Prenatal ultrasound diagnosis of club foot
OUTCOME AND RECOMMENDATIONS FOR COUNSELLING AND FOLLOW-UP

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Club foot was diagnosed by ultrasonography in 91 feet (52 fetuses) at a mean gestational age of 22.1 weeks (14 to 35.6). Outcome was obtained by chart review in 26 women or telephone interview in 26. Feet were classified as normal, positional deformity, isolated club foot or complex club foot.

At initial diagnosis, 69 feet (40 fetuses) were classified as isolated club foot and 22 feet (12 fetuses) as complex club foot. The diagnosis was changed after follow-up ultrasound scan in 13 fetuses (25%), and the final ultrasound diagnosis was normal in one fetus, isolated club foot in 31 fetuses, and complex club foot in 20 fetuses.

At birth, club foot was found in 79 feet in 43 infants for a positive predictive value of 83%. Accuracy of the specific diagnosis of isolated club foot or complex club foot was lower; 63% at the initial ultrasound scan and 73% at the final scan. The difference in diagnostic accuracy between isolated and complex club foot was not statistically significant. In no case was postnatal complex club foot undiagnosed on fetal ultrasound and all inaccuracies were overdiagnoses. Karyotyping was performed in 25 cases. Abnormalities were noted in three fetuses, all with complex club foot and with additional findings on ultrasound.

Club foot is a multiplanar deformity of the lower limb with a prevalence of 1 to 3 per 1000 live births.1 Severity varies widely and includes flexible postural deformity often requiring no treatment; isolated club foot, needing casting and possible surgery, usually with a favourable outcome or complex club foot, associated with syndromic, neuromuscular or chromosomal conditions causing major disability.

The widespread use of ultrasonography during pregnancy and improved techniques have greatly increased the rate of diagnosis of deformities. This has led to the establishment of prenatal clinics in which expectant parents are informed about the outcome and long-term consequences of the condition,2-6 thereby helping them to decide upon the continuation of the pregnancy and cope with the deformity postnatally.

Publications on antenatally diagnosed club foot present a wide variation in the accuracy of ultrasonography, the percentage of complex cases and agreement on further investigation.7-16 The present study aimed to provide further knowledge on these issues.

Patients and Methods
This study included all women referred to the Fetal Abnormality Clinic for abnormalities because of an ultrasonographic diagnosis of club foot between 1996 and 2003.

Fifty-six women (57 fetuses) were counselled. Four women (five fetuses) were excluded because of inadequate documentation, leaving 52 women (52 fetuses). In five pregnancies, there was an unaffected twin.

The women underwent a mean of 2.7 ultrasonographic scans (1 to 7). The women were identified by review of the pregnancy follow-up notes, birth records (if the woman gave birth in our centre), and the children’s records (if the club foot was treated in our paediatric orthopaedic unit). Women who had follow-up and treatment at other centres were interviewed by telephone. Data were collected on details of each sonogram, findings at birth or after abortion, subsequent treatment of the child, and condition at latest follow-up. Feet were classified as normal, post-deformity, isolated club foot or complex club foot.

Statistical analysis was performed with Fisher’s exact test with values for p < 0.05 being regarded as significant.

Results
Club foot was diagnosed in 91 feet at a mean gestational age of 22.1 weeks (14 to 33.6). We
diagnosed 43 at the first scan, six at the second, and three at the third.

At initial diagnosis, 69 feet (40 fetuses) were identified as having isolated club foot and 22 feet (12 fetuses) as complex club foot, a rate of 23%. The diagnosis changed on further ultrasonographs in 13 fetuses (25%) at a mean gestational age of 25.3 weeks. In ten of these, either bilateral or more severe club foot was diagnosed. In the remaining three, the condition appeared less severe. Overall, eight children who were initially classified isolated club foot were reclassified complex club foot. The final prenatal diagnoses were isolated club foot in 53 feet (31 fetuses), complex club foot in 38 feet (20 fetuses), and one foot was reclassified as normal.

There were 42 live births (30 boys and 12 girls). The other ten pregnancies were terminated or stillborn. Postnatal data were obtained from maternal, child and autopsy charts in 26 infants and by telephone interview in 26.

The diagnosis of club foot was confirmed postnatally in 43 children, a positive predictive value of 83% (52 fetuses) for the initial diagnosis and 84% (51 fetuses) for the final diagnosis. The diagnostic accuracy was 63% initially and 73% at final diagnosis.

Of the 40 fetuses with an initial diagnosis of isolated club foot, 26 had isolated club foot at birth (a positive predictive value of 65%). Five of the 14 misdiagnosed fetuses (13%) had normal feet, three (8%) postural deformity and six (16%) complex club foot.

The positive predictive value for the final diagnosis of isolated club foot was 77% (24 of 31). Of the seven misdiagnosed children, three had normal feet (10%) and four (13%) had postural deformity.

Of the 11 children with an initial diagnosis of complex club foot, eight were confirmed at birth and of the 20 fetuses with a final diagnosis of complex club foot, 14 had a complex club foot at birth. The positive predictive values for the initial and final scans were 73% and 70%, respectively. Five misdiagnosed children had isolated club foot at birth and one was normal.

The difference in diagnostic accuracy between isolated and complex club foot was not significant for either the initial or final scan (Fisher's exact test).

Ultrasound diagnosed both complex club foot and associated conditions as follows; five arthrogryposis, four multiple anomalies, two anencephaly, one macrocephaly, one ventricular enlargement, one myelomeningocele, one scoliosis and growth retardation, one leg-length discrepancy and neck widening, one hydronephrosis and small penis (Fig. 1), one congenital knee dislocation (Fig. 2), one polyhydramnios and one Rubinstein-Taybi syndrome. The diagnoses were confirmed in 14 of the 20 cases. Three of the five fetuses diagnosed as arthrogryposis and those with macrocephaly and Rubinstein-Taybi syndrome were found to have idiopathic club foot. The fetus diagnosed with ventriculomegaly had normal feet.
Amniocentesis and karyotyping were performed in 25 pregnancies. Three pathological types were found: one trisomy 18, one deletion on the long arm of 8q and one XXY. All three had been classified as complex club foot by ultrasonography before karyotyping because of additional morphological anomalies.

Live infants were followed to a mean of 39 months (0 to 99). None of the nine children born with postural deformity or no deformity developed late deformity.

The 29 children born with isolated club foot were treated by serial casting. Seven underwent percutaneous heel-cord lengthening, seven a full posterior or posteromedial release and one had secondary surgery.

At latest follow-up, all were fully mobile. Three had residual deformity possibly needing additional surgery.

Of the six live infants with complex club foot, one was undergoing serial casting, two were mobile following extensive surgery, and three died of their underlying disease.

Discussion
The prenatal diagnosis of any fetal deformity causes major anxiety in expectant parents. Well-informed advice can improve parental understanding of the condition and alleviate anxiety. The differentiation between isolated and complex clubfoot at prenatal ultrasonographic diagnosis is very important, as the latter may be associated with other grave conditions, severe lifelong disability or early demise. Therefore, it is also important to inform parents, during counselling, of the limitations of prenatal diagnosis.

In our study, the initial diagnosis of club foot, at a mean gestational age of 22.1 weeks, was confirmed at birth in 83% of cases, a false-positive rate of 17%. Corresponding rates for the final diagnosis were 84% and 16%. The false-positive rate in previous studies has ranged between 0% and 24%, possibly due to the inclusion of postural deformities as a positive postnatal finding. As feet with postural deformity do not require treatment, we do not believe they should be considered as a positive finding at birth.

The reported accuracy of the specific diagnosis of postural deformity, isolated or complex club foot or normal feet varies widely between 43% and 100%, with a mean of 79%. Our rate of 73% at final diagnosis was close to this mean. However, in earlier series, between 5% and 29% of cases of complex club foot were misdiagnosed as isolated club foot, whereas in our study, no case of complex club foot was missed, and all ultrasonographic inaccuracies were overdiaagnoses. The associated condition most overdiagnosed was arthrogryposis, with a 60% (3 of 5) false-positive rate.

The 39% rate of complex club foot in our study was relatively low compared to rates between 22% and 80% (mean of 62%) reported by others. Our rate may reflect the widespread use of ultrasonography in Israel, whereas studies from other areas used selected populations.

Changes in diagnosis during pregnancy were addressed by Bakalis et al who reported that 19% of fetuses initially diagnosed as isolated club foot were reclassified as complex club foot when other defects were found on subsequent scans; another 5.5% classified as isolated club foot were found to have complex club foot postnatally. In our study, the diagnosis changed in 25% of fetuses. Although the change was found to be correct in seven cases and incorrect in six, continued follow-up increased predictive values from 63% to 73% and no diagnosis of complex club foot was missed prenatally. These findings emphasise the need for sequential ultrasonograms after initial diagnosis.

The accuracy of the diagnosis of club foot by stage of pregnancy has been the subject of several reports. Bar Hava et al described a transient club foot-like deformity in early pregnancy. Treadwell et al reported a high rate of false-positive results in third trimester sonograms following earlier normal results in the second trimester and concluded that positional factors may cause a false positive sonogram in the third trimester. In our study, eight of nine infants born with normal feet had a final diagnosis of club foot by the third trimester sonogram; however, in all of them, club foot had also been diagnosed in earlier examinations at weeks 21 to 27. The other infant had been diagnosed with isolated club foot in weeks 26 and 28, but was reclassified correctly as normal at 29 and 34 weeks. We, therefore, do not believe the false-positive results were caused by transient malpositioning but rather by other factors which obscured the diagnosis.

The need for karyotyping after prenatal diagnosis of isolated club foot is controversial. Shipp and Benacerraf found that 5.9% of 87 fetuses with isolated club foot had an abnormal karyotype and concluded that amniocentesis is indicated after that diagnosis. Other investigators came to the opposite conclusion, having found no pathological karyotypes in those fetuses with isolated club foot which they examined.

In the present study, amniocentesis and karyotyping were performed in 25 pregnancies and did not yield any additional information regarding club foot. The three abnormal karyotypes found were in fetuses with additional deformities identified before karyotyping and the results did not alter decision making. Also, a literature and genetic database search revealed 14 chromosomal aberrations causing club foot. Nine have characteristics easily identifiable on ultrasonography and the remaining four are extremely rare. We, therefore, question the need to perform amniocentesis and karyotyping after ultrasonographic diagnosis of isolated club foot.

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References


