Incidence and Sonographic Detection of Fetal Anomalies in Pregnancies Complicated by Polyhydramnios

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OBJECTIVE: The purpose of this study was to evaluate the incidence and sonographic detection of fetal anomaly related with polyhydramnios and to estimate the relationship between fetal anomaly and the severity of polyhydramnios and to compare the idiopathic polyhydramnios with polyhydramnios associated with fetal anomaly.

STUDY DESIGN: This was a prospective study of 60 singleton pregnancies with polyhydramnios. Though all singleton pregnancies with polyhydramnios were included, only the patients with idiopathic polyhydramnios or with fetal anomalies were the object of this study. Idiopathic polyhydramnios was defined as polyhydramnios that is not associated with congenital anomalies, fetal chromosomal abnormalities, fetal infections, maternal diabetes, placental tumors or isoimmunization. Polyhydramnios was defined as 25 cm or greater amniotic fluid index. Polyhydramnios was categorized as mild (amniotic fluid index of 25-29.9 cm) and severe (amniotic fluid index of 30 cm or more). The prevalence of fetal anomalies was analyzed according to the severity of polyhydramnios.

RESULTS: Polyhydramnios was diagnosed in 60 patients. Of the 60 patients, 33 were considered to have idiopathic polyhydramnios (55%), and of the remaining, 22 patients (37%) were associated with fetal anomalies. There was statically significant difference between these two groups in respect to amniotic fluid index. In patients with fetal anomaly, amniotic fluid index was significantly higher than in patient with idiopathic polyhydramnios (p=0.001). In 55 patients, mild polyhydramnios was observed in 43 pregnancies (78%) and severe polyhydramnios was noted in 12 pregnancies (22%). The prevalence of congenital anomaly was increased significantly with the severity of polyhydramnios (p=0.001). The number of fetal anomaly was 11 (25%) in mild polyhydramnios group, 11 (91%) in severe polyhydramnios group.

CONCLUSION: It is reasonable to distinguish between mild and severe polyhydramnios regarding special attention and antenatal follow-up as fetal anomaly is related to the degree of polyhydramnios. Significant risk of fetal anomalies should be considered in patients with severe polyhydramnios (AFI≥ 30 cm).

Key Words: Polyhydramnios, Fetal anomalies, Ultrasonography

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Introduction

Polyhydramnios is defined as an excess of amniotic fluid. It is a relatively common finding in pregnancy and complicates approximately 0.4% to 3.3% of all pregnancies. The etiology of polyhydramnios is multiple, involving both maternal and fetal conditions. Congenital fetal anomalies constitute one of the important etiologic factors associated with polyhydramnios. The other fetal conditions linked with polyhydramnios are isoimmunization, fetal infections (CMV, toxoplasmosis, parovirus, and syphilis), multiple gestation and placental tumors. Up to 60% of cases of polyhydramnios are idiopathic. The gestational or pregestational diabetes are the two reasons of maternal polyhydramnios. Here, we consider only idiopathic polyhydramnios and polyhydramnios associated with fetal anomaly. We aimed to estimate the association between fetal anomaly and the degree of polyhydramnios.

Material and Method

This was a prospective study of singleton pregnancies with antepartum diagnosis of polyhydramnios between April 1, 2005, and through January 30, 2007 at the ultrasound unit in the Department of Obstetrics and Gynecology at the Ondokuz Mayis University. During this period, 2640 pregnant women with living fetuses were evaluated between 20 and 42 weeks of gestation. Though all singleton pregnancies with polyhydramnios were included, only the patients with idiopathic polyhydramnios were analyzed according to the severity of polyhydramnios.

RESULTS: Polyhydramnios was diagnosed in 60 patients. Of the 60 patients, 33 were considered to have idiopathic polyhydramnios (55%), and of the remaining, 22 patients (37%) were associated with fetal anomalies. There was statically significant difference between these two groups in respect to amniotic fluid index. In patients with fetal anomaly, amniotic fluid index was significantly higher than in patient with idiopathic polyhydramnios (p=0.001). In 55 patients, mild polyhydramnios was observed in 43 pregnancies (78%) and severe polyhydramnios was noted in 12 pregnancies (22%). The prevalence of congenital anomaly was increased significantly with the severity of polyhydramnios (p=0.001). The number of fetal anomaly was 11 (25%) in mild polyhydramnios group, 11 (91%) in severe polyhydramnios group.

CONCLUSION: It is reasonable to distinguish between mild and severe polyhydramnios regarding special attention and antenatal follow-up as fetal anomaly is related to the degree of polyhydramnios. Significant risk of fetal anomalies should be considered in patients with severe polyhydramnios (AFI≥ 30 cm).

Key Words: Polyhydramnios, Fetal anomalies, Ultrasonography

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polyhydramnios or with fetal anomalies were the object of this study. Idiopathic polyhydramnios was defined as polyhydramnios that is not associated with congenital anomalies, fetal chromosomal abnormalities, fetal infections, maternal diabetes, placental tumors or isoimmunization. Five patients were excluded from the study. So the study population consisted of 55 consecutive patients with singleton pregnancies, who were idiopathic polyhydramnios or polyhydramnios with congenital anomalies. Subjects were enrolled into the study only after undergoing a normal 1-hour, 50-gr glucose challenge test and/or a normal 3-hour, 100 mg oral glucose tolerance test, using the thresholds of the National Diabetes Data Group. From all study population, TORCH infections and Treponema Pallidum Hemagglutination Test (TPHA) for syphilis were observed by serum tests. All scans were obtained with a real time sector scanner and with the use of 3.5 or 5.0 MHz transducer (Aloka SSD 3500). We used the amniotic fluid index (AFI) for amniotic fluid volume assessment. We assessed the volume by directly measuring the amniotic fluid at the largest vertical pocket in each quadrant of the pregnant abdomen. To measure the amniotic fluid correctly, the transducer was maintained in a position both perpendicular and parallel to the floor. After diagnosis of polyhydramnios, targeted sonography was performed in all cases.

We defined the polyhydramnios as an amniotic fluid index greater than 24 cm, which is ≥97.5 percentile in normal singleton pregnancies. Polyhydramnios was subdivided into categories of severity with mild defined as 25 cm ≤ AFI < 30 cm and severe defined as AFI ≥30 cm. The prevalence of fetal anomalies was analyzed according to the severity of polyhydramnios.

Statistical analyses were performed by x²-test, Student-Newman-Keuls test (NCSS 2007 & PASS 2008 Statistical Software, Utah, USA). Differences between groups were considered significant, when p<0.05.

Results

Among 2640 pregnant women that we conducted ultrasonographic study and prenatal management, 60 patients met the criteria for polyhydramnios. In our population, the incidence of polyhydramnios was estimated as 2.2%. Of the 60 patients with polyhydramnios, 33 were considered to have idiopathic polyhydramnios (55%), and of the remaining, 22 patients (37%) were associated with fetal anomalies. All women with idiopathic polyhydramnios or fetal anomaly had carried serum tests of TORCH infections and Treponema pallidum hemagglutination test (TPHA) for syphilis. One patient had toxoplasma infection. Four patients were diagnosed as gestational diabetes mellitus. So these 5 patients with polyhydramnios were excluded from the study (4 patients with gestational diabetes and 1 patient with toxoplasmosis).

Maternal age ranged from 19 to 44 years with a median age of 27±6 years. It was 28±5 years for idiopathic polyhydramnios group and 27±6 years for fetal anomaly group. There was no difference between patients with idiopathic polyhydramnios and patient with fetal anomaly groups in regard to age (p=0.624). The mean gestational age at the time of antenatal diagnosis of polyhydramnios was 30.8±5 weeks in fetal anomaly group and 30.3±5 weeks in idiopathic polyhydramnios group. Gestational age at which polyhydramnios was detected did not differ between idiopathic polyhydramnios and patient with fetal anomaly groups (p=0.71).

The mean amniotic fluid index was 24.5±1.2 cm in idiopathic polyhydramnios group and 30.2±5.6 in polyhydramnios with fetal anomaly group. There was statically significant difference between these two groups in respect to amniotic fluid index. In patients with fetal anomaly, amniotic fluid index was significantly higher than in patient with idiopathic polyhydramnios (p=0.001). In 55 patients, mild polyhydramnios was observed in 43 pregnancies (78%) and severe polyhydramnios was noted in 12 pregnancies (22%). The mean gestational age at the time of prenatal ultrasonography was 30±5 weeks and did not differ between two groups (p=0.844). There was not significant difference in maternal age and gravidity between these two groups (p=0.624 and p=0.791 respectively). The prevalence of congenital anomaly was increased significantly with the severity of polyhydramnios (p=0.001). The number of fetal anomaly was 11 (25%) in mild polyhydramnios group, 11 (91%) in severe polyhydramnios group (Figure 1). 30 anomalies were detected in 22 fetuses by sonographically. There were 15 (68%) cases with only one anomaly, and 7 (32%) cases with more than one anomaly. Aneuploidy was seen in only one fetus (trisomy 18) among 22 fetuses (4.5%).

Figure 1: The rate of fetal anomaly according to severity of polyhydramnios
The fetal anomalies associated with polyhydramnios were listed in Table 1 according to the organ system involved.

Sonographic detection of fetal anomalies according to degree of polyhydramnios and the relative risk of congenital malformations for severe polyhydramnios were given in Table 2.

Table 1: Sonographic detection of fetal abnormalities in association with polyhydramnios

<table>
<thead>
<tr>
<th>Central Nervous System</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>Spina Bifida</td>
<td>5</td>
</tr>
<tr>
<td>Meningomyelocele</td>
<td>2</td>
</tr>
<tr>
<td>Dandy Walker Malformation</td>
<td>3</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Gastrointestinal</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>Omphalocele</td>
<td>1</td>
</tr>
<tr>
<td>Duodenal Atresia</td>
<td>2</td>
</tr>
<tr>
<td>Esophageal Atresia</td>
<td>3</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Cardiac</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>Pulmonic Stenosis</td>
<td>1</td>
</tr>
</tbody>
</table>

| Thorax                 |       |
|                        |       |

| Fetal hydrops          |       |
| Non-immun Hydrops Fetalis | 7   |

| Musculoskeletal        |       |
| Single Umbilical Artery| 2     |
| Sacral Teratoma        | 1     |

Table 2: Incidence of fetal anomalies in relation to the severity of polyhydramnios and relative risk of congenital malformations for severe polyhydramnios

<table>
<thead>
<tr>
<th>AFI (cm)</th>
<th>Yes n=22</th>
<th>No n=33</th>
<th>*p</th>
</tr>
</thead>
<tbody>
<tr>
<td>25-29.9</td>
<td>11 (25.6%)</td>
<td>32 (74.40%)</td>
<td>0.001**</td>
</tr>
<tr>
<td>≥ 30</td>
<td>11 (91.7%)</td>
<td>1 (8.3%)</td>
<td>3.583 (2.09-6.13)</td>
</tr>
</tbody>
</table>

**p < 0.01 is significant  *: X^2-test; Relative Risk (95% Confidence Interval): 3.583 (2.09-6.13) **

Discussion

Ultrasoundographic assessment of amniotic fluid poses important implications in assessing normal fetal development. For many clinicians, the question becomes whether the polyhydramnios is a sign of abnormality or it is simply idiopathic. Up to 60% of cases of polyhydramnios are idiopathic. Congenital fetal anomalies constitute one of the important etiologic factors associated with polyhydramnios and have an influence on the management and prevalence of adverse pregnancy outcomes. The other conditions that cause polyhydramnios are diabetes, Rh immunization, multiple gestation, placental abnormalities, and infections like CMV, toxoplasmosis, parvovirus, and syphilis. In literature, the most commonly reported congenital anomalies are CNS defects, gastrointestinal defects, cardiovascular defects and urinary system defects.

In our study, central nervous system (CNS) anomalies were the most commonly seen congenital anomaly, accounted for 33% of the all fetus with anomalies. The mechanisms of polyhydramnios vary in fetuses with fetal anomalies. In fetuses with CNS anomalies, the ability of swallowing is impaired. Gastrointestinal anomalies obstruct the flow of fluid through the bowel.

The amount of amniotic fluid volume may be measured by 3 different methods. These methods are measurement of AFI or single deepest pocket, and the subjective assessment of the amniotic fluid volume. Most studies have used AFI as being most sensitive method for detecting polyhydramnios. In literature, polyhydramnios complicates approximately 0.4% to 3.3% of all pregnancies. In our population, the frequency of polyhydramnios was estimated as 2.2%.

The reported anomaly rate in pregnancies with polyhydramnios has ranged from 15% to 50%. The anomaly rate in pregnancies with polyhydramnios is higher in pregnancies referred with complications. In our study the incidence of fetal anomaly in patient with polyhydramnios was detected as 36%. Since our hospital is a central hospital for perinatal medicine in Karadeniz region, most of the patients were referred from other hospitals because of a considered perinatal risk for the fetus or the mother.

With regard to fetal anomalies, it has been observed that the more severe the polyhydramnios the greater the likelihood of a fetal anomaly. We also correlated the severity of polyhydramnios with the prevalence of fetal anomalies. In patients with mild polyhydramnios, fetal anomalies are seen in 12 of 43 patients (35%). In those with severe polyhydramnios, it was 11 of 12 patients (91%).

In literature, only Lazenbik et al who estimated that severe polyhydramnios increases the congenital malformation risks 5.2 times compared with mild polyhydramnios. In our study, the odds ratio for the severe polyhydramnios was estimated as 3.5 (with 95% Confidence Interval: 2.09-6.13) as compared with the mild polyhydramnios.

In conclusion, we have demonstrated that the amount of amniotic fluid volume is an effective diagnostic tool for detection of many severe fetal anomalies in routine ultrasonographic examination. The severity of amniotic fluid volume in polyhydramnios must be taken into account during fetal screening. Significant risk of fetal anomalies should be considered in patients with severe polyhydramnios (AFI ≥ 30 cm).
Polihidramniosun Eşlik Ettiği Gebeliklerde Fatal Anomali Sıklığı ve Ultrasonografik Tespiti

Bu çalışmamız amacımız, ultrasonografik olarak tespit edilen polihidramnios ve fetal anomali insidansını gözden geçirmek, polihidramniosun şiddetini ile fetal anomali arasındaki ilişkiyi tespit etmek ve fetal anomali ile birlikte görülen polihidramniosla idiopatik polihidramniosu karşılaştırmaktır.

Bu çalışma, 60 polihidramnioslu tekil gebeliği içeren prospektif bir çalışmaddır. Polihidramniosu olan tüm tek gebeler çalışmada değerlendirildi. Idiopatik polihidramnios, fetal konje-nital anomali, fetal kromozom anormallikleri, fetal enfeksiyon, maternal diabet, plasental tümörler veya izoimmünizasyon ile ilgili olmayan polihidramnios olarak tanımlandı. Amniotik sıvı indeksi (ASI) 25 cm ve üzeri ise polihidramnios olarak değer-landırıldı. Polihidramnios, hafif (ASI 25-29.9 cm) ve ağır (ASI ≥30 cm) fetal anomali görülme riskini belirgin olarak yüksekledi (p=0.001). Çalışmanın dahil edilen 55 hastanın 43’ünde (%78) polihidramniosunun şiddetine de belirgin bir yükselti (p=0.001) sızdırıldı. Hafif polihidramnios, 12’inde ağır polihidramnios (%22) tespit edildi. Konjenital anomali sıklığı ağır polihidramnios grubunda belirgin olarak yüksekken (p=0.001) hafif polihidramnios grubundaki fetal anomali sayısı 11 (%25) iken, ağır polihidramnios grubunda bu sayı 11 idi (%91).

Hafif ve ağır polihidramniosun birbirinden ayrılanı, antenatal izlem için anlamlı görülmektedir. Çünkü fetal anomalileri görülen polihidramniosunun derecesi ile ilgilidir. Ağır polihidramniosunun (ASI≥30 cm) fetal anomalileri riskini belirgin olarak artırıldığı anlamaktadır.

Anahtar Kelimeler: Polihidramnios, Fetal anomali, Ultrasonografi

References

20. Lazembik N, many A. The severity of polyhydramnios, estimated fetal weight and preterm delivery are independent risk factors for the presence of congenital malformations.