Opis choroby *

Definicja

A rare genetic hyperthyroidism characterized by elevated levels of circulating free thyroid hormones, normal or elevated thyroid-stimulating hormone, decreased peripheral tissue responses to iodothyronine action, and a highly variable clinical phenotype which most commonly includes goiter, resting tachycardia, osteoporosis, short stature, and attention deficit disorder. Some patients may be entirely asymptomatic.

Dane

Klasyfikacja
Choroba

Synonimy
RTHb
Oporność na hormon tarczycy beta
Oporność na hormon tarczycy z powodu mutacji w TRb
Resistance to thyroid hormone beta
Resistance to thyroid hormone due to a mutation in TRb

Kod ORPHA
566243

Kod OMIM
274300

Kod ICD10
E07.8

Kod ICD11
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*Źródło

orphanet