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Can Boys Have Turner Syndrome? An Analysis of 45,X/46,XY Mosaicism

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Background

Individuals with 45,X/46,XY mosaicism raised as boys are not classified as Turner Syndrome (TS), which is a label restricted to phenotypic females.[1] These girls routinely receive growth hormone treatment and are screened for multiple comorbidities associated with TS, but this is not routinely recommended for boys with 45,X mosaicism. We sought to determine if those raised as boys had similar rates of comorbidities compared to those raised as girls.

Methods

Retrospective chart review of patients seen in multidisciplinary DSD clinic with 45,X mosaicism. All had a Y chromosome component. Degree of genital masculinization assessed by External Masculinization score.[2] Height z-scores were recorded before growth hormone treatment. Turner Syndrome screenings[1] were assessed for each patient.

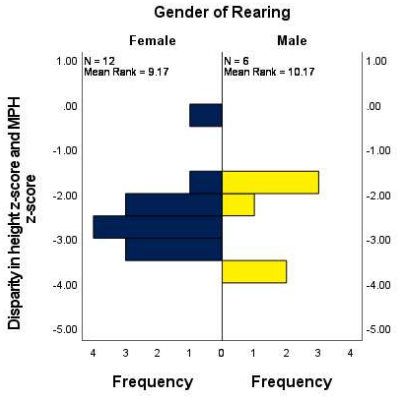
Results

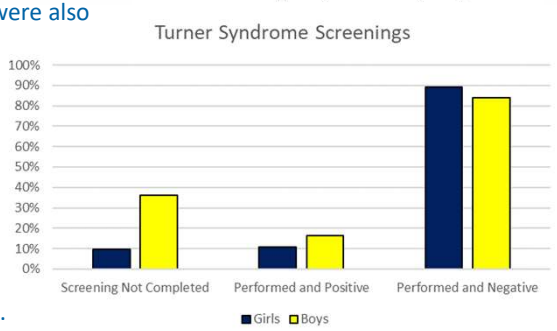
A total of 22 patients were identified (14 girls, 6 boys). One girl was excluded from height analysis for a comorbid diagnosis of congenital adrenal hyperplasia, and one girl did not have MPH available. Mann Whitney U and Chi-Square analysis performed using SPSS (Version 27, IBM).

Girls had less genital ambiguity than boys (p=0.004). All patients had height z-score below mean for age. Z-scores did not significantly differ between girls and boys (p=0.185). This remained true when correcting for MPH (p=0.75).

Screenings included cardiac and renal anomalies, audiology evaluation, and thyroid disease in all children. In those over age 2 yrs, celiac screening was included, and for those older than 10 yrs, A1c, liver function, and 25-OH Vit D were also obtained. Girls were significantly more likely to have screening tests done (p<0.001), but the percentage of abnormal screens of those completed were not significantly different between boys and girls (p=0.405).

Cardiac conditions were common in both groups: 62.5% of boys compared to 78.5% of girls had a cardiac anomaly. There were insufficient number of other screenings to perform individual analysis.





Conclusions

These data suggest that boys with 45,X/46,XY mosaicism have similar rates of comorbidities to girls with the same genotype. Given similar findings in previous studies,[3] a multicenter registry would help clarify and expand these findings. Furthermore, these data suggest the need to screen boys with 45,X mosaicism according to the Turner Syndrome Clinical Practice Guidelines[1] in order to allow for early recognition of comorbidities. Obtaining a karyotype on boys who have features of Turner Syndrome may increase recognition of this condition.

References

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