Pseudoleprechaunism syndrome, Patterson type

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Pseudoleprechaunism syndrome, Patterson type. ORPHA:2976

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.