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Darier Disease: A very rare Genodermatosis

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Abstract

Darier Disease: A very rare Genodermatosis**.** Darier disease (DD) or Darier-white disease, also known as Keratosis follicularis, is an autosomal dominant inherited genodermatosis caused by mutations of ATP2A2 gene. It is characterized by greasy hyperkeratotic papules in seborrheic regions, nail abnormalities and sometimes mucous membrane changes. Oral manifestations primarily affect the palatal and alveolar mucosa. A 24 year old girl presented with greyish coloured warty plaques scattered on the face, neck, ears, trunk, hands, legs, and groins. The lesions were foul smelling. Characteristic V-shaped nicking of the nails with longitudinal ridges parallel to the long axis were seen. Palmer pits were also present. On examining oral mucosa, multiple white papules over the palate giving cobblestone appearance was seen. Patient was also complaining of Photophobia, redness, lacrimation and pain. On slit lamp examination, ulcer of 2x3 mm with Fluorescent stain positive and few anterior chamber cells present. No family history was there of similar illness. Biopsy was taken from hyperkeratotic papules of hand, which shows hyperkeratosis along with central keratin plug, the epithelial rete ridges associated with the lesions appear narrow and elongated. DD is a rare keratinization disorder with skin involvement and oral mucosal lesions. We wanted to present this case because of its rarity. On the basis of Clinical and histopathological finding patient was diagnosed as a case of Darier disease. General measures were advised and genetic counselling was done, including information of inherited condition and risk of transmission to offspring.

Biography:

Harshita Jain has completed her MBBS at the age of 23 years from Peoples University and Ongoing Postdoctoral studies from Teerthankar Mahaveer University in batch of 2018, and complete Dermatology degree by 2021.

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