

## Multiple congenital malformation in a Simental female calf: a case report

O. SMOLEC<sup>1</sup>, J. KOS<sup>1</sup>, D. VNUK<sup>1</sup>, M. STEJSKAL<sup>1</sup>, N. BRKLJACA BOTTEGARO<sup>1</sup>, R. ZOBEL<sup>2</sup>

<sup>1</sup>Faculty of Veterinary Medicine, University of Zagreb, Zagreb, Croatia

<sup>2</sup>Veterinary Practice, Struzec, Croatia

**ABSTRACT:** Congenital anomalies in offspring of natural breedings are often a result of environmental factors, genetic factors, or both. A 21-day-old male Simental female calf was submitted to our clinic with abnormal severe green mucoid nasal discharge from both nostrils. At necropsy, the calf showed multiple defects, including cleft palate, hydrocephalus, diaphragmatic hernia and freemartinism. The cause of these anomalies could not be determined. This report is the first to describe these multiple malformations in a single calf.

**Keywords:** Simental; calf; malformations

Congenital is a descriptive term denoting a condition existing at birth; hence congenital malformations or congenital deformities are defined as abnormalities of structure present at birth. Developmental or congenital abnormalities, defects and anomalies, include functional as well as morphological imperfections. (Rousseaux, 1988). Congenital defects may be compatible with life but are lethal and thus account for an important part of neonatal mortality. Many inherited abnormalities, which result from a change in the DNA base sequence, follow Mendel's principles of independent assortment through chromosomal segregation. Structural genes can be on autosomes or sex chromosomes. Not all inherited abnormalities follow Mendel's principles of dominant and recessive inheritance. Sometimes no simple mode of inheritance can be established and no teratogenic agent is obvious. In such cases, abnormal development may be a result of failure of gene control, failure of cellular and tissue interactions or local environmental effects on gene expression. (Khouri and Gruss, 1983). Many different congenital defects, either of genetic, environmental, or unknown cause, or due to environmental-genetic interaction have been reported in domestic, zoo and feral animals. (Lepoid et al., 1972; Saperstein et al., 1975; Huston et al., 1977, 1978; Lepoid, 1978; Lepoid and Dickenson, 1978).

Congenital defects are reported in all breeds of cattle with variations in the frequency of occurrence. The aetiology of these defects is either genetic (recessive gene) or environmental, and include nutritional deficiencies, endocrine disturbances, extremes of temperature during pregnancy, radiation, drugs, chemicals, toxic plants and infectious diseases (Roberts, 1971). This report describes a unique syndrome of congenital abnormalities in a calf. The number of congenital anomalies was remarkable and included cleft palate, hydrocephalus, diaphragmatic hernia and freemartinism.

### Case description

A 21-day-old female Simental dairy calf was submitted to the Clinic for Surgery, Orthopaedics and Ophthalmology of the Veterinary faculty in Zagreb. The owner reported that the calf had intermittent episodes of coughing and milk dripping from his nose after suckling.

On admission the calf's temperature was 40.9°C, pulse 116 and respiratory rate 88. The breathing was peculiar: the abdomen contracted and the rib cage lifted on expiration; the abdomen expanded and the rib cage lowered on inspiration. Lung sounds were

slightly increased. Mucous membranes were light cyanotic, moist, and the capillary refill time was less than 2 s. There was an abnormal nasal flaring and severe green mucoid nasal discharge from both nostrils. The mandibular lymph nodes were of normal size, and were not painful and tempered. Palpation of the pharynx, larynx and trachea revealed no abnormalities. Comparative percussion of the both sides of the chest was performed and marked dullness in the left half was observed. Auscultation of right side of the lungs revealed enhanced respiratory noises. Thorough auscultation of the opposite side of the chest revealed the presence of typical intestinal sounds: bubbling, gurgling etc.

The calf responded to treatment with trimethoprim sulfa antibiotics and but symptoms recurred once the antibiotics were removed. Two days after submission the calf died.

Necropsy results confirmed the presence of additional external and internal anomalies. The following congenital malformations were observed in Figures 1–4.

## DISCUSSION AND CONCLUSIONS

This calf was unique because of the extent of multisystem malformation. Congenital defects are abnormalities of structure or function present at birth and may account for a high percentage of calf losses from just before to just after calving. The frequency of congenital defects ranges from less than 1 to over 3 percent within herds

The formation of a normal foetus depends on complex intracellular, intercellular and tissue temporal-spatial interactions. Time and space are very important at molecular, cellular, and tissue levels during development. At a molecular level, the times at which certain genes of individual cells are expressed are important in the production of the correct cell mass and cell products necessary for organ formation. (Rousseaux and Wenger, 1985). Each cell carries the hereditary material, genes, on chromosomes. Chromosomes, both sex chromosomes and autosomes, occur in pairs in all body cells (except gametes) that possess a nucleus. Cells containing pairs of chromosomes are called diploid, whereas the gametic form is termed haploid. The chromosomal constitution or karyotype is constant for each species. Abnormal karyotypes usually involve the loss or addition of a chromosome or deletion of part of a chromosome with resulting incorrect gene dosage. This excess or loss of genetic information usually results in phenotypic abnormalities in several organ systems because a single chromosome carries genetic information important to many metabolic and developmental pathways (Shows et al., 1982). The phenotype of a foetus is determined by its genetic constitution and is modified by environmental forces at the molecular, cellular, and organ system levels of development. The production of an abnormal phenotype may be a result of a defect in the genotype, environmental insult, or genotype-environmental interaction. Some defective development occurs because of multiple genetic and/or environmen-

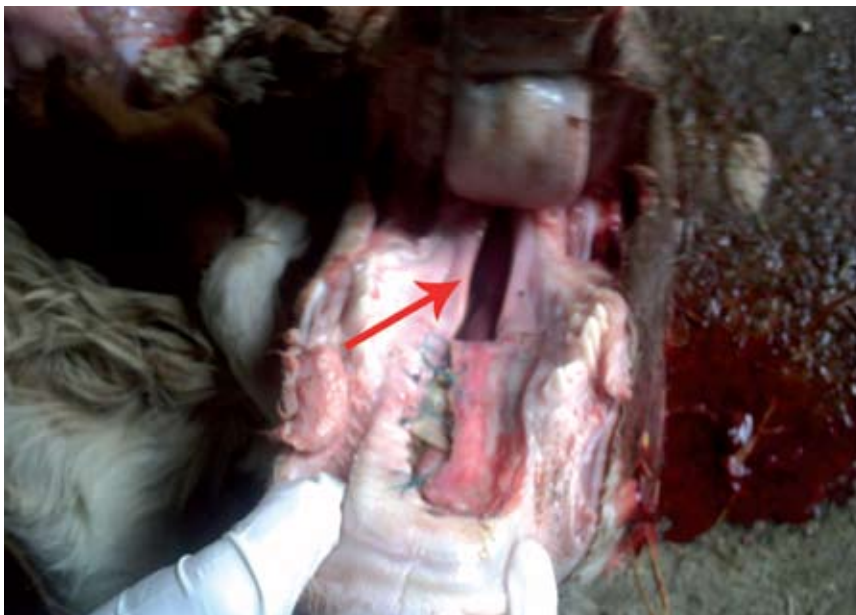


Figure 1. Cleft palate

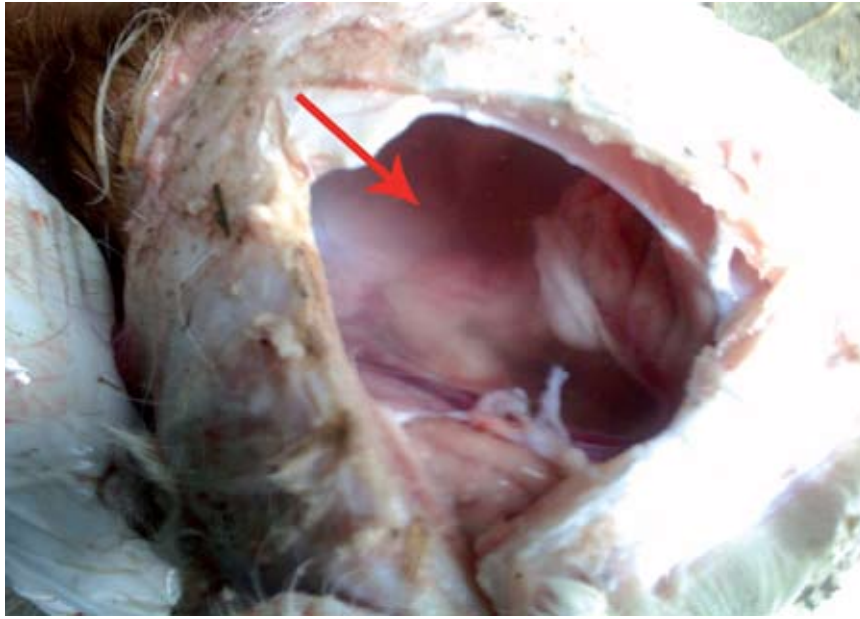


Figure 2. Hydrocephalus

tal factors. Failure of tissues to interact with each other at the correct place and time may also result in abnormalities even though the genotype of the individual is normal and external environmental insults absent (Rousseaux, 1988). The cause of the multitude of developmental anomalies in this calf could not be determined. Many of these anomalies in the bovine population have been associated with genetic factors (transgenes, chromosomes), environmental agents (infections, toxins, fertilization techniques, management), or a combination of factors (Newman et al., 1999). This case report includes descriptions of several unique features in the pathology of bovine fetal malformation.

Combinations of several similar defects have been reported. Internal hydrocephalus can occur concurrently with multiple ocular defects, myopathy, or arthrogryposis in calves. (Lepoid and Dennis, 1987). Cleft palate may occur singly, but in Charolais and Hereford cattle, a recessively inherited syndrome of arthrogryposis and palatoschisis has been described (Lepoid et al., 1974). Clefts of the face are developmental disorders due to failure of closure in facial processes such as the frontonasal maxillary, and mandibular processes. The defects appear in the lateral or median site of the rostral face as cleft lips, jaws, and palates (Morimoto et al., 1999).



Figure 3. Hypoplastic uterus without ovaries

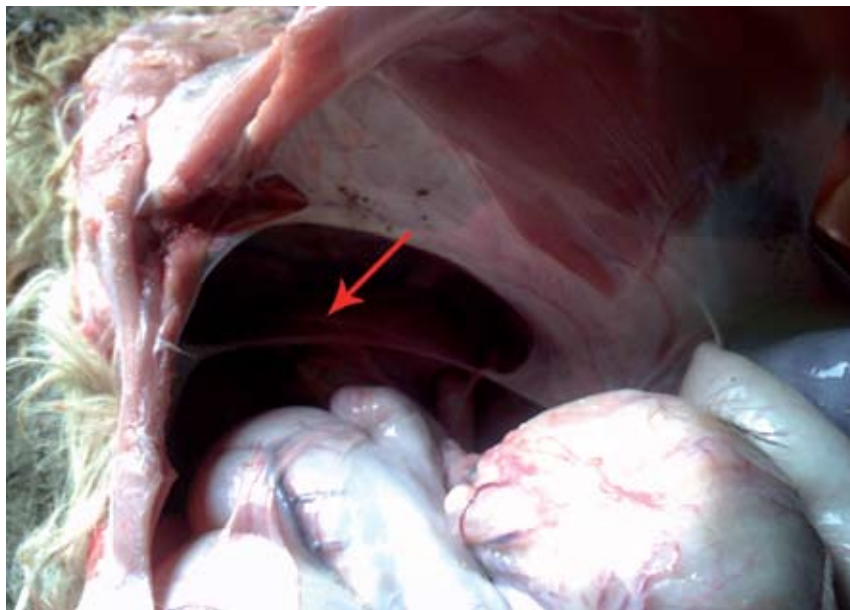


Figure 4. Diaphragmatic hernia

Morphogenesis of the face is very complicated because of the interaction of several organ systems including the central nervous system, upper respiratory and alimentary tract (Pruzansky, 1975). It is known that the foetal organs such as the facial processes, the branchial archs and the developing brain, are intimately connected with the formation of the face in early embryonic life (Pruzansky, 1975; DeMyer, 1975). In the cleft palate, the opening of the bony palate would be a direct change due to disturbance of palatogenesis.

Palatogenesis is accomplished by the initial elevation of the lateral palatine process, which extends ventrally on either side of the tongue, and by the subsequent apposition and fusion of their margins in the early foetal period (Noden and De Lahunta, 1985).

Similarly, a genetic basis has been determined for internal hydrocephalus (a simple autosomal recessive trait in many cattle palates (multifactorial or autosomal inheritance in the Charolais breed; Leech et al., 1978; Rousseaux, 1994). Theoretically, environmental modifications at the cellular level can, through various mechanisms, alter DNA unravelling, transcription, translation, and processing thereby producing morphologically abnormal cells, tissues, and possibly a phenotypically abnormal animal (Felsenfeld, 1978; Breathnach and Chambon, 1981). Hydrocephalus is common in cattle (Cho and Lepoid, 1977).

Congenital hydrocephalus (Water head) is an inherited defect in calves caused by a simple autosomal recessive trait resulting in abnormal accumulation of cerebrospinal fluid within the cerebral

ventricular system characterized by domed skull, poorly developed teeth, depression, blindness and survival for only a few days (Radostits et al., 2005). Defects of the oviducts, uterus, cervix and vagina have been described in several breeds. In many, fusion of the Mullerian ducts is either lacking or exaggerated (Settergren and Galloway, 1965). Freemartins are heifer calves born co-twin to a male. About 93% are affected with variable hypoplasia of organs developing from Mullerian ducts and stimulated development of the Wolffian duct system; they are usually sterile. Bovine freemartinism may not be caused by humoral factors but may be a function of sex chromosome mosaicism. The initial phase of inhibition from day 50 to 75 of gestation is characterised by arrest of gonadal development in the female and regression of Mullerian duct development. The phase of masculinization follows at day (Wijeratne, 1976).

Congenital diaphragmatic hernia does not seem to be one of the rare congenital abnormalities. Since it is difficult to make a diagnosis of this hernia in domestic animals, it is considered that not a few animals suffering from this hernia may have died or been condemned without any definitive diagnosis. (Mitsuo et al., 1989).

More serious, and often overlooked, is a mutant gene that has delayed expression which may become incorporated in the gene pool before much attention is drawn to it (Jolly and Blakemore, 1973). Attempting to define aetiology is important, not only to alleviate the farmer's anxiety, but also to prevent large losses in livestock production and

possible spread of genetic disease in domestic animals. It is important that veterinarians be able to diagnose congenital defects which are of economic significance to the cattle breeding industry. Not only is diagnosis important but practicing veterinarians should be versed in methods for controlling genetically induced defects in cattle.

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## Corresponding Author:

Ozren Smolec, DVM, Ph.D., University of Zagreb, Faculty of Veterinary Medicine, Heinzelova 55, 10000 Zagreb, Croatia  
Tel. +38 512 390 395, E-mail: osmolec@vef.hr