Case report

Congenital ichthyosiform erythroderma with non- syndromic clinical presentation of short stature

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ABSTRACT

In this article we report a 10 years old boy with congenital ichthyosiform erythroderma and short stature.He was born as a collodion baby. Skeletal manifestations were short stature (Height zscore of -5), delayed bone age with normal serum calcium and phosphorus level and upper to lower ratio of 1.09 (U/L=1.09). This case showed an increasing ALP and LDH in several lab data. Lab evaluations of hair, eyes, ears and mental status were all normal. In the skeletal radiographies there was no finding to support Rickets.

The cited features do not match with syndromes that contain congenital ichthyosiform erythroderma such as Netherton syndrome, Tay syndrome, Run syndrome etc.

Please provide your thoughts or other syndromes to consider to aid in the treatment of this child.

INTRODUCTION

Normal growth is the final common pathway of many factors, including endocrine, environmental, nutritional, and genetic influences [1].Growth failure denote a slow growth rate regardless of stature [2]. It can be caused by numerous conditions, such as constitutional delay, Genetic short stature, GH deficiency, Turner syndrome, etc. Epidermolytic hyperkeratosis (Bullous Ichthyosiform Erythroderma) is inherited as an autosomal dominant trait although

many of them are sporadic and characterized by the onset at birth of generalized erythroderma and severe hyperkeratosis[3].

We reported a case of congenital Ichthyosiform Erythroderma with growth retardation.

CASE

In this study a 10 ¬year old Iranian male was referred to the pediatric endocrinology ward, Imam Reza Hospital, Mashhad, Iran, he presented with congenital ichthyosiform erythroderma and growth retardation. In his past medical history, he was born a collodion baby. Then xerosis and thick scales was appeared throughout the body except palmes and soles that improved with emollient partially. About 6 to 7 month ago, bullous lesion and secretory erosions also was appeared on the his skin. His birth height was 48 cm and birth weight was 2850 gr. He started trying to walk in 2 years old, but it took 7-8 months for him to walk. Totally his growth was less than normal children. He was the second child of non consanguineous marriage and normal vaginal delivery. His mother had 2 live born and one abortion. He was admitted in hospital one time for ictere and recived phototherapy and second time for bullous lesion few months ago, but he hadn't other diseases and surgery and admission. In their family history, his sister had a little hearing loss since her birth. But there aren't like this history in other members of his family. In drug history, he only used antibiotic and emollient for skin lesions. On admission, he weighted 22kg (z-score -1.44), his height was 113cm (z-score -5) and upper to lower ratio reported as 1.09. The height of his father is 160cm and his mother is 150cm and his sister is 155cm. On physical exam,vital sign was normal. Laboratory investigation revealed serum Calcium of 11mg/dl, phosphorus of 3.97mg/dl, Alkaline phosphates of 1045mg/dl and Lactate dehydrogenises of 2273 mg/dl. A hematological study showed Ferritin 33 with normal CBC. Urine analysis also was normal. In hormonal profile, a total of T4 9.7micgr/dl, TSH 1.8micIU/ml, FSH 1.4mlU/ml, LH 0.9 mlU/ml, IGF-1 70ng/ml and IGF-BP3 2337ng/ml was recorded, that all of them were in normal ranges for age and race. Serum Albumin (4.26g/dl) and Anti TTG(1.5U/ml) also were in normal range.

In wrist radiography, bone age in ulnar head epiphysis was conformed to 6-7 year old and with no rickets

presentation. So he had delayed bone age. In sonographic study, liver with normal size and increased echogenesity was reported. Sonographic study of Gall bladder and Billiary tract, kidneys, Spleen, Pancreas, Urinary bladder showed they were all normal.
In pathological examination the specimen consisted of skin, hyperkeratosis epidermis and epidermolysis with small intraepidermal bula and increased granular layer thickness and many intracellular keratohyalan granules, that this morphology conform with EpidermolyticHyperkeratosis.
Also Hearing studies and slit lamp testing of the Eyes showed, all parts were normal. Mental status didn't have significant abnormality.
GH provocative tests was not done , because blood sample due to skin lesions can not to take.

DISCUSSION:

Bullous ichthyosiform erythroderma(BIE) is a rare autosomal dominant disorder of keratinization that,in its early phase, is associated with blistering. In pathogenesis, the aggregation of tonofilamants in suprabazal keratinocytes in BIE is suggestive of an underlying genetic defect of keratin synthesis or degenaration, involving keratins K1 and/or K10, as they are distributed only in the suprabazal compartment in normal epidermis. BIE typically presents with fragile skin(epidermolysis), which gives way to progressive hyperkeratosis. A mild, generalized erythroderma is present at birth. Flaccid blisters, peeling and superficial erosions at sites of minor trauma or friction are apparent within the first few hours of life. Frequent misdiagnoses at this stage include staphylococcal scaled skin syndrome and epidermolysis bullosa. The superficial erosions heal rapidly without scarring, and easy blistering ceases in the few months of life. Palmoplantar hyperkeratosis develops in approximately 60% of patiens with BIE and may result in recurrent painful fissures,contractures, sclerodactyly and foot deformity with impaired function. Nail dystrophy is rare, although curvature may result. Severely affected children may be of short stature, although many catch up in adolescence. Rickets due to vitamin D deficiency has been reported.[4]
Several syndromes which include ichthyosis as a constant feature have been established as rare but distinct entities, for example, Netherton

syndrome is characterized by ichthosis, trichrrhexis invagination and other hair shaft abnormality and atopic diathesis [3]

Refsum syndrome includes ichthyosis with polyneuritis, retinit pigmentosa anosmia deafness, bony abnormalities and electrocardiographic changes [3]. Chondrodysplasic punctata is characterized by ichthyosis with cataract, hypertelorism, optic nerve atrophy, disproportionate shortening of the proximal extremities, psychomotor retardation, failure to thrive and spasticity [3].

Other syndromes such as Run syndrome, Tay syndrome, etc, also include ichthyosis.

Thomas P.Senter et al. reported a case of atypical ichthyosiform erythroderma and congenital neurosensory deafness, post natal growth deficiency and other manifestation.

To our knowledge our patient have a congenital ichthyosiform erythroderma with short stature, such that We couldn't find a reason for growth retardation in him.

We have several problems with our case. Initially we couldn't explain the reason of presentation of him based on the other distinct syndromes. Secondary, we couldn't explain the abnormal laboratory results e.g. raised LDH or hyperechogenisity of the liver in sonography. So we hope that we can find a treatable etiology for these problems and so give the treatment of choice to our patient. Finally may be our patient a case report as a new syndrome?

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