Patau's syndrome with aortic coarctation and pulmonary lymphangiectasia

By Manuel Recio Rodríguez, Vicente Martínez de Vega Fernández, Ana Álvarez Vázquez, Javier Urmeneta Ulloa and Raquel Cano Alonso, Hospital Universitario Quironsalud Madrid

Patau's syndrome is the third most common trisomy after trisomy 21 and 18 (1-3). In 80% of cases, the disease is associated with cardiac anomalies like patent ductus arteriosus, atrial septal defect, ventricular septal defect, tetralogy of fallot, pulmonary atresia-stenosis, double outlet right ventricle, and coarctation of the aorta. The smart-sync application allowed us to accurately assess the dimensions of the aortic arch and quantify cardiac function.

CASE PRESENTATION

The presented fetus is at gestational age of 30 weeks with an ultrasound diagnosis of cerebellar hypoplasia, hypoplasia of the corpus callosum, right microphthalmia and enlarged kidneys with small peripheral cysts. The echocardiography study showed dilatation of the right chambers with smaller left chambers and suspicion of aortic coarctation. As the visibility of the aortic arch was limited with echocardiography, a fetal MRI was performed confirming the findings of the fetal echocardiography and showing right microphthalmia.

INVESTIGATION

A fetal MRI was performed at gestational week 32 and confirmed the findings of the fetal echocardiography and also showing right microphthalmia. The fetal cardiac MRI further confirmed the dilatation of the right chambers and decreased size of the left chambers *(Figure 1)*. The ejection fraction of the right and left ventricle is both decreased with 50.77% (normal values 56%-69%) and 52.63% (normal values 69.1 ± 8.4%), respectively.

The ratio of the right cardiac output (0.34 l/min) to the left cardiac output (0.15 l/min) is highly increased with 2.26 compared to the normal value of 1.4.

The presence of aortic coarctation is confirmed. The ascending aorta measured 5.3 mm, the hypoplastic aortic arch is shortened to 2.4 mm (normal values 3.83-4.71 mm) and the proximal descending aorta is 6.4 mm *(Figure 2)*.

DIFFERENTIAL DIAGNOSIS

Fetal cardiac MRI rules out other associated cardiac anomalies and confirms the diagnosis of aortic coarctation. In addition, fetal MRI detected bilateral pulmonary lymphangiectasia predominantly on the left with small left pleural effusion not visible by ultrasound. The confirmed presence of pulmonary lymphangiectasia indicates a poor prognosis *(Figure 3)*.

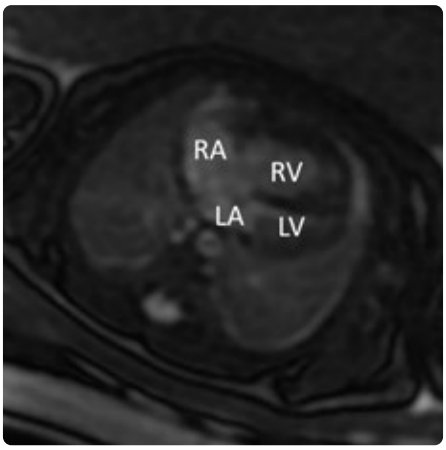

Figure 1: 2D CINE FIESTA four chambers. The fetal cardio MRI study confirmed the dilatation of the right chambers and decreased size of the left chambers. RA: right atrium; RV: right ventricle; LA: left atrium; LV: left ventricle.

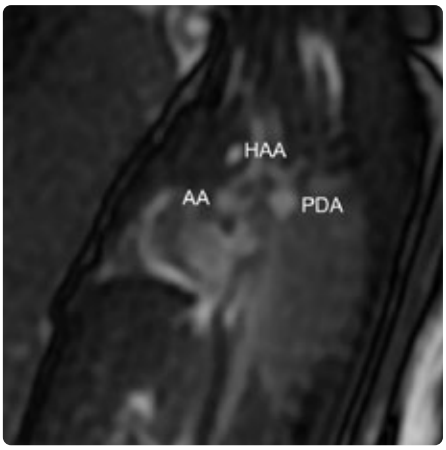

Figure 2: Sagittal 2D CINE FIESTA. The presence of aortic coarctation was confirmed, the ascending aorta (AA) measured 5.3 mm, the hypoplastic aortic arch (HAA) 2.4 mm and the proximal descending aorta (PDA) 6.4 mm.

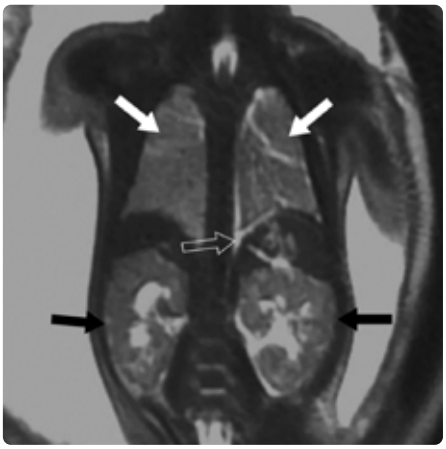

Figure 3: Coronal SSFSE T2. Enlarged kidneys with small peripheral cysts (black arrows). Bilateral pulmonary lymphangiectasia (white arrows) predominantly on the left with small left pleural effusion (empty white arrow), not visible in ultrasound.

TREATMENT

An amniocentesis was performed in gestational week 33 and trisomy 13 (Patau's syndrome) was detected in the karyotype.

OUTCOME AND FOLLOW-UP

According to the knowledge of the examination, the patient decided to terminate the pregnancy.

TAKE HOME MESSAGES

One of the main indications for fetal cardiac MRI is the assessment of the aortic arch and associated extracardiac anomalies such as pulmonary lymphangiectasia. In this case, the fetal cardiac MRI has enabled us to visualize the aortic arch and diagnose the aortic coarctation.

REFERENCES

1. Patau K, Smith DW, Therman E, Inhorn S, Wagner HP. Multiple congenital anomaly caused by an extra autosome. Lancet 1960; 1:790. DOI: 10.1016/s0140-6736(60)90676-0.
2. Ribate M, Puisac B, Ramos F. Trisomía 13 (síndrome de Patau). Asociación Española de Pediatría. 2010;1:91-95.
3. Azamian M, Lalani SR. Cytogenomic Aberrations in Congenital Cardiovascular Malformations. Mol Syndromol 2016;7:51–61