The sonographic identification of fetal gender from 11 to 14 weeks of gestation

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ABSTRACT

Objective To determine the feasibility of correctly identifying fetal gender from 11 to 14 weeks’ gestation.

Methods A prospective cross-sectional study in a university Department of Obstetrics and Gynaecology, London. A total of 524 women from an unselected population underwent a detailed assessment of fetal anatomy at 11–14 weeks of gestation (confirmed by crown–rump length) by means of transabdominal sonography, and transvaginal sonography (26%) when necessary. Fetal gender was identified in the transverse and sagittal planes, and was confirmed at birth.

Results The overall success of correctly assigning fetal gender increased with gestational age from 46% to 75%, 79% and 90% at 11, 12, 13 and 14 weeks, respectively. The ability of the operator to assign fetal gender significantly improved with increasing gestational age (p < 0.0001), being 59%, 87%, 92% and 98% at 11, 12, 13 and 14 weeks, respectively. The accuracy of correctly identifying fetal gender when attempted did not change with gestational age. Fetal gender or the performance of the scan by different operators did not affect the results.

Conclusion Whilst the accuracy of sonographic determination of fetal gender at 11–14 weeks is good, it still falls significantly short of invasive karyotyping tests.

INTRODUCTION

There have been a number of studies using ultrasound to detect and correctly identify fetal gender in the early second and third trimesters. As a consequence of improvements in ultrasound technology, and with the advent of transvaginal sonography, identification of fetal gender starting in the first trimester has been suggested. The most important clinical application of determining fetal gender is in cases with family histories of X-linked disorders such as hemophilia and Duchenne muscular dystrophy. Such conditions are currently diagnosed using chorionic villus sampling (CVS), which is usually carried out after 11 weeks’ gestation. Non-invasive techniques to determine fetal gender in the first trimester need to be more thoroughly investigated in order to avoid fetal losses.

Although gender has been correctly diagnosed by sonography in the first trimester, it is important to establish the accuracy of detecting fetal gender prior to implementation in clinical practice. Should male gender be suggested accurately by sonography in the first trimester, then prenatal diagnosis can be offered at an earlier stage to confirm these findings, allowing the option of an early versus late termination of pregnancy with resulting decrease in surgical complications and psychiatric morbidity. On the other hand, if female gender is suggested, then it may be possible to avoid invasive testing. However, accuracy would need to approach 99% to compare with invasive testing. The aim of this study was to assess the success rates of the sonographic determination of fetal gender in the first trimester.

METHODS

A total of 524 women attending a routine first-trimester scanning clinic were recruited for this study. Scans were arranged by midwives as part of ongoing research into first-trimester detection of fetal anomalies. The midwives were instructed to arrange the scan for 12–13 weeks. There were two elective abortions (for aneuploidy), one intrauterine death and another 74 cases in which the neonatal outcomes could not be obtained. These cases were excluded from the study. Thus, a total of 447 fetuses were included, for which gender assignment was obtained from the neonatal records.

A detailed assessment of the fetal genitalia was attempted initially with transabdominal sonography (5.0-MHz Toshiba PVF-575 MT, Tokyo, Japan) using a combination of transverse and sagittal views. The fetal
male genitalia were identified in the transverse plane when a uniform, dome-shaped structure was seen at the base of the fetal penis (representing the fetal scrotum), a longitudinal, mid-line echogenic line was seen at the base of the fetal penis (representing the median penile raphe) and, in the sagittal plane, the cranial/vertical direction of the fetal phallus was also considered to be a feature of the male fetus/phallus (Figure 1). The female genitalia were identified in the transverse plane by visualizing two or four parallel lines, representing the labia majora and minora, and in the sagittal plane by the caudally directed phallus (considered to represent the clitoris, see Figure 2).

Transvaginal sonography (6.0-MHz Toshiba PVF-621 VT) was employed if visualization of fetal genitalia was not adequate with transabdominal sonography (26%). The scans were performed by one of six clinicians or one of six ultrasonographers who were trained in assessing fetal anatomy and nuchal translucency in the first trimester. The total scanning time was limited to 30 min, with the majority of scans completed within 10 min, and included measurement of nuchal translucency and assessment of fetal anatomy.

Gestational age was calculated from crown–rump length in conjunction with menstrual or ovulation dating. Approval from the Ethics Committee of the hospital was obtained for this study.

Statistical analysis was performed using the Minitab© statistical package; \( \chi^2 \) and logistic regression analyses were performed to determine statistical difference in the results. Fisher’s exact test was used when any of the single tabular values were < 5.

RESULTS

The overall success of correctly identifying fetal gender increased with gestational age from 46% to 75%, 79% and 90% at 11, 12, 13 and 14 weeks, respectively. The ability to assign fetal gender confidently using ultrasonography significantly improved with increasing gestational age from 11 to 14 weeks (\( p < 0.0001 \), Table 1 and Figure 3).

There was no gender-specific significant difference in assigning fetal gender with respect to gestational age (11 weeks, \( p = 0.2 \); 12 weeks, \( p = 0.3 \); 13 weeks, \( p = 0.2 \); 14 weeks, \( p = 0.4 \)), or in the whole group of fetuses (\( p = 0.3 \)). This was confirmed by logistic regression (\( p = 0.8 \)). The number of male and female fetuses in which gender was not

Figure 1  Male fetus in sagittal section at 11 weeks (a). Note the vertical/cranially directed phallus (arrow). Male fetus in transverse section at 12 weeks (b). Note the dome-shaped scrotum in the transverse plane (arrow)

Figure 2  Female fetus in sagittal section at 11 weeks (a). Note the caudally directed phallus in sagittal section (arrow). Female fetus in transverse section at 12 weeks (b). Note the triple parallel lines representing the labia in transverse plane (arrow)
Fetal gender at 11–14 weeks

Table 1 Fetal gender determination in relation to gestational age

<table>
<thead>
<tr>
<th>Gender not assigned</th>
<th>11 weeks</th>
<th>12 weeks</th>
<th>13 weeks</th>
<th>14 weeks</th>
</tr>
</thead>
<tbody>
<tr>
<td>n</td>
<td>%</td>
<td>n</td>
<td>%</td>
<td>n</td>
</tr>
<tr>
<td>Correct male</td>
<td>10/15</td>
<td>67</td>
<td>13/155</td>
<td>8</td>
</tr>
<tr>
<td>Incorrect male</td>
<td>3/15</td>
<td>33</td>
<td>8/74</td>
<td>11</td>
</tr>
<tr>
<td>Correct female</td>
<td>23/30</td>
<td>83</td>
<td>57/69</td>
<td>83</td>
</tr>
<tr>
<td>Incorrect female</td>
<td>5/30</td>
<td>17</td>
<td>12/69</td>
<td>17</td>
</tr>
<tr>
<td>Total correct when attempted</td>
<td>35/45</td>
<td>78</td>
<td>123/143</td>
<td>86</td>
</tr>
<tr>
<td>Total incorrect when attempted</td>
<td>10/45</td>
<td>22</td>
<td>20/143</td>
<td>14</td>
</tr>
<tr>
<td>Overall correct (% of total fetuses)</td>
<td>35/76</td>
<td>46</td>
<td>123/165</td>
<td>75</td>
</tr>
</tbody>
</table>

DISCUSSION

The overall success of correctly identifying fetal gender at 12–14 weeks was found to be 80%. The successful identification of fetal gender increased with gestational age and was highest (90%) at 14 weeks. The ability of the operator to assign male/female genitalia was also found to increase with advancing gestational age. The male or female phenotype did not have a significant effect in assigning fetal gender or in the accuracy of identification. These results are similar to those of other investigators, who found an overall success rate of 85% and 77% between 12 and 14 weeks, using transabdominal sonography. They are also in agreement with earlier work by Bronshtein and colleagues, who found an overall success rate at 13–14 weeks of just under 80% using transvaginal sonography.

A number of factors can affect the correct identification of fetal gender. First, the motivation and skill of the operator are paramount when making confident identification of fetal gender in the first trimester. Second, the resolution of the transvaginal transducer is likely to be superior to a transabdominal transducer. Although transvaginal sonography has been shown to be acceptable to the majority of low-risk women, transabdominal sonography does not involve extra staff (i.e. chaperones). Factors that could inhibit adequate visualization of fetal genitalia include fetal hyperactivity, crowding of the fetal genitalia (legs crossed/cord between legs), unfavorable fetal position, maternal bowel gas shadowing and maternal obesity. Maternal obesity is not, however, a problem when transvaginal sonography is used.

The significant improvement in the confidence of the operators to identify fetal gender with advancing gestational age may be explained by the development and embryology of the fetus. At 6 weeks, the external genitalia of the sexes are similar, and it is impossible to differentiate the two. At this stage, they consist of genital swellings on either side of the genital tubercle and the urethral folds. In the male fetus, the genital tubercle elongates between 6 and 13 weeks, at which time the urethral folds close over the urethral plate to form the penile urethra. During this stage, the urethral tubercle is known as the phallus. It is interesting to note that the genital swellings, which in the male give rise to the scrotum, may be confused with the labia minora and majora (which in the female fetus also develop from the genital swelling).

In a transverse plane at 11–14 weeks, the genital swellings form a dome-shaped scrotum in the male fetus and three or four parallel lines (i.e. the labia). Some investigators reported that a transverse view of the genitalia was more helpful in gestations between 12 and 14 weeks. This is contrary to other investigators, who have found that the direction of the phallus is more helpful in the early ultrasonographic determination of fetal gender. It has been suggested that the direction of the phallus may be vertical/cranially directed in the male fetus because the penile corpora cavernosa are permanently congested and therefore the penis is in a constant erect state.

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Early determination of fetal gender could be of clinical relevance in cases of a positive family history of X-linked disorders such as hemophilia, Duchenne muscular dystrophy and chronic granulomatous disease, possibly avoiding invasive diagnostic techniques with the inevitable small risk of miscarriage. Determining gender in the fetus at mid- and late gestations is probably of little use, apart from management of labor in carriers of hemophilia, in whom invasive procedures and instrumental deliveries should be avoided for potentially affected males\(^5\). Some early studies suggested that the determination of a male fetus might help to enhance pulmonary maturity by the use of steroids\(^1\); however, other investigators showed no difference in such outcomes with regard to fetal gender.\(^17\)

The subtle differences in the growth patterns of male and female fetuses are also so small that determination of gender in order to use separate biometrical reference ranges are impractical in the clinical setting. The level of \(\alpha\)-fetoprotein is significantly lower in female fetuses with Down’s syndrome than their male counterparts, although again this is likely to be of little clinical value.\(^18\) In one study, it was suggested that toxemia may be frequently associated with the male fetus, but again this would be unlikely to alter obstetric management.\(^19\)

Other methods of determining fetal gender include the examination of fetal DNA from the maternal circulation, and this may achieve better sensitivity (94% vs. 67%) in the first trimester compared to the second trimester.\(^20\) Indeed, Sekizawa and co-workers have even developed a method enabling the prenatal diagnosis of Duchenne muscular dystrophy using a single fetal nucleated erythrocyte in maternal blood. Cervical washings and analysis using fluorescent in situ hybridization have been shown to have a sensitivity for detecting a male fetus of around 83%, and this is no better than using ultrasonography. Therapeutic techniques in which X- and Y-chromosome-bearing spermatozoa can be separated by flow cytometry are also being studied, and early results are promising.\(^22\) This may enable intrauterine insemination of X-chromosome-carrying sperm from the husband/partner, so avoiding the need for prenatal diagnosis of fetal gender.

The social implications of antenatal gender identification need to be remembered. In developed countries, it has been shown that the majority (74.7%) of women are in favor of knowing the ultrasonographic interpretation of fetal gender, and the accuracy rate at the 20-week scan is around 97%.\(^3\) However, in developing countries, fetal gender should not be examined as a matter of routine, because of the lack of skilled/available manpower, making routine fetal gender determination impractical.\(^6\)

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**REFERENCES**