

COMORBIDITIES IN TURNER SYNDROME PATIENTS CONTROLLED IN OUR CENTER SINCE THE 80'S

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INTRODUCTION

Turner syndrome (TS) is a genetic condition with different phenotypic expressions depending on karyotype.

Due to **genetic prenatal testing**, its prevalence is getting lower.

AIM

The **objective** was to analyze the presence of different comorbidities associated with TS according to the karyotype and evaluate if there is follow-up in adulthood.

METHOD

Descriptive retrospective study including patients genetically diagnosed with Turner syndrome between 1984 to 2019.

Clinical and follow-up variables

Patients classified according to karyotype (monosomy X/mosaicism/isochromosome)

Follow-up to adulthood, gynecologic screening and pregnancy registered

RESULTS

* 70 patients

- ** 22 prenatal diagnosis (aborted)
- ** 48 postnatal diagnosis → 38 collected patients

* Mean age: 32.6 years old (SD 17.7)

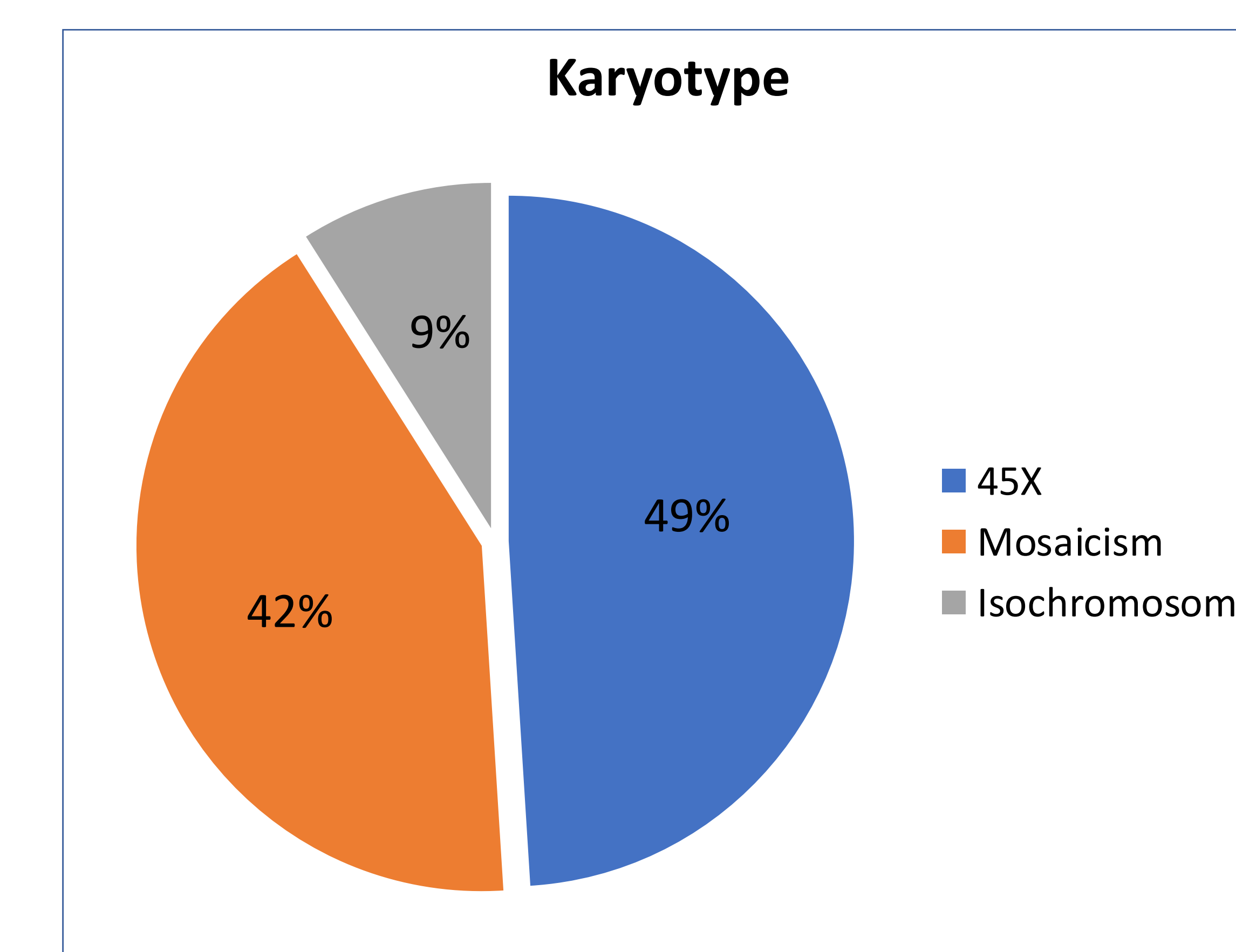
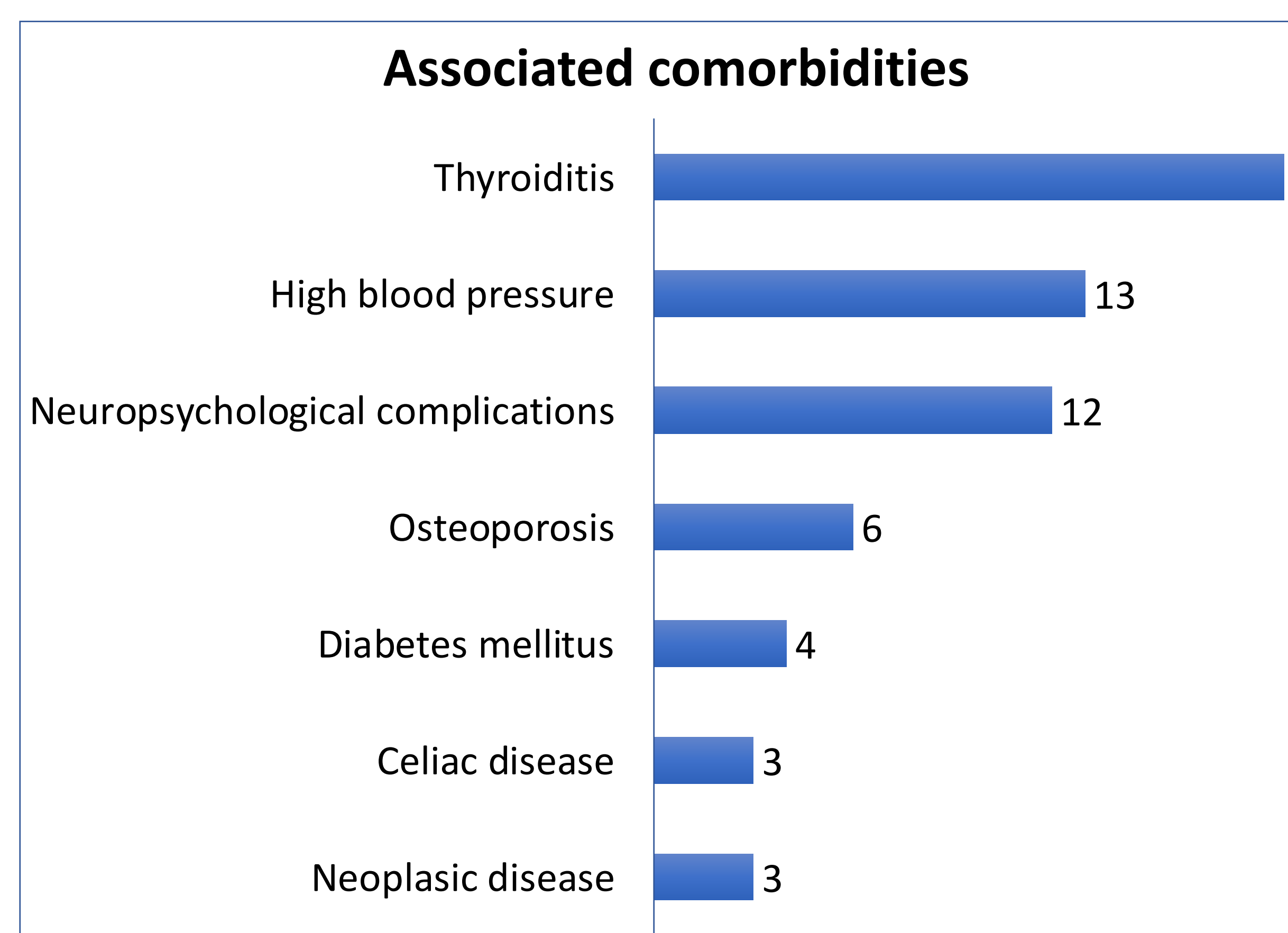
* Prescribed treatment

- ** Growth hormone: 50% of cases (89% of them were born in the last 25 years)
- ** Hormonal replacement therapy: 73% of cases

* Echocardiographic screening: 81.6% of patients (3 cases with aortic root dilatation, 4 with bicuspid aortic valve and 3 with coarctation of the aorta)

* Renal screening: 81.5% of patients (3 cases with renal fusion and 1 case of abnormal renal rotation)

- **Gynaecologic screening:** 63.2% of patients (7.9% achieved pregnancy)
- **Transition from paediatric to adult healthcare:** 52.6% of patients older than 18 years old



• Autoimmune disorders according to karyotype:

- ** Isochromosome: 100% of patients
- ** Monosomy X: 50% of patients
- ** Mosaicism: 60% of patients

CONCLUSIONS

Comorbidities are the main issue in follow-up of TS as they affect most part of the patients

It is important to analyze comorbidities depending on the **karyotype**.

Taking into account prognostic implication of **cardiologic and renal disease**, regular screening is mandatory in all patients.

Since most patients do not have follow-up in adulthood, it is important to take special care on **transition from paediatric to adult healthcare**.

REFERENCES

Gravholt CH et al, on behalf of the International Turner Syndrome Consensus Group. *Clinical practice guidelines for the care of girls and women with Turner syndrome: proceedings from the 2016 Cincinnati International Turner Syndrome Meeting*. Eur. J. Endocrinol. 2017 Sep; 177(3) 1-70.

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