CHAPTER 5

CONCLUSION

In this study, functional candidate genes probably associated with scrotal hernia in pigs were characterized. *TAC1* was assigned to SSC9q12-q14 and *BAX* to SSC6q21. A recent genome-wide linkage analysis and an associated genomic mismatch scanning approach showed evidently associated regions for scrotal hernia on pig chromosomes 3, 6, 7, 12 and 15. Thus, because of the chromosomal assignment, TAC1 gene was excluded from further analyses as a candidate gene for the defect. Where as *BAX* proved to be a functional-positional candidate gene. The *BAX* containing PAC clone was isolated, identified and characterized by sub-cloning and sequencing. A contig of 10,741 bp was sequenced and compared of 5'-UTR and exons 1 to 4. The sequence shares 90% homology with the human gene. The length of the exons is highly conserved between humans and pig. A comparison of the porcine *BAX* coding region with mammalian orthologs revealed nucleotide sequence identities of 94% with *Bos taurus* and 93% with *Homo sapiens*. Microsatellite marker S0220 was detected in the 5'-flanking region. SNPs were detected by a direct comparative sequence analysis of affected and unaffected animals. No polymorphisms were found in the exonic regions. Two single-nucleotide polymorphisms (SNPs) were detected however in the intronic regions (SNPintron1:C8188T, SNPintron3:T8737A). PCR-RFLPs were established for genotyping (*Ear*I for C8188T and *BspH*I for T8737A) of 138 samples of difference breeds and origins. T8737A show significant allele frequency difference between pig breeds. Also C8188T showed significant allele frequency differences between normal and herniated pigs in the German population ($P \leq 0.05$). Thus, C8188T can be taken as a marker for further association analysis in large hernia populations.