

Genome-wide detection and analysis of copy number variation in Italian indigenous chicken breeds

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Advancements in high-density integrated chips have enabled the identification of copy number variations (CNVs) with increased resolution. This study aimed to identify CNVs within the genome of 530 animals of 23 Italian indigenous chicken breeds and explore their distribution. Animals were genotyped using the 600K Affymetrix Axiom HD chip and CNVs were identified using the Hidden Markov Model using the PennCNV software. A significant number of CNVs were observed in the genomes, with higher distribution in the first six chromosomes and predominantly associated with loss types. A total of 1172 CNV regions (CNVRs) composed of 1084 losses, 36 gains, and 54 mixed CNVRs were defined: 711 CNVRs (60.7%) did not involve any genes and 461 CNVRs (39.3%) encompassed one or more genes, including protein-coding genes (87.8%; *DACH1*, *DMD*, *NTS*), miRNAs (9.8%; *MIR7445-1*), and genes of uncertain function (2.4%). Results provided an insight into the genetic variability and diversity of Italian local chicken population using CNVs, highlighted potential functional associations with crucial genes, and suggested that the breeds are indeed reservoirs of genetic diversity.

Keywords: CNV, local population, poultry