Thyroid Disorders in Girls with Turner Syndrome and the Influence of the Underlying Karyotype

Abstract:
Sir

The risk of developing thyroid dysfunction is higher in Turner syndrome (TS) than the general population. In previous studies, the influence of karyotype and thyroid autoantibodies on thyroid disorder in patients with TS has been investigated. We therefore set out to determine the prevalence of thyroid dysfunction in Irish girls with TS. The impact of underlying karyotype and thyroid autoantibodies has also been examined. The presence of thyroid dysfunction was assessed by measuring serum thyroid-stimulating hormone (TSH), thyroxin (T4) and anti-thyroid peroxidase antibodies (TPO-Ab) values. The association between TPO-Ab values and thyroid dysfunction (hypothyroidism and hyperthyroidism) was also assessed.

We studied 32 girls with TS; mean (SD) [range] age 16.7 (2.6) [12.4-20.2] years. Of 32 girls, 14 (43.75 %) had structural X abnormalities, 12 (37.5%) experienced monosomy 45, X and 6 (18.75%) exhibited mosaicism monosomy X. In this group of girls with TS, thyroid abnormalities were reported in 5 of 32 girls (15.6%), of whom 4 of 5 (80%) had hypothyroidism and 1 of 5 (20%) experienced hyperthyroidism. Of 4 girls with hypothyroidism, 2 (50%) had monosomy X chromosome (45, X) and 2 (50%) exhibited isochromosome-X structural abnormalities (45,X;46,X,i(Xq)/47,X,i(Xq),i(Xq) and (46,X,i(X)/46,XX)]. Hyperthyroidism was found in 1 girl with X structural abnormalities (46,X,iel(X)(p11.2;p22.3)). In this study, hypothyroidism occurred in girls with monosomy X chromosome and isochromosome-X structural abnormalities, but not mosaicism monosomy X. In this group of patients with TS, the underlying karyotype does not appear to influence the risk of hypothyroidism (p value = 0.93), whether it would have reach significant level if the number of patients would have been higher is unclear. Further study with larger number of patients may be required to explore this issue. TPO-Ab values were higher than the reference ranges in 8 of 32 girls (25%), of whom 3 (37.5%) exhibited normal thyroid function, 4 (50%) had hypothyroidism and 1 (12.5%) had hyperthyroidism. TPO-Ab showed significant association with thyroid dysfunction (p value < 0.02).

In conclusion, the prevalence of hypothyroidism in girls with TS is higher than the general population and anti-thyroid peroxidase antibodies values, but not the underlying karyotype, appear to influence risk. With respect to the relatively high prevalence of thyroid dysfunction in Irish girls with TS, the potential risks and benefits of thyroid disorder screening should be discussed with young people with TS and / or their families. Our experience underlines the importance of monitoring for thyroid function in patients with TS.

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References