A case of 45,X/47,XXX mosaic Turner syndrome: Clinical manifestation

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Case presentation:

A 12-years-and-5-months-old girl was brought to a pediatric endocrinology clinic to undergo evaluations for short stature and „absent menarche“

Medical history:

* the second child of healthy parents

* born naturally at full term with birth weight of 2.650 kg

* she had no other significant medical history and had achieved normal developmental milestones

* there was no familial history of inherited or congenital disease apart from the parental short stature (father 163.0 cm, mother 144.8 cm, **mid-parental height 147.4±7.5 cm**)

Physical and clinical examination

* ***Auxology:***
Height -124.9 cm **(–3.47 SDS)**
Weight -17.8 kg **(–6.03 of the SD)**

* ***Clinical examination*** :
low hairline
hirsutism on the face and the body
Tanner stage - T III, **P III**, and axillary hair II,
normal external genitalia.

* ***Laboratory test:***

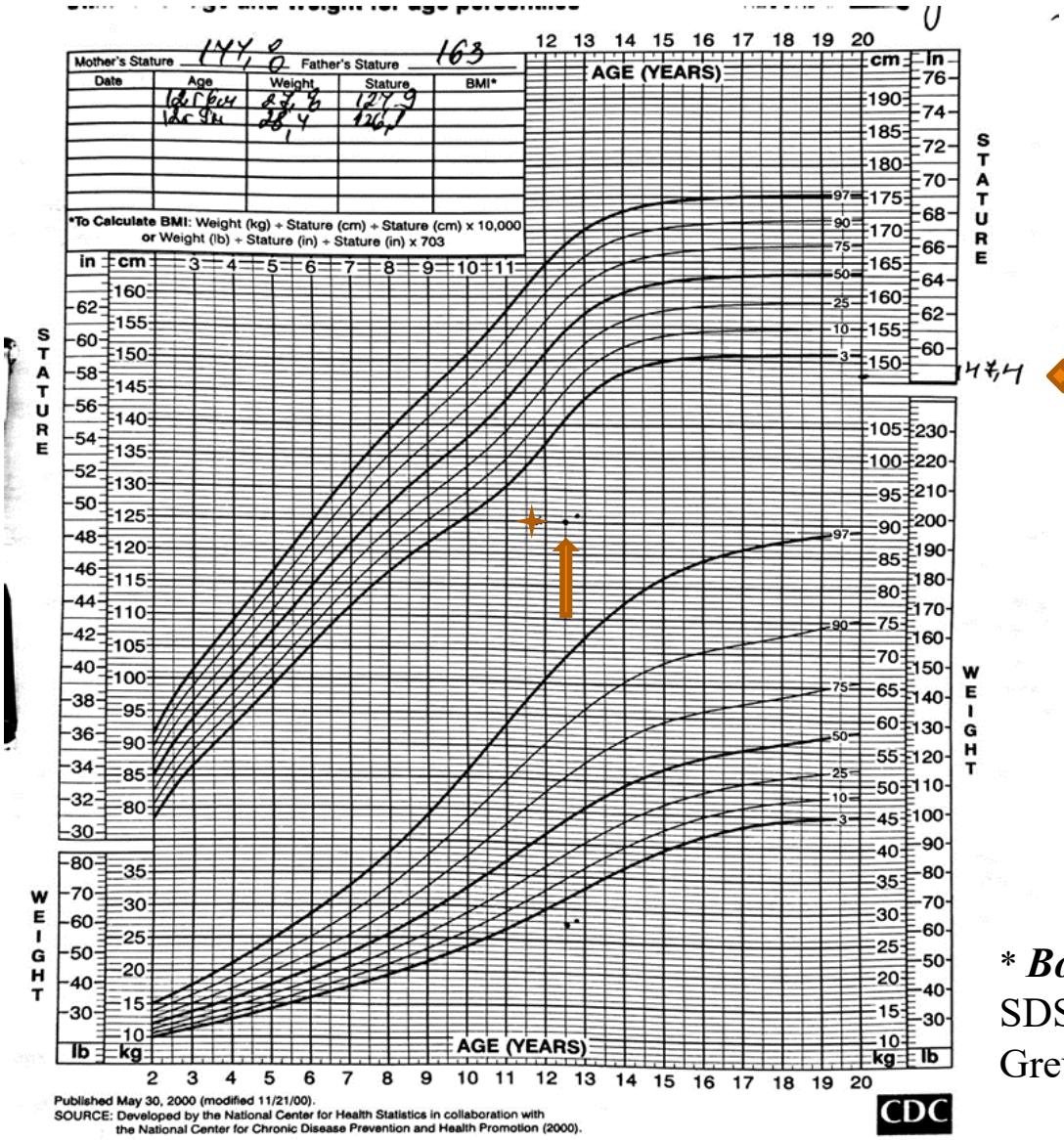
LH 0.206 mIU/mL (1.1-11.6)
FSH 0.522 mIU/mL (2.8-11.3)
Estradiol 81.07 pmol/L (0 –529.2
IGF-1–153 ng/ml (170-527)

* ***Pelvic sonography***: uterus - body: neck 21:17 mm; volume of the right ovary 1.2 cm^3; V left ovary 1 cm^3.

* ***Echocardiography*** found normal aorta and aortic valve.

* ***CT brain*** – normal image

Growth chart



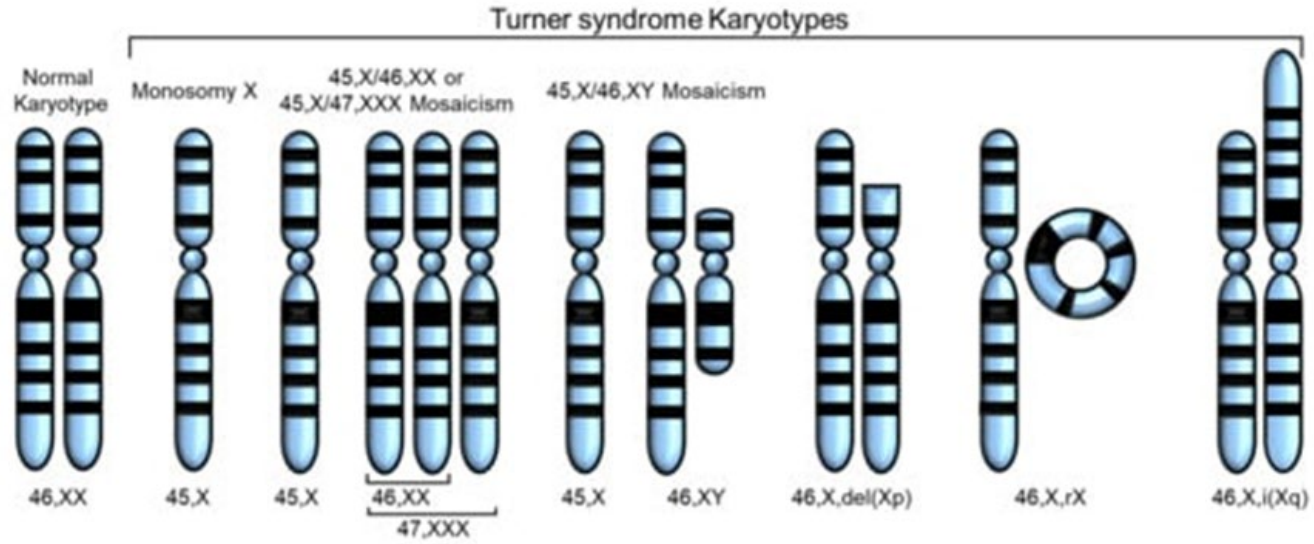
mid-parental height

147,4±7,5 cm

* ***Bone age*** -11.78 years, (-0.59 SDS) according to the Greulich&Pyle (BoneXpert)

Diagnosis

45,X(30 cells)/47,XXX(20 cells)



Treatment

* Growth hormone
* LH-RH (every 3 months)
* Multidisciplinary team

After 3 months

Height ⟶ ⇧ 3 cm

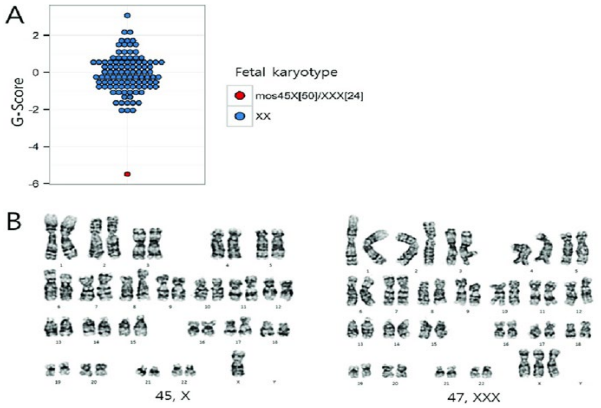
Tanner stage progression (IV)

45,X/47,XXX karyotype

* Turner syndrome (TS) is a common chromosomal disorder affecting approximately 1:2.500 live female births

* Mosaicism is observed in approximately 30% of all TS cases, 3-4% representing the 45X/47XXX karyotype. Such rare cases can present with different phenotypes.

* The presence of the 47XXX cell-line makes them more prone to spontaneous menarche and more fertile, as compared to 45X



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Triple-X syndrome (47,XXX) may be asymptomatic or present with tall stature, microcephaly, epicanthal folds, language learning disabilities and muscular dystonia.

Yoo SH, Ahn MB, Kim SH, Cho WK, Jung MH, Suh BK, Cho K. A case of 45,X/47,XXX mosaic Turner syndrome: Clinical manifestations and effect of growth hormone treatment. J Genet Med 2020;17:47-50. https://doi.org/10.5734/JGM.2020.17.1.47

* The prevalence of a short stature in individuals harboring **45,X/47,XXX** is **64.3%**, which is much lower than that in 45,X monosomy cases (greater than 95%).

* **45,X/47,XXX** mosaicism is diagnosed at a relatively old age compared to the age at which common TS is diagnosed, which may negatively affect the effect of growth hormone treatment.

* Women with **45,X/47,XXX** mosaicism are more likely to retain residual ovarian function. The occurrences of spontaneous puberty and menarche were 88.9% and 77.1% respectively.

* A few rare cases of progressing puberty with mosaic Turner syndrome have been described. The mechanism leading to progressive puberty in this condition is still unknown

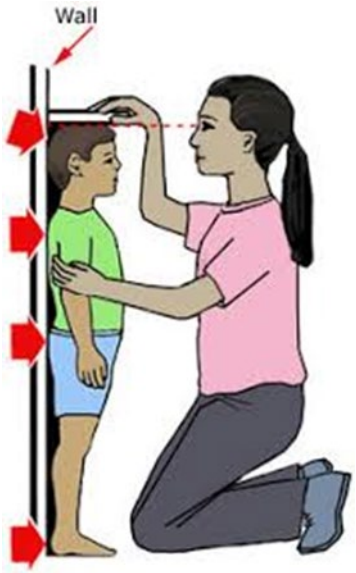
* **45,X/47,XXX** mosaic TS is a very rare genotype; thus, the exact clinical course cannot be predicted. More research is needed to provide specific guidelines to ensure a proper approach for growth, pubertal development, and fertility in TS with 45X/47,XXX karyotypes.

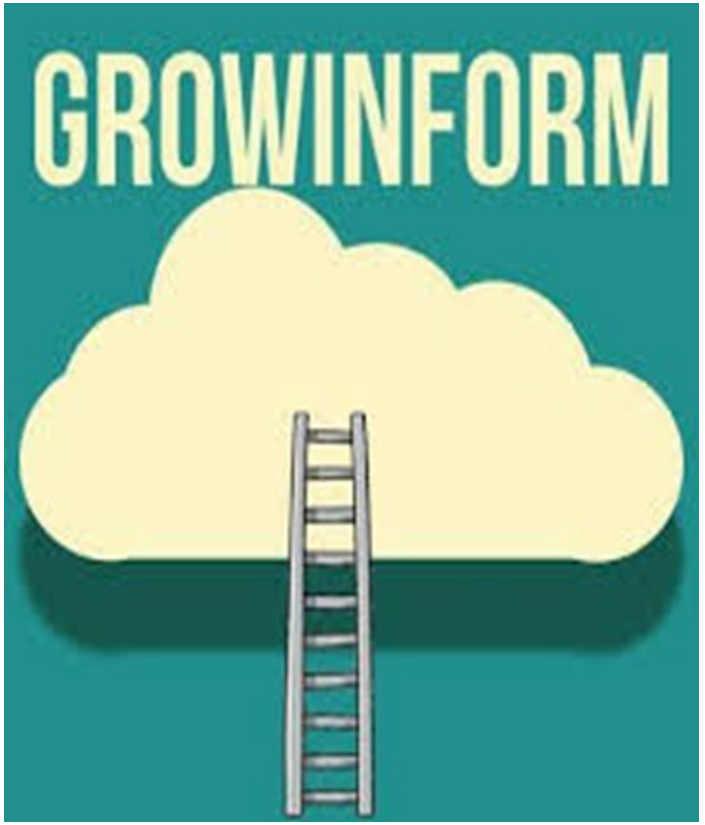
1. A case of 45,X/47,XXX mosaic Turner syndrome: Clinical manifestations and effect of growth hormone treatment. Journal of Genetic Medicine 2020;17:47-50 Published online June 30, 2020; https://doi.org/10.5734/JGM.2020.17.1.47 2020 Korean Society of Medical Genetics and Genomics.

2. Evanchec KA, Rotenstein D. Treatment of precocious puberty in two patients with Turner mosaicism. J Pediatr Endocrinol Metab 2005;18:819-22. 14.

3. Essouabni A, Ahakoud M, Aynaou H, Bouguenouch L, Salhi H, Karim O, Elouahabi H. Rare and Atypical Case of Turner Syndrome With Three Cell Lines. Cureus. 2023 Jun 29;15(6):e41128. doi: 10.7759/cureus.41128. PMID: 37519544; PMCID: PMC10385896.

The present case highlights the importance of short stature as a symptom of a number of childhood diseases.





Regular monitoring of growth in childhood is of great importance for the timely diagnosis of pathological forms and the initiation of correct and successful treatment.

Thank you for your attention!