

Impact of Copy Number Variation on gene expression

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Data set

Source	PRJNA354435	PRJNA403969	Self collected data (S1-S6)
Breed	Duroc x Pietrain	Duroc x Pietrain	Polish Landrace
# pigs	1 female	143 females	6 males
DNA-seq	HiSeq 2500 coverage: 15	-	NovaSeq 6000, coverage: ~15
RNA-seq (Muscle)	HiSeq 2500, >170 mln PE reads	Hiseq 2500, ~40 mln PE reads/sample	NovaSeq 6000, 120-170 mln PE reads

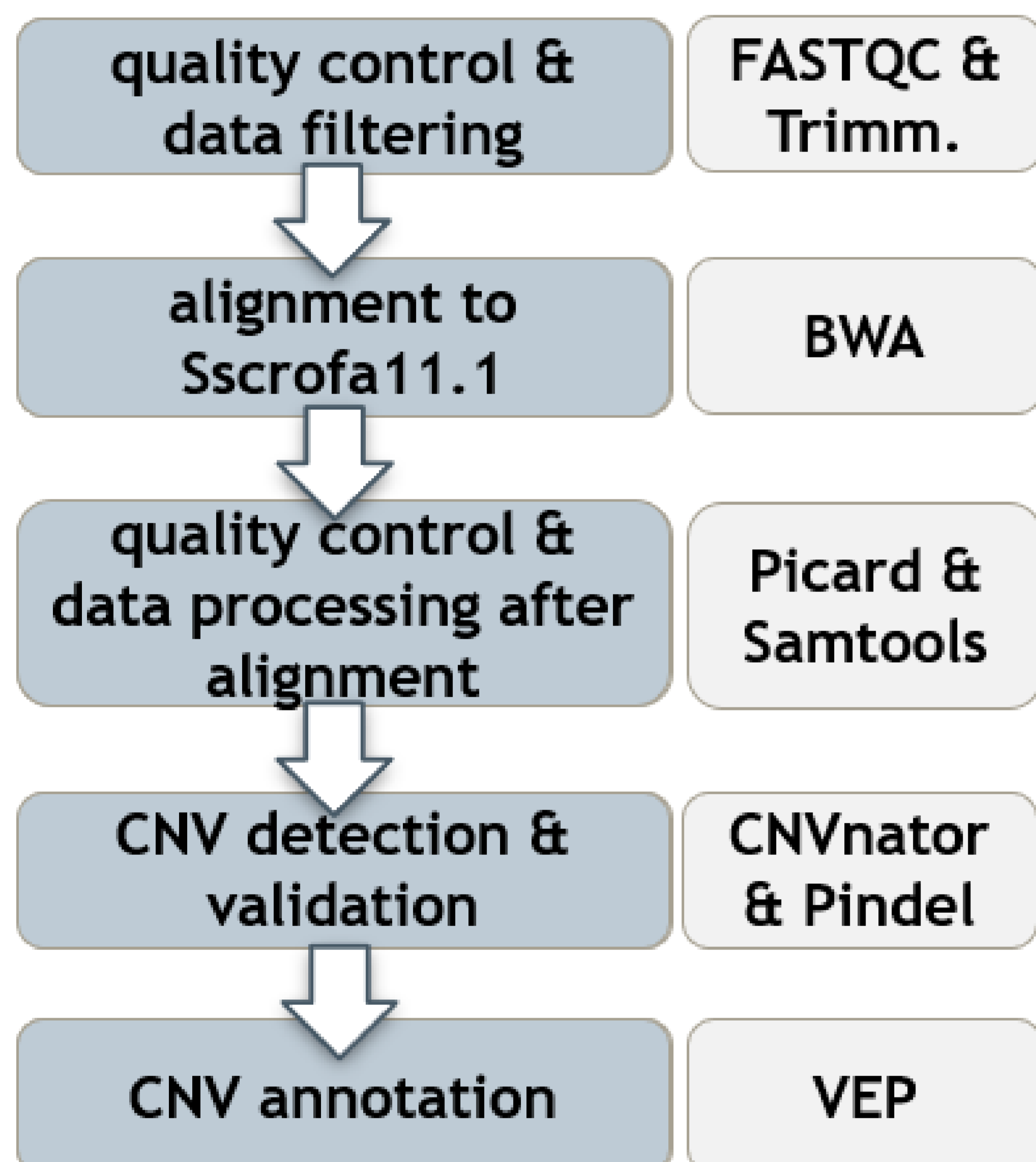
Conclusions

- Deletions have a potential to reduce gene expression.
- Duplications do not change gene expression.
- There is no correlation between the length of CNV and the expression level.

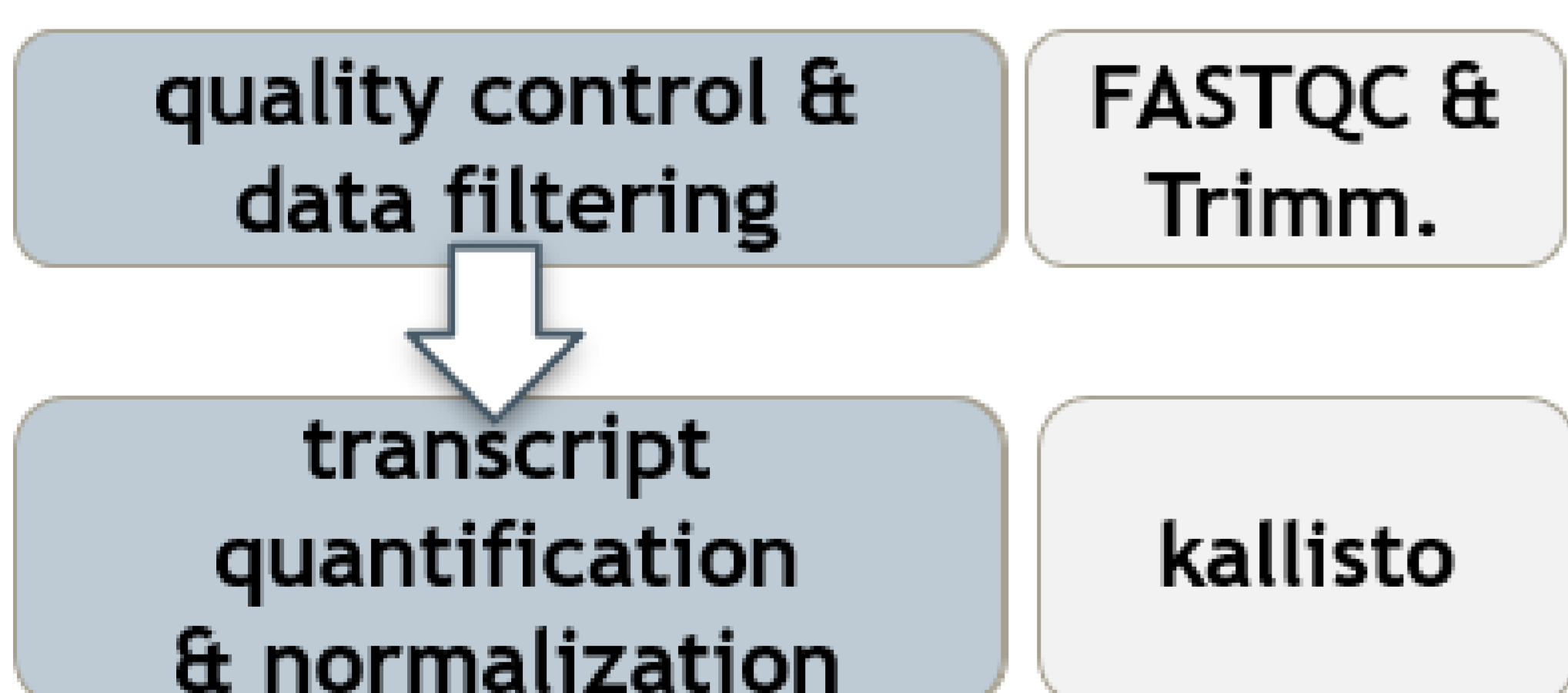
Results

Methods

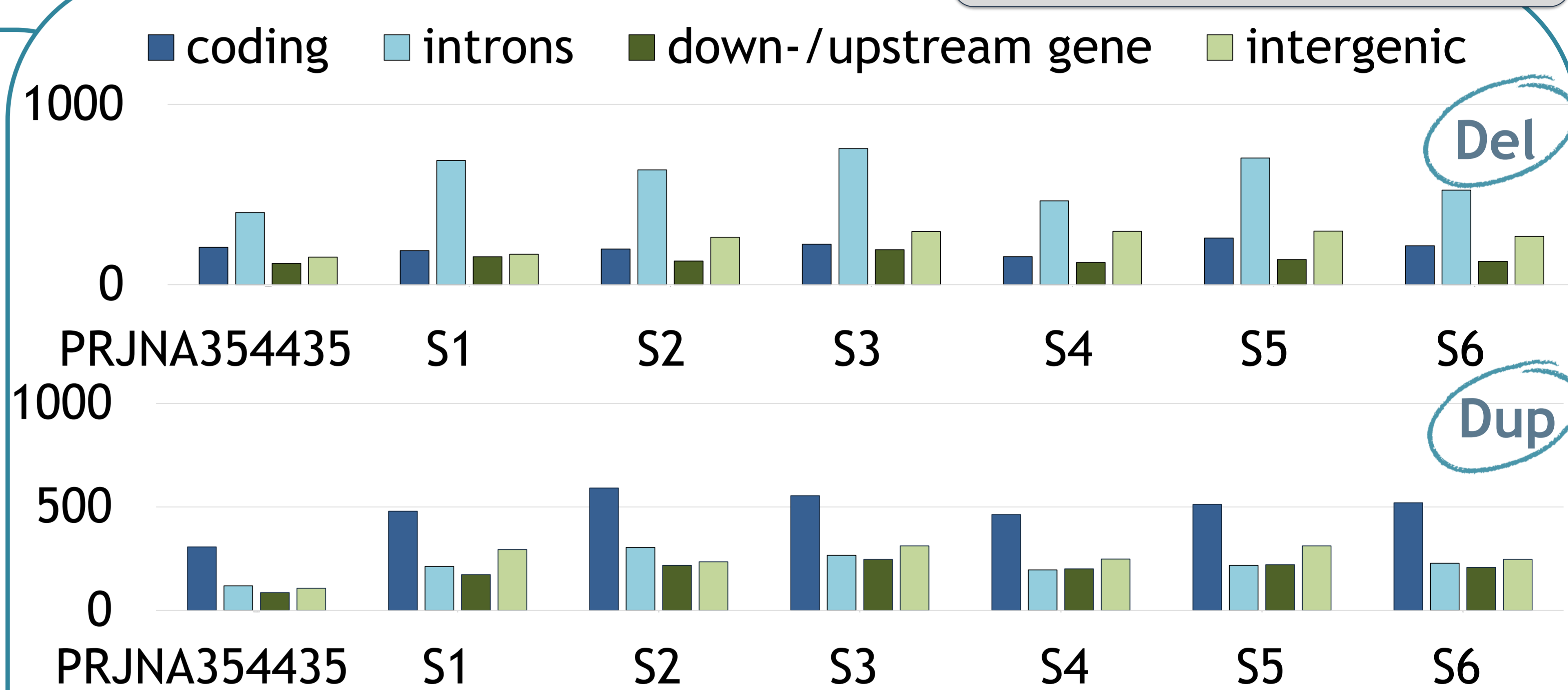
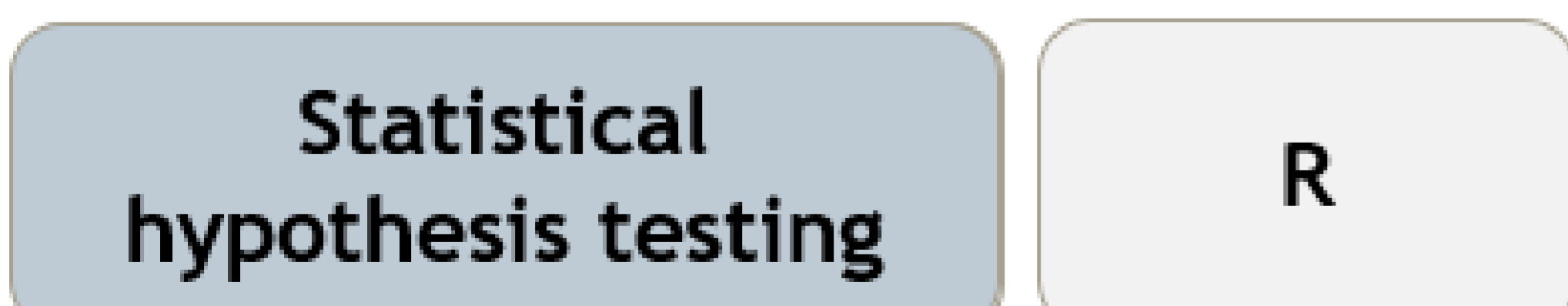
1. CNVs detection



2. Transcript quantification



3. CNV impact on gene expression investigation



Wilcoxon signed-rank test

E_1 - expression of transcripts with CNVs in 1 animal.

E_{143} - average expression of transcripts across 143 animals.

$$H_1: E_1 < E_{143}$$

coding regions	0.002
introns	$1.316 \cdot 10^{-20}$
down-/upstream gene	0.026

$$H_1: E_1 > E_{143}$$

coding regions	0.020
introns	0.999
down-/upstream gene	0.619

$$H_1: E_1 < E_{143}$$

coding regions	0.980
introns	$3.84 \cdot 10^{-5}$
down-/upstream gene	0.389

E - average expression of transcripts without CNVs.

E_{CNV} - average expression of transcripts with CNVs.

$$H_1: E < E_{CNV}$$

coding regions	0.008
introns	$1.355 \cdot 10^{-10}$
down-/upstream gene	0.085

$$H_1: E > E_{CNV}$$

coding regions	0.999
introns	0.846
down-/upstream gene	0.872

$$H_1: E < E_{CNV}$$

coding regions	$8.318 \cdot 10^{-5}$
introns	0.154
down-/upstream gene	0.129

Correlation of CNV length and expression level confirmed only for:

- PRJNA354435, introns (R=0.16, P=0.004)
- S5, coding regions (R=0.78, P=0.004)

- S5, introns (R=0.65, P=0.011)

Acknowledgments

“Modulation of gene expression by CNV in pigs” (N060/0035/20).

