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Description of Turner Syndrome Diagnosis in Nationally Representative Sample: Inspiring New Science in Guiding Healthcare in Turner Syndrome (InsighTS) Registry

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Objectives

Turner syndrome (TS) occurs in ~1 in 2,000 females who are born with partial or complete absence of the second sex chromosome. Inspiring New Science in Guiding Healthcare in Turner Syndrome (InsighTS) is a national collaboration developed to address limitations of prior TS research. With the increased use of non-invasive prenatal screening (NIPS) in routine prenatal care, we sought to use the InsighTS registry to determine whether patients incidentally identified to have TS differ in terms of genetic and demographic variables from those that are diagnosed secondary to symptoms.

Methods

Six institutions with multidisciplinary TS clinics across geographical regions in the United States enrolled patients with a known TS diagnosis, and those with a genetic test available for review (n=201) were included in this analysis. Reason for genetic testing was categorized as incidental if no symptoms led to genetic testing (e.g., NIPS in fetus or mother). Site, race, ethnicity, insurance, karyotype, and timing of diagnosis were compared between incidental and symptomatic diagnostic groups using Chi-square tests; age of enrollment was compared using Mann-Whitney test. Significance was set at $\alpha=0.05$.

Results

This sample is highly diverse with about one third (32.5%) identifying as a person of color (defined as not non-Hispanic white), varied insurance status (36.8% Medicare/Medicaid at enrollment), most enrolling as minors (83.6%), varied timing of TS diagnosis (66.7% postnatally

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at age 6.3 ± 6.8 years), and heterogenous karyotypes (39.3% non-mosaic monosomy X). Overall, 21 (14.4%) were diagnosed via incidental screening. Race ($p=0.53$), ethnicity ($p=0.19$), insurance ($p=0.43$), site of enrollment ($p=0.17$), and karyotype ($p=0.61$) were not different between groups. However, those with an incidental screening diagnosis were more likely to be identified prenatally ($p<0.001$) and enrolled at an earlier age ($p<0.001$).

Conclusions

In a diverse, national sample of girls and women with TS, patients diagnosed incidentally do not differ from those that are diagnosed secondary to symptoms in terms of karyotype or demographic characteristics. As expected, incidentally identified patients were more likely to be diagnosed prenatally and to enroll in the registry at an earlier age. Future directions for this work include increasing the cohort size and analyzing medical outcomes including comorbid diagnoses and longitudinal growth parameters to guide counseling and management for patients incidentally identified.