The importance of high resolution transvaginal sonography in early screening of fetal chromosomal pathology

M. Felicetti, A. Di Domenico, P. Salzano, P. Borrelli, A. Laboccetta, A. L. Borrelli

Ostetrics, Gynacology & Neonatal Department, Faculty of Medicine and Surgery, II University of Naples (Italy)

Summary

Background: Continuous technological improvement made in the field of ultrasound applied to obstetric diagnostics (see tridimensional sonography) [1] has contributed to a better and non-invasive early diagnosis of fetal malformations.

Methods: To evaluate the usefulness of ultrasound in detecting early chromosomal derangements, the authors carried out a high resolution transvaginal sonography (> 6.5 mHz), between the 10th and 14th week of gestational age, on 650 pregnant women at risk for congenital anomalies and afterwards they were subjected to early amniocentesis

Results: Sonographic fetal anomalies were seen in 61 cases (9.3%). The incidence of fetal anomalies in these cases was 52.5%. Trisomies and number of sexual chromosome anomalies were seen, especially, in the cases of cystic septated hygroma and fetal nuchal translucency ≥ 3 mm which are the most frequent sonographic markers of chromosomopathies.

Conclusions: Although further studies are necessary, these findings suggest the usefulness of high resolution transvaginal sonography for the early screening of chromosomopathies.

Key words: Transvaginal sonography; Fetal chromosomal pathology; Chromosome abnormalities.

Introduction

Continuous technological improvement made in the field of ultrasound applied to obstetric diagnostics (see tridimensional sonography) [1] has contributed to a better and non-invasive early diagnosis of fetal malformations.

In this field sonographic diagnostics, using a higher frequency for the high resolution transducers, have improved with the introduction of a new transvaginal probe. (6.5 MHz) [2].

Ultrasound is used successfully in gynaecology [3, 4] and for the early diagnosis of fetal anomalies in the first and early second trimester of pregnancy [5-7].

The aim of our study was to detect whether ultrasound transvaginal diagnostics with high resolution transducers, used during the first trimester of pregnancy, could have an important role in the early non-invasive screening of fetal chromosomal pathologies. We attempted to verify if a correlation exists among fetal anomalies diagnosed with this method and chromosomopathies.

Materials and Methods

Using transvaginal sonography (TVS), from January 1995 to March 2000 we examined 650 pregnant women at risk for congenital anomalies: 494 cases (76%) with advanced maternal age (36 years or more); 91 cases (14%) with personal or family history of congenital anomalies; 65 cases (10%) with maternal disease (diabetes mellitus).

We used The Esaote AU 5 Harmonic with vaginal 6.5 MHz probe. The TVS was performed between the 10th and 14th week of pregnancy and later we performed amniocentesis for the study of fetal karyotype between the 15th and 17th week, and abdominal sonography between the 21st and 24th week.

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Results

Through the early TVS we noticed 61 fetal anomalies (9.3%). The incidence of chromosomal anomalies of the total examined pregnant women in this cases was 52.5% with 32 cases verified by examining of fetal kariotype. The total incidence of identified chromosomal anomalies was 4.9% (see Table 1). The identified anomalies are ordered by increasing incidence (in Table 2): cystic hygroma, 25 cases (41%) (see Figure 1); nuchal translucency ≥ 3 mm, 10 cases (16%) (see Figure 2); pyelectasis, 10 cases (16%), frequently bilateral; choroid plexus cysts, 9 cases (15%); blighted ovum, 5 cases (8%); and anencephaly, 2 cases (3%).

Fifty-three percent of cases of cystic hygroma and 60% of cases of nuchal translucency ≥ 3 mm had some chromosomal anomalies (Table 3). All cases of pyelectasis

Table 1. — *Noted sonographic anomalies and correlated chromosomopathies in 650 pregnancies.*

Sonographic anomalies	Pathological karyotype in cases with sonographic anomalies	Incidence of chromosomal anomalies for the examined pregnant women
61 (9.3%)	32 on 61 → (52.5%)	32 on 650 → (4.9%)

Table 2. — *Features of the noted TVS anomalies.*

Simple cystic hygroma	15	(41%)
Septated cystic hygroma	10	
Nuchal translucency ≥ 3 mm	10	(16%)
Pyelectasis	10	(16%)
Choroid plexus cyst	9	(15%)
Blighted ovum	5	(8%)
Anencephaly	2	(3%)
Total	61	(100%)

Table 3. — *Amniocentesis confirmed chromosomal anomalies in cases of cystic hygroma and nuchal translucency > 3 mm.*

Sonographic anomaly	Number of cases	Karyotype
Simple cystic hygroma	15	21 Trisomy: 2 cases Turner syndrome: 2 cases
Septated cystic hygroma	10	21 Trisomy: 5 cases Turner syndrome: 1 case 18 Trisomy: 2 cases 15 Trisomy: 1 case
Chromosomal anomalies associated with cystic hygroma		13/25 (53%)
Nuchal translucency ≥ 3 mm	10	21 Trisomy: 3 cases Turner syndrome: 1 case 18 Trisomy: 2 cases
Chromosomal anomalies associated with nuchal translucency ≥ 3 mm		6/10 (60%)

healed spontaneously by the second sonographic control or at the end of pregnancy without evidence of fetal pathology and they no had abnormal kariotype; the same thing occurred in the cases of choroid plexus cysts. In the cases of blighted ovum and anencephalia a kariotype study was not performed for spontaneous (blighted ovum) or voluntary (anencephalia) interruption of the pregnancy.

Conclusions

Our study, although performed on a limited number of cases, showed – according to the international literature data [9-11] – the importance of high resolution of transvaginal sonography in the early diagnosis of fetal morphologic anomalies which suggest chromosomal pathology. In particular we found that cystic hygroma and nuchal translucency ≥ 3 mm were sonographic markers which, in a significant percentage of cases, were associated with some chromosomal anomalies. Therefore the use of TVS in the first trimester of pregnancy should be routine screening even for the population with no risk. Those with morphologic anomalies require a fetal karyotype.

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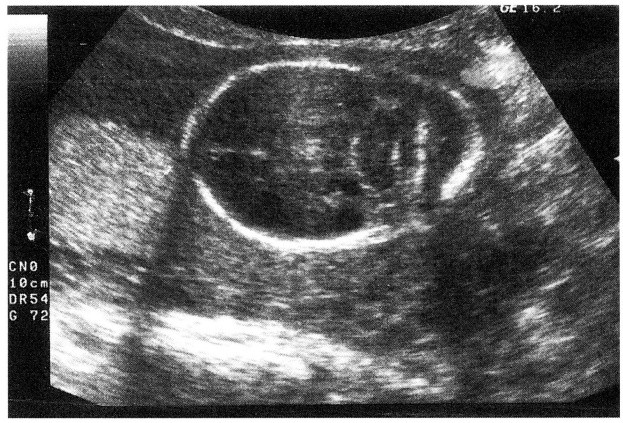

Figure 1. — Simple cystic hygroma.

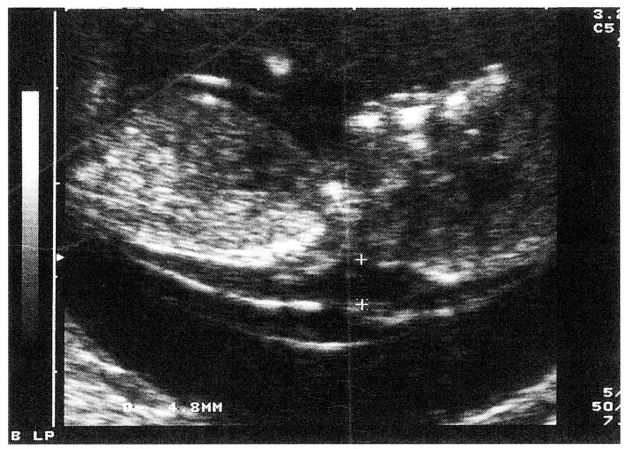

Figure 2. — Nuchal translucency > 3 mm.

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Address reprint requests to:
A. L. BORRELLI, M.D.
Via Giacinto Gigante, 3/B
80136 Napoli (Italy)