ISP-18-7  Prenatal demonstration of distal right coronary artery in normal and growth-restricted fetuses

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[Objective] We aim to investigate clinical efficacy of demonstrated distal right coronary artery (dRCA) by ultrasonography in normal and growth-restricted fetuses. [Methods] Forty-four fetuses of normal growth and 37 of growth-restricted cases were enrolled with approval from the IRB. We employed color or power Doppler to visualize dRCA at the one-third of the apex side of the right ventricular free wall. Pulsed Doppler examination was added to prove characteristic flow patterns in coronary arteries. We investigated the demonstration rate, ratios to fetal biometry, cardiothoracic area ratio (CTAR), ventricular wall thickening, amniotic fluid index, Doppler examinations in umbilical/middle cerebral artery and perinatal outcomes. [Results] Distal RCA was demonstrated frequently in FGR cases (normal 5%, FGR 51%, P<0.001). Amniotic fluid index was smaller in dRCA demonstrated (dRCA (+)) cases (dRCA (-) 12.3cm (7.5, 19.5), dRCA (+) 9.7 cm (2.6, 16.7), P=0.021) (median (max, min)) and CTAR was larger (dRCA (-) 32.4% (30.4, 42.1), dRCA (+) 40.9% (30.2, 52.8), P=0.00088) among FGR cases. We could not prove significant differences in Doppler indices and adverse perinatal outcomes. [Conclusion] Demonstrated dRCA would indicate deterioration of heart function and could be a direct and feasible sign of heart-sparing effect, which was independent of brain-sparing in FGR.

ISP-18-8  In utero stem cell therapy for Osteogenesis Imperfecta : a clinical case report

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Osteogenesis Imperfecta (OI) can be a lethal disorder that can be diagnosed prenatally. Recently, several reports demonstrated clinical benefit of prenatal human fetal mesenchymal stem cell (hMSCs) transplantation on OI patient. We present in utero hMSCs transplantation for OI type IV in our institution. [Case] Our patient was a male fetus identified at 27 weeks of gestation in a 32-year-old woman. The first daughter of this woman was affected with OI type IV with COL1A2 gene mutation. The fetal short long bones were identified with ultrasound. OI was suspected due to the clinical findings and his family history. The mother decided to join an international clinical trial of in utero transplantation of hMSCs for OI at 32 weeks of gestation. The fetus was transplanted hMSCs intravenously. After the transplantation, the fetus developed no bone fractures prenatally. The child was delivered at 37 weeks of gestation by elective cesarean section. A full-body skeletal survey at birth confirmed the presence of bilateral femur fractures. This neonate developed two new femur fractures during the first month after birth. Genotyping of the neonate revealed the same mutation in COL1A2 gene with his sister. [Summary] This is the first Japanese case report of fetal stem cell transplantation for OI. However, it is still a highly experimental therapy, and further studies are needed.

ISP-18-9  A case of X-linked VACTERL-H association with FANCB mutation

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[Introduction] VACTERL-H association is a rare disorder characterized as: Vertebral defects (V), anal atresia (A), cardiac malformation (C), tracheoesophageal fistula (T), esophageal atresia (E), radial or renal dysplasia (R), limb abnormalities (L), hydrocephalus (H). We diagnosed a male fetus as VACTERL-H association. We report his clinical course and counseling for his family. [Case] 24-years-old woman with male fetus with hydrocephalus at 23 weeks' gestation were referred. The fetus was suspected multiple congenital malformation including hydrocephalus, cardiac malformation, duodenal atresia, radial dysplasia, and renal dysplasia. Her two brothers died in the neonatal period with multiple malformations like the fetus. X-linked VACTERL-H association was suspected. During pregnancy, we maintained a counseling with the family. The neonate was born at 37 weeks' gestation. He had hydrocephalus, esophageal atresia, tetralogy of Fallot, duodenal atresia, radial dysplasia, unilateral renal dysplasia, fused vertebra, and anal atresia. Regardless of treatments, he died because of renal failure at the age of 3 months. By exome sequencing, we identified deletion of exon 3 of FANCB, which maps to Xp22.2. This study was approved by IRB of our university. [Discussion] A case of X-linked VACTERL-H association have been reported. According to these literature, FANCB mutation may cause the phenotype.