Nephrocalcinosis in children with X-Linked Hypophosphatemia: prevalence and risks factors

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INTRODUCTION: X-linked hypophosphatemia (XLH) :

- Rare genetical disease (3,9/100000)
- The most commun etiology of hereditary rickets
- Diagnosed with clinical and or radiological signs of rickets, impaired growth velocity, low serum phosphate levels associated with renal phosphate wasting, in the absence of vitamin D or calcium deficiency.
- Conventional treatment is made of phosphate and active vitamin D.
- **Nephrocalcinosis** prevalence is about 25-40% in adult XLH treated patients. It was never characterized in large cohorts of XLH children. **Consequence of the treatment or marker of severity of XLH?**

MAIN OBJECTIVE: to evaluate the prevalence of nephrocalcinosis and identify risk factors associated to the occurrence of this complication.

METHODS:

- **117 children** (74 girls and 43 boys) i.e **the most important cohort of children with XLH.**
- Followed between 2010 and 2019
- **Diagnosis of nephrocalcinosis:** systematic renal ultrasound.
- **Potential risks factors:**
  - **Markers of clinical and biochemical disease severity**, (short stature, lower limb deformities, dental abscesses, craniosynostosis, Chiari malformation, bone fracture, bone surgery and deafness).
  - **Treatment follow up** (observance, duration of treatment, mean dose of treatment during the last four years preceeding the diagnostic of nephrocalcinosis or the last visit).

RESULTS:

Prevalence of nephrocalcinosis: 40,2% (n=47/117)

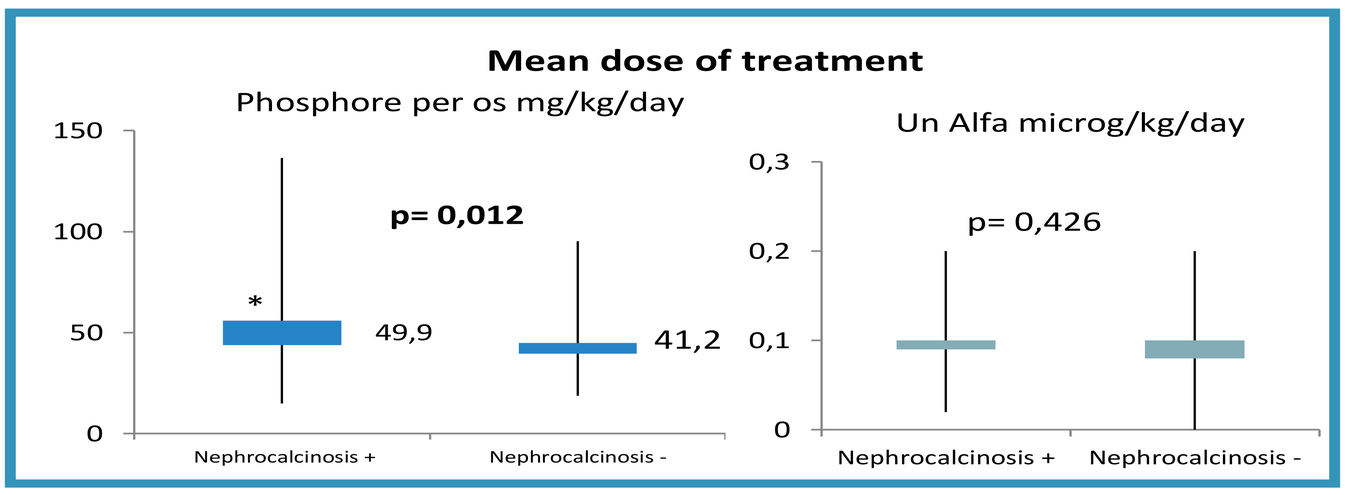
- After 6.6±3.8 years of conventional treatment.
- No impact on renal function

Population caracteristics

	Nephrocalcinosis +	Nephrocalcinosis -	Total
Sexe (F)	28 (60%)	46 (65%)	74 (63%)
Age (years)	8,8	9,9	9,4
Height (cm)	123	131	127
Weight (kg)	29	35	32
PHEX	34 (72%)	47 (66%)	81 (69%)
Familial history	25 (53%)	39 (55%)	64 (55%)

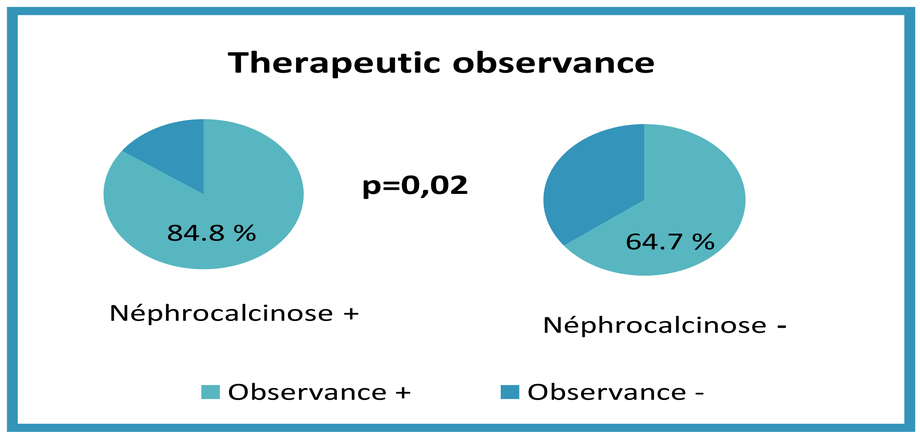
Patients with nephrocalcinosis:

Had significantly higher dose of phosphate supplements …

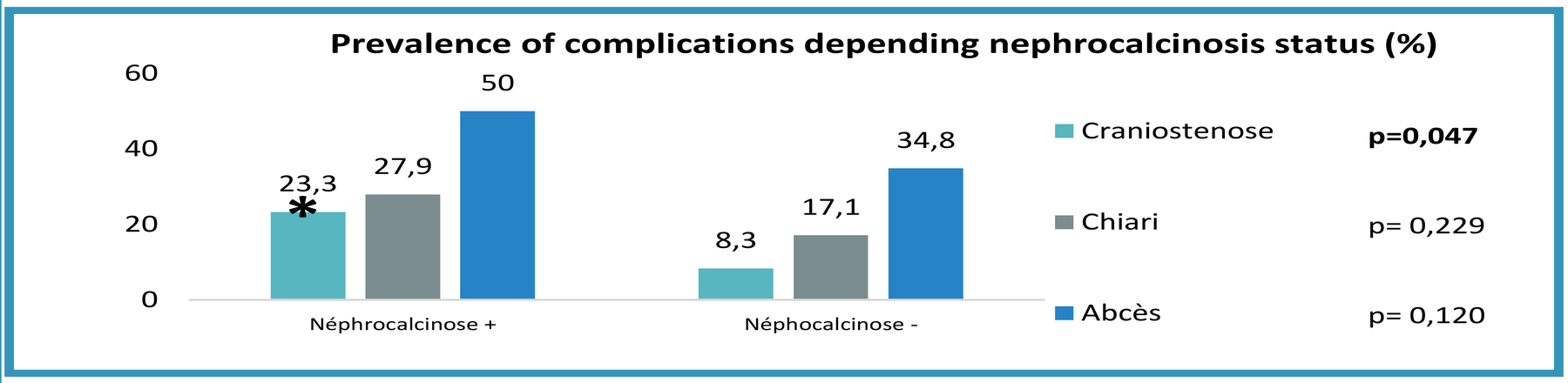


- during the 4 years prior to the study evaluation than patients without nephrocalcinosis, i.e. **49,9±20,5** mg/kg/day versus **41,2±11,4** mg/kg/day, **p=0.012**
- No significant difference between the two groups for the dose of Un Alfa.

Were more observant…



Had more severe complications…



CONCLUSION:

Nephrocalcinosis is a frequent complication without impact on the renal function during childhood (but probable occurrence at adulthood).
Complication of the treatment? Seems to be correlated to higher dose of phosphore supplementation.
Or complication of XLH? Seems to be correlated to more severe complications.