Case Report

A Case History of Gross and Radiological Observations of Agnathia: Otocephaly in a Mehraban Ewe-lamb

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ABSTRACT

Agnathia is one of the first pharyngeal arch deformities referred to as mandibular abnormality. This case report aimed to describe an unusual form of agnathia-otocephaly in a Mehraban ewe-lamb accompanied by other malformations. The scrutiny of the lamb's face indicated a chain of abnormalities in the head region. Lack of mandible, lips, rima oris, oral cavity, tongue, and teeth were recognized. Eyes and ear lobes were normal, but the base of the pinnae met each other ventral to the atlantooccipital joint and formed a single external acoustic meatus there. The nostrils were normally formed, but the philtrum was not formed due to the lack of lips, especially the upper lip. The hyoid apparatus was normally developed. The laryngopharynx had no connection with the nasopharynx and dead end. Also, the nasal cavities ended blindly because of rinopharyngeal aplasia and no choanal foramen. So, the abnormal pharyngeal region caused a non-functional respiratory system followed by death.

Keywords: Agnathia, Congenital, Malformation, Mehraban ewe-lamb

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1. Case History

The congenital defects of pharyngeal arches have been shown in many scientific reports. Agnathia, also known as otocephaly, is one of the first pharyngeal arch deformities, which refer to mandibular abnormality. Agnathia, micrognathia, brachygnathia, parrot beak, bird face, undershot jaw, strophocephalia, and overshot jaw are the scientific terms that are suggested to describe the various types of mandibular defects and applied according to the severity of the presented malformation.

The incidence of agnathia-otocephaly in human populations is low, but on the contrary, small ruminants are prone to this malformation. These defects are mostly along with other abnormalities, especially in the head region (Frixos Pourlis, 2008). The first report on humans was published in 1990 by Brown and Marsh. The otocephaly in small ruminants was described for the first time in 1827 (Dennis & Leipold, 1972). The next

published reported case contains different levels of otocephaly and diverse grading plans with no consensus program (Theret, 1948; Fisher, 1955; McFarland & Deniz, 1964; Smith, 1968; Schneider et al., 1978; Frixos Pourlis, 2008).

Pursuant to classification, breeds of sheep, and grade of agnathia, this malformation in sheep differs amidst the reported studies. In 1972, on scrutiny of spontaneous congenital defects in Merinos breed herds, 25 cases of agnathia (31% deadly anomalies) were detected in 80 lifeless carcasses of newborn lambs (Hughes et al., 1972). According to the study of Dennis and Leipold (1972), in Western Australia, over 3 years, 401 deformed lambs were evaluated, and 74 cases of diverse forms of agnathia (18.3% lethal defects) were recorded.

In the present case report, we describe an unusual form of agnathia-otocephaly in a Mehraban ewe-lamb, accompanied by other malformations.

3. Diagnostic Testing

Anatomical findings

External examination of the lamb showed that the head region was abnormal (Figure 1B). The mandible, lips, rima oris, oral cavity, tongue, and teeth were not formed in the head region. Eyes and ear lobes were normal (Figure 1B), but the base of the pinnae met each other ventral to the atlantooccipital joint and formed a single external acoustic meatus there (Figure 1A). The nostrils were normally formed, but the philtrum was not formed due to the lack of lips, especially the upper lip.

The pharynx was inflated below the throat, ventral to the atlas and axis (Figure 2C and Figure 2D). No other malformations were observed in the external examination of the other body regions, and the trunk, limbs, tail, and neck represented normal structures. After peeling and dissection of the head and cervical areas, the lack of formation of the mandible, oral cavity, tongue, and teeth was confirmed. The maxilla was incompletely organized, and its alveolar process was not developed. The hard palate existed with low thickness. The nasal conchae, ethmoidal conchae, and meatuses were formed ordinarily. But the nasal cavities ended blindly due to aplasia of the rinopharynx and no choanal foramen (Figure 2A). Also, the nasal septum was developed obliquely (Figure 1D). The palatine sinus was the single paranasal sinus which was well distinguishable (Figure 2B).

In the pharynx, the distal end of the esophagus was closed and inflated at the laryngopharynx region (Figure 2, C and D). Also, the megaesophagus was seen in the cervical and thoracic parts of the esophagus (Figure 2D and 3D). Yellowish fluid accumulated at this inflated end of the esophagus (Figure 3B). Laryngeal cartilages were formed normally. The carotid artery and jugular vein were normally formed up to the head on both sides of the neck. But, left and right facial arteries were split on both sides and then met each other at the ventral midline of the head. After going on to the middle of the head, this single fascial artery was ramified left and right lateral nasal arteries.

At the site of carotid artery division in the head area, diverticula or aneurysms were seen on both sides; the left was bigger than the right (Figure 3C). The maxillary artery, a branch of the carotid artery, separated from the carotid artery next to the diverticula and went to the upper parts of the head.

The parotid salivary gland was formed ordinarily, but the sublingual and mandibular salivary glands were not observed. Also, the thymus gland and the trachea at the neck region were clear.

Radiologic observations

In x-ray observations, the lack of mandible, oral cavity, tongue, oropharyngeal and nasopharyngeal cavities was confirmed. Also, the thin frontal bone without sinus was recognized. The occipital condyles, hyoid apparatus, and nasal conchae were seen in the radiograph (Figure 1C and 1D).

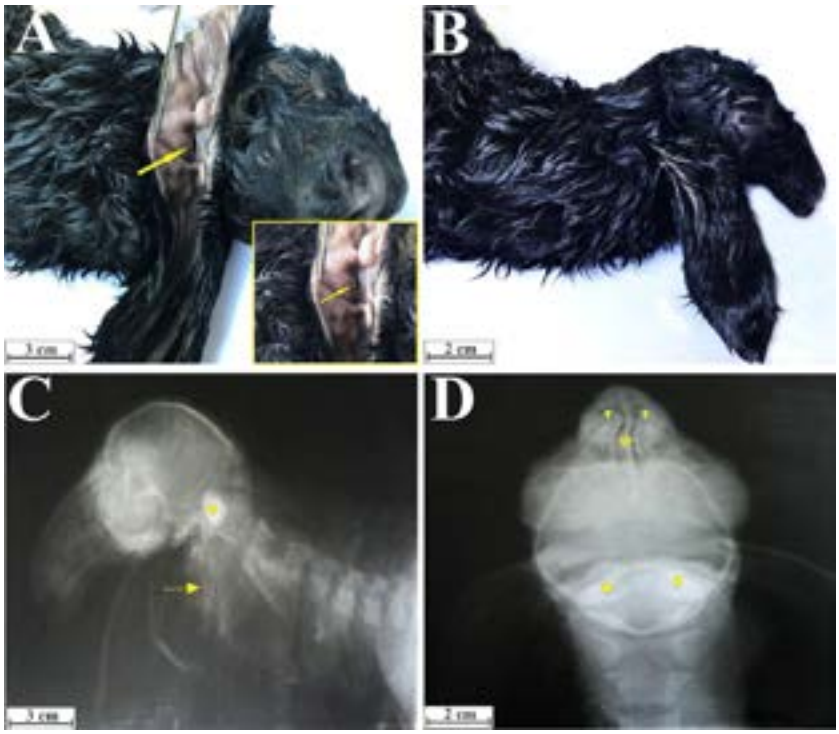

Figure 1. A) Ventral view of the single external acoustic meatus (arrow)

B) Lateral view of the head region

C) Lateral radiograph of the head region demonstrating the lack of mandible, oral cavity, tongue, oropharyngeal, and nasopharyngeal cavities; reduced bone opacity indicating a thin frontal bone without sinuses in the dorsal part of the skull. The occipital condyle (circle) and the hyoid apparatus (dashed arrow) in the caudoventral part of the skull

D) In the dorsoventral view, the occipital condyles (circles), a spiral nasal septum (asterisk), and two deformed nasal conchae (arrow heads)

4. Assessments

The neonatal Mehraban ewe-lamb characterized in this study is a fatal congenital disorder named agnathia. In this case, the oral cavity, tongue, mandible, teeth, ears, and face were malformed. The abnormal pharyngeal region developed in the presented lamb caused a non-functional respiratory system and was followed by early death after birth.

The mandibular absence and related ear and fascial abnormalities were classified in different plots, and the authors categorized the head malformations in various animals (Gladstone & Wakeley, 1923). Dennis and Leipold (1972) categorized the first pharyngeal arch abnormalities morphologically, divided into 5 main types and consisting of all mandibulofacial malformations. This case of first pharyngeal arch abnormality illustrates a moderate form of type 3 otocephaly.

The history of mandibular development is complicated in vertebrates. The momentous pathogenetic mechanism of mandibular abnormalities implicates the ventral part of the first pharyngeal arch, which can be a secondary malformation, and insufficient immigration or proliferation of the neural crest cells arising from the dorsal part of the neural epithelium can be a primary factor to that secondary malformation (Wright & Wagner, 1934). Also, it was proposed that agnathia can result from two factors. The first is a perturbation in neural differentiation in the region of the cranial neural plate, and the second is a malfunction of mesodermal elements, which are needed to induce the neural plate and neural crest derivatives (Pauli et al., 1983). Differently, agnathia can result from a primal deficiency of the mesoderm in mandibular prominence (Juriloff et al., 1985).

Macrì et al. (2008) described the radiological and gross findings of the agnathia condition observed in a male Comisana lamb. Clinical observations showed that the lamb had astoria, proboscis, lack of mandible, bilateral hypoplasia of the maxilla, and slightly prominent fluctuant pharyngeal diverticulum. Also, the pinnae of the

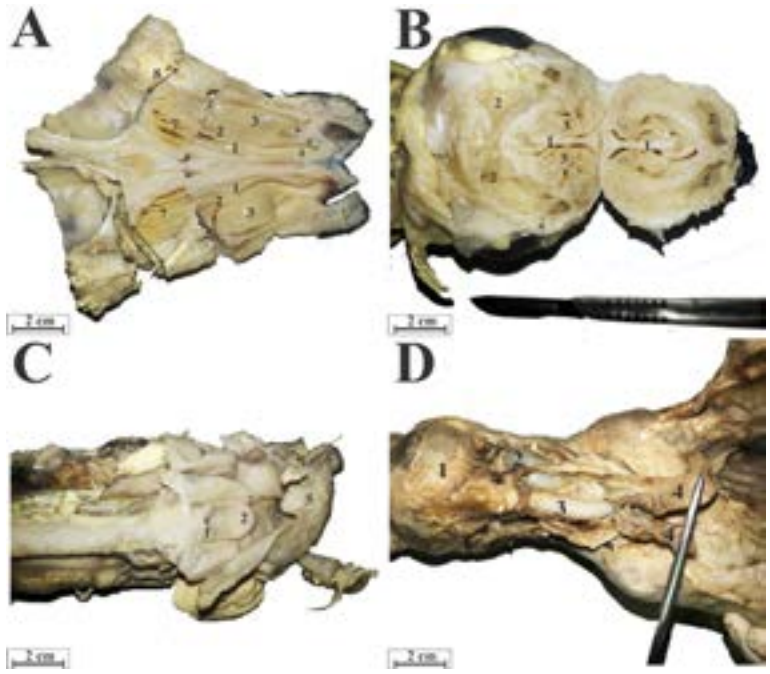

Figure 2. A) Sagittal section of the nasal cavity, 1) Dorsal nasal conchae, 2) Middle nasal conchae, 3) Ventral nasal conchae, 4) Straight fold, 5) Alar fold, 6) Basal fold, 7) Ethmoidal conchae, 8) Blind end of nasal cavity

B) Transverse section of nasal cavity, 1) Nasal septum, 2) Palatine sinus, 3) Nasal conchae

C) Ventral view of the pharynx, 1) Arytenoid cartilage , 2) Epiglottis cartilage, 3) Stylohyoid process, 4) Laryngopharynx, 5) Inflated pharynx

D) Ventral view of the cervical region,1) Inflated pharynx, 2) Megaesophagus, 3) Trachea, 4) Thymus gland

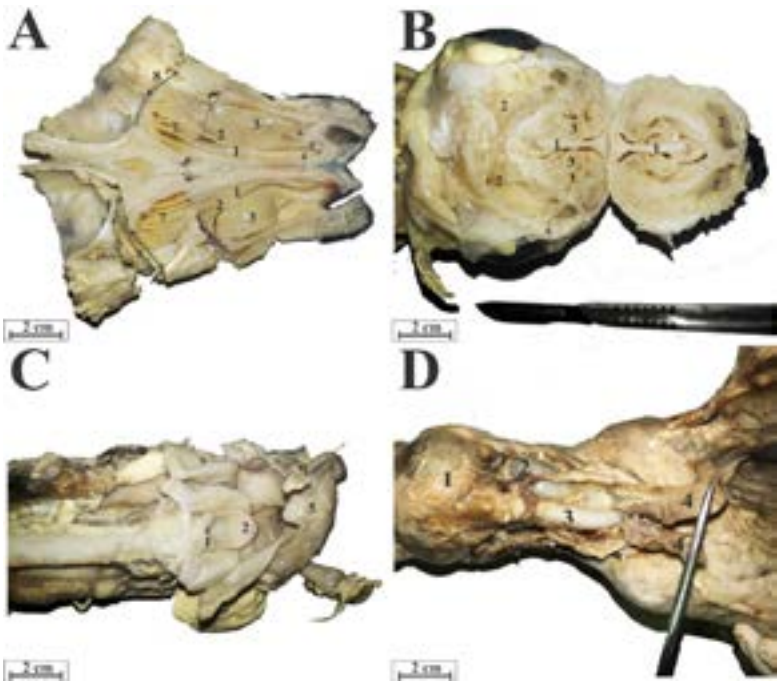

Figure 3. A) The brain with shallow gyri (black asterisk)

B)Ventral view of the pharynx, yellowish fluid (yellow asterisk), glottic cleft (yellow arrow head)

C) Diverticula or aneurism (black arrow) at the site of carotid artery division

D) Lateral view of thoracic cavity, megaesophagus (black dashed arrow)

ears were fused in the ventral midline (synotia) with a common athresic external auditory meatus, similar to our reported case. Furthermore, the absence of the mandible, hyoid bones, tympanic bullae and hypoplasia of the maxilla, laryngopharyngeal diverticulum, and dilation of the esophagus were other observations (Macrì et al., 2008). In the current study, in the head region, mandible, lips, rima oris, oral cavity, tongue, and teeth were not formed, similar to those in a previous report (Macrì et al., 2008).

A female lamb of the Chiotiko breed with severe agnathia-otocephaly has been reported in another study. This report showed a lack of the mandible and related structures, extensive head malformation, synophtalmia-exophthalmia, and synotia (Frixos Pourlis, 2008). Similar to the findings of the present report, drastic encephalon deformity was seen in the cranial cavity. The nasal and ethmoidal conchae were normally developed and distinguishable, while the nasopharyngeal aplasia caused blind termination of the nasal cavities. The nose and nasal septum were a little slant. The skeleton of the female lamb was normal except for the skull. The lower jaw, superficial and masticatory muscles, and oral cavity were completely absent (Frixos Pourlis, 2008). Most reports in this study were also observed in the current report.

The lack of hyoid bones and system was observed in Chiotiko lamb agnathia (Frixos Pourlis, 2008), but the hyoid apparatus was completely developed in our case. In Chiotiko lamb agnathia, the cranial cavity was almost unoccupied (Frixos Pourlis, 2008). Whereas, in Mehraban lamb with agnathia, the cranial cavity was occupied with normal size brain with shallow gyri (Figure 3A).

Vali et al. (2014), using 3-D CT scan methods, reported a lamb was born with an abnormal appearance and mandibular aplasia that died shortly after birth. This report observed mandibular aplasia, skull dysplasia, mandibular aplasia, synophtalmia, and incisive and nasal bone dysplasia Vali et al. (2014). In the present report, the nostrils were normally formed, but the philtrum was not formed due to the lack of lip formation, especially the upper lip.

In Frixos Pourlis (2008) report, the salivary glands were absent, while the parotid salivary gland was formed ordinarily in the current report. Also, the sublingual and mandibular salivary glands were not observed. The lack of the right kidney has been reported in humans and Chiotiko lamb (Pauli et al., 1981; Frixos Pourlis, 2008). Also, one case of unilateral renal agenesis was reported in 74 lambs with agnathic deformity (Dennis & Leipold, 1972). But, in this case, kidneys were formed normally.

Ethical Considerations

Compliance with ethical guidelines

There were no ethical considerations to be considered in this research.

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Authors' contributions

All authors equally contributed to preparing this article.

Conflict of interest

The authors declared no conflict of interest.

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References

Brown, D. M., & Marsh, J. L. (1990). Agnathia and associated malformations: A case report. *The Cleft Palate Journal, 27*(4), 415–418. [PMID]

Dennis, S. M., & Leipold, H. W. (1972). Agnathia in sheep: External observations. *American Journal of Veterinary Research, 33*(2), 339–347. [PMID]

Fisher, H. (1955). Agnathia, a rare lethal malformation in sheep. Hemera Zoa, 62, 260.

Frixos Pourlis, A. (2008). Spontaneous agnathia-otocephaly in a lamb of the Chiotiko breed-a case report. *Veterinary Arhiv, 78*(5), 449-56. [Link]

Gladstone, R. J., & Wakeley, C. P. (1923). Defective Development of the Mandibular Arch: The etiology of arrested development and an inquiry into the question of the inheritance of congenital defects. *Journal of Anatomy*, 57(Pt 2), 149–167. [PMID]

Schneider, H., Schneider, J., & Wassmuth, R. (1978). Chromosomal aberrations and Hb-deficiency in agnathic sheep. *Deutsche Tieraerztliche Wochenschrift*, 85, 472-474.

Hughes, K. L., Haughey, K. G., & Hartley, W. J. (1972). Spontaneous congenital developmental abnormalities observed at necropsy in a large survey of newly born dead lambs. *Teratology, 5*(1), 5-10. [DOI:10.1002/tera.1420050103] [PMID]

Juriloff, D. M., Sulik, K. K., Roderick, T. H., & Hogan, B. K. (1985). Genetic and developmental studies of a new mouse mutation that produces otocephaly. *Journal of Craniofacial Genetics and Developmental Biology, 5*(2), 121-145. [PMID]

Kamiji, T., Takagi, T., Akizuki, T., Kurukata, M., & Ohmori, K. (1991). A long surviving case of holoprosencephaly agnathia series. *British Journal of Plastic Surgery, 44*(5), 386-389. [DOI:10.1016/0007-1226(91)90155-d] [PMID]

Macrì, F., Fascetto, G., Marino, F., Lanteri, G., & Mazzullo, G. (2008). Agnathia in a lamb: Radiological and gross observations. *Large Animal Review, 14*(4), 175-179. [Link]

McFarland, L. Z., & Deniz, E. (1964). Extreme agnathia in a ewe lamb. *The Cornell Veterinarian, 54,* 541-544. [PMID]

Pauli, R. M., Pettersen, J. C., Arya, S., & Gilbert, E. F. (1983). Familial agnathia-holoprosencephaly. *American Journal of Medical Genetics, 14*(4), 677–698. [DOI:10.1002/ajmg.1320140411] [PMID]

Pauli, R. M., Graham, J. M., Jr, & Barr, M., Jr (1981). Agnathia, situs inversus and associated malformations. *Teratology, 23*(1), 85–93. [DOI:10.1002/tera.1420230111] [PMID]

Smith I. D. (1968) Agnathia and micrognathia in the sheep. *Australian Veterinary Journal, 44*(11), 510–511. [DOI:10.1111/j.1751-0813.1968.tb09000.x] [PMID]

Theret, M. (1948). Une mutation léthale recessive chez le mouton. *Recueil de Medecine Veterinaire*, 84, 445-447.

Vali, Y., Soroori, S., & Ragh, M. J. (2014). Three-dimensional CT scan evaluation of agnathia in a lamb. Paper presented at: Proceeding of the 4th international symposium of veterinary surgery (ISVS), 21-23 oct, Mashhad, Iran. [Link]

Wright, S., & Wagner, K. (1934). Types of subnormal development of the head from inbred strains of guinea pigs and their bearing on the classification and interpretation of vertebrate monsters. *American Journal of Anatomy, 54*(3), 383-448. [DOI:10.1002/aja.1000540303]

Yamane, A. (2005). Embryonic and postnatal development of masticatory and tongue muscles. *Cell and Tissue Research, 322*(2), 183-189. [DOI:10.1007/s00441-005-0019-x] [PMID]

گزارش موردی

گزارش بررسی ظاهری و رادیولوژیک آگناتیا اتوسفالی در یک بره نژاد مهربان

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چکیده

زمینه مطالعه: آگناتیا یکی از بدشکلی‌های اولین کمان حلقی است که به ناهنجاری فک پایین منجر می‌شود. هدف از این گزارش تشریح شکل غیرمعمول آگناتیا اتوسفالی در بره میش مهربان بود که با ناهنجاری‌های دیگری همراه بود. بررسی دقیق صورت بره نشان‌دهنده وجود زنجیره‌ای از ناهنجاری‌ها در ناحیه سر بود. عدم وجود فک پایین، لب‌ها، شکاف دهان، حفره دهان، زبان و دندان‌ها تشخیص داده شد. چشم‌ها و لوب‌های گوش نرمال بودند، اما قاعده لاله‌های گوش در ناحیه شکمی مفصل اطلسی-پس‌سری به هم رسیده و یک مجرای گوش خارجی واحد را در قسمت شکمی مفصل اطلسی-پس‌سری تشکیل داده بودند. سوراخ‌های بینی به شکل طبیعی تشکیل شده بود، اما به‌دلیل تشکیل نشدن لب‌ها به‌ویژه لب بالایی، فیلتروم تشکیل نشده بود. دستگاه لامی به‌طور نرمال توسعه یافته بود. ناحیه حنجره‌ای حلق، ارتباطی با ناحیه بینیای حلق نداشت و به‌صورت تهبسته خاتمه یافته بود. همچنین حفره‌های بینی به‌دلیل عدم تشکیل ناحیه بینیای حلق به‌صورت تهبسته خاتمه یافته بود و سوراخ شوان وجود نداشت. بنابراین ناحیه حلقی غیرنرمال باعث ناکارآمدی سیستم تنفسی و به دنبال آن مرگ بره شده بود.

کلیدواژه‌ها: آگناتیا، بدشکلی، مادرزادی، بره میش مهربان

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