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Case Report

Diagnostic problems in fetal visceral heterotaxy syndrome. Sonography vs. autopsy

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ABSTRACT

Introduction: Visceral heterotaxy syndromes (VHS) are defined as abnormalities of the determination of left-right symmetry, described as left or right atrial isomerism (LAI, RAI) [11]. Some cases do not follow classical patterns and may cause diagnostic problems. The aim of this study is to determine whether their features can be helpful or misleading in the diagnosis of VHS.

Material and methods: The study was based on 6 cases diagnosed sonographically and/or in autopsy. The results of examinations were re-evaluated and compared.

Results: Two of 6 fetuses were diagnosed to have right atrial isomerism (RAI), 3 presented left atrial isomerism (LAI) and one case was diagnosed as VHS. Cardiovascular anomalies comprised: abnormal systemic venous connections (6/6), partial anomalous pulmonary venous drainage (PAPVD) (2/6), complete atrioventricular septal defect (cAVSD) (2/6), ventricular left-hand pattern (2/6), ventriculo-arterial discordance (1/6), tricuspid atresia (1/6), mitral atresia (1/6) and tetralogy of Fallot (ToF) (1/6). Fetuses diagnosed as LAI presented complete heart block. In 2 cases of RAI and in 3 cases of LAI isomeric anatomy of the atrial appendages was observed. None of fetuses with LAI presented "polysplenia" and none of fetuses with RAI presented "asplenia". Lungs and main bronchi were isomeric in 3 cases of LAI and in no cases of RAI. One fetus with otocephaly presented atrial situs solitus and multiple cardiac anomalies, situs ambiguus of lungs and bronchi and situs solitus of the abdominal viscera.

Conclusions: There are characteristic features of left and right isomerism, and they may be present in various forms. In some cases extracardiac features of VHS can be seen only on autopsy.

1. Introduction

The word heterotaxy stems from Greek origin, with "heteros" meaning "other" and "taxis" meaning "arrangement" [1]. Heterotaxy syndromes are disorders that involve abnormal lateralization of the viscera, including thoracic and abdominal organs, as well as congenital cardiac malformations. Over the decades a lot of investigators described cases presenting features of visceral heterotaxy syndrome (VHS).

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In 1955 Ivemark first published a study describing asplenia syndrome as "teratologic syndrome of visceral symmetry" associated with cono-truncal anomalies [2]. In 1962 Van Mierop and Wiglesworth proved that not only severe cardiac malformations exist with splenic anomalies, but also atypical lobation of lungs, state of the liver and partial situs inversus may also be observed [3]. Some authors, like Van Praagh, defended the concept of asplenia and polysplenia syndrome and denied the value of atrial situs evaluation [4–6]. Anderson and Becker suggested that morphology of atrial appendages but not entire atriums is relevant [7,8]. Uemura's study correlated morphology of atrial appendages with other cardiac defects characteristic of isomeric left and isomeric right atrial appendages, confirming Anderson's concept [9–11]. Thus, the nomenclature of VHS was established basing on morphology of atrial appendages [12]. However, there exist cases which do not follow the classical patterns described, and may cause diagnostic problems on prenatal as well as on post-mortem examination.

In this study we aimed to find common features of VHS on prenatal as well as on post-mortem subjects and to determine which of them can or cannot be helpful on ultrasound examination.

2. Material and methods

The study was based on 6 cases of VHS diagnosed on ultrasound and/or on post-mortem examination. Prenatal echocardiography is reliable for diagnosis of cardiac malformations and other extracardiac defects since early gestation (17 weeks of gestation) [13,14]. In all patients two-dimensional echocardiaphic images were obtained. In those views pulsewave Doppler and color Doppler were also performed. This technique was used to visualize the flow in ductus venosus and the umbilical vessels. M-mode technique was performed to verify cardiac rhythm. All examinations were performed on an Acuson Sequoia 512 imaging ultrasound system, using a 2–5 MHz transabdominal transducer in order to obtain the best resolution, depending on maternal state and gestational age. In all 6 cases the fetuses or newborns died soon after birth. Autopsy was performed according to the guidelines of sequential segmental analysis of the heart [6,12,15]. All the examinations consisted of two main parts: general evaluation of visceral situs and the morphology of the lungs and the main bronchi, the state of the liver and spleen, and the precise anatomy of the heart. The results of ultrasound and post-mortem examinations were re-evaluated and compared.

3. Results

In four of six cases the reason of admission for prenatal echocardiography was the abnormal imaging of the heart on ultrasound by the obstetrician. In one case it was caused by increased nuchal translucency (NT) on the first trimester sonography. One patient was investigated by cardiologist because of her diabetes mellitus.

Three of six cases were described as a left atrial isomerism on ultrasound examination. The diagnosis was based on morphological features of atria (Fig. 1), the existence an interruption of the hepatic portion of the inferior vena cava with the azygos vein continuation and complete heart block with association to congenital heart disease. One patient with a fetal diagnosis of VHS had balanced atrioventricular septal defect and pulmonary trunk atresia, the second one had ventricular septal defect and transposition of great arteries, and the third had a hypoplastic right ventricle and pulmonary trunk atresia. The autopsy confirmed diagnosis of 3 cases of left atrial isomerism (Figs. 2 and 3). None of them presented polysplenia and in one case the spleen was hypoplastic.

One case of right atrial isomerism was diagnosed in utero as visceral heterotaxy syndrome and associated pulmonary trunk atresia with intact interventricular septum; it was described also on postnatal echocardiography. After birth

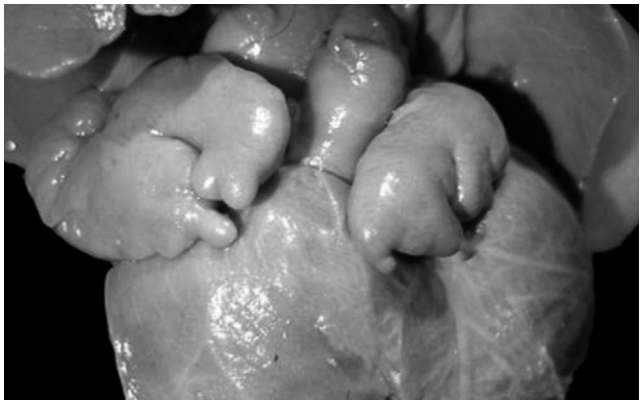

Fig. 1 – Case 3—isomeric left atrial appendages (LAA—left atrial appendage, RAA—right atrial appendage).

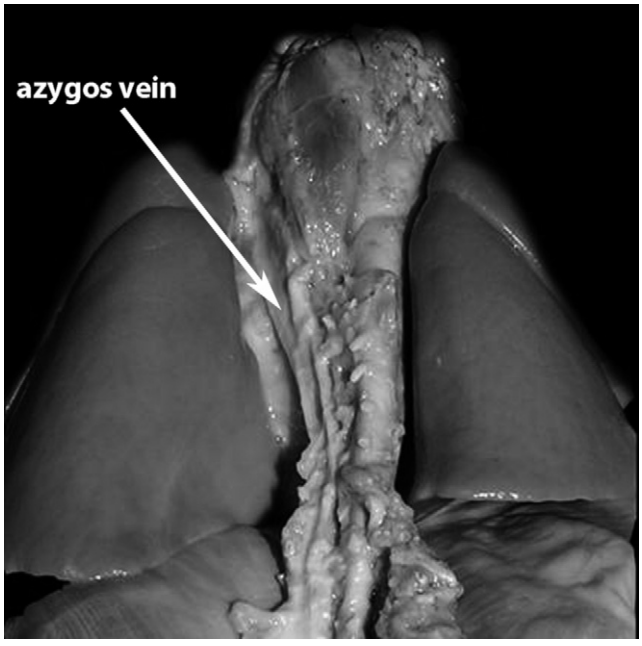

Fig. 2 – Case 2—interrupted inferior vena cava with dilated azygos vein continuation (arrow). Bilateral bilobar lungs.