Efficacy of prenatal ultrasonography in confirmed club foot
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Club foot can be diagnosed by ultrasound of the fetus in more than 60% of cases. We have correlated the accuracy of the prenatal findings in 281 ultrasound surveys with the physical findings after birth and the subsequent treatment in 147 children who were born with club foot.

The earliest week of gestation in which the condition was diagnosed with a high degree of confidence was the 12th and the latest was the 32nd. Not all patients were diagnosed at an early stage. In 29% of fetuses the first ultrasound examination failed to detect the deformity which subsequently became obvious at a later examination. Club foot was diagnosed between 12 and 23 weeks of gestation in 86% of children and between 24 and 32 weeks of gestation in the remaining 14%. Therefore it can be considered to be an early event in gestation (45% identified by the 17th week), a late event (45% detected between 18th and 24th weeks) or a very late event (10% recognised between 25th and 32nd weeks). We cannot exclude, however, the possibility that the late-onset groups may have been diagnosed late because earlier scans were false-negative results. The prenatal ultrasonographic findings were correlated with the physical findings after birth and showed that bilateral involvement was more common than unilateral. There was no significant relationship between the prenatal diagnosis and the postnatal therapeutic approach (i.e., conservative or surgical), or the degree of rigidity of the affected foot.

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Club foot is a relatively common birth defect with an incidence ranging from 0.09% in the newborn population\textsuperscript{1} to 0.43% when diagnosed antenatally by ultrasound.\textsuperscript{2} There is a wide variation of racial incidence\textsuperscript{3} ranging from 0.57 per thousand live births in Asian populations of unmixed blood to 6.81 per thousand in pure-blood Hawaiians\textsuperscript{4} and 6.5 to 7.0 per thousand in Maoris.\textsuperscript{5} Boys are affected twice as often as girls,\textsuperscript{3,6,7} and 50% of cases are bilateral.\textsuperscript{8}

Improvements in obstetric ultrasound have allowed fetal development to be studied from the early stages of gestation and have led to an increase in the detection and evaluation of musculoskeletal anomalies in the developing fetus.\textsuperscript{9} This has resulted in the involvement of paediatric orthopaedic surgeons in prenatal counselling and planning of treatment.\textsuperscript{10} The earliest gestational week for detection of many musculoskeletal anomalies is already known, but we have yet to establish the correlation between early, prenatal diagnosis and the severity of the postnatal findings.\textsuperscript{10} Ultrasound is performed by the transabdominal (TAS) or transvaginal (TVS) approach. Modern, high-resolution TVS ultrasound allows detection of skeletal anomalies up to four weeks earlier than by TAS ultrasound.\textsuperscript{11,12} TVS also gives better visualisation of the fetus, but it can only be used up to 17 weeks of gestation because of its short focal distance. TAS ultrasound is used after 17 weeks. The time recommended for systematic TVS examination is between the 14th and 16th weeks of gestation when many musculoskeletal anomalies can be detected.\textsuperscript{13} The limb buds are first seen on TVS ultrasound at about the eighth week of gestation and the long bones are visible by the 11th week.\textsuperscript{13,14}

The aetiology of club foot is diverse. Most cases are idiopathic, but it may be associated with other structural and chromosomal anomalies.\textsuperscript{15-21} Overdiagnosis may cause unnecessary parental stress and lead to invasive procedures which could place the pregnancy at risk. As a result of the ultrasound findings parents may consider termination of the pregnancy.\textsuperscript{22} The gravity of a false-positive diagnosis of club foot has been previously reported.\textsuperscript{10,18,23}

Our aim therefore was to investigate the accuracy, validity and significance of the prenatal ultrasonographic findings in children who are born with club foot.
Patients and Methods

We undertook a retrospective study of 147 children (106 boys and 41 girls) who were born with isolated idiopathic club foot, and who had been treated and followed up between January 1987 and April 1999. We excluded children with other musculoskeletal anomalies, neurological disorders or chromosomal aberrations. All feet included in the study were screened prenatally by ultrasound. Information on the family and obstetric history and the availability of a detailed ultrasonographic fetal anatomy survey (DSFAS) were obtained from a questionnaire which was completed by the parents. The mothers had all undergone at least one DSFAS. A total of 281 surveys was collected and reviewed. The data included the results of TVS surveys performed up to the 17th week of gestation and TAS surveys from the 18th week. The ultrasonographic surveys were performed by senior obstetricians all of whom are expert in antenatal diagnostic ultrasonography.

All the children were examined by one of us (SW) and each club foot was graded into one of the four groups described by Dimeglio et al.24 For the purpose of this study, flexible feet were assigned to groups 1 and 2 and rigid feet to groups 3 and 4. All had intensive physiotherapy combined with serial casting until the age of three to four months, when they were evaluated clinically and radiologically by Kite’s method.25 The decision to continue conservative treatment or to recommend surgery was based on the clinical and radiological findings. All were followed up for a minimum of 24 months.

Statistical analysis. The relation of possible explanatory variables regarding the time of diagnosis was examined using the chi-squared test, for example the results of more frequent prenatal diagnoses among bilateral cases, the lack of significance of the degree of severity, EMG findings, etc. Possible relations of time of diagnosis to more than one explanatory variable was examined using logistic regression, for example, the comparison of flexible/rigid carried out separately for the bilateral and unilateral cases. The statement that the difference between these two groups was not statistically significant was based on a logistic regression analysis.

Results

All patients included in this study had prenatal ultrasonography at least once. Information on one child was available for 1987, with numbers reaching 23 in 1997 and 21 in 1998 (Table I). A total of 281 surveys was undertaken; 56 children had one survey, 62 had two, 18 had three, 8 had four and 3 had five (Table I).

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Table II summarises the accuracy of antenatal diagnosis. When calculated by patient, only one-third of the children was accurately diagnosed prenatally. There has been a significant improvement in the accuracy of diagnosis with time, reaching over 60% in 1998 to 1999 (Table II). The same trend could be seen when accuracy was assessed by ultrasonographic survey. The diagnosis of club foot was only 42% accurate when the total of 281 surveys from 1987 to 1999 was averaged, but reached between 66% and 70% in 1998 and 1999.

Evaluation of the data from the DSFASs from the 8th to the 39th week of gestation revealed that the earliest
gestational week in which the diagnosis of club foot was made ultrasonographically was the 12th and that the latest was the 32nd (mean, 19th week).

In 29% of the fetuses (15/51), the first ultrasound scan failed to detect the club foot, which was apparent in later scans. The mean gestational week in which an existing club foot was undetected was the 14th (8 to 22). The mean subsequent week of detection in these fetuses was the 22nd (12 to 32).

In 90% of the fetuses, (46/51) who were diagnosed by DSFAS as having club foot, the ultrasonographic diagnosis was made between the 12th and the 24th weeks of gestation. In the remaining five children (10%) it was made between the 24th and the 32nd weeks, suggesting that club foot may develop late during pregnancy, after the second trimester (Table III).

Of 51 children with DSFAS positive for club foot, 23 (45%) were diagnosed by the 17th week of gestation. In a further 23 (45%) the diagnosis was between the 18th and 24th weeks and in the remaining five children between the 25th and 32nd weeks (Table III).

When the positive ultrasonographic surveys were analysed in relation to age at diagnosis, club foot was seen in only 22% of positive scans by the 17th week when TVS was used. This increased by a further 51% by the 24th week, but 27% did not become positive for club foot until the third trimester of pregnancy (Table III).

Analysis of the total number of DSFASs performed (281) showed that the diagnosis was made in 33% by TVS up to the 17th week of gestation, in 39% by TAS up to the end of the 24th week and in 60% by TAS during the third trimester (Table III). This suggests that there are three periods during gestation when club foot may develop: early diagnosed cases (up to the 17th week), late diagnosed cases (up to the 24th week) and very late diagnosed cases (between the 25th and 32nd weeks). In our study, 90% of diagnoses were made by the 24th week, but 10% were diagnosed later. A negative DSFAS for club foot early in pregnancy may not exclude its appearance later. It is not possible to diagnose club foot at an early stage in all patients. In some it may not be seen until after the 24th week.

Table IV compares the postnatal clinical state of the club foot with the gestational age at ultrasonographic detection.

<table>
<thead>
<tr>
<th>Diagnosis at gestational week</th>
<th>Unilateral</th>
<th>Bilateral</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Flexible</td>
<td>Rigid</td>
</tr>
<tr>
<td>12 to 17</td>
<td>3</td>
<td>8</td>
</tr>
<tr>
<td>18 to 24</td>
<td>1</td>
<td>2</td>
</tr>
<tr>
<td>25 to 32</td>
<td>2</td>
<td>1</td>
</tr>
<tr>
<td>Detected prenatally</td>
<td>6</td>
<td>11</td>
</tr>
<tr>
<td>Detected at birth</td>
<td>20</td>
<td>36</td>
</tr>
<tr>
<td>Total</td>
<td>26</td>
<td>47</td>
</tr>
</tbody>
</table>

Table IV. Clinical evaluation of the children with club foot in relation to the gestational age at ultrasonographic detection.
Discussion

The aim of this retrospective study was to investigate the accuracy, validity and significance of a prenatal diagnosis of club foot by ultrasound. Although all patients included in the study had been screened, club foot was only detected prenatally in the minority. The rate of detection has, however, improved over the years.

In our series, 35% of children born with club foot who presented to Dana Children’s Hospital for treatment were accurately diagnosed by prenatal screening. This figure is better than the mean rate of detection of 25% reported recently from Australia. In that series the rate of detection increased to 30% in 1995 while our rate increased to 67.6% in 1998 and to 70% in the first four months of 1999.

The use of prenatal ultrasonography for the detection of club foot has increased steadily during the last decade. Its accuracy is related to several factors such as the skill of the examiner, the quality of the ultrasonographic equipment, gestational age, and the methodology used (TAS, TVS, three-dimensional, colour, etc.). There is a steep learning curve. The latest three-dimensional ultrasonographic equipment may improve the rate of detection of club foot and other anomalies. From our findings we have identified three types of isolated idiopathic club foot based on the age of prenatal detection.

Early club foot. This is detected from the 12th week of gestation (our earliest recorded ultrasonographic detection of club foot) to the 17th week. The diagnosis in this group is based on TVS, and 45% of children with idiopathic club foot at birth were included in this group.

Late-onset club foot. This is detected between the 18th and 24th weeks of gestation and comprises 45% of the children born with club foot and diagnosed prenatally. This group included most of the children whose feet had a normal early ultrasonographic appearance and only later were identified ultrasonographically as having club foot.

Very late-onset club foot. This was seen between the 25th and 32nd weeks of gestation: 10% of the children born with club foot were included in this group.

Although our conclusions are based on retrospective data they are supported by prospective data presented by other authors. We cannot exclude the possibility, however, that the late-onset and very late-onset groups may have been diagnosed late because earlier scans were false-negative results.

Club foot is an infrequent ultrasonographic finding and can be both a transient and a late-onset phenomenon. Over- and underdiagnosis are potential hazards in these situations. It would appear that club foot belongs to a small group of fetal anomalies in which a definitive ultrasonographic diagnosis may sometimes only be possible in the second half of pregnancy. This is well demonstrated by the increasing rate of detection of club foot with time in our series: from 32% when scans were done up to the 17th week of gestation, to 39% between the 18th and 24th weeks, and to 62% in fetuses after 25 weeks of gestation.

There are no previously published reports in which the severity and the rigidity of the club foot have been correlated with the ultrasonographic findings and there are no ultrasonographic criteria which could predict prenatally the severity of the malformation of the foot and assist the paediatric orthopaedic surgeon in prenatal counselling.

It has been shown that a fetus can temporarily turn the foot into a position simulating club foot, but this aberrant positioning is usually expected to readjust during a standard examination of 30 minutes. Some of the previously reported false-positive club feet can be attributed to this transient phenomenon. It would appear therefore that the 22- to 24-week scans should be more reliable for diagnosing club foot, but it may develop even later during pregnancy. Our data suggest that some patients (10%) will develop the condition during this later stage.

Prenatal detection of an apparently isolated club foot is important because it may lead to the discovery of other associated deformities and chromosomal abnormalities. In a recent study, defects involving other systems were found in more than 50% of cases which had been diagnosed antenatally as having an isolated club foot. In another study, most of the associated malformations (20 in 68 patients) were subtle. They were not detected by ultrasonography early in the second trimester and were diagnosed only after delivery.

It is reassuring that in the series of Treadwell et al all the associated anomalies in fetuses with prenatal club foot were detected, but the limitations of ultrasonography must be considered during prenatal counselling. The high correlation between prenatal and postnatal diagnosis in the subgroup of patients with associated anomalies may reflect a more rigid teratogenic defect of the foot in these fetuses. The need to stress the risks and dangers of misinterpreting the ultrasonographic findings cannot be overemphasised. The false-positive rate varies significantly in different studies (0% to 40%) which may be related to differences in the population which was scanned. A higher false-positive rate is usually seen in children with isolated club foot diagnosed later in pregnancy.

In this study we could not control certain relevant factors, such as the skill and experience of the obstetrician or gynaecologist performing the ultrasonographic imaging and the timing of the ultrasonographic examinations. Our sample size is large enough, however, to make it reasonable to assume that these factors would be adequately randomly associated with severity and laterality, thus allowing us to compare those parameters with the time of diagnosis.
No benefits in any form have been received or will be received from a commercial party related directly or indirectly to the subject of this article.

References