Results of orthopaedic surgery in children with X-linked hypophosphatemic rickets (XLHR)

Aliette Gizard[1], Anya Rothenbuhler[2], Zagorka Pejin[3], Georges Finidori[3], Christophe Glorion[3], Benoit de Billy[1], Agnès Linglart[2] et Philippe Wicart[3]

[1]CHU de Besançon, Besançon, France; [2]APHP, CMR Calcium-Phosphore, Plateforme d'Expertise Paris Sud Maladies Rares et Endocrinologie Pédiatrique, Bicêtre Paris Sud, France; [3]APHP, Necker-Enfants Malades, Paris, France

INTRODUCTION

XLHR is due to mutations in the PHEX gene leading to unregulated production of FGF23, hence hypophosphatemia and decreased renal 1,25OH-vitamin D hydroxylation. Amongst other features, XLHR is characterized by leg bowing of variable severity. Phosphate supplements and oral 1,25OH-vitamin D, partially or, in some cases, fully restore the limb straightness. For severe or residual limb deformities, orthopaedic surgery may be recommended.

OBJECTIVE AND METHODS

To retrospectively assess the results of surgical limb correction in in 49 XLHR children (19M, 30F) bearing a PHEX mutation (mean age at diagnosis 6.0 yrs [± 7.1]) who underwent at least one leg surgery.

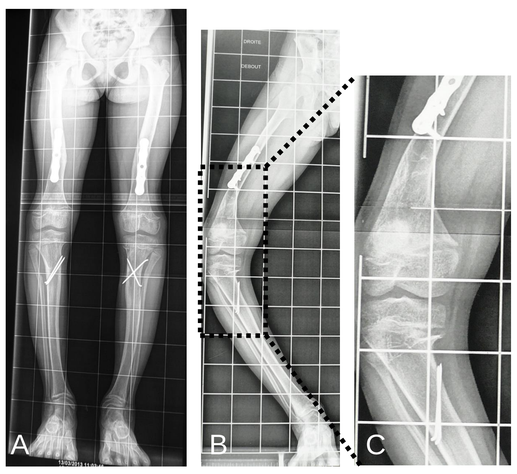

Figure 2: Evolution post surgery in a severe case
A. Correction after surgery (age 7.9 yrs). **B**. Recurrence of the varum below the femoral osteotomy (age 9.5 years). **C**. Enlarged picture of the femoral and tibial physis (framed in B.) showing the medial epiphysiodesis although the surgery was performed at distance from the physis.

RESULTS

42/49 patients (65%) had a *genu varum* at the 1st surgery
→Mean distance between the knees: 11.1 cm [± 6.4].
7/49 patients (35%) had a *genu valgum* at the 1st surgery
→Mean distance between the ankles: 14.3 cm [± 5.7]

Prior to surgery, 42/49 received alfalcalcidol and phosphate 29/49 had a value of Alk. Phos. within the normal range.

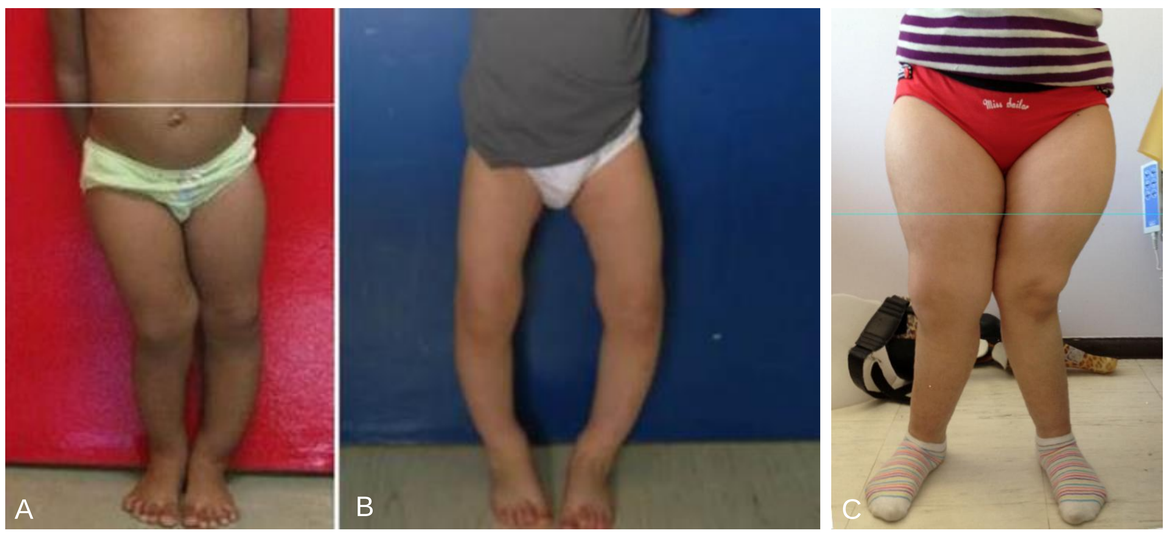

Figure 1: Leg deformities in XLHR
A. Right leg in valgum, left leg in varum. **B.** genu valgum. **C.** Genu valgum

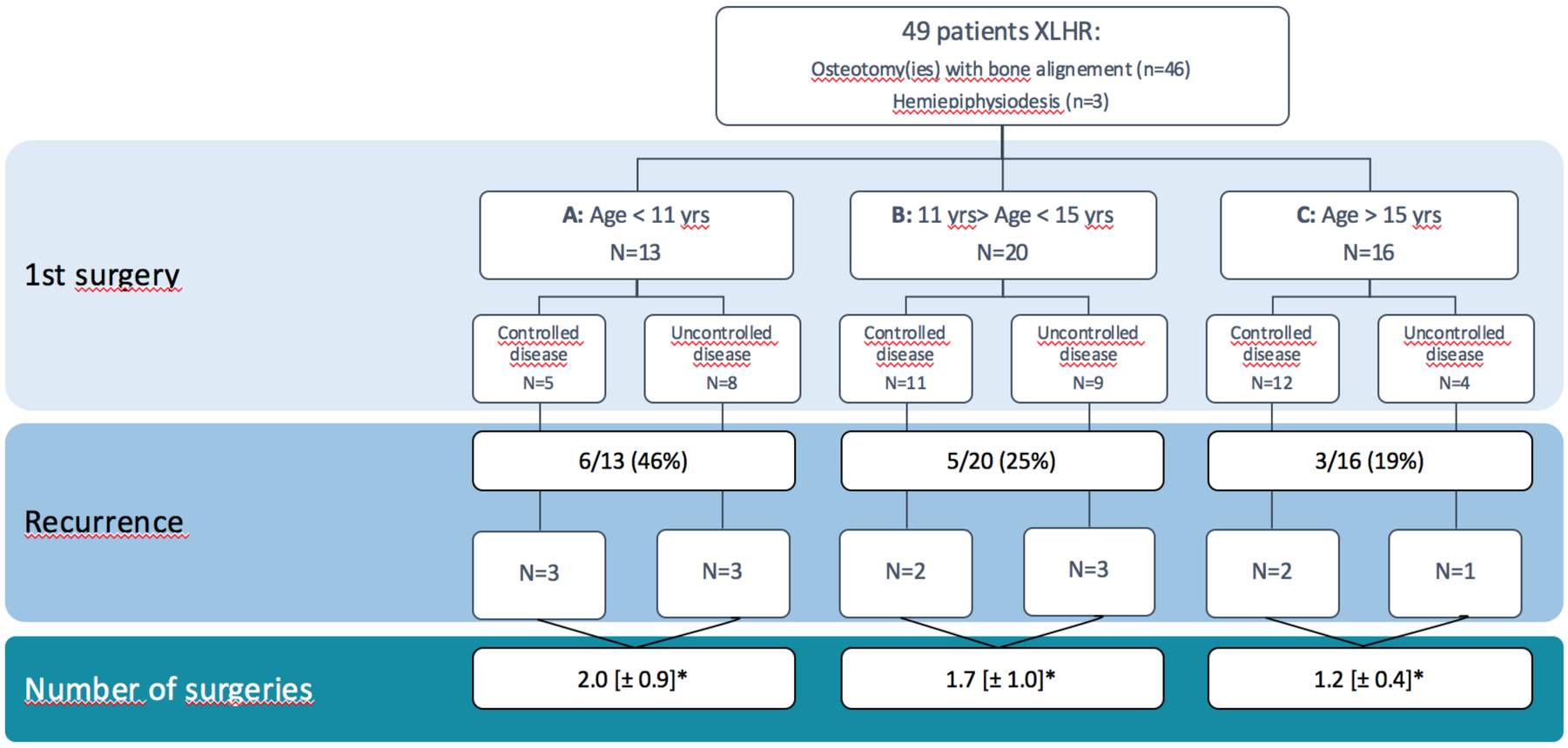

Figure 3: Flow chart of the surgeries and evolution throughout time. Patients are divided in 3 groups (A, B and C) according to their age at the 1st surgery. The number of patients with a controlled or non-controlled rickets is shown. Overall, the mean number of surgeries per patient is shown in the bottom panel. The comparison of the three groups is statistically significant (*).

CONCLUSIONS

We report here the largest series of surgical procedures in XLHR. Our results confirm that phosphate supplements and vitamin D analogues therapy is the first line of treatment in XLHR to correct the leg bowing. Early surgeries are associated with a high risk of relapse of the limb deformity. Such procedures should be recommended, as a multidisciplinary decision, only in patients with severe distortion leading to mechanical joint and ligaments complications, or for residual deformities once growth plates are fused.

REFERENCES

-**Bergwitz C, Jüppner H.** FGF23 and syndromes of abnormal renal phosphate handling. Adv Exp Med Biol. 2012;728:41–64.
-**Linglart A, Biosse-Duplan M, Briot K, Chaussain C, Esterle L, Guillaume-Czitrom S, et al.** Therapeutic management of hypophosphatemic rickets from infancy to adulthood. Endocr Connect. 2014;3(1):R13-30.
-**Carpenter TO, Imel EA, Holm IA, Jan de Beur SM, Insogna KL.** A clinician's guide to X-linked hypophosphatemia. J Bone Miner Res Off J Am Soc Bone Miner Res. 2011 Jul;26(7):1381–8.
-**Mäkitie O, Doria A, Kooh SW, Cole WG, Daneman A, Sochett E.** Early treatment improves growth and biochemical and radiographic outcome in X-linked hypophosphatemic rickets. J Clin Endocrinol Metab. 2003 Aug;88(8):3591–7.

55ESPE 2--4-RFC
Bone & Mineral Metabolism
Agnès LINGLART
DOI: 10.3252/pso.eu.55ESPE.2016
Poster presented at: ESPE 2016 PARIS