

## HEREDITARY DISPROPORTIONATE DWARFISM IN PIGS RESEMBLING CHONDRODYSPLASIA

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**Introduction**

Hereditary disproportionate dwarfism due to developmental disorders in the long tubular bones has been described as a disease entity in several animal species and in man as well. To the authors knowledge a similar condition has never been demonstrated in the pig (1,2).

**Clinical Findings and Genetics**

The case in question occurred in a herd with 25 sows of Danish Landrace. Dwarf pigs were observed in three litters sired by the same boar. Knowledge about the pedigree of the mothers was not available. The boar had in addition sired more than five litters without any affected pigs.

The first signs of a developmental disorder were observed when the pigs were 3 weeks of age. During the suckling period the shortening of the limbs became more and more apparent. After weaning the condition was further aggravated, so that at last the limbs appeared extremely short. In addition, the attachment of the limbs to the body was very loose, and there was an abnormal mobility in the joints causing considerable locomotor disturbance. The resulting discomfort caused reduced weight gain, and at 3 months the affected pigs were only half as high as their normal littermates.

Some dwarfs and some normal littermates were purchased for experimental breeding, but because of locomotor problems (arthrosis) it was possible to bring up only one dwarf - a gilt - to the fertile age. No dwarfs were produced by matings between phenotypically normal relatives. The results of inbreeding experiments with the dwarf gilt and related normal boars are shown in the genealogical diagram, and consistent with a hypothesis of simple recessive inheritance.

Dominant inheritance, autosomal as well as sex linked, can be excluded, since the parents of affected animals in four litters were normal. Likewise, recessive sex linked inheritance can be excluded, as it requires that all affected offspring were males. According to a hypothesis of autosomal recessive inheritance a 3 to 1 segregation is to be expected in matings between non affected carriers. The observation of 7 pigs among 43 offspring from such matings, was in good agreement with the 10.7 affected pigs expected ( $\chi^2 = 1.74$ ,  $P > 0.10$ ). Both normal and dwarf pigs in the litters produced were unthrifty with a long hair coat and reduced weight gain. Many of the pigs died or had to be killed during the suckling period. Dead and killed pigs, except five pigs from 4 litters which died in the early neonatal period, were examined radiologically to verify the clinical diagnosis. A very low fertility was observed after few generations of inbreeding. No dwarfs were observed in litters born

by unrelated breeding pigs in the experimental herd during the investigation period.

**Radiologic Features**

Affected pigs were compared with normal pigs of the same age. Radiographically the defect appeared to involve the long tubular bones. The affected bones were shortened and relatively thicker than normal. The metaphyses were unusually wide and mushroom-shaped, overgrowing the epiphyses. The growth plates were less distinct than in normal pigs. Generally the fore limbs were shorter than the hind limbs. Secondary findings as arthrosis were increasing with the age of the pigs.

**Pathological Findings**

The gross pathological examination confirmed the radiological findings of a developmental disturbance of the long bones. Longitudinal sections through the bones showed uneven, thin growth plates. Secondary degenerative changes (arthrosis) were seen in varying degree. Histologically the growth plates showed an uneven, thin zone of chondrocytes with little tendency to normal columnation and vacuolization and a diminished formation of primary trabeculae.

**Discussion and Conclusion**

This investigation has revealed a condition with abnormal development of the long tubular bones, with the axial skeleton and the skull appearing normal. Similar conditions or conditions also involving other parts of the skeleton have previously been described, using terms as achondroplasia (3), pseudoachondroplastic dysplasia (4) and chondrodystrophy (5,6).

In the actual case the alterations were found exclusively in the extremities and were manifesting themselves by a depressed interstitial growth, while periosteal and perichondral growth appeared normal giving rise to the described malformed bones. Chondrodysplasia is suggested as an appropriate term for this condition. The observations in the herd of origin and in the experimental herd, together with the results of the experimental breedings leave no doubt that the disorder is heritable, probably with a simple autosomal recessive inheritance.

References (1) Huston, R. et al: Vet. Bull. 1978, 48, 645-675. (2) Bille, N. & N.C. Nielsen: Nord. Vet.-Med. 1977, 29, 128-136. (3) Andresen, E. et al: Nord. Vet.-Med. 1974, 26, 681-691. (4) Riser, W.H. et al: J. Amer. Vet. Med. Ass. 1980, 176, 335-341. (5) Baker, J.R. et al: Vet. Rec. 1979, 104, 450-453. (6) Jubb, K.V.F. & P.C. Kennedy: vol. 1, 1970.

Fig. 1. Genealogical diagram showing the relationship between the pigs in the breeding experiment. The symbols mean: square: a male, circle: a female, filled symbol: an affected individual, and shaded symbol: a known carrier.

