Encephalocystocele – Uncommon Diagnosis in Prenatal Medicine

Robert Dankovcik, Vlasta Vyhnalkova, Stela Muranska, Eduard Kucera, Maria Korpova, Anna Plichtova, Maria Miklosova, Vladimir Ferianec, Jan Evangelista Jirasek, Marek Dudas

1 Second Department of Obstetrics and Gynecology, Comenius University and University Hospital, Bratislava, Slovakia; 2 Second Department of Obstetrics and Gynecology, P.J. Safarik University and L. Pasteur University Hospital, Kosice, Slovakia; 3 Center for Prenatal Diagnosis, s.r.o., 4 Department of Forensic Medicine and Pathology, Health Care Surveillance Authority of Slovak Republic, and 5 Fetal Medicine Program, Department of Cell Biology, P.J. Safarik University, Kosice, Slovakia; 6 Department of Gynecology and Obstetrics, Third Faculty of Medicine, Charles University in Prague, and 7 Institute for Care of Mother and Child, Prague, Czech Republic

Encephalocystocele is a developmental malformation characterized by brain herniation accompanied with extracranial cystic protrusion of the ventricular system. This nosological unit is often overlooked and insufficiently classified merely as encephalocele. Herein, two exceptionally clear cases of the parieto-occipital cranioschisis with encephalocystocele and congenital hydrocephalus of the lateral ventricles are documented with 2-dimensional/3-dimensional sonographic images and the corresponding MRI findings. In both cases, prenatal diagnosis was confirmed by autopsy.

Key Words
Cephalocele · Encephalocystocele · Meningocele

Abstract
Encephalocystocele is a developmental malformation characterized by brain herniation accompanied with extracranial cystic protrusion of the ventricular system. This nosological unit is often overlooked and insufficiently classified merely as encephalocele. Herein, two exceptionally clear cases of the parieto-occipital cranioschisis with encephalocystocele and congenital hydrocephalus of the lateral ventricles are documented with 2-dimensional/3-dimensional sonographic images and the corresponding MRI findings. In both cases, prenatal diagnosis was confirmed by autopsy.

Clinical Case 1
Unilateral Encephalocystocele in an Otherwise Normal Brain
A single live fetus [gestation week (GW) 18; healthy mother/age 29, gravida 3, para 2] affected with prominent dilatation of the left lateral ventricle and atrophic brain tissue was identified by sonography (fig. 1 a, b). MRI in GW 24 revealed parieto-occipital encephalocystocele (fig. 1 c, d). Cesarean section at GW 38 delivered a full-term newborn girl, eutrophic, 4,020 g, 51 cm, APGAR score 7/6, normal motorics, head circumference 44 cm. No malformations of the heart, kidney or other organs were found on sonography; small amounts of milk were ingested per os. The soft parieto-occipital protrusion (fig. 1 e, f) showed no cerebrospinal fluid leakage or bleeding. Dyspnea and cyanosis developed 3.5 h postpartum; oxygen inhalation restored blood saturation to 99%, but inoperability was stated by a neurosurgeon. Sudden apnea and bradycardia appeared 5.5 h later, followed by asystolia after another hour. Autopsy revealed a largely normal brain with a normal left frontal lobe and a normal anterior portion of the left lateral ventricle. The expanded posterior part of the left lateral ventricle was covered in thinned differentiated brain tissue and herniated via the distended fontanella occipitalis (fig. 1 g, h).
Fig. 1. Case 1: unilateral encephalocele (*) in the otherwise normal brain. For easy visual comparison, all head and brain images are oriented with the front facing the right side, right brain hemisphere up (all except c, e) or above the image plane/facing the observer (c, e). a Two-dimensional ultrasound at GW 18, horizontal section through the fetal head with normal right lateral ventricle, dilatation of the left lateral ventricle, and left occipital encephalocele (*). b Three-dimensional rendering at the same age and on the same sonographic plane as in a. c Sagittal fast-spin MRI at GW 24 shows a parieto-occipital skull defect (widened and perforated fontanella occipitalis) with encephalocele (asterisk in cystic protrusion of the left ventricle) covered in an additional protruding sac (meningocele, mc) formed by the dura mater (labeled arrowhead). d Horizontal fast T2-weighted MRI at GW 24 reconfirms the herniated brain and meninges together with a posterior part of dilated left lateral ventricle (*). e Dura-covered protruding parieto-occipital encephalocele (*) 1 h after live delivery. f Horizontal two-dimensional sonogram 6 h after the delivery shows persisting extreme dilatation of the ventricular system, with thinned brain tissue on the left side forming a 1-cm-thick layer (arrows). g,h Brain autopsy, basal (inferior) view. Normal overall anatomy and brain thickness was revealed, except for the herniated (*) posterior portion of the left lateral ventricle, which was dilated and surrounded by thinned and smoothened brain tissue (lissencephaly) of the temporal and occipital lobe. c.inf. = Cornu inferior ventriculi lat; c.post. = cornu posterior ventriculi lat; crbl./flip = cerebellum, flipped frontally; F = frontal direction; L = left side; lob. occipit. = lobus occipitalis cerebri; lob. temp. = lobus temporalis cerebri; LV = left lateral ventricle; mc = meningocele; R = right side; RV = right lateral ventricle; sep. pell. = septum pellucidum; thal.sin. = left thalamus.
Clinical Case 2

Unilateral Encephalocystocele Associated with Severe Brain Malformations

Right parieto-occipital encephalocystocele (fig. 2a) was identified by sonography in a single live fetus (GW 20; healthy mother/age 29, gravida 1, para 0). MRI at GW 21 reconfirmed unilateral hemisphere dilatation, with the wall of widely communicat-
at autopsy (asterisk in fig. 1g). This substantiates a speculation that antenatal displacements, folding or perhaps rupture of extremely thinned brain inside the encephalo-cystocelic pouch could imitate meningocele, or otherwise mislead the sonographist. In conclusion, encephalo-cystocele should be routinely suspected and carefully differentiated in all cases of prenatal sonographic diagnosis of any cephalocele.

Acknowledgements

This work was supported by the projects APVV VVCE-0001-07, ASFEU ITMS 26220120024-SEPO and the Slovak Ministry of Health grant No. 2007/65-UPJS-02.

Reference