

# Isolated Fetal Pyelectasis and Chromosomal Abnormalities

Cüneyt E. TANER, İrem ŞENYUVA, Aysun CAMUZCUOĞLU, Gülsen DERİN

İzmir, Turkey

**OBJECTIVES:** Try to find out the frequency of isolated fetal pyelectasis and its association with abnormal fetal karyotypes in a high risk pregnant population

**STUDY DESIGN :** 759 pregnant with advanced maternal age or with increased risk for trisomy 21 in triple tests were enrolled into the study. Ultrasonographic examinations and amniocentesis were performed for all pregnant at 15-21 weeks of their gestations

**RESULTS:** 649 pregnant completed the study protocol. 424 cases had advanced maternal age and 225 cases had increased risk for trisomy 21. There were 15 cases (2.31%) with isolated pyelectasis and these cases had normal karyotypes except a normal variance (46 XX inv9p11q13). There were 14 (3.30%) chromosomal abnormalities in advanced maternal age group and 7 (3.11%) in pregnant with increased risk in triple tests.

**CONCLUSION:** Although increased risk of Down's Syndrome is not high in pregnant with isolated fetal pyelectasis, they should be followed and searched for other findings of aneuploidy.

(*Gynecol Obstet Reprod Med*;13:2 140-142)

**Key Words:** Ultrasonographic Fetal Abnormalities, Chromosomal Abnormalities, Fetal Pyelectasis

## Introduction

Fetuses with chromosomal abnormalities have some defects that can be recognized by second trimester detailed ultrasonography. Dilatation of the fetal renal pelvis in the second trimester is an easily detectable sonographic finding and has gained importance since it may be a marker for aneuploidy and it may be a precursor of potential urinary tract pathology.<sup>1-4</sup>

In this study we tried to find out the frequency of isolated pyelectasis and its association with abnormal fetal karyotypes in a high risk pregnant population.

## Material and Methods

Our study group included 759 singleton pregnant women undergoing genetic amniocentesis because of advanced maternal age (>35 years) and increased risk for trisomy 21 in triple tests. Informed consents were obtained from all pregnant before investigations and the study was approved by our hospital's ethic committee. A detailed abdominal ultrasonographic examination was performed for all pregnant women at 15-21 weeks of their gestations for associated malformations or anatomic abnormalities. Fetal pyelectasis appeared as an un-

chogenic area of one or both kidneys with anteroposterior diameter of renal pelvis (5-10 mm) by transabdominal scan before 22 weeks gestation. All pathologic findings were noted and ultrasonographic examinations repeated at the time of amniocentesis. Amniocentesis were performed for detecting fetal karyotypes at 16-21 weeks gestations with a 22 gauge spinal needle. Karyotypes were determined in all cases by amniotic fluid culture using standard procedures at special genetic laboratories (Gentan or CDF). Cases with negative cultures were excluded. Women having ultrasonographic multiple abnormalities and who subsequently aborted spontaneously or pregnant women without amniocentesis and fetal karyotype and cases where pyelectasis was disappeared were not included in the study group.

## Results

649 pregnant women completed the study: 424 cases had advanced maternal age whereas 225 women had increased risk for trisomy 21. Mean age of the pregnant with advanced maternal age was 37.9 years (range 35-45). And mean age of the pregnant with increased risk in triple test was 28,8 years (range 18-34). There were 15 cases (2.31%) with isolated fetal pyelectasis detected by ultrasonography. 12 of these cases were bilateral and 3 of them were unilateral. There were pyelectasis in left kidney in two cases and one in the right kidney.

Of the 15 cases with isolated fetal pyelectasis 14 cases had normal karyotypes and only a case had normal variance (46 XX inv9p11q13). Karyotype analysis results showed 14 (3.30%) chromosomal abnormality in 424 cases with advanced maternal age. Five of these 14 cases were trisomy

Ministry of Health Aegean Maternity and Education Hospital  
İzmir, Turkey

Corresponding Author: Cüneyt Eftal Taner  
105 Sok. No: 3/17 Göztepe  
İzmir, Turkey  
cuneyt-taner@hotmail.com

Submitted for Publication: 20.11.2007

Accepted for Publication: 13.12.2007

21.225 women with increased risk in triple tests had 7 (3.11%) chromosomal abnormality. One of these abnormal karyotype was trisomy 21. No pyelectasis were detected in any of these fetal Down's Syndrome cases.

## Discussion

Abnormalities of the urinary tract are relatively common accounting for approximately 20% of all fetal malformations (5). Fetal renal pelvic dilatations less than 4 mm in diameter are likely to be normal. Renal pelvic diameters of 5 to 10 mm can be associated with significant renal pathology including pelviureteric function obstruction and reflux and need follow up later in pregnancy and also in the neonatal period. Renal pelvic diameters greater than 1 cm are likely to have significant renal disease and require investigations.<sup>6</sup> Anderson<sup>7</sup> reported that most kidneys with hydronephrosis in later pregnancy and the neonatal period had renal pelvic diameters of less than 10 mm before 23 weeks gestations. They showed that by plotting renal pelvic diameters for obstructed and normal kidneys the measurements were very similar for both groups until 22 weeks after which time they began to diverge. Some authors advised to make serial ultrasound examinations in order to evaluate progression or regression of pyelectasis.<sup>4,8,9</sup> The association of aneuploidy with mild dilatation of the fetal renal pelvis was first reported in 1990 by Benacerraf and et al.<sup>3</sup> They noted that 3.3% of the fetuses with pyelectasis had Down's Syndrome. Some other authors also reported similar association with Down's Syndrome and fetal renal pyelectasis.<sup>2,4,8,10,11</sup>

The mean incidence of fetal pyelectasis in midtrimester was reported to be 0.59-2.9% in the literature.<sup>2,4,8,11-14</sup> In our study we had 15 cases with isolated pyelectasis in 649 high risk pregnant women with an incidence of 2.3%. In a similar study investigating isolated mild pyelectasis in 1093 pregnant women with advanced maternal age the authors reported the incidence of isolated pyelectasis as 5.1% by transvaginal ultrasonography in early pregnancy.<sup>12</sup> Their incidence decreased to 2.9% at the time of amniocentesis. They reported one case of trisomy 21 and one case of triploidy among 56 cases of pyelectasis and concluded that pyelectasis may be transient and not associated with an increased risk of abnormal fetal karyotypes. In our study we also didn't find any abnormal karyotypes in our 15 cases with isolated fetal pyelectasis. In another study investigating 2900 pregnant women for fetal pyelectasis and Down's Syndrome the authors found normal karyotypes in 62 cases of isolated fetal pyelectasis.<sup>15</sup> They recommended prenatal cytogenetic tests in older women and or in presence of associated sonographic abnormalities.

On the other hand Vintzileos<sup>16</sup> reported that 5% of fetuses with isolated pyelectasis had Down's Syndrome. But Wickstrom and Nicolaides<sup>4,11</sup> reported the incidence as 0.6%

and 0.3% in their prospective studies.

In a multicenter study including 737 fetuses with mild pyelectasis the authors found 12 abnormal karyotypes.<sup>10</sup> The overall incidence of aneuploidy in fetuses with pyelectasis was 1.70%. In fetuses where pyelectasis was the only sonographic abnormality seen the incidence of proven aneuploidy was 0.46%. They calculated the aneuploidy risk in the presence of isolated pyelectasis in women >36 years as 2.22% and as 0.33% in women <35 years. The authors concluded that pregnancies complicated by isolated mild pyelectasis increases the risk for aneuploidy in particular trisomy 21. Wickstrom<sup>9</sup> also noted that isolated pyelectasis was associated with an increased risk for Down's Syndrome, beginning at maternal age of 31 years in the interval of 16-20 weeks gestation.

In another study investigating 5944 fetuses for fetal pyelectasis and Down's Syndrome the authors found the predictive value of isolated pyelectasis as one in 340.<sup>2</sup> They concluded that although renal pyelectasis is more common in Down's Syndrome genetic amniocentesis should be reserved for those cases presenting other risk factors such as advanced maternal age, low MSAFP or other sonographic abnormalities. Similary Havutcu<sup>17</sup> reported that fetal pyelectasis in unselected low risk population is not high (1.25%) and should not be an indication for invasive prenatal karyotyping.

We concluded that although increased risk of Down's Syndrome is not high for pregnant women with isolated fetal pyelectasis, they should be followed and searched for other findings of aneuploidy

## References

1. Signorelli M, Cerri V, Taddei F, Groli C, Bianchi UA. Prenatal diagnosis and management of mild fetal pyelectasis: implications for neonatal outcome and follow-up. *Eur J Obstet Gynecol Reprod Biol.* 2005 Feb 1;118(2):154-9.
2. Corteville JE, Dicke JM, Crane JP. Fetal pyelectasis and Down syndrome: is genetic amniocentesis warranted? *Obstet Gynecol.* 1992 May;79(5):770-2.
3. Benacerraf BR, Mandell J, Estroff JA, Harlow BL, Frigoletto FD Jr. Fetal pyelectasis: a possible association with Down syndrome. *Obstet Gynecol.* 1990 Jul;76(1):58-60.
4. Wickstrom EA, Thangavelu M, Parilla BV, Tamura RK, Sabbagha RE. A prospective study of the association between isolated fetal pyelectasis and chromosomal abnormality. *Obstet Gynecol.* 1996 Sep;88(3):379-82.
5. Elder JS. Antenatal hydronephrosis. Fetal and neonatal management. *Pediatr Clin North Am.* 1997 Oct; 44(5): 1299-321.
6. Twining P, Multugo JM, Pilling DW. Textbook of Fetal Abnormalities in: Twining P Urinary Tract Abnormalities

London, Churchill Livingstone 2000; 269-314

7. Anderson N, Clautice-Engle T, Allan R, Abbott G, Wells JE. Detection of obstructive uropathy in the fetus: predictive value of sonographic measurements of renal pelvic diameter at various gestational ages. *AJR Am J Roentgenol.* 1995 Mar;164(3):719-23.
8. Ouzounian JG, Castro MA, Fresquez M, al-Sulyman OM, Kovacs BW. Prognostic significance of antenatally detected fetal pyelectasis. *Ultrasound Obstet Gynecol.* 1996 Jun; 7(6):424-8.
9. Wickstrom E, Maizels M, Sabbagha RE, Tamura RK, Cohen LC, Pergament E. Isolated fetal pyelectasis: assessment of risk for postnatal uropathy and Down syndrome. *Ultrasound Obstet Gynecol.* 1996 Oct;8(4):236-40.
10. Chudleigh PM, Chitty LS, Pembrey M, Campbell S. The association of aneuploidy and mild fetal pyelectasis in an unselected population: the results of a multicenter study. *Ultrasound Obstet Gynecol.* 2001 Mar;17(3):197-202.
11. Nicolaidis KH, Cheng HH, Abbas A, Snijders RJ, Gosden C. Fetal renal defects: associated malformations and chromosomal defects. *Fetal Diagn Ther.* 1992;7(1):1-11.
12. Guariglia L, Rosati P. Isolated mild fetal pyelectasis detected by transvaginal sonography in advanced maternal age. *Obstet Gynecol.* 1998 Nov;92(5):833-6.
13. Sairam S, Al-Habib A, Sasson S, Thilaganathan B. Natural history of fetal hydronephrosis diagnosed on mid-trimester ultrasound. *Ultrasound Obstet Gynecol.* 2001 Mar;17(3):191-6.
14. Dudley JA, Haworth JM, McGraw ME, Frank JD, Tizard EJ. Clinical relevance and implications of antenatal hydronephrosis. *Arch Dis Child Fetal Neonatal Ed.* 1997 Jan;76(1):F31-4.
15. Borrelli AL, Borrelli P, Di Domenico A, Felicetti M, Corcione M, Torella M. The incidence of chromosomal anomalies in fetuses affected by mild renal pyelectasis. *Minerva Ginecol.* 2004 Apr;56(2):137-40.
16. Vintzileos AM, Campbell WA, Rodis JF, Guzman ER, Smulian JC, Knuppel RA. The use of second-trimester genetic sonogram in guiding clinical management of patients at increased risk for fetal trisomy 21. *Obstet Gynecol.* 1996 Jun;87(6):948-52.
17. Havutcu AE, Nikolopoulos G, Adinkra P, Lamont RF. The association between fetal pyelectasis on second trimester ultrasound scan and aneuploidy among 25,586 low risk unselected women. *Prenat Diagn.* 2002 Dec;22(13):1201-6.