

Opis choroby *

Definicja

A rare, systemic disease characterized by normolipidemic mucocutaneous xanthomatosis with histiocytic cells proliferation and secondary deposition of lipid in the dermis. Clinically, multiple, grouped, coalescent, yellowish red to brown papulonodular lesions in the skin and mucous membranes are present. Less often internal organs are affected, in particular pituitary gland and/or hypothalamus. Patients present with characteristic mucocutaneous lesions, diabetes insipidus, dysphagia, dyspnea, hoarseness of voice, and blurred vision.

Dane

Klasyfikacja

Choroba

Synonimy

Montgomery syndrome

Zespół Montgomery'ego

Kod ORPHA

158003

Kod OMIM

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Kod ICD10

D76.3

Kod ICD11

EE81

*Źródło

orphanet