Vomiting, diarrhea and aversion for meat ?

Lysinuric protein intolerance : a case report

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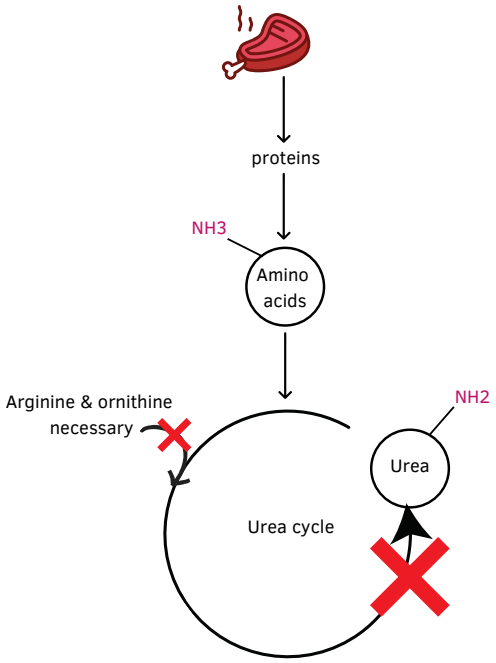
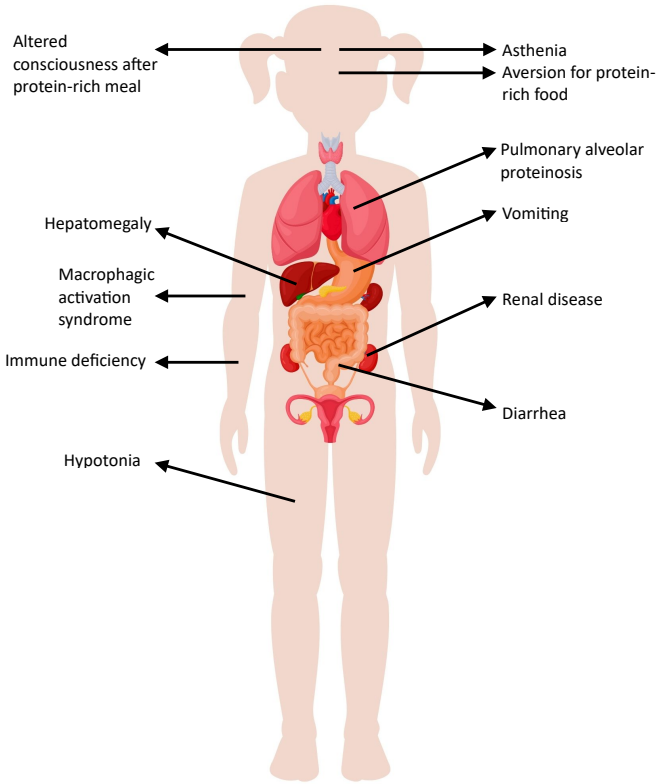
Introduction

- Lysinuric protein intolerance (LPI) is a rare autosomal recessive metabolic disease that affects the SCL7A7 gene coding for the light subunit of an amino acid (AA) transporter (y^+LAT1 protein), present mainly in the **intestinal** and **renal** tubular cells and on **macrophages**
- **Symptoms** include recurrent vomiting and diarrhea, episodes of coma after protein-rich meal, aversion for protein-rich food, failure to thrive, muscular hypotonia
- **Complications** include hyperammonemia with coma, severe lung disease with pulmonary alveolar proteinosis, renal disease, macrophagic activation syndrome (MAS), immune deficiency and osteopenia

Physiopathology

- **Quick reminder** : ingestion of proteins → catabolism of proteins into AA (carrying a toxic NH3 ammonia molecule) → transformation of NH3 into urea (less toxic) → renal elimination
- **In LPI** : ↓ renal and intestinal reabsorption of lysine, arginine and ornithine → ↓ plasmatic concentration of these AA → ↓ substrates for the urea cycle → ↓ urea production = ↓ ammonium excretion → ↑ plasmatic concentration of ammonium





Case report

- 6-year-old girl, arriving from Macedonia
- **Clinical picture :**
  - Recurrent episodes of **vomiting** and **diarrhea** since introduction of formula (2 months old)
  - Episodes of **altered consciousness** after protein-rich meal
  - **Aversion** for meat
  - Fatigue (especially during sport)
  - Multiples infections
  - Important **failure to thrive**
- **Clinical features :**
  - Height <P3, weight <P3, head circumference <P3
  - Flattened face, short philtrum, low set ears
  - Hepatomegaly
- **Laboratory findings :**
  - **Plasma** : anemia (104 g/L), low lysine (35 umol/L) and other AA, hypoproteinemia (pre-albumine 103 mg/L), signs of MAS (ferritine 2'013 ug/L, LDH 980 U/L), normal ammonia (26 umol/L)
  - **Urine** : High lysine (344 umol/mol) and arginine (19 mmol/mol), normal ornithine (6 umol/mol)
- **Treatments :**
  - **Prevent hyperammonemia** by improving the urea cycle **:** low protein diet (0.8-1.5g/kg/d), citrulline +/- sodium benzoate

Conclusion

- LPI is a rare metabolic disease
- Range of symptoms from nausea to coma and death
- Treatment quickly improves symptoms
- Follow-up necessary to prevent pulmonary and renal complications

References

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