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

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Introduction

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

Objective

To describe the clinical presentation, genotype and follow-up of TS patients controlled in the Pediatric Endocrinology department 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

**Associated pathology**

- Obesity
- Cleft lip
- Congenital deafness
- Anal atresia
- Celiac disease

- **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

**Molecular genetics**

- Monosomy XO (9) 45X
- Isochromosomes (10) 45X, 46X(iXq); 46X(iXq)
- Mosaic 46XX (5) 45X, 46X
- 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-

**Adult women (21 cases)**

- 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)

**Medical follow-up**

- The majority (a 89%) carries out analytical controls, but only the 36% has an 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.