View Abstract

SUBMISSION ROLE: General Abstract Submission

CONTROL ID: 3002275

TITLE: Wide and elongated aortic arch: a marker for chromosomal and genetic abnormalities

AUTHORS (FIRST NAME, LAST NAME): Julene S. Carvalho[2, 1], Lital Gordin Kopylov[3, 4]

INSTITUTIONS (ALL): 1. Brompton Centre for Fetal Cardiology, Royal Brompton Hospital, London, United Kingdom.
2. Molecular & Clinical Sciences Research Institute, St George's Hospital University of London, London, United Kingdom.
3. Ultrasound Unit, Department of Obstetrics and Gynecology, Assaf Harofeh Medical Center, Tel Aviv, Israel.
4. Sackler School of Medicine, Tel Aviv University , Tel Aviv, Israel.

ABSTRACT BODY:

Objectives: The significance of a structurally normal but abnormally shaped aortic arch has not been investigated in fetal life. We aim to report the association between abnormal aortic arch morphology and chromosomal and genetic abnormalities.

Methods: This is a retrospective case-control study. We report four cases of an abnormally shaped, wide aortic arch observed at gestational age (GA) of 20 to 23weeks (w), that were subsequently diagnosed with a chromosomal (n=1) or a gene abnormality (n=3). We compared the width of the aortic arch in these cases with that of normal fetuses (n=8, 15 measurements, at 19 to 23w). All measurements were made on a sagittal view of the aorta, from the anterior wall of the ascending aorta to the anterior wall of the descending aorta (outer-to-outer), at the level of the right pulmonary artery. In the control group, aortic width was measured by a single observer who was blinded to the values in the abnormal cases.

Results: The figure shows aortic width in cases and controls at 19-23w and suggests a linear positive relationship between arch diameter and GA (arch width = 0.8864*GA-7.2442). Average aortic width was 11.7mm in controls versus 16.4mm in the cases. The table shows details of the four cases.

Conclusions: Our preliminary data suggest that sagittal views of the aorta may provide additional important information with regard risk of chromosomal and genetic abnormalities.

Additional details

CURRENT TOPIC: Obstetrics

CURRENT SUB-TOPIC: 02. Fetal heart, CHD and cardiac function

PRESENTATION TYPE: No Preference

AWARDS:

KEYWORDS: Fetal heart, Aorta, Genetics, Chromosomal anomalies.

SUPPLEMENTAL DATA: none

TABLE:

Case #	GA	Aortic width (mm)	Other scan findings	Gene or chromosomal abnormality	Outcome
1	21w6d	16.4	Small muscular VSD	Duplication of long arm of chromosome 9	TOP
2	20w4d	19	Dysplastic mitral valve	Neonatal Marfan	TOP
3	20w6d	13.8	No abnormalities	SMAD gene	Alive and well
4	23w0d	16.6	Bilateral SVCs	Genetic syndrome	TOP

d = days; SVC = superior vena cava; TOP = termination of pregnancy; VSD = ventricular septal defect; w = weeks