Outcomes of vitamin D analogues and phosphate supplements in patients with hereditary hypophosphatemic rickets (HHR), comparison with non-treated patients.

Emese Boros (1), Anya Rothenbuhler (2), Claudine Heinrichs (1), Cecile Brachet (1), Laure Estere (2), Peter Kamenicky (3), Pol Harvengt (4), Sylvie Brailly-Tabard (5), Hazar Haidar (5), Celine Gaucher (6), Caroline Silve (2), Charles Gossiome (6), Philippe Wicart (7), Martin Biosse Duplan (6), Frederic Courson (6), Catherine Chaussain (6), Agnes Linglart (2)


- We retrospectively studied 108 adults currently followed or symptomatic first-degree relatives of patients followed at Bicêtre Hospital (Le Kremlin-Bicêtre, France) and Queen Fabiola Hospital (Brussels, Belgium)

- Group 1 - 50 patients who received VDA and phosphate supplements during childhood

- Group 2 - 58 patients who did not received VDA:
  • 27 out of 58 received phosphate supplements
  • 31 out of 58 never had any treatment

- Last available data were recorded from patients’ medical files.

- The diagnosis of HHR was made on biochemical criteria:
  1- low serum phosphate and
  2- low TRP

- The current “conventional” treatment improves height, leg bowing and cortical bone density.

- Corrective surgery is less frequent in patients who received “conventional” treatment during infancy.

- Our results in a large cohort of HHR patients confirm that the use of vitamin D analogues is safe and associated with better long-term outcomes.

- Several features of the disease are not cured and require new therapies

**BACKGROUND**

- Treatment with vitamin D analogues (VDA) and phosphate supplements (“conventional treatment”)

- The evidence of the so-called “conventional treatment” is based on few reports and reviews of cohorts of patients with HHR.

- Despite the current “conventional treatment”, complications include short stature, pseudofractures, due to severe osteomalacia, bone pain, muscle weakness, fatigue, hyperparathyroidism, nephrocalcinosis and enthesopathies.

- Our objective was to compare the adults who received the “conventional treatment” during their childhood with adults who did not, and appraise the burden of the disease in adulthood.

**METHODS**

- We retrospectively studied 108 adults currently followed or symptomatic first-degree relatives of patients followed at Bicêtre Hospital (Le Kremlin-Bicêtre, France) and Queen Fabiola Hospital (Brussels, Belgium)

- Group 1 - 50 patients who received VDA and phosphate supplements during childhood

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**RESULTS**

- Group 1 patients vs group 2 patients:
  • were taller
  • had better correction of leg bowing
  • had less leg corrective surgeries
  • had better dental health.

- VDA treatment was associated with a higher femoral neck T score and lower fracture incidence in adulthood.

- Complications such as nephrocalcinosis and hyperparathyroidism were similar between groups.

**CONCLUSIONS**

- The current “conventional” treatment improves height, leg bowing and cortical bone density.

- Corrective surgery is less frequent in patients who received “conventional” treatment during infancy.

- Our results in a large cohort of HHR patients confirm that the use of vitamin D analogues is safe and associated with better long-term outcomes.

- Several features of the disease are not cured and require new therapies