

Talk:Propionic acidemia

Propionic acidemia is one of the hereditary metabolic disorder (DMT), specifically it is classified as **organic aciduria** . it is Autosomal Recessive.

There is a defect in propionyl-CoA carboxylase (either A or B subunits). Diagnosed in at least four patients in the Czech Republic.

Laboratory findings

At the time of an acute attack, there is ketoacidosis with Ketonuria, hyperammonemia and pancytopenia.

Therapy

During an acute attack, it is necessary to use elimination methods and glucose administration. In the long term, it is necessary to follow a low-protein diet with the supply of AMK (up to valine) and also the administration of ATB (metronidazole) to prevent intestinal colonization. In more severe forms, liver transplantation is indicated.
Template:Doplňte zdroj

Links

related articles

- Dědičné metabolické poruchy
- Organické acidurie
- Ketoacidóza

External links

- http://www.sekk.cz/ELM_ukoncen.pdfencyklopedie/A/LUAEA.htm
- Kyselina propionová (česká wikipedia) (https://cs.wikipedia.org/wiki/Kyselina_propionov%C3%A1%7C)

Template:Navbox - dědičné metabolické poruchy

Kategorie:Patobiochemie Kategorie:Pediatric Kategorie:Genetics