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Promporn Raksaseri

Wuthichai Klomkleaw

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Short Communication

Frontoethmoidal meningoencephalocele with multiple congenital craniofacial and skeletal malformations in a stillborn puppy

Promporn Raksaseri[1] Wuthichai Klomkleaw[1*]

Abstract

A one-and-a-half-year-old Shih-Tzu bitch went into natural labor after full term of her first pregnancy, giving birth to 4 puppies. One of them was a male which died at birth, 104 grams in weight and 12 centimeters in length. This puppy showed several facial abnormalities and skeletal deformities. It was preserved in 10% formalin for anatomical, histopathological, and radiographic studies. Gross anatomical study revealed congenital anomalies of the eyes including anophthalmia of the left eye and microphthalmia of the right eye. Malformations of the ears including anotia of the left ear and microtia of the right ear were observed. Evidence of cleft lips was also noted. There was a membrane-bound mass emerging from the center of the face and replacing the nose. Histopathological sections of the mass revealed evidence of nervous tissues composed of neurons and their supporting cells including microglia and astrocytes. Radiographs showed incomplete formation of frontal bones where the herniation of the mass occurred. These findings were consistent with a diagnosis of frontoethmoidal meningoencephalocele. Additionally, deformities of the 1st to the 3rd cervical vertebrae, the 3rd and lower thoracic vertebrae, and all lumbar vertebrae were found. Rib deformity was observed at both sides and rib fusion was presented on the right side. In conclusion, this study shows a rare case of a stillbirth puppy that possessed frontoethmoidal meningoencephalocele associated with multiple facial and skeletal abnormalities.

Keywords: frontoethmoidal encephalomeningocele, facial anomalies, skeletal abnormalities, inherited, dog

[1]*Department of Anatomy, Faculty of Veterinary Science, Chulalongkorn University, Bangkok 10330, Thailand*
[*]***Correspondence:*** *Wuthichai.K@chula.ac.th*

Introduction

Congenital malformations or congenital anomalies are defect conditions at birth which can be found in many parts of the body. These conditions can be caused by toxic substances, drug toxicity, injury during pregnancy, teratogens, malnutrition of the bitch and genetic defects. In case of canine facial malformations, there are several congenital defects including cyclopion, cleft lip and cleft palate, and frontoethmoidal meningoencephalocele. One of the most common congenital defects of dog face is cleft lip, which might be associated with cleft palate resulting from malformation of the jaw and the face during embryogenesis. Frontoethmoidal meningoencephalocele is a protrusion of intracranial tissues and meninges through the frontal bone (David, 1993; Suwanwela and Suwanwela, 1972). It can be congenital or acquired disorders caused by tumors, hydrocephalus, or other causes (Holmes et al., 2001). The brain tissue can herniate out of the skull through existence of a weak point between the frontal and ethmoidal bones during embryonic development (Dhirawani et al., 2014). In human, frontoethmoidal meningoencephalocele is commonly found with other congenital defects of facial malformation such as cleft lip and cleft palate (Ganapathy et al., 2014). However, it is rarely diagnosed in domestic animals. Other congenital defects of the ears and eyes such as microtia, anotia (Beraud, 2012; Luquetti et al., 2012), microphthalmos, and anophthalmos (Dell, 2010; Shibuya et al., 2000) are also rarely found in dogs, whereas congenital malformations of vertebrae and ribs are frequently found, especially in brachycephalic dogs such as pug and French bulldog (Gutierrez-Quintana et al., 2014). This case will describe the characteristics of frontoethmoidal meningoencephalocele with multiple congenital anomalies of facial and vertebral malformation in a stillborn puppy.

Materials and Methods

Specimen and anatomical study: After full term of first pregnancy, a one-and-a-half-year-old Shih-Tzu bitch previously mated with the same breed went into natural labor at N.P. Animal Hospital, Bangkok, Thailand. One of four puppies had facial abnormalities and died at birth. It was preserved in 10% buffered formalin for further morphological studies. Body length and weight were measured and pictures were taken.

Histopathological study: Briefly, a membrane-bound facial mass located at the center of the puppy's face was removed, cut through a mid-sagittal plane, following the standard protocol for histopathological tissue sectioning, and stained with H&E.

Radiographic study: Total body radiographs were taken in lateral and dorsoventral positions using Universal® X-ray machine model MoD3486-50 at 75 kV, 1/50 second.

Results and Discussion

The puppy was 12 centimeters long and weighed 104 grams. Gross anatomical study revealed anophthalmia of the left eye and micropthalmia of the right eye (Fig. 1a-c). Anophthalmia is characterized by the absence of the globes and ocular tissue within the orbits (Shibuya et al., 2000). The association of anophthalmos with other congenital defects including facial asymmetry and ear deformities is commonly found (Dell, 2010; Samson and Viljoen, 1995). During the first 3 weeks of gestation, optic vessels grow and stimulate neuro-ectodermal tissue to proliferate and form lens placode. Anophthalmos is caused by the stop of optic vessel growth, resulting in no formation of ocular structures (Dell, 2010). Micropthalmos is an eye with size reduction because the growth of optic cup is inhibited. Several factors might cause micropthalmia such as genetic disorder, uterine infection, drug toxicities, and maternal vitamin A deficiency (Kennelly et al., 2011; Tsutsui et al., 1993; Verma and Fitzpatrick, 2007; Williamson and FitzPatrick, 2014). Recently, bilateral micropthalmos has been reported in a Pomeranian puppy (Dell, 2010). Our Shih-tzu puppy had both microtia of the right ear, characterized by underdevelopment of pinna, and anotia of the left ear, characterized by completely undeveloped pinna, which might be associated with abnormalities of the external auditory canal and middle ear (Luquetti et al., 2012) (Fig. 1a-c). Microtia in human has been reported many times and this anomaly was demonstrated for the first time in a 2-month-old male mixed breed dog (Rezaei et al., 2015). In addition, anotia has been reported to be associated with distinct Mendelian autosomal recessive syndrome in St. Bernard (Villagomez and Alonso, 1998). This evidence supports that, at least, anotia is related to genetic disorder.

Interestingly, a mass with the size of 1.4 x 2 cm covered by a clear membrane was found at the center of the face, causing disappearance of the nose and appearance of large cleft lip (Fig. 1a-c). Radiographs revealed incomplete formation of maxillary and frontal bones where the mass protrusion occurred (Fig. 1d). The incomplete frontal bone formation is shown in Fig. 2a after the skin of the puppy's head was dissected. Nasal cavity, nasal septum, ethmoid bone and hard palate were not observed after removal of the entire brain with the mass as shown in Fig. 2b. Meningoencephalocele in our case involved intranasal cavity, similar to most cases of live dogs in a recent report (Lazzerini et al., 2017). H&E staining of mid-sagittal tissue section revealed that the mass was composed of numerous neurons, microglia, and capillaries surrounded by astrocytes. The membrane which encapsulated the mass consisted of fibroblasts and connective tissue (Fig. 3a-b). Encephalocele is characterized by a protrusion of cranial content throughout the cranium according to congenital defects of the skull. In case, herniation containing both brain matter and meninges is called meningoencephalocele. It has been identified into several classifications depending on the location of the skull defect. These include occipital, frontoethmoidal, basal, and cranial vault types, which were proposed by Matson (Fountas et al., 2005). In humans, patients with

frontoethmoidal meningoencephalocele had swelling over the bridge of nose or inner canthus of the eye since birth (Suwanwela and Suwanwela, 1972). Using these criteria, the stillbirth puppy in our study had frontoethmoidal meningoencephalocele, which is rarely found in dogs as opposed to Southeast Asian people, in which it is commonly found with an incidence of 1:5000 of new born babies (David, 1993; Dhirawani et al., 2014). According to history taking, the cause of the abnormality seems not to be from using drugs or being exposed to chemical agents of the bitch. Recently, ethmoidal encephalocele in a 6-month-old cross-bred dog and intranasal meningoencephalocele in a 5-month-old female border collie have been reported. Both dogs showed sign of seizure from cerebral dysfunction (Jeffery, 2005; Martle et al., 2009). The etiology of frontoethmoidal meninegoencephalocele is still unknown. However, failure of the neural plate to transform to the neural tube caused by incomplete separation between the surface ectoderm (epithelium) and neuroectoderm during early gestation period is a possible cause of the defect (Hoving, 2000). Besides genetics, environmental factors such as fungal infection and teratogenic agents can be the causes of encephalocele (Dhirawani et al., 2014).

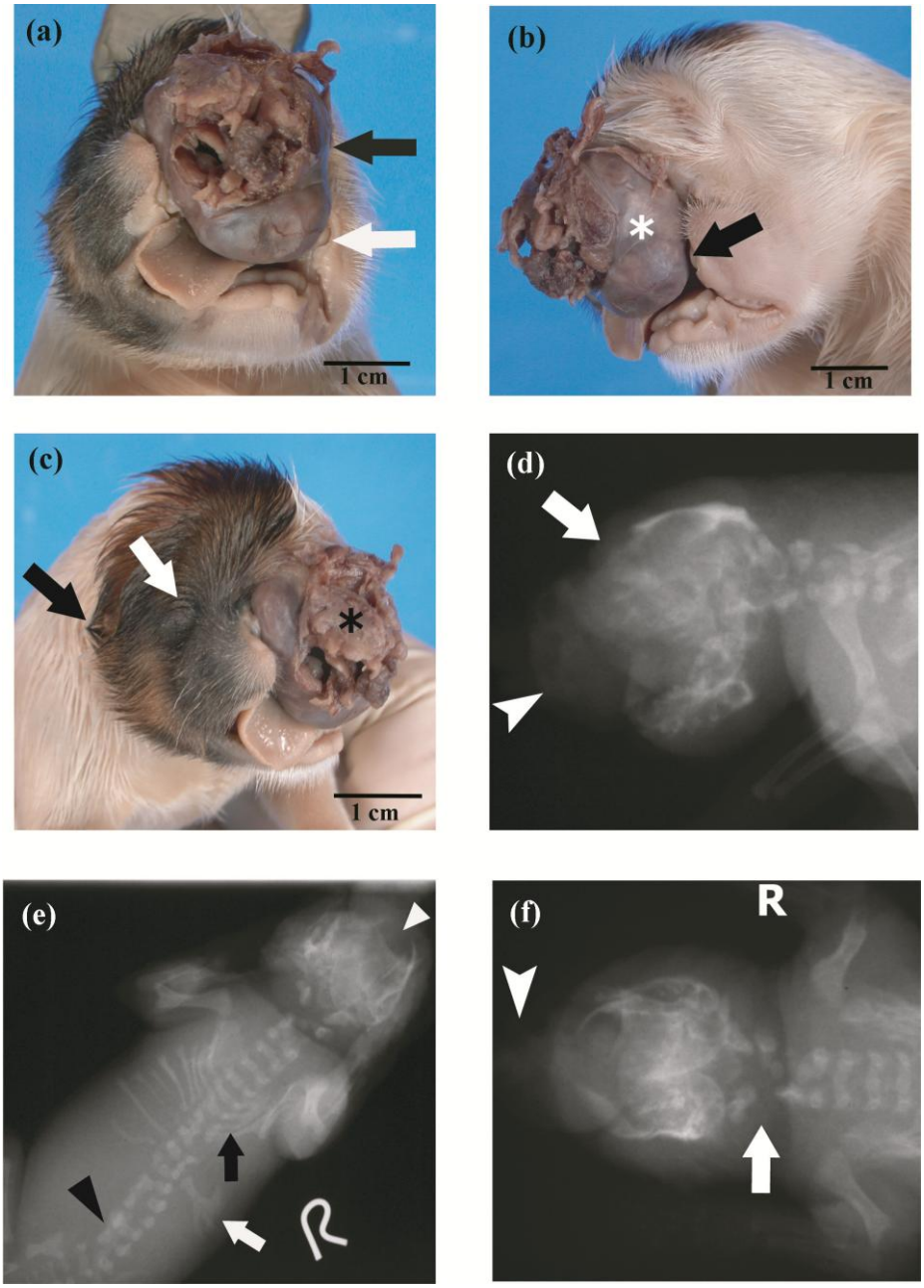


Figure 1 (a) The puppy's face (front view) showing frontoethmoidal meningoencephalocele (black arrow) and cleft lip (white arrow). (b) The puppy's face (left side) showing frontoethmoidal meningoencephalocele (*) and cleft lip (black arrow) under the mass. (c) The puppy's face (right side) showing micropthalmos (white arrow), microtia (black arrow), and frontoethmoidal meniengoencephalocele (*). (d) Radiograph (lateral position) illustrating frontoethmoidal meningoencephalocele (white arrowhead) and incomplete formation of the frontal bone (white arrow). (e) Radiograph (dorsoventral position) showing incomplete formation of the frontal bone (white arrowhead), butterfly lumbar vertebrae (black arrowhead), rib fusion (black and white arrows). R = right side. (f) Radiograph (dorsoventral position) revealing lateral aplasia of the 2nd and 3rd cervical vertebrae (white arrow). White arrowhead pointing at frontoethmoidal meninegoencephalocele. R = right side.

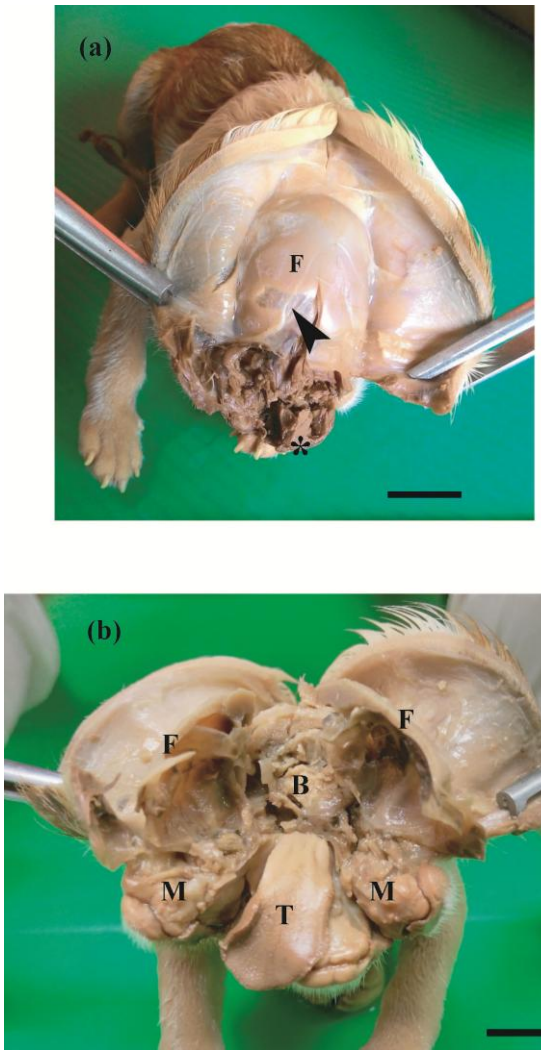
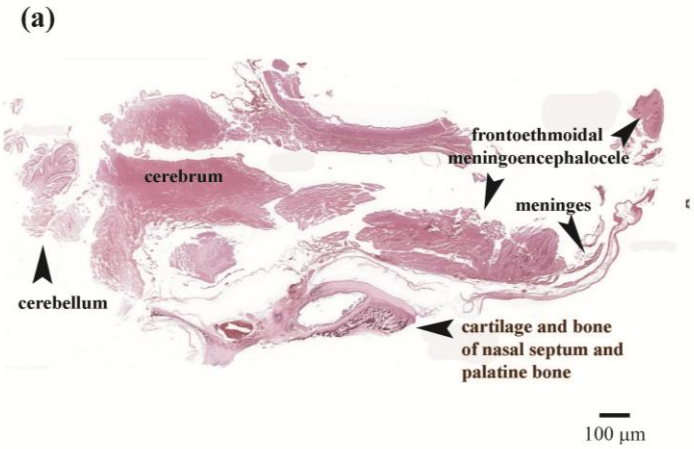


Figure 2 (a) The puppy's head with opening skin showing frontal bone (F) with incomplete formation area (arrowhead). FEEM was still intact (Star). Scale bar = 1 cm. (b) By cutting through the frontal bone, the entire brain and FEEM were removed. Associated structures including nasal cavity, nasal septum, ethmoid bone and hard palate were not observed. T = tongue, F = frontal bone, M = maxilla bone covered with gum and teeth, B = part of brain. Scale bar = 1 cm.



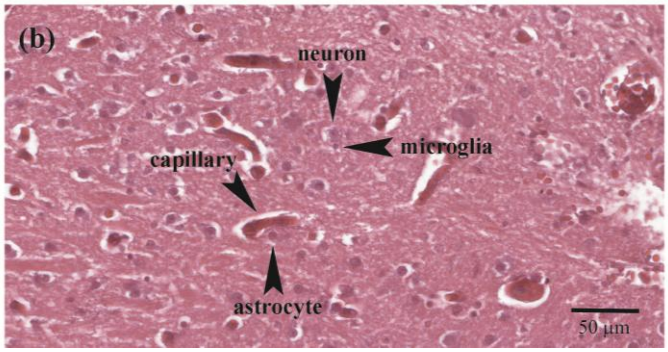


Figure 3 (a) Histopathological sections of the membrane-bound mass protruding through the face (frontoethmoidal meningoencephalocele) showing several parts of the brain, meninges, nasal, and palatine structures. (b) Higher magnification of (a) showing numerous neurons, microglia, astrocytes, and capillaries. Magnification (a) 20x; (b) 400x.

Additionally, lateral aplasia of the 2nd and 3rd cervical vertebrae and ventral and median aplasia of the 3rd to 13th thoracic or butterfly vertebrae were observed. The shape of both left and right ribs was abnormal with fusion on some parts of the right side. Incomplete development of the 7th lumbar vertebra was clearly seen (Fig. 1e-f). Possible mechanisms responsible for the malformation of vertebrae include absence of vertebral vascularization, teratogenic effects, and genetic abnormalities (Westworth and Sturges, 2010).

Taken together, the stillbirth puppy in this report had multiple congenital anomalies including frontoethmoidal meningoencephalocele, anotia, microtia, anophthalmia, microphthalmia, cleft lips, and vertebral malformations. It can be conclude that, at least, genetic disorders may be involved. The developmental defects of this puppy probably occurred during the first 3 weeks of gestation and they were caused by the defective process of neuroectodermal development.

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บทคัดย่อ

โรคงวงช้างและความผิดปกติแต่กำเนิดของใบหน้าและกระดูกโครงร่างในลูกสุนัขตายแรกคลอด

พรหมพร รักษาเสรี[1] วุฒิชัย กลมเกลียว[1*]

แม่สุนัขพันธุ์ชิห์สุอายุ 1 ปีครึ่ง ท้องแรก คลอดลูก 4 ตัวโดยวิธีธรรมชาติเมื่อครบกำหนด มีลูกเพศผู้ 1 ตัว น้ำหนัก 104 กรัม ยาว 12 เซนติเมตร ตายแรกคลอด พร้อมกับความผิดปกติหลายอย่างของใบหน้าและกระดูกโครงร่าง จึงเก็บรักษาในฟอร์มาลิน 10% เพื่อทำการศึกษาทางมหกายวิภาคศาสตร์ จุลพยาธิวิทยา และรังสีวิทยาต่อไป การศึกษาทางมหกายวิภาคพบความผิดปกติแต่กำเนิดของตา โดยไม่มีตาซ้าย ส่วนตาขวามีขนาดเล็กกว่าปกติ พบความผิดปกติของหู โดยไม่มีหูซ้าย ส่วนหูขวามีขนาดเล็กกว่าปกติ พบปากแหว่ง ที่เด่นชัดคือพบก้อนเนื้อสีน้ำตาลมีเยื่อบางใสหุ้มโผล่ออกมากลางใบหน้าแทนที่จมูก ซึ่งเมื่อนำไปศึกษาทางจุลพยาธิวิทยาพบว่าประกอบด้วยเนื้อเยื่อของระบบประสาทส่วนกลาง ได้แก่ เซลล์ประสาทและเซลล์ค้ำจุนชนิดต่าง ๆ รวมถึง microglia และ astrocytes การถ่ายภาพรังสีทั้งตัวพบการเจริญที่ไม่สมบูรณ์ของกระดูก frontal bones ในตำแหน่งที่พบก้อนเนื้อโผล่มากลางใบหน้า ทั้งหมดนี้ยืนยันลักษณะของโรคงวงช้าง (frontoethmoidal meningoencephalocele) นอกจากนี้ ยังพบกระดูกคอท่อนที่ 1-3 กระดูกอกท่อนที่ 3 ลงไป รวมทั้งกระดูกเอวทั้งหมดมีรูปร่างผิดปกติ กระดูกซี่โครงมีรูปร่างผิดปกติทั้งด้านซ้ายและขวา ในขณะที่มีการเชื่อมกันของกระดูกซี่โครงด้านขวาหลายซี่ การศึกษาครั้งนี้เป็นการแสดงภาวะงวงช้างในลูกสุนัขตายแรกคลอดในประเทศไทยที่เกิดร่วมกับความผิดปกติหลายอย่างรวมทั้งใบหน้าและกระดูกโครงร่าง ซึ่งถือเป็นภาวะที่พบได้น้อยมาก

คำสำคัญ: โรคงวงช้าง ใบหน้าผิดปกติ กระดูกโครงร่างผิดปกติ เป็นแต่กำเนิด สุนัข

[1]ภาควิชากายวิภาคศาสตร์ คณะสัตวแพทยศาสตร์ จุฬาลงกรณ์มหาวิทยาลัย ปทุมวัน กรุงเทพฯ 10330

****ผู้รับผิดชอบบทความ E-mail:** Wuthichai.K@chula.ac.th*