DISEASE:
Resistance to thyroid hormone due to a mutation in thyroid hormone receptor alpha

NAME:	Resistance to thyroid hormone due to a mutation in thyroid hormone receptor alpha
DESCRIPTION:	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.
ORPHACODE:	566231
SYNONYMS:	RTHa Resistance to thyroid hormone alpha Resistance to thyroid hormone due to a mutation in TRa
XREF(S):	Orphanet
ANALYTE(S):	THRA
CREATED:	04 Feb 2020 - 15:13
CHANGED:	22 Jun 2023 - 16:14

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