Opis choroby *

Definicja

A rare primary congenital hypothyroidism characterized by a markedly reduced T4/T3 ratio, normal levels of thyroid-stimulating hormone, and a highly variable clinical phenotype, which most commonly includes decreased metabolic rate, bradycardia, chronic constipation, neurodevelopmental delay, and delayed bone age and skeletal abnormalities. Dysmorphic craniofacial features, such as macrocephaly, broad face, flat nose, large tongue, and thick lips, have also been reported. Some patients may show only minimal signs and symptoms.

Dane

Klasyfikacja
Choroba

Synonimy
RTHa
Oporność na hormon tarczycy alpha
Oporność na hormon tarczycy z powodu mutacji w TRa
Resistance to thyroid hormone alpha
Resistance to thyroid hormone due to a mutation in TRa

Kod ORPHA
566231

Kod OMIM
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Kod ICD10
E07.8

Kod ICD11
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*Źródło

orphanet