CLINICAL FEATURES AND GENETIC CONSIDERATIONS OF TURNER SYNDROME: A REVIEW OF OUR CASES

Berrade S[1], Chueca M[1], Zarikian S[1], Mosquera A[1], Ulibarrena N[1], García C[1], Oyarzabal M[1].
[1]Pediatric Endocrinology Departament. Complejo Hospitalario de Navarra. Pamplona. Spain

Introduction

- Turner syndrome (TS) involves a partial or complete loss of an chromosome.
- TS patients have an increased susceptibility to various disorders.

Objetive

To describe the clinical presentation, genotype and folw-up of TS patients controlled in the Pediatric Endocrinology departament of our hospital.

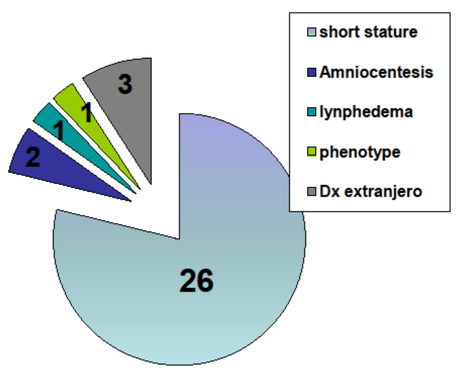
Material and Method

Retrospective study of patients diagnosed with TS at the Navarra Hospital between 1980-2014. Review of medical records.

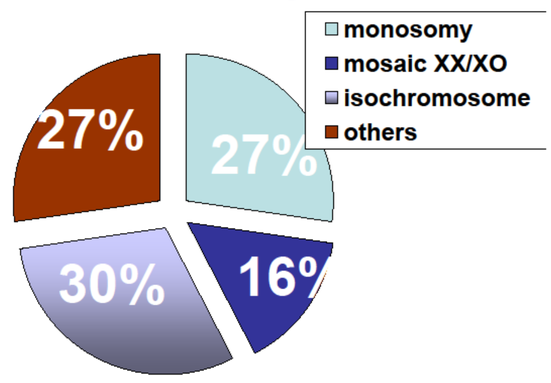
Results

- **Period:** 1980-2014
- **Cases:** *33*
- **Actual mean age:** *22.2 year* (6-47)
- **Age at diagnosis:** *7±3.8* (0-13.5 years)
- **Foreign origin:** 9 cases

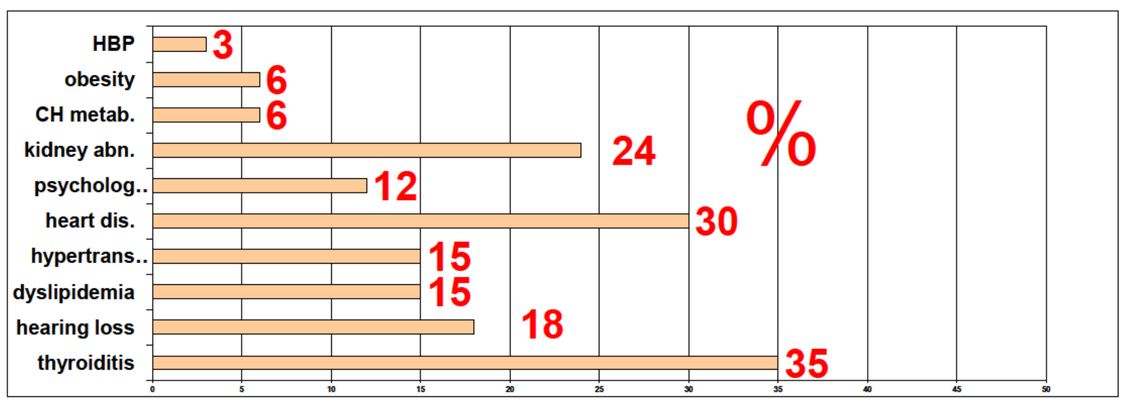
Reason for consultation



Genetic analysis



Associated pathology

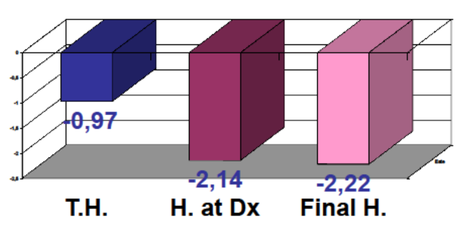


Molecular genetics	
Monosomy XO (9)	45X
Isochromosomes(10)	45X, 46Xi(Xq); 46Xi(Xq)
Mosaic 46XX (5)	45X, 46XX
Complex mosaicism (5)	45X, 46XX, 47XXX
Mosaic ring X (2)	45X, 46Xr(X)
Mosaic 46XY (1)	45X, 46XY
Complex reorganizations (1)	45X, 46X+mar; XO, XX, XXp-

- **Other associated malformations:** congenital hip dislocation (2 cases), cleft lip (1), congenital deafness (1), anal atresia (1), celiac disease (1)
- **1 case death** by dissecting aortic aneurysm

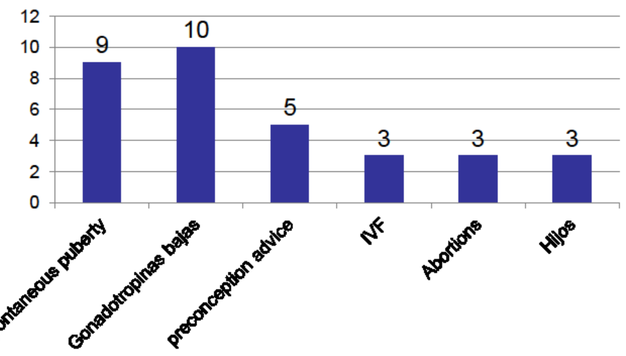
Adult women (21 cases)

Final height



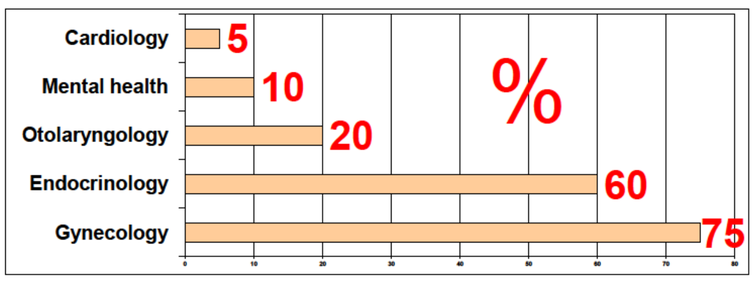
85% received treatment with GH. Mean final stature: 149.4 ± 5.1 cm (138.7-159.3), that means a loss of height regarding its T.H. **-1,25 ± 0,79** (-2,76, +0,59)

Puberty and fertility



The 43% (9 cases) presented spontaneous puberty, 5 women requested for preconception advice, FIV was done in 3 with a result of 5 pregnancies (3 abortions y 2 completed pregnancies) and 1 had one spontaneous pregnancy.

Medical follow-up



The majority (a 89%) carries out analytical controls, but only the 36% has a echocardiography made in the last 5 years and the 26% has a densitometry.

Conclusions

- ❑ The chief complaint that led to the diagnosis of TS was short stature.
- ❑ Genetic analysis reveals a variety of karyotypes, highlighting the presence of monosomy XO and isochromosomes.
- ❑ It is imperative an adequate multidisciplinary follow-up in adults units, to ensure proper screening and management of major complications.

ESPE 2015 1237--P3
Misc 1
Sara Berrade
DOI: 10.3252/pso.eu.54espe.2015
Poster presented at: 54th Annual Meeting 1-3 October ESPE 2015 BARCELONA European Society for Paediatric Endocrinology
PosterSessionOnline.com