Zespół pseudoleprechaunizmu typu Pattersona Kod Orpha: 2976 Kod OMIM: 169170

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

Synonimy
Patterson pseudoleprechaunism syndrome
Patterson syndrome

Kod ORPHA
2976

Kod OMIM
169170

Kod ICD10
E34.8

Kod ICD11
-

*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.