

# Turner Syndrome

## Undiagnosed until adulthood

### - clinical case report

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3<sup>rd</sup> EYES MEETING 2015  
Modena, Italy  
24<sup>th</sup> - 26<sup>th</sup> september 2015

# INTRODUCTION – TURNER SYNDROME

First reference by Morgagni in 1748  
- In 1938, described by Henry Turner

Loss of part or all of an **X chromosome**



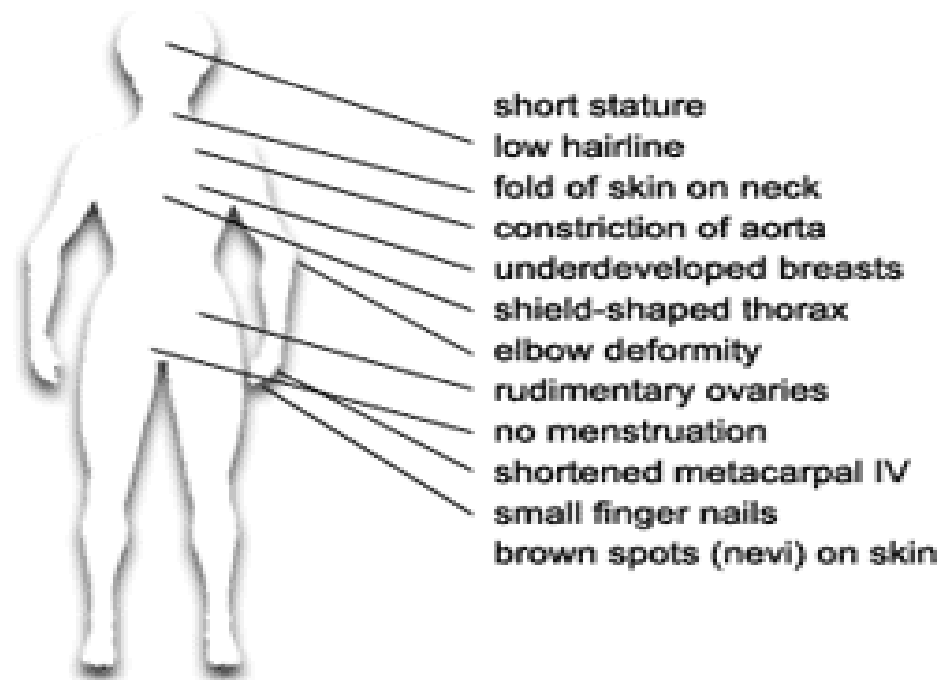
Henry H. Turner

**Most common** sex chromosome abnormality in females.

Occurs in approximately **1 in 2.500 live births.**

# INTRODUCTION – TURNER SYNDROME

- The most characteristic features are: **short stature** and **primary amenorrhea** in young woman.
- Other features:



# CLINICAL CASE

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- 65 year old woman, factory worker.
- Referred to Endocrinology's outpatient clinic by her new general physician because of short stature and primary amenorrhea.

# CLINICAL CASE

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- **Personal medical history:**
  - DM2, diagnosed 20 years ago, hypertension, hypoacusia (use of hearing aid) and hypothyroidism.
- **Usual medication:**
  - Sitagliptin/Metformin 50/1000 mg bid, Lisinopril 10 mg and Levothyroxine 75 mg.
- No relevant family history.

# CLINICAL CASE

- Physical examination:

Height – 134 cm

Weight – 40,5 kg

BP – 127/78 mm hg

No thelarhe. No nevi.

Cardiac A. – s1+s2 rhythmic

Pulmonary A. – Norma



# CLINICAL CASE

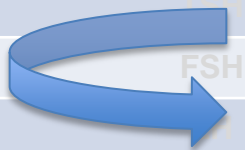
	VALUE	REFERENCE VALUE
Hemoglobin	12.9	11.9-15.6 g/dL
White blood count	8.4x10 <sup>3</sup>	4.0-11.0x10 <sup>3</sup> /uL
Platelets	241x10 <sup>3</sup>	150-400x10 <sup>3</sup> /uL
Creatinine	0.60	0.60-1.20 mg/dL
AST (GOT)	14	15-37 U/L
ALT (TGP)	27	12-78 U/L
Cholesterol - Total	188	< 200 mg/dL
Cholesterol - HDL	58	> 45 mg/dL
Triglycerides	99	< 150 mg/dL
Hba1c	10,0	3.4-5.8 %
TSH	1.880	0.358-3.74 uUI/mL
FSH	31.51	23.0-116.3 mUI/mL
LH	7.18	15.9-54.0 mUI/mL
Estradiol	< 43.31	< 118.2 pmol/L
DHEA-SO4	138.00	35-430 ug/dL
4-Androstenediona	1.01	0.3-3.3 ng/mL
Testosterone	12.90	14-76 ng/dL
HLA DQ2 / DQ8	Negative	

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Intensified treatment with Insulin

# CLINICAL CASE

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**Pelvic ecography** revealed an uterus of small dimensions and the ovaries were not identified.

**Bone densitometry** showed accentuated osteoporosis (T-score - 2.9 and Z-score -1,2)



started alendronic acid+colecalciferol 70 mg/5600U.

**ECG** and **echocardiogram** were normal.

**Renal ultrasound** with no abnormalities.

# CLINICAL CASE

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- Karyotype revealed mosaicism 45,X/46,XX

# DISCUSSION

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- Higher overall mortality, up to a threefold risk increase compared with the general population.
- Yearly follow-up: physical examination, biochemical testing, screening for hearing loss and cardiovascular abnormalities.



**THANK YOU.**