

Outcomes of Controlled Ovarian Hyperstimulation in Patients with Mosaic Turner Syndrome



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BACKGROUND

Turner syndrome (TS) is a chromosomal disorder affecting 1 in 2,500 live births involving partial or complete loss of an X-chromosome, with or without cell-line mosaicism.¹

Mosaic TS refers to a karyotype in which peripheral lymphocytes vary in chromosomal content, often yielding a milder phenotype.¹

TS is characterized by ovarian insufficiency resulting from accelerated oocyte depletion.² To preserve fertility, some individuals with TS may consider assisted reproductive technologies (ART), although robust data on outcomes in this population remain limited.

PURPOSE & OBJECTIVES

Characterize clinical characteristics and assess outcomes in patients with mosaic TS undergoing oocyte cryopreservation (OC), embryo cryopreservation (EC) or embryo creation with fresh embryo transfer (Fresh ET)

MATERIAL & METHODS

- Retrospective study conducted among individuals with mosaic TS (>5% mosaicism 45-X, microdeletion or ring morphology) who underwent controlled ovarian stimulation from 1/1/2009-9/18/2025 at an academic clinic

- Descriptive statistics were used to assess demographic and baseline clinical characteristics. Linear regression was used to assess correlation between different patient and IVF characteristics

Mosaic Turner syndrome stimulation cycles from 1/1/2009-9/18/2025 (93 cycles, 33 patients)

Exclusion:

- Use of donor oocytes (1 cycle)
- Ovulation Induction (7 cycles)
- Mosaicism <5% 45,X (38 cycles)

7 no start/cancelled cycles

Oocyte cryopreservation (22 cycles)

Embryo Cryopreservation (10 cycles)

Fresh Embryo Transfer (8 cycles)

5 Frozen Embryo Transfers

7 Frozen Embryo Transfers

Ovarian stimulation in patients with mosaic Turner syndrome shows promise, with a subset of patients having successfully achieved live births.

Table 1: Demographics among patients with Mosaic TS **Table 2:** Primary outcomes of Oocyte Cryopreservation (OC), Fresh Embryo Transfer Cycles (Fresh ET) and Upfront Embryo Cryopreservation Cycles **Table 3:** Embryo and PGT-A Data for Fresh ET and Upfront EC cycles in patients with Mosaic TS **Table 4:** Obstetric outcomes among patients with Mosaic TS. Statistics reported as either median [Interquartile range] or mean +/- standard deviation.

Table 1: Demographics

Age [years]	34 [34,39]
BMI [kg/m ²]	23 [21, 27]
Height	5'1" ± 5"
Mosaicism [%]	15 [9, 51]
CD2 FSH [IU/L]	7.6 [4, 17]
AMH [ng/dl]	0.92 [0.39, 3.4]
Turner phenotype	65%
Cardiac condition ¹	15%

¹Two patients with aortic coarctation and one with bicuspid aortic valve, all underwent oocyte cryopreservation

Table 2: Outcomes of Controlled Ovarian Stimulation

	OC (N= 22)	Fresh ET (N = 8)	Upfront EC (N=10)
Age [years]	23 [15,36]	34.5 [30, 39]	36 [34,39]
AMH [ng/dl]	0.5 [0.2, 3.3]	2.3 [0.9, 3.7]	0.8 [0.4, 2.7]
Harvested Oocytes [n]	3 [1,6]	10 [5, 17]	12 [4,18]
Mature Oocytes [n]	1 [0,2]	7.5[3, 16]	9 [3, 16]
Fertilization Rate [%]	-	66 [66,88]	87 [64,94]
Cum. Live Birth Rate	-	50%	50%
PGT-A utilization	-	-	6/10

Table 4: Pregnancy Outcomes

Live births per embryo transfer	50% (10/20)
Gestational age [weeks, days]	39w1d ± 1w3d
Birth weight [grams]	3097 ± 429
C-section rate	30% (3/10)

Table 3: Embryo Transfer Characteristics

Fresh ET (N=8)	
Day 3 vs. Day 5 Transfer	75% vs. 25%
Embryos transferred [n]	1 ± 0.9
Upfront EC (N=10)	
Blastulation Rate [%]	32%
Euploid Rate [%]	12.5 [0,40]
Aneuploidy somatic/sex-chromosome/mosaic [%]	65%/10%/25%
Age PGT-A Group [years]	35 [34,39]

CONCLUSIONS

- Controlled ovarian stimulation in individuals with mosaic TS may enable fertility preservation and/or pregnancy with live birth for a subset of patients, despite diminished ovarian reserve and variable ovarian responses
- Percent mosaicism was not associated with AMH, though higher mosaicism was correlated with a lower number of oocytes retrieved
- Euploidy rates were lower than expected in patients with mosaic TS, suggesting that, if feasible, more oocytes may need to be preserved to achieve a reasonable chance of live birth, and PGT-A may be considered on an individualized basis; these findings are preliminary and require validation in larger studies.

CITATIONS

- 1 Gravholt CH, Andersen NH, Christin-Maitre S, et al. Clinical practice guidelines for the care of girls and women with Turner syndrome. Eur J Endocrinol 2024;190(6):G53-G151.
- 2 Viuff M, Gravholt CH. Turner Syndrome and Fertility. Ann Endocrinol (Paris) 2022;83(4):244-249.

RESULTS

Figure 1: Relationship between percent mosaicism and AMH in patients with Mosaic TS **Figure 2:** Relationship between percent mosaicism and number of harvested oocytes in patients with Mosaic TS

Figure 1: Mosaicism vs. AMH

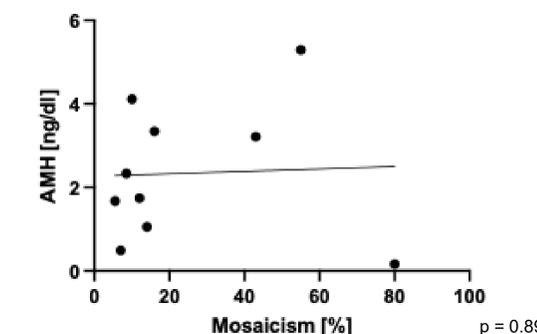
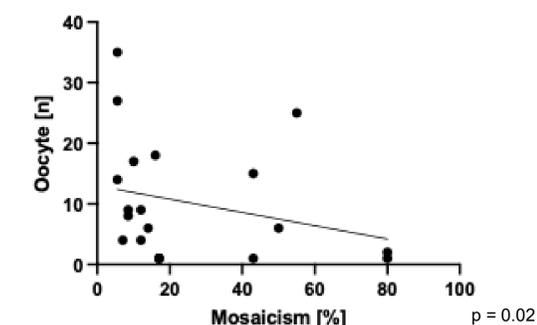


Figure 2: Mosaicism vs. Oocytes Harvested



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