Background

- Intoxication-Type Inborn Errors of Metabolism (IEM) are diseases that affect protein catabolism.
- Treatment requires strict protein restrictive diet.

Aims of the study

To evaluate growth, pubertal status, and protein intake.

Methods

- Longitudinal follow up cohort study
- 213 patients who required a hypoprotidic normocaloric diet since neonatal period or early infancy
- Height was recorded:
  - before 4 years (early infancy, n=189);
  - between 4 and 8 years for girls and 9 years for boys (prepuberty, n=168);
  - after 8/9 years (puberty, n=136).

Results

- Urea Cycle Disease: N=22
- Organic Aciduria: N=29
- MSUD: N=13
- Tyrosinemia: N=5

Conclusions

- In intoxication-type IEM with hypoprotidic diet, may be associated with impaired final height.
- Growth retardation is more frequent in the pubertal subgroup.
- In the pubertal subgroup, growth retardation is more frequent when patient receive amino acid mixture free of pathological precursor.