Prenatal Diagnosis of Fetal Bifid Great Toe

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Prenatal identification of hand and foot anomalies is important in view of their association with chromosomal abnormalities and genetic syndromes.1 In this report we describe the prenatal ultrasonographic diagnosis of a hallux duplication in a second-trimester detailed scan. To our knowledge, this report documents the first case of this rare form of preaxial polydactyly of the foot to be detected prenatally.

CASE REPORT

A 32-year-old woman, gravida 3, para 2, with an unremarkable family and obstetric history, underwent a routine detailed ultrasonographic examination at 19 weeks, menstrual age (MA). This revealed a singleton fetus with biometry consistent with dates, a fundal placenta, and normal amniotic fluid volume. Detailed examination of the fetal anatomy demonstrated that the right great toe was slightly separated from the other toes and showed duplication of its distal portion (Figure 1). No other anomalies were detected. In view of the association of chromosomal abnormalities with polydactyly1 or separation of the great toe,2 the option of amniocentesis was discussed but not accepted by the parents.

After an uncomplicated antenatal course, a healthy infant boy weighing 4018 grams and with Apgar scores of 9 and 9 at 1 and 5 minutes, respectively, was delivered spontaneously at 40 weeks. At pediatric examination, the only abnormality detected was the presence of an appendage on the medial side of the right toe which had a tiny nail (Figure 2A). Radiography confirmed a bifid great toe with a rudimentary distal phalanx (Figure 2B). Excision under general anesthesia at the age of 7 months was performed.

DISCUSSION

This report documents the prenatal diagnosis of a bifid great toe. This condition, one of the rarest forms of preaxial polydactyly, has been noted as part of skeletal dysplasias, genetic syndromes, and chromosomal abnormalities1,3–5 and also in association with maternal diabetes.6 Analogous

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to thumb duplication, bifid great toe has been classified into seven types according to the bony structure of the great toe, ranging from partial duplication of the distal phalanx to complete duplication involving the distal and proximal phalanges and the first metatarsal bone.7

Careful examination of the fetal hands and feet has become an increasingly significant aspect of prenatal ultrasonography.8–14 Our case was detected at a routine second-trimester detailed scan, which in our institution includes visualization of both hands and feet. Prenatal karyotyping was discussed, although the possibility of a chromosomal abnormality was considered unlikely because of the absence of other structural anomalies. Radiographic examination of the right foot after delivery demonstrated that the duplication did not involve the osseous structure of the main great toe but there was a rudimentary phalanx, corresponding, therefore, to one of the milder forms of hallux duplication reported so far. Plastic surgery was performed before walking age in view of the potential problems when the infant would learn to walk and require shoes. Hence, bifid great toe must be treated not only for cosmetic but for functional reasons to avoid growth disturbance and joint stiffness.7

REFERENCES