Coexistence Of Ichthyosis and Ankyloglossia Admitted to the Family Medicine Outpatient Clinic: A Rare Case Report

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Abstract

Family Medicine is a primary care service where undifferentiated patients apply and low prevalence medicine service is provided. Ichthyosis vulgaris is the most common type of ichthyosis, accounting for more than 95% of all ichthyosis cases. The prevalence of this inherited disease, which is caused by a mutation in the filagrin (FLG) gene, is between 1/250 and 1/100. Xerosis, keratosis plaris, palmar hyperlinerity are seen in the patients. Ankyloglossia, on the other hand, is a congenital anomaly that limits the normal movements of the tongue, and its incidence in society varies between 4% and 10.7%. In this case report, for the first time in the literature, the association of ankyloglossia was reported in a patient with ichthyosis. With this case, we tried to draw attention to the importance of undifferentiated case and low prevalence medicine in Family Medicine.

Keywords: Ankyloglossia, family practice, ichthyosis vulgaris, low prevalence medicine

1. Introduction

Ichthyosis originates from the ancient Greek word “ichthys” meaning fish. Because this patients have dry, scaly, thickened skin (Fozia et al., 2021). Ichthyosis is a group of heterogeneous diseases that affect the skin both etiologically and clinically (Fozia et al., 2021). Ichthyoses are divided into two as syndromic and non-syndromic/isolated according to their pathophysiology, mode of inheritance and clinical features. It is inherited autosomal recessive, X-linked or autosomal dominant (Oji & Traupe, 2009; Vahlquist et al., 2017). Ichthyosis vulgaris accounts for more than 95% of all ichthyosis cases (Ran Zhu et al., 2017). Ichthyosis vulgaris is skin keratinization disease due to mutation in Filagrin (FLG) gene (Jaffar et al., 2023). It is characterized by diffuse skin dryness, peeling and scaling of the skin, sometimes erythroderma and histopathologically by hyperkeratosis(Jaffar et al., 2023). Unlike other types of ichthyosis, ichthyosis vulgaris has a good prognosis. The clinical course is good with treatments for dehydrating the skin. In other forms of ichthyosis, genetic treatments and different treatments that are in the trial phase can be applied.(Dorf, Lunen, & Koppelhus, 2021; Nouwen et al., 2022).

Ankyloglossia, also known as tongue tie, is a congenital anomaly that limits the normal movements of the tongue (Rowan-Legg et al., 2015). Its incidence varies between 4% and 10.7% (Segal et al., 2007). The most common symptom in the infancy is feeding difficulties. However, there are also studies showing that ankyloglossia is associated with gastroesophageal reflux, ear pain, growth retardation, and colic (Kotlow, 2011; Siegel, 2016). In older children, speech disorder, sleep apnea, headache, back and neck pain, dental problems may be associated with ankyloglossia (Chinnadurai et al., 2015).

2. Case

A 3-year and 4-month-old boy was brought to the Family Medicine Outpatient Clinic by his family with complaints of sore throat and runny nose for a few days. The patient had no known disease other than ichthyosis vulgaris. His current vaccinations were complete. He had no history of allergies. In her family history, her mother had a non-toxic multinodular goiter, and her father did not have any disease. The patient's siblings did not have any known diseases. At admission, fever: 36.3°C, heart rate: 73/min, arterial blood pressure: 100/60 mmHg, oxygen saturation was 98%. His height was 95 cm (10-25 percentile), body weight was 13 kg (3-10 percentile).

In the physical examination of the patient, there was mild serous discharge from the nose. Oropharynx was slightly reddened, postnasal serous discharge was present. On the ventral surface of the tongue, the frenulum was thick and extended to the apex of the tongue (Fig. 1). The patient's skin was erythematous, dry and squamous. Other system examinations were normal. The patient did not have a speech disorder. The possible risks of ankyloglossia were explained to the patient in detail and the patient was referred to the Ear Nose and Throat Polyclinic.

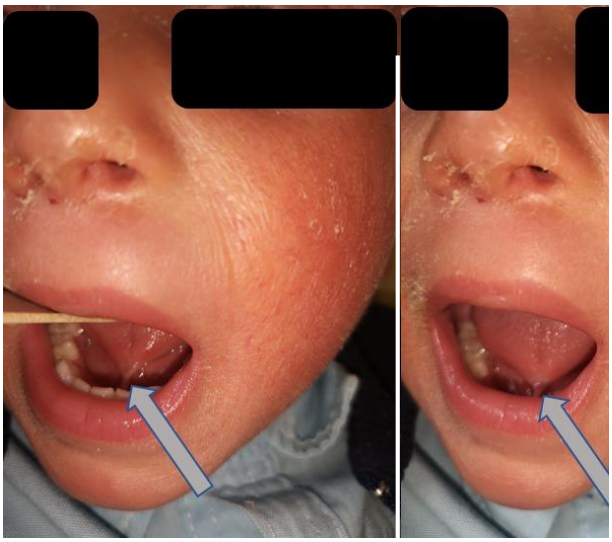

Figure 1. Ankyloglossia in our patient.

3. Discussion

In the consensus statement published by the World Organization of National Colleges, Academies and Academic Associations of General Practitioners/Family Physicians (WONCA) in 2002 and revised in 2011, the basic competencies of the Family Medicine branch were determined under the main headings. These headings are; original problem-solving skills, comprehensive approach, holistic approach, community-oriented, person-centered care and primary care management (Göktaş, 2022).

The number of applications to family medicine for the year 2021 published by the Ministry of Health was approximately 240 million. In the same year, the total number of applications to secondary and tertiary health institutions was approximately 430 million. As can be seen, the role of the family medicine system alone in the provision of health services is quite large. The important function of low prevalence medicine emerges at this point. The family physician can reduce the cumulative burden on the health system by evaluating the patient who applied to him holistically and comprehensively within the scope of core competencies.

Since family medicine is the first point of contact for the undifferentiated patient, it is a low prevalence medicine. Low prevalence medicine requires more comprehensive evaluation, detailed physical examination and a holistic approach. This mission imposes extra responsibility on the Family Medicine branch.

To the best of our knowledge, there is no study in the literature reporting coexistence of ichthyosis vulgaris and ankyloglossia. Ichthyosis can be seen syndromic or non-syndromic/isolated. Ichthyosis vulgaris is the most common and non-syndromic form. It is inherited in an autosomal dominant manner (Oji et al., 2010). Due to the FLG gene mutation, which is the underlying disorder of the disease, hydration is decreased in the stratum corneum layer of the skin. As a result, the skin cannot retain water and the clinic develops accordingly (Irvine et al., 2011; Smith et al., 2006). Ichthyosis vulgaris is a milder clinical type than syndromic ichthyosis types (Karaduman, 2011). The case presented here is ichthyosis vulgaris and its clinical course is moderate.

While ankyloglossia causes difficulties in feeding especially in infancy, it can give different clinical findings with age (Chinnadurai et al., 2015; Kotlow, 2011; Siegel, 2016). According to the history taken in our case, ankyloglossia did not give any clinical twist. However, the patient's weight percentile was below the average.

In the presented case, besides the known diagnosis of ichthyosis vulgaris, the presence of ankyloglossia was detected. There are studies showing that ankyloglossia may cause growth and development retardation (Kotlow, 2011; Siegel, 2016). It can be thought that the low weight percentile of the patient may have been caused by ankyloglossia. For this reason, the patient was referred to the advanced step with a comprehensive and holistic approach.

4. Conclusion

In the context of the fact that the incidence of diseases in primary care will be the same in parallel with the incidence of all diseases in the society, it would be appropriate for family physicians to act in accordance with the low prevalence medicine approach. In this context, since patients are the first point of reference to the health system, the approach of family physicians to the undifferentiated patient in primary health care delivery should include taking a full anamnesis and performing a full physical examination. With this approach, it will be possible to identify possible diseases.

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