DISEASE:
Hyperinsulinism due to INSR deficiency

NAME:	Hyperinsulinism due to INSR deficiency
DESCRIPTION:	A rare autosomal dominant form of familial hyperinsulinism characterized clinically by postprandial hypoglycemia, fasting hyperinsulinemia, and an elevated serum insulin-to-C peptide ratio, and a variable age of onset.
ORPHACODE:	263458
SYNONYMS:	Hyperinsulinemic hypoglycemia due to INSR deficiency Hyperinsulinemic hypoglycemia due to insulin receptor deficiency
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	INSR
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Diabetes neonatal / Maturity onset Diabete of the Young (MODY) / Hyperinsulinism (gene panel)
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Related Laboratories

- Centrum Medische Genetica - UZ Antwerpen

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- insulin receptor

Related Gene Panels

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- Hyperinsulinism (5 genes) - UZA
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