

Opis choroby *

Definicja

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.

Dane

Klasyfikacja

Choroba

Synonimy

Ectodermal dysplasia-short stature syndrome

Zespół dysplazji ektodermalnek i niskiego wzrostu

Zespół niski wzrost, dysplazja paznokci, rogowiec dłoniowo-podeszwowy brzeżny i hiperpigmentacja jamy ustnej

Short stature-nail dysplasia-marginal palmoplantar keratoderma-oral hyperpigmentation syndrome

Kod ORPHA

423454

Kod OMIM

616029

Kod ICD10

Q82.8

Kod ICD11

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*Źródło

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