ENDOCRINOLOGICAL EVALUATION OF GIRLS WITH TURNER SYNDROME ATTENDING ALEXANDRIA UNIVERSITY CHILDREN'S HOSPITAL

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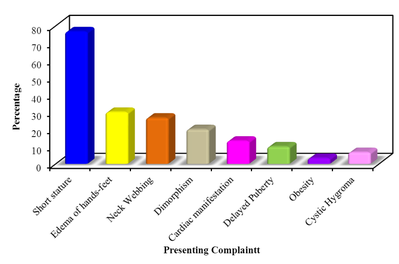
OBJECTIVES

Turner Syndrome (TS) is the consequence of complete or partial absence of one X chromosome in a phenotypic female. The genes involved in Turner phenotype are X-linked genes that escape inactivation. A major locus involved in the control of linear growth has been mapped within the Pseudo-Autosomal Region (PAR1) of the X chromosome. Our aim was to study some hormones with considerable effect on TS and their relation to the genotype of TS.

METHODS

Thirty girls with Turner Syndrome attending Endocrine Clinic of Alexandria University Children's Hospital were subjected to detailed history, clinical examination, karyotyping, and hormonal investigation including luteinizing hormone (LH), follicle stimulating hormone (FSH), estrogen (E2), anti- mullerian hormone (AMH), insulin-like growth factor1 (IGF-1), growth hormone (GH) and thyroid function tests. Bone age & pelvic ultrasound were done.



RESULTS

Age ranged from 3 to 18 years with a mean of 10.98± 4.85 years. Monosomy (45, XO) was the predominant genotype (40.0%) followed by mosaic genotype (36.7%), while isochromosome (46, Xi Xq) was the least (23.3%). Most cases (76.7%) presented with short stature which was significantly more common among mosaic genotype. (p =0.048). Monosomy (45, XO) was the predominant genotype (40.0%). Ultrasound of the ovaries revealed abnormalities in 70% of cases as streak gonads in and infantile ovary, while of the uterus showed hypoplastic form in 56.7% of cases. . LH, FSH, AMH were high in 11 cases (73.3%). FT4 and TSH levels were normal in all studied patients while Anti –TPO (Thyriod peroxidase antibodies) was high in 8 cases (26.7%). No statistically significant differences were found between genotypes regarding hormonal assessment.

CONCLUSIONS

The most common genotypes of Turner Syndrome in the current study are monosomy 45 XO and the mosaicism. Majority of the cases have streak gonads or infantile ovaries with high levels of gonadotropins, AMH and Anti –TPO. Their thyroid functions were normal

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