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Unusual clinical manifestations in Turner syndrome

Akulevich N., Makarava Yu., Ershova L., Kunavitch I.
State Center of Medical Rehabilitation, Minsk, Belarus

57th ESPE 2018 Meeting ATHENS GREECE
27-29 September 2018

Background and Objective

Turner syndrome (TS) is characterized by partly or completely missing of an X chromosome and variability of clinical signs. Our objective is to present 3 Caucasian mosaic TS girls with unusual clinical course.

Methods

Retrospective analyses of the clinical records of TS patients treated by paediatric endocrinologists in our institution.

Results

Table 1 Characteristics of Turner syndrome patients

Feature	Case1 45,XO/46XX	Case2 45,XO/46XX	Case3 46,Xdel(x)(p21)
Age at the 1st examination	8 6/12	8 6/12	5 9/12
Time of follow-up	5,5 years	9,5 years	about 3 years
Reason for the 1st visit to paediatric endocrinologist	weight gain	short stature	short stature
Height (cm/ percentiles for TS) at 1st visit	128,5 cm/ more 97 p.c.	114 cm / 75 p.c.	115,6 cm /75 p.c.
Weight (kg/ percentiles for TS) at 1st visit	42 kg/ more 97 p.c.	20 kg/ 50 p.c.	20,4 kg/ 75 p.c.
UNUSUAL MANIFESTATIONS	spontaneous early puberty – menarche at 9 9/12 regular periods	at16 6/12 y. serous papillary cystic adenoma of the left ovary (surgically removed)	Familial TS – 4 women in 3 generations
Thyroid abnormalities	no	primary hypothyroidism, nodular thyroid lesions, benign cytology at FNBA	no
Cardiac abnormalities	minor	bicuspid aortic valve	minor
Kidney abnormalities	no	kidney duplicaion	no
GH treatment	no	yes (8 4/12 – 15 0/12),	Is under treatment
Laevothyroxine	no	yes	no
Estrogen replacement	never	never spontaneous menarche at 13 8/12.	no spontaneous telarche at 9 6/12
Metabolic problems	acanthosis nigricans, glucose intolerance, hyperinsulinemia	no	no
Coeliac disease	no	no	no
Final height (cm if achieved/ percentiles for TS)	142,5 cm/ 97 p.c.	152 cm/ more 97 p.c.	132 cm/ 97 p.c. (GV 9 cm/year)

Conclusion

Girls with TS may have normal functioning ovarian tissue, with approximately 30–40% entering puberty, but only about 1% being fertile. Our observations demonstrate a differences in mosaic TS clinical presentation and stress the need of a careful multi-disciplinary follow-up for such girls with a concern for their future health including fertility.

The authors have nothing to disclose

Correspondence for Dr. Natallia Akulevich: natamedical@mail.ru

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