

Rare Skin Diseases: Treatment and Diagnosis

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ABSTRACT

A skin disease, also known as cutaneous condition, is any medical condition that affects the integumentary system—the organ system that encloses the body and involves skin, hair, nails, and associated muscle and glands. The main feature of this device is as a buffer against the external world. Skin disease, any of the diseases or disorders that affect the human skin. They have a wide range of cause's skin rash caused by Lyme disease rashes and hives, for example, are visible changes in the texture of the skin that may indicate a severe disease.

Keywords: Skin; Blau syndrome; Argyria; Diagnosis

DESCRIPTION

The skin is the largest organ of the human body. There are a number of conditions that can affect the skin. Some of them are common, while others are rare. Many people may have experienced eczema or hives, for instance. However, some skin diseases affect far fewer people. Many of these are inherited conditions. The epidermis is the skin's most superficial layer, consisting of many strata: stratum corneum, stratum lucidum, stratum granulosum, stratum spinosum, and stratum basale. Since the epidermis lacks a direct blood supply, nutrients are delivered to these layers through diffusion from the dermis.

LIST OF RARE SKIN DISEASES

Blau syndrome

Blau Syndrome is an inflammatory condition that affects the skin, eyes, and joints and is caused by an autosomal dominant genetic mutation. A mutation in the NOD2 (CARD15) gene causes it. Early onset cutaneous sarcoidosis, granulomatous arthritis, and uveitis are common symptoms that appear before the age of four. The main cause of blau syndrome is the discovery that the CARD15/NOD2 gene is involved in the BS gene defect has prompted several researchers to investigate how this gene functions as part of the innate immune system, which reacts to bacterial polysaccharides like muramyl dipeptide to activate signalling pathways that cause cytokine responses and

protect the organism. Overexpression appears to be the result of a genetic mutation in BS [1].

Treatment: Treatment has included the usual anti-inflammatory drugs such as adrenal glucocorticoids, anti-metabolites and also biological agents such as anti-TNF and infliximab all with varying degrees of success.

Actinic prurigo

Actinic prurigo is a rare sunlight-induced, pruritic, papular or nodular skin eruption. Some medical experts use the term actinic prurigo to denote a rare photodermatosis that develops in childhood and is chronic and persistent; this rare photodermatosis, associated with the human leukocyte antigen HLA-DR4, is often called "Familial polymorphous light eruption of American Indians" or "Hereditary polymorphous light eruption of American Indians" but some experts consider it to be a variant of the syndrome known as polymorphous light eruption (PMLE). The disorder involves a type IV hypersensitivity reaction driven by both Th1 and Th2 inflammatory pathways, the latter of which leads to secretion of IL-4, IL-5, IL-13, and production of B cells, IgE, and IgG4 [2].

Treatment: Currently there is no cure for actinic prurigo, and treatment focuses on relieving the dermatologic symptoms, by way of topical steroid creams or systemic immunosuppressants. Prescribed treatments include:

- Topical creams such as Tacrolimus and Betamethasone.
- Systemic immunosuppressants such as Prednisone.

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- In some cases, Thalidomide has proven to be effective in controlling the symptoms of actinic prurigo.

Peeling skin syndrome

Peeling Skin Syndrome (PSS) is a group of rare inherited skin disorders in which the normal gradual process of invisible shedding of the outermost skin layers is hastened and/or aggravated. Factors such as heat and friction may worsen the peeling, which can affect only the extremities or the entire body. PSS symptoms include skin shedding or peeling, usually painless, blistering, itching and skin reddening. Peeling skin syndrome is a relatively rare clinical case with pathology of apparently normal skin that needs clinical details to reach accurate diagnoses [3].

Treatment: There is no cure for acral peeling skin syndrome. Treatment is centered on preventing skin damage and addressing symptoms as they occur. Emollients are often used to reduce skin peeling.

Erythropoietic protoporphyria

Erythropoietic protoporphyria is caused by a partial deficiency of ferrochelatase, which is the last enzyme in the heme biosynthesis pathway [4]. Erythropoietic protoporphyria (EPP) is a form of porphyria that can be painful and has a range of severity. It is caused by a lack of the enzyme ferrochelatase, which results in abnormally high levels of protoporphyrin in red blood cells (erythrocytes), plasma, skin, and liver. The severity varies significantly from one person to another person. Some symptoms of this skin condition include skin pain upon exposure to the sun, with prolonged exposure, redness and swelling of the skin.

There is no cure for erythropoietic protoporphyria. People must limit sun exposure to prevent a painful reaction.

WHEN TO APPROACH DOCTORS

Some of these conditions are hereditary, which means that people have them from birth or develop them early on in

childhood. People whose skin is bothering them because of a rash or other ailment should consult a dermatologist. Most rashes are not a serious cause for concern. If any of one is having below symptoms then contact a doctor.

- It affects the entire body
- They also have a fever
- The rash happened suddenly and is spreading
- There is blistering or open sores
- It is painful
- There is an infection

CONCLUSION

Most rare skin conditions have a genetic link, and people inherit them in some way. However, sometimes people may develop a rare skin condition later in life. People who are worried about their skin health should talk with a dermatologist. If a person feels bothered by something on their skin, it is worth speaking with a healthcare professional to get a proper diagnosis.

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