

## Concordance analysis: from QTL to candidate causative mutations

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## Concordance analysis: from QTL to candidate causative mutations

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Although a large number of QTL have been detected for various traits in dairy cattle, the causative mutations underlying these QTL have only in a few cases been detected. QTL regions contain a large number of polymorphisms and thus candidate mutations. Our goal was to narrow down from QTL region to a limited number of candidate mutations by a concordance analysis. The principle behind this analysis is that if a mutation is the causative mutation underlying a QTL, it should be homozygous when an animal is homozygous for the QTL and heterozygous when an animal is heterozygous for the QTL.

QTL regions were selected using Bayes C for various quantitative traits, including milk yield and composition, fertility, somatic cell count and conformation traits, based on the posterior inclusion probability summed over intervals of 40 markers. For each selected region, QTL statuses were determined using marker effects estimated by Bayes C. A list of polymorphisms in the QTL region was obtained and subsequently, polymorphisms whose status was non-concordant with the QTL status were discarded.

The success of the analysis depended both on the accuracy of the QTL status prediction and on the linkage disequilibrium in the QTL region. When several QTL are present in the same region, it becomes more difficult and sometimes impossible to accurately predict the QTL status. Since an accurate QTL status is required for the concordance analysis, the analysis could only be performed for QTL with an accurate status prediction. Furthermore, for regions with strong linkage disequilibrium, most polymorphisms in the region will be in concordance with each other. As a result, hardly any polymorphisms could be discarded. In such a situation, validation across breeds will help to reduce the amount of linkage disequilibrium. For other regions, however, it was possible to reduce the number of polymorphisms from more than 30,000 in the QTL region to less than 100, located in only one or two genes, showing the potential of the analysis in the search for causative mutations.

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