

Possibilities of evolution in distal renal tubular acidosis - observation of two patients in a family

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Summary: Distal renal tubular acidosis is a rare genetic disease with the possibility of a favourable outcome.

Introduction: Distal renal tubular acidosis is a rare genetic disease, inherited in a predominantly autosomal recessive manner, but the autosomal dominant inheritance is also possible, with a potential evolution towards severe complications or an infaust evolution in the absence of therapy.

Clinical observation: In a family, two children, a boy and a girl, were diagnosed with distal renal tubular acidosis. Diagnosis was made at the age of 18 months, 8 months, respectively. Both children had polyuria and growth delay. Diagnosis was established based on the following criteria: clinical (polyuria, bone deformities, statural retardation), bio-humoral (metabolic acidosis, dyselectrolytemia: hypopotassemia, hypophosphatemia, hyperpotassiuria, hyperphosphaturia, high alkaline phosphatase), and imaging (bone mineralization disorders, nephrocalcinosis). The evolution of the two cases was different: in the case of the boy, inconsistent treatment caused renal failure, resulting in death at the age of 11 years. In the case of the girl, treatment (consisting in the preparation of sodium bicarbonate, potassium chloride, disodium phosphate, and vitamin D) although accepted only at the age of 9½ years, when some of the disease complications (nephrocalcinosis, hypophosphatemic rickets, and short stature) were already present, the results were satisfactory (resumption of statural growth, weight gain, clear improvement in Astrup parameters and normalization of the ionogram).

Conclusions: The observation presented emphasizes the possibility of a favourable outcome in a severe genetic disease.

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