CASE REPORT OLGU SUNUMU

Case Report of Rickets Due to Vitamin D Deficiency Associated Ichthyosis

İktiyozis İlişkili Vitamin D Eksikliğine Bağlı Rikets Olgu Sunumu

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Abstract

Exposure of keratinocytes in the skin to sunlight is an important source of vitamin D. Vitamin D deficiency can occur in skin diseases involving keratinization disorders such as ichthyosis. In this case, aged six years and six months, ichthyosis-related symptoms include erythema, widespread desquamation and xerosis throughout the body, along with rickets-related frontal bossing, widening of the wrists and Harrison's groove. The investigations revealed that rickets was associated with vitamin D deficiency. This report emphasizes the need for screening for vitamin D deficiency and prophylaxis in patients with chronic skin diseases.

Öz

Ciltteki keratinositlerin güneş ışığına maruz kalması önemli bir vitamin D kaynağıdır. Vitamin D eksikliği, iktiyoz gibi keratinizasyon bozukluklarını içeren cilt hastalıklarında görülebilir. Bu altı yaş altı aylık olguda, iktiyozise bağlı vücutta yaygın kızarıklık, pullanma ve kurulukla beraber raşitizmle ilişkili alın çıkıntısı, bileklerde genişleme ve Harrison oluğu tespit edilmiştir. Olguda yapılan tetkikler sonucu vitamin D eksikliğine bağlı rikets olduğu saptandı. Bu olgu raporu kronik cilt hastalıkları olan hastalarda vitamin D eksikliği taraması ve profilaksisinin gerekliliğini vurgulamaktadır.

Keywords
Ichthyosis, vitamin D, rickets, hypocalcemia

Anahtar kelimeler
İktiyozis, Vitamin D, rikets, hipokalsemi

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Introduction

Rickets is a clinical condition characterized by a mineralization defect of the epiphyseal plates. Pathological conditions affecting vitamin D, calcium, and phosphorus metabolism, which are essential for mineralization, can lead to rickets. The most common cause is vitamin D deficiency (1). Clinical findings are mostly seen in the skeletal system, such as forearm deformities in crawling children, "O" or "X" shaped legs in toddlers, "rachitic rosary" due to enlargement at the costochondral junctions, prominence in the frontal part of the skull, and delayed tooth eruption. Extra-skeletal findings such as muscle weakness, hypotonia, tetany, laryngospasm, and growth retardation may also occur (2).

The main source of vitamin D is the conversion of 7-dehydrocholesterol to cholecalciferol (vitamin D3) in keratinocytes after exposure to sunlight (3). Therefore, various skin diseases, especially keratinization disorders, can impair

vitamin D metabolism and lead to vitamin D deficiency. Cases of vitamin D deficiency have been reported in many skin diseases such as atopic dermatitis, xeroderma pigmentosum, and ichthyosis (4). Here, a case of rickets due to ichthyosis vulgaris is presented due to its rarity.

Case Report

A 6-year and 6-month-old male patient presented to the pediatric neurology outpatient clinic with complaints of inability to walk. On physical examination, his body weight was 13.2 kg (SDS Z score -4.2), and height was 95 cm (SDS Z score: -4.9). On head and neck examination, there was mild erythema on the face, ectropion, and widespread, superficial, white powdery scales. The frontal region of the skull was prominent. Only the two upper incisors and molars were present, and the teeth were hypoplastic. Pectus carinatum deformity, Harrison's groove, and widening of the wrists were observed. There was widespread desquamation and xerosis throughout the body. Neurological examination revealed spasticity in both lower extremities, while other system examinations were normal. The patient's physical examination findings are shown in Figure 1 and Figure 2.

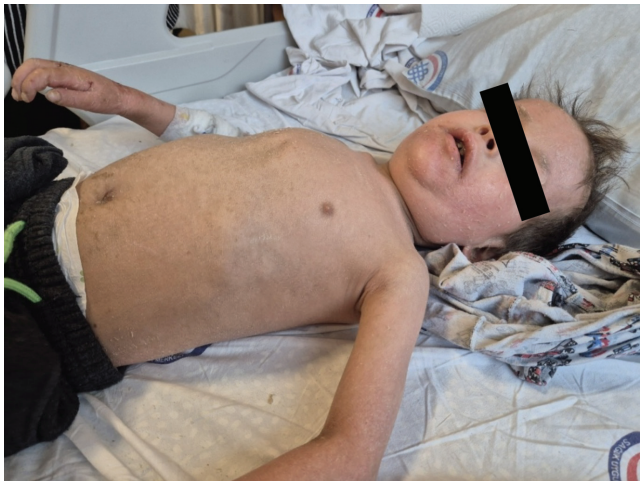

Figure 1. Widespread desquamation and xerosis throughout the body

In his medical history, it was learned that he was born at 33 weeks by normal delivery, weighing 1800 grams, and was hospitalized in the neonatal intensive care unit for one month due to prematurity. He was diagnosed with ichthyosis vulgaris in the neonatal period and had been receiving psoretin treatment for 3 years. The patient, who used bilateral cochlear implants due to hearing loss, was being followed by pediatric neurology and pediatric genetics units at another center. There was no consanguinity between the parents. His siblings were alive and healthy.

Laboratory tests revealed alkaline phosphatase 1179 U/L (142-335), calcium 6.02 mg/dl (8.8-10.8), albumin 4.19 mg/dl (3.5-5.2), phosphorus 3.78 mg/dl (3.3-5.6), magnesium 1.76 mg/dl (1.6-2.6), parathyroid hormone 597 ng/l, and 25-hydroxy vitamin D 3.26 mg/L. X-ray of the left wrist showed a cup-shaped appearance and fraying. Bone age was compatible with 3.5-4 years (Figure 3).

Based on the current examination and laboratory findings, the patient was evaluated as having rickets due to vitamin D deficiency. After a single dose of intravenous

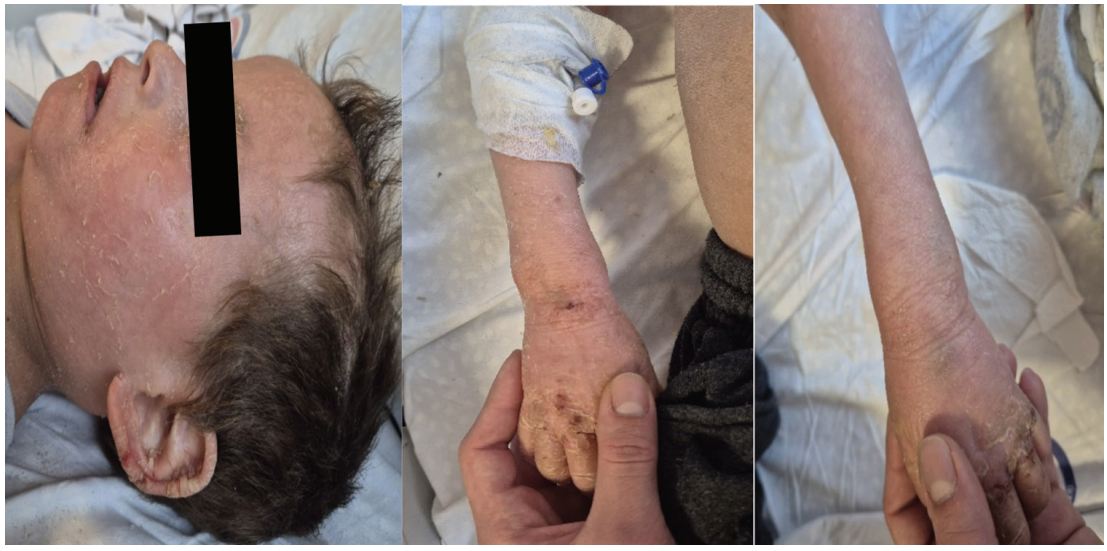

Figure 2. Frontal bossing and widening of the wrists

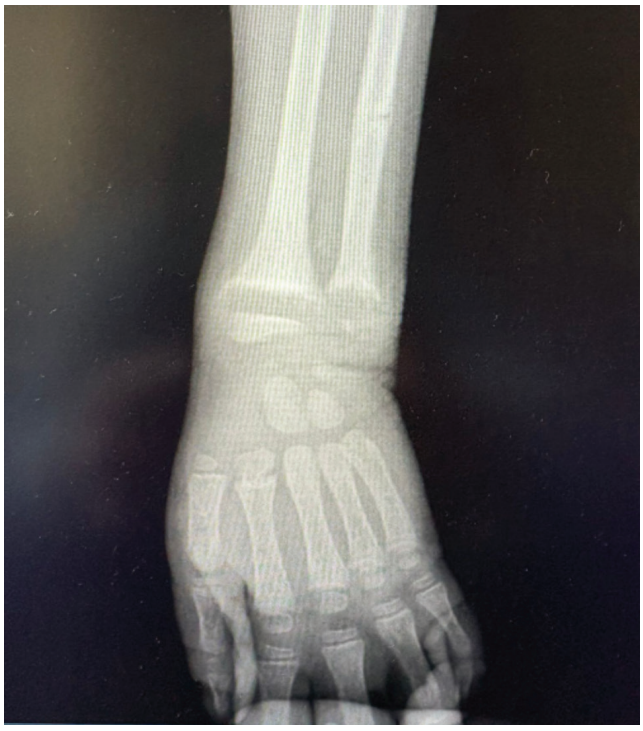

Figure 3. X ray of upper limb showed marked cupping and fraying of distal metaphyses

calcium, 75 mg/kg elemental calcium and 5000 IU/day vitamin D supplementation were started. Upon normalization of calcium levels in follow-up blood tests, the patient was discharged with oral treatments arranged and routine follow-ups planned. At the one-month follow-up, calcium was 9.3 mg/dl, alkaline phosphatase 739 U/L (142-335), and parathyroid hormone 114 ng/l.

Discussion

Here, a rare case of rickets due to ichthyosis vulgaris is discussed. The most common cause of rickets worldwide is vitamin D deficiency. The most common risk factors for vitamin D deficiency are winter and spring seasons, dark skin color, covered clothing, reduced sun exposure, insufficient dietary intake, and malabsorption. Nutritional causes and low sun exposure are the most prominent factors (5). One of the important steps in vitamin D metabolism begins in the skin. Even if sun exposure and nutrition are normal, vitamin D deficiency can develop in pathological skin conditions (4,6). Numerous studies have demonstrated an association between low vitamin D levels and conditions such as vitiligo, psoriasis, rosacea and, in particular, atopic dermatitis in children (6-8). In this case, rickets due to ichthyosis was considered.

Ichthyoses are a group of skin keratinization disorders characterized by dry, rough skin and prominent scaling. They exhibit an autosomal semi-dominant inheritance pattern with a milder phenotype in heterozygotes due to loss-of-function mutations in the filaggrin gene (FLG). Clinical findings include xerosis, keratosis pilaris, palmar hyperlinearity, and a predisposition to atopic disorders (9). Risk factors for the development of rickets in patients with ichthyosis include impaired vitamin D synthesis in diseased epidermis, poor penetration of ultraviolet B rays into the epidermis due to widespread scaling, reduced sun exposure due to discomfort from sunlight, dark skin, loss of calcium through the skin, and the use of systemic retinoids that reduce intestinal calcium absorption (4). Ichthyosis has its own risk factors and shows some differences from nutritional rickets. Known differences include consanguinity between parents, similar skin lesions in siblings, and discomfort from sun and heat (10). In this case, consistent with the literature, there were risk factors such as skin findings, retinoid use, consanguinity between parents, discomfort from sunlight, and features distinguishing it from nutritional rickets.

No typical skin lesions are associated with vitamin D deficiency. However, alopecia is observed in vitamin D-dependent rickets type 2. The most important difference between these two pathological conditions is that 1,25-dihydroxyvitamin D levels are high in vitamin D-dependent rickets and, despite treatment for rickets, the alopecia does not improve (11). Vitamin D deficiency has been demonstrated in inflammatory skin diseases characterised by itching, such as ichthyosis, and numerous studies have reported the potential benefits of using vitamin D topically or systemically (6).

In children with rickets, secondary hyperparathyroidism develops due to decreased vitamin D and hypocalcemia. Among the different combinations of threshold levels of serum 25-hydroxyvitamin D and parathyroid hormone, the risk of developing rickets in children with congenital ichthyosis is higher when serum 25-hydroxyvitamin D is ≤8 ng/ml and parathyroid hormone is ≥75 pg/ml (12). In this case, consistent with the literature, parathyroid hormone was 597 ng/l and 25-hydroxyvitamin D was 3.26 ng/ml.

It is recommended that patients with ichthyosis be advised about adequate sun exposure, have their vitamin D levels checked even in the absence of clinical signs of rickets, and receive lifelong “rickets prophylaxis” with periodic vitamin D supplementation(4,10).

Conclusion

Because the risk of developing rickets is quite high in cases of ichthyosis vulgaris, vitamin D levels should be monitored during follow-up. Preventing serious complications with oral treatment or prophylaxis is easier than dealing with the complications of rickets.

Footnotes

Conflict of Interest: The authors reported no potential conflict of interest.

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