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Darier disease

Darier disease is also known as 'keratosis follicularis'. It is a rare genetic disorder that is manifested predominantly by skin changes. Onset of skin changes is usually in adolescence and the disease is usually chronic. It is inherited in an autosomal dominant pattern, which means that a single gene passed from one parent causes the condition. The chance of a child inheriting the abnormal gene if one parent is affected is 1 in 2 (50%) but not all people with the abnormal gene will develop symptoms of the disease.

Recently the abnormal gene in Darier disease has been identified as *ATP2A2*, found on chromosome 12q23-24.1. This gene codes for the SERCA enzyme or pump (SarcoEndoplasmic Reticulum Calcium-ATPase) that is required to transport calcium within the cell. The exact mechanism by which this abnormal gene causes the disease is still under investigation but it appears that the way in which skin cells join together may be disrupted. The skin cells (keratinocytes) stick together via structures called desmosomes and it seems the desmosomes do not assemble properly if there is insufficient calcium.

How is it diagnosed?

Usually Darier disease is diagnosed by its appearance and the family history, but it is often mistaken for other skin problems.

Diagnosis may require a [skin biopsy](#). The histology is characteristic, known as "focal acantholytic dyskeratosis" associated with varying degrees of papillomatosis (skin thickening). The pathology is similar in [Grover's disease](#) (transient acantholytic dermatosis).

Clinical Features

The symptoms and signs of Darier disease vary markedly between individuals. Some have very subtle signs that are asymptomatic and found only on careful inspection. Others have extensive lesions which can cause considerable distress to the affected individual. In an affected person the severity of the disease can fluctuate over time.

Skin Rash

The skin lesions are characterised by persistent, greasy, scaly papules (small bumps) which tend to occur over the "seborrhoeic" areas of the face (scalp margins, forehead, ears, around the nostrils and sides of nose, eyebrows, and beard area), neck, and central chest and back. The flexures (within natural folds of the skin around joints such as armpits and groins) and skin under breasts and between buttocks are also commonly affected. The papules have a firm, harsh feel like coarse sandpaper and may be skin-coloured, yellow-brown or brown in colour. If several of the small papules grow together they may form larger warty lesions which can become quite smelly within skin folds. The scalp is often affected with a heavily crusted rash which can be similar to [seborrhoeic dermatitis](#) but is usually harsher to the touch.

Atypical presentations of this disease are common. Some patients may have flat, freckle-like lesions. Others may have very large, raised, warty lesions. [Acne conglobata](#) (cystic acne) may occur. A blistered pattern is recognised. Some patients develop a linear pattern of rash, often following the lines of embryonal development of the skin (dermatomal distribution).

Darier's disease



Darier disease on the mid-back



A more severe case



Result of one month's acitretin



Scalp disease



Facial cysts



Darier disease complicated by herpes simplex infection

Lesions on the hands and nails

Small pits (tiny indentations) on the palms and soles may occur and are very characteristic of Darier disease. Small warty lesions or areas of bleeding under the skin can also be seen on the palms and soles as well as the tops of the hands and feet. These are known as keratosis follicularis, and also sometimes called acrokeratosis verruciformis.

Most patients with Darier disease will have longitudinal broad stripes of white and reddish colour on the nails. A V-shaped nick at the free edge of the nail is also very suggestive of Darier disease.

Darier's disease



Palmar pits



Nail disease



Nail disease

Lesions affecting the mucous membranes

Mucous membranes are the red, moist linings of all body surfaces except the skin (eg. mouth, oesophagus, rectum, vulva, vagina). Patients with Darier disease may uncommonly have a white cobblestone pattern or small papules affecting the mucous membranes. Overgrowth of the gums is also seen.

Prognosis and Complications

Most patients will develop signs of the disease (even if subtle) before the age of 30 years. Many patients will have

a mild form of the disease that, although present, will go unnoticed throughout life. Patients with more severe disease will notice a chronic relapsing–remitting pattern to their signs and symptoms although cases of spontaneous resolution of signs have been reported. The rash is often exacerbated by sunlight (a reaction which may be delayed eg. on return from holiday), and occasionally by corticosteroid use (although this may be useful for other patients).

Bacterial infection can cause flares. Widespread infection of the skin the skin with the [herpes simplex](#) (coldsore) virus is a well recognised complication. This can cause a severe flare and patients can feel quite unwell.

In most patients general health remains good regardless of the severity of the disease.

Treatment

Treatment is required only if there are troublesome symptoms. For patients with mild disease simple [moisturisers](#), [sun protection](#) and selection of the right clothing to avoid heat and sweating are usually sufficient.

Dermabrasion (sanding off the surface of the skin) may be effective in localised disease. Localised Darier disease may also be treated successfully with [topical retinoids](#).

Secondary bacterial infection (usually due to [Staphylococcus aureus](#)) should be treated with antibiotics, and herpes simplex with antiviral agents.

If symptoms are particularly severe then a trial of an oral retinoid medication such as [acitretin](#) or [isotretinoin](#) may be effective. [Ciclosporin](#) has been reported to be effective in a few patients.

Related information

On DermNet NZ:

Other websites:

- [Darier Disease Resource Site](#)
- [Keratosis follicularis](#) (Darier disease): from emedicine dermatology, the online textbook

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DermNet does not provide an on–line consultation service.

If you have any concerns with your skin or its treatment, see a [dermatologist](#) for advice.

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