



CONCURRENT ARHINIA AND CHOANAL ATRESIA IN A DAY OLD MALE KID

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Summary

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Arhinia is a congenital nasal developmental anomaly that is seldom reported in literature, and especially not reported in domestic animals. This report describes a case of concurrent occurrence of congenital arhinia and choanal atresia in a day-old male kid which had no external nares with the nasal bones fused together with the nasal processes of the premaxillae presenting as a conical shaped rhinal structure with a tapering rostral apex and an occluded nasal vestibule. A bilateral osseous choanal atresia was also seen at the pharynx. Additionally, craniofacial and brain anomalies presented in this condition with right lateral deviation of the face and the absence of olfactory apparatus including olfactory bulbs, tracts and nerves and a vestigial *trigonum olfactorium* were noted. This, to the best of our knowledge is the first report in the literature indexed in the Medline of concurrent occurrence of congenital arhinia and choanal atresia in a goat.

Key words: choanal atresia, congenital arhinia, craniofacial anomalies, goat

Congenital abnormalities are developmental errors that occur in various animal species during pregnancy. These developmental inaccuracies or mistakes have been reported to have a wide range of possible defects that may affect single structure or function and sometimes, the defects may affect combination of structures and functions which may eventually lead to economic losses through reproductive waste

and enhanced perinatal mortalities (Alhaji *et al.*, 2013). Development of the nose is complex and begins when the frontonasal processes appear during the first trimester of gestation; they elevate into the dorsum and apex of the nasal cavity (Petrova & Lobko, 1977; Kim *et al.*, 2004). Ovoid thickenings of ectodermal origin called nasal placodes invaginate to form the nasal pits which deepen dorso-caudally to

form the nasal part of the oronasal cavity. Lateral and medial processes are formed from mesenchymal proliferations in the margins of the nasal pits. These processes subsequently fuse forming the nose, nasal septum and the nasal alae (Steding, 2008). An epithelial plug is formed which fills the nasal cavity which then dissolves later in gestational life. Ectodermal cells of the nasal placodes differentiate to form primary sensory neurons. These cells develop axons which then project to the olfactory bulb which contain secondary neurons (Kim *et al.*, 2004). Anomalies of the nose that may arise during development range from complete aplasia of the nose to duplications and nasal masses (Fijałkowska & Antoszewski, 2016; Funamura & Tollefson, 2016). Arhinia is an example of such congenital nasal deformities (Loose *et al.*, 2004; Funamura & Tollefson, 2016).

Arhinia is the congenital partial or complete absence of the soft tissue of the nose and nasal structures (Baruah *et al.*, 2014). It may be associated with other cranio-facial anomalies such as hypertelorism, microphthalmia, eyelid coloboma, facial clefts, choanal atresia, microtia and midline defects (Akkuzu *et al.*, 2007; Méndez-Gallart *et al.*, 2009). Choanal atresia, on the other hand, refers to the unilateral or bilateral anatomical closure of the choanal openings. It may be membranous, osseous or mixed. However, it usually presents as mixed or osseous (Assanasen & Metheetrairut, 2009). It may lead to severe respiratory distress and subsequently cyanosis. The choanae develop in the first trimester of gestation following rupture of the vertical epithelial fold between the olfactory groove and the roof of the stomodeum (Steding, 2008). This condition is relatively common in Alpacas and uncommon in humans and other

mammals (Reed *et al.*, 2010). The cause of choanal atresia is unknown, however, several theories have been proposed to explain its pathogenesis: persistent buccopharyngeal membrane, incomplete resorption of the nasopharyngeal mesoderm and local misdirection of neural crest cell migration (Andaloro & La Mantia, 2019; Kurosaka, 2019). Diagnosis is done through physical examination, radiography, endoscopy, ultrasonography and computed tomography. Treatment depends on the type of choanal atresia; if bilateral, surgery is necessary. The aim of surgical approach is to open up the choana. Two approaches are mainly used – the transnasal endoscopic approach and the transpalatal approach (Tusaliu *et al.*, 2015).

Case presentation

On the 19th of September, 2019, a day-old male Kalahari red × West African Dwarf cross kid was presented to the Veterinary Clinic of Institute of Food Security, Environmental Resources and Agricultural Research (IFSERAR) of the Federal University of Agriculture Abeokuta, Ogun State, Nigeria. The kid was born at full term without assistance, weighing 1.8 kg and was the result of its dam's second pregnancy with the kid being the only offspring of this pregnancy. There had been no previous history of congenital abnormality. The kid was observed to be weak, frail and was unable to suckle postpartum.

On careful observation, the kid presented with facial malformations, most notably the absence of an external nares (Fig. 1). In addition, there was a right lateral deviation of the upper jaw structures and hard palate (Fig. 1 and 2). Clinically, the kid was dyspnoeic and only struggled to breathe through the oral cavity with the

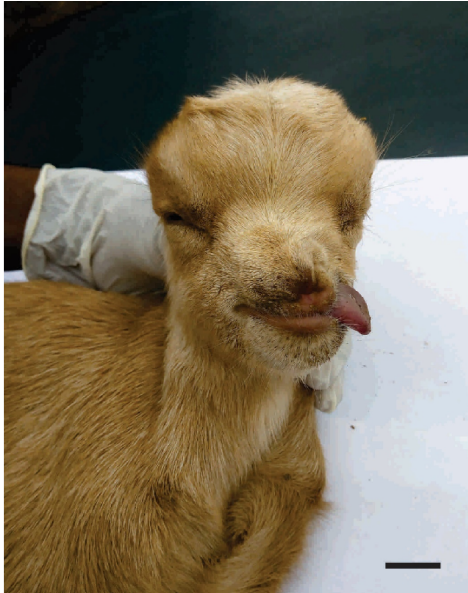


Fig. 1. Front view of the head showing absence of nostrils with right lateral deviation of the upper jaw (bar 4 cm).

tongue protruding and displaced laterally. After a few minutes, the kid passed away and a post-mortem exam was carried out.

Post-mortem findings were centered around facial deformities. A mid-sagittal skin incision was made on the face from the frontal region to the tip of the philtrum on the upper lip. The skin and connective tissues were reflected to examine the underlying bony structures of the face. The right and left nasal bones were fused along their medial borders. Also, the rostral extremities of the nasal bones were fused together with the nasal process of the premaxilla bones, thus presenting as a conical shaped structure with a tapering apex and an occluded nasal vestibule (Fig. 3). Rostral to this, the dorsolateral and ventrolateral nasal cartilages appeared as a fused non-patent mass. On further examination of the pharynx, a bilateral bony occlusion of the choana was noticed, al-

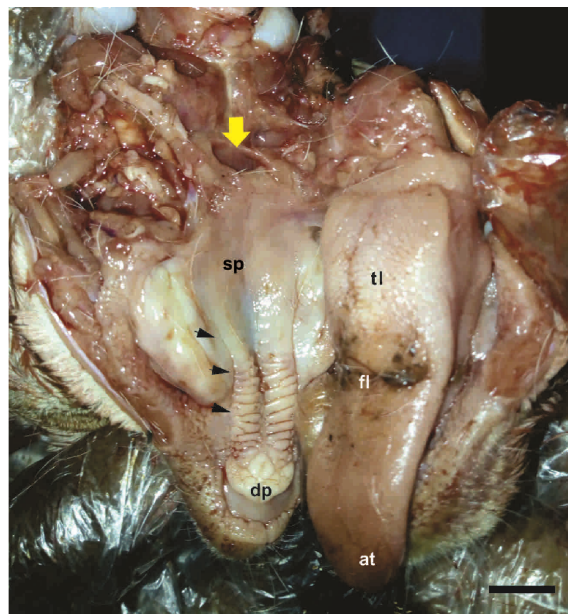


Fig. 2. Dissected oropharynx of the kid showing a deviated hard palate (black arrows). An osseous choanal obstruction was noted with a patent choanal membrane (yellow arrow); at: apex of tongue; dp: dental pad; fl: *fossa linguae*; sp: soft palate; tl: *torus linguae* (bar 1 cm).

though the choanal membrane opening to the pharynx was patent (Fig. 2).

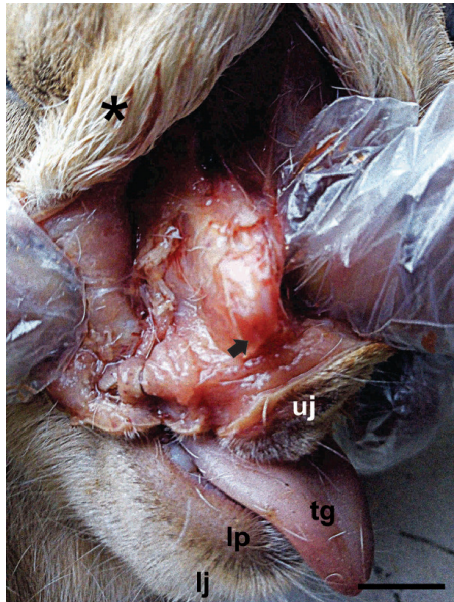


Fig. 3. The bony and cartilaginous frame work of the nose appearing as a conical shaped mass with a tapering apex with no patent nasal vestibule (black arrow); lj: lower jaw; lp: lower lip; tg: tongue; uj: upper jaw; asterisk (*): Incised and reflected skin over the nasal region (bar 1 cm).

Afterwards, the midline skin incision was extended further backwards from the frontonasal area to the nuchal region to expose the underlying cranium. The cranial bones were inspected grossly with all appearing apparently well formed. The kid's brain was then exposed, as described by Mustapha *et al.* (2019), by careful chipping of the cranial bones beginning from the foramen magnum and progressing rostrally to the temporal, parietal and frontal bones. The brain was then gently reflected from its base and exteriorised after severing the cranial nerves. On observation of the brain, the gross structures

of the brain appeared normal except for the rhinencephalic region. The brain had no olfactory bulbs and tracts. Also, the medial and lateral olfactory striae were not delineated from the vestigial *trigonum olfactorium* (Fig. 4 and 5).

Arhinia is a rare congenital malformation during embryogenesis characterised by the lack of or absence of soft tissues of the nose and nasal structures (Takci *et al.*, 2013). Losee *et al.* (2004) classified arhinia as a type I congenital nasal anomaly characterised by hypoplasia and atrophy, representing paucity, atrophy, or underdevelopments of skin, subcutaneous tissue, muscle, cartilage, and/or bone. The post mortem observations in this case report were consistent with features of arhinia as the kid had no external nares and nasal vestibule with a malformed bony and cartilaginous framework of the nasal cavity.

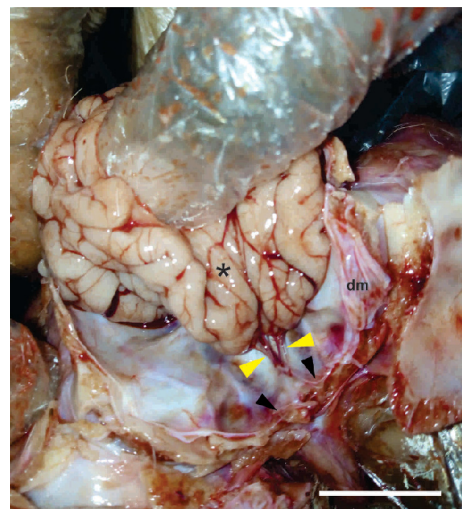


Fig. 4. Rostradorsal view of the brain showing well-formed cerebral cortices, sulci and gyri. Note the paired vestiges of the olfactory nerves (yellow arrowheads) extending from the cribriform plate of the ethmoid bone (black arrowheads) to the rostral floor of the brain; asterisk: cerebral hemisphere; dm: dura mater (bar 1 cm).

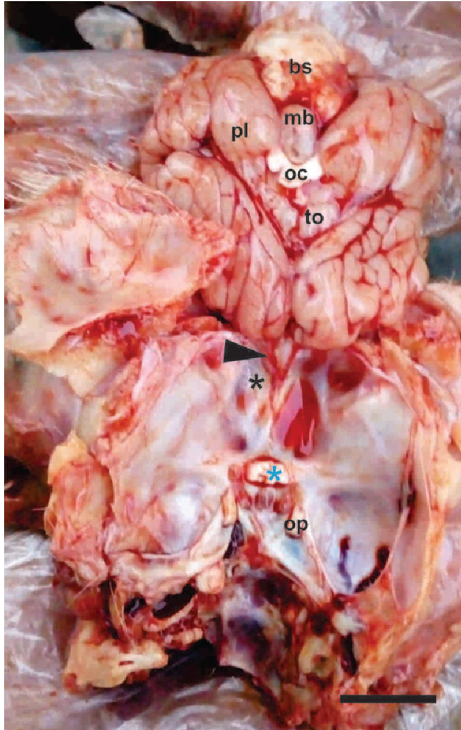


Fig. 5. Rostroventral view of the reflected kid's brain with no olfactory bulbs, tracts and striae. Note the vestigial *trigone olfactorium* (to). Notice the area cribriformis (black asterisk) with no olfactory bulb. Arrowhead: vestigial olfactory nerve; blue asterisk: infundibular stalk with pituitary body lodged in the sella turcica (sphenoid bone); bs: brain stem; mb: mammillary body; oc: optic chiasma; op: optic nerve (severed); pl: piriform lobe; (bar 1 cm).

This condition is known to be accompanied by both craniofacial and brain anomalies such as choanal atresia, absence of olfactory bulbs and/or nerves, hypertelorism, microphthalmia, eyelid coloboma, facial clefts, microtia and other midline defects (Losee *et al.*, 2004; Akkuzu *et al.*, 2007; Marinov *et al.*, 2007; Méndez-Gallart *et al.*, 2009; Funamura & Tollefson, 2016). In this case, the accompanying malformations observed include

absence of olfactory bulbs in the brain, facial distortion consequent to the right lateral deviations of the upper jaw and bilateral osseous choanal atresia. Choanal atresia is a congenital condition characterised by a bony and/or membranous obstruction of the internal nares, which may be unilateral or bilateral. Alhaji *et al.*, (2013) reported a case of congenital anophthalmia and choanal atresia in a two-month old kid, however the type of choanal atresia was not stated. As most neonates are considered obligate nasal breathers because of the relatively elevated larynx when compared to adults, airway obstruction in arhinia poses a serious threat to life, as it may lead to cyanosis and consequently death (Goyal *et al.*, 2008). This may have been the most likely cause of this kid's death.

Nasal development is the result of a complex embryologic patterning and fusion of multiple primordial structures. Loss of signalling proteins or failure of migration or proliferation can result in structural anomalies with significant cosmetic and functional consequences (Funamura & Tollefson, 2016). The pathogenesis and aetiology of this condition is not well understood, however, several postulations have been made. It may result from failure of development of the nasal placodes leading to undeveloped nasal prominences and septum. This could also secondarily lead to failure of development of the olfactory bulb and tract due to the lack of stimulation from the developing nose and olfactory epithelium (Olsen *et al.*, 2001; Mondal & Prasad, 2016). Although the development of the olfactory bulb and olfactory epithelium is simultaneous and independent at first, it becomes interrelated when the axons from the olfactory epithelium innervates the olfactory bulb (Treloar *et al.*, 2010).

Diagnosis of this condition can be made during gestation with the use of ultrasound (Olsen *et al.*, 2001). Management is difficult and involves a multidisciplinary approach as well as expert neonatal care. This includes orogastric feeding and temporal tracheostomy as well as maxillary osteotomy, vertical facial distraction and nasal reconstruction permanently (Feledy *et al.*, 2004; Brusati & Colletti, 2012).

Arhinia is generally a rare congenital condition with only about 45 cases recorded in history from 1931 till date in humans (Mondal & Prasad, 2016). Fewer incidences of this anomaly have been reported in animals. For instance, Schulze & Distl, (2006) reported a case of arhinia and cyclopia in a calf while Sutaria *et al.* (2012) and Patel *et al.* (2019) documented the same in buffalo calves. Cyclopia, arhinia and hermaphroditism have also been reported in a Saanen kid (Karan *et al.*, 2011). To the best of our knowledge, concurrent congenital arhinia and bilateral osseous choanal atresia in goats have not been documented in literature. This may likely be the first case observed in goats and is hereby reported.

Since congenital arhinia is considered sporadic with hereditary transmission not yet documented, farmers do not necessarily need to cull or prevent from breeding dams that deliver kids with this congenital anomaly. Further investigations should be carried out in order to determine the exact cause(s) and predisposing factors in order to institute preventive and adaptive management measures to minimise economic losses by farmers and breeders.

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