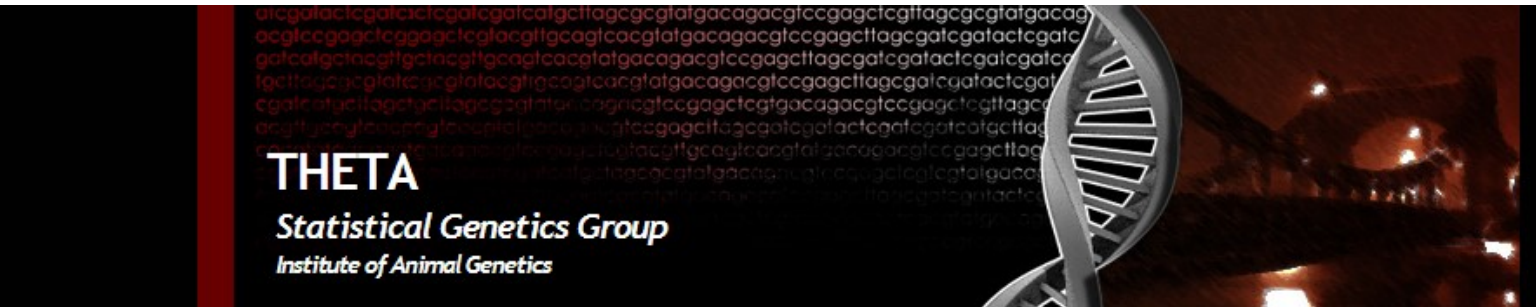


<http://theta.edu.pl/>

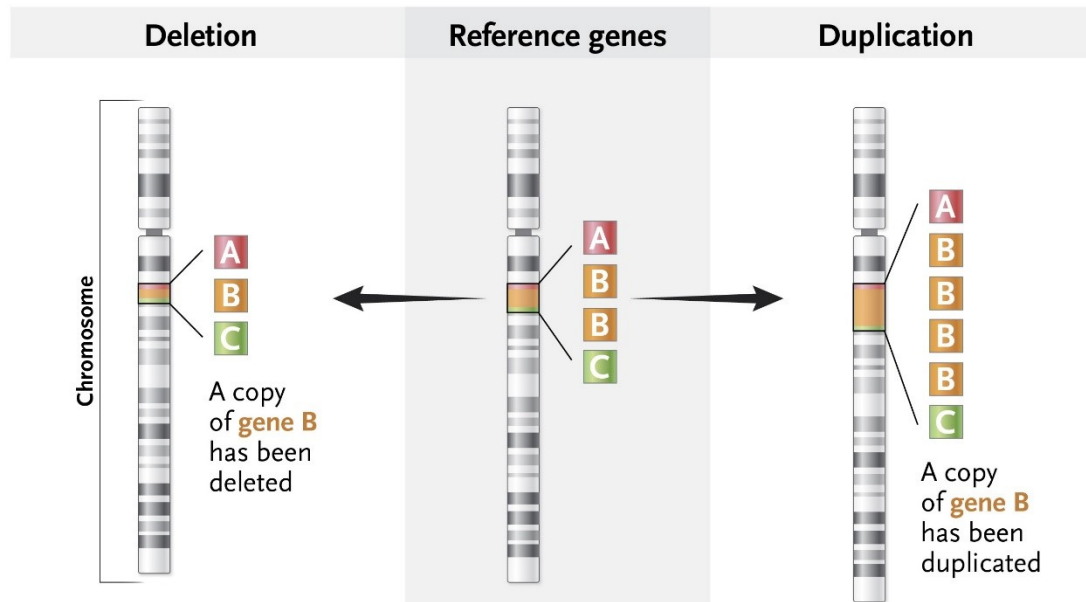


# Modulation of gene expression by CNVs

M. Mielczarek, M. Fraszczak, J.Szyda

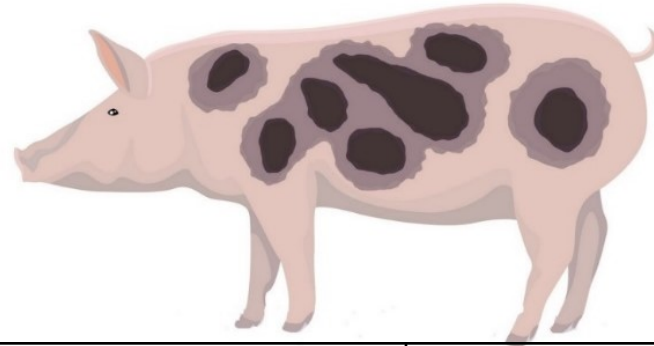
# Background & Objectives

- Genome-wide CNVs detection
- **CNV impact on gene expression**



source: <https://illustrated-glossary.nejm.org>

# Dataset



NCBI BioProject	PRJNA354435	PRJNA403969
Breed	Duroc x Pietrain	Duroc x Pietrain
# individuals	<b>1</b> female	<b>168</b> females
DNA-seq	Illumina HiSeq 2500 150×2 PE average coverage: 15	
RNA-seq	Illumina HiSeq 2500 <b>Muscle</b> 171,810,298 PE reads	Illumina Hiseq 2500 and 2000 <b>Muscle</b> 43,873,302 – 39,455,116 PE reads

# Methods

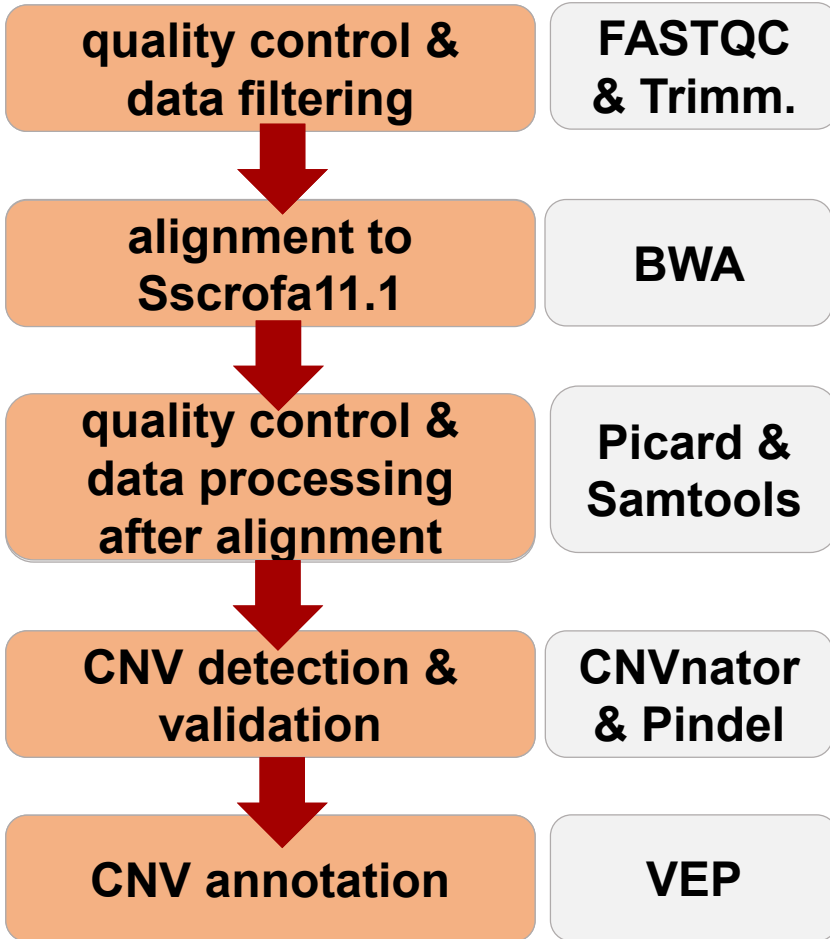
1. CNVs detection (DNA)

2. Transcript quantification (RNA)

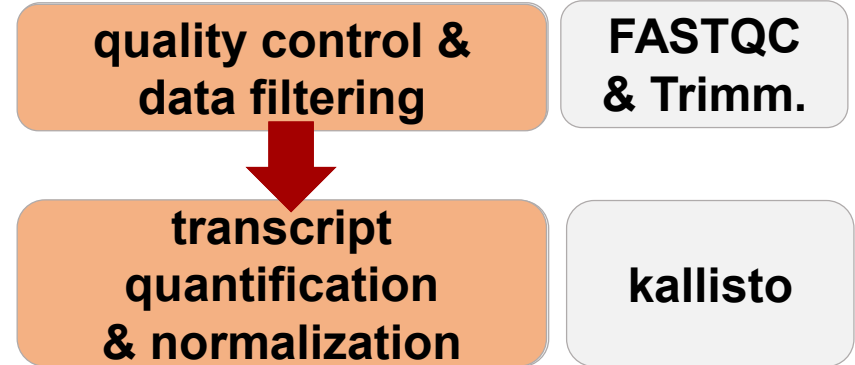
3. CNV impact on gene expression  
investigation

# Methods

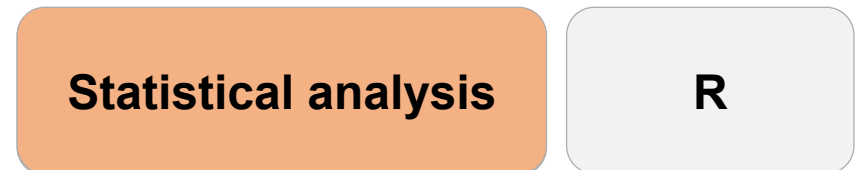
## 1. CNVs detection (DNA)



## 2. Transcript quantification (RNA)



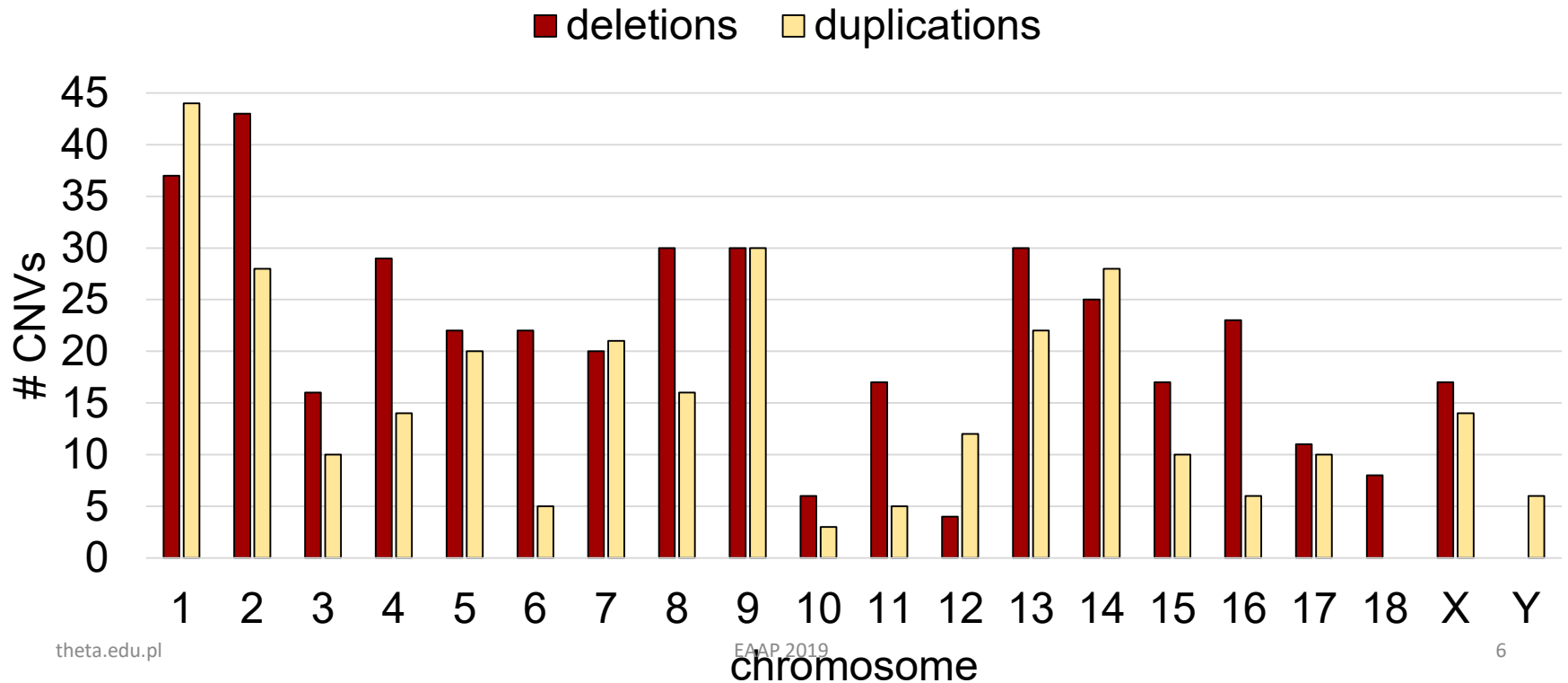
## 3. CNV impact on gene expression investigation



# Results

CNVs characterization → the number of CNVs

- 407 deletions
- 304 duplications

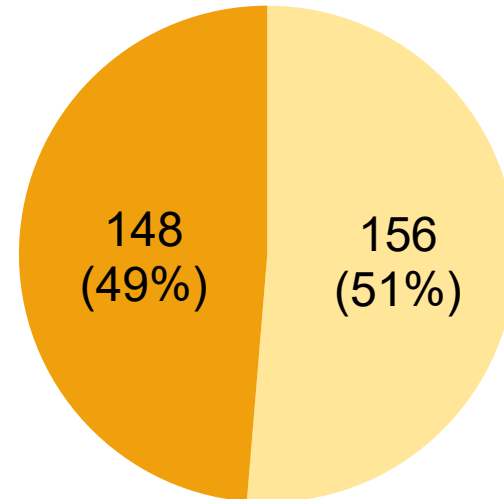
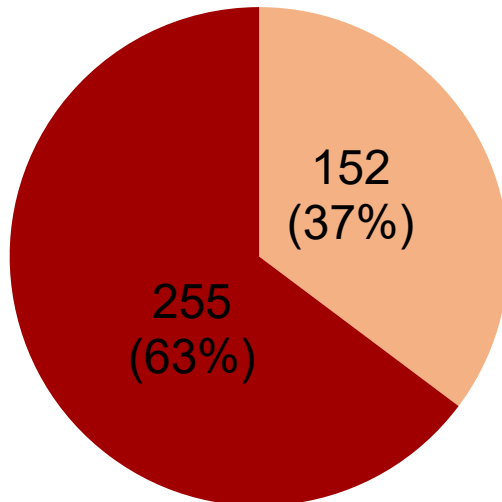


# Results

CNVs characterization → **the number of CNVs**

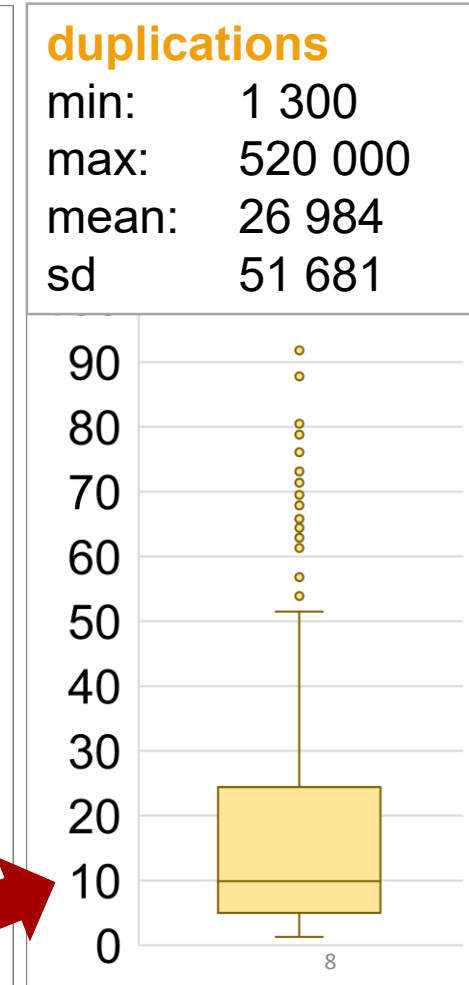
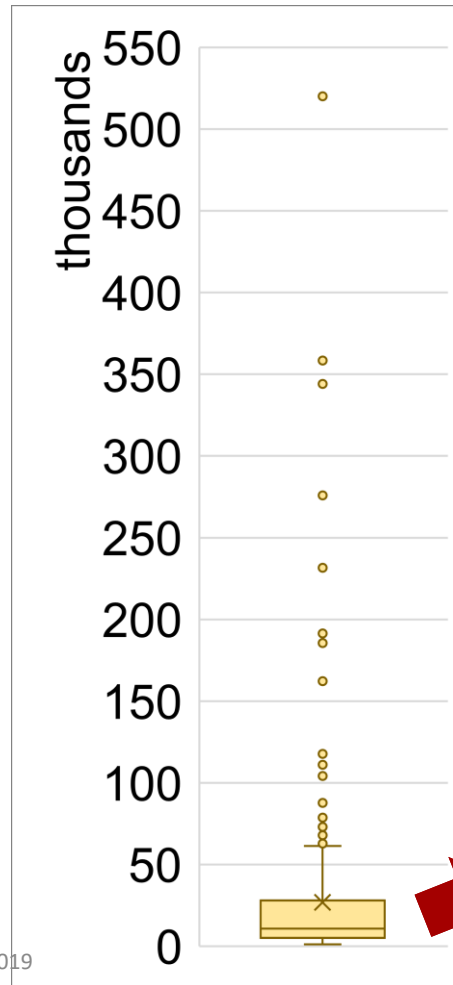
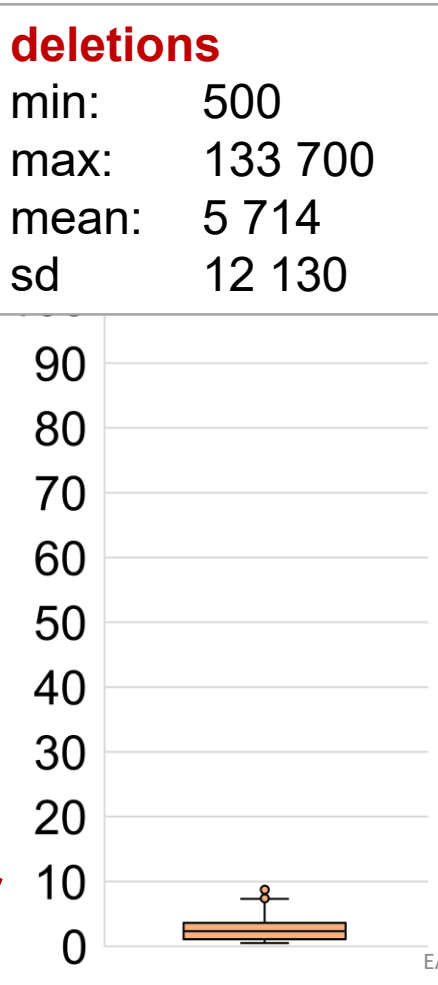
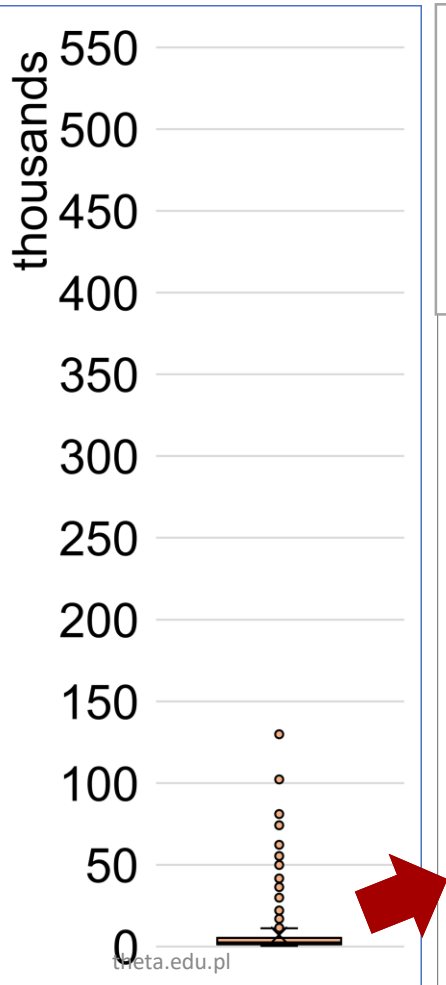
- 407 deletions
- 304 duplications

■ deletions   ■ duplications



# Results

CNVs characterization → CNVs length (bp)





# Results

**Deletions** impact → **Wilcoxon signed-rank test**

$$H_0: Y_{CNV} \geq \bar{Y}$$

$$H_1: Y_{CNV} < \bar{Y}$$

$Y_{CNV}$  - expression of a transcript overlapping with CNV

$\bar{Y}$  - population averaged expression

→ *Transcript expression is lower if deletion appears*  
( $p = 0.0088$ )

$$H_0: Y_R \geq \bar{Y}_R$$

$$H_1: Y_R < \bar{Y}_R$$

$Y_R$  - expression of a random transcript with no CNV

$\bar{Y}_R$  - population averaged expression of a random transcript with no CNV

→ *Transcript expression is not lower in the investigated sow*  
( $p = 0.217$ )

# Results

**Duplications** impact → **Wilcoxon signed-rank test**

$$H_0: Y_{CNV} \leq \bar{Y}$$

$$H_1: Y_{CNV} > \bar{Y}$$

$Y_{CNV}$  - expression of a transcript overlapping with CNV

$\bar{Y}$  - population averaged expression

→ *Transcript expression is not higher if duplication appears*  
( $p = 0.9885$ )

$$H_0: Y_R \leq \bar{Y}_R$$

$$H_1: Y_R > \bar{Y}_R$$

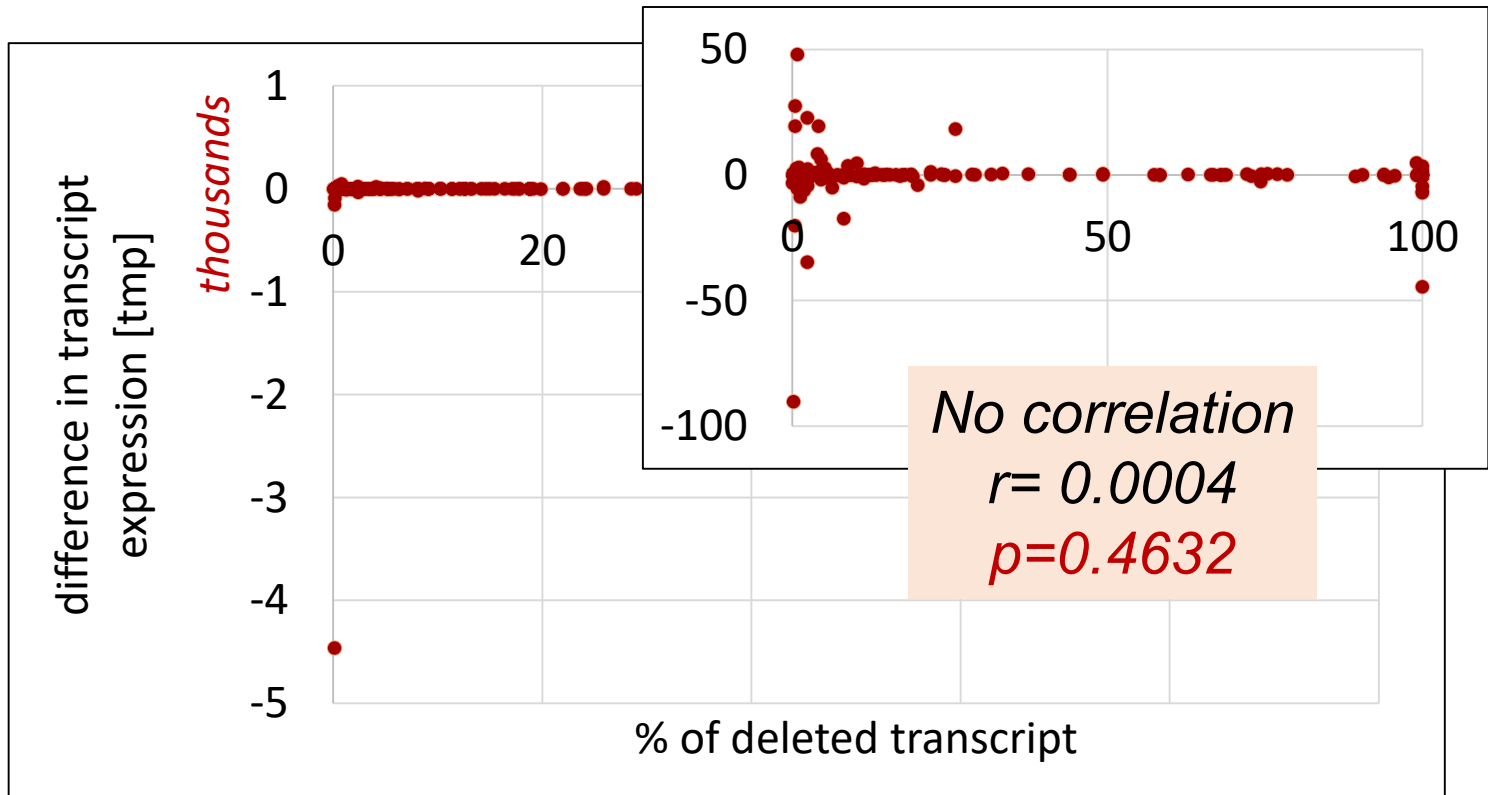
$Y_R$  - expression of a random transcript with no CNV

$\bar{Y}_R$  - population averaged expression of a random transcript with no CNV

→ *Transcript expression is not higher in the investigated sow*  
( $p = 0.7556$ )

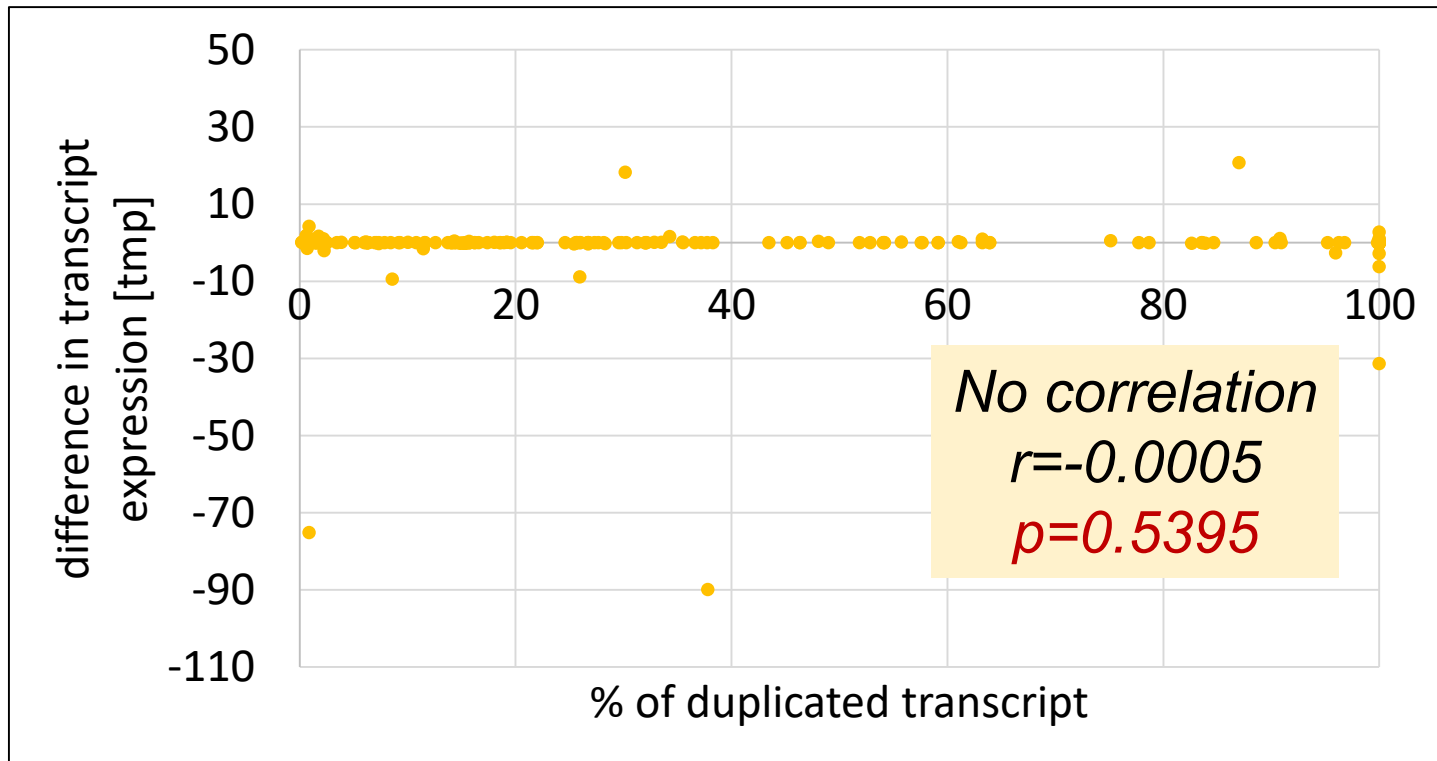
# Results

CNVs impact → **size of a deleted transcript**



# Results

CNVs impact → **size of duplicated transcript**



# Conclusions

- **Deletions** have a potential to reduce gene expression
- **Duplications** does not change gene expression
  
- There is no correlation between the size of **deletions/duplications** and gene expression level

# Acknowledgements

Leading National Research  
Centre in Poland

Poznan Supercomputing  
and Networking Center



Polish National  
Science Centre

**Thank you!**