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HEREDITARY SKIN DISEASES AND MODERN TREATMENT METHODS

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Abstract
Hereditary skin diseases are a diverse group of genetic disorders that affect the structure and function of the skin, hair, nails, and other associated tissues. These conditions may be present at birth or develop during childhood and can significantly influence patients' physical health and quality of life. Common hereditary skin disorders include epidermolysis bullosa, ichthyosis, albinism, and inherited forms of palmoplantar keratoderma. This study examines the genetic and clinical characteristics of hereditary skin diseases, modern diagnostic approaches, and recent advances in treatment. Particular attention is paid to molecular diagnostics, personalized medicine, gene therapy, biological treatments, and multidisciplinary patient care. Recent developments in molecular medicine demonstrate that targeted therapies can improve wound healing and reduce disease-related complications in selected hereditary skin conditions.

Keywords: Hereditary skin diseases, genetic disorders, dermatology, epidermolysis bullosa, ichthyosis, molecular diagnosis, gene therapy, biological therapy, personalized medicine.

Introduction

Hereditary skin diseases represent a significant group of medical conditions caused by genetic mutations that affect the development, structure, and functioning of the skin. The skin is the largest organ of the human body and performs several essential functions, including protection against external factors, regulation of body temperature, prevention of water loss, and immune defense. Genetic abnormalities affecting skin proteins, enzymes, and cellular processes may lead to chronic dermatological disorders.

Some hereditary skin diseases are relatively mild, while others can cause severe skin damage, recurrent infections, pain, impaired mobility, and complications involving other organs. Therefore, early diagnosis, appropriate medical care, and continuous monitoring are essential for managing these conditions.

One of the most significant hereditary skin disorders is epidermolysis bullosa (EB). It is a group of genetic diseases characterized by extreme skin fragility and the formation of blisters following minor mechanical trauma. Depending on the affected gene and the level of skin separation, different forms of epidermolysis bullosa can occur. Severe cases may lead to chronic wounds, scarring, nutritional problems, and an increased risk of skin cancer.

Another important condition is ichthyosis, a group of inherited disorders characterized by abnormal skin scaling and excessive dryness. The skin may become thick, rough, and covered with scale-like

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structures. Some forms are associated with mutations affecting proteins responsible for skin barrier formation and epidermal development.

Albinism is a genetic condition associated with reduced or absent melanin production. It affects the pigmentation of the skin, hair, and eyes. Individuals with albinism have increased sensitivity to ultraviolet radiation and a higher risk of developing skin damage and skin cancer. Regular dermatological examinations and effective sun protection are particularly important.

Inherited forms of palmoplantar keratoderma are characterized by excessive thickening of the skin on the palms and soles. Depending on the genetic subtype, the condition may cause discomfort, painful cracks, and difficulties in walking or performing everyday activities.

Modern Diagnostic Methods The diagnosis of hereditary skin diseases requires a combination of clinical examination, family history assessment, and specialized laboratory investigations.

Modern diagnostic methods include:

1. Clinical and dermatological examination: Assessment of skin lesions, blistering, pigmentation changes, scaling, and other characteristic symptoms.
2. Genetic testing: Identification of mutations responsible for inherited skin disorders through targeted genetic testing or next-generation sequencing.
3. Skin biopsy: Histological examination of skin tissue to identify structural abnormalities.
4. Immunofluorescence mapping: Evaluation of specific proteins and structural components of the skin, particularly in certain forms of epidermolysis bullosa.
5. Molecular diagnostics: Analysis of genetic and molecular mechanisms underlying the disease.

Genetic diagnosis is particularly important because different genetic mutations may produce similar clinical symptoms but require different approaches to management.

Modern Treatment Methods Treatment of hereditary skin diseases depends on the specific diagnosis, severity of symptoms, genetic characteristics, and individual needs of the patient. Although many inherited dermatological conditions do not yet have a complete cure, modern medicine offers several approaches to symptom management and targeted treatment.

Supportive and symptomatic treatment Basic treatment includes moisturizing the skin, reducing friction, preventing infections, treating inflammation, and protecting damaged skin. In patients with epidermolysis bullosa, specialized wound dressings and careful wound management are particularly important.

Topical and systemic therapy Depending on the disease, dermatologists may prescribe emollients, keratolytic agents, topical anti-inflammatory medicines, or other medications. In selected forms of ichthyosis, retinoids may be considered under specialist supervision, taking into account their potential adverse effects.

Biological and targeted therapies Advances in molecular medicine have contributed to the development of treatments that target specific biological pathways. For example, dupilumab has been investigated in patients with Netherton syndrome, a rare inherited skin disorder associated with severe skin barrier dysfunction and inflammation. However, evidence remains limited, and its use for this condition requires individualized specialist assessment.

Gene therapy Gene therapy is one of the most promising developments in the treatment of inherited skin diseases. It aims to introduce functional genetic material into affected cells to compensate for a disease-causing genetic defect. In May 2023, the United States Food and Drug Administration

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approved beremagene geperpavec (Vyjuvek), a topical gene therapy for wounds in patients with dystrophic epidermolysis bullosa associated with mutations in the COL7A1 gene [1].

Regenerative medicine and cell-based approaches Research into stem cells, tissue engineering, and genetically modified skin grafts has created new possibilities for treating severe inherited skin conditions. These approaches aim to restore damaged skin tissue and improve its structural and functional properties.

Conclusion

Hereditary skin diseases are complex genetic disorders that require early diagnosis, individualized treatment, and long-term medical supervision. Modern dermatology increasingly relies on molecular diagnostics, genetic testing, targeted medicines, and gene therapy to improve patient outcomes.

The analysis demonstrates that gene therapy has already become a clinical treatment option for selected wounds associated with dystrophic epidermolysis bullosa. Biological therapies may also provide benefits for certain inherited inflammatory skin disorders, although further clinical research is necessary.

The integration of dermatology, medical genetics, molecular biology, and regenerative medicine is expected to play an important role in the future development of personalized treatment strategies. At the same time, continuous medical monitoring, prevention of complications, and improvement of patients' quality of life remain essential components of care.

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