韓國家畜繁殖學會誌 25(4) : 295~304 (2001)
Korean J. Animal Reprod.

Detection of Imprinted Quantitative Traits Loci (QTL) for Reproductive and Growth Traits in Region of IGF Ⅱ Gene on Pig Chromosome 2

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ABSTRACT

Characterization of quantitative trait loci (QTL) was investigated in the experimental crosses between Berkshire and Yorkshire breed. A total of 525 F_2 progenies from 65 matting of F_1 parents were produced. Phenotypic measurements included average daily gain (ADG), average back fat thickness (ABF), and loin eye area (LEA). To identify the presence of QTL for reproductive performance, birth weight (BWT) and body weight at 16 days (16DAY) were included as indirect trait. QTL segregation was deduced using 8 markers assigned to chromosome 2 (SSC2). Quantitative trait locus analyses were performed using interval mapping by regression under line-cross model. Presence of imprinting was tested under the statistical model that separated the expression of paternally and maternally inherited alleles. To set the evidence of QTL presence, significance thresholds were derived by permutation following statistical tests, respectively. Genome scan revealed significant evidence for three quantitative trait loci (QTL) affecting growth and body compositions, of which two were identified to be QTL with imprinting expression mode near the IGF Ⅱ gene region. For average back fat thickness (ABF), a paternally expressed QTL was found on chromosome 2 (SSC2). A paternally expressed QTL affecting loin eye area (LEA) was found in the region of SSC2 where evidence of imprinted QTL was found for average back fat thickness (ABF). For average daily gain (ADG), QTL expressed with Mendelian mode was found on chromosome 2 (SS2). Also, QTL affecting average daily gain (ADG), was identified to be expressed with Mendelian express mode.

(Key words : QTL mapping, Imprinting, IGF Ⅱ, Reproductive trait, Swine)

Ⅰ. INTRODUCTION

The use of genetic marker has made it widely enable to dissect quantitative trait variation. Due to the availability of large number of polymorphic markers, it is now possible to scan a complete genome for genes affecting economic traits, so-called quantitative trait loci (QTL). And then QTL underlying the genetic variance of economic traits in the livestock are mapping in chromosomal regions. Saturated genetic marker maps are being used to map individual gene affecting quantitative traits. Accordingly, whole genome scans have derived as number of genomic regions containing quantitative trait loci and also provided with a better insight into the mode of inheritance for those traits. Recently, several studies showed that non-Mende-

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lian form of gene expression could be searched using the genome scans. And there was the number of reports that QTL, which exhibit no Mendelian inheritance, was found in pigs. As explicit example of non-Mendelian gene expression, imprinted gene near the IGF-2 locus on chromosome 2, which affects muscle mass and fat deposition in pigs, was identified (Nezer et al., 1999; Jeon et al., 999). Only a few reports have identified QTL for reproduction in swine. Rathje et al. (1997) reported a possible QTL on SSC8 associated with the ovulation rate. Hirooka et al. (2001) reported highly significant evidence for three QTL affecting teat number on SSC 2, 10 and 12 using a whole-genome scan, of which two were imprinted. Paternally expressed QTL were found on SSC 2 and 12. Also recently much attention has been paid for genomic imprinting in respect of identification of gene expression pattern or characteristics for medical application as well as animal breeding. In spite of import implication, there was only a few evidence of imprinted gene or its expression mode by limitation of approach difficulties in human and experimental population. In livestock, evidence for non-Mendelian gene expression was reported for one specific chromosomal region in sheep and pigs (Nezer et al., 1999; Jeon et al., 1999).

But de Koning et al. (2000) first presented result of a genome-wide systematical approach to detect imprinted regions for multifactorial traits. They detected gametic imprinting for QTL in swine F_2 cross based on a comparison of level of significance paternal and maternal imprinting effects against no-QTL model. But they did not test imprinting against Mendelian model, which is needed to identify deviations from Mendelian inheritance. It was clear evidence of imprinted gene that by crossing a domestic strain to its wild boar ancestor, the version of IGF-2 in the domestic strain contributes greatly to variance in muscle mass. If imprinting is common phenomena than previous thought, it is needed to investigate whether imprinting happen to be or not in previous published candidate gene for application of marker assisted selection. And it is also needed to include statistical test for imprinting in human and animal genetic result in both genome scan and evaluating candidate gene. It is needed to develop and apply tests that can be done to identify deviations between Mendelian and parental competition model. We conducted to develop and evaluate statistical models and test to identify evidence for imprinted QTL in chromosomal region that IGF Ⅱ gene was mapped.

Ⅱ. MATERIALS AND METHODS

1. Experimental Population and Data

Two Berkshire grand sires and nine Yorkshire grand dams were used to produce nine F_1 litters. From the F_1 litters, 8 boars and 28 gilts were chosen to produce F_2 animals. The QTL mapping resource population with 525 F_2 progenies was produced by swine breed cross of three generations. Resource population structure with F_2 of breed origin probabilities was showed (Fig. 1). A total of 525 F_2 progenies were genotyped for 8 polymorphic

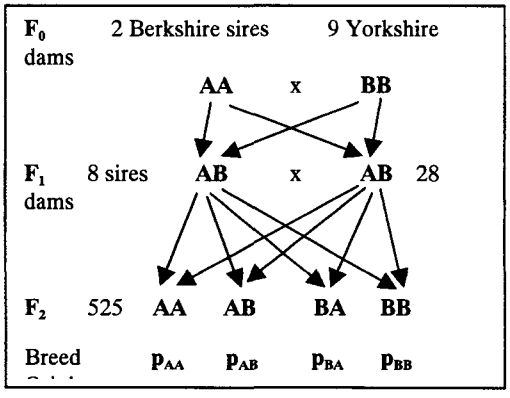


Fig. 1. Resource population used and outline of allele transmission pathway.

Table 1. Means and standard deviations for traits of interest measured on 525 F_2 animals and expected differences between breed means (Berkshire minus Yorkshire)[a]

Traits analyzed for QTL mapping	Mean	Std Dev	Berk-York[a]
Average daily gain (kg/day)	0.69	0.074	0.005
Average back fat (cm)	3.31	0.641	NA
Loin eye area (cm^2)	35.59	5.684	−5.548
Birth weight (kg)	1.55	0.325	NA[b]
Body weigh at 16 days (kg)	4.95	1.311	NA

[a]: Expected difference between breed means based on twice the difference observed in crossbreds in NPPC genetic evaluation program (Goodwin and Burroughs, 1995).

[b] NA: Not available

microsatellite markers for chromosome 2 (SSC2). Traits recorded for the purpose of the present paper were average back fat thickness (ABF), average daily gain (ADG), and loin eye area (LEA), birth weight (BWT) and body weight at 16 days (16 DAY). See Table 1 for a description of the traits.

2. Statistical Model

Statistical model for providing the evidence of imprinting and also identifying whether QTL seems to be paternally or maternally expressed in genome, the model for imprinting was reparameterized to enable a direct test for the contribution of the paternally and maternally inherited effect by de Korning et al. (2000). For every F_2 offspring, we inferred the probabilities of inheriting two alleles of first founder breed (P_{AA}), two alleles of alternative breed (P_{BB}) or one from each founder breed (P_{AB}, P_{BA}) at 1-cM intervals across the genome. P_{AB} is the probability that F_2 progeny can have the paternal allele originated from breed A and maternal allele from B. P_{BA} is the probability that F_2 progeny can also have the paternal allele originated from breed B and maternal allele from A. Also, breed origin probabilities of paternal and maternal alleles received by an F_2 progeny were determined at each chromosomal position based on marker data (Fig. 1). Breed origin probabilities can be used to derive conditional probability of additive effect ($P_a = P_{11} - P_{22}$), dominance effect ($P_d = P_{12} + P_{21}$), paternal contribution ($P_{pat} = [P_{11} + P_{12}] - [P_{22} + P_{21}]$), and maternal contribution ($P_{mat} = [P_{11} + P_{21}] - [P_{22} + P_{12}]$) coefficients. Under the traditional line cross approach, the expected performances of offspring that written in term of the additive (a) and dominance (d) contributions are estimated using the regression of the phenotypes on the P_a and P_d.

$$Y_j = m + a\ P_{aj} + d\ P_{dj} + e_j$$

Mendelian model (Mend)

However, when accounting for the grandparental origin of the alleles by using the multiple marker information, it is available to calculate probabilities of the two alleles in an offspring according to four possible genotypes in the F_2 generation. Additional extension model can be fitted with exclusive paternal or maternal expression using the conditional probabilities. To separate contribution of parents, we introduced the probabilities that individual inherited one founder breed allele from its father (P_{pat}) or from its mother (P_{mat}) (de Korning et al., 2000). A saturated full model (Full), which include conditional probabilities of paternal (P_{pat}), maternal (P_{mat}) and a dominance (P_d) contributions, was fitted at 1-cM intervals across the genome. The expected performances of offspring that written in term of

the paternal (a_{pat}) , maternal (a_{mat}) and dominance (d) contributions also were estimated using the regression of the phenotypes on the P_{pat}, P_{mat} and P_d.

$$Y_j = m + a_{pat}\ P_{pat(j)} + a_{mat}\ P_{mat(j)} + d\ P_{d(j)} + e_j$$

Full Imprinting model (Full)

$$Y_j = m + a_{pat}\ P_{pat(j)} + e_j$$

Paternal imprinting model (Pat)

$$Y_j = m + a_{mat}\ P_{mat(j)} + e_j$$

Maternal imprinting model (Mat)

Due to outbred cross that two types of heterozygotes can be distinguishable, fitting the imprinting genetic model is possible by tracing the four alleles segregation.

3. Statistical Test and Testing Procedure

Statistical test and testing procedure Maximum likelihood analysis was used for ordering marker loci relative to each other with the CRIMAP program (version 2.4) (Green et al., 1990; Malek et al., 2001). For QTL analysis, interval mapping procedure using least squares regression method was applied under line cross concept, where founder breed or lines are assume to be fitted for alternative alleles at the QTL affecting the traits of interest. The statistical model used included sex and slaughter date plus the covariable live weight. A single QTL was fitted in all cases by regressing on additive and dominance coefficients for the QTL at each putative position of the QTL (every 1 cM). Additive and dominance coefficients at a given position of the QTL were derived based on marker data following the procedure of Haley and Knott (1994). In order to estimate paternal and maternal contribution coefficients, we followed extension of traditional Mendelian expression model suggested by Knott et al. (1998) and de Korning (2000). To test QTL presence, we conducted some of different tests; Mendelian vs. No QTL model (Men/Null), full imprinting vs No QTL model (Full/Null), exclusive paternal vs No QTL model (Pat/Null) and maternal vs No QTL model (Mat/Null). And to test mode of inheritance of QTL, we also conducted other tests under full imprinting vs Mendelian model (Full/Men). This extended additional model including paternal and maternal component and dominance effect of putative QTL was tested against no QTL presence. If significant, best parental competition model (paternal or maternal) were inferred by testing full imprinting against Mendelian inheritance mode.

4. Derivation of Significance Thresholds

The method of permutation test outlined by Churchill and Doerge (1994) was used to determine significance thresholds for the F statistic to control Type I error rate at the chromosome-wise level. And to apply to hypotheses test between each alternative genetic model that proposed in this study, we conducted to do specialized permutation test. Permuted data sets were created by randomly shuffling marker data against phenotypic plus fixed effects data for the F_2 individuals. By each inferred genetic models, we intended to test; (1) Mendelian model vs no QTL, (2) Imprinting saturated model vs no QTL, (3) only paternally expressed model vs no QTL, (4) only paternally expressed model vs no QTL, permutation tests were carried out as same way according to tests. For test of imprinting vs. Mendelian model, permutated data set produced by randomly switching coefficients of paternal (P_{pat}) with that of maternal (P_{mat}) within individual with a 50% probability. Each permuted data set was then analyzed using the least squares regression interval mapping method and the maximum value of the F statistic was recorded. For multiple test thresholds, the maximum F statistics were recorded. A total of 20,000 permutated data sets were analyzed, resulting in 20,000 maximum F statistics per trait and every hypothesis test with different alternative mo-

del. Significance thresholds to control type I error rate at a level? on the chromosome-wise level were determined by ranking the maximum F statistics and determining the value of the F statistic that marked the $(1-\alpha)*100^{th}$ percentile.

Ⅲ. RESULTS AND DISCUSSION

Arithmetic mean and standard deviation of traits measured on the F_2 animals are listed in Table 1. Marker mapping results of chromosome 2 are presented in Table 2. The 8 markers genotyped in this study represent reasonable chromosomal coverage. Chromosome-wide significance thresholds (5 % and 1%) were derived by 20,000 replicates of permutation test, based on the each test model for between genetic models. Threshold of 5% chromosome-wide against the no QTL model on traits were range 5.0 to 5.2 for Mendelian model. Thresholds for chromosome-wide significance at 5% level in the test of Mendelian model correspond approximately to suggestive significance at the genome -wise level (de Koning et al., 1999; Lander and Kruglyak, 1995). For full imprinting, there is no deviation of threshold derived by permutation for each traits. And thresholds were range from 7.2 to 7.1 for paternal and 7.2 and 7.3 for maternal model. Thresholds of full imprinting model were range 7.1 to 7.7 against the Mendelian model. Individual chromosome significance at 5% level, as determined by the permutation test, differed slightly by trait, but more substantially by tests with specific statistical mode. Threshold differences between traits are due to differences in phenotypic distributions and random sampling (Spelman et al., 1996). Also thresholds were derived from permutation test of some kind of test model that was suggested for identification of best genetic inheritance mode of putative QTL. In particularly, threshold of critical value obtained under the paternally or maternally only expressed model against no QTL were showed very higher than any other those of genetic model tests. Also thresholds for test of maternal and paternal only expressed model are very similar. And then critical value derived on each paternal or maternal model may be allow to application for each test. Under the various genetic models, QTL analysis resulted in evidence for presence of QTL affecting reproductive and growth traits on SSC2 (Table 3). There was no significant QTL for birth weight and body weight at weaning (16day). De Koning et al. (2000) developed methods to detect imprinted QTL in a genome scan and found evidence for several traits in an F_2 swine breed cross. They compared the significance of paternal and maternal imprinting effects against a no-QTL model but did not test imprinting against a Mendelian inheritance model, which is needed to determine significant deviations from Mendelian inheritance. To develop the procedure for identifying the imprinting, we performed various test that can be suggested by combination of dissected genetic components (additive and dominance effect in Mendelian inheritance model, and paternal and maternal component and dominance effect in full imprinting model). It was possible to carry out five types of

Table 2. Markers used in the QTL mapping on chromosome 2

Marker	Position (cM)	Number of allele	IIC[a]
SW2623	0.0	5	0.90
SW2445	27.9	4	0.89
SW766	71.3	3	0.73
SW2157	86.3	6	0.89
SW1408	90.1	6	0.44
SW1844	111.6	3	0.72
SWR308	136.9	5	0.86
S0036	143.3	6	0.97

[a] Information content based on data for given marker only.

Table 3. QTL detection results for tests[b] by statistical model on growth and body composition traits

[a]Location		[b]Tests for presence of QTL				Tests for mode of inheritance
Trait	(cM)	Men	Full	Pat	Mat	Full/Men
ABF	7	3.43	5.16*	15.31**	2.54	8.54*
ADG	87	8.31**	5.53*	6.39	8.82*	1.69
LEA	6	4.45**	5.68*	15.11**	2.30	8.15*

[a] : Best position estimated on the most appropriate genetic model for the QTL.

[b] : Men : Mendelnkian vs no QTL, Full : Full imprinting vs no QTL, Pat : Parternal vs no QTL, Mat : Maternal vs no QTL, Full/Men; Full imprinting vs Mendelian.

* : Significant at the 5% chromosome-wise level by permutation test of inferred genetic model.

** : Significant at the 1% chromosome-wise level by permutation test of inferred genetic model.

hypothesis test. Two paternally expressed QTL, affecting respectively average back fat thickness (ADF) and loin eye area (LEA), map to the same region on SSC2. In this same region, it was said that many of significant evidence for imprinting QTL were found (de Koning, 2001; Hirooka, 2001; Nezer et al., 1999). Especially, it was identified that IGF-Ⅱ gene located in this region was expressed with paternal inheritance mode and can be a strong candidate gene for body composition traits (Jeon et al., 1999). Even though evidence of QTL presence was found from test of imprinting vs. no QTL model, there was no significant evidence of imprinted QTL. And then it may not be selected to have evidence of imprinted QTL (Table 3) for ADG. Thus, consideration of only tests against the null model (no QTL) would lead to wrong conclusion. In these results of test, we need additional tests for identifying the best inheritance mode of putative QTL for each trait. Testing full imprinting model against Mendelian model seems to be allowed to derive conclusion for ambiguous statistical inference. For ABF and LEA, test of full imprinting against Mendelian model showed putative evidence of imprinted QTL. From these some kind of tests, evidence of imprinted QTL for ABF and LEA can be inferred in distal location of SSC2. In this area, IGF-Ⅱ gene is previously mapped. Our result allows to conform findings imprinted QTL of Nezer et al. (1999) and Jeon et al. (1999). Table 4 shows

Table 4. Location and characterization of QTL affecting trait

Trait	Position(cM)	Genetic model[a]	Test statistics[b]	a(s.e.)[c]	d(s.e)
ABF	7	Paternal	15.3**	0.100(0.041)	
ADG	87	Mendelian	8.3**	0.015(0.000)	0.010(0.007)
LEA	6	Paternal	15.1**	−0.991(0.250)	

**: Significant at the 1% chromosome-wise level by permutation test including the inferred genetic model.

[a] : Genetic model that is most appropriate for QTL.

[b] : Test statistics for the inferred genetic model vs. the Ho of no QTL.

[c] : Estimated QTL effects for the inferred genetic model. The additive effect 'a' is expressed as the deviation of Berkshire allele and the dominance effect 'd' is expressed as the deviation of the heterozygous animals from the mean of the homozygotes.

the estimated effects for the significant QTL affecting reproductive and growth traits. For the QTL affecting loin eye area on SSC2, Yorkshire alleles were superior to Berkshire alleles.

To illustrate the presence of QTL for each trait, graphical comparison of results obtained under full imprinting, Mendelian, paternal and maternal model were showed in Fig. 2, 3 and 4. For ADG, peak point indicating presence of QTL under all test of imprinting and Mendelian model exceeded the threshold at the 5% chromosome-wide level. For ABF and LEA, we could not find presence of QTL that exceeded the threshold under Mendelian mode (Fig. 3, and 4). But peak point that exceeded the threshold at 5% chromosome-wide level, were showed for those trait under the test of imprinting model. Curves (Fig. 2, 3 and 4) represent F-statistics profile at every cM position under different genetic models against null model (no QTL). All tests conducted for genetic model (Mendelian, full imprinting, and parental and maternal expressed

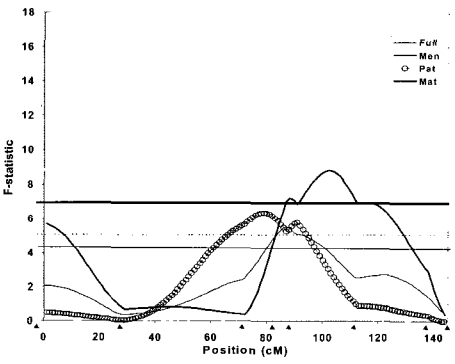


Fig. 2. Test statistics profiles for presence of QTL ADG in pig genome. Three horizontal lines are provided for 5% chromosome-wide threshold for Mendelian vs. no QTL (doted line), 5% chromosome-wide threshold for full imprinting vs. no QTL (thin line), 5% chromosome -wide threshold for paternal or maternal vs. no QTL model (solid line).

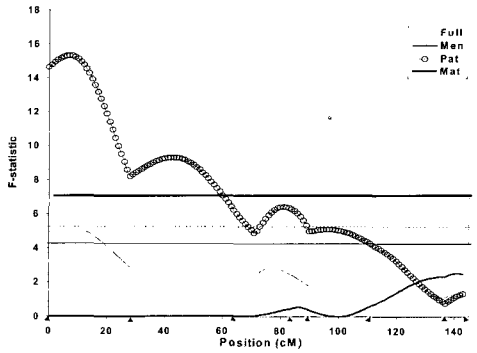


Fig. 3. Test statistics profiles for presence of QTL ABF in pig genome. Three horizontal lines are provided for 5% chromosome-wide threshold for Mendelian vs. no QTL (doted line), 5% chromosome-wide threshold for full imprinting vs. no QTL (thin line), 5% chromosome -wide threshold for paternal or maternal vs. no QTL model (solid line).

model) showed evidence on QTL at chromosome -wide significance level (5%) based on the permu-

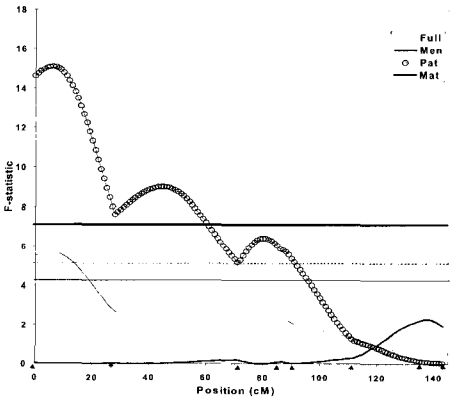
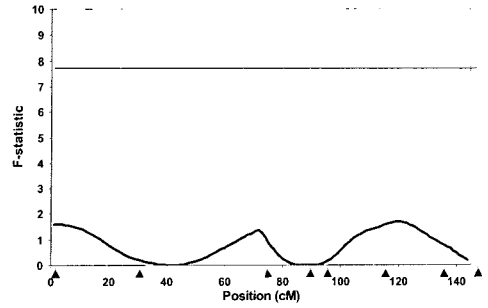
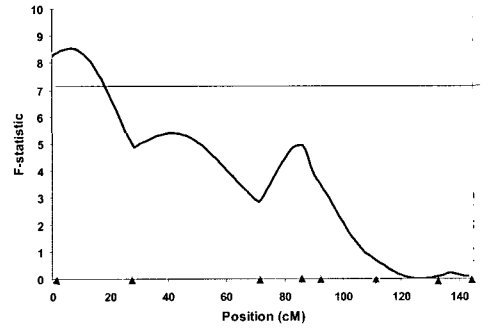


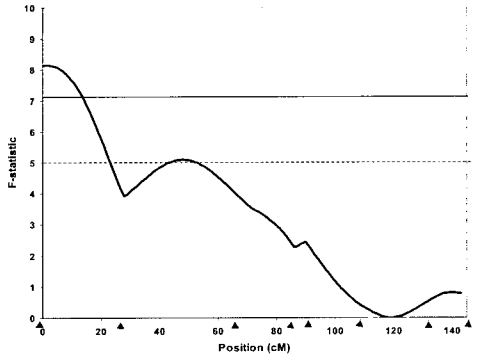
Fig. 4. Test statistics profiles for presence of QTL LEA in pig genome. Three horizontal lines are provided for 5% chromosome-wide threshold for Mendelian vs. no QTL (doted line), 5% chromosome-wide threshold for full imprinting vs. no QTL (thin line), 5% chromosome -wide threshold for paternal or maternal vs. no QTL model (solid line).



(a) ADG



(b) ABF



(c) LEA

Fig. 5. Test statistics profiles for mode of QTL inheritance a) ADG, b) ABF, and c) LEA in pig genome. Three horizontal lines are provided for 5% chromosome-wide threshold for full imprinting vs. Mendelian (solid line).

tation test by trait. And another graphical profiles were present for confirming the test for mode of inheritance (Fig. 5). For ADG (a), peak point indicating imprinting inheritance mode of QTL, did not exceed threshold provided by permutation test under imprinting vs. Mendelian model. For ABF (b) and LEA (c), peak points that exceeded threshold were showed in Fig. 5. Fig. 5 illustrated how the best genetic model was inferred from analysis as clue that reveal QTL inheritance mode.

To derived imprinting test threshold under the various genetic models, specialized permutation test were conducted using real phenotypic data. Especially, only test against the no QTL model can lead to the wrong conclusion. In order to identify imprinted QTL, it seems to be needed to do the two steps of statistical procedure. Firstly, test for presence of QTL against no QTL model would be previously conducted for every trait. Secondly, for evidence of imprinting QTL expression mode, comparison of full imprinting model against Mendelian model should be tried. Also, imprinting model can reveal QTL not detected by Mendelian model. Finding of paternal imprinted QTL with effect on back fat and muscularity in this study were showed compatible trends with previous finding (Jeon et al., 1999; Nezer et al., 1999).

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요 약

돼지 염색체상의 IGF Ⅱ 유전자 인접 부위에서 번식 및 성장형질에 연관된 Imprinting 양적형질 유전자 좌위(QTL)의 탐색

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양적형질 유전자 좌위 (QTL)의 탐색과 이들의 발현 양상 규명을 위해 Berkshire종과 Yorkshire종 간의 교배를 통해 생산된 F_2 실험집단에서 regression interval mapping이 이루어졌다. 모두 525마리의 F_2 자손들에서 일당 증체량, 평균 등지방 두께, 배장근 단면적이 표현형으로 조사되어 분석에 이용되었으며 모돈의 번식능력에 관련된 QTL 존재 여부 추정을 위해 간접 형질로 인정되고 있는 생시체중과 이유시 체중을 분석에 포함하였다. 양적형질의 분리 여부를 추론하기 위하여 돼지의 2번 염색체에서 8종의 microsatellite 표지인자가 선택되어 유전자형이 조사되었다. 각각의 유전적 모델에서 산출된 통계량으로부터 QTL 존재 여부와 특정 QTL 발현 양상에 대한 여부를 나타낼 수 있는 인정되는 수준의 type I 오차율을 제어할 수 있는 임계값 (threshold)을 permutation test에 의해 제시하였다. QTL의 존재와 그 QTL의 Imprinting 여부는 부계와 모계를 통해 원가계 1세대의 대립유전자가 전달되는 과정에서 발현되는 특성을 분리시키는 통계적 모형을 설정하여 검정 통계량을 산출하였다. 분석에 이용된 3가지 형질과 연관된 3종류의 QTL 존재 가능성을 돼지의 2번 염색체에서 확인하였으며, 이들 중 평균 등지방 두께와 배장근 단면적에 각각 영향을 미칠 것으로 추론된 2종류의 QTL 발현은 정상적인 Mendelian 유전양식을 따르지 않고 imprinting된다는 증거를 얻어냈다. 또한 이들 imprinting되는 QTL은 이미 imprinting 표현 양식을 가진다고 알려진 IGF Ⅱ 유전자의 위치와 거의 동일한 염색체상의 지점에서 부계로 전달되는 QTL만이 발현되는 특징을 보이는 것으로 밝혀졌다. 한편 Mendelian 모형과 imprinting 모형 모두에서 유의적인 임계값 이상을 보이는 검정 통계량이 산출된 일당 증체량 연관 QTL은 두 모형 간의 적정성 분석을 위한 검정을 통해 Mendelian 양식을 따른 것으로 최종 확인되었다.

(접수일자 : 2001. 9. 30. / 채택일자 : 2001. 10. 23.)