



ENDOCRINE DISORDERS IN WOMEN WITH TURNER SYNDROME

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INTRODUCTION: Turner syndrome (TS) is the most common chromosomal abnormality in women. It occurs in 1/2,500 to 1/3,000 live-born females and results from a **total or partial absence of the X chromosome**. The clinical manifestations are diverse and TS is accompanied by multiple medical problems.

METHODS: We analyzed retrospectively **6 cases of women with TS aged 20-66 years**, treated at Endocrinology Department between 2003 and 2014, to describe endocrine disorders in such patients. In the studied subjects, the diagnosis of TS was made in childhood (1 patient), in adolescence (3 cases) or in adulthood (2 women), based on clinical symptoms as well as genetic tests.

RESULTS:

3 women with short stature received **recombinant human growth hormone**. The average height was 152.5 ± 6.53 cm. None of patients were obese (BMI: 24.12 ± 2.88 kg/m²). Sex **hormone replacement therapy** was used by 4 women, 2 the oldest non-treated patients suffered from **osteoporosis**. Subjects with hypothyroidism due to Hashimoto's thyroiditis used **levothyroxine**. Patient with **type 1 diabetes** had many diabetic complications such as: coronary heart disease, cerebrovascular disease, peripheral vascular disease, sensory neuropathy, diabetic amyotrophy, glaucoma, cataract.

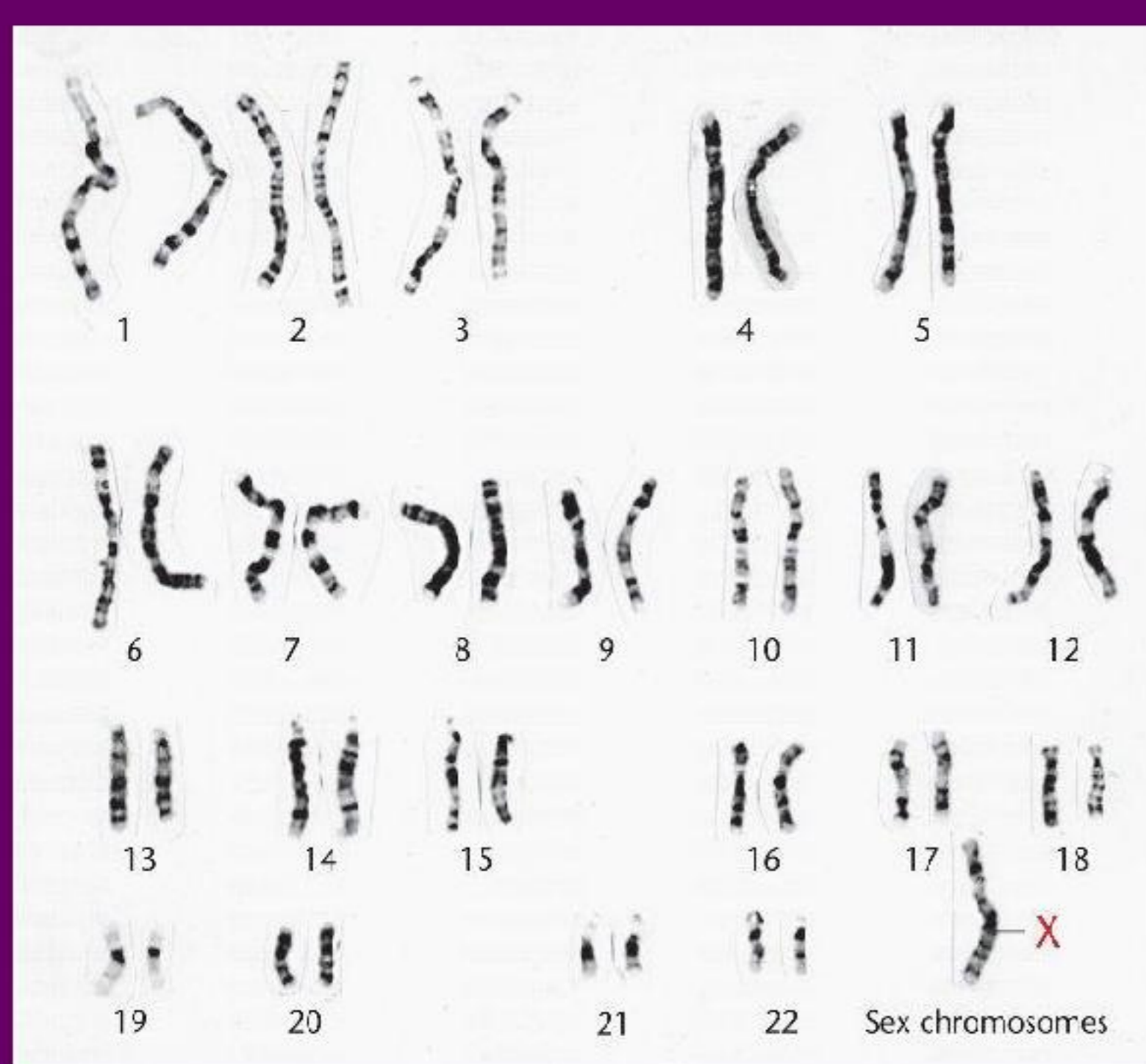


Fig. 1. Karyotype of Turner syndrome
(Ref. www.geoset.info)

| ENDOCRINE DISORDERS in TS | NUMBER of PATIENTS (n=6) |
|---|--------------------------|
| • short stature | 6 |
| • primary amenorrhea | 5 |
| • premature ovarian insufficiency | 1 |
| • clitoromegaly | 1 |
| • leukoplakia vulvae | 1 |
| • hypothyroidism due to Hashimoto's thyroiditis | 5 |
| • diabetes mellitus (type1 and type 2) | 2 |
| • non-functioning pituitary adenoma | 1 |
| • osteoporosis | 2 |

Moreover other **non-endocrine diseases** were observed, including: **coeliac disease** (2 cases), **hypertension** (3 patients), **mitral incompetence and tricuspid regurgitation** (1 woman), **impaired hearing** (2 subjects), **ectopic or horseshoe kidney** (2 cases).

The **intellectual performance** was within the normal range in 4 women, 2 patients were mentally retarded.

CONCLUSIONS: Most of subjects with TS have hormonal disorders. Therefore, **endocrine care is needed to optimize patients' treatment**.

