Mode of Clinical Presentation and Delayed Diagnosis of Turner Syndrome

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Background

Early diagnosis of girls with Turner syndrome (TS) is essential to provide timely intervention and support. Based on the findings by Savendahl and Davenport[1], the proposed guidelines for screening girls for TS were used (Table1).

Table 1: Guidelines for screening for TS[1]

Any girl with one or more of the following*:

1. **Unexplained short stature (height <5th percentile)**
2. Webbed neck
3. Peripheral lymphoedema
4. Coarctation of the aorta
5. Delayed puberty

OR

Any girl who has at least two or more of the following:

1. Nail dysplasia
2. High arched palate
3. Short 4th metacarpal
4. Strabismus

* Other suggestive features include: non-verbal learning disability, epicanthial folds, ptosis, cubitus vagus, multiple naevi, renal malformations, bicuspid AV, recurrent OM and need for glasses

Aim

The aim of the study was to determine the age and clinical features at the time of presentation to identify potential delays in diagnosis of TS.

Method

Retrospective data collection on age at diagnosis, reason for karyotype analysis and presenting clinical features was collected from the medical records of 67 girls with TS.

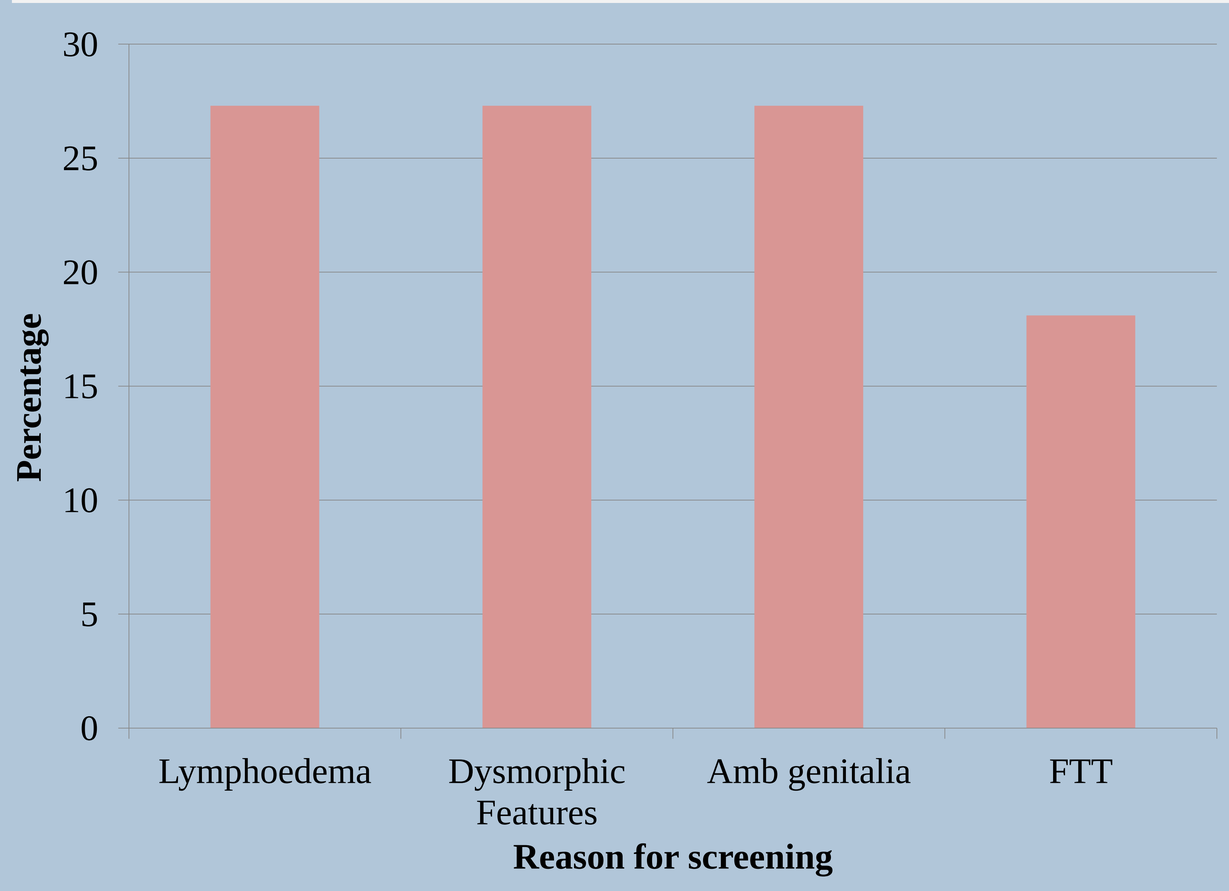

Figure 1: Initial clinical presentation for patients diagnosed during infancy (under 1 year of age)

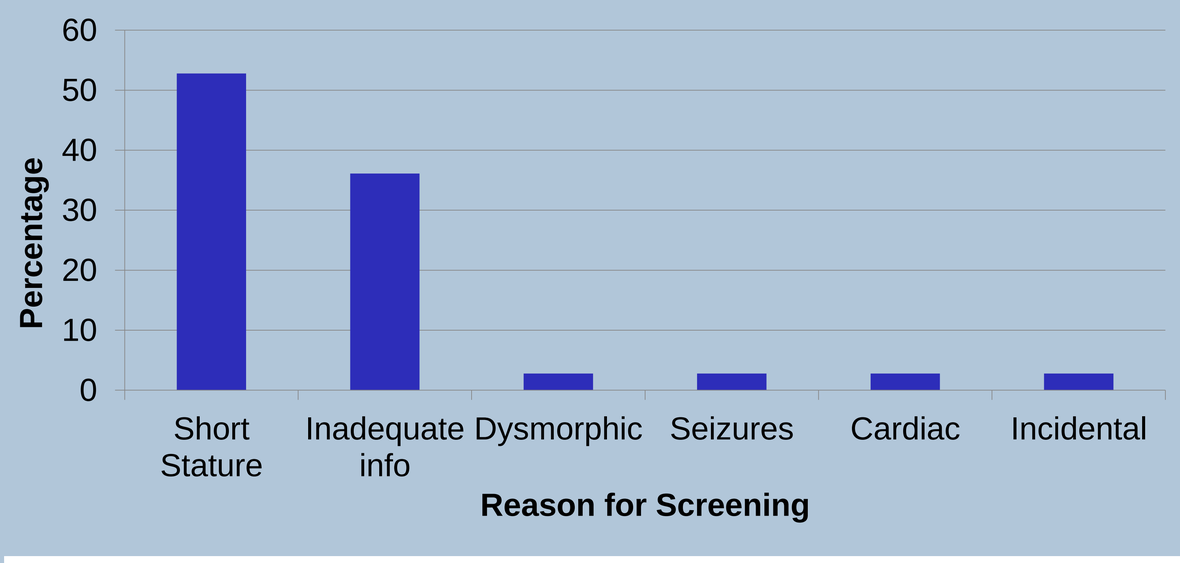

Figure 2: Initial clinical presentation for patients diagnosed in their childhood years (aged 1-12 years)

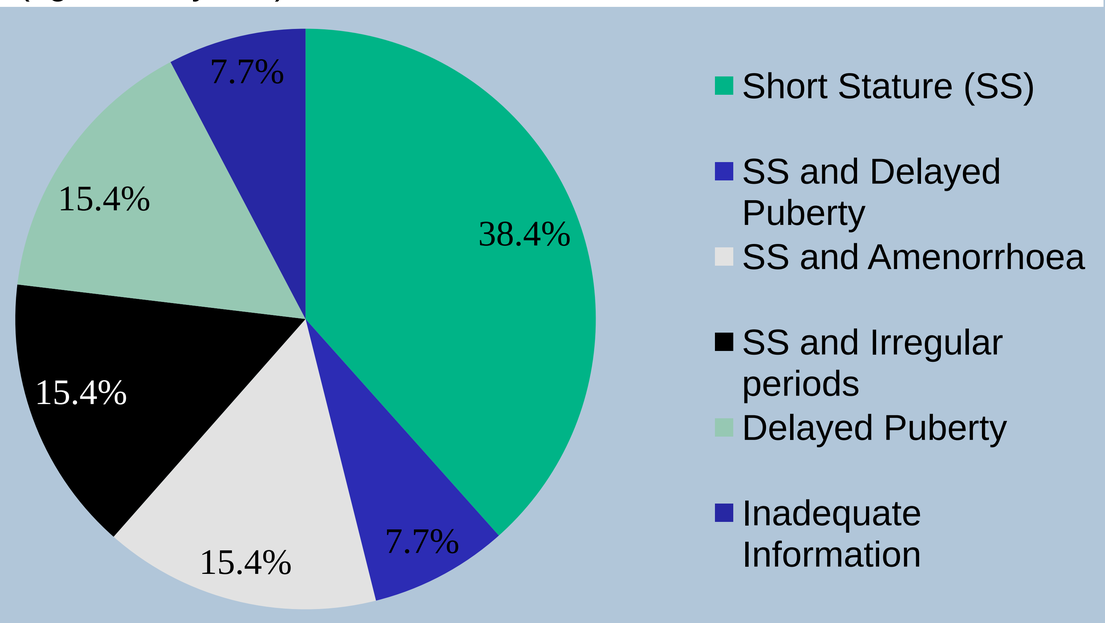

Figure 3: Initial clinical presentation for patients diagnosed in their adolescent years (aged 12-18 years)

Results

- Mean age of diagnosis was 5.89 (±5.3) years, ranging from pre-natal to 17.9 years.
- Overall only 42% of the girls were diagnosed before the age of 5 years.
- 10% were diagnosed antenatally (29% secondary to maternal age).
- 16% of the patients were diagnosed in infancy (Figure 1).
- 54% were diagnosed in childhood (Figure 2). 6 of the 36 children should have been screened at birth if using the screening guidelines.
- Finally, 20% were diagnosed between the ages of 12 and 18 years old (Figure 3). 2 of these adolescents should have been screened at birth if using the proposed guidelines.
- At least 12% of girls fulfilled the criteria for earlier screening but were diagnosed only at a later age (mean age= 8.78 years).
- The actual duration of delay in children presenting with short stature could not be ascertained due to lack of height measurements prior to seeking specialist opinion.

Conclusions

- The majority of girls with TS were diagnosed only after the age of 5 years.
- Short stature triggered evaluation for most patients diagnosed in childhood and adolescence.
- Lack of community height-screening programme and lack of awareness could have led to potential delays in diagnosing TS.
- New strategies for earlier detection of TS are needed.

References

1. Sävendahl L and Davenport M. Delayed diagnoses of Turner's syndrome: Proposed guidelines for change. *Journal of Paediatrics*. 2000; 455-59.