Case Report

Association of turner syndrome with hypothyroidism

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To create awareness about association of turner syndrome with hypothyroidism. TS affects 1/2,500 female births and is due to the absence/structurally abnormal sex chromosome and characterized by short stature and oestrogen deficiency, secondary to ovarian dysgenesis. Approximately $^1/_3$ has a thyroid disorder, usually hypothyroidism. In most cases, it is caused by an immune system attack (Hashimoto's thyroiditis). To create awareness about association of turner syndrome with hypothyroidism. A 14 years old female presented with short stature, primary amenorrhea, non-development of breast, webbed neck, no pubic hair with sexual infantilism with disinterest in school. Due to this phenotype, turner syndrome was suspected. There was fatigue, exhaustion, feeling cold, constipation, muscle cramps, dry, coarse, itchy skin, thinning hair. Diagnosis is confirmed by the presence of a 45 X cell line. Thyroiditis with hypothyroidism is confirmed by presence of elevated TSH 17.22 ulU/ml (0.35-5.50), normal T3 & T4 and elevated anti-TPO. Atria *et al*[1] 1948 reported the post-mortem findings of a small thyroid gland with lymphocytic infiltration in a young TS woman. The association has since been confirmed in TS with a high incidence of Hashimoto's disease and elevated thyroid antibodies[2-8]. S Livadas *et al*[9] analysed 84 girls with TS and Hypothyroidism was detected in 24% and hyperthyroidism in 2.5%. There is association of turner syndrome with thyroid disorder, usually hypothyroidism.

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Key words **: Turner syndrome, hypothyroidism, thyroiditis.**

Turner syndrome (TS) affects approximately 1 out of every 2,500 female live births worldwide. It embraces a broad spectrum of features, from major heart defects to minor cosmetic issues. Turner syndrome is due to the absence of a sex chromosome or the presence of a structurally abnormal sex chromosome in a phenotypic female. It is characterized by short stature and oestrogen deficiency, secondary to ovarian dysgenesis. Approximately $^1/_3$ of individuals with Turner syndrome have a thyroid disorder, usually hypothyroidism. Symptoms of this condition include decreased energy, dry skin, cold-intolerance and poor growth. In most cases, it is caused by an immune system attack on the thyroid gland (also known as Hashimoto's thyroiditis). This case report is presented here to create awareness about association of turner syndrome with hypothyroidism (Fig 1).

Case Report

A fourteen years old female presented with short stature, primary amenorrhea, non-development of breast, webbed neck, no pubic hair with sexual infantilism & disinterest in school studies. Due to this phenotype, turner syndrome was suspected. There was fatigue, exhaustion, feeling cold, especially in the extremities, constipation, muscle cramps, dry, coarse, itchy skin, thinning hair. Diagnosis is confirmed by the presence of a 45 X cell line. Thyroiditis with hypothyroidism is confirmed by presence of elevated TSH 17.22 uIU/ml (0.35-5.50), normal T3 1.00 ng/ml (0.60-1.81) & T4 8.00 ug/dl (5.01-12.45) and anti-thyroglobulin (anti-TG) and anti-thyroid peroxidase (anti-TPO) antibodies were measured using chemiluminescent immunometric assay method and they were 72 (normal range 0 to 60 IU/ml) in the serum.

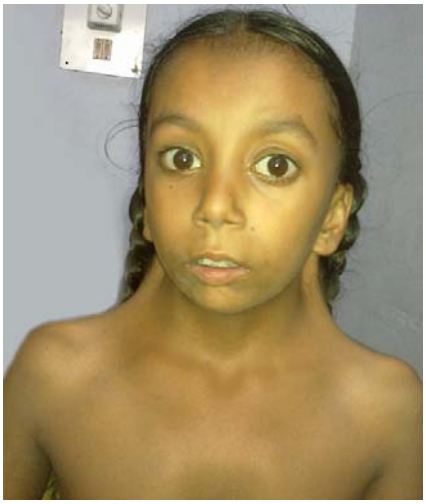

Fig 1 — Showing turner syndrome with hypothyroidism

Discussion

The relationship between thyroid disease and TS was first suggested by Atria *et al*[1] in 1948 when they reported the post-mortem findings of a small thyroid gland with lymphocytic infiltration in a young TS woman. The association has since been confirmed in TS and in gonadal failure with a high incidence of Hashimoto's disease and elevated thyroid antibodies[2-4]. A positive family history has been documented by Wilson *et al*[5] who found an elevated antibody titre in both TS patients and their mothers. Radetti G *et al*[6] studied 478 patients, mean age 15.5 (3.6-25.3) years, suffering from Turner's syndrome, in order to determine the frequency of autoimmune thyroiditis. They found 106 (22.2%) patients positive for antithyroid antibodies (AT-Ab) and of those 49 (10%) also had positive ultrasound findings, and were therefore considered to be affected by thyroiditis. This frequency was significantly higher ($p<0.001$) than that seen in the normal population. Chiovato L *et*

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al[7] searched for thyroid disease in patients with TS. Seventy-five unselected TS patients were studied. Sera were tested for thyroid hormones, thyrotropin (TSH), thyroglobulin and thyroperoxidase antibodies. Eight had autoimmune thyroiditis (AT) (six with subclinical and two with overt hypothyroidism and one with euthyroidism) and one had Graves' disease. During the period 1995-2003, 177 women, aged 16-71 year, with TS were studied at the university departments of Internal Medicine and Obstetrics and Gynecology in Göteborg and Malmö, Sweden[8]. These women were compared with randomly selected controls. At baseline, 15 (16%) TS women were treated for hypothyroidism, and elevated serum TSH was found in another eight (9%). As a result, hypothyroidism was more common in women with TS (25%) than in controls (2%; P<0.0001). After the 5-year follow-up, an additional 11 (16%) developed hypothyroidism, of whom four (36%) had elevated thyroid peroxidase. Altogether, 34 (37%) TS women had hypothyroidism after the 5-year follow-up. S Livadas *et al*[9] analysed 84 girls with TS attending their clinic. The mean age ± standard deviation (SD) at their initial evaluation was 10.3 ± 3.7 years (range, 0.5 to 19 years) and the mean period of observation was 8.4 ± 4.4 years. Hypothyroidism was detected in 24% of the studied subjects and hyperthyroidism in 2.5%. Elevated values of thyroid autoantibodies were detected in 42% of girls with TS. According to this study, thyroid function should be evaluated yearly in girls with TS past the age of 8 years and more frequently in those with positive thyroid auto antibodies.

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