

# PRENATAL ULTRASOUND DIAGNOSIS OF CONGENITAL TALIPES EQUINOVARUS IN BOGOTA (COLOMBIA) BETWEEN 2003 AND 2012

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## ABSTRACT

**Introduction:** Congenital Talipes Equinovarus (CTEV) or clubfoot is one of the most common congenital abnormalities<sup>1, 2</sup>. Early diagnosis by means of ultrasonography allows an opportune intervention and improves the deformity's correction prognosis.

**Goal:** To describe patients diagnosed with CTEV by means of prenatal sonographies between 2003 and 2012 in Bogotá (Colombia) at both the Instituto de Ortopedia Infantil Roosevelt (IOIR) and one of the authors' private office.

**Methods:** A descriptive, retrospective study on the focus population was made. The equality of the data of the quantitative variables in distance measure was analysed by the Kolmogorov–Smirnov test. For the variables “prenatal diagnoses” and “days from the start of the treatment” the Mann–Whitney U test was used. Finally, an analysis was made by means of the SPSS Statistics software package, version 18.0.

**Results:** 178 patients met the selection criteria. 34.3% of the patients had a prenatal diagnosis by ultrasonography (n=61). Regarding the number of prenatal ultrasounds performed, there were statistically significant differences between the patients with a CTEV prenatal diagnoses and those whose diagnoses came after birth, being higher in the first group (p<0.001). The number of days before the treatment started once the pre or postnatal diagnosis was done was also a subject of study. Significant differences were found in the treatment

start between patients with a prenatal diagnosis (mean of 9.9 days) and those diagnosed after birth (mean of 30 days) (p<0.001).

**Conclusions:** prenatal diagnosis by foetal ultrasonography contributes to an early detection of musculoskeletal abnormalities such as CTEV and promotes an early intervention of the patient.

## INTRODUCTION

Congenital Talipes Equinovarus (CTEV) or clubfoot is one of the most common congenital abnormalities<sup>1</sup>. Although most populations show an incidence of approximately 1-2 cases for every 1000 born alive infants, a study made in three Colombian cities (Bogotá, Ubaté and Manizales) found an incidence of 2.5 cases for every 1000 born alive infants<sup>2</sup>.

CTEV prenatal diagnosis can be performed whether in early stages of gestation (from the 12<sup>th</sup> week by a transvaginal ultrasound exam) or in late stages (from the 3<sup>rd</sup> trimester on by means of an abdominal ultrasound)<sup>3,4,5,6</sup>. However, most cases are diagnosed between the 18<sup>th</sup> and the 20<sup>th</sup> week<sup>10</sup>. Once a CTEV diagnoses is confirmed, the possibility of a transient deformity (transient CTEV) must always be considered, as described by Bar-Hava et al.<sup>11</sup>. His study identified CTEV cases in which the deformity was diagnosed at the end of the 1<sup>st</sup> trimester or the beginning of the second but disappeared in later ultrasound examinations.

On the other hand, there have been described cases diagnosed on a late stage of gestation (week 22<sup>nd</sup> to 24<sup>th</sup>) with initially normal ultrasounds, which suggests a late development of the abnormality. It should be noted that such cases almost always correspond to CTEV of a postural origin<sup>12</sup>.

Incidence of CTEV diagnosed before birth varies significantly in published studies, with a range going from 0.43%<sup>7</sup> to 59,8%<sup>12</sup>. In Colombia such incidence has been estimated with a presence of 12.54 / 10.000 and a detection rate of 6.67%<sup>15</sup>, which is lower than estimates obtained by other studies globally ( 63%)<sup>16</sup> and in Latin America (19.1%)<sup>17</sup>.

Additionally, the frequency of false positives also ranges from 0% to 40%, especially when it comes to CTEV of the postural type diagnosed by ultrasound on the third trimester of gestation<sup>12</sup>. It is also known that the rate of

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false positives is higher in unilateral cases (29%) than in bilateral ones (7%)<sup>7,8,9,10,12,16,18,19,20,21,22</sup>.

Furthermore, with regard to the prognosis of the deformity treatment, there are publications such as the one produced by Bakalis et al. in 2002 that reported a worse prognosis for the prenatally diagnosed patients, since they were associated with a higher possibility of rigid CTEVs of difficult management<sup>7,13,25,27,28,29</sup>.

Even if it is hard to establish before birth the severity of the deformity and the need for surgery of patients with CTEV, it has been proved that patients diagnosed prenatally can be subjected to earlier and less invasive postnatal procedures than those diagnosed after birth<sup>13,14,16,23,24</sup> (Bar-On et al., 2005; Cohen-Overbeek et al., 2006).

## **MATERIALS AND METHODS**

A descriptive retrospective study was made having as selection criteria the patients diagnosed with CTEV at the IOIR and at the practice of one of the authors (PR) between 2003 and 2012. Variables associated with prenatal diagnosis were identified such as number of ultrasound examinations performed, gestational age at the time of the ultrasonography, performance of amniocentesis, associated congenital malformations, family background, genetic counselling and beginning of treatment. The study was approved by the Committee of Clinical Practice and Research Ethics at the IOIR.

### **Data analysis**

Qualitative variables were represented as percentages. Subsequently, the equality of the data of the quantitative variables in distance measure was analysed by the Kolmogorov–Smirnov test. For the variables “prenatal diagnosis” and “days from the start of the treatment” the Mann–Whitney U test was used. Finally, an analysis was made by means of the SPSS Statistics software package, version 18.0.

## **RESULTS**

178 patients diagnosed with CTEV at the IOIR and at the practice of one of the authors between 2003 and 2008 met the selection criteria. 30.9% (n=55) were female whereas 69.1% were male (n=123).

Prenatal ultrasound diagnosis was achieved in 34.3% of the patients (n=61). In 13.1% (n=8) of the cases the diagnosis came in the 1<sup>st</sup> trimester, in 62.3% (n=38) during the 2<sup>nd</sup> and in 23% (n=14) during the 3<sup>rd</sup> one.

Among the patients prenatally diagnosed, 26% showed associated abnormalities, while in contrast the percentage increased to 42% (p<0.10) in the patients without a prenatal diagnosis. The reported abnormalities included hip dysplasia, arthrogyposis, various syndromes and fibular hemimelia.

A relation with the performance of amniocentesis was investigated. It was found that 21% (n=13) of the patients prenatally diagnosed with CTEV had an amniocentesis done, in comparison to only a 4% (n=5) (p<0.001) for the group diagnosed after birth.

31% of the patients had genetic counselling once the CTEV prenatal diagnosis was done, whereas this type of counselling was given to 17% of the patients diagnosed after birth (p<0.031).

Regarding the number of prenatal ultrasounds performed, there were statistically significant differences between the patients with a CTEV prenatal diagnosis (mean of 4.5) and those whose diagnosis came after birth (mean of 3), being higher in the first group (p<0.001).

The number of days before the treatment started once the pre or postnatal diagnosis was done was also a subject of study. Significant differences were found in the treatment start between patients with a prenatal diagnosis (mean of 9.9 days) and those diagnosed after birth (mean of 30 days) (p<0.001).

## **DISCUSSION**

Prenatal diagnosis of CTEV has repercussions on the mother and family's psychological state and in some cases it can modify the pregnancy's course. Even if the deformity's progress cannot be modified in utero, most mothers admit as useful the fact of being aware of it before their child's birth<sup>26</sup>. Although it is true that a prenatal diagnosis leaves some questions unresolved (such as rigidity) and that a false positive remains a possibility, it does allow the mother to start the treatment soon after the birth and to seek genetic counselling.

A study made at the IOIR (to be published), aiming to evaluate the results of the Ponseti method on children with clubfoot, found that the main factor leading to good results was not the rigidity level of the deformity, but an early start of the treatment<sup>30</sup>. This finding gives great importance to prenatal diagnosis of CTEV.

As far as we are concerned, the major use of a prenatal diagnosis is an early start of the treatment, which is, according to our studies, a decisive factor in the patient's final results.

The convenience of amniocentesis performance in order to look for additional malformations proposes an interesting discussion. Nevertheless we recommend first seeking genetic counselling; in the absence of further alterations amniocentesis may be discarded.

The greatest limitation of the present study is the data collection. It was collected by asking the patients' mothers, which represents a recall bias.

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