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Review Article

Vulgaris Ichthyosis Genetic Predisposition and Histopathological Features: A Review Article

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Abstract

Introduction: Ichthyosis is a skin disease characterized by changes in the skin, especially thickening of the stratum corneum, xerosis cutis, and the form of scaly skin all over the body. Ichthyosis is a disease that is rare, in many case studies, ichthyosis has an unfavorable prognosis in certain types and results in death in the patient over a short period of time. The etiology of ichthyosis is generally caused by gene mutations during fetal development and is largely inherited by hereditary history, whereas ichthyosis is associated with systemic diseases that interfere with the keratinization process of the human epidermal, causing ichthyosis. The histopathological features that are often found in ichthyosis vary, depending on the type of ichthyosis. Some of the features that can be found include hyperkeratosis, hypergranulosis, epidermolysis, orthokeratosis, parakeratosis, acanthosis and so on. Given the importance of histopathological examination in establishing cases of ichthyosis, an understanding of vulgaris ichthyosis needs to be further understood.

Discussion: Ichthyosis vulgaris is an inherited autosomal disease that usually appears in early childhood. The clinical features of ichthyosis vulgaris include dry, scaly skin, and mild itching of the skin. Dry, scaly skin is usually heaviest on the feet, but may also occur on the arms, hands, and midsection, as well as the face and scalp. People with this condition may also have many fine lines over the palms of the hands. The ridges of the hands and feet are usually not involved. More severe clinical features occur because of mutations in both prophylagrine alleles and these disorders are inherited semidominantly The histopathological features that can be found in ichthyosis vulgaris are the thickening of the stratum corneum, thinning or loss of the stratum granulosum, the presence of perivascular lymphohistiocytic infiltrates in the dermis with normal stratum spinosum.

Conclusion: Ichthyosis vulgaris is an inherited autosomal disease, with the highest incidence among ichthyosis. Ichthyosis vulgaris, contribute for about 95% of all cases of ichthyosis with incidence 1: 250 - 1: 1,000. Typical features of Ichthyosis vulgaris, the common histopathological features are hyperkeratosis without the presence of stratum granulosum and loss of rete ridges.

Keywords: Etiology; Genetic; Histopathological

Introduction

Ichthyosis is a skin disease characterized by changes in the skin, especially thickening of the stratum corneum, xerosis cutis, and the form of scaly skin all over the body. The term ichthyosis or ichthyosis comes from the Greek word ichthys which means fish, because this condition resembles fish scales. Ichthyosis is a disease that is rare. In many case studies, ichthyosis has an unfavorable prognosis in certain types and results in death in the patient over a short period of time. The etiology of ichthyosis is generally caused by gene mutations during fetal development and is largely inher-

ited by hereditary history, whereas ichthyosis is associated with systemic diseases that interfere with the keratinization process of the human epidermal, causing ichthyosis [1,2].

Based on the degree of severity, ichthyosis is divided into the most severe forms, namely harlequin ichthyosis (HI), lamellar ichthyosis (LI), congenital ichthroderma (CIE), epidermolytic hyperkeratosis (EHK), recessive X-linked ichthyosis (RXLI) to the lightest form. are ichthyosis vulgaris (IV), and Siemens bullous ichthyosis (SEI). Hereditary (congenital) ichthyosis includes lamellar ichthyosis, epidermolytic hyperkeratosis, and X-linked ichthyosis. Whereas acquired ichthyosis is usually associated with systemic disease. Ichthyosis with the highest incidence is ichthyosis vulgaris. Ichthyosis vulgaris, accounts for about 95% of all cases of ichthyosis. Incidence 1: 250 - 1: 1,000 [3,4].

The histopathological features that are often found in ichthyosis vary, depending on the type of ichthyosis. Some of the features that can be found include hyperkeratosis, hypergranulosis, epidermolysis, orthokeratosis, parakeratosis, acanthosis and so on [5,6]. Given the importance of histopathological examination in establishing cases of ichthyosis, an understanding of vulgaris ichthyosis needs to be further understood.

Discussion

Etiology of vulgaris ichthyosis (IV)

Ichthyosis vulgaris is an inherited autosomal disease that usually appears in early childhood, which is between 3 - 12 months of age. In some studies it was caused by biochemicals, this could only have an effect on the skin. Decreased production of amino acids and some ionic metabolism can reduce water levels in the stratum corneum so that it can cause dry skin and can aggravate this disease, there is no other effect on lipid production that affects ichthyosis vulgaris.

Normal gene expression in profilaggrin can first be detected in the granulosum stratum. In ichthyosis vulgaris, profilaggrin expression is absent or lacking in the epidermis. This biochemical abnormality correlates with the amount of keratohyalin decreased and the severity of the clinical condition. Analysis of keratinocyte culture has shown reduced profilaggrin mRNA. Compared with normal amounts, a study found only 50% of profilaggrin mRNA and 10% of the profilaggrin protein were present. Studies have shown that defective posttranscriptional regulation leads to decreased stability of profilaggrin mRNA [2].

Although the skin in hereditary ichthyosis vulgaris looks and feels normal at birth, it gradually becomes rough and dry in early childhood. The skin that tends to be scaly is the most prominent symptom that is on the extensor surface of the extremities and is absent on the flexor surface. The forehead and cheeks may be affected earlier, but usually the scales of the skin diminish with age [3].

The clinical features of ichthyosis vulgaris include dry, scaly skin and mild itching of the skin. Dry, scaly skin is usually heaviest on the feet, but may also occur on the arms, hands and midsection, as well as the face and scalp. People with this condition may also have many fine lines over the palms of the hands. The ridges of the hands and feet are usually not involved [3]. More severe clinical features occur because of mutations in both prophylagrine alleles and these disorders are inherited semidominantly. Individuals with mutations in one profilaggrin only (heterozygous) allele show mild clinical features occurring in about 60 % cases of ichthyosis vulgaris. The prevalence of this clinical disorder reaches 1 in 80 births in the Anglo-European population [3].

Figure 1: Vulgaris ichthyosis. (A) Thin white scales on lower abdomen, (B) Hyperlinear palms; The first image shows the manifestations of ichthyosis vulgaris in individuals with mutations in both profilaggrin gene alleles, while the second image shows milder manifestations of ichthyosis vulgaris in individuals with mutations in the profilaggrin gene in one allele [3].

Although classic ichthyosis vulgaris is usually absent at birth, the clinical signs of this disorder can manifest itself in the first year of life, with onset at 3 months of age in 40% of cases. Classical ichthyosis vulgaris is characterized by the presence of prominent scales accompanied by smooth, grayish white scales. which are often found larger and more frequent on the surface of the extensor area. There may be fine white scales in a large area, especially in the lower extremities [3]. The absence of the fold area is a characteristic clinical feature of ichthyosis vulgaris which is very helpful in differentiating it from other forms of ichthyosis. The diaper area is less likely to be involved as well [7].

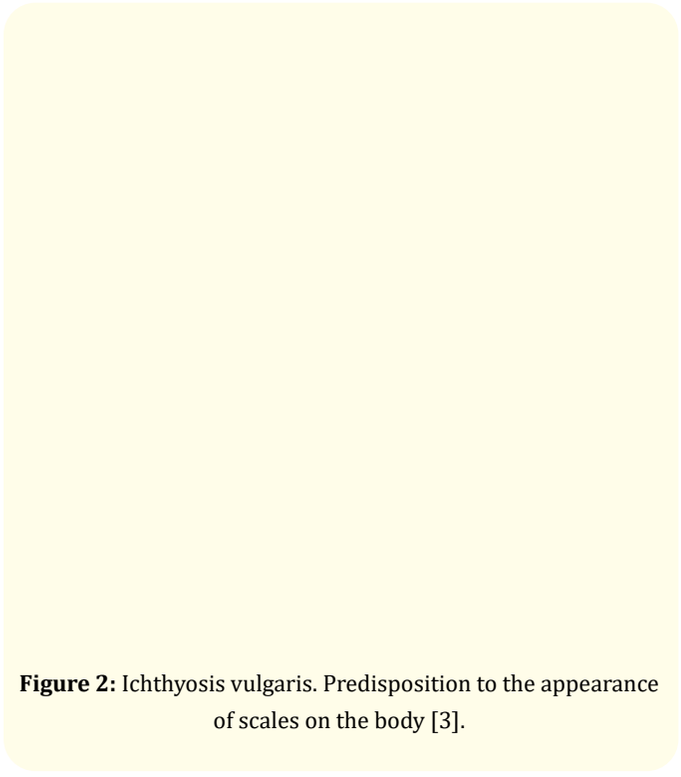

Figure 2: Ichthyosis vulgaris. Predisposition to the appearance of scales on the body [3].

Histopathological examination

The histopathological features that can be found in ichthyosis vulgaris are the thickening of the stratum corneum, thinning or loss of the stratum granulosum, the presence of perivascular lymphohistiocytic infiltrates in the dermis with normal stratum spinosum. Meanwhile, immunohistochemical examination using anti-filaggrin monoclonal antibody showed a marked decrease in the amount of stained filaggrin, indicating a decrease in filaggrin expression in the epidermis in patients with ichthyosis vulgaris [3].

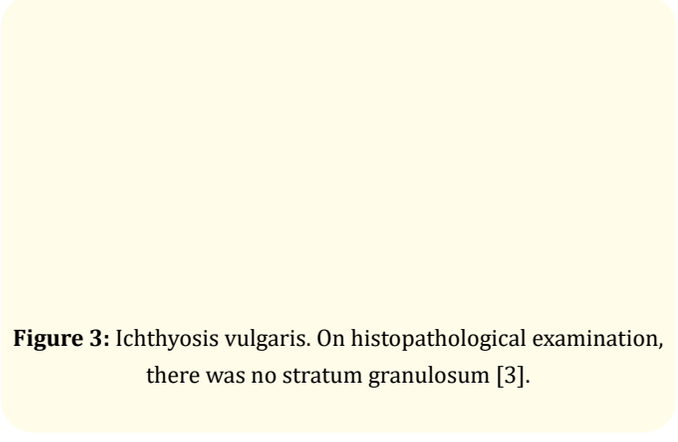

Figure 3: Ichthyosis vulgaris. On histopathological examination, there was no stratum granulosum [3].

The differential diagnosis of ichthyosis vulgaris includes fetal ichthyosis, atopic dermatitis, lamellar ichthyosis, allergic contact dermatitis, recessive x-linked ichthyosis, irritant contact dermatitis, impetigo, and drug eruptions. All of these differential diagnoses can be distinguished based on the pattern of inheritance and from the type and distribution of scales that appear [8].

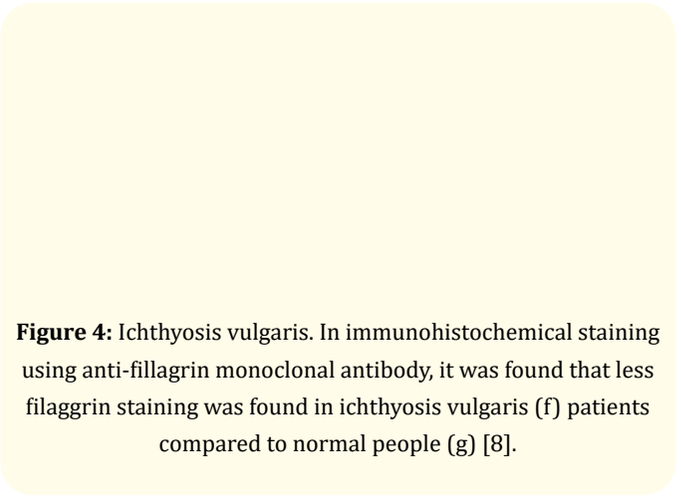

Figure 4: Ichthyosis vulgaris. In immunohistochemical staining using anti-fillagrin monoclonal antibody, it was found that less filaggrin staining was found in ichthyosis vulgaris (f) patients compared to normal people (g) [8].

Complications caused by ichthyosis vulgaris include: fissures in the extremities, secondary infections, and conditions associated with systemic disease. The prognosis for hereditary ichthyosis vulgaris is good with increasing age, whereas ichthyosis vulgaris prognosis depends on the degree of severity of the underlying systemic disease [10].

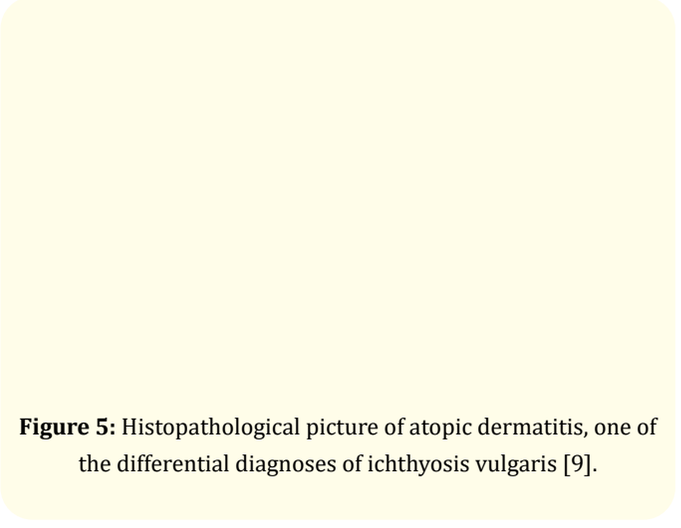

Figure 5: Histopathological picture of atopic dermatitis, one of the differential diagnoses of ichthyosis vulgaris [9].

Conclusion

Ichthyosis vulgaris is an inherited autosomal disease, with the highest incidence among ichthyosis. Ichthyosis vulgaris, contribute for about 95% of all cases of ichthyosis with incidence 1: 250 - 1: 1,000. Typical features of Ichthyosis vulgaris, the common histo-

pathological features are hyperkeratosis without the presence of stratum granulosum and loss of rete ridges.

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