthyosiform erythroderma (NBCIE).[5]

Non bullous congenital ichthyosiform erythroderma (NBCIE) is a rare with skin abnormalities tend to be less severe than those in harlequin ichthyosis.[6] NBCIE is an autosomal recessive ichthyosis, which occurs in 1 in 300,000 births.[7] NBCIE, appears as generalized erythroderma with fine white scales that gradually replace the collodion membrane.[7,8] Other associations include ectropion, eclabium, scalp alopecia, hyperhidrosis with heat intolerance, and nail dystrophy.[9] Clinical presentation, pattern of inheritance, and laboratory evaluation may establish a diagnosis, which can assist in prognosis and genetic counseling.[10]

Because of the rareness of this disease and lack of knowledge about the specific clinical characteristics, it usually happens to be treated on the basis that it is another disease. Therefore, the researchers here decided that they should make a report of this disease and thus will enrich medical scientific research with more clinical characteristics of this disease.

CASE REPORT

A 17-years-old male came to clinic with his mother complaining of generalized dryness and scaling and hard thick palms and soles since birth. Moreover, his mother mention that he has not been sweating since birth. He tried a lot of topical treatment that prescribed by different dermatological doctors without any improvement.

Family history showed that there is consanguinity and his sister (19 years old) and his brother (30 years old) suffer with similar condition as well but no diagnosis. This family is living in rural area.

There was no history of any blister formation at any time, at birth or later.

During his mother pregnancy of him, there was no history of maternal complications or any drug exposure and no history of allergies.

Pedigree analysis for the patient's family (Figure 1) shows that, the disease itself is autosomal recessive and the patient' parents carry the abnormal gene (circle: female, square: male). However, this family has 10 kids (6 males and 4 females) two males and one female are affected (dark square: affected male, dark circle affected female).

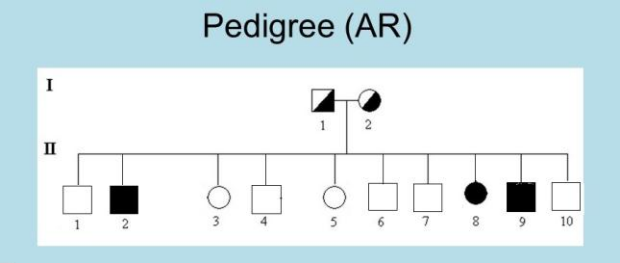

Figure 1: Pedigree analysis for the patient's family.

Detailed physical examination

Patient look well with no ecctropion nor eclabion

There is generalized intense erythema with fin white powdery scales.

There is transgradient diffuse fissuring keratoderma involving both palms and soles.

The nails are normal; there is no scarring alopecia and normal oral mucosa.

Systemic examination was carried out which revealed no abnormalities.

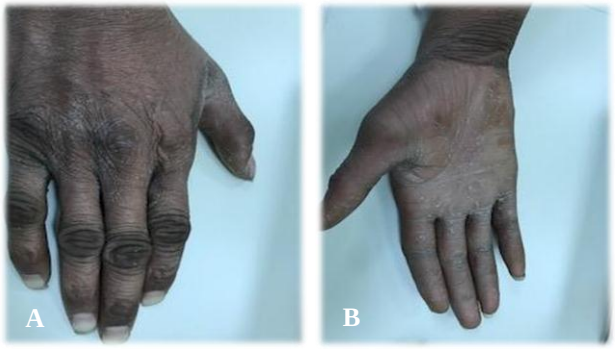

Figure 2 (A and B): Skin dryness of upper arms of the case (back and palms of the patient's hands).

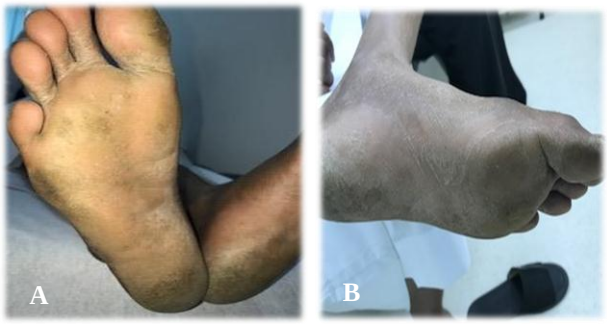

Figure 3 (A and B): Skin dryness of lower limbs of the case (soles of the patient's feet).

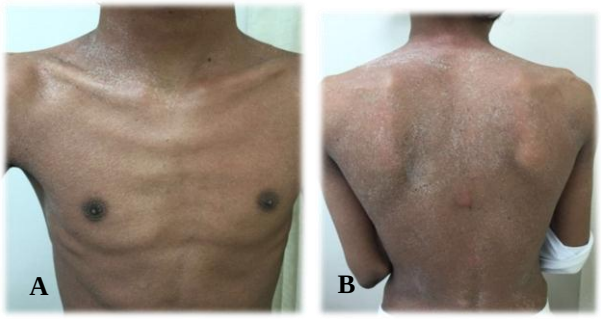

Figure 4 (A and B): Over the anterior and posterior trunk; the skin is dry and erythematous with fine white scales.

Detailed investigations

As a routine lab work and to start medication (complete blood count, liver profile, lipid profile and urinalysis) requested for the patient. All the results of these investigations were normal.

Genetic analysis was ordered as well and it shows abnormal findings and identified the homozygous or hemizygous variant ABCA12 c.

Detailed treatment

Patient was on topical treatment (emollient creams and keratolytic creams) for long time with no improvement because of miss diagnosed from the beginning.

The dermatology team in King Fahd armed forces hospital (KFAFH) discussed the case and decided to give the patient the following:

1) Acitretin oral tablet 10 mg once daily.
2) Follow up appointment after one month.

After one month, the patient got excellent improvement in terms of no more skin scale and no more thickening of keratoderma in palms and soles and the patient actually start to sweat normally.

The dermatology team put the patient on regular follow up to monitor the improvement and side effects of the acitretin.

*Invitation of other affected family members was done and the same investigations and treatment approach done for them.

DISCUSSION

Non bullous congenital ichthyosiform erythroderma is a rare type of ichthyosis that occurs in 1 in 200,000 to 300,000 births.[11] NBCIE is an autosomal recessive genetic disorder. This means you need to inherit a defective pair of genes (one from each parent) to show the symptoms. Parents who are carriers of the defective genes show no symptoms but their children have a 25% chance of having NBCIE.

Non syndromic autosomal recessive ichthyosis have been divided into two major clinical entities, nonbullous congenital ichthyosiform erythroderma (NBCIE) and lamellar ichthyosis (LI). Although the management plan will not change as that much but the nature of scaling and intensity of erythroderma are important clinical features that distinguish between NBCIE and LI. In NBCIE, the entire body is covered in erythroderma skin with fine white or light grey scales and feathery. Ectropion and eclabium are frequently seen, but not severe as LI.[7]

Histopathology examination is not specific for NBCIE. However, lamellar ichthyosis (LI) will be found moderate to mild acanthosis, mild parakeratosis, stratum corneum thickness at least twice than NBCIE.[12]

The management of all types of dry skin consists of retention of water loss, rehydration, and softening of skin, and alleviation of scalliness and associated pruritus. Patients can (NBCIE patients can use) topical application of keratolytic agents, emollients and topical or systemic retinoids. Oral retinoids such as isotretinoin and acitretin have led to dramatic improvement in some pediatric patients with the ichthyosis, but should be used with caution because of side effect that limit long term therapy, particularly liver toxicity. These systemic agents are generally reserved for use in adolescents and adults with more severe ichthyosis disorders that do not show a satisfactory response to topical agents.[8]

Diagnosis of NBCIE is established from history taking, clinical features, and gene analysis. Systemic Retinoid (isotretinoin and acitretin) are very effective, but careful monitoring for toxicity is required. Only severe cases may require intermittent therapy.

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