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45X/47XXX Mosaicism and progressive puberty

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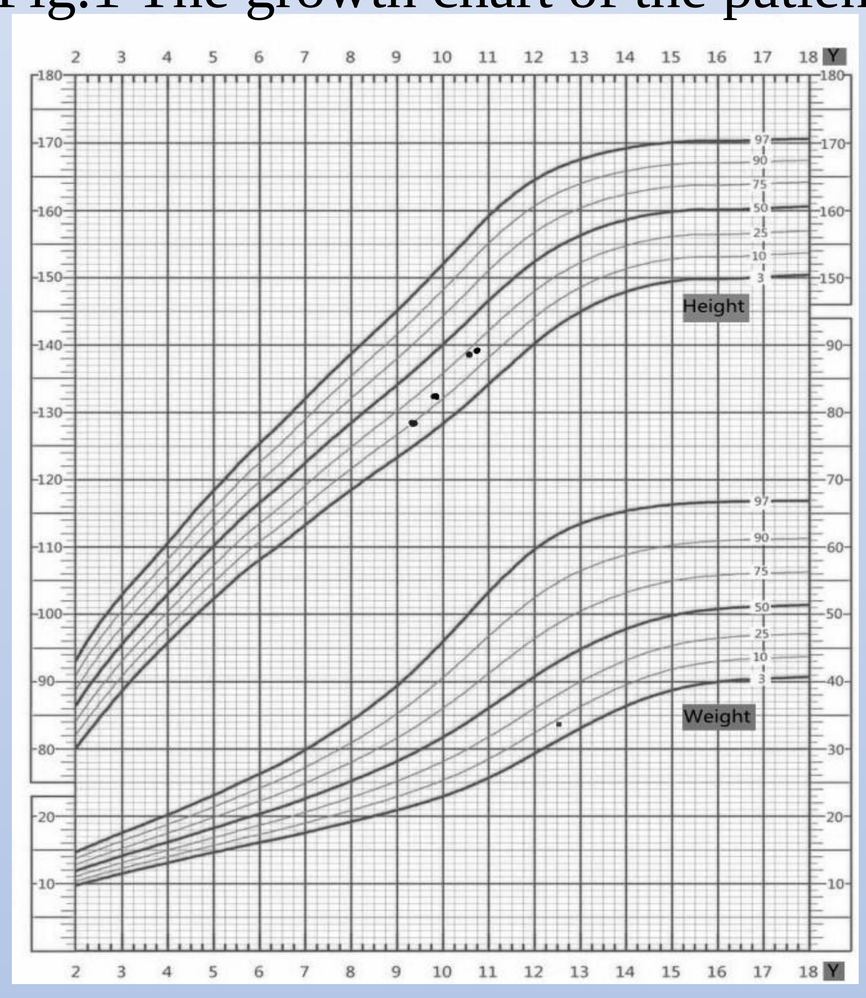
Introduction

The most common clinical features of Turner's syndrome include short stature and ovarian failure. The degree to which each individual is affected varies widely. Mosaic forms of TS tend to have improved prognoses and milder phenotypes, the rarer karyotype 45X/47XXX(about 2% of those with TS)also results in more mildly affected girls . The clinical and laboratory findings in a patient with mosaic Turner syndrome (TS) and precocious sexual development are described here.

- **Design**: Case report
- **Result:** A 9 years 4 months old girl was presented with growth retardation. Chromosome analysis revealed a mosaic karyotype 45X(90%）/47XXX（10%）. She presented with normal height but much lower than the mid-parental height. During annual check-ups, her growth rate was accelerated without growth hormone treatment, her physical examination revealed a Tanner stage II to stage IV and menarche occurred spontaneously within 14 months(Tab.1and Fig.1)

Fig.1 The growth chart of the patient



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

- Table 1. Clinical data and laboratory results of the patient.

Age	Height (cm)	Brest stage	Pubic hair	LH (IU/L)	FSH (IU/L)	E2 (pg/ml)
9y5m	128.5	II	I	8.26	35.47	63.54
9y10m	132	III	II	1.05	10.39	46.72
10y1m	134	III	III	2.16	14.17	89.5
10y7m	138.5	IV	IV	3.37	6.86	166.1

Reference:

1. L. Tauchmanovà, R. Rossi, M. Pulcrano , L. Tarantino, C. G. Lombardi. Turner's syndrome mosaicism 45X/47XXX: An interesting natural history. J. Endocrinol. Invest. 24: 811-815, 2001.
2.Evanchec KA, Rotenstein D. Treatment of precocious puberty in two patients with Turner mosaicism. J Pediatr Endocrinol Metab 2005;18:819-22. 14.