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Prenatal diagnosis of thrombocytopenia-absent radius syndrome

Abstract: The prenatal diagnosis of thrombocytopenia-absent radius (TAR) syndrome by ultrasound is established. The sonographic findings showed bilateral absence of the radii and club hands with normal thumbs and metacarpals. Thrombocytopenia was identified from the postabortal cord blood. Three-dimensional computerized tomography images confirmed the sonographic diagnosis. This report, to our knowledge, is one of the few cases published in the literature about the prenatal diagnosis of TAR syndrome.

Keywords: Club hands; radius; thrombocytopenia.

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Introduction

Thrombocytopenia-absent radius (TAR) syndrome is an autosomal recessive disorder characterized by bilateral radial aplasia with the presence of both thumbs and hypomegakaryocytic thrombocytopenia [4]. The presence of the thumbs distinguishes TAR syndrome from other disorders featuring radial aplasia, which are usually associated with absent thumbs. The frequency of TAR syndrome is 0.42 per 100,000 live births. If patients survive the first 2 years of life, the life expectancy is normal. The main cause of mortality is hemorrhage. Here, we present a prenatal diagnosis of TAR syndrome and the postmortem three-dimensional computerized tomography (3D CT) images of the abortus.

Case report

A 23-year-old gravida 2, para 1 woman presented to our clinic at 22 weeks’ gestation. No significant characteristics in the family history were noted. Down syndrome screening was negative in the fetus at 17 weeks (estimated risks: <1:10,000 for trisomy 21, 1:1148 for neural tube defects, and 1:7550 for trisomy 18). Ultrasonographic examination showed bilateral absent radius associated with bilateral ulna hypoplasia and club hands (Figure 1). Both thumbs and metacarpals were present in the fetus. No other abnormalities were detected. TAR syndrome was suspected. Karyotype analysis was offered to the patient but her family refused. Genetic counseling was given to the patient about radial agenesis syndromes. After the counseling, the family decided to terminate the pregnancy. Therapeutic abortion was performed, and subsequent radiographic, 3D CT (Figures 2 and 3), and macroscopic (male abortus weighing 470 g) findings were identified. Thrombocytopenia (platelets 59,000 cells/mL³) in the cord blood confirmed the prenatal diagnosis.

Discussion

The major abnormalities associated with TAR syndrome include thrombocytopenia and radial absence. Limb anomalies can affect both upper and lower limbs, although upper limb involvement tends to be more severe than lower limb involvement. The upper limbs may have hypoplasia or absence of the ulnae, humeri, and shoulder girdles. Fingers may show syndactyly, and fifth-finger clinodactyly is common. The lower limbs are affected in almost half of those with TAR syndrome, with common findings of hip dislocation, coxa valga, femoral and/or tibial torsion, genu varum, and patellar absence. The thumbs of patients with TAR syndrome are of near-normal size, but are somewhat wider and flatter than usual [1]. Other anomalies of the skeleton (upper and lower limbs, ribs, and vertebrae), heart, and genitourinary system (renal anomalies and agenesis of the uterus, cervix, and upper part of the vagina) can occur. Congenital heart disease, principally tetralogy of Fallot and septal defects, is present in one-third of cases [2]. With early prenatal diagnosis, as was possible in this case, termination of pregnancy can be performed before fetal viability. TAR syndrome is an inherited autosomal recessive trait, although alternative modes of transmission have been reported [3].
In our case, the main sonographic findings that raised the possibility of TAR syndrome were the bilateral absence of the radii, with normal thumbs and metacarpals (Figure 1). However, fetal platelet count is usually mandatory to establish the diagnosis of TAR syndrome and to differentiate it from other syndromes involving malformations of the upper limbs, such as Roberts, Holt-Oram, and Fanconi syndromes. It appears that routine ultrasonographic assessment of the long bones in addition to measurement of femur length may yield a certain diagnosis. Postmortem radiologic images also confirmed the extremity defects in this case.

Although this condition is not uniformly fatal, its severe morbidity and high mortality make early prenatal diagnosis of the disease desirable. Prenatal detection of the disease can lead to more appropriate management, including intrauterine platelet transfusion to prevent fetal hemorrhage in labor, as well as a planned atraumatic delivery. If patients survive the first 2 years of life, the life expectancy is normal. The main cause of mortality is hemorrhage. Bleeding, especially intracranial hemorrhage, may result in significant morbidity. The prognosis regarding hand and upper extremity function is usually good, providing radial aplasia is the only skeletal abnormality.

3D ultrasounds may soon become a part of routine care. Indeed, many hospitals and clinics already provide 3D ultrasounds to pregnant women as a courtesy procedure. 3D ultrasounds are already being used to detect fetal anomalies of the heart. In the near future, for actual neurological and behavioral testing of the fetus to help diagnose or rule out cerebral palsy and other anomalies, 3D ultrasounds will be used.

We conclude that TAR syndrome can be readily diagnosed prenatally with ultrasound and, if severe thrombocytopenia is confirmed by cordocentesis, therapeutic termination of pregnancy can be offered.

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