



CASE REPORT

Prenatal Diagnosis of Achondroplasia with Ultrasound, Three-Dimensional Computed Tomography and Molecular Methods

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KEY WORDS

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A 29-year-old female was transferred to our clinic because of short femurs (<2 percentile) on ultrasound scan. She had nonspecific medical and obstetrical history. The ultrasound scan at 32 weeks of gestational age revealed rhizomelic shortening of the extremities, frontal bossing and the over rounded metaphyseal - epiphyseal interface at the femur ends while connecting to diaphysis, also called "collar hoop" sign. A 3D helical computed tomography (3D-HCT) scan reported rhizomelic limbs, narrowing of the interpediculate distance of the lumbosacral spine, rounded iliac wings and bilateral "collar hoop" sign of the proximal femurs. All these findings led to diagnosis of achondroplasia, which was confirmed by DNA testing. A well, active male baby was born. Postnatal x-ray also confirmed antenatal findings of ultrasound and 3D-HCT. © 2012, Elsevier Taiwan LLC and the Chinese Taipei Society of Ultrasound in Medicine.

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Introduction

Achondroplasia is one of the best known and most common types of nonlethal skeletal chondrodysplasia¹. The incidence is about one in 10,000 to 30,000 [1–5]. Before the

relationship between fibroblast growth factor receptor III (FGFR3) gene and this disease was identified it was hard to differentiate between variant types of chondrodysplasias, such as metatropic dysplasia, Ellis-van Creveld syndrome, or diastrophic dysplasia [6,7]. Nowadays, because prenatal ultrasound examinations are routinely performed, more fetuses influenced by this disease are identified. The gold standard method of diagnosis is DNA testing for mutations of FGFR3 [1,2]. If family history is not present, early diagnosis is hard because of the late appearance of ultrasound

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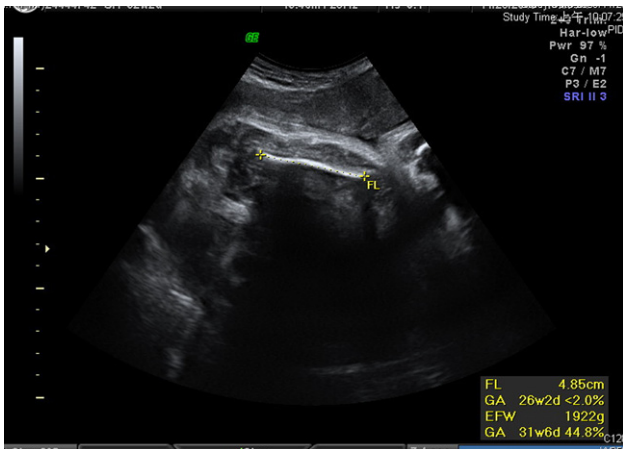


Fig. 1 Two-dimensional ultrasound at 32 weeks' gestation revealed rhizomelic shortening of the extremities. Femur length (FL) = 4.85 cm; about 26 weeks' gestation.

signs of this disease. We report a case of prenatal diagnosis of achondroplasia with ultrasound, three-dimensional helical computed tomography (3D-HCT) and genetic testing.

Case

A 29-year-old female, gravida 2 para 0, was transferred to our clinic for further examination because of short femurs (<2 percentile) seen on ultrasound examination. This woman had unremarkable medical and obstetrical history. She had regular antenatal examinations at our hospital branch and the fetus had normal growth pattern before 24 weeks of gestational age. The ultrasound scan at 32 weeks of gestational age revealed rhizomelic shortening of the extremities (Fig. 1), frontal bossing and the over rounded metaphyseal-epiphyseal interface at the femur ends with wide angle 143.55°, while connecting to diaphysis (Fig. 2). This also called "collar hoop" sign. After 6 days, 3D-HCT

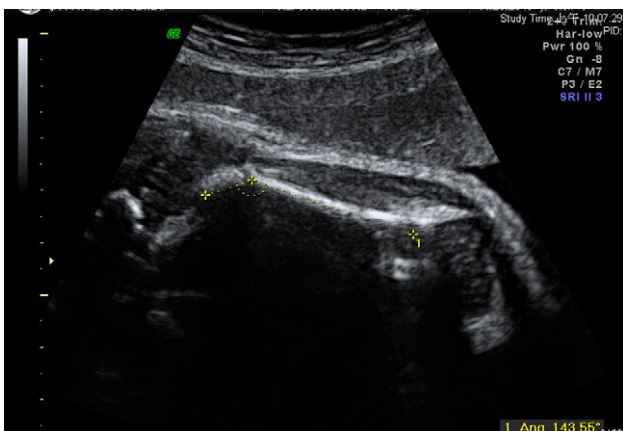


Fig. 2 Two-dimensional ultrasound at 32 weeks' gestation reported an over-rounded metaphyseal-epiphyseal interface at the femur ends with wide angle 143.55° while connecting to diaphysis. This also called "collar hoop" sign.

scan was done, which also revealed rhizomelic limbs, narrowing of the interpediculate distance of the lumbosacral spine, rounded iliac wings and bilateral "collar hoop" sign of the proximal femurs (Fig. 3). All these findings led to the diagnosis of achondroplasia, which was confirmed because heterozygote mutation in Exon 8 of fibroblast growth factor receptor III (FGFR3) was found in genetic molecular testing of amniotic fluid (Fig. 4). After counseling, patient and her husband chose to keep pregnancy. A well, active male baby was born by elective cesarean section. Postnatal radiograph was also compatible with antenatal findings of ultrasound and 3D-HCT (Fig. 5).

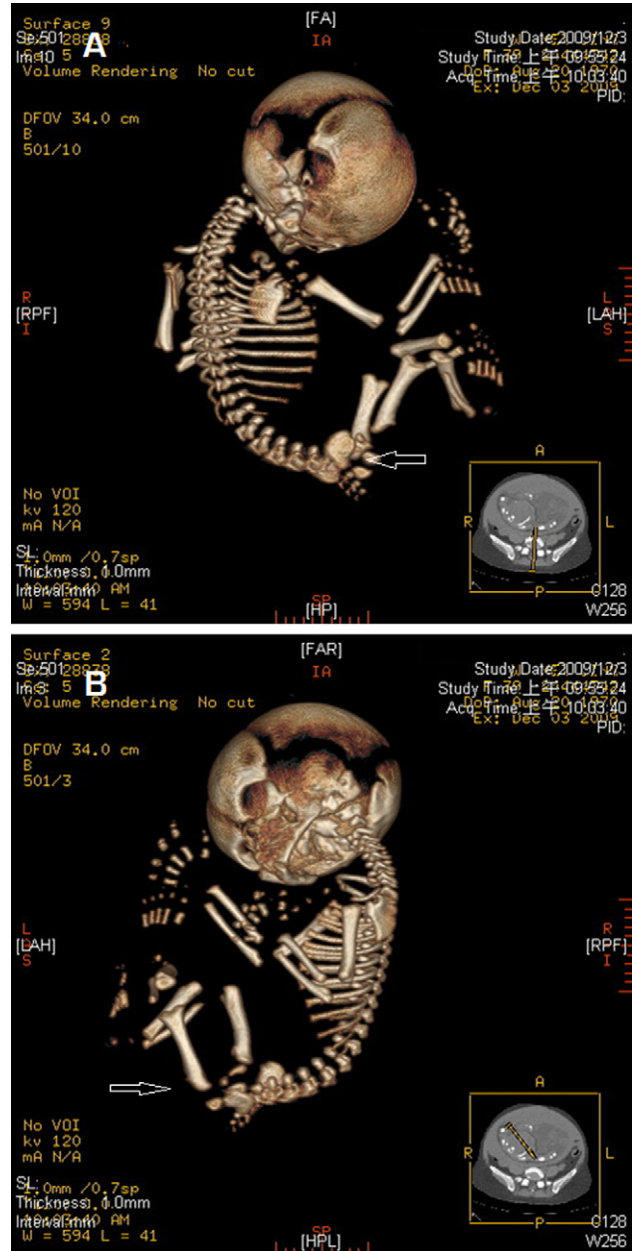


Fig. 3 Three-dimensional helical computed tomography scan at near 33 weeks' gestation revealed (A) rhizomelic limbs, rounded iliac wings (arrowhead) and (B) bilateral "collar hoop" sign of the proximal femurs (arrowhead).

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