


ISP-18-4 Perinatal diagnosis and outcome of fetal skeletal dysplasia : a report of 8 cases

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[Introduction] The incidence of fetal skeletal dysplasia (SD) is 2/10,000 delivery and about 50% of them display adverse outcomes. We reported here diagnosis and perinatal outcome of cases with SD. [Materials and Methods] We experienced 8 cases of SD between 2012 and 2014 and all cases were diagnosed by ultrasonography and three-dimensional computed tomography. We investigated delivery methods, fetal and neonatal outcomes in these cases. [Results] A remarkable short femoral length was found in six cases between 15 weeks and 36 weeks. One previously suspected case with fetal heart disease was later confirmed with SD. One case was identified as Desbuquois syndrome. Imaging modalities revealed 3 cases with osteogenesis imperfect (OI), 2 cases with thanatophoric dysplasia (TD), one case each of achondroplasia (ACH) and campomelic dysplasia (CD). Five cases survived after hospital discharge and 3 cases (OI type 2A and 2 of TD) died soon after birth. Caesarian section was performed in 6 cases, and two cases had vaginal delivery. One case with prenatal diagnosis of OI later conformed as CD after birth. One case of ACH was complicated with trisomy 21. [Conclusion] A variable perinatal outcome was observed in our cases with fetal skeletal dysplasia. An accurate imaging diagnosis of SD is necessary for making delivery decision and careful perinatal management in clinical practice.

ISP-18-5 Sirenomelia associated with an abdominal cyst and a single umbilical artery

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[Introduction] Sirenomelia is a rare congenital anomalies characterized by fusion of the lower limbs. Two main pathogenic hypothesis are proposed, the defective blastogenesis hypothesis and vascular steal theory. [Case report] A 40-year-old woman, para0, An sonography revealed an abdominal mass in fetus at GA13weeks. At GA14weeks oligohydramnios was seen and a single blood vessel flow alongside the abdominal mass coursing into the umbilical artery. At GA17weeks abdominal mass was enlarged to 46*16mm. Close observation of the limbs were limited by the oligohydramnios. Infant prognosis was considered poor due to the sever oligohydramnios, patient chose a termination. An infant weighing 226g was delivered vaginally at GA19weeks. Infant showed bulging abdomen, fusion of lower limbs, anorectal atresia, absent external genitalia, and diagnosed as Sirenomelia. Prenatal diagnosis was missed. On autopsy, there was a pulmonary hypoplasia, duodenal atresia, absence of right kidney, abdominal cyst that revealed by sonography proved to be saccular cloaca ; cranial part was intestinal epithelial tissue and caudal part was bladder. [Conclusion] Detection of the lower limbs by sonography and an assessment of the aberrant abdominal vascular of the fetus using color and power Doppler could be useful for diagnosis. Prenatal diagnosis of Sirenomelia is beneficial for family to predict prognosis of infant.

ISP-18-6 The survey of thrombocytopenia in newborn babyAiiiku Hospital¹, The University of Tokyo Hospital², Saitama Medical University³Takahiro Yamashita¹, Takeshi Nagamatsu², Yoshimasa Kamei³, Tomoko Adachi¹, Yutaka Osuga², Takashi Okai¹, Masao Nakabayashi¹, Tomoyuki Fujii²

[Objective] It is well known that maternal ITP can cause thrombocytopenia in the newborn baby. Recently, neonatal allo-immune thrombocytopenia (NAIT) has been noticed as another cause of thrombocytopenia. Because the complete blood count (CBC) is not routinely tested for newborn baby, the frequency of NAIT has not been known. In this study, we analyzed the CBC data of the newborn babies hospitalized in NICU. [Methods] This is a retrospective study. Since 2012 to 2014, the 218 newborn babies hospitalized in NICU in our hospital. The 207 babies' platelet count was analyzed. We did not apply this study to the ethics committee because this is a retrospective study of the medical records. [Results] Thirty three babies (15.9%) showed the platelet count under 150,000/ μ L. Nine babies had a chromosomal abnormality, eight babies were born from mothers suffered from PIH or HELLP syndrome. The other 16 babies' causes of low platelet count were unclear. The babies of NAIT might be included in these 16 babies (7.7%). There were no cases who presented symptoms of low platelet such as subcutaneous bleeding. [Conclusion] About one sixth of newborn babies were found to show a low platelet count, and 7.7% babies could be NAIT. This is a pilot study and the additional study is needed to unveil the actual frequencies of NAIT among the newborn babies.