

## 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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