A Case Report of Ichthyosis Vulgaris with Arthropathy and Ophthalmic Findings


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Abstract

Observation: Ichthyoses are a group of genetic disorders of keratinization. Ichthyosis vulgaris is a disease that mainly affects skin and hyperkeratosis and scales over a large part of the body surface. Ichthyoses may sometimes cause eye disorders, but eye findings are seen rarely in ichthyosis vulgaris. Joint findings might be seen in some syndromes accompanied with ichthyoses, as well as joint disorders could be accompanied with ichthyoses. But ichthyosis vulgaris does not lead to joint involvement. To our knowledge there haven’t been a reported case of joint contracture associated with ichthyosis vulgaris in the literature. Here we report an 18-year-old female patient of ichthyosis vulgaris associated with arthropathy, joint contractures, ectropion and lagophthalmus.

Introduction

Ichthyoses are a group of genetic disorders of keratinization characterized by hyperkeratosis and scales over a large part of the body surface. Ichthyoses are divided into two main groups as syndromic and nonsyndromic. Six distinct types of inherited non syndromic ichthyosis are known as ichthyosis vulgaris, non-bullous congenital ichthyosiform erythroderma, lamellar ichthyosis bullous congenital ichthyosiform erythroderma, X-linked ichthyosis and harlequin ichthyosis [1]. Ichthyosis vulgaris is a disease that mainly affects only skin and does not lead to joint involvement. Sometimes it can cause ophthalmic disorders. Progressive subepithelial cicatrization and abnormal cornification of eyelid skin causes progressive ectropion in both eyelids,

Figure 1. Thick scales on her back and extremities
which leads to restrictive lagophthalmos and corneal complications. An 18-year-old female patient of ichthyosis vulgaris associated with ectropion, arthropathy and joint contractures is reported.

Case Report

An 18-year-old woman presented to our clinic with complaints of erythema and scaling of her body. Changes in the hands of patients had started at an early age and since five years of age she gradually wasn’t able to use her hands. Her parents had a consanguineous marriage and she had a 9-year-old sister with similar symptoms in her family. Furthermore the patient was complaining of erythema and tearing in both eyes.

On dermatological examination there were thick scales on her trunk, back and extremities and fine scales on her face (Figure 1). Flexural areas of her extremities were less prominent. There was mild palmo-plantar hyperkeratosis. Hypohidrosis or alopecia were not present. Atopic dermatitis or keratosis pilaris was not present. Her skin biopsy was nonspecific and showed compact hyperkeratosis, focal parakeratosis, acanthosis, elongation of rete ridges and normal dermis.

Although ophthalmologic examination was normal while the eyes were open, minimal ectropion in both lower eyelids and restriction in both upper eyelids were determined when eyes were closed (Figure 2). Therefore lagophthalmus was occurred. Bell’s phenomenon was well in both eyes. The corneal epithelium, intraocular pressure and fundus examination were normal for both eyes.

Laboratory investigations including rheumatoid factor, anti cyclic citrullinated peptide antibody, anti nuclear antibody, anti-Extractable Nuclear Antigen profile, C-reactive protein, 25-hydroxy vitamin D, complete blood count, routine biochemistry, lipid profile, hepatitis A, hepatitis B and
hepatitis C markers (IgG and IgM) were done. All of the laboratory investigations were in the range of normal limits or negative.

Hand radiographs were performed for joint symptoms. The X-ray of the hands revealed narrowing of the interphalangeal spaces, subluxations, contractures and ulnar deviation of 2, 3, 4, and 5th metacarpophalangeal (MCP) joints, and narrowing of interphalangeal spaces of proximal interphalangeal (PIP) joints in both hands. Flexion contractures were observed in 2, 4, 5th distal interphalangeal (DIP) joints of the left hand and in 2, 5th DIP joints of the right hand. There were acro-osteolysis of the some of the distal phalanxes of both hands (Figure 3). These radiographic findings suggested us that this case had nonspecific arthropathy and joint contractures (Figure 4).

Treatment with 35mg/day acitretin and topical emollients were started. Skin lesions showed improvement. Stretching exercises were begun and surgical release of the flexion deformities of the hands was planned. The patient started on artificial tear eye drops. Patient is still under follow-up and doing well.

Discussion

Autosomal co-dominant ichthyosis vulgaris usually appears within the first year of life. Pruritus, palmar and plantar hyperlinearity, atopic dermatitis, and keratosis pilaris are often seen in ichthyosis vulgaris. The cause of the disease is filagrin gene mutations [2,3]. Our patient was normal at birth and her skin lesions had began after 5-6 months from birth. Presence of lesions on entire of the trunk was compatible with ichthyosis vulgaris.

Ichthyosis vulgaris is a disease that usually confined to the skin, but occasionally may be accompanied by eye disorders like the other types of ichthyosis. Cicatricial ectropion is a common ophthalmic feature of patients with ichthyoses. Progressive subepithelial cicatrization and abnormal cornification of eyelid skin cause progressive ectropion in both eyelids, which leads to restrictive lagophthalmus and corneal complications [4]. Although the presence of upper eyelid restriction and mild lower ectropion, normal Bell’s phenomenon may explain the good condition of the ocular surface in our patient.

Some rare syndromes are associated with “vulgar” ichthyoses and these syndromes may be associated with articular manifestations. Multiple sulfatase deficiency and Refsum disease are syndromes associated with ichthyosis vulgaris [3]. Several joint disorders associating with ichthyoses have been reported such as digital contracture with palmoplantar keratoderma [5], two cases of ichthyosiform erythroderma associated with severe bilateral genu valgum [6], a stillborn female with joint contractures, subcutaneous oedema, ectropion and extensive peeling of skin [7], joint hyperextansibility in a patient with Sjögren-Larsson syndrome [8], acroosteolytic changes and Jaccoud arthropathy in a patient with keratitis, ichthyosis, deafness (KID) syndrome [9], flexion contractures of extremities, bilateral talipes equinovarus and hands fixed in a clenched position in a stillborn female with Neu-Laxova syndrome [10]. We could not find any reported case in the literature about joint contracture associated with ichthyosis vulgaris.

Ichthyosis vulgaris is usually a benign disease and only rarely is accompanied by other systemic findings. Since ichthyosis vulgaris may also present with many syndromes, it is very important to make a through systemic examination. In this case, severe ichthyosis vulgaris had been associated with eye symptoms and arthropathy and joint contractures.

References


