



HERD HEALTH MANAGEMENT & ECONOMY

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A NOVEL CONGENITAL SYNDROME WITH PALATOSCHISIS IN PIGLETS IN THE PROGENY OF A SINGLE PIETRAIN BOAR COULD BE EXPLAINED BY A *DE NOVO* MUTATION

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Introduction

Palatoschisis are oronasal fistulas, and often supposed to be inherited resulting in death.

Material & Methods

In a satellite farrowing farm of a sow pool system, piglets with palatoschisis were noticed in six out of thirty litters. A DNA-based examination was conducted, revealing that all affected litters were progeny of the same Pietrain boar. Thirteen affected piglets, males and females, had a unilaterale or bilaterale palatoschisis. All sows were from the same origin and had received the same feed. All littermates and their parents were sampled and the affected piglets were finally euthanized.

Results

A necropsy was performed, showing that all male animals were unilateral or bilateral cryptorchids, eleven animals of both sex had renal cysts and further different congenital defects were found single cases. A genetic aetiology due to a dominant inheritance with incomplete penetrance or a mosaic germline mutation was suspected as all recorded cases were indeed progeny of the same boar. Single nucleotide polymorphism genotyping data of 13 cases and their parents were used to map the defect in the porcine genome. Significant genetic linkage was obtained for three regions located on different chromosomes. Whole genome sequencing of an affected piglet and a normal fullsib revealed a total of 46 variants affecting the coding sequence of annotated genes which are present in heterozygous state in the affected piglet only.

Discussion & Conclusion

Systematic surveillance is needed to identify genetic defects as early as possible and to avoid the occurrence of further losses. The ultimate option in terms of prevention is to cull the boar in question once identified. For dominantly inherited disorders occurring within the offspring of single asymptomatic boar mosaicism could be suspected. The largely improved genomic resources in pigs facilitate the identification of the responsible boar and finally to unravel the responsible mutations.