Darier Disease

Other Names

Darier white disease; Darier's disease; Keratosis follicularis

Categories

Congenital and Genetic Diseases; Skin Diseases.

Signs and Symptoms

Darier disease is a skin condition characterised by wart like blemishes on the body. These are usually coloured (yellow-brown or brown), hard to the touch, mildly greasy and can emit a strong odour.

Common sites for blemishes are the scalp, forehead, upper arms, chest, back, knees, elbows, and behind the ear. Pits (depressions in the skin) may be present in the palms of the hands and soles of the feet. The mucous membranes may also be affected including the mouth (palate), inside the cheeks, gums, and throat, the vulva and rectum.

Red and white streaks in the nails, with irregular texture, can occur.

Darier disease symptoms usually appear in late childhood to early adulthood. A person with the condition may have 'flare-ups' where more symptoms are present and periods with fewer blemishes.

Other characteristics *may* include:

- Mild intellectual disability
- Epilepsy
- Depression
- Learning and behavioural difficulties.

Another form of Darier disease is the linear or segmental form which has blemishes on localised areas of the skin; these blemishes are not as widespread as typical Darier disease and may only occur on one side of the body.

Cause/Inheritance

- Darier disease is an inherited autosomal dominant condition which means that one copy of the altered gene in each cell is enough to cause the condition.
- Darier disease may be inherited from one affected parent.
- Diagnosis may require a skin biopsy.
- In some cases, the condition may be a new mutation in the gene and there is no history in the family.
- Genetic testing may identify a mutation in the ATP2A2 which can be used to confirm a diagnosis

Treatment and Management

Treatment and management of Darier disease is based on symptoms and may include avoiding the sun and heat or using sunscreen and wearing cool cotton clothing. Other measures may include (depending on impact of side effects):

- Use of moisturisers with urea or lactic acid to reduce scaling
- Topical steroids
- Topical medications
- Oral retinoids
- Oral antibiotics
- Surgical or laser removal may be appropriate for smaller, severely affected areas.

Support Contacts International

FIRST Foundation of Ichthyosis and Related Skin Types http://www.firstskinfoundation.org/

Facebook Support

Darier's Disease Support Group https://www.facebook.com/groups/21860644983/

Sources

Genetics Home Reference https://ghr.nlm.nih.gov/condition/darier-disease
National Organisation for Rare Disorders https://rarediseases.org/gard-rare-disease/6243/darier-disease/
Genetic and Rare Diseases Information Center https://rarediseases.info.nih.gov/diseases/6243/darier-disease
Orphanet https://www.orpha.net/consor/cgi-bin/Disease_Search.php?lng=EN&data_id=323&Disease_Disease_Search_diseaseGroup=darier&Disease_Disease_Search_diseaseType=Pat&Disease(s)/group%20of%20diseases=Darier-disease&title=Darier%20disease&search=Disease_Search_Simple
The Australasian College of Dermatologists https://www.dermcoll.edu.au/atoz/dariers-disease/

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