DESCRIPTION FOR THE GENERAL PUBLIC

Hernia is a condition in which internal organs (most often intestines) protrude through an opening in the layers of muscle and connective tissue. The occurrence of umbilical hernia in pigs is a serious breeding problem affecting the animal's welfare and results in economic loss. It also has an effect on the sanitary quality of the carcass, sometimes even resulting in its disqualification from use. The frequency of hernia in different pig populations has been estimated to be approximately 2%. The heritability of this trait oscillates around 0.3, meaning that it seems to be justified to seek the genetic background of this disorder. Current knowledge of umbilical hernias in pigs is scarce, and mostly based on genome-wide association studies. Reports have shown a number of different genomic regions involved in hernia pathogenesis, but indicated few markers, which explained no more than 10% of the variability. To our best knowledge, until now no analysis of transcriptome profile and protein levels were undertaken in pigs suffering from umbilical hernia. We have thus **hypothesized** that the expression profiles in muscle and connective tissues localized near umbilicus are different in affected and unaffected animals. Moreover, we anticipate that the differences in transcript level may be caused by genetic factors such as polymorphisms and CpG methylation alternations in regulatory elements of these genes. We believe that this complex approach will result in the identification of markers associated with umbilical hernia in pigs.

The aim of this study is to perform a comprehensive genomic analysis of transcriptional profiling in two tissues types near the umbilicus. The selected after global RNA sequencing (RNA-seq) genes with different expression profile will be next studied using real-time PCR, Western blot and by immunostaining of the *in vitro* cultured cells derived from connective and muscle tissues of the two studied groups. Moreover, to find potential causes of transcriptional alternations, we will be searching for polymorphisms in regulatory regions by Sanger sequencing (in terms of single nucleotide polymorphisms and small insertions and deletions) and by droplet-digital PCR (for copy number variation polymorphisms). Simultaneously, we will also determine the DNA methylation level of the selected genes though bisulfite conversion followed by pyrosequencing.

It is expected that this study will allow us to identify genes with different expression profile as well as find potential genetic and epigenetic mechanisms regulating this expression that contribute to umbilical hernia pathogenesis in pigs. This may prove essential for breeders seeking to eliminate the carriers of undesired alleles from the gene pool, and thus reduce economic losses. Moreover, since the pig is considered a large animal model in the biomedical sciences, the results may be valuable for similar problems in other livestock species.