J. Fetal Med. (September 2019) 6:133–137
https://doi.org/10.1007/s40556-019-00211-z

ORIGINAL ARTICLE

Prenatal Diagnosis of Atretic Occipital Cephalocele: A Case Report

Çiğdem Kunt İşgüder[1] · Özlem Koşar Can[2] · Mine Kanat Pektaş[3] · Erkan Gökçe[4]

Received: 20 May 2019 / Revised: 23 June 2019 / Accepted: 8 July 2019 / Published online: 25 July 2019

Abstract Atretic cephaloceles refer to the congenital herniation of meningeal and vestigial tissues such as arachnoid, glial or neural rests. These small, skin covered subscalp lesions usually appear within a few centimetres of the lambda and nearly half of them have a parietal situation, the remaining half have occipital, parieto-occipital, frontal, asterion, and sincipital locations. Atretic cephaloceles can be isolated or associated with congenital syndromes, agenesis of corpus callosum, grey matter heterotopias, ventriculomegaly, mental retardation, developmental delay, epilepsy, spasticity, speech difficulty, strabismus, optic nerve atrophy, microphthalmia, enophthalmos, cleft palate, hypertelorism, congenital cardiac and vascular defects, renal agenesis, hearing problems, congenital lobar emphysema, and muscular anomalies. This case report describes a newborn which has been diagnosed with atretic occipital cephalocele prenatally and also bilateral cochlear hypoplasia postnatally.

Keywords Atretic cephalocele · Cochlear hypoplasia · Occipital

✉ Çiğdem Kunt İşgüder
cidos_55@hotmail.com

[1] Department of Obstetrics and Gynecology, Faulty of Medicine Istanbul, Istanbul University, 34093 Istanbul, Turkey

[2] Department of Obstetrics and Gynecology, Faculty of Medicine, Pamukkale University, Denizli, Turkey

[3] Department of Obstetrics and Gynecology, Faculty of Medicine, Afyonkarahisar University of Health Sciences, Afyonkarahisar, Turkey

[4] Department of Radiology, Faculty of Medicine, Gaziosmanpaşa University, Tokat, Turkey

Introduction

A cephalocele is defined as the congenital herniation of intracranial structures through a skull defect while an encephalocele is a cephalocele which contains central nervous system tissue. An atretic cephalocele is the congenital herniation of meningeal and vestigial tissues such as arachnoid, glial or neural rests. These rudimentary lesions are also called as atypical or rudimentary meningoceles, meningoceles manque', cutaneous meningiomas or meningeal heterotopias. Historically, atretic encephaloceles have been named as hamartomas as they are fibrous structures that are not associated with central nervous system. Although these malformations have better prognosis than encephaloceles, their clinical course may be complicated with co-existing pathologies [1–3].

This case report describes a newborn which has been diagnosed with atretic occipital cephalocele prenatally and also bilateral cochlear hypoplasia postnatally.

Case Report

A 28-year-old, multiparous woman with 18-week-old pregnancy was admitted to the study center due to routine follow up. The patient revealed that she had two uneventful vaginal deliveries and her marriage was not consanguineous. She had no known history of smoking, alcohol intake or drug use and there was also nothing particular in her medical or surgical history. It was learnt that the patient had a normal pregnancy with folic acid intake in the first trimester and she did not undergo any screening test for Down syndrome on her own will.

Obstetric ultrasonography showed a fetus with biometric measurements that were compatible with 18 weeks of

gestation as well as a normal placenta and amniotic fluid index. On the other hand, mild ventriculomegaly of 11 mm and an occipital solid mass of 2 × 2 cm were detected (Fig. 1a). The patient and her spouse were informed about these findings and the couple decided to continue with the pregnancy. The patient and her spouse were offered at the 18th-week genetic test for ventriculomegaly and cephalocele, but for religious reasons, the patient did not accept genetic testing. The patient and her spouse were informed also about the need for detailed fetal screening to look for other systemic anomalies. Detailed fetal anomaly screening with ultrasonography and MRI scans were planned for the 22nd gestational week. However, the patient did not come to the study center at 22 weeks of gestation. Since the patient and her spouse did not want pregnancy termination, they did not accept any detailed screening and diagnostic tests. However, sonographic examination failed to visualize this occipital mass at 35th gestational week and magnetic resonance imaging (MRI) was performed to indicate the presence of a 2 cm-wide subcutaneous formation over the defect in the occipital bone. Since this formation had no integrity with the cranial compartments and all intracranial structures were normal, a preliminary diagnosis of atretic occipital cephalocele was made (Fig. 1c, d).

Three weeks later, a female neonate with a birth weight of 3100 g, first minute Apgar score of 7 and fifth minute Apgar score of 9 was delivered vaginally. Physical examination was normal except a soft protuberance of 2 × 2 cm which was located on the occipital bone and covered by hairy skin. MRI at the 23rd postnatal day clearly demonstrated a solid mass of 2 × 2 cm over the osseous defect in the occipital bone and bilateral cochlear hypoplasia. Therefore, the newborn was diagnosed with an atretic occipital cephalocele (Fig. 1e, f). Four months later, computed tomography demonstrated the atretic cephalocele sac and the underlying osseous defect in the occipital bone (Fig. 2).

An ultrasonographic examination of the atretic cephalocele at 20 months of age pointed out that the defect in the underlying occipital bone persisted (Fig. 1b). Clinical follow-up until 24 months of age showed normal neurological development of the newborn except hearing impairment. Although surgical removal of the atretic cephalocele was recommended electively, the parents of the patient refused the procedure.

Discussion

The exact pathogenesis of atretic cephaloceles is unknown but several hypotheses have been put forward. These malformative lesions might develop as a true intrauterine regression of a cephalocele, a neural crest remnant, a re-

Fig. 1 **a** Obstetric ultrasonography showed an occipital solid mass of 2 × 2 cm. **b** An ultrasonographic examination of the atretic cephalocele at 20 months of age pointed out that the defect in the underlying occipital bone persisted. **c** Magnetic resonance imaging at 35th week of gestation indicates the presence of a 2 cm-wide subcutaneous formation over the defect in the occipital bone. **d** This formation has no integrity with the cranial compartments and all intracranial structures were normal. **e** Magnetic resonance imaging at postnatal 23rd day clearly demonstrates a solid mass of 2 × 2 cm over the defect in the occipital bone and bilateral cochlear hypoplasia. **f** The newborn has been diagnosed with an occipital atretic cephalocele ▶

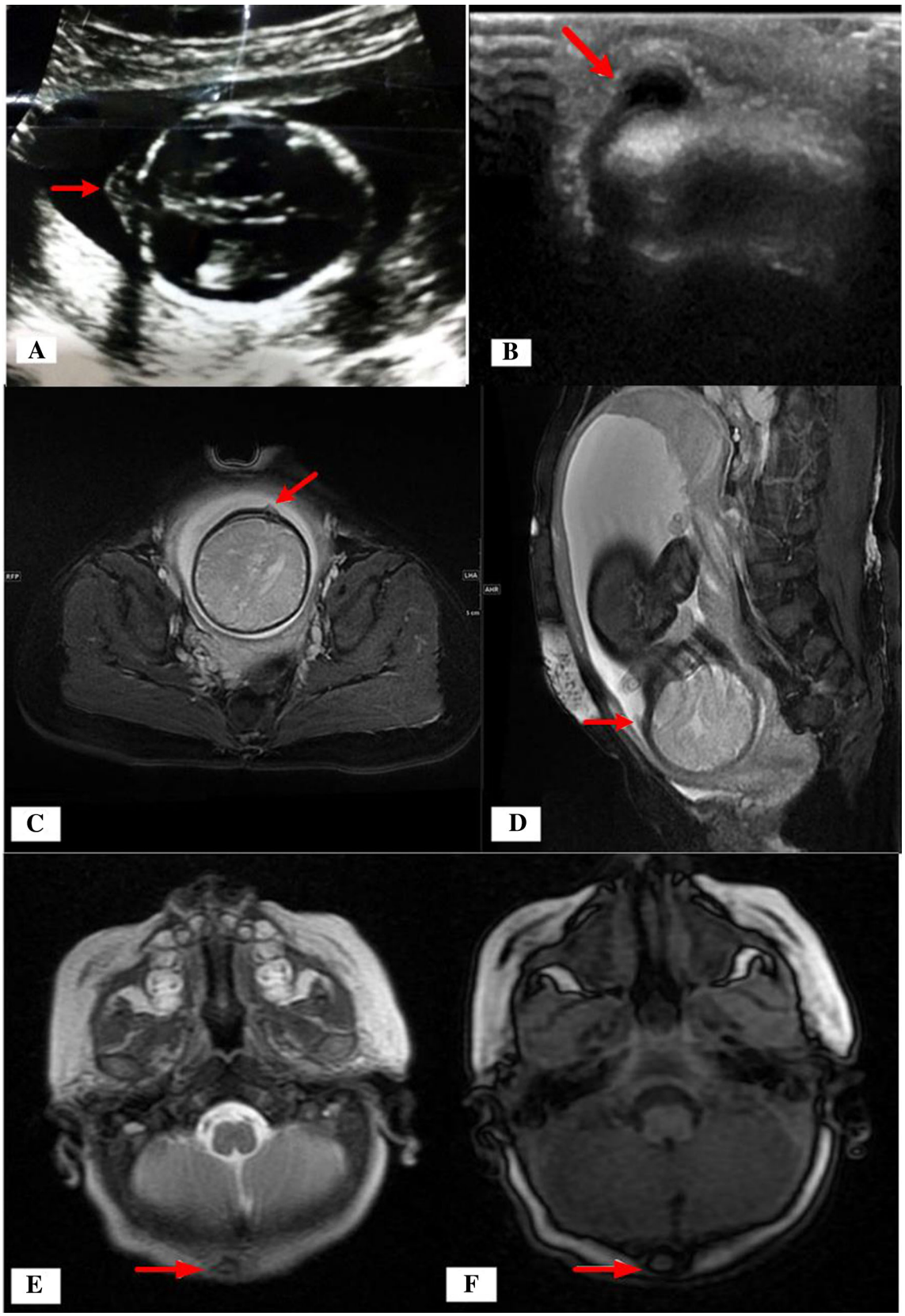
opened neural tube, a persistence of a nuchal bleb, or a sequel of primary mesenchymal injury [3, 4].

A body of evidence for the embryological hypothesis is that atretic cephaloceles consist of the remnant nuchal blebs that may be related with the transient overdistension of the rhombencephalic vesicle. Since skin and the neural tissues are derived from the ectoderm, associated skin abnormalities and concomitant cranial or spinal malformations point out a common etiology [4, 5]. Radiological follow-up in this case suggested that true intrauterine regression of an encephalocele might be the underlying cause. However, more data is needed to draw a conclusion about the etiopathogenesis.

The existence of familial cases and specification of parental consanguinity in affected newborns have indicated genetic factors as the underlying cause. When the existence of familial cases, amniocentesis should be performed in such cases to determine the underlying genetic factors. Moreover, environmental factors such as vitamin A hypervitaminosis, teratogens, X-ray, folic acid antagonists, trypan blue, triamcinolone, parental malnutrition and diabetes have been proposed as possible mechanisms for atretic cephaloceles [5, 6].

Atretic cephaloceles make up 25% to 56% of all types of cephaloceles. These subscalp lesions appear as hamartomatous structures which are generally covered with atrophic and alopecic skin. Rarely, dark hair aggregation is noted over the lesions. Nearly half of the atretic cephaloceles are usually situated within a few centimeters of the lambda over the parietal bone. The remaining half of the atretic cephaloceles has occipital, parieto-occipital, frontal, asterion, and sincipital locations. The size of these lesions varies between 1 cm and 3.5 cm and these masses may enlarge with Valsalva maneuver if there is a communication with ventricles or subarachnoid space [7–9].

Additional congenital anomalies may be accompanied in up to 50% of cephalocele cases. In such cases, detailed fetal screening with ultrasound is required because of the possibility of additional congenital abnormalities such as spina bifida. Variety of the neural tube defects can be related to multifactorial, sporadic or genetically. Also, it is believed that most neural tube defects occur as


A
B
C
D
E
F

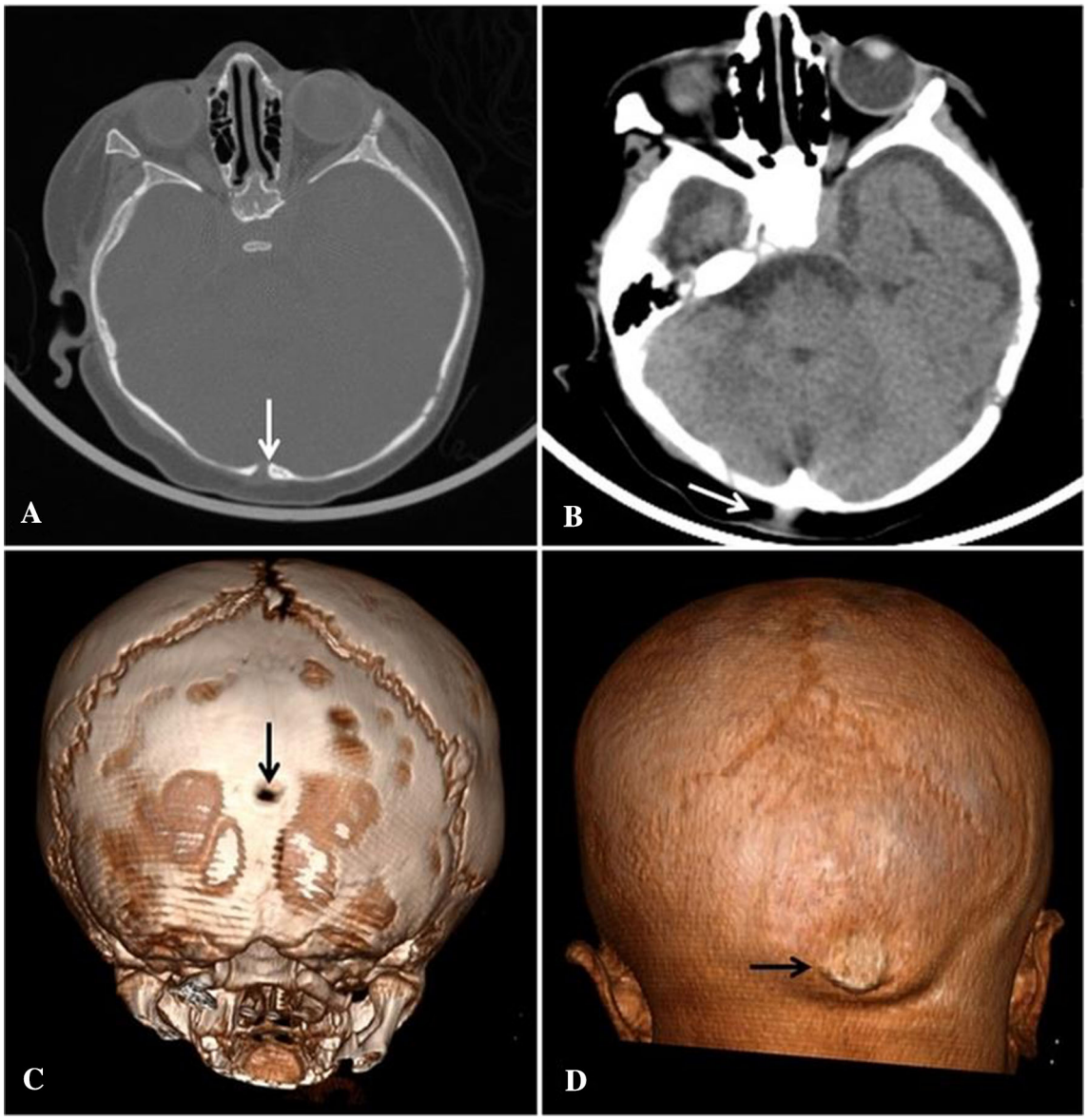


Fig. 2 Computed tomography at fourth month of postnatal life demonstrated the atretic cephalocele sac and the underlying defect in the occipital bone

multifactorial disorders, cephaloceles are more frequently related to specific syndromes including aneuploidy, sporadic deformation sequences and Mendelian disorders [10]. On the other hand, fetal ultrasonographic findings of significant ventriculomegaly are generally correlated with important neurological problems. Fetal ultrasound screening may demonstrate ventriculomegaly, microcephaly, oligohydramnios, intracranial calcifications and intrauterine growth restriction. If there are findings other than cephalocele such as ventriculomegaly detected in fetal screening by ultrasound which, as in this presented case, genetic diagnostic tests such as amniocentesis and cordocentesis can be performed to detect other congenital abnormalities [11, 12]. Definitive ultrasonographic diagnosis is critical in ensuring prognosis and providing suitable genetic counseling. Also, MRI and computed tomography can be used to evaluate the concomitant central nervous system anomalies in patients with atretic cephaloceles. These anomalies include holoprosencephaly, porencephaly, schizencephaly, dolichocephaly, macrocephaly, multiple intracranial cysts, agyria, hypointensity of white matter, corpus callosum hypo- or agenesis, absence of septum pellicidum, intracranial lipomas, pineal body enlargement, vermicular agenesis, dropping down of occipital lobes, split pons, hypoplasia of tentorium cerebella, peaking of tentorium, spinning-top configuration of tentorial incisura, hydrocephalus, gray matter heterotopias, ventricular diverticulum, parietal foramina, prominence of

brain cisterns, Chiari types II–III malformations, and anomalous arteries and draining veins [13, 14].

The patients with atretic cephaloceles may have distinct facial features such as epicanthal folds and upslanted palpebral fissures. These lesions can be associated with congenital syndromes, mental retardation, developmental delay, seizures, spasticity, speech difficulty, hypertelorism, strabismus, optic nerve atrophy, microphthalmia, enophthalmos, retroocular cyst, cleft palate, tetralogy of Fallot, renal agenesis, hearing problems, congenital lobar emphysema, and muscular anomalies [14, 15]. The patient in this case report has been also diagnosed with cochlear hypoplasia and, thus, hearing loss.

Differential diagnosis of atretic cephaloceles comprise cutis aplasia congenita, dermoid cysts, epidermoid cysts, traumatic cysts, sebaceous cysts, lipomas, tumors, vascular malformations, cephalhematomas, infections and sinus pericranii. On the other hand, atretic cephaloceles with concurrent dermoid cyst or sinus pericranii have been rarely reported [1, 3].

The clinical outcomes of the patients with atretic cephaloceles depend on their initial neurological status and the presence of the co-existing central nervous system anomalies and systemic morbidities. As expected, the patients with isolated atretic cephaloceles have more favorable prognosis. It has been speculated that the patients with occipital atretic cephaloceles may have better neurological outcomes than those with parietal lesions [1–3].

Expectant management and surgery can be adopted for the management of patients with atretic cephaloceles. Asymptomatic cases can be observed whereas surgical treatment is indicated for cosmetic reasons, histopathological confirmation and cases complicated with erosion, infection, and pain. Whenever surgery is planned, a complete neuroradiological workup should be carefully performed to reveal the content of the lesion and its possible communication with interior structures. Unless vascular anomalies have been identified preoperatively, surgery has low morbidity [7–9]. As the patient described here was asymptomatic, her parents refused a surgical intervention. This case report should remind the obstetricians that prenatal ultrasonography and fetal MRI can be conveniently and efficiently used to examine fetuses with atretic cephaloceles.

Compliance with Ethical Standards

Conflict of interest The authors declare that they have no conflict of interest.

Informed Consent Informed consent was obtained from all individual participants included in the study.

References

1. Şengöz A, Kaya M, Yildirim CH, Tasdemiroglu E. Atretic cephalocele in adults. Acta Neurochir (Wien). 2011;153(11):2275–7.
2. Bick DS, Brockland JJ, Scott AR. A scalp lesion with intracranial extension. Atretic cephalocele. JAMA Otolaryngol Head Neck Surg. 2015;141(3):289–90.
3. Demir MK, Çolak A, Ekşi MŞ, Özcan-Ekşi EE, Akakın A, Yılmaz B. Atretic cephaloceles: a comprehensive analysis of historical cohort. Childs Nerv Syst. 2016;32(12):2327–37.
4. Hsu SW, Chaloupka JC. Atretic parietal cephalocele associated with sinus pericranii: embryological consideration. Brain Dev. 2012;34(4):325–8.
5. Fukuyama M, Tanese K, Yasuda F, Hayashi Y, Tanikawa A. Two cases of atretic cephalocele, and histological evaluation of skin appendages in the surrounding skin. Clin Exp Dermatol. 2016;41(1):48–52.
6. Carvalho DR, Giuliani LR, Simão GN, Santos AC, Pina-Neto JM. Autosomal dominant atretic cephalocele with phenotype variability: report of a Brazilian family with six affected in four generations. Am J Med Genet A. 2006;140(13):1458–62.
7. Wong SL, Law HL, Tan S. Atretic cephalocele—an uncommon cause of cystic scalp mass. Malays J Med Sci. 2010;17(3):61–3.
8. Santos SF, Ramos H, Irañeta A, Conceição C. Atretic parietal encephalocoele. BMJ Case Rep. 2016. https://doi.org/10.1136/bcr-2016-215812.
9. Güzel A, Tatli M, Er U, Bavbek M. Atretic parietal cephalocele. Pediatr Neurosurg. 2007;43(1):72–3.
10. Steven JW, Donnenfeld AE. Syndromes identified in fetuses with prenatally diagnosed cephaloceles. Prenat Diagn. 1994;14:839–43.
11. Fox NS, Monteagudo A, Kuller JA, Craigo S, Norton ME. Mild fetal ventriculomegaly: diagnosis, evaluation, and management. Am J Obstet Gynecol. 2018;219(1):B2–9.
12. Wyldes M, Watkinson M. Isolated mild fetal ventriculomegaly. Arch Dis Child Fetal Neonatal Ed. 2004;89:F9–13.
13. Leykamm S, Wessling B, Mühlenbruch G. Atretic cephalocele and associated anomalies in a newborn child. Clin Neuroradiol. 2013;23(1):37–40.
14. Siverino RO, Guarrera V, Attinà G, Chiaramonte R, Milone P, Chiaramonte I. Parietal atretic cephalocele: associated cerebral anomalies identified by CT and MR imaging. Neuroradiol J. 2015;28(2):217–21.
15. Morioka T, Hashiguchi K, Samura K, Yoshida F, Miyagi Y, Yoshiura T, Suzuki SO, Sasaki T. Detailed anatomy of intracranial venous anomalies associated with atretic parietal cephaloceles revealed by high-resolution 3D-CISS and high-field T2-weighted reversed MR images. Childs Nerv Syst. 2009;25(3):309–15.