SHORT COMMUNICATION

PRENATAL DIAGNOSIS OF DIASTEMATOMYELIA: CASE REPORTS AND REVIEW OF THE LITERATURE

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SUMMARY

Diastematomyelia is a rare malformation characterized by complete or incomplete division of the spinal cord by osseous or fibrocartilaginous septum. Most cases are seen in association with other anomalies of the vertebral column such as spina bifida, kyphoscoliosis, butterfly vertebra, and hemivertebra. In this report we describe two cases of isolated diastematomyelia detected at routine second-trimester detailed ultrasound scan, the most striking feature being the detection of an echogenic focus in the posterior aspect of the spine in association with widening of the interpedicular vertebral space. The prenatal literature is reviewed to assess the clinical significance of this finding.

INTRODUCTION

Diastematomyelia is a rare form of spinal dysraphism characterized by splitting of the spinal cord, which results in a sagittal spinal cleft and enlargement of the spinal canal (Goldberg et al., 1984; Pang et al., 1992). This anomaly is frequently seen in association with defects of the vertebral column such as spina bifida, kyphoscoliosis, butterfly vertebra, and hemivertebra (Goldberg et al., 1984; Pang et al., 1992). In this report we present two cases of isolated diastematomyelia detected at routine second-trimester ultrasonography and review the prenatal literature to assess the clinical significance of this finding.

Case Reports

Case 1

A 28-year-old woman, gravida 4, para 3, underwent a routine scan at 21 weeks’ gestation. Her obstetric history included one neonatal death at 3 days due to hypoplastic left heart and one infant who died at 18 months from a rhabdomyosarcoma. Ultrasonographic examination revealed a singleton fetus with biometry consistent with dates, a high posterior placenta, and normal amniotic fluid volume. Examination of the fetal spine revealed a widening of the lumbar spine with an echogenic focus at the level of L3 without discontinuity of the skin (Fig. 1). No other anomalies were detected. Maternal serum a-fetoprotein levels were within the normal range and subsequent fetal echocardiography was reported as normal. The parents were counselled by a multiprofessional team regarding the implication of diastematomyelia and elected to continue the pregnancy.

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Follow-up ultrasonographic examinations at 23, 30, and 36 weeks showed normal fetal growth with no changes in the appearance of the spine. At 41 weeks, a female infant weighing 3834 g and with Apgar scores of 9 and 10 at 1 and 5 min, respectively, was delivered spontaneously. At neonatal examination there was a minor palpable abnormality at the level of L2/3. Radiographic studies showed widening of the interpedicular space in the lumbar region with an extra piece of bone in the spinal canal but no other spinal deformity. Paediatric follow-up showed no neurological dysfunction over a period of 18 months.

Case 2

A 32-year-old woman, gravida 2, para 1, was referred at 21 weeks’ gestation because of the suspicion of spina bifida occulta. Her family and obstetric history were unremarkable and the maternal serum α-fetoprotein level at 16 weeks was within the normal range. Ultrasonographic examination at referral revealed a singleton fetus with biometry consistent with dates, a high posterior placenta, and normal amniotic fluid volume. Examination of the fetal spine confirmed widening of the lower lumbar spine but intact overlying skin, and an echogenic focus at the level of L4 (Fig. 2). No other anomalies were detected and, in particular, there were no cranial or cerebellar signs suggestive of spina bifida (Nicolaides et al., 1986). At follow-up examination at 25 weeks, the appearances of the fetal spine remained unchanged. A 2608 g healthy male infant was delivered spontaneously at 36 weeks. Physical examination of the neonate showed a small lumbar dimple, and subsequent evaluations confirmed the prenatal diagnosis. No significant neurological
Table I—Prenatal diagnosis of diastematomyelia. Review of the literature

<table>
<thead>
<tr>
<th>Authors</th>
<th>Maternal age (years)</th>
<th>Gestational age (weeks)</th>
<th>Indication</th>
<th>Additional ultrasonographic findings</th>
<th>Pregnancy outcome</th>
</tr>
</thead>
<tbody>
<tr>
<td>Williams and Barth, 1985</td>
<td>26</td>
<td>23</td>
<td>Hydrocephaly</td>
<td>Spina bifida</td>
<td>TOP Liveborn, small foot, short leg, butterfly vertebra, spinal surgery</td>
</tr>
<tr>
<td>Winter et al., 1989</td>
<td>18</td>
<td>15</td>
<td>Routine</td>
<td>None</td>
<td>Liveborn, butterfly vertebrae, hemivertebrae, asymptomatic</td>
</tr>
<tr>
<td></td>
<td>21</td>
<td>20</td>
<td>Routine</td>
<td>None</td>
<td>TOP</td>
</tr>
<tr>
<td></td>
<td>19</td>
<td>16</td>
<td>Routine</td>
<td>None</td>
<td>TOP</td>
</tr>
<tr>
<td></td>
<td>23</td>
<td>18</td>
<td>Routine</td>
<td>None</td>
<td>TOP</td>
</tr>
<tr>
<td>Caspi et al., 1990</td>
<td>29</td>
<td>19</td>
<td>?Spina bifida</td>
<td>Spina bifida</td>
<td>TOP</td>
</tr>
<tr>
<td>Pachi et al., 1992</td>
<td>30</td>
<td>20</td>
<td>Maternal diabetes</td>
<td>Scoliosis</td>
<td>TOP</td>
</tr>
<tr>
<td>Boulot et al., 1993</td>
<td>25</td>
<td>33</td>
<td>Abnormal fetal spine</td>
<td>Spina bifida,* kyphosis</td>
<td>TOP</td>
</tr>
<tr>
<td>Anderson et al., 1994</td>
<td>NS</td>
<td>NS</td>
<td>Routine</td>
<td>None</td>
<td>TOP</td>
</tr>
<tr>
<td></td>
<td>NS</td>
<td>NS</td>
<td>Routine</td>
<td>None</td>
<td>Fetal demise 25 weeks</td>
</tr>
<tr>
<td></td>
<td>NS</td>
<td>NS</td>
<td>Routine</td>
<td>None</td>
<td>Liveborn, spinal surgery</td>
</tr>
<tr>
<td></td>
<td>NS</td>
<td>NS</td>
<td>Routine</td>
<td>None</td>
<td>Liveborn, asymptomatic</td>
</tr>
<tr>
<td>Current report</td>
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<td>21</td>
<td>Routine</td>
<td>None</td>
<td>Liveborn, asymptomatic</td>
</tr>
<tr>
<td></td>
<td>32</td>
<td>21</td>
<td>Routine</td>
<td>None</td>
<td>Liveborn, asymptomatic</td>
</tr>
</tbody>
</table>

NS=not stated; TOP=termination of pregnancy.
*Normal a-fetoprotein levels.
abnormality has developed over an 18-month follow-up period.

**DISCUSSION**

In this report we describe two cases of isolated diastematomyelia detected by routine second-trimester ultrasonography, the most striking finding being the detection of an echogenic focus in the posterior aspect of the spine in association with widening of the interpedicular vertebral space. Although in both cases the initial concern was whether spina bifida was present or not, the absence of a lemon-shaped head and a banana-shaped cerebellum (Nicolaides et al., 1986), the visualization of an intact skin overlying the defect, and a normal second-trimester maternal serum α-fetoprotein level were reassuring. Similarly, the normal appearance of the spinal curvature also ruled out other vertebral anomalies such as butterfly vertebra or hemivertebra (Harrison et al., 1992; Zelop et al., 1993). Therefore, in both cases the fetal prognosis was considered favourable. Indeed, the pregnancy proceeded uneventfully to term in both cases and the infants have been asymptomatic from the neurological and orthopaedic point of view up to 18 months of age.

To our knowledge, prenatal diagnosis of diastematomyelia has been previously reported in 13 cases (Williams and Barth, 1985; Winter et al., 1989; Caspi et al., 1990; Pachi et al., 1992; Boulot et al., 1993; Anderson et al., 1994). Table I displays the principal ultrasonographic findings and the pregnancy outcome of all these cases, and includes the two reported here. In five cases, there were severe associated vertebral defects detectable by ultrasonography (Williams and Barth, 1985; Caspi et al., 1990; Pachi et al., 1992; Boulot et al., 1993; Anderson et al., 1994), including four with spina bifida and one with scoliosis; all these pregnancies were terminated. Isolated diastematomyelia was reported in eight fetuses in two reports (Winter et al., 1989; Anderson et al., 1994). Of these, three pregnancies were terminated and one resulted in a stillbirth at 25 weeks unrelated to the spinal defect. Therefore, including our two cases, information regarding the paediatric outcome of isolated diastematomyelia detected during the second trimester of pregnancy is available from six cases. A butterfly vertebra was subsequently detected at follow-up scan in one fetus that proved to have a spinal deformity, a small foot, and shortening of the leg. In another, postnatal studies disclosed butterfly and hemivertebrae, but the infant had no neurological involvement. Two infants underwent spinal surgery to remove intradural fibrous bands, including the infant with the abnormal leg, and all the others have been free of neurological and orthopaedic symptoms.

Identification of an extra echogenic posterior focus in the spinal canal has been reported as a highly specific prenatal sign of diastematomyelia (Winter et al., 1989; Anderson et al., 1994). This finding has been detected prospectively in 0·06 per cent of 10 070 obstetric ultrasonographic examinations, and in all cases, the prenatal diagnosis was confirmed after delivery (Anderson et al., 1994). However, it should be stressed that the diagnosis can be easily missed in otherwise normal fetuses and only discovered when the infants develop neurological symptoms later on in life. Once the diagnosis of diastematomyelia is suspected prenatally, a focused ultrasonographic examination of the abnormal segment should be performed in order to exclude spina bifida and vertebral abnormalities. In this respect, it is noteworthy that in two of the four cases previously reported in which diastematomyelia was associated with spina bifida, the α-fetoprotein levels were within the normal range (Boulot et al., 1993; Anderson et al., 1994), and in two of the cases in which no other associated abnormalities were detected prenatally, vertebral anomalies were found at postnatal evaluations, one of which had severe orthopaedic problems (Winter et al., 1989). Nevertheless, according to our experience and the reported prenatal series (Winter et al., 1989; Anderson et al., 1994), it seems that diastematomyelia without other ultrasonographic detectable anomalies carries a good prognosis, but the parents should be counselled by a multidisciplinary team and be aware of the neurological and orthopaedic involvement occasionally seen in these cases.

**REFERENCES**


