

Opis choroby *

Definicja

Pseudoleprechaunism syndrome, Patterson type is a rare, genetic, adrenal disorder characterized by congenital bronzed hyperpigmentation, cutis laxa of the hands and feet, body disproportion (comprising large hands, feet, nose and ears), hirsutism and severe intellectual disability. Patients additionally present hyperadrenocorticism, cushingoid features, premature adrenarche and diabetes mellitus, as well as skeletal deformities (not present at birth and which progress with age). There have been no further descriptions in the literature since 1981.

Dane

Klasyfikacja

Zespół wad wrodzonych
Patterson pseudoleprechaunism syndrome
Patterson syndrome

Synonimy

Kod ORPHA

2976

Kod OMIM

169170

Kod ICD10

E34.8

Kod ICD11

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*Źródło

orphanet