

Results:

1. In the isolated group, only 1 genetic result was found to be suspected of pathogenicity. In the non-isolated group, the genetic results of 10 fetuses were found to have abnormal pathogenicity. The difference in pathogenicity between the two groups was statistically significant.
2. In the isolated group, 7 fetuses were terminated, and 5 fetuses were delivered at term.

Follow-up after delivery: 1 baby had increased muscle tone in his lower limbs and dorsiflexion of both feet, no obvious retardation of growth and intelligence were found in the other 4 babies. In non-isolated group, 19 fetuses were terminated, 5 fetuses were delivered at term. Follow-up after delivery: 1 baby suffered from seizures; 1 baby had bilateral tibialis anterior muscle tension and excessive flexion of both feet, requiring rehabilitation treatment; 1 baby died 10 days after birth due to dysphagia; 2 babies were mentally retarded.

Conclusions: When the fetuses with corpus callosum hypoplasia and partial absence were associated with other structural abnormalities, the probability of genetic abnormalities was 52% (10/23), and almost 100% of the children had poor long-term prognosis according to our study. The incidence of genetic abnormalities in the group of isolated corpus callosum abnormalities were 10% (1/10), and 80% of the children had a normal long-term prognosis. The probabilities of genetic abnormality and long-term poor prognosis of fetuses with non-isolated corpus callosum abnormalities are significantly higher than those of fetuses with isolated callosal abnormalities.

EP09.13

‘The face predicts the brain’: a case report of Alobar holoprosencephaly

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Holoprosencephaly is a rare spectrum of cerebral and facial malformation that can be detected by routine ultrasound during antenatal care. Its prevalence is estimated at 1/10,000 live births and stillbirths and 1/250 products of conception; the distribution is worldwide. A 31-year-old gravida presented for routine obstetric ultrasonography. There were no symptoms or signs suggestive of any clinical condition. Diabetes mellitus was discovered during the prenatal work-up, and she was not on any medications. There was no consanguinity between parents. Ultrasound revealed a single live fetus with a calculated gestational age of 14 weeks. Both thalami fused and corpus callosum, interhemispheric fissure, cavum septum pellucidum and 3rd ventricle were absent. The cerebellum is clearly hypoplastic with ocular sketches, absence of nose. These features were found in association with the presence of polydactyly. Taking into consideration the mother's will, our approach was to terminate the pregnancy. The postnatal examination was concordant with the ultrasound data.

Holoprosencephaly is a condition that occurs in the first two or three weeks of pregnancy and results in abnormal development of the brain. The short-term treatment and outcome depends on the severity of HPE and any associated features. It's symptomatic and supportive and requires multidisciplinary management.

EP10: NEURAL TUBE DEFECTS AND CRANIAL ANOMALIES

EP10.01

A case report of fetal spinal meningocele

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Neural tube defects are congenital malformations of the CNS resulting from defective closure of the neural tube during early embryogenesis between 3rd and 4th week of intrauterine life. It involves defect in the skull, vertebral column, the spinal cord and other portion of CNS. It occurs about 1 to 5 per 1000 live births. Myelomeningocele is most common and severe form of spina bifida cystica, characterised by protrusion of spinal cord through the open vertebrae into the amniotic fluid.

We present a case of a 25-year-old pregnant woman who came into our hospital in the 17th week of pregnancy with characteristic ultrasound signs for the presence of fetal spina bifida. Lumbosacral meningocele was present. The anterior part of the head started to develop “the lemon sign” and cerebellum showed “the banana sign”. Dilated lateral ventricle was also present. There was anamnesis for folic acid supplementation starting at 6 weeks of gestation. We performed induction of labour with oxytocine and termination of this pregnancy. The autopsy report showed the presence of spina bifida in the lumbosacral region, with a defect of the skin and the vertebral arches, measuring 1.7 × 1 cm, with protrusion of meninges and the spinal cord.

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

EP10.02

Pathophysiology and intrauterine appearance of unusual causes of fetal acrania-exencephaly-anencephaly sequence

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Objectives: Diagnosis of fetal acrania-exencephaly-anencephaly sequence (AEAS) is usually straightforward, however early detection and classification of causative conditions prior to disaggregation of exposed dysplastic brain tissue remains challenging. We aimed to further characterise this lethal fetal condition and give further insights into the pathophysiology.

Methods: A systematic literature search has been conducted and tabulated all cases representing an unusual shape of the fetal cranial pole caused by a segmental amniotic rupture and added six new