Case Report

Three-Dimensional Ultrasound Findings in Cornelia de Lange Syndrome: A Case Report

Yoichiro Akahori, Hisashi Masuyama, Yumi Masumoto, and Yuji Hiramatsu

Department of Obstetrics and Gynecology, Okayama University Graduate School of Medicine, Dentistry and Pharmaceutical Sciences, Okayama 700-8558, Japan

Correspondence should be addressed to Hisashi Masuyama, masuyama@cc.okayama-u.ac.jp

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Introduction. The objective is to report a case of Cornelia de Lange syndrome (CdLS) diagnosed by detailed observations using three-dimensional sonography. Case Report. A 33-year-old healthy multipara was referred to our hospital at 34-week gestation after severe fetal growth restriction, congenital heart anomaly, and antebrachium abnormality were diagnosed during the third trimester. Further sonography diagnosis on cardiac abnormalities diagnosed the existence of ventricular septal defect in the outflow tract, atroventricularis communis, and truncus arteriosus communis where the pulmonary artery branched from the common arterial trunk. As for abnormalities of the forearms, ectrodactylia and monodactylism were suspected and the abnormalities were observed sterically by using three-dimensional sonography. A 1986 g (1.07 percentile) male newborn was delivered by assisted breech extraction at 37-week gestation. After birth, from characteristic facies including bushy eyebrow, broad nasal bridge, micrognathia, and abnormalities of the forearms (ectrodactylia and monodactylism), the case was diagnosed with CdLS. Conclusion. Through detailed observation including abnormalities of fingers, we could exemplify this very rare disease as an antenatal diagnoses for fetal growth retardation.

1. Introduction

Cornelia de Lange syndrome (CdLS) is a rare fetal malformation characterized by severe fetal growth arrest, specific facial features, and major malformations (particularly the cardiac, gastrointestinal, and musculoskeletal systems). It has been estimated to occur in about 1 : 10,000 individuals but, as more mildly affected individuals have been reported, its actual prevalence may be much more [1–3].

This paper will report a case where Cornelia de Lange syndrome was suspected due to a diagnosis of fetal growth restriction, abnormalities of upper arms, and congenital heart diseases by detailed observations using three-dimensional sonography, and Cornelia de Lange syndrome was diagnosed with characteristic facies after birth.

2. Case Presentation

A 33-year-old healthy multipara, with no family history of Cornelia de Lange syndrome or exposure to teratogenic drugs, was referred to our hospital at 34-week gestation after severe fetal growth restriction (FGR), congenital heart anomaly, and antebrachium abnormality were diagnosed during the third trimester. In our hospital, fetal anomaly was examined in detail, using two-dimensional (2D) and three-dimensional (3D) ultrasonography (Voluson E8, GE health care Japan Co., Ltd., Tokyo, Japan). Fetal growth was restricted and type of FGR was symmetric FGR.

Further sonography diagnosis on cardiac abnormalities diagnosed the existence of ventricular septal defect in the outflow tract, atroventricularis communis, and truncus arteriosus communis.
Figure 1: Color Doppler findings of fetal heart anomaly. (a) Ventricular septal defect (arrow): atroventricularis communis (arrow head), (b) truncus arteriosus communis where the pulmonary artery branched from the common arterial trunk: truncus arteriosus communis (arrow), pulmonary artery (arrow head).

diagnostic for CdLS, and misdiagnosis is not common. The diagnosis, based solely on clinical assessment and recognition of the specific characteristics of this syndrome, is dependent on the identification of the distinctive facial features. Typical phenotypic findings include hirsutism, synophrys, long eyelashes, microcephaly, anteverted nostrils, prominent philtrum, and variable abnormalities of upper limbs.

It is not unusual in CdLS for congenital heart diseases (ventricular or atrial septal defects, aortic or pulmonary stenoses, tetralogy of Fallot, atroventricular canal, single ventricle, and aortopulmonary window) to be associated with each other. In many cases of CdLS, the antenatal diagnosis is made after postnatal evaluation [5]. After detailed evaluation of the fetal heart, this case was able to be diagnosed with truncus arteriosus communis. As the association of cardiac abnormalities expects life prognosis of CdLS, antenatal diagnosis of cardiac abnormalities is especially important.

There are several reports on antenatal diagnosis of CdLS [5–9]. By using intrauterine growth retardation as a trigger for antenatal diagnosis, CdLS is diagnosed by abnormalities of upper arms, congenital heart diseases, and characteristic facies. Recently, with the advancement of medical sonography equipment, antenatal diagnosis can be conducted more stentially using three-dimensional sonography. The past records also described the effectiveness of three-dimensional sonography for antenatal diagnosis of CdLS [8–10]. In this case, intrauterine growth retardation triggered detailed examinations; facial abnormalities and ectrodactyly and monodactyly were stentially observed using three-dimensional sonography, which was effective for antenatal diagnosis of CdLS. As CdLS is accompanied by
facial and upper arm abnormalities, three-dimensional sonography which observes a fetus sterically could be effective for antenatal diagnosis.

We experienced one case of Cornelia de Lange syndrome. Through detailed observation including abnormalities of fingers, we could exemplify this very rare disease as an antenatal diagnosis for intrauterine growth retardation.

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