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Nail and teeth abnormalities-marginal palmoplantar keratoderma-oral hyperpigmentation syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Nail and teeth abnormalities-marginal palmoplantar keratoderma-oral hyperpigmentation syndrome. ORPHA:423454*

Nail and teeth abnormalities-marginal palmoplantar keratoderma-oral hyperpigmentation syndrome is a rare genetic ectodermal dysplasia syndrome characterized by short stature, nail dystrophy and/or nail loss, oral mucosa and/or tongue hyperpigmentation, dentition abnormalities (delayed teeth eruption, hypodontia, enamel hypoplasia), keratoderma on the margins of the palms and soles and focal hyperkeratosis on the dorsum of the hands and feet. Additionally, dysphagia with esophageal strictures, sensorineural deafness, bronchial asthma and severe iron-deficiency anemia have been observed.