



THE UNIVERSITY of EDINBURGH The Royal (Dick) School of Veterinary Studies

Genome-wide association studies for body weight in broilers using sequencing and SNP chip data

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Aim of this presentation

• To compare the SNP signals between the GWAS using the SNP chip and whole-genome sequencing (WGS) data.

Material and methods (1)

SNP chip data:

60k animals and 45,834 autosomal SNPs (chr: 1-28)

After quality control (QC) :

- 56635 samples (29865 females and 26770 males) remained with call rate >0.90,
- 44,959 autosomal SNPs remained with call rate >0.95, minor allele frequency >0.05 and HWE test pvalue> 0.000001

Software: PLINK

Sequence data:

- We used the same 60k animals as for chip data.
- These SNP genotypes were imputed to sequence level for a total of 12,806,034 variants with high levels of accuracy (>0.95).
- Genotype imputation was conducted with Beagle software.

Material and Methods (2)

GWAS using chip and sequence data, respectively

- Software: GCTA
- Same Linear mixed model: Each time, a single locus mixed (additive) model GWAS:
- SNPs (fitted as fixed effect covariates) and adjusted BW for the fixed effects (no covariates)
- Animals' relationships were included in the mixed model as random effects via the genomic relationship matrix (GRM).
- Genome-wide significant SNP: FDR p-value < 0.05
- Significant markers were annotated using Ensembl Variant Effect Predictor (VEP) tool against the gene annotation database and genome reference sequence file from Ensembl for *Gallus gallus*.

Results by GWAS using chip data

In total: 11 genome-wide significant SNPs



Results by VEP for the significant markers using SNP

Position of the marker (bp)	Chromoso me	P-value (FDR p- value)	Consequence type of variant	Positional candidate gene(s) including significant marker						
31128475	2	1.83379e-06 (0.009)	intron	GPNMB						
31294098	2	7.25205e-06 (0.027)	intron	TRA2A						
111130624	2	8.01306e-07 (0.007)	Upstream gene variant	LOC112531857						
111156392	2	1.60245e-06 (0.009)	intron	LYN						
111182748	2	1.51017e-06 (0.009)	intron	KIF20AL						
111287788	2	1.20372e-07 (0.002)	intron	CHCHD7						
111314287	2	1.18994e-06 (0.008)	intron	CHCHD7,SDR16C5						
111452734	2	2.61663e-07 (0.003)	intron	Non-coding RNA gene						
111597499	2	4.70903e-07 (0.005)	intron	IMPAD1						
1236869	11	5.79365e-06 (0.023)	intron	Non-coding RNA gene						
1288067	11	5.599e-06 (0.023)	intron	PLEKHG4						

Results by VEP for the significant markers using SNP

Position of the marker (bp)	Chromoso me	P-value (FDR p- value)	Consequence type of variant		Position includin _{	270 sequence variants were lied within GPNMB gene		
31128475	2	1.83379e-06 (0.009)	intron	GPNMB				
31294098	2	7.25205e-06 (0.027)	intron	GPNMB (alycoprotein nmb)				
111130624	2	8.01306e-07 (0.007)	Upstream gene variant	gen dep	gene regulates abdominal fat deposition in chickens (Wang			
111156392	2	1.60245e-06 (0.009)	intron	et al., 2022) Poult Sci: 10.1016/j.psj.2022.102216.				
111182748	2	1.51017e-06 (0.009)	intron					
111287788	2	1.20372e-07 (0.002)	intron		CHCHD7			
111314287	2	1.18994e-06 (0.008)	intron			HCHD7,SDR16C5		
111452734	2	2.61663e-07 (0.003)	intron		none			
111597499	2	4.70903e-07 (0.005)	intron		IMPAD1			
1236869	11	5.79365e-06 (0.023)	intron		none			
1288067	11	5.599e-06 (0.023)	intron		PLEKHG4	1		

Common markers between GWAS using SNP vs. sequence data





- 3 markers on GGA2 and GGA11 were common between the GWAS
 - In total: 44 significant SNPs were identified by GWAS using sequence data for GGA2 while 531 SNPs were found for GGA11.

GWAS using sequence data highlighted more significant variants e.g.



GGA8 (243 SNPs), GGA19 (2 SNPs), GGA20 (122 SNPs), GGA23 (3 SNPs)

Conclusions

- With the use of sequencing data, results were similar to those obtained with SNP chip data. However, additional GWAS signals were found.
- Our results suggest an improvement in power and precision when using WGS data. However, the use of sequence data is still time-consuming.
- In spite of the identified markers for BW, the high proportion of genetic variance attributed to regions harbouring non-significant SNPs supports the hypothesis that the genetic architecture of BW is polygenic.
- Under the assumption of polygenicity, a "large" number of genes with "small" effects is expected to control BW.
- Next step: Genomic prediction

Thank you for attention!

Aviagen is acknowledged for data provision

