

Neonate with meroacrania: radiological findings and review of the literature

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ABSTRACT

Acrania is a developmental abnormality characterized by a partial or complete absence of calvaria with complete but abnormal development of brain tissue. Acrania is a relatively common malformation and affects about 1 in 1000 newborns. Meroacrania refers to absence of the cranium with the exception of the occipital bone. Brain stem and cerebellum develop normally, but cerebral parenchyma tissue is covered with a thin membrane and severely dysmorphic supratentorial brain is also seen. The other system findings are normal. Magnetic resonance imaging findings of one neonate with meroacrania have been reported in medical literature. Other radiographic and computed tomography findings have not yet been reported. We report a female neonate with meroacrania with discussion of etiology, pathogenesis, radiological findings, and differential diagnosis.

Key words: • meroacrania • x-ray • computed tomography • magnetic resonance imaging

Acrania is a developmental abnormality characterized by a partial or complete absence of calvaria with complete but abnormal development of brain tissue. Acrania is an example of neural tube defects (NTDs). NTDs are relatively common malformations and affect about 1 in 1000 newborns (1). Meroacrania refers to a condition in which the basis cranii bones (occipital bone, foramen magnum, clivus) are normal, but the other cranial bones are not developed. We present a case of meroacrania in which the brainstem and cerebellum developed normally, but cerebral parenchyma was covered with a thin membrane, and brain tissue was severely dysmorphic.

Prenatal diagnosis and intrauterine monitoring of meroacrania are possible with ultrasound. The differential diagnosis includes anencephaly and large cephaloceles. There is little detail on the radiological findings in this disorder. There is one reported case with magnetic resonance imaging (MRI); other radiographic and computed tomography (CT) findings have not previously been reported.

We report a female neonate with meroacrania and discuss etiology, pathogenesis, radiological findings, and differential diagnosis.

Case report

This infant was the first alive infant born to a 24-year-old mother by spontaneous vaginal delivery in the hospital. Following delivery, the baby cried, and there was no hypoxia. The mother was of low socioeconomic status and had not been able to afford prenatal care or prenatal ultrasound. There was no history of any illnesses or drug usage during the pregnancy. Because she came to our hospital urgently, there was no known intrauterine pre-diagnosis.

After birth, the newborn was pre-diagnosed with acrania-anencephaly. Birthweight was 3200 g and length was 50 cm. There was no bony structure in the temporal, frontal, or parietal regions. Cerebral parenchymal tissue was covered with a thin membrane (Fig. 1). There was also an atypical facial appearance, cleft lip, and pes equinovarus deformity. No other abnormalities were noted.

On anteroposterior skull radiograph, there was no evidence of cranial bones except occipital bone (Fig. 2). Cranial axial CT showed that the basis cranii bones (occipital bone, foramen magnum, clivus) were normal, but other cranial bones were not developed. It was also noted that the brainstem and cerebellum were developed; and supratentorial views showed cystic dilatation and thinning of the cerebral parenchyma (Fig. 3). Coronal and sagittal MRI images showed normal cerebellum and cranio-cervical junction. There were dilatations in the supratentorial ventricular system, and the brain was severely dysmorphic (Fig. 4).

The infant died three days after delivery.

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Received 11 May 2007; revision requested 27 June 2007; revision received 24 July 2007; accepted 29 July 2007.

Published online 10 November 2009
DOI 10.4261/1305-3825.DIR.1075-07.1



Figure 1. Postpartum macroscopic picture of the case: cranial bone structures were not developed, and the brain tissue was covered with dura mater.



Figure 2. The anteroposterior cranial radiograph shows presence of the occipital bone and craniocervical junction bones but no development of other cranial bone structures.

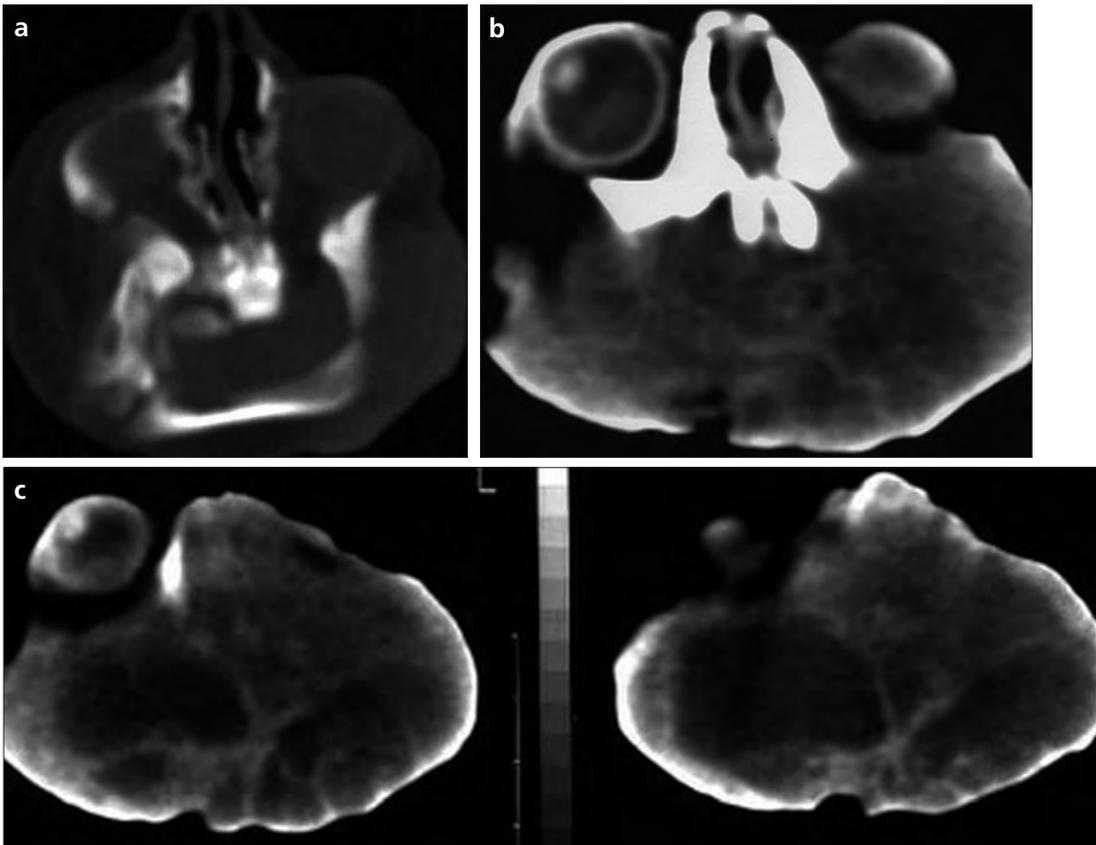


Figure 3. a–c. Axial cranial CT image (a) shows normal development of the occipital bones. In the upper axial CT sections (b), it was found that temporal bones were not developed. In the cross-sections at the vertex levels (c), it was found that frontal and parietal bones were not developed, and brain parenchyma showed cystic dilatations.

Discussion

Meroacrania refers to absence of the cranium with the exception of the occipital bone. Brainstem and cerebellum are developed normally, but cerebral

parenchymal tissue is covered with a thin membrane, and brain tissue is severely dysmorphic. There is no sex preponderance in meroacrania (2). Like other forms of NTD, there is a multi-

factorial pattern of transmission with interaction of multiple genes and environmental factors, although neither the genes nor the environmental factors are well characterized. It may be

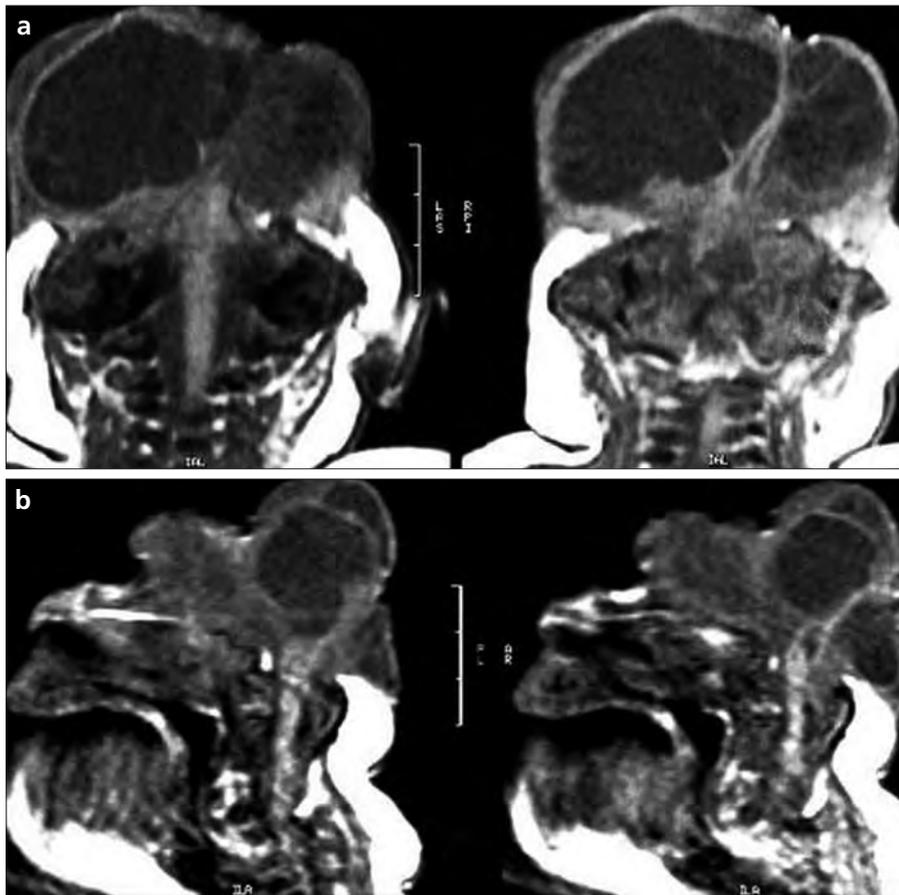


Figure 4. a, b. Coronal (a) and sagittal (b) postcontrast T1-weighted MR images show normal cerebellum and cranial-cervical junction. Posterior fossa bones were not developed, and the structure of the posterior fossa was deformed. There were supratentorial dilatations in the ventricular system.

part of a more complex process involving single-gene defects or disruption of the amniotic membrane (3).

Meroacrania is lethal because of the severe brain malformation that is present. This is a rare congenital anomaly resulting from failure of the mesenchyme to migrate under the ectoderm overlying the brain tissue to form the bony tissue over the cerebral hemispheres.

The pathogenesis is not clearly understood. Normally during embryological development, after the closure of the anterior neural pore (around the fourth week), migration of the mesenchymal tissue under the ectoderm underlying the future cerebral hemispheres takes place. The ectoderm forms the skin and scalp, while the mesenchyme gives rise to the muscle and bone. The most commonly accepted theory suggests that acalvaria is a post-neurulation defect, that is, it results from faulty migration of the mes-

enchyme with normal placement of the embryonic ectoderm. There is thus an absence of the calvaria but an intact layer of skin over the brain parenchyma (4). The brain tissue is covered by only a thin membrane and is therefore exposed to the amniotic fluid. The cerebral hemispheres, although present, are anatomically disorganized. The cerebellum and the base of the skull, however, are well developed (1, 3).

Other theories suggest that it results because of the primary non-closure of the neural tube or may be a part of a spectrum of anencephaly. The disorder is etiologically and pathogenetically heterogeneous, and its prevention by ingestion of folic acid has not been described.

Associated anomalies are frequent and include NTDs, cleft lip, cardiac defects, clubfoot, bilateral absence of orbital floors, and omphalocele.

Prenatal diagnosis and intrauterine monitoring of this anomaly are pos-

sible with ultrasound (5, 6). The biochemical and cytological examination of amniotic fluid samples may suggest the presence of a NTD. Cytological examination of amniotic fluid obtained by amniocentesis has confirmed the presence of neural cells in anencephalic fetuses, in which acrania was diagnosed originally in the first trimester (7). The crown-rump length is smaller in fetuses with anencephaly than in healthy control fetuses (8). It has been confirmed that the specific body segment that is decreased in size is the mentovertebral diameter (crown-chin length), attributable to progressive destruction of the brain as pregnancy progresses (9). Increased echogenicity of amniotic fluid in the first trimester should be considered a warning sign for the acrania-anencephaly sequence. This further supports the hypothesis of progressive destruction of the brain as a causative factor of this condition, in which anencephaly is the end of a spectrum that appears in the first trimester as fetal acrania. Sonographers and clinicians should be aware of this indirect sign in fetuses with acrania because of increasing interest in incorporating nuchal translucency screening at 11 to 14 weeks' gestation into clinical practice (10).

The differential diagnosis of acrania-meroacrania includes anencephaly and large cephaloceles. In anencephaly, cerebral tissue is completely absent; while in cephaloceles, the cranial vault can always be detected, and a part of the brain is intracranial. A distinction should also be made between acrania and conditions characterized by lack of mineralization of the skull bones such as hypophosphatasia and osteogenesis imperfecta type II. In these skeletal dysplasias, the intracranial anatomy is normal, and a thick layer of tissue representing soft tissues and unossified bone surrounds the brain. Bowing, fractures, and shortening of long bones are usually present (6).

Meroacrania is a lethal malformation. Following a sonographic diagnosis of meroacrania, termination of the pregnancy should be offered to the couple. Pathologic examination of the fetus should be performed in all cases to establish the final diagnosis. This is extremely important for genetic counseling. After one infant with anencephaly, risk is 2% to 5% in subsequent pregnancies (6, 11). There-

fore, the diagnosis of acrania warrants assessment of the parents and proband in order to provide accurate genetic counseling. Because acrania may be a field defect or a mutation in the paired-class homeobox-containing gene *Cart1*, folic acid supplementation during pregnancy or even before conception may suppress the phenotypic expression of acrania, as has been suggested in mice (12).

There is little radiologic information about meroacrania in the literature. Tubbs and Blount reported MRI and surgical findings of a female neonate (13). They reported that sagittal T1 weighted MRI of the patient that showed a vertical clivus and severely dysmorphic supratentorial brain and an anteriorly placed non-skin-covered herniation of brain tissue.

In conclusion, merocrania is a rare lethal congenital anomaly. This is the second published report of radiological findings in a neonate with meroacra-

nia. Fetal ultrasound is needed for in-utero diagnosis; if cranial or spinal abnormalities are found, fetal MRI can provide more detailed information about fetal anomalies.

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