Appraising genetic resources in pigs

Chris Moran

Department of Animal Science, University of Sydney, NSW 2006, Australia.

Introduction

Molecular genetics continues to provide increasingly powerful tools for sophisticated genetic studies in domestic animals. These tools permit determination of breed origins and analyses of evolutionary relationships. Molecular genetic tools also permit sensitive analyses of gene expression. Large scale EST (Expressed Sequence Tag) projects in the pig in the United States and Europe are providing the resources for development of expression arrays, containing potentially all genes in the pig. These arrays will enable identification of genes responding to important environmental stimuli such as disease and nutrition and management and which by inference have an important role in production and health. The tools of molecular biology also are enabling the identification by mapping of the genes responsible for a substantial component of economically important variation in domestic species such as the pig. Rothschild and Soller (1999), in a previous contribution to this symposium, have discussed the candidate gene strategy for identifying molecular variants at gene loci controlling variation in economically important performance in domestic animals. This talk will address a number of related issues beginning with the question of measurement, conservation and protection of genetic resources in the pig, so crucial in the long run to providing the raw materials for improvement of domestic animals. Without a sensible strategy for conserving genetic variation, it is obvious that there will be nothing to detect in gene mapping or candidate gene studies and that genetic improvement of pigs will not be possible. Interestingly, the results of gene and quantitative trait locus (QTL) mapping studies have actually provided real and quantifiable incentives for conservation of porcine genetic resources, especially for breeds quite inferior in relation to current production standards.

In Australia, we do not have the wide range of pig breeds available in

Asia and Europe, which could potentially contribute to long term improvement of pig productivity and health. Nevertheless we have embarked on QTL studies focussing on current commercial populations and have had some success in recognising QTL within these available resources. The results of these QTL studies will be briefly discussed.

Pig Breed Diversity Studies

Various methods and molecular tools are available for assessment of breed diversity and relationships. Since the taxa involved in domestic breed studies are very closely related in evolutionary terms, it is important to use rapidly evolving marker systems to assess their relationships.

Microsatellite markers for biodiversity

Microsatellite loci experience very high mutation rates, making them very suitable for assessment of close relationships, as well as providing very good markers of intra-breed diversity. FAO and ISAG have jointly recommended standard panels of microsatellite markers for measurement of domestic animal diversity in most economically important species (http://dad.fao.org/). These have been applied to the study of breed relationships in pigs by several laboratories including Asian pig breeds (Li, Chen, Moran, Fan, Zhao and Peng, in press). Analysis of very large sets of microsatellite data in biodiversity studies provides a particular challenge. The traditional approach has been to collapse all the data into a single distance measure and then use this to attempt to determine the evolutionary relationships of the populations and breeds under study. It is obvious that compression of data from 27 microsatellite loci, each with 10 to 20 alleles, into a single genetic distance measure, which does not accurately reflect the actual genotypic differences or variation in the precision of gene frequency estimates, loses a large amount of information. Improved methods of analysis are required to get the full benefit of these data sets. Other problems include the possible or actual existence of introgression and migration between populations, which violate the assumptions of most phylogenetic analyses.

Table1. Matrix of Nei's (1978) unbiased genetic distance among four Chinese pig breeds and an Australian commercial population (From Li et al., in press)

	EH	TC	QP	WH	AC
EH	***				
TC	0.739	***			
QP	0.564	0.475	***		
WH	1.046	0.832	0.812	***	
AC	0.857	0.897	1.029	1.438	***

Erhualian (EH), Tongcheng (TC), Qingping (QP), Wannanhua (WH), Australian commercial population (AC)
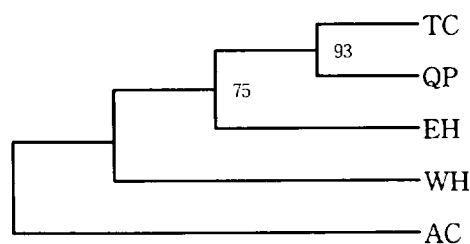

Figure 1. UPGMA dendrogram of the five breeds.

Based on Nei's unbiased genetic distances from Table 1. TC, QP and WH are traditionally classified as Central China type pigs, whereas EH is classified as a Lower-Changjiang River Basin Type. The microsatellite- derived dendrogram is inconsistent with this traditional classification. The digits at nodes are the percent occurrence in 1000 bootstrap replicates. (From Li et al., in press)

Mitochondrial D-loop sequence

Another rapidly evolving system, suitable for analysing relationships of recently diverged taxa, is the mitochondrial D-loop system. The DNA sequences within and near the D-loop evolve about 10 times faster than nuclear genes, due to higher mutation rate and also smaller effective population size for mitochondrial genes. Such sequences have been used for analysing relationships in domestic animals including horses (Kim et al., 1999). mtDNA D-loop sequence has also been used to analyse the relationships of pigs, including Asian Wild Boar and their domestic relatives (Okumura et al. 1996). Giuffra et al. (2000) have analysed mtDNA D-loop and mitochondrial cytochrome B sequence data on a limited set of European and Asian breeds. However, the most comprehensive study, using a large sample of Asian and European breeds, is that of Kim, Lee, Li, Zhang, Lee, Gongora and Moran (submitted). 1036 bp of mtD-loop sequence, excluding the D-loop repeats, was analysed in twenty-nine breeds or populations of pigs. These comprised seventeen newly generated sequences and twelve published sequences. Sequences were analysed from eight European breed animals, including European Wild Boar and

twenty Asian-breed animals, including Japanese Wild Boar and a single feral pig of unknown origin from Australia. Results of this and the Giuffra et al. (2000) study are discussed below

Independent domestication of Asian and European Breeds

The results of Kim et al. (submitted) show clearly that Asian and European clades can be unequivocally distinguished. Furthermore the lowest branching member in the Asian clade is the Japanese Wild Boar and the lowest branching member in the European clade is the European Wild Boar. This strongly implies that Asian domestic pigs were domesticated from an Asian Wild Boar ancestor independently of European domestic pigs, which were domesticated from a European Wild Boar ancestor. A similar conclusion was drawn by Giuffra et al. (2000) on a much smaller samples of European and Asian breeds, but confirmed by analysis of both types of mitochondrial sequence data and also by analysis of MC1R, TYR and GPI pseudogene sequences. Giuffra et al. (2000) concluded that the time since divergence of the ancestors of European and Asian breeds is between 500,000 and about 2.7 million years before present

Domestication of pigs is estimated to have occurred about 9,000 years before present. Thus it is clear that that the common ancestors of Asian domestic pigs and European domestic pigs are much further back in time than was previously thought. Kim et al. (submitted) estimate a divergence time between Asian and European lineages of 57,000 years BP, but up to 3 million years BP based on the peccary outgroup used in their study.

Interestingly, similar studies in other domestic species have provided evidence of separate domestication events in Asia and Europe. Loftus et al. (1994) provided evidence of two independent domestications of cattle. Hiendleder et al. (1998) showed that distinct Asian and European clades existed in sheep. Using mitochondrial sequence, Vila et al. (1997) invoked multiple independent domestications of dogs.

Introgression

Giuffra et al. (2000) noted that one of their Large White mt D-loop sequence clustered with Asian breed sequences and the other clustered with the European breeds. This implied that some Large White animals have inherited Asian mitochondrial genomes, presumably introduced into the breed early in its foundation. Further, they

found that Asian mtDNA haplotypes could be found in Large White, Landrace and Duroc pigs, providing evidence of recent introgression of Asian genes probably in the last 200 years. Kim et al. (submitted) found also that their Berkshire and Large White sequences clustered with Asian pig mitochondrial sequence. Jones (1998) cited historical evidence that the Chinese breeds contributed significantly to the development of the Berkshire, and the mtDNA data fully support this. Thus the mitochondrial sequences have provided evidence of the historical contribution of Asian pigs to the development of important European breeds several centuries ago.

Two hundred or more years ago, European animal breeders were exploiting newly imported germplasm from Asia and incorporated some of this into the foundation stocks of newly emerging breeds which have subsequently domi-nated pig production in Europe, America and Australia. The dominance of these "European" breeds due to highly effective selective breeding programs is such that in Asia the reverse phenomenon to that occurring in Europe two or three centuries ago is now happening. Asian animal breeders and producers are replacing traditional Asian breeds with high performance European breeds. Even where attempts

have been made to halt or reverse this process, as in the Cheju native breed in Korea, European mitochondrial sequence has been maintained in some animals despite recovery of a phenotypically "Cheju-like" population derived from hybrids (Kim et al., submitted).

Feral pigs-repository of biodiversity

In remote and inaccessible parts of Australia, there are populations of feral animals, derived from escaped domestic stock. Feral populations of horses, cattle, goats, camels or pigs are found, sometimes reaching very large numbers. In general, these animals are considered as pests, as they can attack and damage crops and as they sometimes provide a reservoir of disease and a potential route of disease transmission to domestic stock. Occasionally these animals provide unexpected reservoirs of useful genes. For example, about 25 years ago it was discovered that some feral goats in Australia produced relatively large amounts of high quality cashmere and a small Australian cashmere industry was established, with foundation breeding stock collected from the wild. Feral pigs are very widespread throughout Australia from the tropical

north to the temperate south and even on some islands around the coast of Australia. Their only benefit is providing sport for hunters and a very small export market for "game" meat.

Mitochondrial DNA sequence was obtained from animals derived from a feral pig population, descended from two animals released on Kangaroo Island in South Australia in 1803 (Cooper, 1954). Kim et al. (submitted) included these animals, from the highly inbred Westran line produced in Sydney for transplantation research, as additional putatively "European samples". However, phylogenetic analysis showed that their mitochondrial DNA sequence clustered with Asian pigs, not with European pigs as expected. The sequence did not cluster closely with either the Large White or Berkshire sequences within the Asian clade, suggesting that the Asian mitochondrial sequence was not derived from these sources. This suggests that pigs imported from Asia may have contributed to the feral pigs found in Australia. Feral pigs in Australia may not be predominantly of European-breed origin, as most people intuitively suspect. These Asian origin animals may have come directly from Asia or possibly from Polynesian sources in the early days of colonisation of Australia. There are historical reports of the release in tropical northern Australia of pigs from Timor in the early 19th century, but so far, no reports have been found of Asian pigs being introduced into southern parts of Australia. Determining the origin of the Westran pigs remains an interesting historical and genetical puzzle.

However, it is clear that the feral pigs in Australia provide an untapped and uncharacterised repository of porcine biodiversity. By analogy with the cashmere-producing feral goats, perhaps feral pigs in Australia may have some unsuspected but valuable features. It is planned in future to systematically analyse mitochondrial DNA sequences from samples of feral pigs from as many regions as possible in Australia to get a better picture of this diversity and to determine whether it is acting as an unplanned and unmaintained but valuable germplasm bank.

Conservation

Porcine diversity studies are very interesting and provide fascinating insights into the relationships and evolution of pigs. However, the ultimate justification for performing biodiversity analyses is to determine priorities for conservation of genetic

resources. Given that there is now very good evidence for separate domestication of pigs in Asia and Europe, it is clear that strong independent conservation efforts must be applied to Asian pigs. A fundamental principle of conservation is that effort to preserve should be proportional to distinctiveness of the populations. For example, in a group of endangered populations or breeds, most effort should be applied to preserving the most distinct population and less effort should be spent on groups of more similar populations. Loss of the most distinct and dissimilar population will lead to a much greater depletion of genetic resources than the loss of a population or breed with close relatives. Weitzman (1992, 1993) presents a detailed discussion of the principle of using diversity theory for prioritising conservation efforts.

Given the strong dissimilarity of Asian and European pigs, their independent domestication and long period of separate evolution dating from the much more distant common ancestor of European and Asian Wild Boar, strong priority must be given to conservation of Asian pig genetic resources. In the face of increasing ii. national pressure to use mainly high performing European breeds and their derivatives and the loss of Asian

native breeds, this priority is becoming critical.

Cryptic QTLs provide a rational basis for rare breed conservation

A frequently asked and very sensible question is "Why bother to conserve a very poorly performing breed, if it's performance is very substantially lower than current elite commercial stocks?" Any attempt to use such a breed would drag the performance of the crossbred population so severely downward as to make its utilisation in crossing or introgression apparently very undesirable. The standard answer to this question has always been along the vague and general lines that these non-competitive populations might contain hidden genetic resources valuable in the face of a new environmental challenge – the more genetically distinct the line, the greater the chance it will contain a valuable hidden resource. A frequently cited scenario is the arrival of a new disease challenge to which the homogeneous elite stocks are particularly susceptible, leading to potential catastrophe. If only the lowly performing breeds had been conserved, they could have been screened for resistance. Any resistance genes found could then have been introgressed

into the elite stocks to meet the new disease or environmental challenge. Of course, this scenario does have strong validity, supported by numerous examples in plant breeding where a novel plant pathogen has arisen and devastated homogeneous high performance lines of plants. Wild relatives and lowly performing lines of plants have provided the necessary genes for resistance to "revitalise" the susceptible highly productive line. However, in animals, we now have for the first time direct evidence for the existence of valuable genes and genetic variants in lowly performing stocks, hidden behind a screen of complementary unfavourable alleles at other loci. Gene mapping has provided us with the first direct evidence for this and many of the first and best examples in domestic animals come from pigs.

Most of the initial QTL mapping resource families in Europe, Japan and the United States were made by crossing lines of pigs differing as much as possible for traits of economic importance. It was argued that these lines were more likely to differ by QTL alleles of relatively large effect and that these QTL variants could be detected in resource populations of reasonable size. Thus crosses were made between breeds like Pietrain, highly muscular, low in fat and with high growth rates and excellent food conversion efficiency, and breeds like Meishan, which excel in reproductive performance but are extremely poor in every other regard, particularly fatness. While this experimental design was aimed at maximising the chance of detecting any QTL at all, it was also a very good design for detecting the existence of cryptic QTL alleles.

What are cryptic QTL alleles? This term has been used to describe favourable alleles present in an otherwise very unfavourable genetic background. It has been attributed to Professor Morris Soller (Weller, pers. comm.) and was first used in the literature by Weller, Soller and Brody (1989). Plant breeders also use the term "transgressive segregation" to describe the inheritance of such alleles in crosses. DeVincente and Tanksley (1993) provided the first example of the use of gene mapping in plants to detect transgressive segregation in tomatoes. They found that 36% of the QTL detected in a cross between cultivated tomatoes and a wild relative were transgressive. This means that for over a third of the QTL detected, the wild parent had the most favourable allele for a range of agronomic and quality traits. The results supported the claim by Miller and Tanksley (1990) that 95% of useful

genetic variation in tomatoes is found in wild relatives, none of which have any direct utility as they produce small or inedible fruit. Useful transgressive alleles derived from wild relative have been found in many species of plants including rice (Xiao et al., 1998). However, it is important to note that transgressive segregation is not only observed when crosses are made between lines showing large differences in performance. In maize for example, Stuber, Edwards and Wendel (1987) have observed transgressive segregation in intraspecific crosses where there were not extreme differences between the performances of the parents. Tanksley and McCouch (1997) have emphasised the value to the plant breeder of a germplasm bank, especially in conjunction with a genetic map allowing the recognition and exploitation of cryptic variation. This is a message which animal breeders should heed.

In animals, the occurrence of a QTL allele for leanness in a very fat breed would provide a good example of a cryptic QTL. If these favourable alleles could be extracted from the unfavourable genetic background in which they currently exist, the introgressed allele could be exploited in the genetic background of the highly performing breed. Fortunately the same genetic

markers used to recognise these cryptic QTL alleles in gene mapping studies could be used for marker-assisted introgression into the genetic background of the high performing breed. Thus gene mapping has not only provided a mechanism for recognition of hidden genetic merit within an unfavourable genetic background, but has also provided the tools to enable its capture and exploitation.

One of the first examples of cryptic QTLs in domestic animals was found in a study of pigs in the mid 1990s. A research group at Hohenheim University analysed an F2 resource population constructed using the very lean Pietrain and the very fat Meishan breeds. Initially they scanned chromosome 7 using 10 microsatellite markers. It was found that fatness, meat quality, stress resistance and carcass composition traits were significantly influenced by QTLs on chromosome 7, each accounting for 3.5% to 14.2% of the phenotypic variance. The QTLs were mapped in a region spanning the MHC. Contrary to what was predicted from parental phenotype, the Meishan QTL alleles were associated with a leaner carcass than Pietrain alleles. The results have recently been reported (Geldermann et al., 1999). Many other studies have subsequently verified the presence of the cryptic QTL for

fatness in other Meishan by European breed crosses

Probably the best systematic dissection of QTL, including cryptic QTL, contributing to differences between extremely different lines of animals is that of Knott et al. (1998), investigating the loci responsible for the very large difference in small intestine length between Large White pigs and European Wild Boar. Large White pigs have intestines measuring more than 21 metres in length, over four metres longer than the intestines of Wild Boar. Intestine length is positively correlated with growth and is believed to influence food conversion efficiency.

Table 2. QTL alleles contributing to the difference in intestine length between Large White and European Wild Boar. The effect of the Wild Boar allele is shown(From Knott et al., 1998)

Chromosome	Additive effect(cm)	Dominance effect(cm)
4	-63.6	+25.3
5	-46.5	-23.1
8	-39.7	-52.9
12	-6.8	-11.7
14	-18.6	-88.1
16	-57.0	+30.9
17	+54.5	+7.7

Using an F2 QTL mapping resource, Knott et al. (1998) were able to identify 7 significant QTL on different chromosomes (Table 2), explaining a large proportion of the difference between Large White and Wild Boar. As expected, most of the Wild Boar alleles had a reducing effect on intestine length. However, the QTL on chromosome 17 is clearly a cryptic QTL, since the Wild Boar allele has a large increasing effect on intestine length. The cumulative effect of the decreasing alleles in Wild Boar is -232.2 cm, but when the cryptic allele is taken into account, the cumulative additive effect is only -177.7 cm. If the Wild Boar chromosome 7 could be introgressed into the Large White breed, the intestine length would be expected to increase by over one metre. It is not clear that there would be a practical benefit from such an exercise but it does demonstrate that beneficial alleles of large effect can be hidden in otherwise very unpromising genetic backgrounds. If we allow such "invisible" genetic resources to slip away by extinction of rare and unproductive breeds, the consequences in the long run could be severe. It is obvious also that different poorly performing breeds are very likely to have different cryptic alleles hidden within them, especially if thy are genetically distinct on other criteria. Of course, it has long been recognised that superior lines of animals may owe

their superiority to alleles at different loci in which case the genetic basis of superiority is cryptically different.

The message is clear. Phenotypic evaluation is a very poor and misleading guide to the genetic resources within a breed. Breeds shown by biodiversity studies, such as mitochondrial sequence analysis or microsatellite markers, to be most dissimilar and genetically distinct from current elite breeds are more likely to contain novel cryptic alleles. The conservation of all breeds, but with a special emphasis on the most distinct breeds, is very important for maintaining the resources for future breed improvement. Asian pig breeds, with their long history of separate evolution from European breeds apart from some relatively recent introgression, have very great promise for providing cryptic alleles of value for improvement of existing elite European breeds which currently dominate world production.

QTL Studies in Australia

In Australia, we do not have easy access to the wealth of breed resources available to animal breeders in other parts of the world. None of the common domestic species are native to Australia and stringent quarantine regulations to prevent the introduction of animal diseases make it difficult or impossible to import animal germplasm as well. Importation of pigs into Australia is virtually impossible because of these quarantine restrictions and is certainly not feasible within the financial constraints of a scientific research program. For this reason, QTL mapping work on pigs in Australia was initiated using commercial lines of animals. This constraint has meant that the Australian studies are perhaps less likely a priori to detect QTLs. However, those detected and of sufficient size and economic relevance can be immediately exploited within current breeding programs without the need for extensive backcrossing and introgression.

Conventional Animal Improvement – PigBLUP

Performance-based animal breeding, especially if it uses sophisticated statistical methodology like Best linear Unbiased Prediction (BLUP), is a robust and demonstrably successful method of increasing the economic productivity of animals. In Australia, the Animal Genetics and Breeding Unit at the University of New England in Armidale has been a world leader in the adoption and utilisation of BLUP-based breeding methodology for many

species. For pig breeders, a computer package called PigBLUP has been produced and is widely used throughout the industry for purposes of record keeping, breeding value estimation and selection decisions (http://abri.une.edu. au/pigblup.htm). PigBLUP is continually upgraded and improved both in relation to user preferences and utility and also scientific advances. QTL studies performed at the University of Sydney, in conjunction with collaborators at the University of New England and the University of Melbourne, have the pragmatic aim of enhancing pig breeding efficiency. If the studies demonstrate the occurrence of QTL variation which is economically exploitable, then an additional feature will be added to PiGBLUP to allow incorporation of marker and QTL information into future evaluation and choice of parents for use in breeding.

Overview of QTL studies at University of Sydney

All QTL studies performed at the University of Sydney have involved an industry partner, Bunge Meat Industries Pty Ltd. Bunge Meat Industries is a very large integrated breeding and production company with one large piggery with approximately 24,000 sows and several smaller separate piggeries with about 6,000 sows. The very large scale of the Bunge operation means that it has a large veterinary and scientific staff, familiar with the requirements of scientific research, and able to breed and performance test large numbers of animals for QTL mapping projects in a relatively short period of time. The two major QTL studies undertaken to date have employed a sire design. In other words, a selected group of sires were mated with large numbers of sows and large sire progeny groups have been performance tested for numerous production and meat quality-related traits. Genotyping for the purpose of genome scanning or for analysis of targeted regions has been and continues to be performed at the University of Sydney. From our first study, involving four large sire families, approximately 70% of the genome has been scanned. Marker informativeness is of course lower in such studies than in the analyses of F2 populations between wide crosses. It is sometimes frustrating to find a particular genomic interval in a particular sire where a series of markers are found to be uninformative, when all other sires have proved informative for that region. The other important difference between this type

of study and the typical wide-cross QTL analysis is that it is assumed in wide cross studies that the two extreme breeds are fixed for alternative QTL alleles. Thus all F1 parents are heterozygous and informative with respect to the QTL. However, when sires are more-or-less randomly chosen from a commercial population, one has no idea in advance whether they are heterozygous for a QTL or not. This means that only a proportion of the

Table 3. Estimated effects of QTL ((SE) affecting growth, meat quality and carcase.

Effects estimated by single trait mapping and using a) Lander and Kruglyak (1995) significance thresholds and b) less stringent Churchill and Doerge (1994) empirical thresholds.

a). Trait*	Boar	Effect of allele substitution, δ	b). Trait*	Boar	Effect of allele substitution, δ
ADG1	1	60 (15) gm	ADG2	2	217 (52) gm
ADG3	4	104 (30) gm	ADG2	2	140 (54) gm
FD3/4	1	1.43 (.36) mm	ADG2	1	138 (44) gm
FDP2	3	1.85 (.55) mm	ADG3	3	63 (19) gm
HCP2	1	1.73 (.48) mm	ADG3	3	77.8 (24) gm
HCP2	1	1.86 (.46) mm	ADG3	4	75 (27) gm
HAM	1	-.44 (.14) kg	DFDINT	2	.26 (.13)
HAM	2	-.43 (.13) kg	FDP2	2	1.26 (.41) mm
HAM	4	1.39 (.57) kg	HCP2	4	-3.67 (1.0) mm
HAM	4	1.42 (.75) kg	HCP2	2	1.45 (.77) mm
HAM	3	1.02 (.17) kg	MD3/4	2	3.75 (1.02) mm
HAM	4	1.43 (.25) kg	LW	4	1.08 (.52) kg
LW	4	1.84 (.53) kg	IMF	4	.93 (.63)
LW	3	1.56 (.30) kg	CLD	2	6.2 (2.03)
LW	4	1.62 (.46) kg	CSP	1	2.33 (1.29)
IMF	4	.94 (.27)	PH24	1	-.10 (.03)
CLD	1	3.46 (.87)	PH45	3	.32 (.16)
PH24	2	.13 (.04)	PH45	3	.30 (.12)
PH45	3	.34 (.12)			

*ADG 1, 2 and 3, average daily gain from 3 to 18 weeks, from 18 to 22 weeks and lifetime respectively; DFDINT, daily feed intake 18-22 weeks; FD3/4, MD3/4, FDP2 and HCP2, fat or muscle depth at 3 and 4th last ribs or P2 position measured with real time ultrasound or Hennesy Chong probe; HAM, LW, weight of back left leg slash boned or bone in; IMF, intramuscular fat percentage; CLD, CSP, colour of longissimus dorsi or superior spiralis muscle; PH45, PH24, muscle pH 45 minutes or 24 hours after slaughter.

sires, depending on the QTL allele frequencies, will be heterozygous for a QTL and thus capable of revealing its existence by its segregation among the progeny. In our first study involving four sire families, virtually all QTL were detected in only one sire from four, with only an occasional hint of support for existence of a QTL at a particular site coming from more than one sire.

Analysis of QTL data is a particular challenge. Dr Richard Kerr at the Animal Genetics and Breeding Unit at the University of New England is developing methodology and software for QTL analyses as part of the Australian QTL mapping project. This includes software for performing multiple trait analyses (quite a few traits are closely related, for example different measures of fatness, where it makes sense to attempt a joint analysis of the data) or multiple family analyses (to attempt to increases the precision of detection of QTL within particular families).

In general, the results of the first QTL study on Australian commercial populations (Table 3) has provided very encouraging indications of the existence of QTL for a range of growth and quality traits. The study currently underway aims to independently confirm the existence of these and other QTL but has also included a much larger set of trait measurements, including more measures of meat quality.

Conclusions

Biodiversity analyses, using microsatellite markers, mitochondrial D-loop or other sequences, have provided quantitative measures of diversity. While many breeds remain to be included within global studies of biodiversity, these data will ultimately enable efficient conservation strategies. QTL mapping in animals has provided strong empirical support for the validity of conservation of rare and endangered breeds of pigs and other domestic species. The cryptic genetic resources within breeds of animals are beginning to be exposed by gene mapping and QTL analyses and are strongly reinforced by similar results from many species of economically important plants.

Positional candidates

Ideally the full exploitation of QTL will be achieved when the underlying gene and genetic variants are recognised, enabling direct DNA-based

selection of the favourable alleles. Soller and Rothschild (1999) last year spoke about the candidate gene approach for identifying useful genetic variants for use in breeding programs. The powerful combination of map position of a QTL in a domestic animal in concert with a detailed genomic sequence from the equivalent region in human or mouse will enable much more refined identification of positional candidate genes, eliminating the vast majority of potential candidates based only on physiological function. With the first draft of the human genome already available and multiple-pass annotated versions likely to be available in the near future, the human genome sequence provides an enormous and untapped resource of huge benefit to animal breeders and geneticists.

Data mining the genome initiatives

The latest efforts in bioinformatics are directed at mining the human and model organism genome sequences. At one level, this is simply to identify and interpret and annotate the human sequence. From the point of view of animal geneticists, the opportunity is available for finding a fast track to identification of genes underlying

economically important phenotypic variation in domestic animals.

Functional Genomics – reverse data mining

Interestingly, this opportunity for animal geneticists to mine the human genome sequence also represents a complementary opportunity for human geneticists to reverse mine the large amounts of functional phenotypic data being accumulated in large and well designed animal QTL mapping projects. Humans are very poor subjects for QTL studies as family sizes are small and pedigree data only become available fortuitously and opportunistically rather than by design as in animals. There is intense interest among human geneticists in exploiting the results of animal studies for providing functional significance to human sequences.

The future – single nucleotide polymorphisms (SNPs)

The human genome initiative is moving on to a new phase of identification of functionally significant variation. Low cost, high throughput automated methods of detection of this

sequence diversity, mainly in the form of single nucleotide polymorphisms, are being developed by many organisations. These methods have the ultimate objective of detecting all functionally relevant sequence variants for all traits. Perhaps there may be a time in the future when such methods can be used for screening the cryptic genetic resources of rare and endangered breeds. Instead of using functionally irrelevant markers such as microsatellites and mitochondrial D-loop sequence variants which merely indicate evolutionary distinctiveness, we will be able to screen a breed with a SNP panel capable of recognising all functionally important variants at all loci. Then a fully informed decision could be made about conservation and exploitation of the genetic resources of the breed.

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