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신장병증, 통앞뇌증 및 양대혈관우심기시를 동반한 태아 1예

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A Case of Fetal Nephropathy, Holoprosencephaly and DORV

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We diagnosed a case of multiple fetal malformations in utero by the ultrasonography. The patient was referred at the 21 weeks of gestation because of oligohydramnios. Both fetal kidneys were hyperechoic suggesting fetal nephropathy. There was a common communication of the right and left ventricular cavity in the frontal lobe of fetal brain. One eye was placed in the middle of the face. The right ventricle of the fetal heart was dilated while the left ventricle was shrunken. Both aorta and pulmonary artery rose from the right ventricle. The pulmonary artery was smaller than aorta. Autopsy was taken after the termination of the pregnancy. Both kidneys are enlarged and revealed to the renal dysplasia under the microscopic examination. The gross findings of the brain and face were consistent with the diagnosis of holoprosencephaly. Double outlet right ventricle (DORV) was confirmed in the heart. It had a non-committed ventricular septal defect (VSD), and pulmonary stenosis.

We report this case with the review of the related articles.

Key words: Fetal nephropathy, Holoprosencephaly, Double outlet right ventricle

Introduction

We report a case of fetal nephropathy, holoprosencephaly and double outlet right ventricle (DORV), which was diagnosed prenatally and confirmed by following an autopsy on the terminated fetus. We review the related articles and emphasize the autopsy to confirm the prenatal diagnosis of the multiple fetal anomalies.

Case report

A 33 years-old-age nulliparous woman was referred because of oligohydramnios at 21 weeks of gestation. Nuchal translucency was not checked at the time of referral. Several fetal anomalies were found by the ultrasound examination. Both fetal kidneys were hyperechoic (Fig. 1). There was a common communication of the right and left ventricular cavity in the frontal lobe of fetal brain (Fig. 2). One eye placed in the middle of the face. The right ventricle of the fetal heart was dilated while the left ventricle is shrunken. Both aorta and pulmonary artery arose from the right ventricle (Fig. 3). The pulmonary artery was smaller than aorta. There is no evidence of the heart failure. We diagnosed fetal bilateral renal dysplasia, holoprosencephaly, and DORV (Double Outlet Right Ventricle) according to these findings. It made us to decide to terminate the pregnancy.

Autopsy was taken after the termination of the pregnancy. The autopsy showed both kidneys enlargement, 15 mm×20 mm×30 mm, compare to normal kidneys at GA 21weeks, 10

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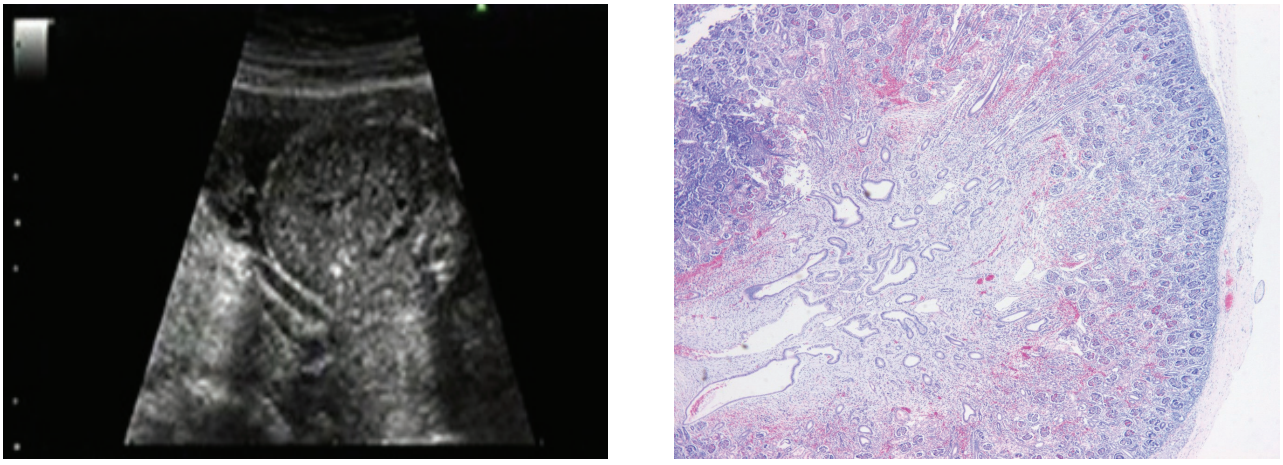


Fig. 1. Hyperechoic kidneys reveal the dysplasia. There are nephrogenesis even the unconnected atretic ureter.

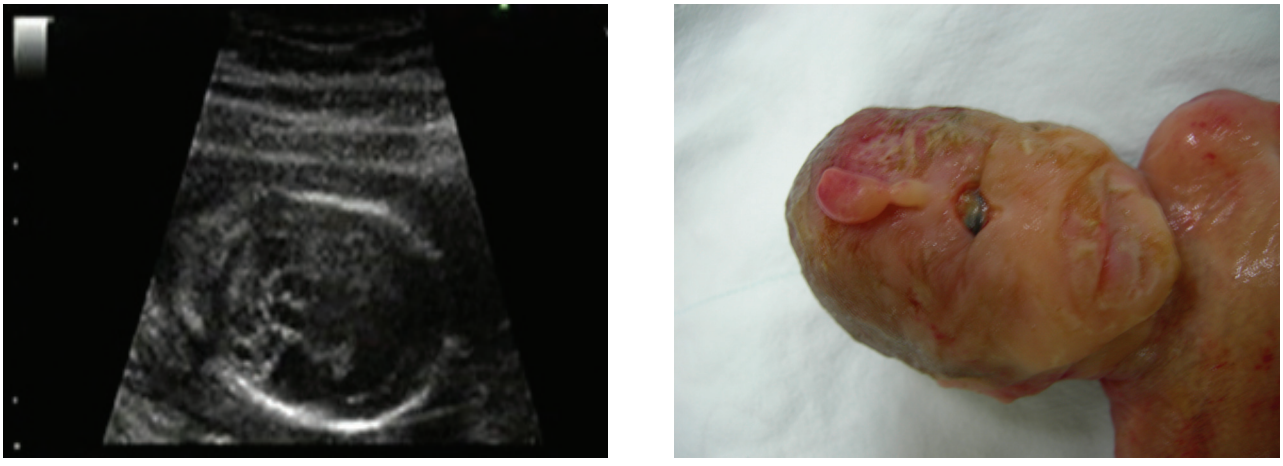


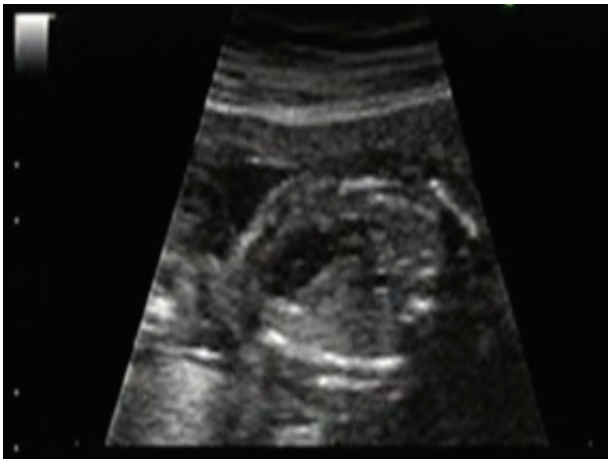
Fig. 2. The single fused ventricle is shown at the right side of the sonographic picture. The typical facial structural abnormalities are captured.

mm×12 mm×22 mm, and revealed to the renal tubular dysgenesis under the microscopic examination (Fig. 1). The brain grossly correlated with holoprosencephaly (Fig. 2). DORV was confirmed in the heart (Fig. 3). It had a non-committed ventricular septal defect (VSD) and pulmonary stenosis.

Comment

The mesonephric duct should be connected with the metanephric mesoderm, which will be the nephron, to develop as the normal urinary system. The failure of the process leads the ureteral atresia and several types of the renal dysplasia, consequently. The normal nephrogenesis may occur before the secondary changes of the dysplasia.[1] Multicystic dysplastic kidney is an extreme form of the dysplasia. If oligohydramnios and bilateral abnormalities are present, the pulmonary hypoplasia is inevitable.

The holoprosencephaly is the anomaly that results from absent or incomplete division of the embryonic forebrain, the prosencephalon. The etiology of the holoprosencephaly is autosomal dominant, recessive, and monogenic inheritance as well as infection (cytomegalovirus, toxoplasmosis), toxic



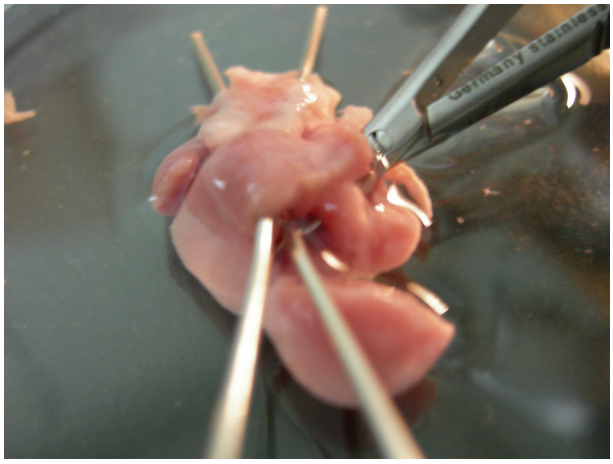


Fig. 3. Both great arteries arise from the right ventricle (RV) in a parallel fashion. The iron bars show the aorta and pulmonary artery arise from the RV and the needle holder passes through the VSD.

(hydantoin), and maternal condition such as gestational diabetes. Although chromosomal study is needed because the incidence of syndrome due to single chromosome combined with multiple congenital anomaly is 18~25% of all holoprosencephaly,[2] we could not carry out in this case because guardian refused.

The nonchromosomal, nonsyndromic holoprosencephaly carries an empiric recurrence risk of 6% because it is sporadic.[3] Two sonographic criteria for the prenatal diagnosis are intracranial abnormalities and structural abnormalities of the face such as cyclopia, cebocephly, ethmocephaly, hypotelorism, median cleft lip, single ventricular cavity, thalamic fusion, absence of median structure, and microcephaly. Extracranial abnormalities occur in approximately 50% of cases.[4] The most common anomalies are meningomyelocele, renal dysplasia, cardiac defects, and polydactyly. Only 3% of holoprosencephalic babies survive long enough to be considered a live birth. Considerations for the management of the pregnancy include elective termination of pregnancy.

The ventricular outflow tract is a common site of congenital cardiac defect. An association exists between Double-outlet right ventricle (DORV) and both trisomies 13 and 18. Also the presence of preexisting maternal diabetes mellitus seems to be a significant risk factor for the development of DORV.[5] DORV is a type of ventriculoarterial connection in which both great arteries arise from the morphological right ventricle. It may happen because of the arrest of normal rotation of the outflow tracts. The aortic outflow tract and pulmonary outflow tract tend to ascend in a parallel fashion. In almost all cases, a large ventricular septal defect (VSD) coexists. DORV is classified based on the location of the VSD; a subaortic, subpulmonary, doubly commited which means a large defect opens beneath both arterial valves, and non-committed VSD which means a defect is remote from both arterial valves. The atrioventricular septal defect is another form of non-committed defect. Additional cardiac malformations are almost always present including a VSD most commonly, atrial septal defect, pulmonary stenosis, coarctation of the aorta, and anomalous venous return. Noncardiac malformations may also present such as trisomy 18 or 13, tracheoesophageal fistula, cardiosplenic syndrome, and orofacial cleft.[6,7]

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「국문초록」

임신 21주에 양수과소증으로 전원된 환자에서 정밀 초음파를 시행하였다. 태아의 양측 콩팥은 초음파 음영이 증가되어 있었고 태아 신장병증을 의심하게 했다. 태아의 좌우 뇌실은 하나로 연결되어 보였고, 얼굴에서는 한 개의 눈이 정 중앙에 위치하고 있었다. 태아의 폐동맥과 대동맥은 모두 우심실에서 나오고, 폐동맥은 대동맥보다 좁아 보였다. 좌심실은 우심실보다 현저히 작았다.

이에 태아 신장병증, 통앞뇌증, 양대혈관우심실기시로 산전에 진단하였고, 환자 및 보호자와 상의 후 임신을 종결하였으며, 분만 후 시행한 부검에서 위의 기형들을 확인하였다

중심단어: 태아 신장병증, 통앞뇌증, 양대혈관우심기시