

Opis choroby *

Definicja

Short stature-delayed bone age due to thyroid hormone metabolism deficiency is a rare, genetic congenital hypothyroidism disorder characterized by mild global developmental delay in childhood, short stature, delayed bone age, and abnormal thyroid and selenium levels in serum (high total and free T4 concentrations, low T3, high reverse T3, normal to high TSH, decreased selenium). Intellectual disability, primary infertility, hypotonia, muscle weakness, and impaired hearing have also been reported.

Dane

Klasyfikacja

Choroba

Kod ORPHA

171706

Kod OMIM

609698

Kod ICD10

E03.1

Kod ICD11

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*Źródło

orphanet