



Analysis of Various Genetic Skin Disorders Composed of Heterogeneity of Cell Molecules

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DESCRIPTION

Hereditary skin disease program specializes in the diagnosis, treatment and management of people with complex hereditary diseases that primarily affect the skin. Many of the patients will personally examine and diagnosed by a team of pediatric dermatologists and geneticists.

The skin is composed of a variety of cell types that express specific molecules and have distinct properties that facilitate complex interactions and cell-cell communication that are essential for maintaining the structural integrity of the skin. Importantly, a single mutation in any one of these molecules disrupts the overall composition and function of these essential networks, resulting in cell isolation, blistering, as seen in hereditary skin disorders. And can lead to other prominent phenotypes. Over the last few decades, the genetic basis of many monogenic skin disorders has been elucidated using classical genetic techniques. Importantly, the results of these studies shed light on important genetic and molecular interactions with many classes of molecules that give the skin its firm but flexible properties. With the advent of the Human Genome Project, next generation sequencing techniques, and several other recently developed methods, significant advances have been made in the analysis of the genetic structure of complex non-Mendel type skin disorders.

The structural integrity of human skin is protected by a large number of cells. Mutations in any of these cells can completely disrupt the function of these important networks, causing the cell isolation, blistering, and other phenotypes found in skin diseases. Mendel's human skin disorders include pigment disorders, keratosis, bullous disorders, and other hereditary skin disorders that follow Mendel's genetic patterns. Some skin diseases of Mendel's laws are genetically and standard heterogeneous. Therefore, accurate genetic diagnosis of skin diseases according to Mendel's laws is important to provide patients with information on disease progression, prognosis, and management.

Genetic Disorders of the skin contains mostly Albinism, Ectodermal dysplasias, Ehlers-Danlos syndrome (classic type), Ichthyoses, Incontinentia pigmenti, Tuberous sclerosis, Premature aging syndromes.

Albinism: Albinism is a rare hereditary disorder that is not born with normal amounts of melanin pigment. Melanin is a chemical in the body that determines the color of skin, hair and eyes. Most people with albinism have very thin skin, hair, and eyes. They are prone to sunburn and skin cancer.

Ectoderm dysplasia: Ectoderm dysplasia is a group of conditions with abnormal development of skin, hair, nails, teeth, or sweat glands. Different types of ectoderm dysplasia are caused by mutations or deletions of specific genes on different chromosomes. Ectoderm dysplasia is caused by a genetic defect and can be inherited or inherited by the family.

Ehlers-Danlos syndrome: Ehlers-Danlos syndrome is a group of hereditary diseases that primarily affect connective tissues such as the skin, joints, and walls of blood vessels. Connective tissue is a complex mixture of proteins and other substances that gives strength and elasticity to the underlying structure of the body.

Ichthyosis: Ichthyosis is a condition that causes widespread, persistent, thick, dry "fish scale" skin. The skin of people with ichthyosis is rough, dry, and scaly and requires regular moisturization.

Incontinentia pigmenti: Incontinentia pigmenti is characterized by skin abnormalities that develop in childhood and young adulthood. Many affected babies develop a papular rash at birth and early infancy, which heals and continues to grow warty skin.

Tuberous sclerosis: Tuberous sclerosis is a rare hereditary disease, also known as tuberous sclerosis, in which predominantly benign tumors occur in different parts of the body. Tumors most commonly affect the brain, skin, kidneys, heart, eyes, and lungs.

Premature aging syndrome: Aging is an inevitable consequence of human life, with a gradual decline in the function of cells,

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Received: 01-Jul-2022, Manuscript No. HGCR-22-17499; **Editor assigned:** 05-Jul-2022, PreQC No. HGCR-22-17499 (PQ); **Reviewed:** 18-Jul-2022, QC No. HGCR-22-17499; **Revised:** 25-Jul-2022, Manuscript No. HGCR-22-17499 (R); **Published:** 01-Aug-2022, DOI: 10.35248/2161-1041.22.11.220

Citation: Nikos H (2022) Analysis of Various Genetic Skin Disorders Composed of Heterogeneity of Cell Molecules. Hereditary Genet. 11: 220.

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tissues and organisms, increasing the risk of developing chronic diseases. Premature aging syndromes, also known as progeroid syndrome.

The rare skin disorder is morgellons, in which small fibres and particles emerge from a wound on the skin, creating a sensation

of crawling on the skin. Little is known about the condition, but according to the Morgellons Research Foundation, it affects more than 14,000 families.