

Choanal Atresia/Multiple Malformation Syndrome in Alpaca

Aníbal G. Armién and Kent Reed

College of Veterinary Medicine, University of Minnesota

Choanal Atresia (CA) is a malformation characterized by an abnormal development of the nasal passages which provide the airway connection between the nose and larynx. The major clinical presentation of CA in camelids is persistent dyspnea (Fowler, 1989). Blockage of the choanae, either partial or complete, is especially problematic for newborn cria (primary nasal breathers). Crias with CA have a severe prognosis with no practical treatment alternative. As a result, euthanasia is the only option to relieve the animal from prolonged suffering. CA is rarely reported in most companion animals, but is relatively common in new world camelids occurring in 0.48% and 0.75% of live births in alpacas and llamas (Johnson, 2008), respectively. In all these species clinical and pathological exploration has normally focused on the choanal defects (Fowler, 1989; Hogan et al., 1995; Coolman et al., 1998; Khoo et al., 2007). However, associated cardiac, gastrointestinal, skeletal or neural anomalies are reported in 33% of the choanal atresia cases in llamas (Leipold et al. 1994). Lately, we are finding evidence that familial and sporadic cases of choanal atresia in alpacas are frequently associated with multiple defects including multiple subtle to marked, cranial bone deformations, hypoplasia and aplasia of cranial nerves and encephalic structures, brain heterotopy, heart defects as well as other defects. The etiology of CA is poorly understood. Evidence suggests that the condition is a genetically inherited, recessive or perhaps polygenic trait. We hypothesize that choanal atresia in alpacas and llamas is part of a multiple malformation complex, which shares major clinical criteria with a similar syndrome (CHARGE) in humans, caused by a gene mutation (Sanlaville and Verloes 2007). We are currently characterizing multiple malformations associated with choanal atresia and working to determine whether in analogy to the human syndrome, a specific gene mutation (*CHD7*) is the cause of the syndrome in alpacas and or llamas. Recently, a sequence of *CHD7* coding region was obtained from a non-affected cria. The alpaca *CHD7* mRNA sequence was 97.9% similar to that of the human, with the greatest sequence difference being an insertion in exon 38. This study provides the genomic reagents necessary to determine if mutations in *CHD7* are the cause of CA in camelids (Reed et al 2010).

Coolman BR, Marretta SM, McKiernan BC, and Zachary JF. Choanal atresia and secondary nasopharyngeal stenosis in a dog. *J Am Anim Hosp Assoc*, 1998, 34(6):497-501.

- Fowler ME. Congenital/hereditary Conditions. *In: Medicine and Surgery of South American Camelids*, Iowa State University Press, Ames, IA, 1989.
- Hogan PM, Embertson RM, and Hunt RJ. Unilateral choanal atresia in a foal. *J Am Vet Med Assoc*, 1995, 207(4): 471-473.
- Johnson LW. 2008. Choanal atresia and wry face in camelids. Abstract. 1st International Workshop on Camelid Genetics.
- Khoo AM, Marchevsky AM, Barrs VR, and Beatty JA. Choanal atresia in a Himalayan cat—first reported case and successful treatment. *J Feline Med Surg*, 2007, 9(4): 346-349.
- Leipold HW, Hiraga T, Johnson LW. Congenital defects in the llama. *Vet Clin North Am Food Anim Pract*, 1994,10: 401-420.
- Reed K.M., & Chaves L.D. (2008) Simple sequence repeats for genetic studies of alpaca. *Animal Biotechnology* 19, 243-309.
- Reed K.M., Bauer M.M., Mendoza K.M., and Armién A.G.: A Candidate Gene for Choanal Atresia in Alpaca. *Genome*, 2010. **In Press**
- Sanlaville D, and Verloes A. CHARGE syndrome: An update. *Eur J Hum Genet*, 2007, 15: 389-399

Author address:

Anibal G. Armien, DVM, MSc, PhD, Diplomate, ACVP
Associate Professor of Veterinary Pathology
Minnesota Veterinary Diagnostic Laboratory,
College of Veterinary Medicine
University of Minnesota
1333 Gortner Ave
St. Paul, MN 55108
Office phone: 612 625 8270
Fax: 612 624 8707
<http://www.cvm.umn.edu/vdl/ourservices/ElectronMicroscopy/home.html>