

The porcine *TBC1D1* gene: mapping, SNP identification, and association study with meat, carcass and production traits in Italian Large White pigs

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ABSTRACT

TBC1D1 [TBC1 (tre-2/USP6, BUB2, cdc16) domain family, member 1] is a Rab-GTPase-activating related protein playing a key role in whole body glucose omeostasis. It is implicated in regulating the trafficking of GLUT4 storage vesicles to cell surface in response to insulin and AMPK-activating stimuli in skeletal muscle. In humans, mutations in the *TBC1D1* gene confer higher risk for obesity and type 2 diabetes. In mice, a functional mutation in this gene causes body weight reduction and lower adipose tissue deposition. According to these evidences and based on the biological functions of the *TBC1D1* protein, the porcine *TBC1D1* gene is a strong candidate for fat and lean meat deposition in pigs. Mining NCBI Trace Archive using BLASTN and the human *TBC1D1* cDNA sequence, we identified sequence traces of the homologous porcine gene. Primers were designed to amplify a fragment of the porcine gene encompassing parts of intron 1 and intron 2 and the intervening exon 2. Radiation hybrid mapping positioned the porcine *TBC1D1* gene on chromosome 8, confirming comparative mapping data with the human. Resequencing this fragment in a panel of 16 pigs of different breeds identified 5 single nucleotide polymorphisms. One of them was genotyped by high resolution melting analysis and/or by PCR-RFLP to evaluate allele frequencies in 6 different pig breeds. Furthermore, this polymorphism was analysed in 100 Italian Large White sib-tested pigs with extreme estimated breeding value (EBV; 50 with most positive and 50 with most negative values) for back fat thickness (BFT), 100 Large White sib-tested pigs with extreme EBV (50 with most positive and 50 with most negative values) for lean cuts (LC), and 239 sib-tested Italian Large White pigs not selected by any phenotypic or genotypic criteria and for which EBV for BFT, LC, average daily gain, ham weight (HW), and feed:gain ratio (FGR) were calculated. Fisher's exact test for allele frequency differences between the two groups of pigs of positive and negative EBV was significant ($P < 0.05$) for both BFT and LC selected animals. Association analysis between the *TBC1D1* genotypes and EBV in the 239 pigs obtained with the GLM procedure of SAS identified significant results ($P < 0.05$) for FGR, HW and LC. Together, these results might indicate an effect of this gene on muscle mass deposition in Italian Large White pigs.