

Diagnosis workup of an IEM-linked hypoglycaemia in adults

Glycaemia < 0.55 g/L or 3mmol/L (venous sample))

Usual causes ?

- Drugs**
Malicious hypoglycaemia
Antibody to insulin
Antibody to insulin receptor
Alcohol
Cortisol (GH) deficiency
Cachexia, Sepsis
Hepatic, renal or cardiac failure
IGF-2 or big-IGF-2 by mesenchymal tumors

Timeline ?

Fasting

Postprandial

Exercise-induced

no suspicion IEM

suspicion IEM

Investigations (4)

Reactive Hypoglycaemia ? (6)

Suspicion Inherited Fructose Intolerance ?

Fasting-test (2)

hypoglycaemia

insulinoma (3)

no insulinoma

normal (5)

GSD suspicion ?
oral glucose tolerance test with lactates levels

Metabolic Fasting-test (up to 48 or 72h if no abnormalities)

yes

no

Molecular diagnosis

Mixed-meal test

Exercise test

Muscle symptom
CPK (high)
Ketone -
Free Fatty Acid (FFA) +++
(FFA/βOH butyrate > 2.5)

Alanine +++
Glycerol +++
Lactates ++
Ketones +

Hepatomegaly
Lactates ++
Ketones +

ENDOGENOUS HYPERINSULINISM (3)

FAO DISORDER

GLUCONEOGENESIS DISORDER

GLYCOGENOLYSIS DISORDER

INHERITED FRUCTOSE INTOLERANCE

- NIPHS
Glucokinase activating mutation ? (SUR1/Kir6.2, SCHAD, GDH in adult ?)
- CDG Id (Ia, Ib in adult ?)

MCT1

GSD: glycogen storage disease; FAO: defects of fatty acid oxidation; NIPHS: non-insulinoma pancreatogenic hypoglycaemia syndrome; MCT 1: monocarboxylate transporter 1.