QTL Analyses across Studies and Designs

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Abstract:

Livestock genomic research has developed in parallel with similar research on humans and other model species. Much of the current focus of research is on the identification of loci controlling variation in complex traits, particularly quantitative trait loci (QTL). Compared to research on humans, livestock research has less well developed technological resources and as yet has limited sequence data, but benefits from access to large well-recorded and pedigreed populations and the ability to undertake designed crosses. In addition, the similarity in structure and sequence between different mammalian species allows ready transfer of comparative information between species. Taking pigs as an example, initial QTL studies focused on crosses between genetically diverse breeds, particularly wild boar or Chinese Meishan crossed with European commercial pigs. QTL with major effects on traits such as growth rate and fatness have been mapped in such studies. Meta-analysis of data from different studies has been used to confirm the presence of the major QTL and improve estimates of their position and effect as well as to identify new QTL. One advantage of studies based on outbred crosses is the ability to detect QTL showing parent of origin effects characteristic of imprinting. One such QTL with effects on lean tissue content is well documented in pigs, but further meta-analysis has failed to confirm the widespread occurrence of such effects that had been reported in a previous study of pigs. An area of active research is the detection of QTL involved in epistatic interactions for which there is increasing evidence in model species and in livestock. However, for some individual QTL it has been demonstrated that markers can be used to move alleles from one genetic background to another with little evidence of major changes in the effects of the QTL. This demonstrates that, at least for some QTL, epistatic interactions with the background genotype are not substantial. Studies within outbred populations of pigs, rather than in designed crosses, also demonstrate the presence of segregating QTL. In some cases these coincide with the position of QTL found in diverse crosses. An interesting topic for debate and future research is why QTL with major effect remain segregating after long-term directional selection for the traits that they influence. Ongoing research is focusing on the use of linkage disequilibrium mapping in these outbred populations for high precision mapping of QTL in order to aid identification of the causative polymorphisms at the DNA level. New traits, especially genetic resistance to disease, are also under study. For the future, we can anticipate that the flow of information between mammalian species will continue to grow and understanding of the genetics of complex traits such as obesity and disease susceptibility in livestock will increasingly inform studies of human variation.