Interrupted inferior vena cava in fetuses with omphalocele. Case series and review of the literature

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Background and aim: Congenital abdominal wall defects (omphalocele or exomphalos and gastrochisis) occur in approximately 1 of 2000 live births. In omphalocele, congenital heart disease is reported in 15-45% of cases. Associated abnormalities of systemic veins including abnormal angulation or interrupted inferior vena cava (Int-IVC) have occasionally been reported in children but only rarely has this been documented in the fetus. We report a case series of prenatal diagnosis of Int-IVC in association with omphalocele and review the literature.

Method: All cases of omphalocele referred for fetal echocardiography (FE) between 1997 and 2012 were identified from our fetal database. Pre and post-natal medical records were reviewed. A literature search was performed to identify any previous relevant publications.

Results: Of approximately 9,000 fetuses referred for FE 33 had an omphalocele. Mean gestational age at FE was 21+3 weeks (range: 17+5 to 24+0). Seven of the 33 cases (21%) were shown to have an Int-IVC with azygos continuation to a right-sided superior vena cava (SVC). In six, the heart was structurally normal. One fetus had a ventricular septal defect and suspected coarctation. In all seven, the defect was large, containing liver in 6/7 cases. Since 1975, there have been only 11 publications reporting systemic venous abnormalities in association with omphalocele. We identified 10 cases (with possible cases being reported >once) in >30 years prior to this study. Two of the 10 cases had been misdiagnosed as having an abnormal situs based on int-IVC but post-mortem data confirmed situs to be normal.

Conclusion: In this study we have documented the relatively common association between a large omphalocele and Int-IVC with azygos continuation to the SVC. We speculate that such an association occurs early in pregnancy as the systemic venous system develops at the same time as abdominal wall defects. Int-IVC in the presence of omphalocele is a developmental abnormality rather than an abnormality of visceral or atrial situs. This may have implications at the time of surgical repair and will influence route of any future cardiac catheterisation.