

**0057 - PUBERTAL GONADAL FUNCTION IN 45,X/46,XY CHROMOSOMAL MOSAICISM.
EXPLORING PREDICTIVE VALUE OF MINIPUBERTAL DATA.**

Mode: Research paper

Thematic Unit: DSD

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Background/Objectives: Minipuberty (MP) represents a diagnostic window to assess early gonadal function, particularly in patients with disorders of sex development (DSD); however, its predictive value for gonadal function during puberty (P) remains unclear. Chromosomal mosaicism 45,X/46,XY (DSDc) represents an interesting model to evaluate the scope of this correlation. Our aim was to characterize the clinical and hormonal profiles during P and MP in patients with DSDc and to evaluate their correlation.

Methods: Retrospective review of clinical records of DSDc patients (excluding Turner Syndrome) followed at a tertiary pediatric center (1987-2025). Hormonal profiles were compared with normal male (M) and female (F) controls. Paired P and MP samples of male-assigned patients were correlated.

Results: MP data were available for 25 DSDc patients, assessed at a median age of 24.5 days (9-153). Karyotypes were 45,X/46,X with Y-chromosome structural defects (72%); 45,X/46,XY (20%); 45,Xsry+ (4%); and other mosaicisms (4%). In DSDc LH (3.63 MIU/ml vs. M 2.08 and F 0.07) and testosterone (1.47 ng/ml vs. M 0.73) values were significantly higher than in controls (p

Conclusion: Puberty and minipuberty in DSDc suggest predominant impaired testicular tubular function. Early FSH levels correlated with pubertal gonadotropin secretion, suggesting a potential role as an early marker of gonadal reserve. Minipuberty represents a valuable but limited window into future gonadal function.

