

# File:Screen Shot 2022-02-22 at 9.46.32 AM.png

Isovaleric aciduria

Leucin

transaminase, decarboxylase

Isovaleryl-CoA + glycine

Isovalerylglycine

glycinová acyltransferasa

Isovaleryl-CoA dehydrogenasa

β-methylcrotonyl-CoA

Scheme of metabolic disorder in isovaleric aciduria

Clinical picture

Metabolic ketoacidosis, dehydration, hyperammonaemia, ketonuria, hypocalcaemia, hepatomegaly, vomiting, hyper / hypoglycaemia and failure

Cause

Isovaleryl-CoA dehydrogenase deficiency

Diagnostics

Analysis of isovalerylglycine in amniotic fluid or enzymatic analysis in chorionic villi

Prognosis

The long-term projection of IVA is good. Death in an acidotic episode can occur at any age.

Classifications and references

OMIM 243500


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