

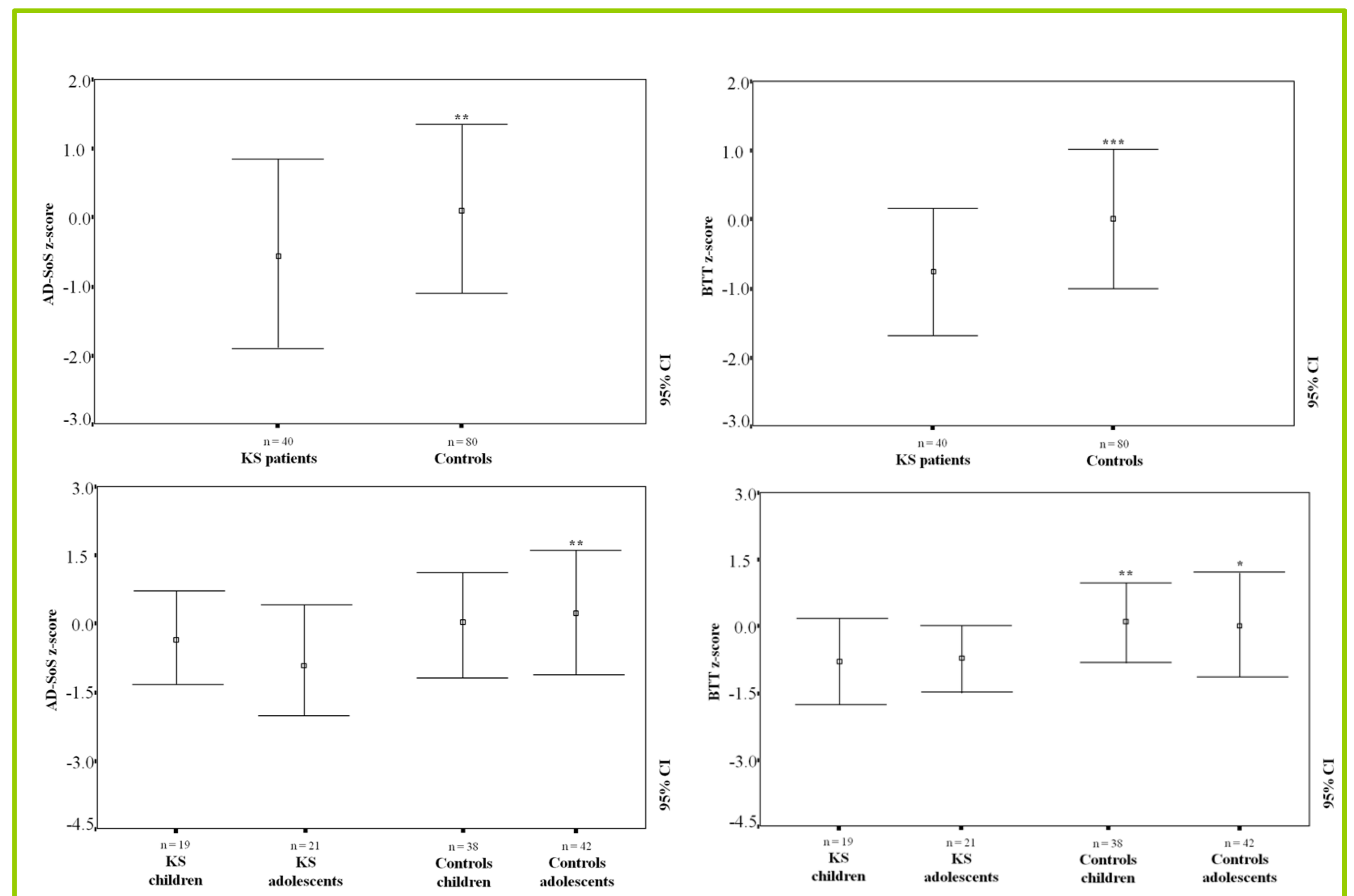
## INTRODUCTION

Klinefelter syndrome (KS) is a common chromosomal disorder with a prevalence of 120-153 per 100,000 live-born male births and is characterized by the presence of one extra X chromosome in a male phenotype (47, XXY karyotype). The major finding is represented by primary hypogonadism due to seminiferous tubule dysgenesis and androgen deficiency, resulting in a progressive testicular failure. Testosterone is an important hormone to achieve bone maturation and adequate peak bone mass. In KS metabolic bone disorders have been reported and low testosterone may be responsible of decreased bone mineral density (BMD). On the contrary, testosterone therapy did not necessarily increase bone mass, and low bone mass was also observed in patients with normal testosterone levels. The purpose of our study was to evaluate bone mineral status and metabolism in a cohort of KS children and adolescents.

## METHODS

40 KS children (mean age  $13.7 \pm 3.8$  years) evaluated to Meyer's Children Hospital of Florence from 2013 to 2015 were cross-sectionally studied and the following parameters were compared to 80 age- and body-size-matched control group :

- $Ca^{2+}$ , total Calcium,  $P_i$ , PTH,
- 25(OH)D and  $1,25(OH)_2D$ ,
- osteocalcin,
- bone ALP,
- LH, FSH, Testosterone
- urinary deoxypyridinoline
- phalangeal amplitude-dependent speed of sound (AD-SoS) and bone transmission time (BTT) z-scores



## RESULTS

- KS children and adolescents showed a significantly reduced AD-SoS ( $P < 0.005$ ) and BTT ( $P < 0.0005$ ) z-scores than controls. These metabolic bone disorders were present already in prepubertal subjects.
- AD-SoS and BTT z-scores correlated negatively with PTH levels ( $P < 0.005$ ) and with age ( $P < 0.0001$ ) and positively with 25(OH)D levels ( $P < 0.005$ ).
- KS patients presented significantly higher PTH levels ( $P < 0.0001$ ) and significantly lower 25(OH)D ( $P < 0.0001$ ), osteocalcin ( $P < 0.05$ ), and bone alkaline phosphatase levels ( $P < 0.005$ ).
- PTH correlated significantly with age ( $P < 0.0001$ ), calcium ( $P < 0.005$ ), LH ( $P < 0.0001$ ), FSH ( $P < 0.0001$ ), total testosterone ( $P < 0.0001$ ), vitamin D ( $P = 0.04$ ), and osteocalcin ( $P = 0.002$ ) levels.

## CONCLUSIONS

Our study showed that subjects with KS have a significant reduction in bone mineral status and an impaired bone metabolism compared to controls, focusing on the need of a close follow up in these subjects.

## REFERENCES

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