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Ventricular Septal Defect with Left Pulmonary Artery arising from Ductus Arteriosus in a Subject with 22q11.2 Deletion Syndrome

Gioia Mastromoro[1], Walter Vignaroli[2], Davide Curione[2], Aurelio Secinaro[2], Antonio Pizzuti[1], Paolo Guccione[2], Gianluca Brancaccio[2], Bruno Marino[1], Riccardo Lubrano[1], Flavia Ventriglia[1]

[1] Sapienza University of Rome, Italy, Rome, Italy
[2] Bambino Gesù Pediatric Hospital, Rome, Italy

Conotruncal heart defect is one of the most common findings in 22q11.2 deletion syndrome (22q11DS) patients, due to ectomesenchimal tissue migration anomalies.

Pulmonary arteries (PAs) anomalies are more frequent in patients with conotruncal defects and 22q11DS.

The junction between left pulmonary artery (LPA) and ductus arteriosus (DA) seems to be at risk of additional anomalies.

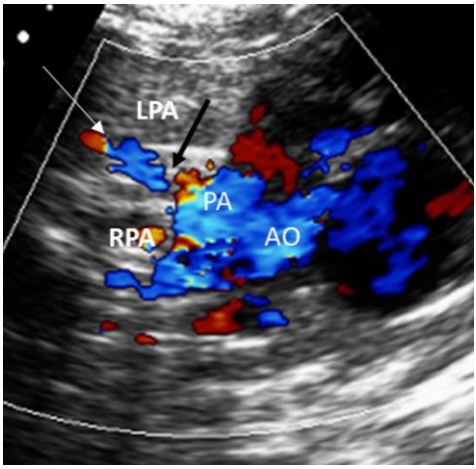
We report a rare case of **PAs discontinuity in a fetus with ventricular septal defect (VSD) and 22q11DS**, in which prenatal diagnosis had had a pivotal role for a proper and timely surgical management.

A woman was referred for fetal echocardiographic follow-up because the fetus was previously diagnosed with conotruncal heart defect.

Chromosomal microarray analysis, after amniocentesis, detected **22q11.2 deletion**.

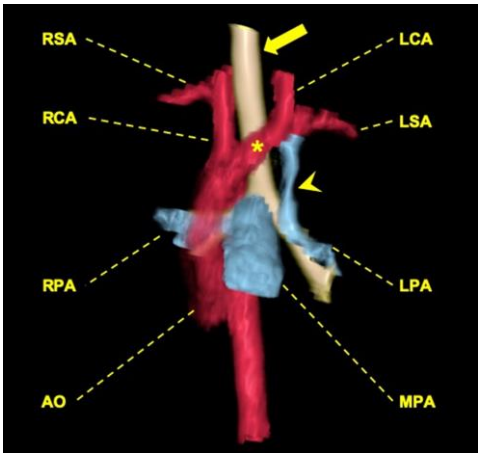
37w2d → large **VSD, without right outflow tract obstruction, right aortic arch (RAA)** and thymic hypoplasia.

There was a **minimum discontinuity between main PA and LPA and a reverse flow in LPA coming from DA was detected.**



An early surgical treatment was suggested. After Cesarean section, echocardiography confirmed malaligned subaortic VSD, RAA and discontinuous PAs. **A continuous flow, referable to DA, was detected into the LPA.**

CT scan confirmed the diagnosis and a left DA arising near left common carotid artery origin and suppling the LPA was identified. A right DA connecting the RAA and the RPA was detected.



Anomalies of LPA, such as stenosis, hypoplasia, absence, or crossing, were quite frequently reported in 22q11DS patients with conotruncal defects, including Tetralogy of Fallot and Truncus Arteriosus.

In this fetus, if the LPA anomalous origin was not diagnosed and repaired in neonatal period, the blood from DA to the left lung would have diminished and the distal portion of the LPA would have become atretic.

Despite PAs anomalies are uncommon findings in fetuses with VSD, **this case highlights how the knowledge and the early recognition of 22q11DS should suggest a particular attention to the morphology of the aortic arch and to the PAs anatomy, guiding to an early and precise treatment and improve the prognosis.**

gioia.mastromoro@uniroma1.it