Chromosomal findings and subsequent genotype-phenotype correlations have provided an insight into the variability in growth and development, ovarian function, and phenotypic abnormalities seen in Turner syndrome patients. Chromosomal mosaicism, a relatively common phenomenon in Turner syndrome, impacts the phenotype and clinical outcome. We present prenatal and follow-up findings on one of the least common types of Turner mosaicism. In 1992, prenatal chromosome studies performed in our laboratory showed a 45,X/47,XXX karyotype in the amniotic fluid indicative of Turner mosaicism. The child was clinically normal at birth with no problems during the newborn period. At 11 months of age, the baby girl had normal echocardiogram, no dysmorphic features, and normal developmental milestones. At the time of clinical follow-up at 12 years of age, cytogenetic studies from were performed again on peripheral blood which confirmed the prenatal findings of 45,X[20]/47,XXX[30] with similar percentages. Clinical evaluation revealed normal intellect, normal phenotype with no dysmorphic features, and normal growth and development for age. Pelvic ultrasound was also normal. An endocrine evaluation is in progress. Existing reports show that girls with 45,X/47,XXX are only mildly affected, with good preservation of ovarian function, and the present case supports these findings. Due to the rarity of this particular type of Turner mosaicism, additional reports with follow-up studies help to provide supportive evidence for phenotypic and clinical outcome expectations and facilitate prenatal counseling and patient care.

**CONCLUSION**

- Genetically the patient has the rare 45,X/47,XXX mosaic state of Turner syndrome
- Phenotypically the patient shows none of the characteristics of Turner syndrome
- The patient has no apparent characteristics of 47,XXX
- This adds to the evidence that 45,X/47,XXX Turner syndrome cases are less severe
- The extra X in 47,XXX cells may offset the negative effects of loss of X in 45,X cells
- Additional reports of 45,X/47,XXX cases will help prove or disprove this theory
- Endocrine studies are ongoing and may reveal abnormal hormone levels
- Development in this patient suggests that she will be fertile without hormone treatment

**INTRODUCTION**

- Turner syndrome is defined as a loss or structural abnormality of one X chromosome
- Variants include mosaic states such as 45,X/46,XX or 45,X/47,XXX
- Occurs in about 1:2000 live female births
- Characteristics include short stature, dysmorphic features, and ovarian dysgenesis
- Estrogen therapy is usually required for normal puberty and fertility
- Severity is highly variable, making counseling and treatment difficult
- Evidence suggests the 45,X/47,XXX variant causes the fewest clinical problems
- Forty percent of Turner syndrome cases are mosaic; of these, 1% are 45,X/47,XXX
- Since this variant is so rare, not enough cases have been studied to be conclusive
- Correlating severity with the type of variant will lead to better counseling and treatment

**RESULTS**

- Chromosome studies on the amniotic fluid indicated the 45,X/47,XXX variant
- No phenotypic abnormalities were found during clinical evaluation at 11 months of age
- At 12 years of age, chromosome studies confirmed the 45,X/47,XXX karyotype
- Clinical evaluation at 12 years of age found no abnormalities

**CONCLUSION**

- Genetically the patient has the rare 45,X/47,XXX mosaic state of Turner syndrome
- Phenotypically the patient shows none of the characteristics of Turner syndrome
- The patient has no apparent characteristics of 47,XXX
- This adds to the evidence that 45,X/47,XXX Turner syndrome cases are less severe
- The extra X in 47,XXX cells may offset the negative effects of loss of X in 45,X cells
- Additional reports of 45,X/47,XXX cases will help prove or disprove this theory
- Endocrine studies are ongoing and may reveal abnormal hormone levels
- Development in this patient suggests that she will be fertile without hormone treatment

---

**Patient Genotype**

- Turner syndrome phenotype
  - Short Stature
  - Craniofacial dysmorphisms
  - Renal Failure
  - Ovarian Dysgenesis
  - Learning Disabilities

- Phenotype of the 45,X/47,XXX patient
  - Average Stature
  - Normal Cranial and Head Circumference
  - Normal Renal Ultrasound Findings
  - Normal Uterus and Ovaries
  - An “excellent” student, well behaved

---

**Standard Growth Curve**

- ◦ = Typical patient with 45,X Turner syndrome
- ● = 45,X/47,XXX Patient

GH = Growth Hormone therapy

Adapted from: www.meadjohnson.com/professional/newsletters/ppv4n2a.html