# Triple X Syndrome: An Evaluation of Bone Mineral Status and Metabolism

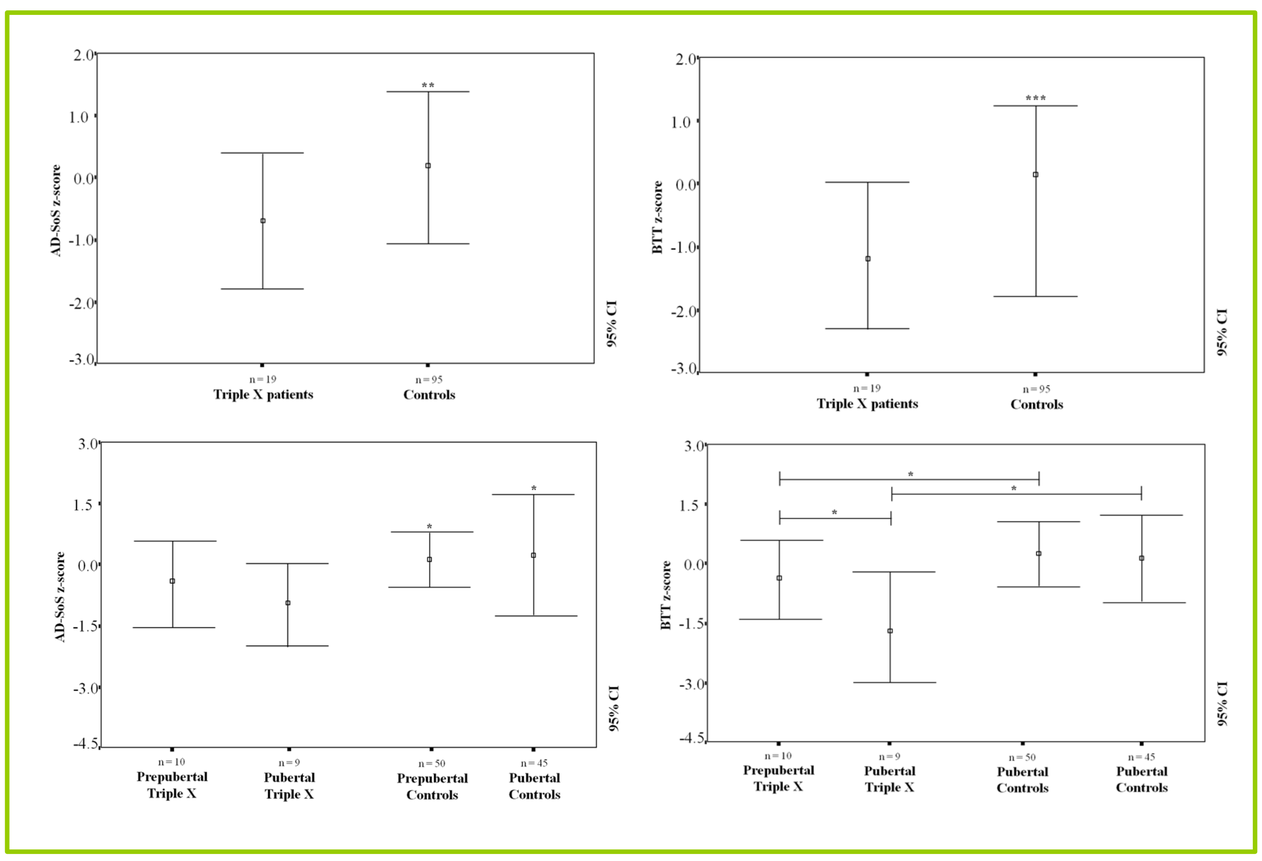
## INTRODUCTION

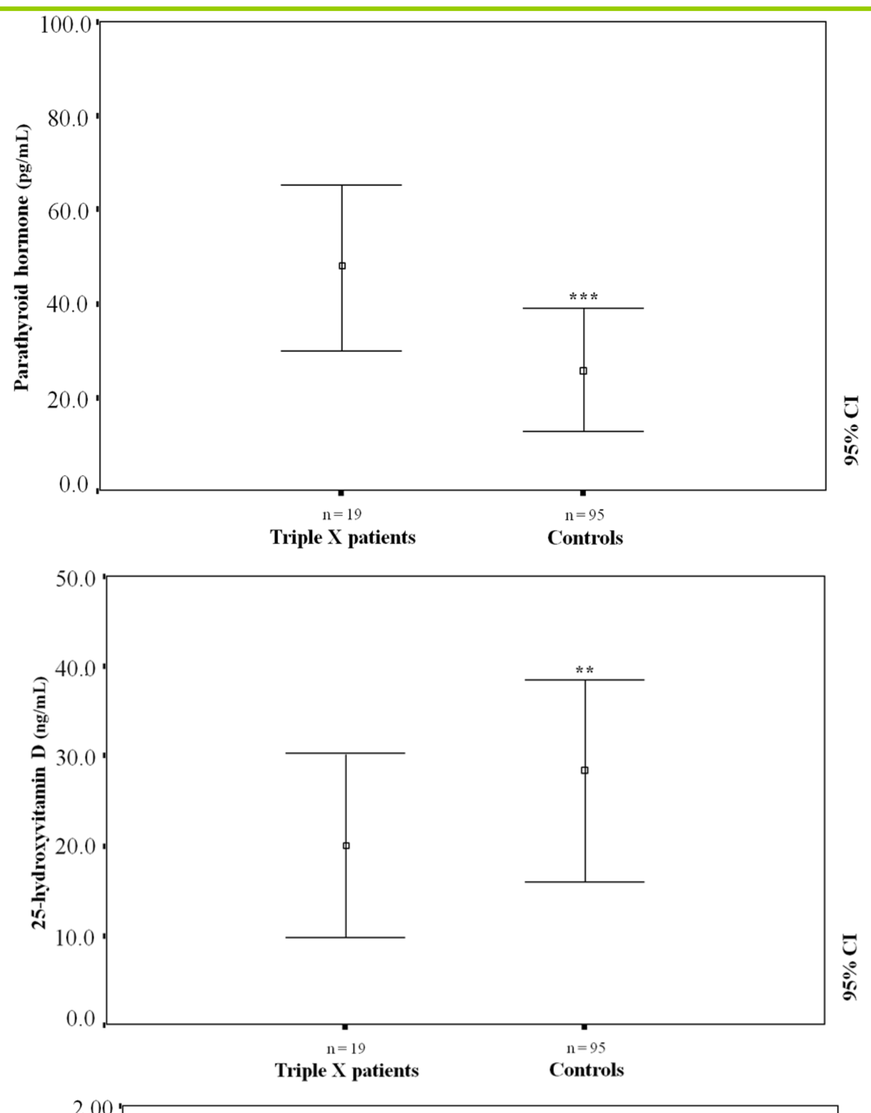
Triple X Syndrome (47, XXX) is due to a non-disjunction during the meiotic period and affects as many as 1 in 1,000 live female births. It's often misdiagnosed because of the mild and variable phenotype. This girls tend to be tall and thin and reproductive capability is generally not affected, although premature ovarian failure seems to be more prevalent. It is also described increased frequency of bilateral polycystic kidney, urethral stricture, menstrual irregularity, ovarian dysgenesis. No study has considered the effect of a supernumerary X chromosome on bone mineral status and bone metabolism, which is impaired in other chromosomal disorders. Therefore, the purpose of our study was to evaluate bone mineral status and bone metabolism in a cohort of patients with non-mosaic triple X syndrome.

## METHODS

Nineteen girls with non-mosaic Triple X syndrome were evaluated at Meyer's Children's Hospital of Florencefrom 2013 to 2015 and cross-sectionally studied. The following parameters were compared to an age- and body-size-matched control group

- Ca $^{2+}$,total Calcium, $P_i$
- PTH
- 25(OH)D and1,25$(OH)_2$ D,
- osteocalcin,
- bone ALP,
- urinary deoxypyridinoline
- phalangeal amplitude-dependent speed of sound (AD-SoS) and bone transmission time (BTT) z-scores




## RESULTS

Triple X patients showed significantly reduced AD-SoS ($p<0.005$) and BTT z-scores ($p<0.0001$) than the controls. These results persisted when we divided the sample into pre-pubertal and pubertal patients ($p < 0.05$). Our patients also had significantly lower ionised calcium and 25(OH)D ($p<0.005$), wile phosphate ($p<0.0001$) and PTH ($p < 0.0001$) levels were higher.
AD-SoS and BTT z-scores values were significantly inversely correlated with age ($p < 0.005$), PTH ($p < 0.005$), and 25(OH)D ($p < 0.005$) levels.

## CONCLUSIONS

Our study showed that subjects with non-mosaic triple X syndrome 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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