

DISEASE:
Congenital hyperinsulinism due to HNF4A deficiency

NAME:	Congenital hyperinsulinism due to HNF4A deficiency
DESCRIPTION:	A form of diazoxide-sensitive diffuse congenital hyperinsulinism due to HNF4A deficiency and, characterized by macrosomia, transient or persistent hyperinsulinemic hypoglycemia (HH), responsiveness to diazoxide and a propensity to develop maturity-onset diabetes of the young subtype 1 (MODY).
ORPHACODE:	263455
SYNONYMS:	Hyperinsulinemic hypoglycemia due to HNF4A deficiency
XREF(S):	<u>Orphanet</u>
ANALYTE(S):	<u>HNF4A</u>
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: <http://gentest.healthdata.be/disease/1103>

RELATED CONTENT

Related Genetic Tests

- Diabetes neonatal / Maturity onset Diabete of the Young (MODY) / Hyperinsulinism (gene panel)
- Hyperinsulinism (gene panel)
- Maturity onset Diabete of the Young (MODY), type 5 / Renal cysts and diabetes syndrome (gene panel)

Related Laboratories

- Centrum Medische Genetica - UZ Antwerpen

Related Analytes

- hepatocyte nuclear factor 4 alpha

Related Gene Panels

- Diabetes neonatal / Maturity onset Diabete of the Young (MODY) / Hyperinsulinism (genepanel) - UZA
- Hyperinsulinism (5 genes) - UZA
- MODY (7 genes) - UZA

Source URL: <http://gentest.healthdata.be/disease/1103>