Prevalence and prenatal ultrasound detection of clubfoot in a non-selected population: an analysis of 549,931 births in Tuscany

Viola Seravalli1, Anna Pierini2,3, Fabrizio Bianchi2,3, Sabrina Giglio4,5, Francesca L. Vellucci6, and Ettore Cariati1

1Medical Surgical Fetal-Neonatal Department, “A. Meyer” University Children’s Hospital, Florence, Italy, 2CNR Institute of Clinical Physiology, Unit of Environmental Epidemiology, Pisa, Italy, 3CNR-Tuscany Region “Gabriele Monasterio” Foundation, Unit of Epidemiology, Pisa, Italy, 4Department of Clinical and Experimental Biomedical Sciences, University of Florence, Florence, Italy, 5Medical Genetics Unit, Meyer Children’s University Hospital, Florence, Italy, and 6Department of Molecular and Developmental Medicine, Obstetrics and Gynecology, University of Siena, Siena, Italy

Abstract

Objective: To evaluate the prevalence and prenatal ultrasound detection of clubfoot in Tuscany during a period of 20 years.

Methods: This is a descriptive analysis on data from the Tuscan register of congenital defects, covering a 20-year period from 1992 to 2011. The Tuscan registry of congenital defects is a population-based register for the epidemiologic surveillance of congenital anomalies. The study included all cases of pre- or postnatally diagnosed clubfoot (isolated clubfoot and cases associated with other congenital defects). Overall prevalence and pre-natal detection rates were calculated.

Results: Among the 549,931 deliveries recorded in Tuscany between 1992 and 2011, 858 cases of clubfoot were registered, with a prevalence of 1.56/1000. Seventy-eight percent of cases were isolated. The detection rate was higher when the defect was associated with other anomalies compared to isolated forms. Over the study period, there was a substantial improvement in the prenatal detection of clubfoot (from 11 to 31% overall). For isolated forms, detection rate improved from 4 to 16%, and for cases associated with other congenital defects, it increased from 43 to 73%.

Conclusion: Prevalence of clubfoot in Tuscany is 1.56 per 1000 births, in agreement with the incidence reported in epidemiological studies in Europe. Prenatal detection of clubfoot improved over time. The detection rate was higher in cases associated with other anomalies.

Keywords

Clubfoot, congenital abnormalities, prenatal diagnosis, ultrasound

Introduction

Clubfoot, or talipes equinovarus, is a positional deformity of the fetal foot resulting in the foot being fixed in adduction, supination and varus position. It is characterized by a subluxation of the talo-calcaneo-navicular joint, with underdevelopment of the soft tissues on the medial side of the foot and frequently of the calf and peroneal muscles [1]. It is one of the most common congenital birth defects, with an incidence of approximately one per 1000 newborns [2]. It is almost twice as common in males than in females [3]. The incidence may vary between countries and population, suggesting differences in genetic disposition [4].

The foot development is influenced by many factors, such as neuromuscular conditions, genetic syndromes, aneuploidy, amniotic fluid volume, multiple gestations and hereditary factors [4–6].

In most cases the defect is isolated and the exact etiology is unknown (idiopathic). In 20% of cases, clubfoot is associated with distal arthrogryposis, congenital myotonic dystrophy, myelomeningocele, amniotic band sequence or other genetic syndromes, such as trisomy 18 or chromosome 22q11 deletion syndrome [7]. In addition, some studies highlight the importance of early limb developmental pathways in clubfoot etiology, as for example the anterior tibial artery hypoplasia present in more than 80% of clubfoot patients though the genetic basis of this abnormality is unknown [8].

Clubfoot can be diagnosed prenatally by ultrasonography. Even if its diagnosis in utero has been reported as early as 12 weeks of gestation [9], it is more commonly detected at 19–23 weeks, during the routine second-trimester ultrasound. Overall detection of clubfoot prenatally is reported to be around 60% [9,10].

The objective of our study was to evaluate the prevalence of clubfoot in a non-selected population in Tuscany during 20 years and to study trends in prenatal detection rate over time.
Materials and methods
This is a descriptive analysis on data from the Tuscan register of congenital defects, covering a 20-year period from January 1992 to December 2011. The Tuscan registry of congenital defects is a population-based register for the epidemiologic surveillance of congenital anomalies.

A detailed description of the registry, method of case ascertainment, data collection and processing is available elsewhere (http://www.eurocat-network.eu/content/Reg-Des-Tuscany.pdf). The study included all cases of pre- or postnatally diagnosed clubfoot (both cases of isolated clubfoot and those associated with other congenital defects). Prenatal diagnosis of clubfoot was based on ultrasound performed by physicians. All cases of clubfoot were confirmed postnatally either through a physical examination by a pediatrician or at autopsy. The study included all deliveries occurred in Tuscany during the study period, including miscarriages, terminations of pregnancy for fetal anomaly (TOPFA) following prenatal diagnosis and intrauterine fetal deaths (FD) of fetuses beyond 16 weeks’ gestation.

All cases were coded according to the International Classification of Diseases (ICD) version 9 with 1-digit BPA extension (from 1992 to 2001) or version 10 (from 2002 to 2009) [11,12].

Cases can have one syndrome and/or up to eight malformation codes. The data were extracted from the Tuscan database on the basis of the ICD/BPA codes (754.5, 754.6, 754.7, Q66.0–Q66.8) assigned to cases of clubfoot.

In Tuscany, routine fetal ultrasound examinations are offered to all pregnant women between 10 and 12 weeks of gestation.

The study period was divided into four 5-year periods: the first period from 1992 to 1996, the second from 1997 to 2001, the third from 2002 to 2006 and the fourth from 2007 to 2011. Prevalence and detection rates were calculated for each period. Prevalence rates were calculated as: number of cases (live births + FD + TOPFA)/number of births (live and stillbirths). The reason why we chose to divide the study period in these four intervals is that during the last 20 years important changes in prenatal diagnosis occurred: first, an increase in the number of women having access to prenatal ultrasound, and second, an improvement in the skills of the examiners and in the ultrasound technology, which led to an increase in the detection rate of fetal anomalies in general.

Results
A total of 549,931 deliveries occurred in Tuscany between 1992 and 2011. Data on prevalence of clubfoot and detection rates are reported in Tables 1 to 3. A total of 858 cases of clubfoot, isolated or associated to other congenital defects, were reported, with a prevalence of 1.56/1000 (Table 1). Prenatal diagnosis was overall made in 168 cases (19.7%) and showed an increase over the years.

Of the 858 cases of clubfoot, 672 (78.3%) were isolated (idiopathic), with a prevalence of 1.22/1000 (Table 2) and a prenatal diagnosis was made in 68 cases of 666 (10.2%) (in six cases the time at which the diagnosis was made was not reported). The other 186 cases were associated with other congenital defects (Table 3), with a prevalence of 0.34/1000 and the prenatal diagnosis was made in 100 cases (53.8%). Of these 186 cases, 83 (3.8% of total cases) were associated with a chromosomal abnormality. The type of abnormality and the outcome is reported in Table 4.

In order to show the improvement of prenatal diagnosis throughout the years, Tables 1–3 report data grouped by period. The detection rate of clubfoot showed a substantial increase over the study period, from 11.4% in the period 1992–1996 to 31.2% in the period 2007–2011. The increase in the detection rate was observed for both the isolated forms (from 3.6 to 15.9%) and the cases associated with other congenital defects (from 42.9 to 72.7%).

Concerning the period in which the defect was detected, of the 858 cases, 168 had prenatal diagnosis, 634 were diagnosed at birth, 30 in the first week of life, 8 in the first month of life, 5 in the first year of life, 4 were detected after spontaneous abortion and 2 were detected at autopsy. In 7 cases the time of diagnosis was not reported.

Therefore, in a large percentage of cases the diagnosis of clubfoot was performed after birth while less frequently the

---

### Table 1. Total cases of clubfoot.

<table>
<thead>
<tr>
<th>Period</th>
<th>Total live births</th>
<th>Cases</th>
<th>Prevalence (on 1000 births)</th>
<th>Prenatal diagnosis</th>
<th>Detection rate**</th>
</tr>
</thead>
<tbody>
<tr>
<td>1992–1996</td>
<td>123,738</td>
<td>211</td>
<td>1.7</td>
<td>24</td>
<td>11.4%</td>
</tr>
<tr>
<td>1997–2001</td>
<td>129,710</td>
<td>226</td>
<td>1.74</td>
<td>41</td>
<td>18.3%</td>
</tr>
<tr>
<td>2002–2006</td>
<td>142,776</td>
<td>213</td>
<td>1.49</td>
<td>39</td>
<td>18.3%</td>
</tr>
<tr>
<td>2007–2011</td>
<td>153,707</td>
<td>208</td>
<td>1.35</td>
<td>64</td>
<td>31.2%</td>
</tr>
<tr>
<td>Total</td>
<td>549,931</td>
<td>858*</td>
<td>1.56</td>
<td>168</td>
<td>19.7%</td>
</tr>
</tbody>
</table>

*In seven cases the time at which the diagnosis was made was not reported.
**Percentages are calculated on the total cases with known time of diagnosis.

### Table 2. Cases of isolated clubfoot.

<table>
<thead>
<tr>
<th>Period</th>
<th>Total live births</th>
<th>Cases</th>
<th>Prevalence (on 1000 births)</th>
<th>Prenatal diagnosis</th>
<th>Detection rate**</th>
</tr>
</thead>
<tbody>
<tr>
<td>1992–1996</td>
<td>123,738</td>
<td>169</td>
<td>1.36</td>
<td>6</td>
<td>3.6%</td>
</tr>
<tr>
<td>1997–2001</td>
<td>129,710</td>
<td>178</td>
<td>1.37</td>
<td>18</td>
<td>10.3%</td>
</tr>
<tr>
<td>2002–2006</td>
<td>142,776</td>
<td>172</td>
<td>1.2</td>
<td>20</td>
<td>11.6%</td>
</tr>
<tr>
<td>2007–2011</td>
<td>153,707</td>
<td>153</td>
<td>0.99</td>
<td>24</td>
<td>15.9%</td>
</tr>
<tr>
<td>Total</td>
<td>549,931</td>
<td>672*</td>
<td>1.22</td>
<td>68</td>
<td>10.2%</td>
</tr>
</tbody>
</table>

*In six cases the time at which the diagnosis was made was not reported (one in the period 1992–1996, three in the period 1997-2001 and two in the period 2007–2011).
**Percentages are calculated on the total cases with known time of diagnosis.

### Table 3. Cases of clubfoot associated with other congenital defects.

<table>
<thead>
<tr>
<th>Period</th>
<th>Total live births</th>
<th>Cases</th>
<th>Prevalence (on 1000 births)</th>
<th>Prenatal diagnosis</th>
<th>Detection rate**</th>
</tr>
</thead>
<tbody>
<tr>
<td>1992–1996</td>
<td>123,738</td>
<td>42</td>
<td>0.34</td>
<td>18</td>
<td>42.9%</td>
</tr>
<tr>
<td>1997–2001</td>
<td>129,710</td>
<td>48</td>
<td>0.37</td>
<td>23</td>
<td>47.9%</td>
</tr>
<tr>
<td>2002–2006</td>
<td>142,776</td>
<td>41</td>
<td>0.29</td>
<td>19</td>
<td>46.3%</td>
</tr>
<tr>
<td>2007–2011</td>
<td>153,707</td>
<td>55*</td>
<td>0.35</td>
<td>40</td>
<td>72.7%</td>
</tr>
<tr>
<td>Total</td>
<td>549,931</td>
<td>186*</td>
<td>0.34</td>
<td>100</td>
<td>54.1%</td>
</tr>
</tbody>
</table>

*In one case the time at which the diagnosis was made was not reported (in the period 2007–2011).
**Percentages are calculated on the total cases with known time of diagnosis.
For personal use only.

For personal use only.

For personal use only.

For personal use only.

For personal use only.

For personal use only.

For personal use only.
In summary, our results demonstrate an improvement in prenatal diagnosis of clubfoot over the years, in accordance with other studies [9,20]. One obvious explanation for the improved detection rates over the years is the improvement in ultrasound equipment. However, still in the larger percentage of cases the diagnosis of clubfoot is performed after birth: this highlights the importance of continuous teaching and training of ultrasound personnel, in order to detect this defect. Accuracy of prenatal diagnosis of this condition is important in order to exclude any other associated abnormalities.

Declaration of interest

The authors report no conflicts of interest.

References