

DARIER'S DISEASE

What are the aims of this leaflet?

This leaflet has been written to help you understand more about Darier's disease. It tells you what it is, what causes it, what can be done about it, and where you can find out more about it.

What is Darier's disease?

It is a rare inherited skin condition characterised by the loss of binding (acantholysis) between skin surface cells and there is also some thickening of the skin. It is also known as Keratosis Follicularis. The nails and mouth may also be affected.

What causes Darier's disease?

It is the result of an abnormality in the gene involved in calcium transport within cells. The defective calcium transport leads to reduced cell binding. Normally, in the outer layer (epidermis) of the skin, the skin cells are held together like bricks cemented in a wall. In Darier's disease the sticky junctions that hold the skin cells together are not made properly, and the skin may be easily irritated and becomes inflamed and weeping. It is not due to an allergy and it is not contagious (catching). It can be made worse by sunlight, skin friction, excessive sweating, and secondary infection. Some women are aware of their skin flaring before their period.

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 in childhood. Occasionally, there may be no family history of the condition. This may be because the disease is so mild in parents, it has gone unnoticed, or sometimes the abnormality may develop for the first time as a new genetic defect. Some people with the genetic abnormality do not develop the skin rash. The severity and extent of the disease may vary considerably within a single family. If a person is badly affected it does not necessarily mean that other family members who inherit the condition will also get severe disease.

What are the symptoms of Darier's disease?

Some patients have very mild disease which does not bother them at all. But for others it can be quite troublesome. The main symptoms are of itching and soreness. The affected skin may smell unpleasant, particularly in moist areas. This is probably

caused by increased numbers of ordinary skin bacteria growing in the inflamed affected skin.

There is an increased chance of developing both bacterial and viral skin infections if you have the condition and secondary infection can cause the skin to flare. It is important to know that affected individuals are more at risk of getting widespread "cold sore" virus (herpes simplex) infection of their skin. If the skin suddenly gets worse and is much more painful than usual this may be a sign of a herpes simplex infection. Contact your GP urgently if this happens.

What does it look like?

The skin rash usually appears in childhood / teenage years, but onset may be delayed until adulthood. The rash tends to involve the areas of the skin where there is the most grease 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 brownish, yellowish or red papules (small lumps) to larger thickened patches which may be scaling. The skin folds particularly in the groin area and underneath the breast are also often affected and in these areas the skin can become raw and weepy. The fingernails are usually affected. There may be red or white lines running the length of the nails with some notching at the ends of the nail. Nail changes and/or flat "warts" on the backs of the hands are often present in childhood, well 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 (a biopsy) under a local anaesthetic and it is examined under a microscope in the laboratory.

Can Darier's disease be cured?

No, there is no cure, but there are many ways of helping it (see below). A quarter of patients notice that the condition improves as they get older. Some people find that the sun causes their Darier's disease to flare up. Some women notice that it worsens around the time of their periods.

How can Darier's 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 flares.

Topical treatments:

Moisturising creams may relieve some of the itching and irritation. Sometimes corticosteroid creams are helpful if the skin is very itchy. Antibiotic creams can be used if the skin becomes infected. Careful washing is important and in addition,

antiseptic solutions for the bath, and antiseptic creams may help, particularly if there is a problem with odour.

For localised disease, topical retinoid creams may be helpful. However, irritation is a limiting factor, so emollients and topical corticosteroids can be used in combination with these to reduce irritation.

Topical 5-fluorouracil, which is used to treat other skin conditions (e.g. Actinic Keratosis), has been used effectively in some cases of Darier's disease.

Oral Treatments:

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

For more severe disease treatment with Retinoid tablets such as Acitretin or Isotretinoin may be tried. Ciclosporin is sometimes used 'off-licence' to help control Darier's disease. You can discuss these with your dermatologist.

Other useful treatments:

Laser treatment of very thick areas has been reported to be successful.

Photodynamic therapy that is used to treat other skin conditions (e.g. Actinic Keratosis) has been used effectively in some cases of Darier's disease.

Surgical excision or Dermabrasion (removing surface layers) of very thick areas has been used occasionally.

What can I do?

Most people with Darier's disease lead full, normal lives and have no other medical problems. Less than a quarter need any time off work or school because the skin is a problem. However, it is sensible to try to avoid excessive heat, sweat, humidity, sunlight, and mechanical trauma as they can make the skin worse.

Where can I get more information about Darier's disease?

<http://www.dermnetnz.org/dna.darier/darier.html>

<http://www.patient.co.uk/directory/dariers-disease>