X-Linked Ichthyosis, Dextrocardia And Septal Defect In A Pair Of Dizygotic Twin

Dizigotik İkiz Eşlerinden Birisinde Görülen X'e Bağlı İktiyozis, Dekstrokardi ve Septal Defektler

Yrd.Doç.Dr. Osman BAŞPINAR[1], Prof.Dr. Yavuz ÇOŞKUN[2], Prof.Dr. Metin KILINÇ[2]
Arş.Gör. Derya AYDIN[2], Doç.Dr. Serhat İNALÖZ[3]

[1]Gaziantep University Medical School Department of Pediatric Cardiology
[2]Gaziantep University Medical School Department of Pediatrics
[3]Gaziantep University Medical School Department of Dermatology

Abstract

The ichthyosis may be found isolated or in association with other genetic defects. A two-month-old in a pair of dizygotic twin boy with ichthyosiform dermatosis, dyspnea, a pansystolic precordial murmur at the right sternal border was referred to our clinics. On physical examination the infant showed ichthyosis, situs inversus totalis, lung diseases, cryptorchidism, and inguinal hernia. An echocardiographic study revealed mirror-image dextrocardia, a large perimembranous outlet ventricular septal defect, subvalvular aortic stenosis and pulmonary hypertension. The other pair of twin girl had no skin changes. We believe this patient represents a new manifestation of ichthyosis with a unique appearance.

Key Words: Ichthyosis, dextrocardia, congenital heart defects, twin

Özet

İktiyozis, izole veya diğer genetik defektlerle birarada bulunabilir. Dizigotik ikiz eşi iki aylık bebekte iktiyoziform dermatozis, dispne, sağ sternal kenarda pansistolik üfürüm duyulması üzerine kliniğimize değerlendirme için sevk edilmişti. Muayenesinde iktiyozis, situs inversus totalis, akciğer hastalığı, kriptoorşidizm ve inguinal herni saptandı. Ekokardiyografide ayna hayali dekstrokardi, geniş perimembranöz outlet ventriküler septal defekt, subvalvular aort darlığı ve pulmoner hipertansiyon saptandı. Cilt ve kalp problemi diğer ikiz eşinde yoktu. Literatür taramasında iktiyozise ait bu klinik duruma daha önce rastlanılmamıştır.

Anahtar kelimeler: İktiyozis, dekstrokardi, doğumsal kalp defekti, ikiz

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INTRODUCTION

X-linked ichthyosis (XLI) is hereditary hyperkeratotic dermatosis in which steroid sulfatase is deficient (1-3). The mode of inheritance is X-linked recessive. Disease is usually present at birth, and ichthyosiform scales tend to be large and dark. They are prominent on the extensor surfaces of the extremities, although there is significant involvement of the flexural areas. The front of the trunk may be affected more severely than the back (1-3). Ichthyosis may be part of a more widespread congenital abnormality. In the review of the literature, ichthyosis and situs inversus totalis in a pair of diyzgotic twin is not found together. We present a two-month-old boy in a pair of twin with ichthyosis, situs inversus totalis and other congenital abnormalities.

Yazışma Adresi:
Yrd.Doç.Dr. Osman BAŞPINAR
Gaziantep Üniversitesi Tıp Fakültesi Pediatri AD
Adres: Gaziantep Üniversitesi Tıp Fakültesi 27310 Gaziantep
Tel: 0342 360 60 60
Fax. 0342 360 39 28
E-mail:osmanbaspinar@hotmail.com

CASE REPORT

A two-month-old boy in a pair of diyzgotic twin was referred for skin problems, lung diseases and cardiac abnormalities. His height and weight was 25-50th percentile. The patient had a pruritic ichthyosiform eruption since at birth (Fig. 1). There was no family history of a congenital disease. The patient did not have any medical care up to that time. He had congestive heart failure and pneumonia findings with hepatomegaly, dyspnea and tachycardia. His heart rate was 165/min, respiratory rate 50/min, temperature 37°C, and blood pressure 60/40 mmHg. Normal heart sounds and grade III/VI pansystolic murmur were at the apical region on the right side of the chest. His liver was palpated three cm below at the left costal margin. He also had cryptorchidism, proximal hypospadias and a large left inguinal hernia. The chest roentgenogram revealed pneumonic infiltration and dextrocardia (Fig. 2). The electrocardiogram showed dextrocardia findings with normal age-related changes. The patient underwent transthoracic echocardiography, which demonstrated a mirror-image dextrocardia, a large perimembranous ventricular septal defect, subvalvular aortic stenosis, moderate pulmonary hypertension. Abdominal ultrasonography confirmed that situs inversus totalis.

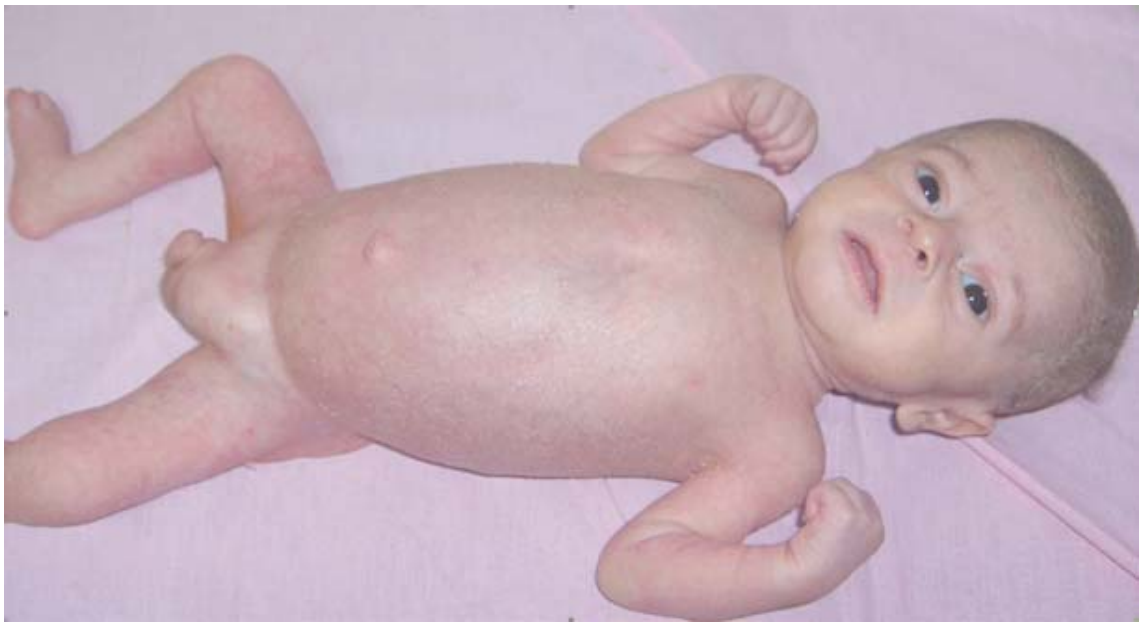

Figure 1. Note the scales involving the body of the patient with X-linked ichthyosis.

He required oxygen and was treated with fluid restriction, furosemide, digoxin, captopril and antibiotic therapy. Routine laboratory testing revealed marked leukocytosis and higher sedimentation rate, and C-reactive protein levels. Other serum biochemistry parameters, metabolic values, blood culture results, and viral and bacterial serological studies were within normal limits. Detailed ophthalmological, neurological and audiometric examinations were normal. He was discharged with inotropic support therapy. At the follow up, cardiac catheterization and operation had been planned, but the patient was referred to our pediatric emergency unit for aspiration pneumonia after three weeks. Immediate suctioning of the airway and administering oxygen were done, but the patient was not survived. The other pair of twin was not same sex and also she had no cardiac diseases and skin problems.

DISCUSSION

Ichthyosis may be found isolated or in association with other genetic defects. These genetic disorders are rare. The following syndromes with ichthyosis may be considered in the differential diagnosis. These syndromes are KID (keratitis, ichthyosis, and deafness), CHIME (colobomas of the eye, heart defects, ichthyosiform dermatosis, mental retardation, and ear defects), Netherton (ichthyosis, erythroderma, hair shaft defects, atopic features), Sjögren-Larsson (ichthyosis, spastic diplegia, pigmentary retinopathy, and mental retardation), Refsum disease (ichthyosis and pigmentary retinopathy) (3-7).

Major features of these syndromes except cardiac diseases were absent in our patient. Although cardiac involvements are not a universal manifestation of the CHIME syndrome, there was not different syndrome consisting ichthyosis and cardiac defects (7). But the other phenotypic features of CHIME syndrome were not absent in the patient.

XLI in girls is rare and has only been described in three instances (8). Therefore, he had XLI features; his twin was no skin problems. Traupe and Happle (1) were found that a causal relationship cryptorchidism and XLI.

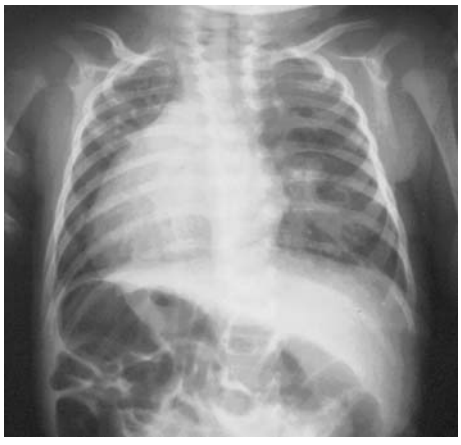

Figure 2. Chest X-ray of the patient. Note the dectrocardia, situs inversus totalis and pulmonary congestion findings.

The patient had bilateral cryptorchidism and a large left inguinal hernia as usual. There is no cure for ichthyosis so we directed at decreasing symptoms regarding skin problems. Cardiac problems were need surgical correction, and then operation was planned. To our knowledge, this is the first report of ichthyosis, a mirror-image dextrocardia, situs inversus totalis, a large ventricular septal defect and subvalvular aortic stenosis. These abnormalities and its relationship to ichthyosis are unknown. This case presented emphasizes the broad range of abnormalities in ichthyosis. These multiple system involvement suggests an event early in the course of fetal development, the specific cause of this condition remains unknown. At the clinical practice should be aware of the possibility. We conclude that the manifestations of ichthyosis are more extensive and cardiac features should be evaluated not only with electrocardiography but also with echocardiography.

REFERENCES

1.Traupe H, Happle R. Clinical spectrum of steroid sulfatase deficiency: X-linked recessive ichthyosis, birth complications, and cryptorchidism. Eur J Pediatr. 1983;140:19-21.

2.Bruno L, Bocanegra O, Magnelli N. Recessive X-linked ichthyosis associated with hypertrophic pyloric stenosis: a chance occurrence? Clin Exp Dermatol. 2003;28:74-6.

3.Okano M, Tanaka H. X-linked ichthyosis associated with congenital dislocation of the hip. Int J Dermatol. 2001;40: 340-2.

4.Miteva L. Keratitis, ichthyosis, and deafness (KID) syndrome. Pediatr Dermatol. 2002;19:513-6.

5.Tursen U, Kaya TI, Ikizoglu G, Aktekin M, Aras N. Genetic syndrome with ichthyosis: congenital ichthyosis, follicular atrophoderma, hypotrichosis, and woolly hair; second report. Br J Dermatol. 2002;147:604-6.

6.Zunich J, Esterly NB, Holbrook KA, Kaye CI. Congenital migratory ichthyosiform dermatosis with neurologic and ophthalmologic abnormalities. Arch Dermatol. 1985;121:1149-56.

7.Schnur RE, Greenbaum BH, Heymann WR, Christensen K, Buck AS, Reid CS. Acute lymphoblastic leukemia in a child with the CHIME neuroectodermal dysplasia syndrome. Am J Med Genet. 1997;72:24-9.

8.Thauvin-Robinet C, Lambert D, Vaillant G, Caillier P, Donzel A, Cusin V, et al. X-linked recessive ichthyosis in a girl: strategy for identifying the causal mechanism. Br J Dermatol. 2005;152:191-3.