Effects of the *CTSD* g.70G>A and *IGF2* intron3 g.3072G>A polymorphisms on meat production and carcass traits in pigs: evidences of the presence of additional QTN close to the imprinted *IGF2* region of chromosome 2

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Abstract n. 5254

60th Annual Meeting of the European Federation of Animal Science, Fira de Barcelona, Barcelona, Spain, August 24-27, 2009

INTRODUCTION

The insulin-like growth factor 2 (*IGF2*) intron3-g.3072G>A substitution is considered the causative mutation of the imprinted QTL affecting muscle mass and fat deposition reported on porcine chromosome 2 (Van Laere *et al.*, 2003). In the same chromosome region, we assigned by linkage mapping, the cathepsin D (*CTSD*) gene, a lysosomal proteinase, for which we previously identified a single nucleotide polymorphism in the 3'-untranslated region (g.70G>A) (Russo *et al.*, 2008).

The aim of this study was to clarify if the reported effects of the *CTSD* marker on several production traits in pigs were due to linkage disequilibrium with the *IGF2* mutation or to a direct effect of the *CTSD* polymorphism on these traits.

MATERIAL AND METHODS

ANIMALS

a. 270 Italian Large White sib-tested pigs.
b. 311 Italian Duroc sib-tested pigs.

TRAITS

Estimated Breeding Values (EBVs) for Average Daily Gain (ADG), Back Fat Thickness (BFT), Lean Cuts (LC), Ham Weight (HW) (only for the Italian Large White population) and Feed:Gain Ratio (FGR) were calculated.

DNA MARKERS

a.The *IGF2* intron3-g.3072G>A substitution, reported by Van Laere *et al.*, 2003, was analyzed by PCR-RFLP technique.
b.The mutation previously identified in *CTSD* gene was investigated as described in Russo *et al.*, 2008.

ASSOCIATION ANALYSIS

Association analysis between genotypes at the investigated loci and EBVs in these animals was performed using the GLM procedure of SAS. The analyses were carried out independently for the two groups of sib-tested Italian pigs.

RESULTS

In Italian Large White breed, highly significant results were obtained for the *IGF2* mutation for all the EBVs, with the most relevant one for lean content ($P = 2.2e^{-18}$). In Italian Duroc pigs, no association was found between this marker and the EBVs, probably due to the low number of heterozygous and g.3072GG animals. However the effects were in the same direction.

Association analysis between the *CTSD* polymorphism and ADG, LC, BFT and FGR in Italian Duroc pigs (considering only the *CTSD* g.70AG and g.70AA animals that were g.3072AA for the *IGF2* gene) showed significant results for all the EBVs ($P < 0.001$), according to the results we obtained in Italian Large White pigs reported in Russo *et al.*, 2008 (*Table 1*).

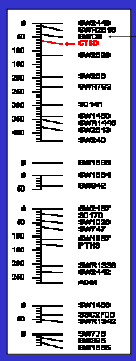
Table 1. Association analysis between the *CTSD* g.70G>A genotypes and estimated breeding values (EBVs) in Italian Large White and Italian Duroc pigs.

		Italian Large White			*Italian Duroc*		
Gene	**Traits**	**Genotypes**		***P****	**Genotypes**		***P****
CTSD		g.70GA (36)[§]	g.70AA (231)		g.70GA (42)[§]	g.70AA (253)	
		LSM (s.e)	LSM (s.e)		LSM (s.e)	LSM (s.e)	
	EBV ADG	+16.825 (4.070)	+36.169 (1.693)	*<0.0001*	+15.419 (4.486)	+32.996 (1.821)	**0.0008**
	EBV BFT	-0.220 (0.588)	-2.394 (0.2447)	*0.0007*	0.286 (0.601)	-2.196 (0.248)	**0.0002**
	EBV LC	+0.869 (0.283)	+2.188 (0.118)	*<0.0001*	+0.867 (0.308)	+2.158 (0.127)	**0.0001**
	EBV FGR	-0.064 (0.024)	-0.163 (0.010)	*0.0001*	-0.082 (0.029)	-0.176 (0.010)	**0.0008**

* Only significant ($P < 0.05$) associations are shown. § The number of animals for each genotypic class is indicated in parenthesis.

Differences between the *CTSD* genotype least square means for the EBVs in Italian Large White pigs carrying the *IGF2* intron3-g.3072GG genotype (no. of pigs 59) were significant for LC and HW ($P < 0.10$) (data not shown).

RH map of ***CTSD*** gene on **SSC2**



Radiation hybrid mapping indicated *Swc9*, a microsatellite in the 3'-UTR of porcine *IGF2* gene, as the closest marker to the *CTSD* gene.

CONCLUSIONS

We verified for the first time in the Italian Large White breed the effects of the IGF2 alleles on production traits.

In Italian Duroc pigs, for which no confounding effects could be attributed to the IGF2 intron3-g.3072G>A mutation, the CTSD g.70A allele is confirmed to be the favourable allele, showing the higher ADG, higher LC, lower BFT and best FGR.

These results suggest a separate effect of the CTSD g.70G>A substitution from the IGF2 intron3-g.3072G>A mutation, supporting the hypothesis of the existence of additional QTL in the IGF2 region of SSC2.

Marker assisted selection already applied using the IGF2 mutation could increase in efficiency adding information on the CTSD genotype.

Acknowledgements: We thank ANAS for providing samples and data. This work was supported by Italian MiPAAF SelMol project.