

Outcome of antenatally diagnosed talipes equinovarus in an unselected obstetric population

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ABSTRACT

Objective To investigate the natural history and outcome of fetal talipes diagnosed by routine ultrasound scanning at 18–23 weeks' gestation.

Patients and methods This was a retrospective study of 103 228 pregnancies undergoing routine ultrasound scanning at 18–23 weeks' gestation. A computer search was made to identify all cases of fetal talipes and the records of these patients were examined to determine the incidence of other defects and pregnancy outcome.

Results The incidence of fetal talipes following routine ultrasound examination was 0.10% (107/103 228) and was bilateral in 64 (59.8%) and unilateral in 43 (40.2%) cases. In 52 (48.6%) cases, talipes was of complex etiology, as it was associated with other defects, while, in 55 (51.4%) cases, it was classified as idiopathic. In 19% of cases, an initial diagnosis of idiopathic talipes was changed to complex, because of the subsequent development of associated features. Perinatal death and long-term neurodevelopmental or musculoskeletal problems were significantly more common when the talipes was complex rather than idiopathic (odds ratio, 150; 95% confidence interval, 34–665). Adverse outcomes were also seen more frequently with bilateral compared to unilateral talipes (odds ratio, 3.44; 95% confidence interval, 1.50–7.90).

Conclusion The outcome of antenatally detected talipes is mainly dependent on the presence or absence of other defects. A significant proportion of cases, thought to be idiopathic at presentation, will develop associated complex features when reassessed on subsequent scans or postnatally.

INTRODUCTION

Talipes equinovarus (club foot) is a congenital deformity of the joints of the ankle. Despite its prevalence of approximately

1 per 1000 live births^{1,2}, fetal talipes is a relatively poorly studied congenital anomaly. Information relating to the postnatal outcome of fetal talipes is limited because previous studies were conducted on small, preselected populations with a lack of uniformity in the ultrasound diagnostic criteria and poor assessment of neonatal outcome^{3–9}. The aim of this study was to investigate the natural history and outcome of fetal talipes detected at 18–23 weeks' gestation in a large, unselected obstetric population.

PATIENTS AND METHODS

This was a retrospective study of routine ultrasound scanning at 18–23 weeks' gestation in 103 228 women who were booked for antenatal care at three London teaching hospitals between January 1991 and October 2000.

On ultrasound examination at 18–23 weeks' gestation, a full structural survey of the fetus was performed. Fetal talipes was diagnosed if the two long bones of the lower leg (tibia and fibula) were seen in the same plane as the lateral aspect of the foot, with the foot extended and inverted. In all affected cases, the woman was referred to a fetal medicine specialist for further assessment and, in cases with additional defects, fetal karyotyping was performed. Fetal talipes was classified as complex if there were associated anomalies detected on ultrasound or if the fetal karyotype was abnormal. In the absence of associated features, antenatally diagnosed talipes equinovarus was classified as idiopathic. When subsequent antenatal or postnatal follow-up of idiopathic talipes revealed associated neurodevelopmental or physical abnormalities, the classification was changed to complex talipes equinovarus.

A computer search was made to identify all cases of fetal talipes and the records of these patients only were examined to determine the incidence of other defects and pregnancy outcome. Delivery details were collected from the statutory

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Table 1 Causes of complex fetal talipes equinovarus

Classification	Number	Type
Musculoskeletal	3	Three unspecified musculoskeletal syndromes, resulting in residual neonatal defects of poor general tone, joint and underdeveloped muscles
Chromosomal	9	One trisomy 21 One triploidy
Neurological	20	Seven trisomy 18 Three ventriculomegaly Three unspecified neurological disorders with developmental delay Six neural tube defects
Syndromic	20	Eight arthrogryposis multiplex congenita One CHARGE syndrome One Potters sequence One body stalk anomaly Two amniotic band syndromes Six skeletal dysplasias (three with thanatophoric dwarfism) Nine unspecified syndromes with multiple congenital anomalies

CHARGE, Coloboma–heart defect–atresia of nasal choanae–restricted growth–genital hypoplasia–ear defect syndrome.

birth records submitted following the delivery of all neonates. Chromosomal abnormalities were identified from a review of the regional cytogenetic database. Follow-up of affected live-born infants was carried out by a review of orthopedic surgery records on infants. General practitioners and parents were contacted either by telephone or post in order to complete a structured questionnaire, when local hospital information was incomplete. Stillbirth, neonatal death and long-term neurodevelopmental or generalized musculoskeletal problems defined poor pregnancy outcome.

RESULTS

During the study period, 103 228 pregnant women had routine scans at 18–23 weeks' gestation. There were 107 cases of talipes equinovarus giving an antenatal incidence of 0.10% (95% confidence interval, 0.08–0.12). Talipes was diagnosed antenatally as bilateral in 64 (59.8%) and unilateral in 43 (40.2%) cases and as complex in 49 (45.8%) and idiopathic in 58 (54.2%) cases.

Complex talipes was classified according to the putative cause; chromosomal abnormalities, single gene disorders, musculoskeletal or neurological abnormalities (Table 1). The talipes was correctly identified as complex in etiology at the 18–23-week scan in 75% (39/52) of cases, and on repeat ultrasound after 24 weeks' in 19.2% (10/52) of cases. In another three pregnancies, the diagnosis was changed to complex because of a postnatal diagnosis of significant neurodevelopmental delay (Figure 1). In 10 pregnancies considered to be idiopathic antenatally, the diagnosis was classified postnatally as postural talipes, as it was presumed to have occurred as a consequence of a crowded intrauterine environment. In four pregnancies in the idiopathic group, the antenatal diagnosis of talipes was not confirmed postnatally, resulting in a false-positive rate of 3.7%. In three cases, subsequent scans failed to confirm the finding of fetal talipes equinovarus and, in the fourth case, the diagnosis was suspected at 29 weeks' gestation.

There were 65 (60.7%) live births, 33 (30.8%) terminations of pregnancy, 3 (2.8%) intrauterine deaths and six (5.6%)

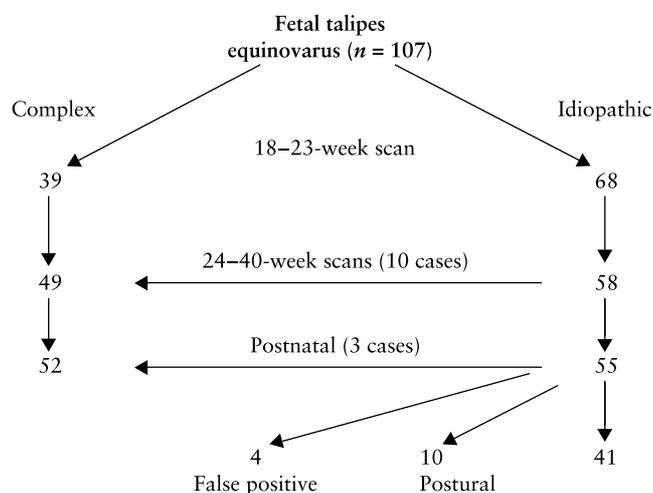


Figure 1 Flowchart showing the classification of talipes equinovarus diagnosed at the 18–23-week ultrasound scan, and any changes made on subsequent scans or postnatally.

neonatal deaths. The relationships between pregnancy outcome and classification are shown in Tables 2 and 3 and Figures 2 and 3. Bilateral talipes was found more frequently in the complex compared to the idiopathic group (Table 2, Figure 2). Poor outcome, as defined by stillbirth, neonatal death or long-term neurodevelopmental or musculoskeletal problems, was significantly more common with complex and bilateral talipes equinovarus (Table 3, Figures 3 and 4).

DISCUSSION

This study reports on the natural history and postnatal outcome for fetal talipes equinovarus diagnosed by routine mid-trimester ultrasound in a large, unselected obstetric population.

Incidence and laterality

The incidence of talipes equinovarus of 1 per 1000 pregnancies is lower than that estimated from postnatal studies, but

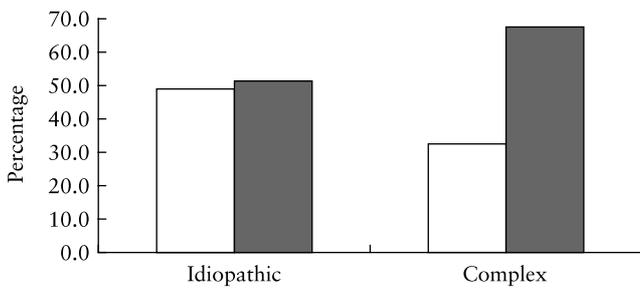


Figure 2 Laterality of fetal talipes equinovarus shown according to etiological classification. □, Unilateral; ■, bilateral.

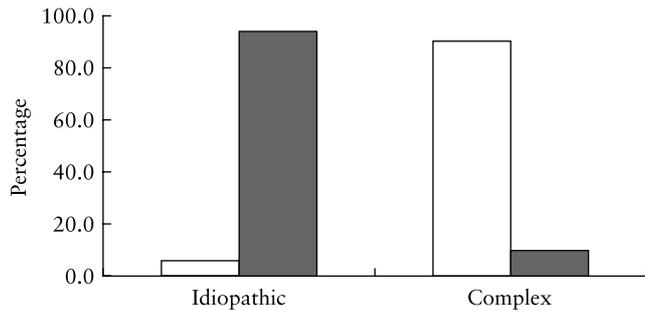


Figure 3 Pregnancy outcome according to etiological classification. Poor outcomes include all terminations of pregnancy, stillbirths, neonatal deaths and long-term neurodevelopmental or musculoskeletal disorders. □, Poor outcome; ■, good outcome.

Table 2 Number of unilateral and bilateral fetal talipes equinovarus according to etiological classification

Classification	Unilateral	Bilateral	Odds ratio (95% confidence interval)
Idiopathic	25	26	0.505 (0.227–1.12)
Complex	17	35	
Total	42	61	

Table 3 Outcome of pregnancy according to etiological classification. Poor outcomes include all terminations of pregnancy, stillbirths, neonatal deaths and long-term neurodevelopmental or musculoskeletal problems

Classification and laterality	Poor outcome	Good outcome	Observed difference (95% confidence interval)
Idiopathic	3*	48	150 (34–665)
Complex	47	5	
Unilateral	13	29	3.44 (1.50–7.90)
Bilateral	37	24	

*Two neonatal deaths due to complications of prematurity and one termination of pregnancy. Two cases were bilateral and one case was unilateral.

similar to that quoted in previous ultrasound surveys (0.4–4 per 1000)^{1–4}. As follow-up was only performed on those pregnancies diagnosed with talipes equinovarus on antenatal ultrasound, the incidence of talipes diagnosed after birth is not known. Antenatal studies underestimate the prevalence of postural talipes equinovarus because the ultrasound features of talipes are often subtle and they are usually of late

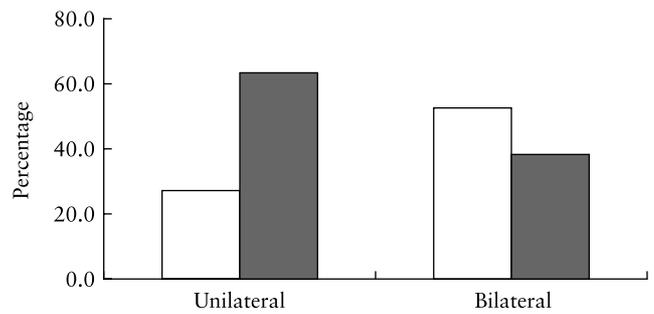


Figure 4 Pregnancy outcome according to laterality. Poor outcomes are as defined in Figure 3. □, Poor outcome; ■, good outcome.

onset, thereby remaining undetected on the 18–23-week scan^{4,8,10}. This explanation is supported by the finding that postnatally diagnosed talipes is quite frequently postural in origin. Since the majority of postural talipes equinovarus cases are unilateral in origin, the latter hypothesis also explains our finding of a higher incidence of bilateral compared to unilateral talipes; postnatally, unilateral and bilateral talipes equinovarus are thought to occur at equivalent frequencies¹¹. An alternative explanation for the discrepancy in laterality between the antenatal and postnatal periods is that bilateral talipes is more likely to be associated with a poor outcome, resulting in stillbirth or termination of pregnancy.

Etiology

The incidence of fetal abnormality, be they chromosomal, structural or neuromuscular, is much higher in our study population than was previously reported in postnatal studies⁵. The 52 cases with a complex etiology had the following classifications: 20 neurological, 20 genetic, nine chromosomal and three musculoskeletal syndromes. There were no chromosomal abnormalities in the group classified as idiopathic talipes equinovarus on ultrasound. Although the majority of aneuploid pregnancies (seven of nine) were trisomy 18, this total number is lower than expected, probably because all three hospitals performed routine nuchal translucency screening and most aneuploidies would have been diagnosed and terminated in the first trimester. The value of karyotyping in the presence of isolated talipes in a previously screened population is therefore questionable. The majority of talipes cases with complex etiologies were bilateral, suggesting that when there are distant associated features such as with neural tube defects, it is more likely that both legs are affected. Another hypothesis would be that the bilateral cases are due to early onset of some genetic, muscular or neurological abnormality, and therefore both sides are more likely to be affected.

The gestation at which the etiology, rather than the diagnosis of the talipes, is confirmed is of considerable importance. Most of the complex cases (75%) were diagnosed when presenting at 18–23 weeks’ gestation. The remainder were diagnosed when the classification was changed from idiopathic to complex, either when scanned again after 24 weeks’ gestation (19%) or postnatally (6%). In this study, of the 68 cases of talipes equinovarus thought to be idiopathic at the 18–23-week

scan, 13 (19%) were subsequently found to be complex in etiology. Hence, when apparently isolated talipes is noted at the 18–23-week scan, parents should be counseled about the possibility of associated problems being noted on later scans or after birth. The majority of complex cases diagnosed late were progressive neurological or musculoskeletal conditions.

Outcome

The postnatal outcome of talipes equinovarus is a reflection of our current understanding of this fetal abnormality, and is invaluable in counseling parents. Of the complex cases, only 10% were live births with a normal outcome at the time of follow-up, while the remainder resulted in termination of pregnancy, stillbirth, neonatal death or significant postnatal neurodevelopmental problems. In contrast, 94% of idiopathic cases had normal pregnancy outcomes. The only losses that occurred in the latter two groups were incidental, either due to premature delivery or termination of pregnancy for unrelated reasons. The majority of the pregnancies with a poor outcome (10/13) had a diagnosis of complex etiology made at presentation, and the three remaining losses were incidental to the diagnosis of talipes. The diagnosis of bilateral talipes equinovarus, however, had a poor outcome in 58% of pregnancies, as they were far more likely to have a complex etiology.

In conclusion, there are significant differences in the prevalence of complex, idiopathic and postural talipes, depending on whether fetal talipes is unilateral or bilateral. The prognosis was favorable in 94% of fetuses with a diagnosis of idiopathic talipes, but resulted in termination of pregnancy, stillbirth or long-term handicap in the majority (90%) of cases with complex talipes. Importantly, the classification was changed from idiopathic to complex in 19% of cases when scanned again after 24 weeks' gestation or postnatally. This study clearly delineates the natural history of fetal talipes equinovarus diagnosed by routine mid-trimester ultrasound in a routine, low-risk obstetric population. Fetal medicine

practitioners should be aware that the outcome of fetal talipes equinovarus is significantly worse than previously reported. The fetal/neonatal prognosis is largely dependent on the presence or absence of associated features.

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