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A complex malformation in a pig: case report and review of the literature

Eine seltene Entwicklungsstörung beim Schwein: Fallbericht und Literaturübersicht

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Summary

Congenital defects like myofibrillar dysplasia (splayleg), umbilical and inguinal hernias, cryptorchism, intersexes, and anal atresia occur relatively frequently in swine. On the other hand, some developmental anomalies like double monsters are very rare. The present paper reports a rare case of a congenital complex malformation including polymelia, duplicitas coli partialis and recti, atresia ani et fistula rectogenitalis, duplicitas corpori uteri, cervicis, vaginae et vulvae and duplicitas vesicae, urethrae et renalis. A plausible interpretation concerning the etiology is that the anomalies arose from unequal partial twinning.

The pig has been healthy and inconspicuous. Although no anus was formed defecation took place via a fistula to one of the vaginas. Posture and behaviour of the pig were normal. Cytogenetic analysis of blood lymphocytes revealed no numerical or gross structural anomalies. There have been no further piglets with developmental disorders in the same litter, in a second litter of the same parents and in other twelve litters by the same boar.

Keywords: pig, complex malformation, polymelia, partial duplication of urogenital tract, duplication of rectum, atresia ani, rectovaginal fistula.

Zusammenfassung

Angeborene Defekte wie Myofibrilläre Dysplasie (Spreizferkel), Nabel- und Leistenbrüche, Kryptorchismus, Zwitterbildungen und Atresia ani werden beim Schwein relativ häufig beobachtet. Daneben finden sich Entwicklungsstörungen wie Doppelbildungen, die extrem selten auftreten. Die vorliegende Arbeit beschreibt den Fall einer seltenen angeborenen Entwicklungsstörung mit Pygomelie, Duplicitas coli partialis et recti, Atresia ani et fistula rectogenitalis, Duplicitas corpori uteri, cervicis, vaginae et vulvae und Duplicitas vesicae, urethrae et renalis. Als plausibler ätiologischer Entstehungsmechanismus kann eine inäquale, partielle Zwillingsbildung angenommen werden. Das betroffene Schwein schien - abgesehen von den äußerlich erkennbaren Anzeichen gesund und unauffällig. Trotz Analatresie wurde normaler Kot aus einer der beiden Vaginas abgesetzt. Die zytogenetische Untersuchung ergab keine Hinweise auf numerische oder grobstrukturelle Abweichungen im Karyogramm. Im betroffenen Wurf, in einem zweiten Wurf derselben Eltern sowie in 12 Würfen aus Anpaarung desselben Ebers an andere Sauen traten keine Formen von Missbildungen auf.

Schlüsselworte: Schwein, Entwicklungsstörungen

Introduction

Developmental malformations occur relatively frequently in swine, especially in comparison to other domestic species. The main defects are myofibrillar dysplasia (splayleg), umbilical and inguinal hernias, cryptorchism, intersexes, and anal atresia (PRIESTER et al. 1970, MULLEY and EDWARDS 1984, EDWARDS and MULLEY 1999). The overall incidence for developmental defects in piglets in Germany was 2.07 % (THALLER et al. 1996). In a large survey on more than 60.000 piglets from 190 German Landrace boars and over 170.000 from Pietrain boars from artificial insemination stations in Bavaria, 1.79 % and 1.97 % of the Landrace and Pietrain progeny had defects, respectively (WILLEKE and PESCHKE 1992). Frequencies depend on breed and population and have been decreased throughout the last decades (BEISSNER et al. 2003a). A polygenic mode of inheritance seems to be most likely in many defects (BEISSNER et al. 2003b) and present research is focussing on the evaluation of the molecular background of such defects. But not all congenital defects are heritable. Such defects are collectively termed spontaneous defects if no environmental factors are considered to be involved as teratogenic agents.

Among congenital defects, disorders of the limbs are very common, while polydactyly is rather rare in the pig (MULLEY and EDWARDS 1984). Excessive phalanges may be due to disturbed morphogenesis. They may also be manifested as duplications of phalanges or by accessory limbs at different anatomical regions, e.g. the interposition of an excess phalanx between two normal phalanges (SZABO 1989). Accessory limbs can be found in the region of the cranium (cephalomelia), the epigastric area (epigastromelia), the perineum (perineomelia), the back (notomelia) or the pelvic region (polymelia). Cases have been found in calves (SCHÖNFELDER et al. 2003) and ewes (HIRAGA and DENNIS 1993), goats (RAMADAN et al. 1998), poultry (HOFFMANN 1968) and in humans (e.g. RIVERA et al. 1999) but they are extremely rare in swine. Only one case of Polymelia in a piglet has been described (HAMORI 1983). Accessory limbs are always smaller than normal limbs, with stiff joints and sparse muscles without innervation (POHLMEYER 1974).

Among congenital diseases of the urogenital system, partial or complete agenesis or metabolic disorders of the organs have been occasionally described (HOEFLIGER 1971). According to PRIESTER et al. (1970), pigs have a three times greater risk for urogenital defects than cattle or horses. Duplications of kidneys have not been described in pigs, but there have been sporadic cases of bladder duplication, which were often accompanied by a

duplication of the genital system (SZABO 1989). Two cervixes and two completely separate uterine horns have been described in pigs by NALBANDOV (1958).

Case description

We describe the case of a five months old female Landrace x Pietrain fattening pig. The pig has been healthy and inconspicuous in its herd, without clinical signs and with normal performance. Because of an accessory limb in the region of the pelvis (Fig. 1), the animal was transferred to the Department of Veterinary Clinical Sciences at the University of Giessen for detailed examination.

Clinical examination

Posture and conformation of the pig reflected a healthy appearance. The nutritional and the developmental status of the animal were excellent. Behaviour was undisturbed. The pig showed normal stance and movement. The skin was unblemished and pink. The hair was smooth and flat. Heart rate, breathing frequency and rectal temperature were 80/min., 20/min., and 38.6°C, respectively. There were no hints on disturbances of the respiratory and circulatory system. Although no anus was formed, defecation was normal in frequency, and took place via a fistula to one of the vaginas. There were two vulvas formed, both involved in urination. Neither vulval discharge nor dysuria, were visible. Urine was dimmish.

Serology

All serological traits investigated were within the physiological range (data not shown).

Macroscopy

The two accessory hind limbs (pygomelia) were fused up to the pastern joint, and a joint-like structure formed the articulation to the pelvis in the area of the symphysis (Fig. 1). The accessory limbs showed dactylogryposis. An anal orificium was not visible (atresia ani). Vulva and vagina were doubled. The distal part of the colon on a length of 1.4 m and the rectum were duplicated. Both large intestines opened to the cervix and vagina, respectively. There were two urinary bladders (Fig. 2), each with its own urether. The right kidney was dislocated to the entrance of the pelvis. The organ had the shape of a horseshoe kidney, a

duplication of the kidney. There were two uteri, each with one horn and one ovary. Lesions or malformations in other organs and tissue were not detectable.

Overall diagnosis:

1. Polymelia et dactylogryposis
2. Duplicitas coli partialis et recti
3. Atresia ani et fistula rectogenitalis
4. Duplicitas corpori uteri, cervicis, vaginae et vulvae
5. Duplicitas vesicae, urethrae et renalis

Cytogenetic investigation

Whole blood cultures were set up according to standard procedures (BARCH 1991). Metaphase preparation included hypotonic treatment (0.075 M KCl) at 37 °C for 18 min., fixation (3:1, methanol : acetic acid) and chromosome spreading using hot steam (HENEGARIU et al. 2001). Slides were stained with 5 % Giemsa (Merck, Darmstadt, Germany) in Sørensen buffer (pH 6.8).

Cytogenetic analysis revealed a species specific karyotype, exhibiting a female gonosomal complement (2n = 38, XX). No numerical or gross structural anomalies were detected in a total of 97 metaphases screened.

Discussion

We describe here a very rare complex of malformation. Besides congenital anomalies like splayleg, hernia inguinalis, hernia umbilicalis, myoclonia congenita and atresia ani, the singular malformations of the present case are themselves very infrequent and hardly described in the literature. There is just one example for polymelia, and a few examples for duplicitas uteri and duplicitas vesicae in the literature. The combination of malformations as desribed in the present paper has not been published before. Thus, the case would not play any role in practice. Similar cases might lead to problems at birth and to early death of the piglet due to atresia ani, even in females. In our case, the fistula between rectum and cervix/vagina was wide enough to allow defecation. However, the contamination of the

urogenital system with faeces resembles a high risk to develop serious infection and inflammation. Posture, conformation and the general condition of the pig were still excellent.

The reasons that lead to these combined developmental disorders are disputable. In only 13% of defects in pigs, the cause was known or believed to be heritable, or a known environmental or teratogenic agent was identified. The cause was unknown or classified as potentially heritable in 75% of the defects (HUSTON et al. 1978). Thus the majority of defects has multifactorial causes with complex interactions between a genetic liability and one or a number of environmental agents (FRASER 1959). The reaction of embryos and fetuses varies individually and according to their exact developmental stage. The most sensitive phase for developmental dearrangements is during organogenesis (day 13/14 to day 35), with days 14 to 25 bearing the highest risk (WRATHALL 1971).

Atresia ani is one of the most common defects in pigs, with incidences of 0.4 to 0.6 % depending on breed and population. The defect is generally accepted as heritable with varying modes of transmission. Defects of the female genital tract are common in pigs (EINARSSON and GUSTAFSSON 1970). The authors reported partial duplication of the vagina in 4 % of slaughter gilts in Sweden. In most cases the cause is unknown or suspected to be heritable.

The pathoanatomical features of the propositus are not associated with gross alterations of chromosomes detectable on the basis of classical cytogenetic techniques. This finding corresponds to the fact that as far as we are aware no reports exist, connecting malformations as those observed to chromosomal anomalies.

Because each of the particular malformations described in the propositus rarely occurs in pigs, it seems unlikely that all of them developed independently due to heritable factors by chance. Therefore we interpret the coexisting malformations as a result of a partial unequal twinning event. From that point of view the dysmorphy can be attributed to an autosit-parasite situation. It remains unclear, whether the propositus was monozygotic due to incomplete fission or dizygotic due to fusion. While most of conjoined twins are regarded as the consequence of incomplete fission, at least one case in man has been reported, where an autosite-parasite couple shared all parental alleles at different loci (LOGRONO et al. 1997). Notably in that case body axes were perpendicular to each other. In other cases of conjoined twins of various morphologic classes monozygosity has been demonstrated (e.g. in cattle [SCHULZE et al. 2006]). The phenotypic appearance of our propositus is not indicative of one or the other, nor was it analysed with regard to zygosity. So the pathoetiology remains speculative.

According to the farmer, no further congenital anomalies were observed in the originating herd, neither in the litter concerned, nor in a second litter of the same mating and in other twelve litters by the same boar.

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Figures

Fig. 1a (left)/b (right): Caudal view of the pig: pygomelia, short accessoric legs with dactylogryposis.

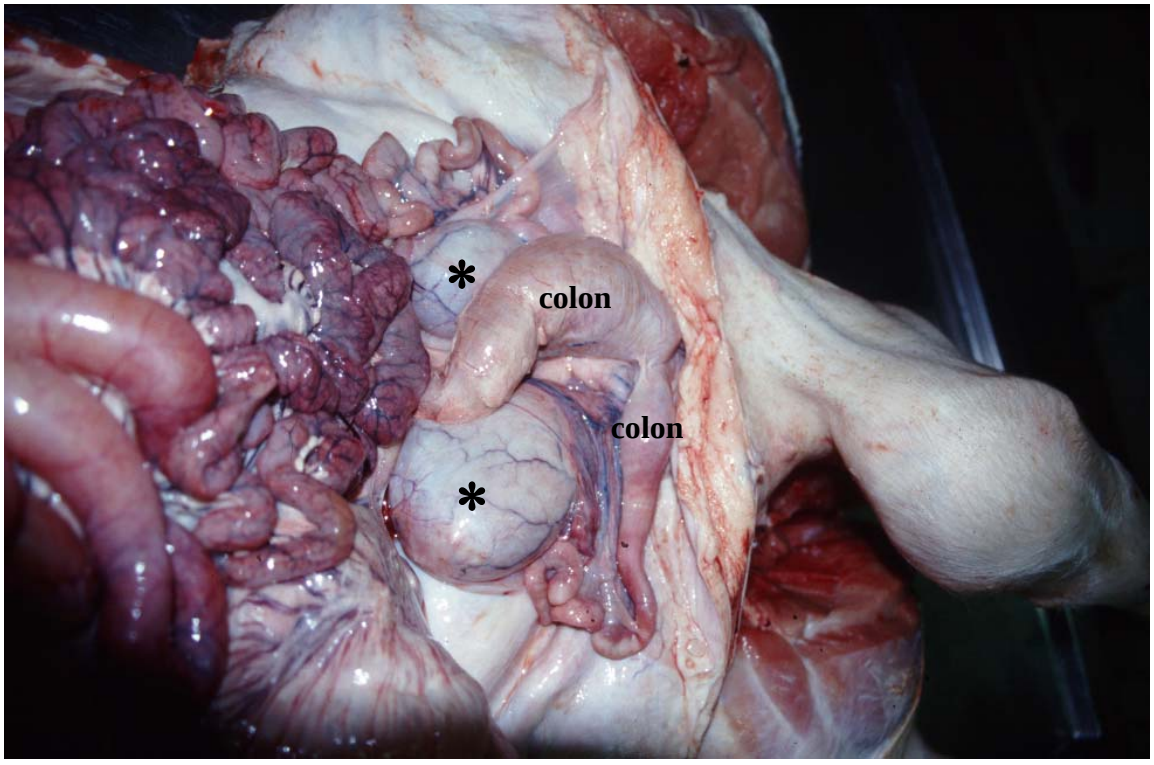

Fig. 2: Duplicitas vesicae (∗) and duplicitas coli.