Supporting information can be found in the online version of this abstract

P04.12
Split notocord syndrome associated with neuroenteric fistula

Y. Dogan, G. Yucesoy, S. Ozkan

Kocaeli University, Kocaeli, Turkey

Split notocord syndrome is an extremely rare form of spinal dysraphism with an endoectodermal fistula opening in the dorsal area, associated with gastrointestinal tract, central nervous system and urogenital anomalies. We present a case of split notocord syndrome with meningomyelocele and dorsal herniation of bowels in thoracolumbosacral region.

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P04.13
Agenesis of the cavum septum pellucidum in the third trimester of pregnancy

I. Pelayo[1], L.L. Abarca[1], I. Tamarit[2], M. Recio[3], G. Nadal[1], J. Lazaro[1]

[1]*Obstetrics, H. Ramón y Cajal, Madrid, Spain;* [2]*Obstetrics, H. Universitario Quiron, Madrid, Spain;* [3]*Radiology, H. Universitario Quiron, Madrid, Spain*

Introduction: The visualization of the cavum septum pellucidum is an important marker of neuroencephalic proper development. The suspicion of absence requires to perform a throrough study of brain structures. In case of absence, the prognosis of affected children is variable.

Case Report A 34 year old woman primigravida of 29 weeks of pregnancy, arrives to perform a ultrasound report. Prior control of gestation in a private center without significant abnormalities. First trimester ultrasound: Nuchal Translucency: 1.1 mm (CRL: 62 mm < P95) First trimester combined screening: low risk (combined risk trisomy 21: 1/542; trisomy 18 < 1/10.000). In the third trimester ultrasound it was seen a defect in the middle line of the brain. No cavum septum pellucidum is displayed, with extensive communication between the lateral ventricles anteriorly. The corpus callosum is present. Ventricular atriae appear in a limit size (9.7 mm).

In MRI there was seen the absence of cavum septum pellucidum, as well as a deformity of the lateral ventricles and mild ventriculomegaly (10.7 and 11.7 mm). There is no evidence of agenesis or dysgenesis of the corpus callosum. It also appeared to be a nonfusion of frontal lobes suggesting lobar holoprosencephaly.

The patient decides to finish her pregnancy in France. The necropsy showed isolated septal agenesis without ventricular abnormalities. The corpus callosum was present. No other abnormalities including ocular or optic chiasm alterations were present. The karyotype of amniotic fluid was normal (46 XY).

P04.14
Embryonic diagnosis of encephalocele: a case report

S. Ramakrishnan[1,2], M. Bharadhi[1], I. Suresh[2], S. Seshadri[2]

[1]*Fetal Medicine Department, Mediscan Ashvita Fetal Medicine Centre, Trichy, India;* [2]*Fetal Medicine Department, Mediscan Systems, Chennai, India*

Encephalocele is a type of neural tube defect that occurs very early in fetal life and can be effectively diagnosed by prenatal ultrasound. Here we present a case report, where an early diagnosis was made in the embryonic period.

Mrs. S.V a 22 year old, a second gravida presented at 8 + 5 weeks in her second pregnancy. Their first pregnancy was terminated at 20 weeks due to ultrasound diagnosis of encephalocele, polydactyly and polycystic kidneys. The couple had elected to terminate the pregnancy and the diagnosis was confirmed after delivery by a fetopathological examination. In their current pregnancy, at 8-9 weeks ultrasound examination revealed an abnormal posterior fossa raising a suspicion of encephalocele. She was re-assessed at 11 weeks, as examination of the limbs and kidneys would be difficult in the embryonic period and ultrasound examination confirmed the diagnosis of Encephalocele. Additionally there was polydactyly. The kidneys were examined, however polycystic kidneys were not clearly discernable at that stage. Based on the above findings a recurrence of Meckel Gruber Syndrome was made. The couple were counseled regarding the above findings and they opted for termination of pregnancy.

The case is presented because there are no reports of early diagnosis of encephalocele as early as 8 -9 weeks of gestation in the embryonic period on literature survey. Further this report emphasizes the fact that many abnormalities can be identified in the first trimester itself at the time of examination of nuchal translucency.

When an occipital encephalocele is encountered on pre-natal ultrasound, it is important to examine the remainder of the fetus for abnormalities such as polydactyly and cystic dysplasia of the kidneys so that a proper diagnosis is made.This case is unique because a diagnosis of an abnormal posterior fossa was made at 8–9 weeks.In the current case, the family history of a previously affected child made the diagnosis possible in the first trimester. This case illustrates the importance of a detailed anatomical survey at an 11–14 weeks scan.

P04.15
Craniosynostosis in the third trimester of pregnancy

I. Pelayo[1], L.L. Abarca[1], I. Tamarit[2], M. Recio[3], G. Nadal[1], J. Lazaro[1]

[1]*Obstetricia, Hospital Ramon y Cajal, Madrid, Spain;* [2]*Obstetricia, H. Universitario Quiron, Madrid, Spain;* [3]*Radiology, H. Universitario Quiron, Madrid, Spain*

Introduction: craniosynostosis is the premature fusion of one or more cranial sutures causing a cranial deformity. It appears in 0.2/10.000 rn. These include the type II Pfeiffer syndrome consisting of a osteochondrodysplasia and craniosynostosis and Crouzon S. characterized by the triad of cranial deformities, facial abnormalities and exophthalmos.

Case Report: a pregnant woman, 39 years old, in the third trimester of pregnancy (31 + 4 weeks of gestation), obese (BMI: 36) who comes to control visit. Previous pregnancy tests without clinically relevant changes.

On her 1st trimester ultrasound Nuchal translucency was normal (CRL: 71 mm, 13 + 2 weeks gestation, 1.4 mm < P95). First trimester combined screening: low risk (combined risk trisomy 21: 1/2001; trisomy 18 < 1/10.000).

On her second trimester ultrasound only a persistent right umbilical vein was objectified, without other significant abnormalities.

In the 3rd trimester ultrasound (2D -3D) were also detected: cranial deformity with "cloverleaf skull": Craniosynostosis at the