Polyhydramnios: A warning sign in the prenatal ultrasound diagnosis of foetal malformation?

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KEYWORDS
Polyhydramnios; Foetal malformation; Prenatal diagnosis; Obstetric ultrasonography

Abstract
Purpose: To propose polyhydramnios seen during prenatal diagnosis as a warning sign of foetal malformation.

 Patients and methods: A retrospective multicentre study over a three-year period carried out in Ivory Coast and Burkina Faso. We reviewed 3903 obstetric ultrasound reports. All cases of foetal malformation and polyhydramnios were counted. The instances of foetal malformation associated with polyhydramnios were compared to those of foetal malformation without polyhydramnios and to polyhydramnios only.

 Results: A list of 72 cases of polyhydramnios was made (equating to 1.8%). In 55 cases (76.4%), polyhydramnios was combined with foetal malformation. These were lethal abnormalities in 33 cases and non-lethal in 22 cases. In 17 cases, polyhydramnios was not associated with any foetal malformations and in eight cases, foetal malformation was discovered in the absence of polyhydramnios. Polyhydramnios had a positive predictive value of 76.4% for the presence of foetal malformation. The negative predictive value was 99.8%. Sensitivity was 87.3% and specificity was 99.5%.

 Conclusion: Polyhydramnios is a highly sensitive and specific sign for prenatal diagnosis of foetal malformation. If it is identified, then this should lead to a very careful search for foetal malformation.

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Ultrasonography has become a crucial investigation during pregnancy because it is harmless, easily available, and inexpensive, and it has entered into the day-to-day practice of specialists in reproductive health [1]. The investigation has different objectives depending on the stage of pregnancy. According to the literature, at least three ultrasound scans should be carried out during a normal pregnancy [2]. The one carried out in the first trimester is diagnostic and for dating the pregnancy while the purpose of the one in the third trimester is thought to be useful for establishing a prognosis for vaginal delivery. Between these two, the second trimester ultrasound scan is used to look for foetal malformations. However, it has not always proved easy to detect foetal malformations and a good number of these have remained unseen in spite of repeated ultrasound scans [3,4]. In these cases, the alpha-fetoprotein test, amniocentesis, or even magnetic resonance imaging are complementary investigations that are needed and often requested in addition to sonography [5]. These additional investigations are expensive and are not always available in all countries. This means that the morphology ultrasound scan remains the only effective examination for prenatal screening of severe foetal malformations (those that require medical termination of pregnancy) or minor foetal abnormalities (for which the parents will need to be psychologically prepared). For this reason, it is important to simplify the protocol for the ultrasound scan during pregnancy by establishing direct and/or indirect signs that should alert the sonographer to the possibility of foetal malformation. One of these signs is, in our view, polyhydramnios. It is defined as an excess of amniotic fluid and there are a number of known causes. Furthermore, its frequent association with foetal malformations has often been observed in our practice and in the literature [6–8].

The purpose of this article is to propose polyhydramnios as a warning sign of foetal malformation to be used in the sonographic prenatal diagnosis of these abnormalities.

Patients and methods

Our retrospective multicentre study was carried out over 3 years (from January 2009 to December 2011). It took place in Ivory Coast (Yopougon University Hospital, Naglé Clinic and Yamoussoukro Regional University Hospital) and in Burkina Faso (Soro-Sanou University Hospital and Saint Léopold Clinic in Bobo-Dioulasso). It involved reviewing reports from obstetric ultrasound scans carried out during the study period in these different centres. We were interested in all cases of polyhydramnios and foetal malformation detected on sonography during the second (2T) and third (3T) trimesters. The cases of polyhydramnios found together with foetal malformations were compared to those of isolated foetal malformations and to cases of polyhydramnios only. We calculated the positive predictive value, the negative predictive value, the sensitivity, and the specificity of polyhydramnios in prenatally diagnosing foetal malformations. The method proposed by Chamberlain et al. [9] was used to identify polyhydramnios (Fig. 1), which involves measuring the vertical diameter of the largest fluid pocket, as was the amniotic fluid index (AFI) used by Phelan et al. [10], which is based on the measurements of all four quadrants. Any pocket of amniotic fluid greater than or equal to 80 mm and/or any AFI greater than or equal to 250 mm was considered to be polyhydramnios. There was said to be moderate polyhydramnios if the depth of the fluid pocket was below 160 mm and the AFI was below 350 mm. If found to be in excess of these figures, polyhydramnios was considered to be severe (Fig. 1). Foetal malformations were classified as minor morphologic abnormalities (viable) and major malformations (lethal). Malformations seen in the first trimester were excluded from our study because we were interested in cases in which first trimester sonography was either not carried out or was suboptimal in terms of early screening for malformations, in contrast to the good practice recommendations in Western nations [11,12]. Epidemiological data (age, sex, trimester of pregnancy), the reason for the ultrasound scan, and the results that we made use of were marked on the sonogram reports. The post-natal or post-medical termination of pregnancy outcome was identified from the birth registers of the various maternity wards and the Neurosurgery and Paediatric surgery departments of the hospitals involved. Data was processed using the software packages Word and Epi-info 6.0.

Figure 1. (a, b): severe polyhydramnios revealed using the method of Phelan et al. [10] (amniotic fluid index measurement = 671 mm) in a 25-year-old patient in the second trimester of pregnancy.
Polyhydramnios and prenatal diagnosis of foetal malformation

Table 1 Distribution of cases of polyhydramnios according to their severity and the method by which amniotic fluid was quantified.

<table>
<thead>
<tr>
<th>Method</th>
<th>Largest amniotic fluid pocket (Chamberlain)</th>
<th>Phelan’s amniotic fluid index</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Polyhydramnios</td>
<td>Moderate (8–16 cm)</td>
<td>Severe (≥ 16 cm)</td>
<td></td>
</tr>
<tr>
<td>Number</td>
<td>9</td>
<td>51</td>
<td>72</td>
</tr>
<tr>
<td>Percentage</td>
<td>12.5</td>
<td>70.8</td>
<td>100</td>
</tr>
<tr>
<td></td>
<td>Moderate (25–35 cm)</td>
<td>Severe (≥ 35 cm)</td>
<td></td>
</tr>
<tr>
<td></td>
<td>4</td>
<td>8</td>
<td></td>
</tr>
<tr>
<td></td>
<td>5.6</td>
<td>11.1</td>
<td></td>
</tr>
</tbody>
</table>

Results

Our results are summarised in Tables 1–3.

While the study was underway, 3903 obstetric ultrasound scans were carried out in women in their second and third trimesters. Among these, 72 cases of polyhydramnios (equating to 1.8%) and 63 instances of foetal malformation (1.6%) were identified. The mean age of the patients was 23.6 years with extreme values of 19 and 40 years. These sonograms were conducted in the second trimester in 37 cases (51.4%) and the third trimester in 35 cases (48.6%). In 60 of these cases (83.3%), polyhydramnios was determined using the Chamberlain method with a mean amniotic fluid pocket depth of 157.8 mm. The Phelan method was only used in 12 cases (16.7%). The mean of the AFI was 335 mm. In 81.9% of cases, polyhydramnios was found to be severe, and in 18.1% of cases, it was moderate. In 55 cases (76.4%), polyhydramnios was associated with foetal malformation. In 33 cases (60%), these were major foetal abnormalities, while they were minor abnormalities in 22 cases (40%). The major abnormalities included 32 cases of anencephaly (Figs. 2 and 3) and one case of thanatophoric dysplasia. The minor abnormalities were eight cases of myelomeningocele, five cases of spina bifida, seven abdominal masses (three renal masses, three cases of omphalocele, one case of duodenal atresia) and two cases of hydrocephalus. There were 17 cases of polyhydramnios that were not associated with a foetal malformation and eight cases in which a foetal malformation was discovered without being associated with polyhydramnios (four cardiac abnormalities, one omphalocele, two cases of cleft lip and palate, and one of achondroplasia). The positive predictive value of polyhydramnios for a foetal malformation was 76.4%. The negative predictive value was 99.8%. Sensitivity was 87.3% and specificity was 99.5%. Because there had been no patient consent, no medical termination of pregnancy was practised in the cases of major foetal malformation. There were 16 cases (48.5%) of foetal death in utero, four cases (12.1%) of vaginally delivered anencephalic neonates (Fig. 3) and one case

Table 2 Distribution of foetal malformations according to the severity of polyhydramnios.

<table>
<thead>
<tr>
<th>Polyhydramnios</th>
<th>Foetal malformation identified</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Number</td>
<td>Percentage</td>
</tr>
<tr>
<td>Moderate</td>
<td>5</td>
<td>7.9</td>
</tr>
<tr>
<td>Severe</td>
<td>58</td>
<td>92.1</td>
</tr>
<tr>
<td>Total</td>
<td>63</td>
<td>100</td>
</tr>
</tbody>
</table>

Table 3 Significance of polyhydramnios in the prenatal identification of foetal malformations.

<table>
<thead>
<tr>
<th>Malformation</th>
<th>No malformation</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Polyhydramnios</td>
<td>55</td>
<td>17</td>
</tr>
<tr>
<td>No polyhydramnios</td>
<td>8</td>
<td>3823</td>
</tr>
<tr>
<td>Total</td>
<td>63</td>
<td>3840</td>
</tr>
</tbody>
</table>


Figure 2. Same patient, ultrasound scan shows a severe foetal malformation: anencephaly.

Figure 3. Anencephalic newborn delivered vaginally after the mother refused medical termination of pregnancy.
(3%) of a vaginally delivered neonate with thanatophoric dysplasia. The other 12 cases of major foetal malformation (36.4%) were lost to follow-up. All of the minor foetal malformations identified were confirmed post-natally. The neonates were managed in the Paediatric Surgery and Neurosurgery departments.

Discussion
According to the literature, polyhydramnios complicates between 0.5 and 3% of pregnancies [13]. In our study, it was identified in 1.8% of cases. It is diagnosed by clinical examination and sonography. Clinically, there is a rapid increase in fundal height and tension of the uterine walls. On abdominal palpation, the fluid wave sign is seen. On vaginal examination, the foetal head may present when subjected to movement [2]. There are several possible causes of polyhydramnios as pregnancy progresses. These causes are known, and include diabetes, rhesus isoimmunisation, chromosome abnormalities, foetal anaemia, viral infections (toxoplasmosis, rubella, CMV, herpes etc.) and foetal malformations [13]. The most serious of these causes is foetal malformation, which can lead to psychological difficulties for the parents and perinatal morbidity/mortality or severe psychomotor impairment in the child [14]. It is therefore necessary to carry out effective prenatal screening for these malformations, with a view to psychologically preparing parents and managing minor abnormalities, as well as carrying out medical termination of pregnancy in major foetal malformations. However, checking for foetal malformations, and especially minor abnormalities, is a difficult process that requires a trained, qualified and, above all, highly focused technician. These criteria are not always met since, according to Lawrence et al. [3], 2 to 3% of neonates are born with an abnormality that would have been detectable on sonography. Dashe et al. [4] found in their study that 11% of malformations were not detected on prenatal sonography. A sign that is easier to detect on sonography and that is often connected to foetal malformation is polyhydramnios. According to Cabrol et al. [13] sonography is the gold standard examination for the positive diagnosis of polyhydramnios. It consists of a subjective assessment (impression that the foetus appears to be swimming) or an objective evaluation (depth of the largest amniotic fluid pocket or the AFI). In our study, in 55 cases (76.4%), polyhydramnios was associated with foetal malformation. The positive predictive value, negative predictive value, sensitivity, and specificity for polyhydramnios when foetal malformation was present were all very high. Specifically, they were 76.4%, 99.8%, 87.3% and 99.5% respectively. Our results are in agreement with those of Dashe et al. [4] who brought together a retrospective series of 672 cases of polyhydramnios, with a 79% rate of foetal malformation. They differ, however, from those of Kouakou et al. [6] who found a positive predictive value of 42.6% in a study that, like ours, was carried out in Ivory Coast. The difference can be explained by the fact that in this study (which was based on the physical examination of neonates), there was a foetal death rate of 44.4% and Kouakou et al. [6] did not state whether the cause of this foetal mortality was connected to major foetal abnormality. According to Perthus et al. [15], foetal malformations cause in excess of 20% of foetal deaths. In our study, 48.5% of the major foetal malformations led to in utero foetal death. In Israel, Golan et al. [8] identified a positive predictive value of 14.2%. Although this is a major difference, the author did draw the attention of sonographers to the association between polyhydramnios and congenital malformations. There appears to be a higher incidence of foetal malformation associated with polyhydramnios in Africa in contrast to that which is seen in developed countries due to infectious diseases (rubella, toxoplasmosis etc.) and poverty (folic acid supplements are not used and pregnancy is inadequately monitored with sonography either not carried out at all or only a single obstetric ultrasound scan in the second or third trimester). This lack of first trimester sonography explains the presence of neural tube defects identified in the second and third trimesters of pregnancy in our study. This difference could also be explained by the severity of the cases of polyhydramnios found in our study. According to the literature, the risk of foetal malformation is directly correlated to the extent of polyhydramnios. In a series of 105 cases of polyhydramnios, Damato et al. [16] identified malformations in 63% of these but while the rate was 50% when including women whose deepest fluid pocket was between 8 and 9.5 cm, it rose to 88% when including only cases of fluid pockets in excess of 16 cm. In our study, 81.9% of the cases of polyhydramnios we saw were severe and the foetal malformations observed were neural tube defects (anencephaly, spina bifida, hydrocephalus etc.). Abnormalities of the central nervous system, according to the literature, are the most common cause of malformation encountered in polyhydramnios [7,17,18]. Excess amniotic fluid is thought to be caused by fluid production through the meninges, potentially combined with reduced loss of amniotic fluid because the foetus has difficulty swallowing. In our study, the patients whose foetus had a major abnormality refused medical termination of pregnancy and 36.4% of them were lost to follow-up. This observation shows that it is essential to offer suitable psychological management for women whose pregnancies present foetal malformation [19]. This was not so for the population in our study.

Conclusion
Foetal malformation may be rare (1.6% in our study), but it is a sufficiently serious phenomenon to warrant precise and early prenatal diagnosis. To this end, medical imaging now enhanced by the use of foetal MRI can complement laboratory tests, while occupying a predominant place in screening. Obstetric sonography is a dedicated routine examination. Although it is operator-dependent, consistency should be improved by incorporating the sign of polyhydramnios into the screening for foetal malformations. Our study demonstrates that polyhydramnios has very high sensitivity, specificity, and positive and negative predictive values when foetal malformations are present. If polyhydramnios is seen on obstetric sonography, this should lead to sonographer to look routinely and extremely carefully for any foetal malformations, especially in countries where there is a high prevalence of infectious diseases with neural cell tropism, such as toxoplasmosis and rubella.
We propose that polyhydramnios should be considered to be a warning sign for the sonographer that prompts them to look extremely closely for foetal malformations that have not been detected in the first trimester of pregnancy.

Disclosure of interest

The authors declare that they have no conflicts of interest concerning this article.

References