

HEIGHT IN INBORN ERRORS OF METABOLISM REQUIRING HYPOPROTIDIC DIET: A LONGITUDINAL FOLLOW UP STUDY ABOUT 213 PATIENTS

K. Busiah^{1,2}, C. Roda³, A. Brassier², C. Pontoizeau⁴, C. Ottolenghi⁴, M. Piketty⁵, A-S. Crosnier⁶, L. Perin⁶, Y. Le Bouc⁶, J-B Arnoux², I. Netchine⁶, P. de Lonlay².

¹ Pediatric endocrinology, diabetology and obesity unit; DFME, CHUV, Lausanne, Suisse

² Reference Center of Inherited Metabolic Diseases, Hospital Necker Enfants Malades, APHP, Imagine institute, Filière G2M, metabERN, INEM, University Paris Descartes, Paris, France

³ Université Paris Descartes, Sorbonne Paris Cité, EA 4064, Epidémiologie environnementale, Paris, France

⁴ Metabolic biochemistry, Hospital Necker Enfants Malades, APHP, Imagine institute, Filière G2M, metabERN, University Paris Descartes, Paris, France

⁵ Service des Explorations Fonctionnelles, Hospital Necker Enfants Malades, APHP, Paris, France

⁶ Service des Explorations Fonctionnelles, Hospital Trousseau, APHP, Paris, France

Background

Intoxication-Type Inborn Errors of Metabolism (IEM) are diseases that affect proteins 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

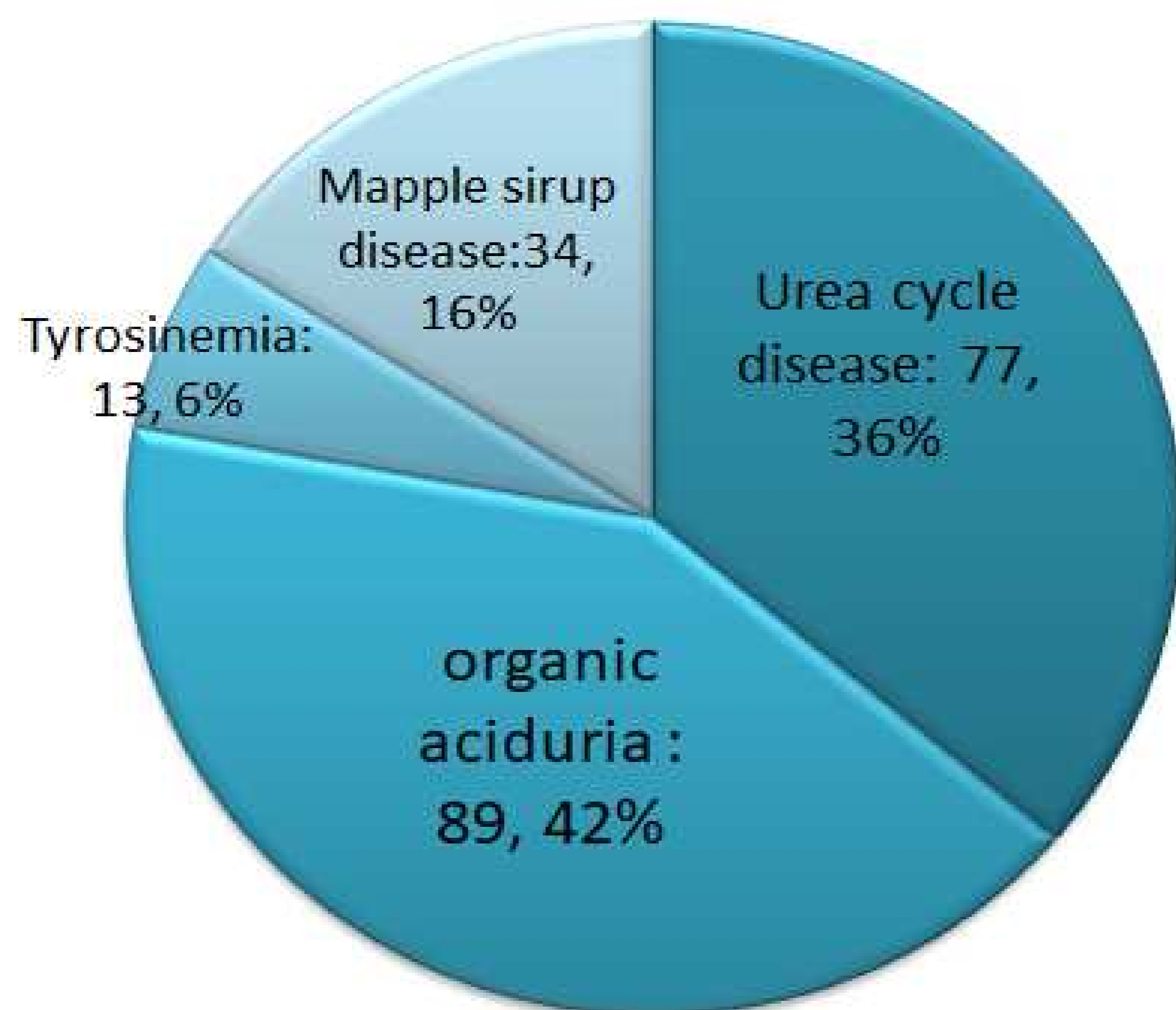


Figure 1: Diagnosis of IEM included

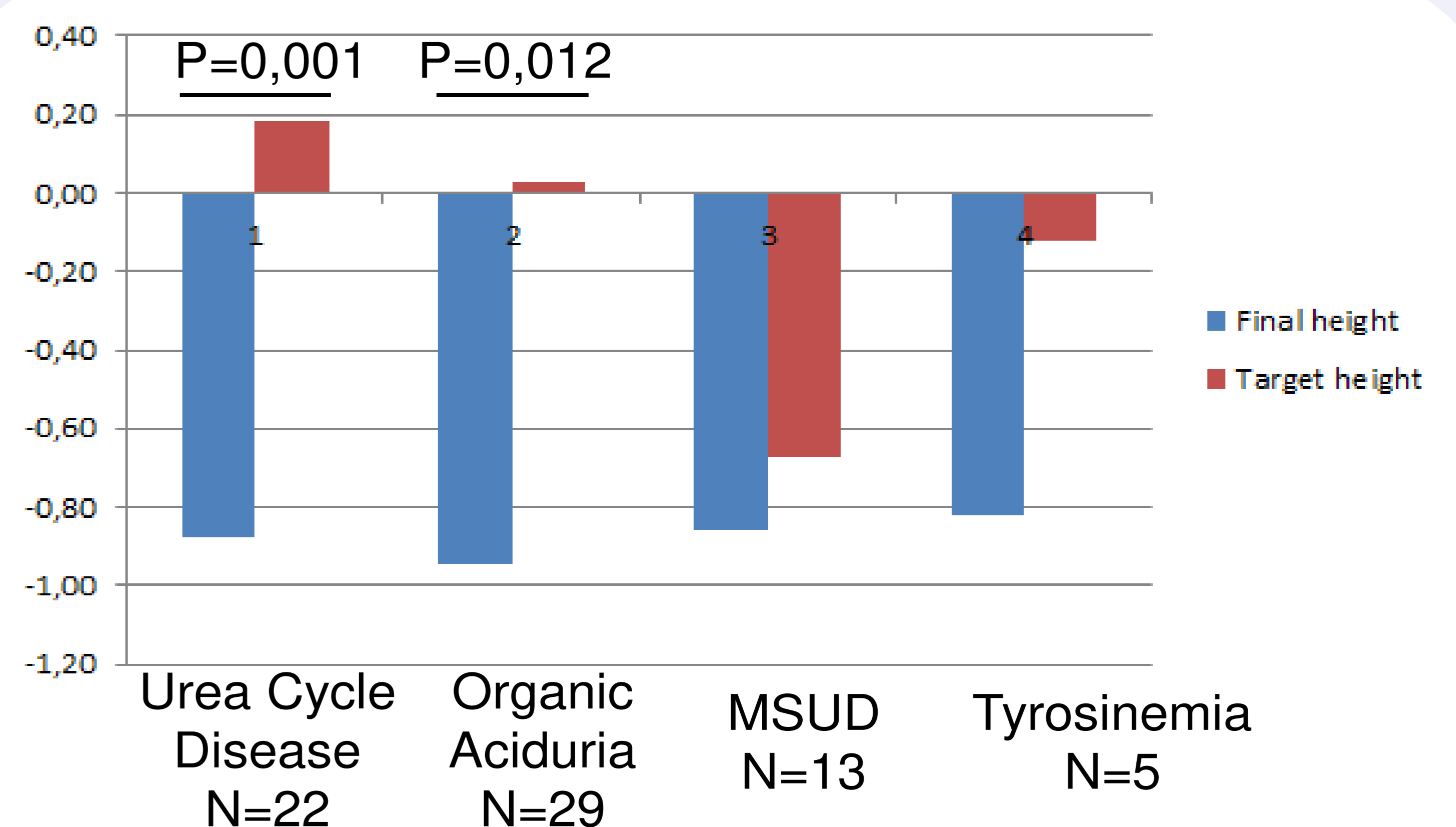


Figure 2: Final height (SDs) according to diagnosis

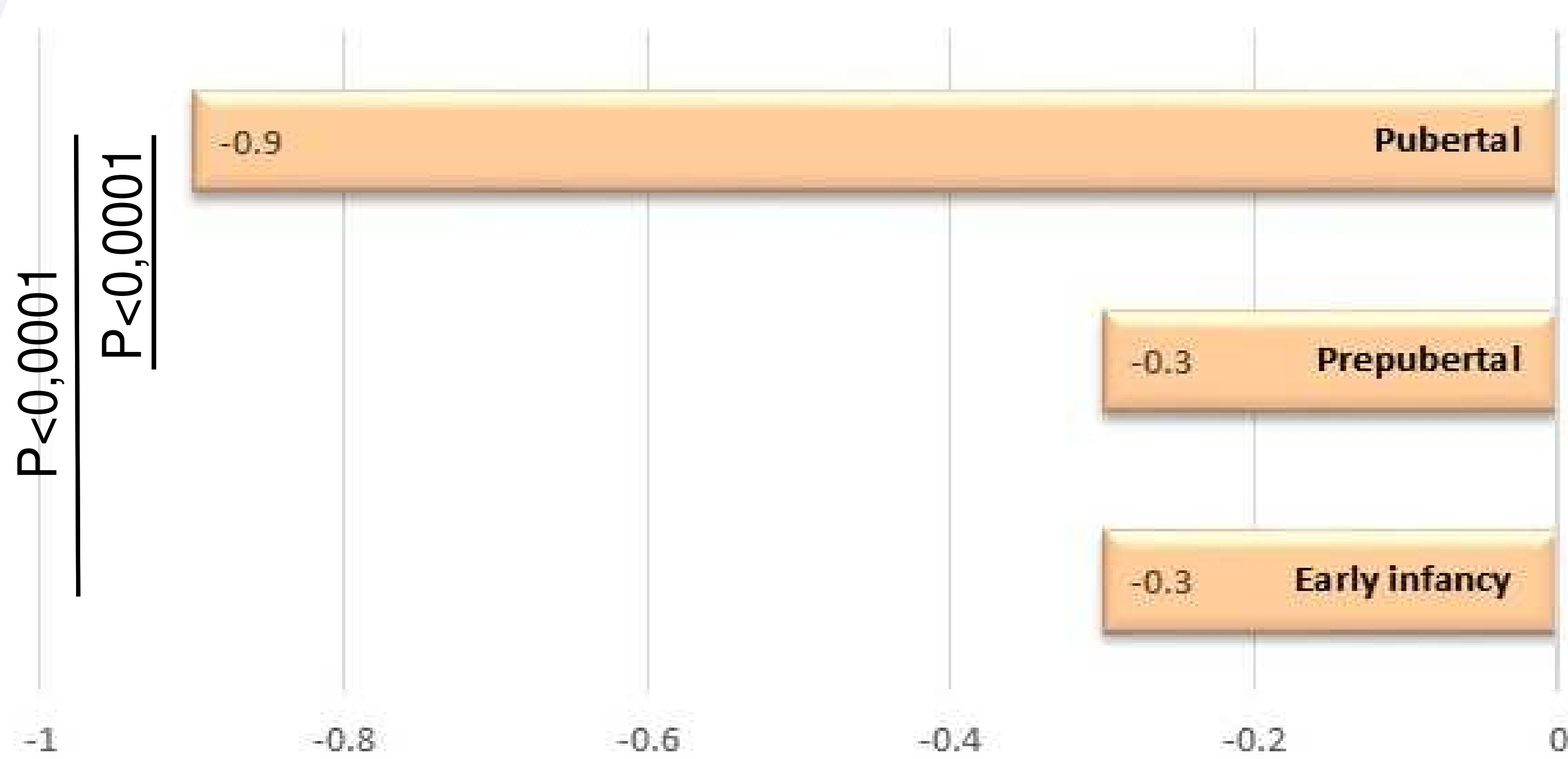


Figure 3: Height (SDs) according to age subgroups

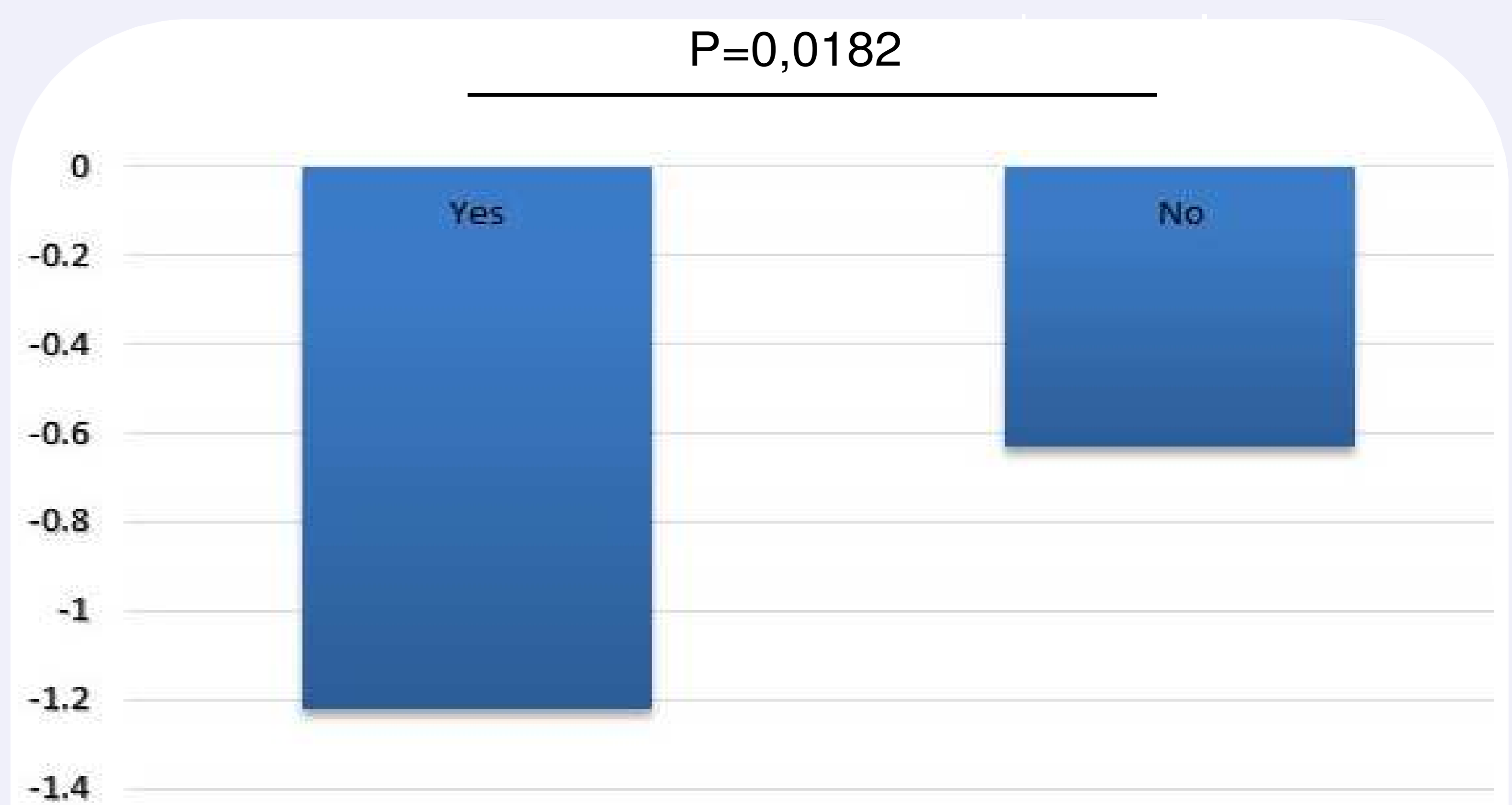


Figure 4: Height (SDs) in pubertal subgroup according to additional amino acid mixture

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.