A Rare Form Genetic Abnormality in Turner Syndrome

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PERSONAL PAST HISTORY
No history of menses
No secondary sexual characteristics
♀, 17.9 y.o. at presentation
No recent changes in weight

Examination
✓ Height 158.3 cm (-1.05 SD), MPH=161.5 cm. (SD= -0.75 DS)
✓ W=51.3 kg., BMI= 19.38 kg/m²
✓ low-set ears, widely spaced nipples, cubitus valgus
✓ BP=110/60 mmHg= equal in both arms.
✓ Female external genitalia Tanner stage B1P2(A2)

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

What’s first?
Initial approach
gonadotropin status and skeletal age

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)

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

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