CEOG Clinical and Experimental Obstetrics & Gynecology

A case of prenatally diagnosed Uhl's anomaly

M.J. Kim[1], J.Y. Bae[2], W.J. Seong[2]

[1]Yeungnam University Hospital, Department of Obstetrics and Gynecology, Dae Gu
[2]Kyungpook National University Hospital, Graduate School of Medicine, Department of Obstetrics and Gynecology, Dae Gu (South Korea)

Summary

Background: Uhl's anomaly is an extremely rare cardiac defect characterized by absence of the myocardium of the right ventricle. Until now, only three cases have been diagnosed or have showed suspicious diagnosis in prenatal period. *Case:* A 28-year-old nulliparous woman was referred to the present hospital for counseling the risk of drug medication. The authors found dilatation of the right ventricle and thinning of the right ventricular wall in the fetus at 25 weeks gestation. No other structural abnormalities were found concerning the great arteries and all heart valves demonstrated normal function. Uhl's anomaly was suspected on fetal echocardiography and it was confirmed postnatally by echocardiography and computed tomography (CT). The infant showed stable condition during neonatal period and is doing well in the ambulatory care after three-years follow up. *Conclusion:* Although the outcomes of Uhl`s anomaly are generally unfavorable, the duration of survival shows wide variation according to the cardiac function. To estimate the postnatal outcomes, it is highly recommended to perform the accurate differential diagnosis by using fetal echocardiography during pregnancy.

Key words: Echocardiography; Uhl`s anomaly.

Introduction

Uhl's anomaly is characterized by absence of the myocardium of the right ventricle. It is a very rare cardiac dysplasia first reported by Henry Uhl in 1952. The missing myocardial layer results in the formation of a very thin ventricular wall, also described as "parchment" heart, where there is no interposing adipose tissue and also no evidence of inflammation or necrosis [1]. Approximately 100 studies have been published in PubMed, but only three cases were diagnosed or showed suspicious diagnosis in prenatal period [2-4]. In the present case the authors found dilatation of the right ventricle and thinning of the right ventricular wall in the fetus at 25 weeks. Uhl's anomaly was suspected on fetal echocardiography and it was confirmed postnatally by echocardiography and computed tomography (CT).

Case Report

A 28-year-old nulliparous woman was referred to the present hospital at ten weeks for counseling regarding pregnancy related risks due to the consumption of two tablets of trimebutine maleate consumed at week 6. Two tablets of trimebutine maleate (FDA category D) were administered at six weeks of pregnancy. Quad test showed low risk at 16 weeks of pregnancy and no abnormal cardiac findings were observed. At 25 weeks of pregnancy, however, the dilatation of the right ventricle was marked on the targeted sonography with diameter of 1.44 cm, while diameter of the left ventricle was 0.86 cm. Apical trabeculation was observed, but the lateral wall of the right ventricle was much thinner than that of the left ventricle (Figure 1). No other structural abnormalities were found concerning the great arteries and all heart valves demonstrated normal function. Uhl's anomaly was suspected. Pre- and post-natal complications such as arrhythmia, hear failure, and sudden cardiac arrest were explained to the patient. The patient wanted to prolong the pregnancy, and there were no other interval changes during sonographic examinations timely performed every two weeks. In order to provide the best possible intensive care for the newborn, the labor was induced using prostaglandin E2 pessary and oxytocin at 38+3 weeks. The patient delivered a 2,670 g female with Apgar score of 8 at one minute and 9 at five minutes. No abnormal cardiopulmonary symptoms or signs were found during the postnatal intensive care. An initial chest radiograph noted marked cardiomegaly. Echocardiography of the newborn immediately after birth verified the prenatal findings: moderately dilated right inlet and wall thickness of the right ventricle was 2.8 mm. Ostium secundum type atrial septal defect with diameter of eight mm was also observed and cone-shaped patent ductus arteriosus in 1 x 2 x 3.4 mm size was found. Regurgitation of the tricuspid valve or any other cardiac dysfunction was not found. Pro-BNP (brain natriuretic peptide) was increased to 3,426 pg/ml in the blood test. A chest CT was carried out on the third day post-partum. Dilatation of the right ventricle and myocardial thinning of the lateral wall were observed. Size of the right atrium was adequate and no other unexpected cardiac abnormality was found. Diagnosis of Uhl's anomaly was confirmed by echocardiography and chest CT (Figures 2, 3). The infant showed stable condition in general and was discharged from the hospital at seven days after birth. The infant showed normal weight increase and stable body condition in the ambulatory care after three years.

Discussion

Uhl's anomaly is an extremely rare cardiac dysplasia that may cause a sudden death. Only 84 cases had been reported

Revised manuscript accepted for publication December 18, 2013

Clin. Exp. Obstet. Gynecol. - ISSN: 0390-6663
XLII, n. 2, 2015
doi: 10.12891/ceog1844.2015

7847050 Canada Inc.
www.irog.net

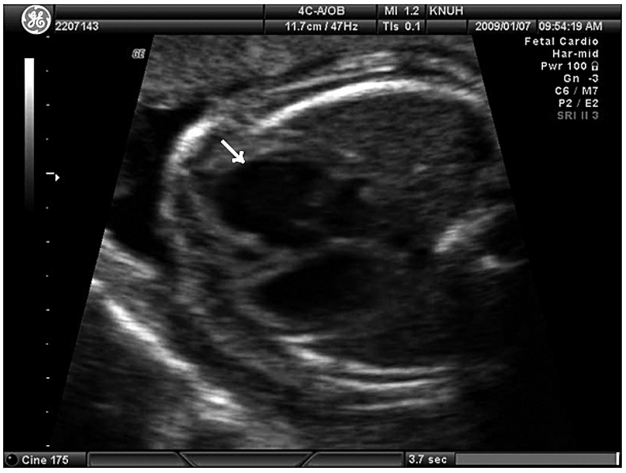

Figure 1. — Fetal echocardiography at 25 weeks gestation shows moderately dilatated right ventricle and thin right ventricular wall.

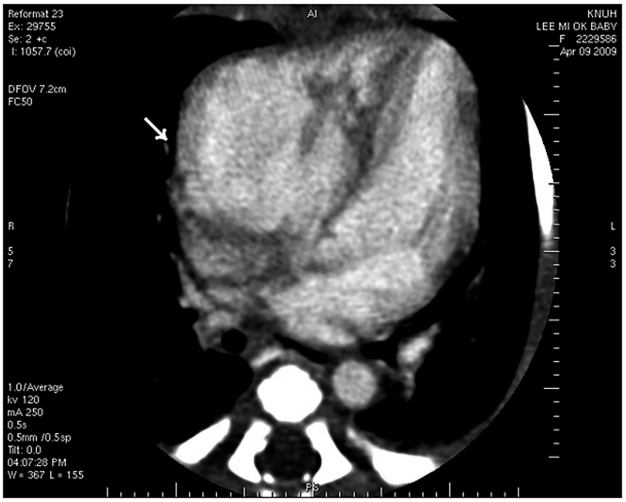

Figure 2. — Postnatal CT findings confirm very thin lateral wall of the right ventricle in a transverse cardiac image (arrow).

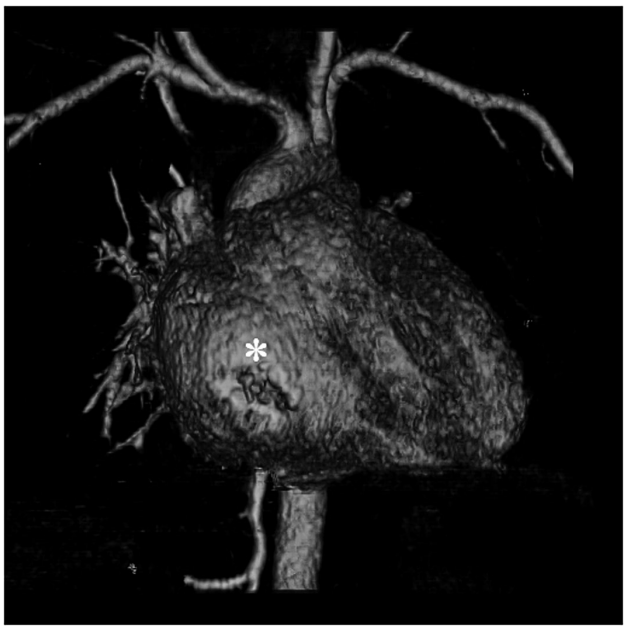

Figure 3. — Postnatal CT findings confirm very thin lateral wall of the right ventricle in a three-dimensional image (asterisk).

until 1993, and its etiology is not yet clearly determined. It is characterized by absence of the myocardium in the right ventricular lateral wall. The right ventricle is composed of apposing endocardial and epicardial surfaces with thin wall and ventricular cavity dilatation. Apart from the absence of myocardium in the right ventricle, the septomarginal trabeculation and papillary muscles of the tricuspid valve are normal. Patients' age at the time of death ranged from one day after birth to 84 years with an average of 15 years. No difference of incidence was found between males and females [5].

Majority of the cases are known to be sporadic but some cases were familial. The underlying cause is not yet determined, but some hypothesize that it is caused by primary agenesis or selective apoptosis of the cell, while others hypothesize that it is caused by recessive heredity of damaged genes or by incomplete expression of dominant genes. Exposure of toxic substances or infection sources may be another cause of the disease as well. It is important to distinguish between Uhl's anomaly and arrhythmogenic right ventricular dysplasia (ARVD). In ARVD, myocardium of the right ventricle is partially replaced with fibrofatty layer and endocardium and epicardium appear separate. Contrary to Uhl's anomaly which is mostly diagnosed in infancy, majority of ARVD cases are diagnosed in adulthood. The typical symptoms of ARVD are palpitation, syncope, ventricular tachycardia, cardiac arrest, and sudden death often related with exercise [5]. Uhl`s anomaly should also not be confused with hypoplasia of the right heart. Uhl's anomaly is rarely associated with other congenital cardiac malformations, and arrhythmias or heart block is not the general symptom of this disease.

In the past, Uhl's anomaly was mainly diagnosed by autopsy. However, prenatal and postnatal diagnoses have been increasing due to the recent development of ultrasonography and echocardiography. Total of three cases have reported the prenatal diagnosis or suspicion of the prenatal diagnosis (Table 1). In the first case on prenatal diagnosis reported by Wager *et al*., the high similarity of Ulh's anomaly and Ebstein's anomaly were shown in which pulmonary atresia was also present. However, the tricuspid valve in Wager's case was normally located in the atrioventricular groove [2]. It is uncertain whether the thinning of the right ventricular wall is prior to or a secondary reaction resulted by the dysplastic tricuspid or pulmonary valve. Some suggest to watch for the secondary reaction and not to diagnose it as an original Uhl's anomaly if other cardiac

Table 1. — *Cases of Uhl's anomaly which were diagnosed or showed suspicious diagnosis in prenatal period.*

Autour	GA	RV	RA	TV	PV	Associated anomalies	Prenatal diagnosis	Prognosis
Wager, et al. 1988	24	Large, thin	Large	Dysplastic	Atresia	None	Ebstein's Anomaly	Expired at 1st postpartum day
Benson, et al. 1995	18+5	Large, thin	Large	TR	Normal	None	Ebstein's Anomaly	Termination at 21+4 weeks
Cardaropoli, et al. 2006	31	Large, thin	Normal	TR	Normal	Pericardial effusion, hydramnios	Uhl's anomaly	Neonatal resuscitation, Asymptomatic for 1 year
This case	25	Large, thin	Normal	Normal	Normal	None	Uhl's anomaly	Asymptomatic for 6 months

GA: gestational age (weeks), RV: right ventricle, RA: right atrium, TV: tricuspid valve, TR: tricuspid regurgitation, PV: pulmonary valve.

malformations are associated [5]. There is another case reported on Uhl's anomaly diagnosed at 31 weeks of pregnancy. Unlike the previous cases, their case showed no dilatation of the right atrium, but excessive tricuspid regurgitation with pericardial effusion and thrombus was found. Heart failure at the time of birth was treated by dopamine and heparin, and further evaluation was followed for a year. No unexpected outcome was revealed [4].

The present case is the only report among the prenatal diagnosed cases that has not shown any abnormal cardiopulmonary symptoms or signs and stayed in subclinical condition. Follow-ups on this case are still ongoing. The possible reasons for this are; firstly, unlike the other previous cases, this case showed the typical signs of Uhl's anomaly such as occurrence of dilatation or wall thinning only in the right ventricle without any other dysfunctions in the tricuspid valve or the pulmonary valve. Secondly, no dilatation was found in the other right atrium. This supports the earlier study of Benson *et al*. describing dilatation of the right atrium and level of the cardiac compromise as the key factors for outcome of the Uhl's anomaly [3]. Thirdly, the reason for maintenance of the ventricular function may be because it involved only the partial region of the lateral wall in the right ventricle. Virtually, the apical trabeculations observed in prenatal period persisted even in the postnatal period.

The main cause of sudden death in Uhl's anomaly is either congestive heart failure or critical ventricular arrhythmia. Therefore, periodical observation and conservative treatment on the congestive heart failure should be performed. Implantable cardioverter defibrillator is recommended to prevent sudden death and heart transplantation should be considered when Fontan-type circulation has developed. A successful treatment in emergency situation has been reported using the following procedures: the tricuspid valve closure and a bidirectional Glenn shunt with atrial septectomy combining a partial right ventriculectomy, namely, "one-and-a-half ventricular repair" [6]. The outcomes of Uhl's anomaly are generally unfavorable. However, if it is a single lesion and only partial loss is suspicious, similar to the present case, it can be in the subclinical phase even at maturity [7]. Therefore, in order to estimate the postnatal outcomes, it is highly recommended to perform the accurate differential diagnosis by using fetal echocardiography at pregnancy.

References

[1] Uhl H.S.: "A previously undescribed congenital malformation of the heart: almost total absence of the myocardium of the right ventricle". *Bull Johns Hopkins Hosp.*, 1952, *91*, 197.

[2] Wager G.P., Couser R.J., Edwards O.P., Gmach C., Bessinger B. Jr.: "Antenatal ultrasound findings in a case of Uhl's anomaly". *Am. J. Perinatol.*, 1988, *5*, 164.

[3] Benson C.B., Brown D.L., Roberts D.J.: "Uhl's anomaly of the heart mimicking Ebstein's anomaly in utero". *J. Ultrasound Med.*, 1995, *14*, 781.

[4] Cardaropoli D., Russo M.G., Paladini D., Pisacane C., Caputo S., Giliberti P., Calabrò R.: "Prenatal echocardiography in a case of Uhl's anomaly". *Ultrasound Obstet. Gynecol.*, 2006, *27*, 713.

[5] Gerlis L.M., Schmidt-Ott S.C., Ho S.Y., Anderson R.H.: "Dysplastic conditions of the right ventricular myocardium: Uhl's anomaly vs arrhythmogenic right ventricular dysplasia". *Br. Heart J.*, 1993, 69, 142.

[6] Azhari N., Assaqqat M., Bulbul Z.: "Successful surgical repair of Uhl's anomaly". *Cardiol. Young*, 2002, *12*, 192.

[7] Güler N., Demirbag R., Eryonucu B., Gül A.: "A case of successful six consecutive deliveries in a 41-year-old woman with Uhl's anomaly". *Int. J. Cardiol.*, 2003, *87*, 283.

Address reprint requests to:
W. J. SEONG, M.D.
Kyungpook National University Hospital,
Department of Obstetrics and Gynecology,
Samduk 2 Ga 50, Jung-Gu, Dae Gu (South Korea)
e-mail: wjseong@knu.ac.kr