Is There More to SNP Chips than Just SNPs? Detecting Copy Number Variation in Chickens

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Summary and Implications

SNP chips can be used to provide genomic information on copy number variations (CNV) in addition to information on the SNPs (single nucleotide polymorphism) for which they were originally designed. Although some CNVs may be missed compared to methods specifically designed for CVN detection, CNVs derived from SNP chips can contribute valuable information on additional genetic variation without additional cost.

Introduction

SNP chips have been designed to interrogate the genome for tens or hundreds of thousands of genetic markers called single nucleotide polymorphisms (SNPs) simultaneously at a relatively low cost per unit of genomic information. They have enabled incorporating genomic information into poultry breeding programs. Information from SNP chips has been collected on thousands of individuals in the major breeding companies. However, genetic variation goes beyond SNPs. Another important source of genetic variation defined as large scale duplications or deletions relative to the reference genome is called Copy Number Variation (CNV). Although not specifically designed for that purpose, unusual types of sample clustering can be used to detect CNVs from SNP chips. In this study we used data from publicly available 600k, and custom made 50k and 42k SNP chips to detect CVNs in chickens.

Materials and Methods

Genotype clusters obtained for a typical high-quality SNP, segregating at moderate frequency, are shown in Figure 1. Three clearly distinct clusters can be identified, representing the AA, AB and BB genotypes. In contrast to a SNP genotype, a CNV will present itself as having more than 3 clusters (Figure 2). Detecting cluster patterns that represent CNVs enables extracting additional information on genetic variation from SNP chips without additional cost. The Axiom® CNV Summary Tools, combined with the PennCNV software, were used to identify CNVs in 18,719 chickens from 4 pure lines (2 white egg layers and 2 white egg layers) and one commercial cross.

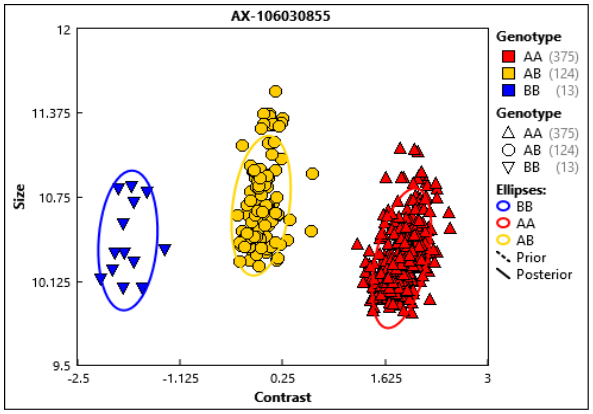

Figure 1. High quality, SNP genotypes (3 clusters)

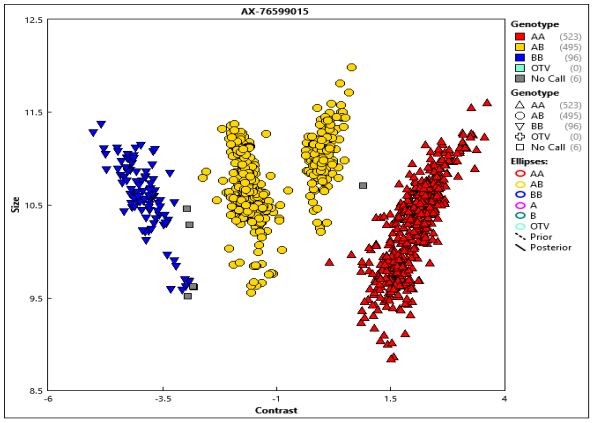

Figure 2. Segregating CNV (more than 3 clusters)

Results and Discussion

In total 19,525 CNVs were detected, with an average length of 51.1 kilobases. Compared to the high-density panel (600k), the medium density panels detected less CNVs but with higher average length. This is likely due to the higher threshold for detection and also the design of custom chips, which intentionally avoids genomic locations with unusual clustering patterns. More CNVs were detected in brown egg layers than in white egg layers.

Acknowledgments

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