

Opis choroby *

Definicja

A rare PIK3CA-related overgrowth syndrome disease characterized by segmental and progressive overgrowth, predominantly involving the adipose tissue, or a mixture of adipose and fibrous tissue, with variable involvement of subcutaneous and muscular tissue, as well as skeletal overgrowth. Overgrowth severity and range is highly variable, although frequently it is asymmetric and disproportionate, it affects lower extremities more than the upper ones, and progresses in a distal to proximal pattern. Congenital overgrowth is typically associated.

Dane

Klasyfikacja

Choroba

Kod ORPHA

314662

Kod OMIM

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Kod ICD10

Q87.3

Kod ICD11

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*Źródło

orphanet