Aim: Hypophosphatemic rickets (HR) is a rare renal phosphate wasting disorder commonly related to X-linked form, caused by PHEX mutations and it treatment and follow-up is challenging due to imperfect treatment options. We aimed to present a nationwide data on HR with initial and follow-up data on the patients presented to the pediatric endocrinology clinics.

Method: CEDD-NET Data were used Inclusion criteria: Diagnosed between 0 to 18 years of age Hypophosphatemia Renal phosphate wasting Exclusion criteria: Calcitriopenic Rickets

Results: From 24 centers, 158 patients, before the age of 18 years, were included in the study data. Genetic analysis (n:75) showed PHEX mutation in 80%. The mean follow-up period was 6.7±2.4 years.

In follow-up: First 3 years treatment response (N:91) of patients, mild increase in P (from 2.6±0.6 to 2.7±0.6, 2.8±0.7 and 2.8±0.7 mg/dl), decrease in ALP (from 786±522 to 627±449, 561±319 and 546±327 U/L) and, elevation in PTH levels (from 68±28 to 84±77, 79±66 and 93±99 pg/ml) had been detected (from intitial to 1st, 2nd and 3rd year, respectively).

36% of the patients showed complete or partial improvement in leg deformities.

Conclusion: HR treatment and follow-up is challenging and higher calciotriol doses could improve bone deformities. However, higher treatment doses leading nephrocalcinosis without any change in serum levels, suggesting given higher doses lead higher phosphaturia probably through the stimulation FGFR23. Safer and more efficacious therapies are needed.