ISUOG

View Abstract

SUBMISSION ROLE: General Abstract Submission CONTROL ID: 3002275

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

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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-toouter), 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.

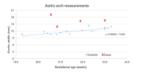
Additional details CURRENT TOPIC: Obstetrics CURRENT SUB-TOPIC: 02. Fetal heart, CHD and cardiac function **PRESENTATION TYPE:** No Preference AWARDS: KEYWORDS: Fetal heart, Aorta, Genetics, Chromosomal anomalies.

SUPPLEMENTAL DATA: none E:

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| 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



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Product version number 4.15.1 (Build 33). Build date Jun 28, 2018 11:52:46. Server c559prhs1as