

47,XYY

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=ABSTRACT=

A Case of Full Term Delivery of a Child with 47,XYY Subsequent to Prenatal Diagnosis at Midtrimester

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47,XYY is a rare sex chromosomal disorder. Approximately 1.45 per 1,000 live births have on XYY chromosome pattern. The extra Y chromosome is paternal in origin and results from nondisjunction in the second meiotic division. Although the phenotype is normal on the newborn, an increased incidence of minor anomalies has been reported. Recently, a 37-year-old primigravid woman received amniocentesis at 17 weeks gestation at a private clinic and was diagnosed as having a fetus with 47,XYY. We performed amniocentesis again at 20 weeks of pregnancy and confirmed fetal karyotype to be 47,XYY using the conventional cytogenetics and fluorescence in situ hybridization (FISH) techniques. As she did not want to terminate her pregnancy, she was put under antenatal care but ended up in vaginal delivery in 40 weeks. As a result of physical examination, the neonate showed a normal phenotype except for a mild hypospadias and a simian crease.

Key Words : 47,XYY, Prenatal diagnosis, Nondisjunction, FISH

47,XYY	가	(spermatogenesis)	FISH	47,XYY
2	(meiotic division)	(nondisjunction)		
		1967		
Price	Whatmore			
	Ratcliffe			
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		^{1,2}	:	, 37 .
1980		XYY	:	17 5
		가 ³		47,XYY

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-Full term delivery of a child with 47,XYY subsequent to prenatal diagnosis-

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 17 5
 47,XYY
 (alpha-fetoprotein) 16.20 ug/ml
 (1.26 MoM) 20
 FISH 47,XYY
 (Fig. 1, 2, 3)
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 G-banding 47,XYY
 (Fig. 1) DNA probe
 FISH 47,XYY (Fig. 2, 3)



Fig. 1. Apparent 47,XYY karyotype detected at amniocentesis (G-banding)

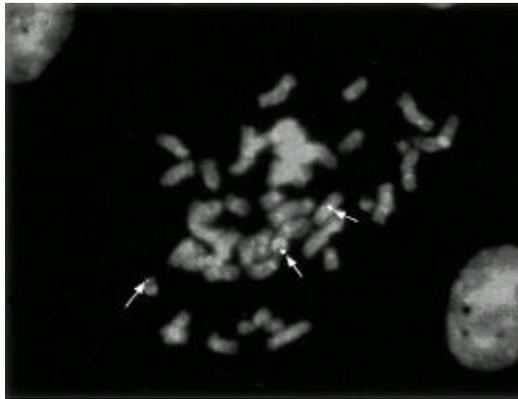


Fig. 2. Pictures of FISH using DXZ1 probe for X chromosome and SRY probe for Y chromosome in amniotic cells. Probe for X and Y chromosome is labeled with spectrum green (short arrow) and spectrum orange (long arrow), respectively

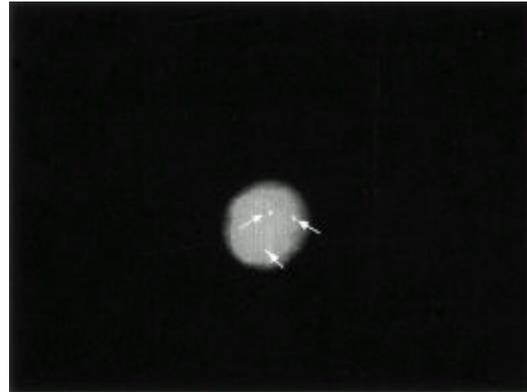


Fig. 3. FISH images of interphase demonstrating two chromosomes Y (orange) and a X chromosome (green)

:17
 47,XYY
 20
 47,XYY
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 (triple test) 17
 20 110/60 mmHg
 20 + 5 FISH
 32
 31 2 ,
 29 3
 33 가
 (nonstress test)
 36 12.1 gm/dL,
 36.0%, 10,920/mm³, 212,000/mm³
 , X
 :2001 4 11 40 6
 1.5 cm 10
 120/80 mmHg, 74 , 160 cm,
 70 kg ,
 . 14.1 gm/dL,
 40.6%, 28,520/mm³, 93.2%,
 215,000/mm³ . 5
 30 12