CHARACTERISATION OF QUANTITATIVE TRAIT LOCI FOR GROWTH AND MEAT QUALITY IN A BREED CROSS IN SWINE

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INTRODUCTION

Genome scans have enabled the detection genomic regions with quantitative trait loci (QTL) for economic traits. While most studies have focused on QTL with a Mendelian mode of expression, recent studies have detected QTL for which expression depends on parental origin, i.e. gametic imprinting. Knott *et al.* (1998) were the first to search for imprinted QTL in a genome scan. They inferred imprinting when effects differed significantly from Mendelian expression. Jeon *et al.* (1999) and Nezer *et al*. (1999) found paternal imprinting for muscularity in the *IGF*2 region of chromosome 2 in pigs. De Koning *et al*. (2000, 2001) modified the approach of Knott *et al.* (1998) and reported a large number of imprinted QTL for growth and meat quality traits in pigs. They inferred imprinting if the contribution of only one parental allele was significant. The purpose of this study was to further develop tests for imprinting and to characterize QTL for growth and meat quality traits in a Berkshire-Yorkshire cross family.

MATERIAL AND METHODS

The three-generation Berkshire-Yorkshire family described by Malek *et al.* (2001a,b) was used. The family consisted of 527 progeny from 8 F_1 boars and 26 F_1 gilts and was phenotyped for 40 traits and genotyped for 133 markers. An additional 35 markers were genotyped for the current analysis. The following subset of traits was used: average backfat thickness (BF), average daily gain (ADG) and loin eye area (LEA) to characterize growth and composition, average glycolytic potential (GP), average drip loss (DRIP), water holding capacity (WHC), loin pH at 24 (PH24) and 48 h (PH48), light reflectance (Minolta L) in the loin at 24 (L24) and 48 h (L48) as objective measures of meat quality, and color (COLOR), firmness (FIRM) and marbling (MARB), as subjective measures of quality.

Line cross least squares regression interval mapping (Haley *et al.* 1994) was used for QTL mapping. To differentiate between expression of paternal and maternal alleles, the model was parameterized following De Koning *et al.* (2000) and four reduced models were derived, as shown in Figure 1. All models were fitted at each 1 cM position and the decision tree in Figure 1 was used to identify QTL and to determine their mode of inheritance. Mendelian expression was used as the base model and imprinting was only declared if there was significant evidence that the effects of the paternal and maternal alleles at a given position were not equal.

Significance thresholds at the chromosome-wise level were derived for each test based on 10,000 data permutations. For tests of Full/Men (Figure 1), permutations were conducted by switching $P_{pat(j)}$ and $P_{mat(j)}$ with 50% probability within each individual to create data under the Mendelian model. For tests of Full/Pat, $P_{mat(j)}$ and $P_{d(j)}$ were shuffled across individuals to create data under paternal imprinting. $P_{pat(j)}$ and $P_{d(j)}$ were shuffled for the Full/Mat test.

Models fitted and tested at each 1 cM :

Full model [A]:

$$y_j = \mu + a_{pat} P_{pat(j)} + a_{mat} P_{mat(j)} + dP_{d(j)} + e_j$$

Null model: $a_{pat} = a_{mat} = d = 0$

$$y_j = \mu + e_j$$

Mendelian model: $a_{pat} = a_{mat} = ½\ a$

$$y_j = \mu + ½\ a\ (P_{pat(j)} + P_{mat(j)}) + dP_{d(j)} + e_j$$

Paternal imprinting model: $a_{mat} = d = 0$

$$y_j = \mu + a_{pat} P_{pat(j)} + e_j$$

Maternal imprinting model: $a_{pat} = d = 0$

$$y_j = \mu + a_{mat} P_{mat(j)} + e_j$$

[A] a_{pat}, a_{mat}, and d are paternal, maternal, and dominance effects at the QTL. $P_{pat(j)}$, $P_{mat(j)}$, and $P_{d(j)}$ are breed origin probability coefficients (De Koning *et al.* 2000).

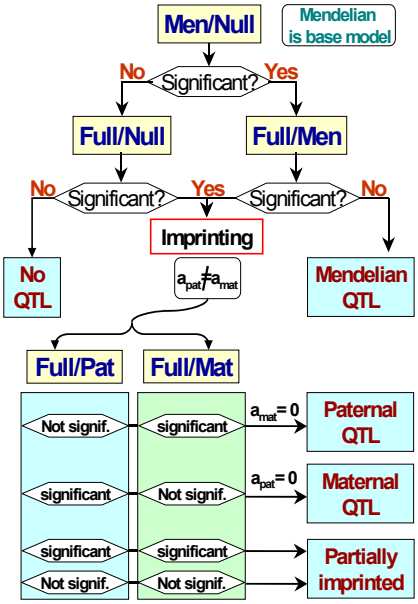

Figure 1. Models and decision tree of tests to detect and characterize QTL

RESULTS AND DISCUSSION

QTL detected at the 5% chromosome-wise level are shown in Tables 1 and 2.

Table 1. QTL detected for growth and composition and their mode of inheritance

SSC	Trait	Position (cM)	Test of alternative/null hypothesis					Genetic model
			Men/Null	Full/Null	Full/Men	Full/Pat	Full/Mat	
1	BF	73	*	**	*		**	Paternal [A]
1	LEA	32	**	**			**	Mendelian [A]
1	LEA	74		*			*	Paternal [A]
1	LEA	108	*	*				Mendelian
2	ADG	87	**	*		*	*	Partial
2	BF	8		*	*		*	Paternal [A]
2	LEA	5		**	*		**	Paternal [A]
4	ADG	120	*			*		Mendelian [A]
4	LEA	98	**	**			**	Mendelian [A]
5	BF	117	**	**		*		Mendelian [A]
7	BF	55	**	**		**		Mendelian [A]
8	ADG	48	*	*			*	Mendelian [A]
9	ADG	120	*					Mendelian
9	LEA	37	*				*	Mendelian [A]
10	LEA	85		**	**		*	Paternal [A]
13	BF	28	*					Mendelian
18	BF	5	*					Mendelian

* significant at ≤5% chromosome-wise level; ** significant at ≤1% chromosome-wise level;
[A] Imprinting detected using the test of De Koning *et al.* (2000)

Table 2. QTL detected for meat quality and their mode of inheritance

SSC	Trait	Position (cM)	Test of alternative/null hypothesis					Genetic model
			Men/Null	Full/Null	Full/Men	Full/Pat	Full/Mat	
1	DRIP	88	*	*				Mendelian
1	COLOR	102	*					Mendelian
1	PH48	95	*					Mendelian
1	MARB	34	*					Mendelian
1	MARB	57	**	**				Mendelian
2	DRIP	42	**	**			**	Mendelian [A]
2	DRIP	125	*	*			*	Mendelian [A]
2	FIRM	56	*			*		Mendelian [A]
2	FIRM	86	*					Mendelian
2	L24	76	*					Mendelian
2	L48	129	**	*				Mendelian
2	PH48	121		*	*	*		Maternal [A]
2	WHC	70	*					Mendelian
2	WHC	141	*					Mendelian
4	L48	146	*					Mendelian
5	DRIP	29		*			*	Paternal [A]
5	L24	120	*			*		Mendelian [A]
5	PH24	120	*	*		*		Mendelian [A]
5	L48	120	*			*		Mendelian [A]
5	PH48	80	*	*				Mendelian
5	PH48	105	*	*				Mendelian
5	WHC	120	**	*		*		Mendelian [A]
7	L48	82	*	*			*	Mendelian [A]
8	MARB	51	*					Mendelian
9	DRIP	95		*		*		Maternal [A]
9	L48	95		*		*		Maternal [A]
10	MARB	1		*		*		Maternal [A]
11	DRIP	0	*					Mendelian
11	GP	0	*					Mendelian
12	COLOR	60	*	*			*	Mendelian [A]
13	L48	81	*					Mendelian
13	WHC	45	*					Mendelian
15	DRIP	52	**	**		*		Mendelian [A]
15	GP	74	*	**		**		Mendelian [A]
15	PH24	79	**	**		**		Mendelian [A]
15	L48	70	**	**		*		Mendelian [A]
15	PH48	46	**	**		**		Mendelian [A]
17	GP	84	*					Mendelian
17	COLOR	86	**	**		**		Mendelian [A]
17	L48	85	**	**		*		Mendelian [A]
18	L24	28	**	*		**	**	Mendelian [A]

* significant at ≤5% chromosome-wise level; ** significant at ≤1% chromosome-wise level;
[A] Imprinting detected using the test of De Koning *et al.* (2000)

Results of a basic scan with the Mendelian model were similar to results of Malek *et al.* (2000a,b). Fitting the extended models and testing based on the decision tree of Figure 1, detected imprinted QTL on chromosomes 1, 2, 5, 9, and 10. Some of these were not detected

under the Mendelian model. Tests for imprinting following De Koning *et al.* (2000) were applied also, by inferring paternal imprinting if Full/Mat was significant and Full/Pat not, and *vice versa* for maternal imprinting. This resulted in a much larger number of imprinted QTL (Tables 1 and 2). Imprinted QTL detected by the decision tree of Figure 1 were also imprinted based on tests of De Koning *et al*. (2000), except for a QTL with partial imprinting for ADG on chromosome 2. The paternally imprinted QTL for BF and LEA at the distal end of chromosome (Table 2) confirm results of Nezer *et al*. (1999), Jeon *et al.* (1999), and De Koning *et al.* (2000). These QTL could be associated with *IGF2*, although *IGF2* is 5 to 10 cM proximal to our first marker on chromosome 2. Maternal imprinting was detected for DRIP and L48 on SSC9 (Table 1). De Koning *et al.* (2001) detected maternal imprinting for shear force and pH in a similar region.

CONCLUSIONS

Detection of imprinting should be based on tests against Mendelian expression. Such tests lead to substantially less evidence of imprinting than tests used by De Koning *et al.* (2000). Imprinting models can detect QTL that may remain undetected with Mendelian models. Several QTL for growth, composition, and meat quality traits were detected in a Berkshire-Yorkshire cross, but only few were found to be imprinted. Although imprinting results should be interpreted with caution because the number of F_1 parents was limited (De Koning *et al.*, 2001), several were confirmed by literature, in particular paternal imprinting near *IGF2*.

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