

Opis choroby *

Definicja

Growth hormone insensitivity syndrome (GHIS) is a group of diseases characterized by marked short stature associated with normal or elevated growth hormone (GH) concentrations, which fail to respond to exogenous GH administration. GHIS comprises growth delay due to IGF-1 deficiency, growth delay due to IGF-1 resistance, Laron syndrome, short stature due to STAT5b deficiency and primary acid-labile subunit (ALS) deficiency (see these terms).

Dane

Klasyfikacja

Kategoria

Synonimy

GHIS

GHIS

Niski wzrost z powodu defektu receptora dla hormonu wzrostu lub szlaku poreceptorowego

Short stature due to a defect in growth hormone receptor or post-receptor pathway

Kod ORPHA

181393

Kod OMIM

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Kod ICD10

E34.3

Kod ICD11

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*Źródło

orphanet