Severe hypoplasia of the right femur, ipsilateral agenesia of the fibula and twisted right foot in a 24-week-old fetus with proximal femoral focal deficiency (PFFD)

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Summary

A case of a 24-week-old fetus of non consanguineous parents with an ultrasonographic diagnosis of a short right femur, ipsilateral agenesia of the fibula and a twisted right foot is described.

Cordocentesis revealed a normal 46XY karyotype. The parents were informed on treatment options and after psychological counselling they decided to undergo an abortion.

Post mortem examination confirmed the diagnosis of a severe right limb malformation.

Key words: Unilateral femoral hypoplasia; Agenesia of fibula; Twisted foot; Proximal femoral focal deficiency (PFFD); Prenatal diagnosis.

Introduction

Proximal femoral focal deficiency (PFFD) and fibular agenesia or hypoplasia (FAH) are rare congenital anomalies of unknown etiology.

Anoxia, ischemia [1, 2], chemicals [2, 3], radiation, viral infections, hormone changes, and mechanical trauma [1, 2] have been involved in the etiopathogenesis.

PFFD occurs during the first two to seven weeks of gestation, the most important period of limb bud development.

Due to the typical PFFD characteristics, diagnosis is possible at birth. However physical examination cannot define the severity of the defect. X-rays are required to determine severity and classification.

The new modern prenatal diagnostic techniques have permitted diagnosis in an increasing number of cases before birth, which is very important for the psychological acceptance of the defect by the parents.

In this paper we describe a case of severe hypoplasia of the right femur associated with ipsilateral agenesia of the fibula and a twisted right foot diagnosed by ultrasound.

Case Report

A 36-year-old woman in her first pregnancy was referred to the Maternity Care Unit of University of Rome "La Sapienza" at 24 weeks of gestation for a routine ultrasonographic (US) examination. No family history of congenital malformation or consanguinity with the partner was present, no viral infections or mechanical trauma, and no medication or X-ray examination were performed during the first trimester of pregnancy. Ultrasound at ten weeks of gestation showed a normal finding. When the patient was referred to our service we diagnosed a short right femur, ipsilateral agenesia of the fibula and a twisted right foot by an Aloka SSD 2000. The upper limbs and the lower left limb had a length at the 50th percentile for gestational age according to the standard growth curves (METZ 1987). Left femoral length was 41 mm versus 37 mm of the right femur (Figure 1). No ecographic evidence of defects of the internal organs was recorded.

The patient underwent cordocentesis. The kariotype analysis revealed a normal 46XY karyotype and the plasmatic alpha-fetus protein levels were within the normal range according to the gestational period (14,100 mg/ml; range 9500-251,000 mg/ml).

On the basis of the ultrasonographic findings, the couple was informed about the severe lower right limb malformation of the fetus and about the therapeutical possibilities at birth.

After a detailed discussion of this pathology followed by psychological counselling, the parents decided to undergo an abortion.

Abortion was induced by intravaginal application of geneprost (1 mg). The fetus weighed 650 grams and was submitted to X-ray examination and autopsy. X-ray examination confirmed the ultrasonographic features of a right limb malformation (Figure 2). No abnormalities of the internal organs, upper limbs or left lower limb were demonstrated at autopsy.

Discussion

The case we present here clearly shows a congenital unilateral hypoplasia of the femur, which is one of the main signs of four uncommon malformations: proximal femoral focal deficiency (PFFD) [2, 4, 5, 6], femur-fibula-ulna (FFU) complex [7]; femoral hypoplasia-unusual facies syndrome [8] and limb/pelvis-hypoplasia/aplasia syndrome [9]. PFFD is characterized by the failure of normal development of the proximal extremity of the femur. Several classifications have been proposed for PFFD but the most commonly used was described by Aitken in the 1960's [10].

Aitken classified this pathology in four classes (A, B, C, D) based on the X-ray results at birth. Type A and B show elements of the proximal femur, i.e., acetabulum and head, whereas type C and D show no evidence of acetabulum and consequently no head or neck of the femur. Class A is the less severe while class D is the most severe.

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All the classes present clinically with a thigh that has the appearance of a ship's funnel and is almost flexed, abducted and externally rotated. In the newborn it is not uncommon for the ankle on the affected side to lie at the level of the knee joint on the normal side.

Other skeletal anomalies of the limbs, such as a/hypoplasia of the patella, subluxation of the knee and absence of the forth-fifth finger can be associated with PFFD [11].

PFFD and FAH are distinct malformations of the lower limb, but 70-80% [10] of PFFD also experience fibula defects ranging from mild tissue defects to complete absence of the fibula and deformed foot. Therefore complete absence of the fibula on the affected side is frequently associated with PFFD, and an overlap of clinical features suggests that these anomalies may derive from an alteration of the lower limb developmental field as Lewin and Opitz described [12].

In fact, the fibular field involves development of the fibula, of the fibular portion of the foot, of the proximal portion of the femur, and of the acetabulum and pubic bone [13, 14, 15]. The acetabulum and the femur head and neck develop from a common block of cartilage with a subsequent cleft, to create a hip joint cavity. The cleft gradually separates until a definite joint cavity is formed. The development of the acetabulum and femur head are mutually dependent [16, 17, 18].

We can postulate that the cause of this error is due to anoxia affecting the embryonic lower extremity during the formation of the cleft which later will represent the hip joint.

The aim of prenatal diagnosis should not lead to on abortion, even if the birth of a baby with this limb deficiency is always unexpected and shocking, but it should alert the parents in order to achieve acceptance of the event. Dealing with PFFD treatment, a large number of options have been described over the years, and even though uniformity of opinion has not been reached, the goal of treatment is to provide optimal functions and an acceptable appearance.

The treatment plan is based on three basic factors:

1) bilateral or unilateral anomaly;

2) severity (length discrepancy, Aitken class);

3) presence or absence of a functional foot and ankle.

Bilateral PFFD usually requires only prosthetic treatment.

Treatment options for unilateral PFFD are:

1) limb lengthening or shoe lift;

2) prosthesis with or without surgery;

3) Van Nes rotation.

The simplest treatment for the mildest case is to use a shoe lift. Another option for the mildest cases is limb lengthening, even if the validity of this technique is widely debated. Positive reports come from the Maryland Center for Limb Lengthening and Reconstruction.

What kind of quality of life would the baby in our case have had if he had been born relying exclusively on the anatomical data.

We consider that the new possibilities in prenatal diagnosis offer the opportunity to re-analyse these circumstances according to the personality of the parents. In our experience only in cases of a weak and somewhat egotistic personality does the prenatal diagnosis of severe anomalies which are compatible with a normal social life lead to selective abortion.

What is important is to consider the prenatal diagnosis with its therapeutic goals so that parents are able to maintain a positive attitude towards these babies, trying to consider them as normal babies with problems.

A multidisciplinary group should include a psychiatrist and a pediatric surgeon, who has to be an expert on prenatal diagnosis.

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