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◇ CASE REPORT ◇

Prenatal skeletal sonographic findings of Jeune syndrome

Junko MOCHIZUKI[1], Yuji KANAI[1], Yoko ONISHI[1], Yoko KISHIMOTO[2], Kenichiro HATA[2], Nobuya UNNO[1]

Abstract

Jeune syndrome is a ciliary disorder characterized by short ribs, polydactyly, and multisystem organ abnormalities. This is a case report of a fetus terminated at 19 weeks of gestation because of short limbs, short ribs, and ascites. The fetus was found to have different mutations in both alleles of the *DYNC2H1* and was diagnosed as having Jeune syndrome (MIM #613091) caused by compound heterozygotes. We present and describe skeletal images at 18 weeks of gestation recorded using three-dimensional sonography and helical computed tomography.

Keywords

Jeune syndrome, short rib-polydactyly syndrome, ciliary disorder, prenatal, three-dimensional sonography

1. Introduction

Jeune syndrome, classified as a short rib-polydactyly syndrome (SRP), is a ciliary disorder with skeletal and multisystem organ abnormalities. Also known as asphyxiating thoracic dystrophy, it was first described by Jeune et al. in 1955[1–4]. Its frequency is estimated to be between 1 in 100, 000 to 130, 000 live births[5, 6]. It is a genetically inherited autosomal recessive disorder[6–8]. Here, we report prenatal skeletal images from a case of Jeune syndrome recorded with three-dimensional (3D) sonography and helical computed tomography (CT) at 18 weeks of gestation. We also describe the characteristics of Jeune syndrome as a ciliary disorder that is relatively new concept of a group of disorders.

2. Case report

A 34-year-old woman, gravida 2 para 1, and her 33-year-old husband are not a consanguineous couple. Neither of them had a notable medical history. The woman previously gave birth to a healthy 3, 608-g baby boy at 39 weeks of gestation.

During the second pregnancy, however, a sonographic examination performed at 17 weeks of gestation at the previous hospital revealed that the fetus had ascites and short limbs, at which time the woman was referred to our hospital.

Sonographic findings of the fetus at 18 weeks of gestation obtained using a Voluson E 8 scanner (GE Healthcare, Milwaukee, MI, USA) revealed the following: biparietal diameter, 41 mm (+1. 4 SD); abdominal circumference, 139 mm (+2. 1 SD); thoracic circumference, 74 mm; abdominal-to-thoracic circumference ratio, 0. 53 ; femur right/left, 12. 8 mm/12. 8 mm (−4. 6 SD/−4. 6 SD); humerus right/left, 10 mm/ 12 mm (−5. 9 SD/−4. 8 SD); and radius right/left, 12 mm/11 mm (−3. 9 SD/−3. 8 SD). The long bones were shorter than normal, but no distinct bowing deformity or bone fractures were observed. The fetus had short ribs, a narrow thorax, and an expanded abdomen with ascites (**Fig. 1**). The

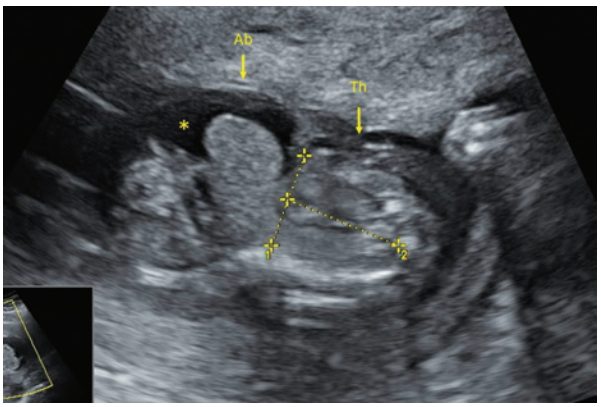

Fig. 1 Two-dimensional sonography of the fetus at 18 weeks of gestation. Sagittal view. Narrow thorax (Th) and expanded abdomen (Ad) with ascites*.

[1]Department of Obstetrics and Gynecology, Center for Perinatal Care, Child Health and Development, Kitasato University Hospital, 1−15−1 Kitasato, Minami, Sagamihara, Kanagawa 252−0375, Japan, [2]National Center for Child Health and Development, 2−10−1 Okura, Setagaya, Tokyo 157−8535, Japan

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