Anatomical study of a case of holoprosencephaly in a cat

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INTRODUCTION

Holoprosencephaly (HPE) is a complex brain malformation resulting from incomplete cleavage of the prosencephalon. HPE leads to face and neurological defects of variable intensity, ranging from a relatively normal facial appearance to cyclopia, the most severe form of HPE. Although it has a low prevalence, cases have been seen in both humans and different animal species. The etiology is diverse and unknown with accuracy, but chromosomal and genetic defects and teratogenic factors have been described. The prognosis may vary depending on the severity of the malformations in each case.

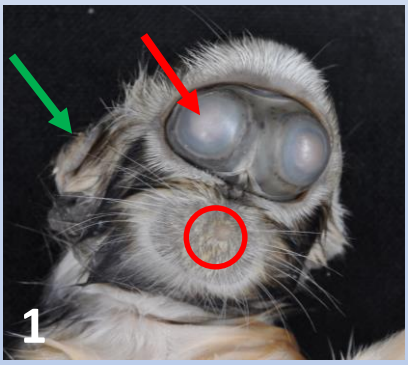
OBJECTIVES

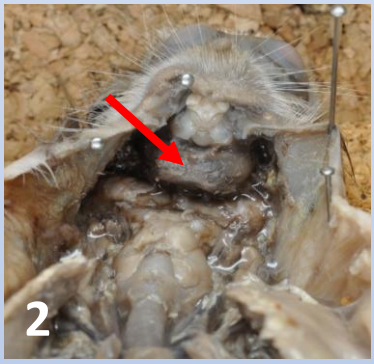
- Diagnose the case with precision based on the facial malformation observed in a cat and bibliography research.
- Observe possible internal pathologies associated with facial malformation.
- Study ontogeny development of the malformation.

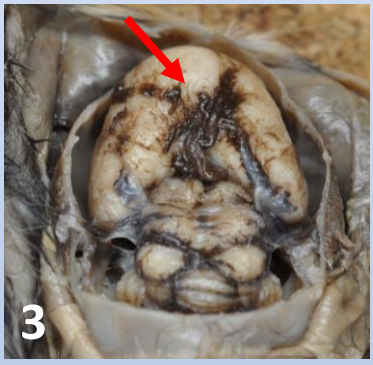
MATERIAL AND METHODS

Female cat presenting congenital craniofacial malformations (Figure 1). External and internal examination, dissection and 3D reconstruction were performed at the Veterinary Hospital and the Veterinary Anatomy Unit of the UAB.

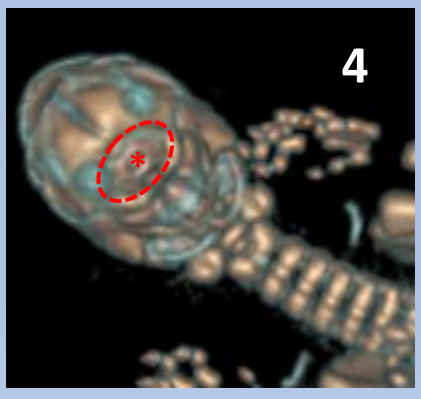
RESULTS AND DISCUSSION







External examination reveals the presence of a **single central orbit** with two incomplete and exophthalmic **fused eyeballs** (Fig. 1, red arrow). Synophtalmia is common in alobar HPE. A **small external nasal structure** (Fig.1, circle) can be seen below the orbit, **low-set ears** (Fig. 1, green arrow) and the mouth with a reduced size. The **jaw** (Fig. 2) and the maxilla were smaller than usual and microglossia and the absence of the nasal cavity were noted. These features are of otocephaly. The dissection showed the internal organs, which were normal. There were a **fusion of the cerebral hemispheres** (Fig. 3) and an absence of olfactory bulbs, the ethmoid bone and **cerebral convolutions** (Fig. 3), typical features of an alobar HPE.



In 3D reconstruction, a **single central orbit** (Fig. 4, circle) with a **single central optic canal** can be seen (*). Also noteworthy is the **fused frontal bone** without interfrontal suture (Fig. 5).

CONCLUSIONS

- The definitive diagnosis of the case is an alobar holoprosencephaly with synophtalmia and otocephaly.
- The animal has no internal pathology associated with alobar HPE.