AUDITING PRESENTATION, INVESTIGATIONS AND MANAGEMENT OF TURNER'S SYNDROME

P3-P227

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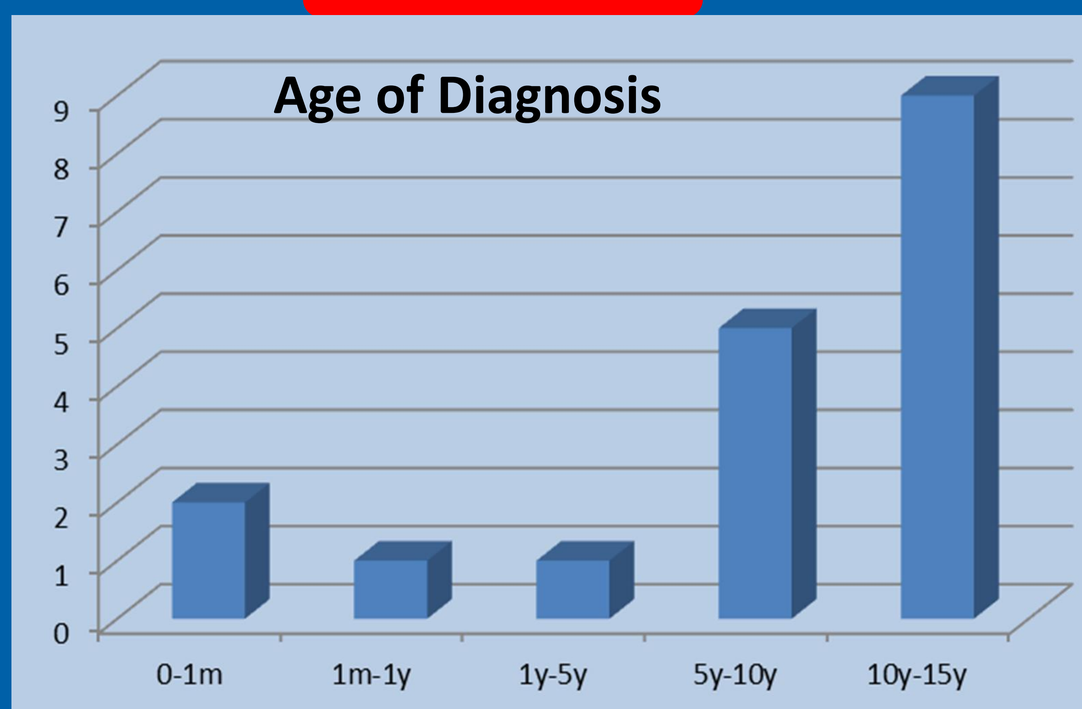
Introduction and Objectives

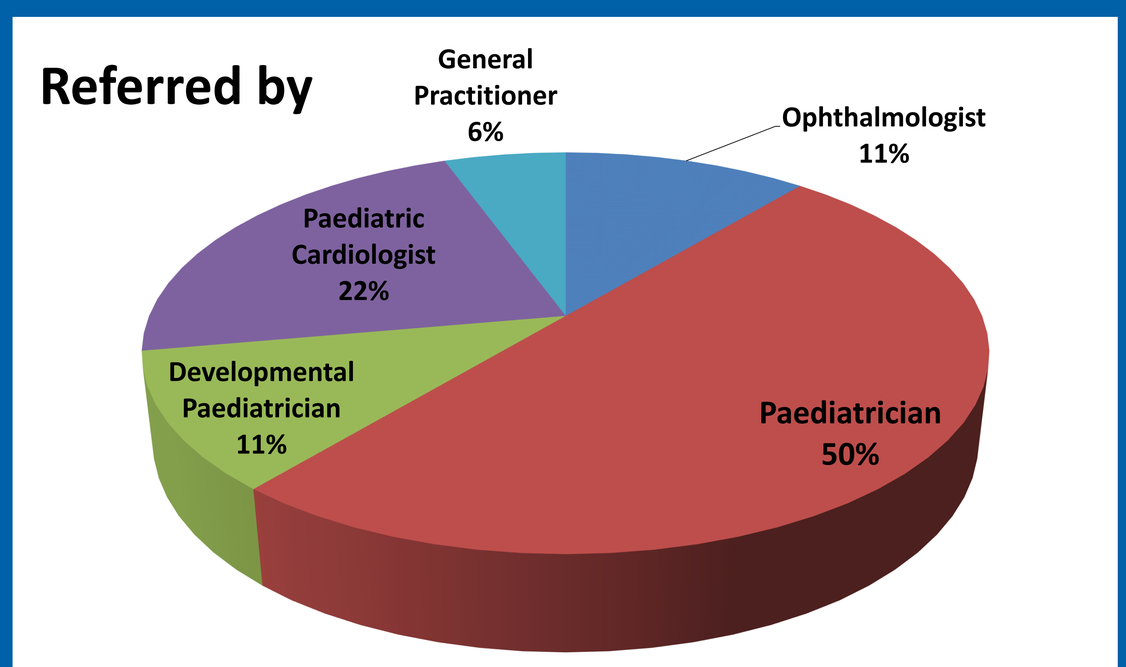
Turner's syndrome (TS) is the most common genetic disorder in females affecting approximately 1 in 2500 live female births, as a result of partial or complete X chromosomal monosomy. Age at diagnosis is important to start growth hormone at younger age to attain optimal adult height and to decide on hormone replacement therapy(HRT) .This audit evaluated trends in presentation, age at diagnosis and investigation findings of children with TS at presentation and overall surveillance.

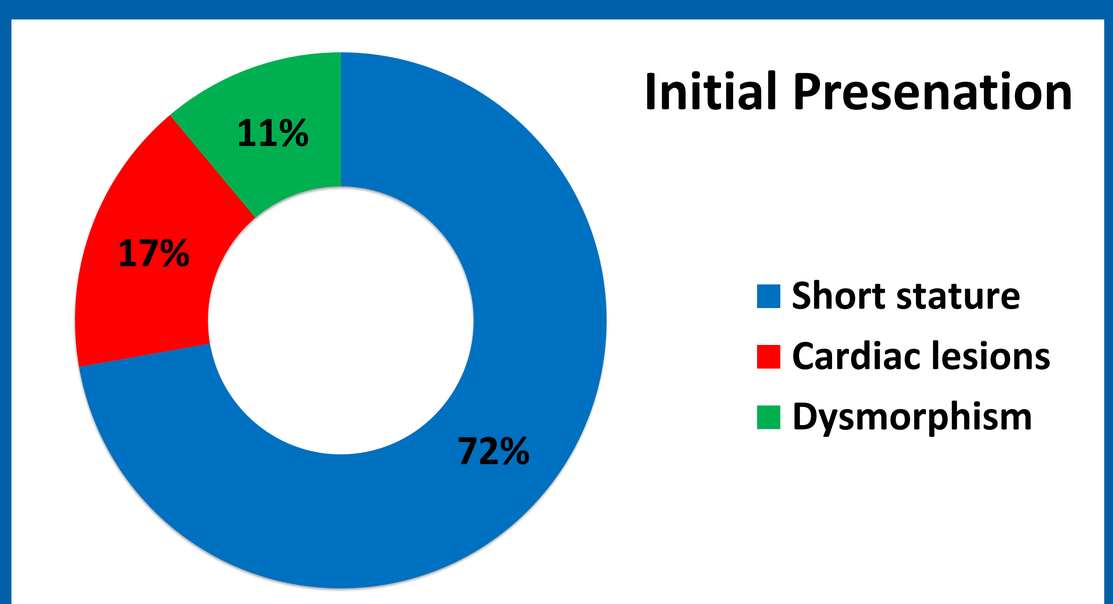
Method

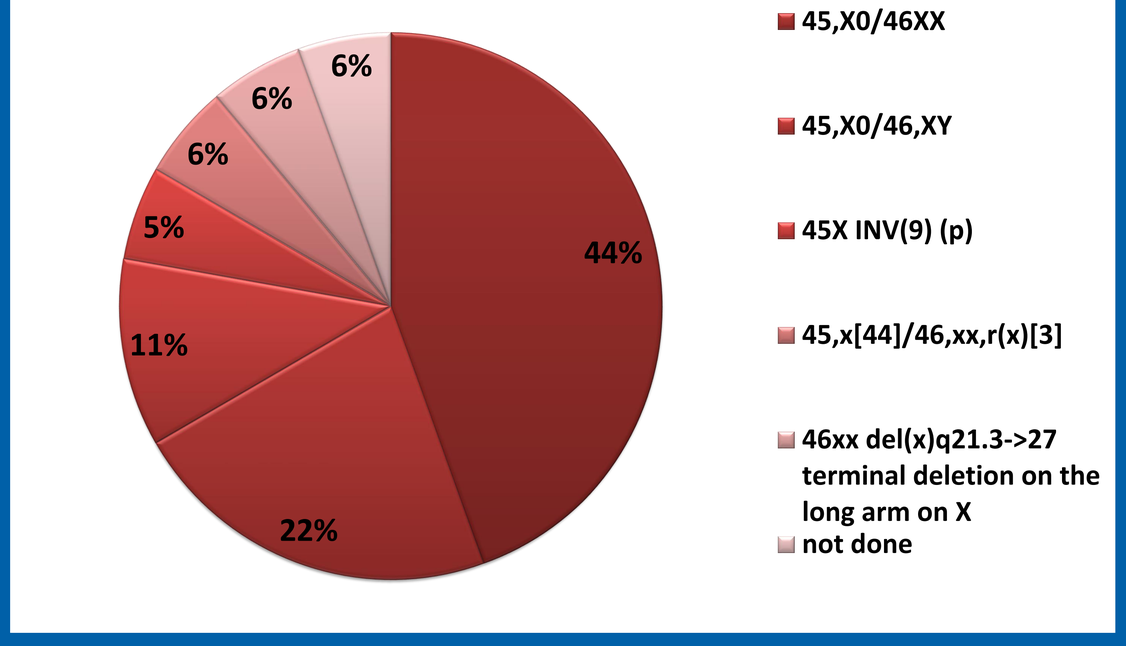
Retrospective data analysis carried out on children diagnosed with Turner syndrome and followed up in Paediatric Endocrinology Clinic in Tertiary care Hospital for Children. Patient data (n=18) were analysed to evaluate trends in diagnosis and to audit clinical presentation, investigations and management.

Results



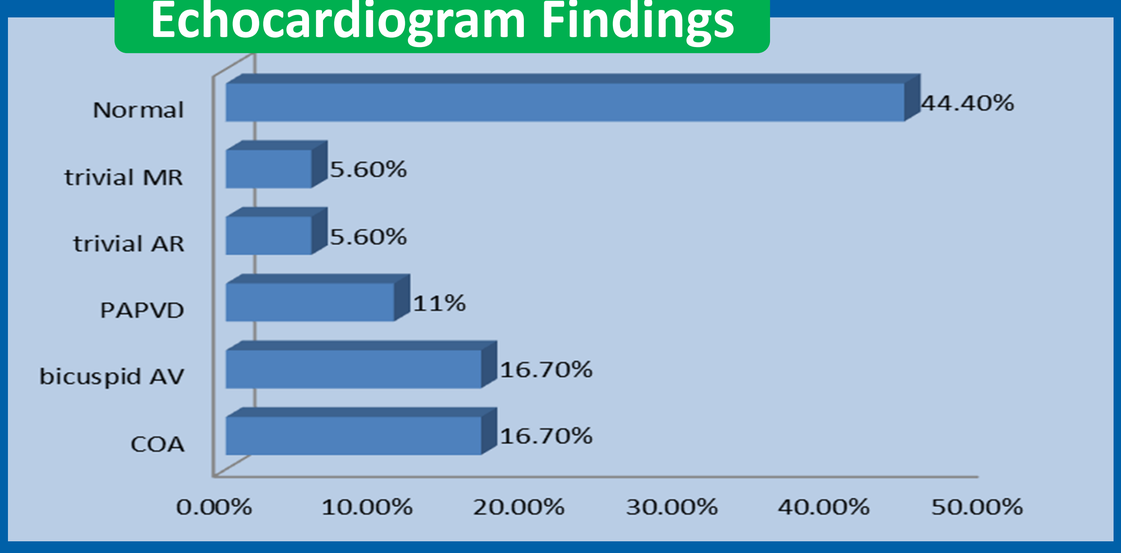






Ultrasound scan findings

Majority had small uterus and atretic ovaries and only two girls (11%) had renal anomalies



Management

- 72 % of them on growth hormone therapy.
- 16% on hormone replacement therapy
- Two of them underwent bilateral gonadectomy as they carry Y chromosome material.
- Abnormal thyroid functions found in 22% and currently on thyroxine.
- Screening for hearing detected 22% of girls with mild to moderate hearing impairment
- Ophthalmologic assessment found two girls(11%) with strabismus and two (11%) with refractory errors.

Conclusion

This study shows delay in diagnosing and referring patients with TS commonly after 11years of age which limit growth hormone therapy and delay in detecting other system involvement.