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INTRODUCTION

X-linked hypophosphatemic rickets (XLHR) is due to mutations in the PHEX gene inducing increased levels of fibroblast growth factor 23 (FGF23), phosphate wasting, hence rickets. FGF23 is suspected to be as an important metabolic regulator of glucose and lipid metabolism.

Objective : To describe the metabolic profile (body mass index, blood pressure, glucid and lipid profile) in patients with XLHR and evaluate the correlation between FGF23 levels and metabolic biomarkers.

RESULTS

55 XLH patients (17 boys and 38 girls) (PHEX mutated) were included (mean age : 8.0 ± 4.3 years ; range 0.3-18 years).

Each subject was classified based on International Obesity Taskforce (IOTF) cut off values of BMI for age (Figure 1) and sex as overweight (IOTF >25) or obese (IOTF >30).

Mean IOTF of study population was 23.9 ± 3.2 . 42% of patients had IOTF > 25 of which 12/55 (22%) were overweight (IOTF mean : 25.9 ± 1.13) and 11/55 (20%) were obese (IOTF mean: 31.2 ± 3) (Figure 2).

When stratified by age, children show a dramatic increase of BMI z-score (SDS) over time (Figure 1).

Metabolic data were correlated with FGF23 level ($p=0.0237$) (Figure 3).

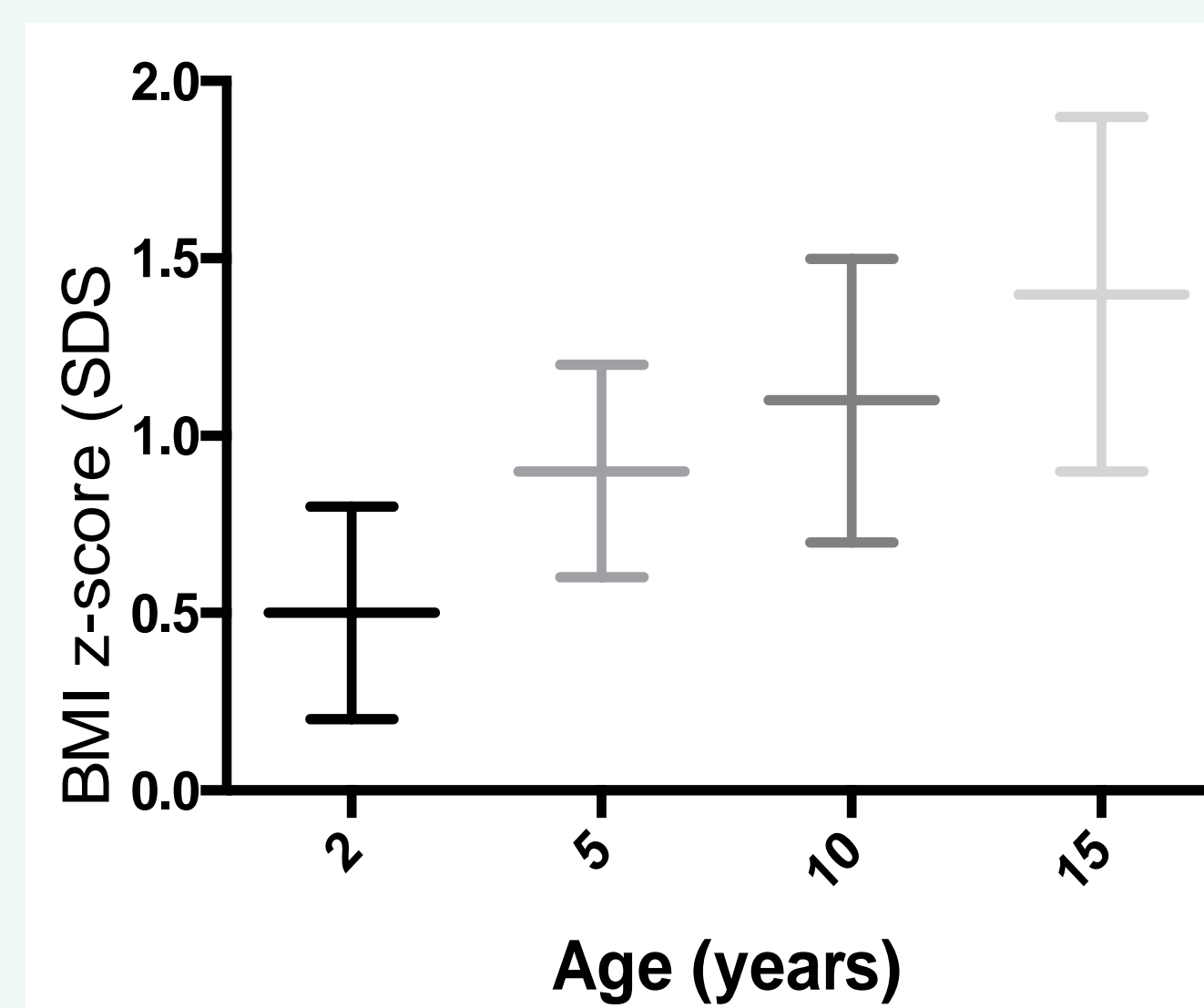


Figure 1

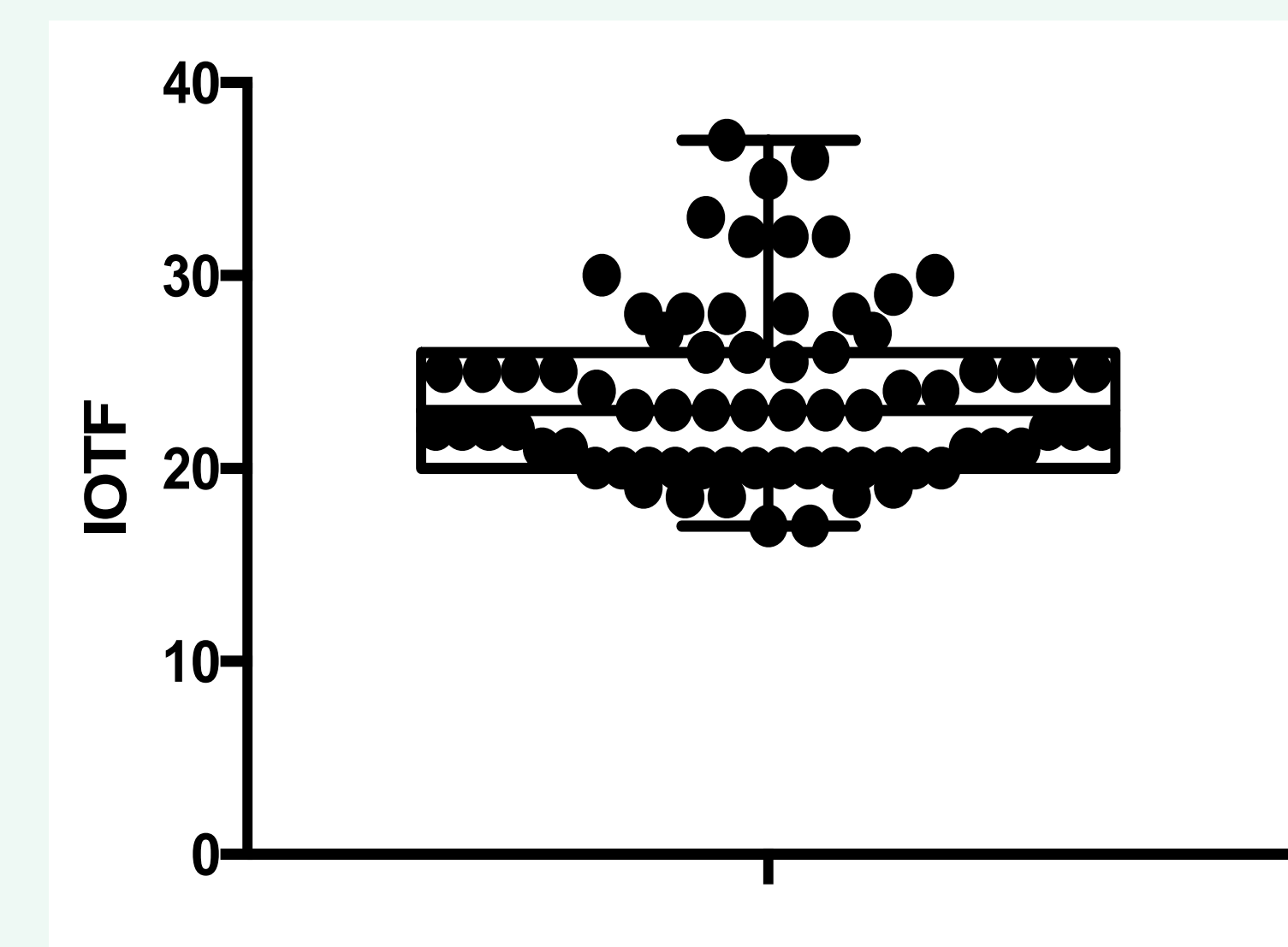


Figure 2

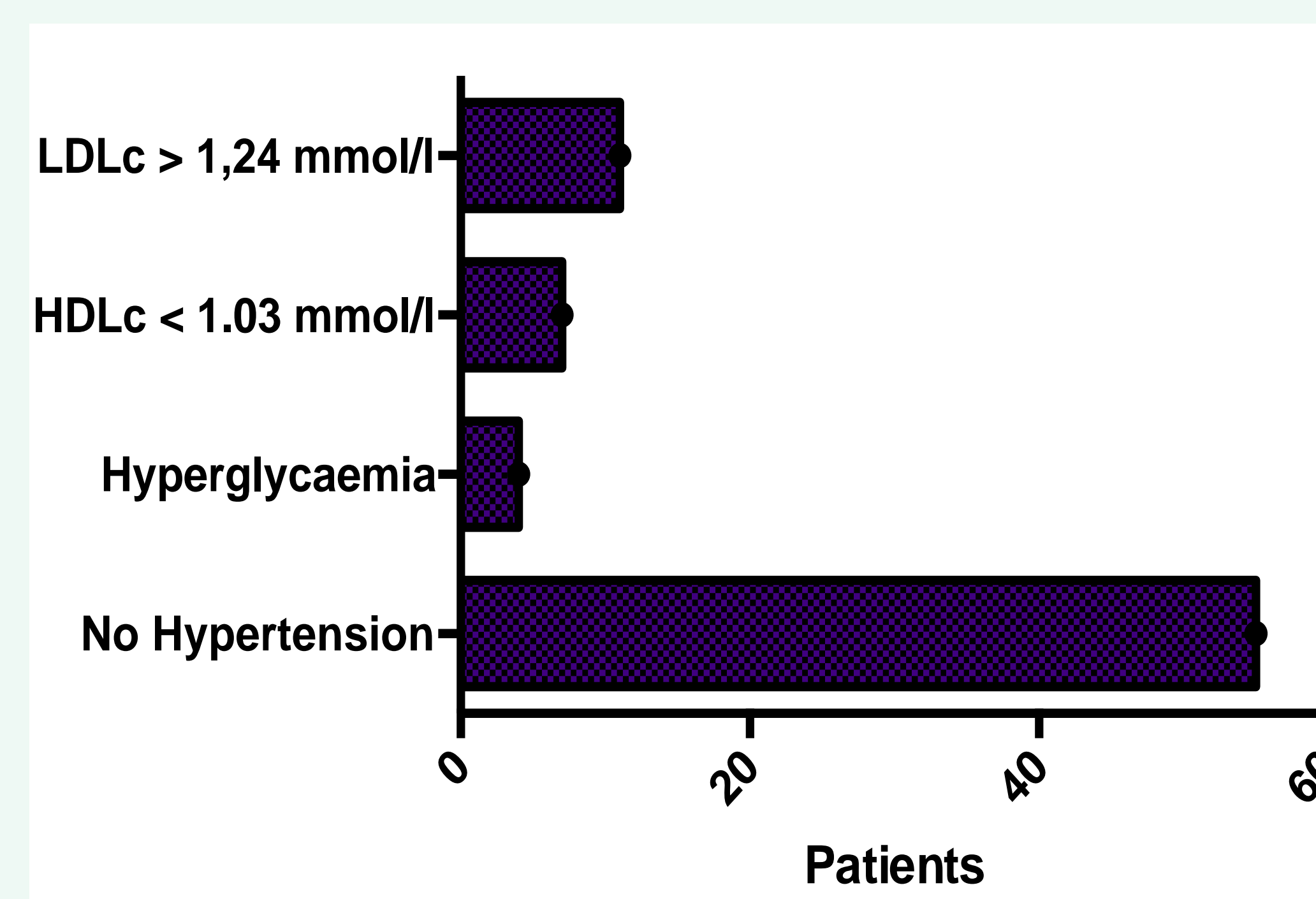


Figure 3

CONCLUSION

This pilot study shows that children with XLHR gain too much weight during childhood, and that there may be an association between FGF23 and the development of metabolic syndrome. Further investigations are needed in a larger cohort of children and in adults to define the specific roles of FGF23 and PHEX in the development of metabolic syndrome.

