



DARIER DISEASE

What are the aims of this leaflet?

This leaflet has been written to help you understand more about Darier disease. It tells you what it is, what causes it and what can be done about it including treatment options. It provides other sources where you can find out more information about it.

What is Darier disease?

It is a rare inherited skin condition, estimated to affect 1 to 4 people per 100,000 of the population and is characterised by a change in the way skin cells (keratinocytes) stick together within the upper layer of the skin (epidermis). This leads to changes in the skin and nails, and sometimes inside the mouth can be affected. Other names for Darier disease include Darier-White disease and Keratosis Follicularis.

What causes Darier disease?

The movement of calcium within cells is disrupted, leading to a change in the way skin cells are held together. Normally, these cells are held together like bricks cemented in a wall. In Darier disease, the 'cement' that holds the skin cells together is weakened, so the cells separate easily and do not form a good barrier against the outside world. This causes the skin to become easily irritated, inflamed and it may begin to weep. It is not due to an allergy and it is not contagious (catching). Darier disease is often aggravated by heat, sunlight, skin friction, excessive sweating, and it can make the skin more prone to infection. Some females may notice their skin flares before their period. Certain prescribed medications (usually taken by mouth) may also make the rash worse.

Is it hereditary?

Yes. It is inherited in a pattern known as dominant inheritance. This means that there is a 1 in 2 (50:50) chance that each child of an affected parent will inherit the condition. It affects both men and women equally and often begins to show in

childhood. Sometimes, there may be no history of any other family member being affected. This may be because the disease is so mild in the parents that it has gone unnoticed, or because the genetic abnormality has developed in someone for the first time. Some people may have inherited the genetic abnormality but do not develop any skin problems. The severity and extent of the condition may vary considerably within a family. If one person is severely affected it does not necessarily mean that other family members will be severely affected.

What are the symptoms of Darier disease?

For some people with Darier disease, the skin condition is mild and does not require medical attention. In a minority of patients, it can be more severe and medical help may be required. Affected areas may be itchy and sore and the skin may have an unpleasant odour, particularly in more moist areas such as the underarm and groin. This is caused by skin bacteria.

People with Darier disease have an increased chance of developing bacterial, fungal and viral skin infections. These infections can cause the skin condition to flare up. It is important to know that affected individuals are more at risk of getting widespread "cold sores" (herpes simplex virus). If the skin condition suddenly gets worse and is much more painful than usual this may indicate herpes simplex virus infection, even if there are no blisters or vesicles (small fluid filled bubbles in the skin) as usually seen in 'cold sores' affecting the lip. Seek urgent medical advice if this happens.

What does it look like?

The rash usually appears in childhood / teenage years, but in some people, it is not present until adulthood. It mainly affects areas of the skin where there is the most sweat production (sebaceous areas) which typically include the face, scalp, chest, neck, and upper back. The appearance of the rash varies from small scattered, slightly greasy or waxy brownish, yellowish or sometimes red skin lumps (papules), to larger thickened patches which may be crusty or scaly. In the skin folds, particularly in the groin area and underneath the breasts, the skin may be more prone to becoming raw and starting to weep. The fingernails are usually affected with red or white lines running the length of the nails and small notches at the ends of the nail. Nail changes and/or flat "warts" on the backs of the hands can often be seen in children with Darier disease several years before there are any other skin changes. Pits or small areas of hard skin occur on the palms of the hands and less often the soles of the feet. Occasionally there may be small spots inside the mouth and these may give the roof of the mouth a rough feeling.

How will it be diagnosed?

The diagnosis can often be made on the appearance of the rash and the fact that it runs in families. To confirm the diagnosis, your dermatologist may take a small sample of skin (called a biopsy) that will be examined under a microscope in the laboratory.

Can Darier disease be cured?

No, there is no cure, but there are many ways of managing the affected skin. A quarter of patients notice that the condition may improve over time. There are treatments available to try and manage the condition, but it can recur and worsen again after treatment.

How can Darier disease be treated?

If there are no symptoms, treatment is not required. Simple measures such as wearing cotton clothing, minimising sweating and using sun protection may reduce symptoms.

Topical treatments:

Moisturising creams may relieve some of the itching and irritation and may help reduce scaling of the skin. Sometimes corticosteroid creams are prescribed and can be helpful if the skin is very itchy. Antibiotic creams may be used if the skin becomes infected. Antiseptic solutions for washing and antiseptic creams may be helpful, particularly if there is a problem with odour. Creams and washes containing antiseptics such as chlorhexidine may help reduce the tendency to get secondary infection.

For small areas of affected skin, retinoid creams may be of benefit. They can, however, cause irritation which may limit their use and they must not be used during pregnancy. Topical 5-fluorouracil, which is licensed for the treatment of other skin conditions (e.g. Actinic Keratosis), has been used effectively in some cases of Darier disease. Other topical agents that have helped in some cases of Darier disease include calcineurin inhibitors (pimecrolimus cream and tacrolimus ointment).

Oral Treatments:

If bacterial infection is severe, oral antibiotics may be required and widespread cold sore infections (herpes simplex) require treatment with oral anti-viral treatment, usually acyclovir.

For more severe disease, treatment with the oral Retinoids [acitretin](#) or [isotretinoin](#) may be tried. These are usually given long term to keep the skin under control. They must not be used during pregnancy. Ciclosporin is sometimes used 'off-licence' to help control Darier disease (a license for a drug says how it should be used; 'off-

license' means that it is used in a way that is not described in this license). Doxycycline, an antibiotic sometimes used for skin infections and acne has demonstrated positive effects in Darier disease but this is also an 'off-licence' use.

Other useful treatments:

Laser treatment or resurfacing of very thick areas has been reported to be successful and may be offered by some dermatology departments. Surgical excision or Dermabrasion (removing surface layers) of very thick areas has been used occasionally.

Photodynamic therapy, a type of photochemotherapy delivered in dermatology departments, that is used to treat other skin conditions (e.g. Actinic Keratosis) has been used effectively in some cases of Darier disease.

Botulinum toxin injections has been reported in a small number of patients and may be helpful for severe Darier disease. This can be helpful in reducing sweating and can be discussed with your dermatologist.

What can I do?

Most people with Darier disease lead a normal life and have no other medical problems. Simple measures recommended to try and keep the skin condition controlled have been mentioned above. It is important to monitor and inspect your skin regularly and if you notice any changes or have any concerns please report these to your GP or Dermatologist.

Where can I get more information about Darier disease?

<https://www.dermnetnz.org/topics/darier-disease>

This leaflet aims to provide accurate information about the subject and is a consensus of the views held by representatives of the British Association of Dermatologists: individual patient circumstances may differ, which might alter both the advice and course of therapy given to you by your doctor.

This leaflet has been assessed for readability by the British Association of Dermatologists' Patient Information Lay Review Panel

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