INTRODUCTION

- Turner Syndrome (TS) is a genetic condition that occurs in ~1 in 2000 females who have a partial or complete absence of the second sex chromosome.
- TS may occur with mosaicism for a typical 46,XX (20% of TS) and/or 47,XXX (~3% of TS) cell lines.
- Literature on mosaic 45,X/47,XXX are limited to case reports and series.
- The objective of this study was to describe the phenotype of 45,X/47,XXX and compare to non-mosaic TS (45,X) and mosaic for an XX cell line (45,X/46,XX).

METHODS

- 246 females with TS were enrolled into the InsighTS Registry, a prospective study of TS in six large academic institutions.
- Diagnostic, birth, and neonatal histories were obtained from medical records.
- Individuals with karyotypes of 45,X, 45,X/46,XX, and 45,X/47,XXX were included in this analysis.
- All three karyotype groups were compared using ANOVA for continuous variables and Chi-squared or Fisher’s Exact for categorical variables; pairwise comparisons were conducted when group differences were significant (alpha<0.05).

LIMITATIONS AND FUTURE DIRECTIONS

- Small sample size for the 45,X/47,XXX group; convenience sample may not be generalizable.
- Chart reviews may lead to errors of omission or commission from medical record.
- Enrollment continues for the InsighTS registry and future data collection includes comorbidities, interventions, and patient-centered outcomes.

CONCLUSIONS

- Within the InsighTS registry, 7% of girls with TS have mosaicism for a trisomy X cell line.
- Girls with 45,X/47,XXX showed similar reasons for postnatal diagnosis and perinatal history to those with 45,X/46,XX but differed from those with non-mosaic TS.

REFERENCES