

With the advent of prenatal diagnosis, 45X/46XY mosaicism is ascertained on a fairly regular basis. The question is whether 45X/46XY mosaicism is associated with Turner syndrome, infertility, ambiguous genitalia, or any other problems. Until recently, selection for testing for 45X/46XY mosaicism has been based on the presence of unusual postnatal features.

The authors have taken advantage of current prenatal testing procedures to conduct an unbiased survey of 45X/46XY incidence by sending a questionnaire to an international sample of 730 cytogenetic laboratories. A total of 92 cases of prenatal diagnosis of 45X/46XY mosaicism were reported. There was good clinical information on 76 cases; 75 were phenotypically male and 1 was female. Three of the phenotypic males had hypospadias, and the phenotypic female had clitoromegaly. Many of the cases had been terminated prenatally at the parents' request, and gonad histology was done in 11 cases. Of these, 3 (27%) had abnormal testicular development, but only 1 had abnormal external genitalia. Of the 75 "males," 5 had other congenital abnormalities of consequence. The authors found no relationship between the degree of mosaicism observed at prenatal diagnosis and the severity of abnormalities.

The percentage of 45X cells ranged from 1% to 98%; the majority had less than 50% 45X cells and (probably for this reason) presented with a normal male phenotype rather than a Turner phenotype.

Long-term follow-up of 45X/46XY patients is not available, and information concerning long-term stature, pubertal development, tumor risk, and fertility is needed. However, this study suggests that most patients with 45X/46XY karyotype (95%) have normal male genitalia, in contrast with previous postnatal studies. Dysgenetic gonads appear to occur in about 25% of cases, but whether this figure would be higher by

puberty is not yet known. Dysgenetic gonads in normal-appearing males who have never had chromosome studies may be a source of infertility or gonadoblastomas in the general population.

Chang HJ, et al. *Am J Hum Genet* 1990;46:156-167.

**Editor's Comment**—*This is an important study in view of the frequent use of prenatal diagnosis and concomitant finding of 45X/46XY karyotypes. This study suggests that most cases will do well, but clearly long-term follow-up is needed. Also of interest, it appears that approximately half of the families where this diagnosis was made prenatally have terminated the fetus. These decisions were probably based on expectations of poor outcome ensuing from previous, biased, reports, whereas the actual prognosis may not be so unfavorable for such children.*

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