

Anti-Müllerian Hormone: a marker of premature ovarian insufficiency in girls with Turner syndrome

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Background:

Patients with Turner syndrome (TS) typically exhibit short stature and gonadal dysgenesis with pubertal delay and infertility. Up to 30% of these girls will have spontaneous pubertal development, however only 2% achieve a spontaneous pregnancy. Biochemical markers reflecting ovarian reserve in girls and adolescents with TS are therefore needed.

Objective:

Assessment of ovarian reserve in girls and adolescents with TS using serum anti-Müllerian hormone (AMH) and comparing this value with other markers, including serum follicle-stimulating hormone (FSH), number of ovarian follicles (OF) on ultrasound and karyotype.

Methods and results:

METHODS:

► Prospective study

TS girls followed at the Pediatric Endocrinology Unit of a Portuguese General Hospital.

► Period of study

April-August 2013.

► Data analyzed*

Clinical data: age, Tanner stage, spontaneous menarche

Karyotype

Hormone measurements: serum AMH, FSH

Transabdominal gynecologic

ultrasound: number of ovarian follicles.

*Girls with previous or ongoing hormonal replacement therapy were excluded.

► Statistical analysis

Statistical significance was tested with χ^2 test. A p -value <0.05 was considered statistically significant.

RESULTS:

Table 1. Characteristics of studied patients

Number (n)	20
Karyotype	
45,X	8 (40%)
45,X/46,XX	3 (15%)
Other	9 (45%)
Age at testing (y)	
Mean	9.94
Median/SD	8.82 ± 3.81
Minimum	3.45
Maximum	15.22
Spontaneous puberty, n (of 11)^a	
8-13y	1 (9.1%)
>13y	4 (36.4%)
Spontaneous menarche, n (of 11)^a	1 (9.1%)
Measurable AMH, n	13 (65%)
AMH level if measurable, ng/mL (mean/SD)	0.83 ± 1.1

^a girls < 8 y were excluded.

Table 2. Age, karyotype and pubertal development

Age group	n	karyotype		Spontaneous puberty	Spontaneous menarche
		Monosomy	Mosaicism, SAs		
< 8 y	9	2	7	-	-
8-13 y	5	2	3	1	0
> 13 y	6	4	2	4	1

SAs: structural abnormalities in one of the X chromosomes.

Table 6. Markers of ovarian reserve grouped by age and karyotype

Age	n	Normal serum AMH		Normal serum FSH		Presence of OF	
		Monos.	Mosaicic, SAs	Monos.	Mosaicic, SAs	Monos.	Mosaicic, SAs
< 8 y	9	0/2	3/7	0/2	3/7	0/2	5/7
8-13 y	5	0/2	1/3	0/2	1/3	0/2	1/3
> 13 y	6	1/4	2/2	1/4	2/2	2/4	2/2

Table 3. Relationship between FSH and AMH values (n)

	FSH ↑	Normal FSH	P
AMH ↓	13	0	<0,001
Normal AMH	0	7	

Table 4. Relationship between serum AMH and OF (n)

	Absence of OF	Presence of OF	P
AMH ↓	9	4	=0,057
Normal AMH	1	6	

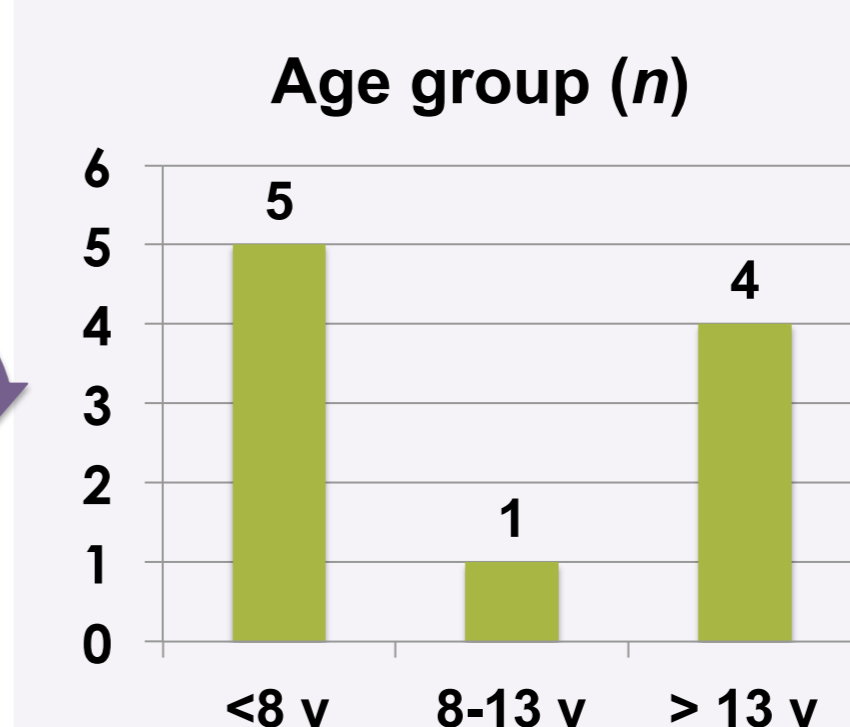
OF in 86% of girls with normal AMH, absence of OF in 69% of girls with low AMH level.

Table 5. Relationship between karyotype and OF (n)

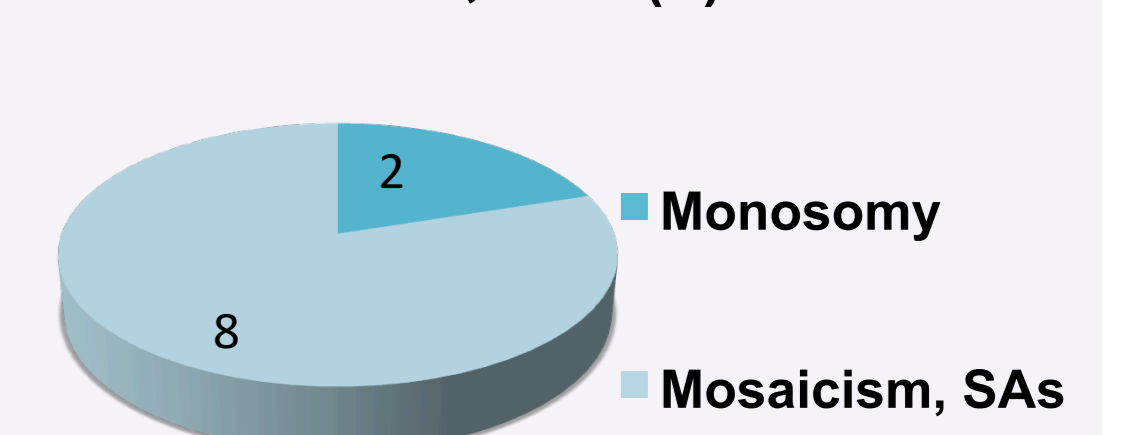
	Absence of OF	Presence of OF	P
Monosomy	6	2	=0,158
Mosaicism, SAs	4	8	

Ovarian follicles were detectable in only 25% of girls with karyotype 45,X and in 67% with karyotype 45,X/46,XX or other cytogenetic abnormalities.

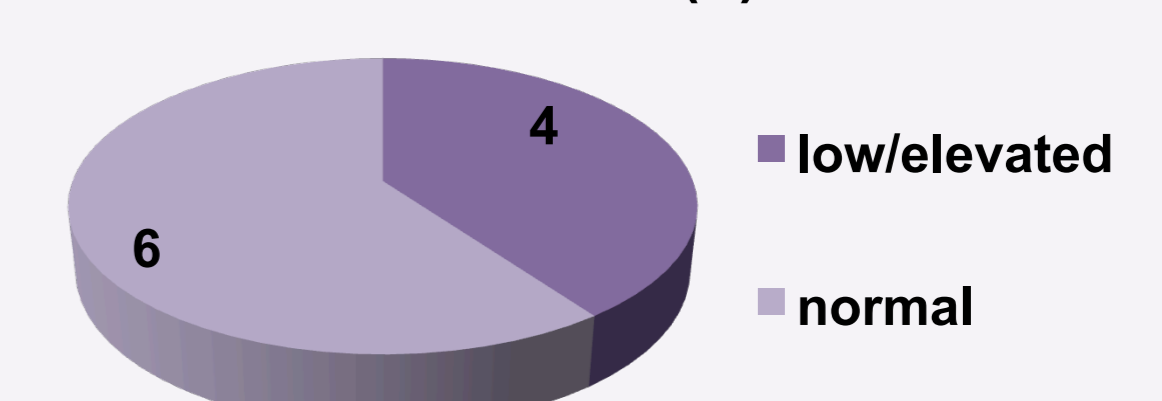
Characteristics of girls with OF (n=10)



Monosomy vs. Mosaicism, SAs (n)



Serum AMH/FSH (n)



Discussion:

Serum AMH correlated well with serum FSH and appears to be a useful marker of the follicle pool. Nevertheless, complementary imaging study is still needed. Karyotype is a good predictive marker of premature ovarian insufficiency when considered together with other parameters.