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A Rare Form Genetic Abnormality in Turner Syndrome

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♀, 17.9 y.o. at presentation
No history of menses
No secondary sexual characteristics

PERSONAL PAST HISTORY
Born premature (28 w) , AGA
No significant past history
No recent changes in weight

FAMILY HISTORY
History of delayed puberty
(mother: menarche 16 y.o.)
father's growth spurt 14 y.o.

Examination

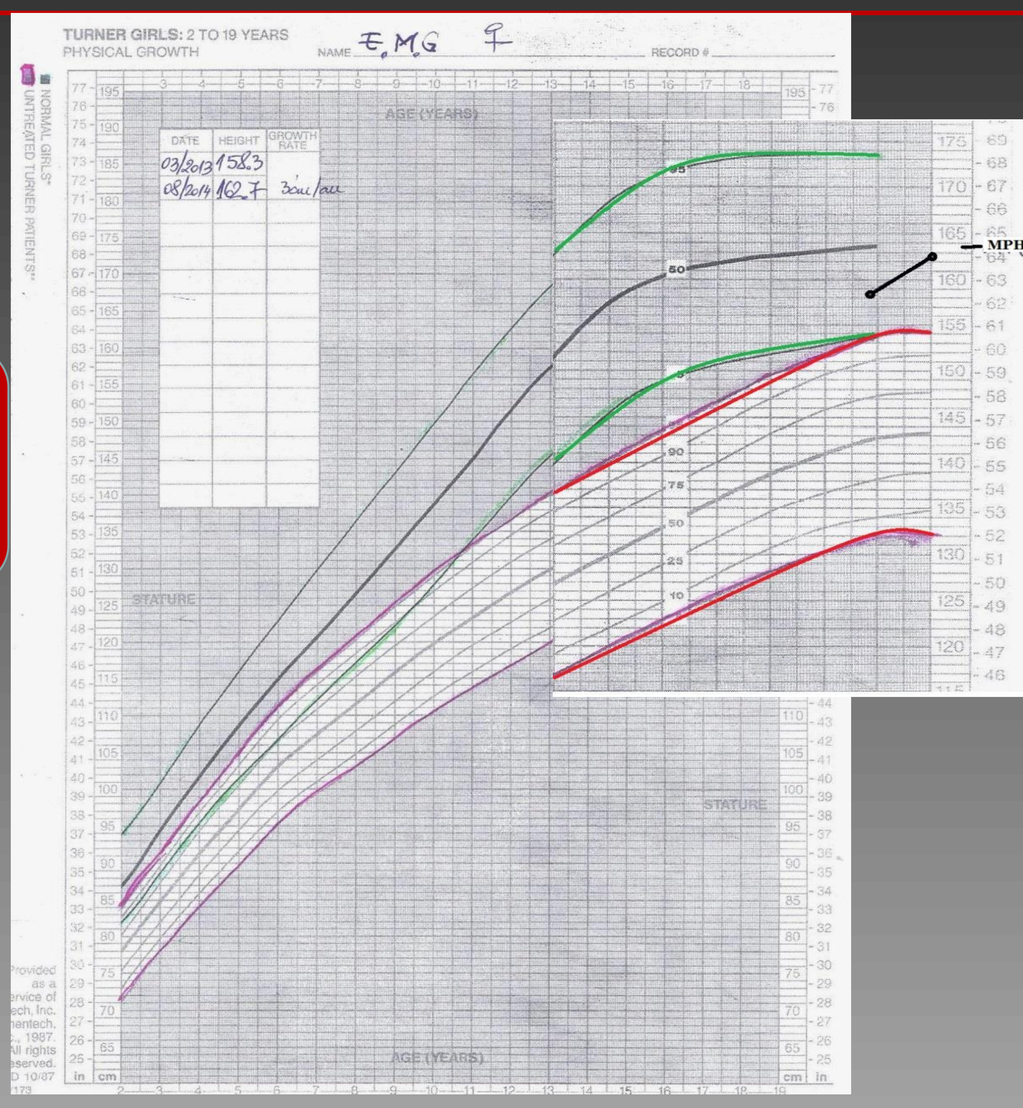
- ✓ Height 158.3 cm (-1.05 SD), MPH=161.5 cm. (SD= -0.75 DS)
- ✓ W= 51.3 kg., BMI= 19.38 kg/m^2
- ✓ low-set ears, widely spaced nipples, cubitus valgus
- ✓ BP=110/60 mmHg- equal in both arms,
- ✓ Female external genitalia Tanner stage B1P2(A2)

What's first?
Initial approach

gonadotropin status and skeletal age

Bone Age Greulich &Pyle: 15 years

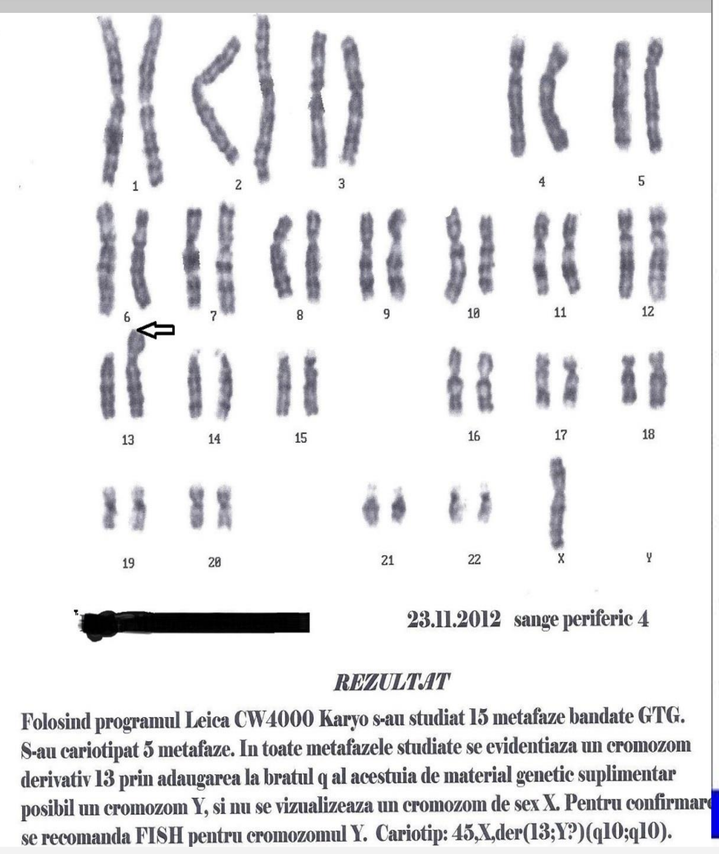
Parameter	value	normal range
FSH	65.39 mIU/mL	3.5-12.5
LH	44.05 mIU/mL	2.4-12.6
E2	8.43 (pg/mL)	43.8-211
testosterone	0.31 ng/mL	0.1-0.75
AMH	0.08	
PRL	16.37 ng/mL	4.79-23.3
fT4	11.64 pmoL/L	12-22
TSH	1.55	0.5-4.5
ATPO	<10 UI/mL	<35

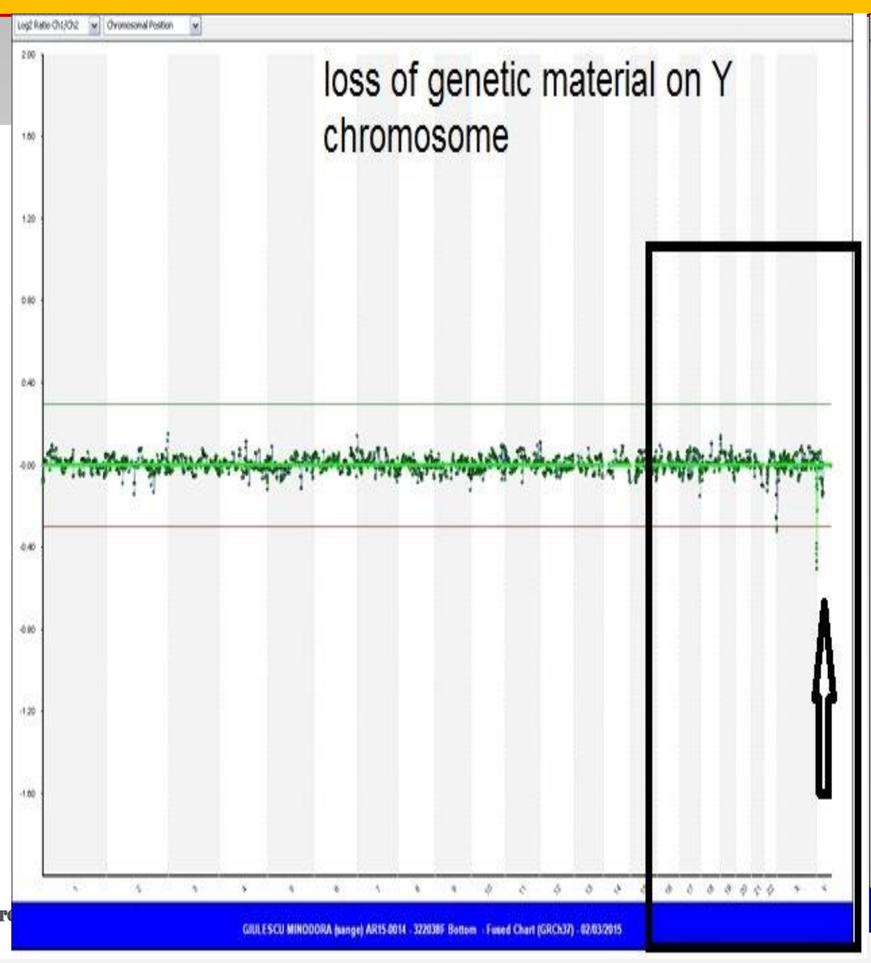


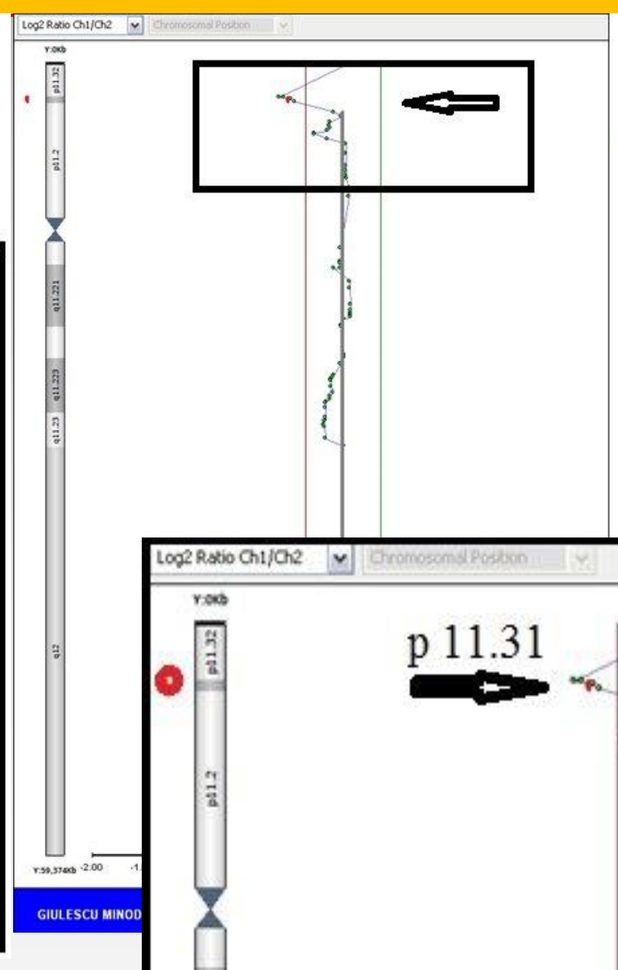
Pelvic US : small uterus 26 / 5.5 mm, undifferentiated endometrium, negative for gonads or pelvic tumors.
Abdominal CT: uterus 1.92 / 1.13 / 1.76 cm. (2 mL); no gonads or abdominal tumors; normal adrenals

Short stature is the most constant clinical finding in Turner Syndrome due to loss of one copy of SHOX gene (located Xp22.33 or Yp11.32)

Karyotype: Turner Syndrome 45X, der (13;Y)(q10;q10)







Y chromosome: complete deletion of p11.31 band

Array CGH - report

ISCN

ISCN	Size
arr Yp11.31(2,565,871-2,871,490x0, 3,025,075)	305,6

Detailed Region Summary

Type:	Assessment:
Position:	Chr24: 2,565,871 -> 2,871,490
Band:	Yp11.31 -> Yp11.31
Size:	305.6 Kb

arrYp11.31(2,565,871-2,871,490x0, 3,025,075)

From our knowledge this is the first report of Y-autosome translocation together with SRY deletion in a female phenotype. The classical cytogenetics showed Turner Syndrome together with Y chromosome material. Further investigations were performed because of her normal stature and female phenotype.

Y Ch. specific sequences in karyotype of girls with TS vary from 2% to 12%- depending on methodology applied
Risk of gonadoblastoma : up to 30% . Prophylactic gonadectomy is advisable. Genes implicated in gonadoblastoma: TSPY (Yp11.22), POU5F1 (OCT4: 6p21.31). SRY gene (Yp11.31) is not directly implicated in gonadoblastoma

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