Abstract

Congenital talipes equinovarus (clubfoot) is a relatively common congenital deformity, affecting about 1 to 3 per thousand live births, generally idiopathic but sometimes associating different conditions: chromosomal anomalies, neuromuscular or urological diseases. Nowadays, the diagnosis is made prenatally, by having a detailed fetal ultrasound scan. The standard care of this condition is a non-surgical treatment according to Ponseti method; it can be initiated soon after birth and consist in manipulation of the foot and application of plaster casts, once a week, for about 4 to 6 weeks, followed by Achilles tendon tenotomy and a final cast for 3 weeks; after that, the corrected position must be maintained by applying a brace (boots connected by a bar) the child must wear inconstantly during the day, but until 4-5 years of age. The major benefit of prenatal diagnose is related to the family counseling, as long as having a baby with this kind of structural malformation will generate a spectrum of emotions to the parents and relatives; having the opportunity to prenatally present the baby's condition to the parents and discuss a treatment which is highly effective and non-invasive is very important in terms of increasing parental confidence and gaining their cooperation; those aspects are important both for psychologically reasons, preparing the family to have a baby with a visible structural malformation, but also to accept an excellent but demanding treatment, and obtaining active involvement of the family in the children's treatment: getting them prepared for seeing the baby’s leg/legs in plaster casts, several visits to the clinic, limitation in the family life during the treatment and more important transferring them the responsibility of the home treatment (the compliance to the bracing protocol in order to maintain the correction).

Keywords: Prenatal diagnosis; clubfoot; Ponseti; psychological distress.

1. Introduction

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Congenital talipes equinovarus (CTVE), or clubfoot, is one of the most common congenital malformations, affecting about 1 to 3 children per thousand live births. It is a three-dimensional structural deformity of the foot and ankle represented by equinus and varus of the heel, supination and adduction of the forefoot forming plantar cavus. Typically an isolated defect, it can also be part of a syndrome and associate other abnormalities: chromosomal anomalies, neuromuscular, urological, myotonic dystrophy, myelomeningocele, distal arthrogryposis. The etiopathogenesis of equinovarus foot has not been well established. It is, without a doubt, a multifactorial pathological process that involves maternal exposures (smoking has been shown to correlate with CTVE) (Honein, 2000) and genetic variation. Multiple studies demonstrated the importance of the vascular and muscular development, noting the absence of the anterior tibial artery, and its derivative in feet with CTVE. (Levinsohn, 1991)

2. Methods of diagnosis

The lower limb buds appear around week 5 of gestational age and in the 7th week they extend and rotate medially about 90 degrees. The ossification centers of long bones appear by week 12. Also, in the first two months of pregnancy, the foot is physiologically positioned in equinus and adduction, making it impossible to assess, at this point, if the deformity is present or not. By the beginning of month four (around the 18th week) the foot should be dorsiflexed, pronated and in mild supination. The gestational age at diagnosis range typically from 12 to 39 weeks (Mammen, 2004). The optimal momentum to perform the ultrasound for detecting CTVE in considered between 14 and 16 weeks of gestation. In our country the morphology screening ultrasound is done at 20-24 weeks gestational age, while other centers perform it between weeks 18-20. Based on the moment of ultrasound diagnosis, four types of CTVE have been described: early onset (14-16 weeks), late onset (after 20 weeks), transient talipes (initial abnormality results in normal later scans) and incomplete (inconclusive ultrasounds result in about 5% true CTEV). The sensitivity and specificity of ultrasound detection of CTEV varies, some authors suggesting that a false positive rate is higher if unilateral isolated clubfoot is suspected. (Wientroub, 1999)

At this point, the only prenatal diagnosis remains the ultrasound scan. Additional image information, after a positive diagnosis via ultrasound is not recommended or justified since there has been no correlation between the intratero aspect of the foot and the severity of the clubfoot at birth. Some authors tried to find predictive factors in the prenatal diagnosis but with no results. In a study performed by Tillet et al. 26% of the prenatally diagnosed CTEV had a mild deformity that no treatment was required. (Tillett, 2000)

Another available and routinely performed scan is 3D ultrasound (Fig. 1). In addition to conventional 2D ultrasound, a 3D scan may better describe the abnormal positions of the fetal limbs, and a real-time assessment of fetal movement has also been proven useful in the confirmation of arthrogryposis, which sometimes, clubfoot associates with (I-Wen Lin, 2008). In addition to confirming a previously suspected abnormality, a 3D ultrasound makes it possible for the parents to understand and cope with the condition, better than from an abstract description from the physician.

In theory, other diagnostic imaging is available, but it is also not indicated. A CT scan for example would only
offer information about the bone position and not show soft tissue well, but the main concern remains the unacceptable high dose of radiation, making it an unavailable option for prenatal diagnosis. Magnetic resonance imaging may show foot abnormalities and some information about the soft tissue structure, but there haven’t been any studies showing the effects on fetus exposure to MRI.

3. Associated anomalies

In most cases, the CTEV is an isolated and unilateral deformity. Having a prenatal diagnosis of bilateral CTEV may raise suspicion of other associated abnormalities, making it a syndromic CTEV. In bilateral and severe cases of clubfoot there has been demonstrated a higher incidence of associated anomalies such as: urogenital abnormalities, cardiac or neural tube defects, arthrogryposis and other muscular dystrophies (Cooke, 2008). There is a conflicting opinion in literature about performing an amniocentesis when CTEV is diagnosed. In our country, if the deformity is isolated, and no other structural anomalies are detected, amniocentesis is not recommended. Some authors have demonstrated that even isolated CTEV could have abnormal karyotypes, about 6%, arguing that a recommended amniocentesis in this cases are justified because of the subtle associated malformations that are detectable only later in the pregnancy (Shipp, 1998). We believe that this percent is fairly small and that the risks involving amniocentesis does not justify a routine karyotype evaluation in isolated CTEV.

4. Prenatal counseling

Prenatal diagnosis demands prenatal counseling, and the main purpose is to inform and reassure parents that, although we have an incomplete picture about this deformity’s etiopathology, it is completely treatable, manageable and the results obtained are excellent in literature, as well as in our experience treating this deformity.

The moment of the diagnosis of any fetal abnormality is a sensible one. The range of emotions the parents and family go through is wide and unpredictable. Many studies have demonstrated that parents usually perceive the ultrasound examination as a confirmation of a healthy fetus, making any abnormality, however benign, always a surprise, and a reason for distress (Eurenius, 1997).

The priority of prenatal counseling should be to inform the parents as soon as possible about the problems they have to face, but also to reassure them that CTEV, although a structural and obvious malformation, is treatable and the results obtained are usually excellent. There are many aspects to consider when performing prenatal counseling, one of them being to gain the trust of the parents by giving them time to analyze the information given, answering all their questions and being available when they need further clarifications. A multidisciplinary team is needed formed by the obstetrician, pediatric orthopedic surgeon and other specialized caregivers.

After the prenatal diagnosis of CTEV, studies have shown that parents go through a wide range of emotions such as depression, anger, shock, denial, fear, self-blame, guilt, sorrow, grief, confusion, despair, hostility and even emotional breakdown (Lawoko, 2004). It is well known that prenatal stress and anxiety is an important issue, and that the effects on the fetus were constantly demonstrated by numerous studies. O’Connor et. al (2002, 2003) revealed that emotional problems in children correlate with maternal stress during late gestation, while Van den Bergh and Marcoen (2004) suggested that cognitive functions of toddlers are related to maternal anxiety at 12-22 weeks of pregnancy, this being the moment when early neuron differentiation and proliferation occurs. This makes the prenatal counseling a difficult challenge, and giving the information should not be in any case delayed.

Congenital talipes equinovarus is not, in any case a life-threatening condition. It is however a structural one and usually concerns the parents about the quality of life of their unborn child. The gestation age at diagnosis is on average around 22 weeks and when isolated, parents must be well informed that it is treatable and in no way has an indication for terminating the pregnancy. If a syndromic CTEV is detected, the severity of the fetal anomalies, early detection and maternal age seem to have the most influence in a woman’s decision to continue or not the pregnancy. It is the physician’s role to reassure the parents that CTEV, in its self is not a disabling condition on the long term, but also to inform them that the treatment implies frequent visits to the doctor, patience and compliance.

Parents should also be prepared for when the baby arrives, understanding the aspect of the deformity in order to minimize the surprise factor. At this point the imaging scans come in handy, particularly the 3D ultrasound. The management of clubfoot should also be centered on the concerns of the parents, addressing their worries about quality of life. The emotional impact on the parents is also important and giving abstract information is usually not
5. Treatment

The prenatal counseling should also offer enough information about the treatment options and the results obtained by performing them. The treatment, in case of idiopathic clubfoot involves serial casting manipulation every 5-7 days soon after birth. (Fig. 2a) Usually, depending on the severity, there are 5 to 7 casts necessary. Unfortunately, there are no predictive prenatal factors to determine the severity of clubfoot, thus making prenatal counseling difficult. After the casting is completed, obtaining a normal hypercorrection, the Achilles tenotomy is performed followed by another 3 weeks cast. (Cooke, 2008) (Fig. 2b). This routine can, to some families, have an important impact financially, because of the frequent visits to the hospital, and is necessary to correctly inform the parents about the whole course of treatment.

![Fig. 2. (a) aspect of the first cast; (b) clinical aspect at the end of casting; (c) bracing.](image)

The next phase of the treatment is just as important, if not more, and it represents the bracing (Fig. 2c). The parents have to be engaged to respecting the indications and to be well informed that the healing process does not end when the casts are removed. Prenatal counseling is closely followed by postnatal management, and prepares the parents for what’s to come, but also reassures them about the nature of the deformity.

6. Conclusion

Congenital clubfoot can be easily prenatally diagnosed by ultrasound scan; the 3D imaging is useful in helping parents understanding the deformity but not to be used as severity factor; having an antenatal diagnose of this condition is helpful in planning the treatment, reducing the parents’ anxiety and increasing confidence. The availability of the caregivers is important, as well as the understanding of the emotions the parents go through. Taking special attention to diminish the mothers stress and anxiety is as important as planning a course of treatment even before the baby is born.

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