Analysis of the CNV inheritance in swine genome based on combined Illumina and Nanopore data

M. Frąszczak[1], M. Mielczarek[1,2], M. Kaźmierczak[1], T. Strzała[1], B. Nowak[1], J.Szyda[1,2]

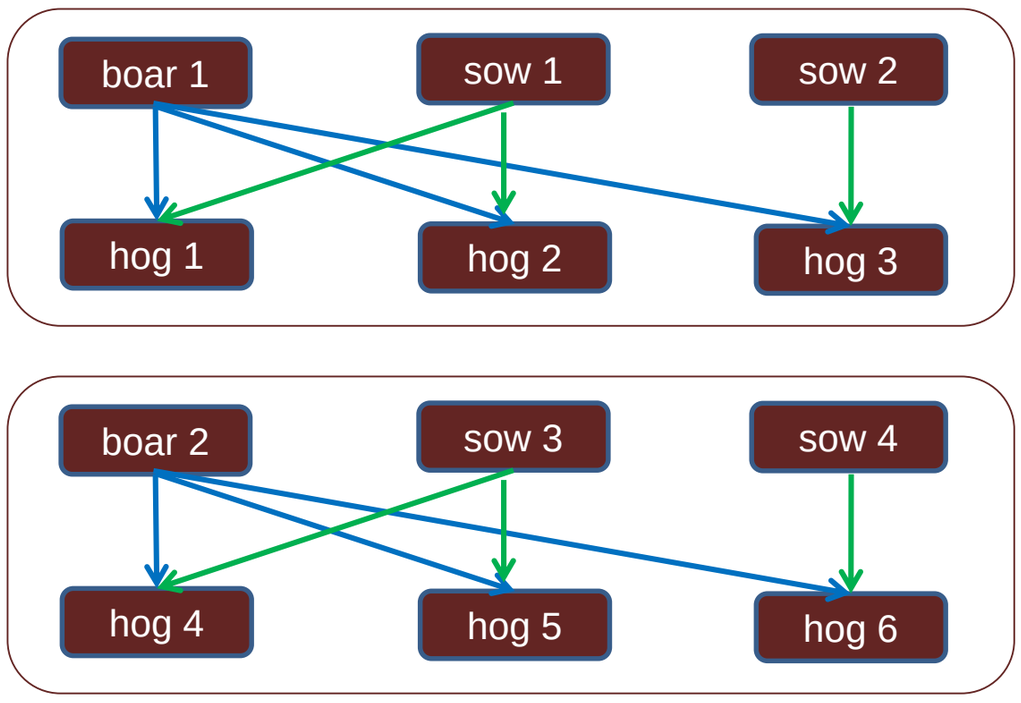
[1]Biostatistics group, Wroclaw University of Environmental and Life Sciences
[2]National Research Institute of Animal Production

CONCLUSIONS

- **CNVs formed *de novo* account for 2%-12% of all CNVs**
- **deletions formed *de novo* are shorter than inherited**
- full siblings are more simmilar each other in structure of CNVs than to other individuals
- duplications in genes are considered to be essential sources of evolutionary innovation and duplications with beneficial effects are likely to be kept
- deletions bring severe consequences by interrupting gene products and causing loss of the biological functions; polymorphisms with large negative effects are likely to be removed from the population by natural selection
- methods based on short read sequencing detected more CNVs than using Oxford Nanopore Technology

Material

- **Whole genome sequence with IluminaHiSeq2000 + Oxford Nanopore Technology**
- **12 swines (6 trios):**



Methods

Bioinformatics pipeline

- **Alignment to Sscrofa11.1 (BWA-MEM / minimap2)**
- **Post alignment filtering (Picard & Samtools)**
- **CNV detection (CNVnator, Pindel, Sniffles, CuteSV)**
- **Functional annotation (VeP)**

Statistical analysis

- **Descriptive statistics**
- **Permutation tests**
- **Wilcoxon sum of rank tests**
- **Multidimensional scalling**
- **Correspondence analysis**
- **Test for fraction**
- **Enrichment analysis**
- **Bonferroni correction**

Results

- Most of deletions (duplications) arise *de novo* is located on chromosome 3 (16)
- Less CNVs arise *de novo* than inherited (del: P=0.008, dup: P=0.006)

Fraction of CNVs formed *de novo*

	hog 1	hog 2	hog 3	hog 4	hog 5	hog 6
deletion	0.123	0.138	0.159	0.118	0.089	0.099
duplication	0.068	0.086	0.061	0.034	0.026	0.051

Fraction of *de novo* deletions depends on animal (P=0.0027).
No significant differences in fraction of *de novo* duplications (P=0.018).

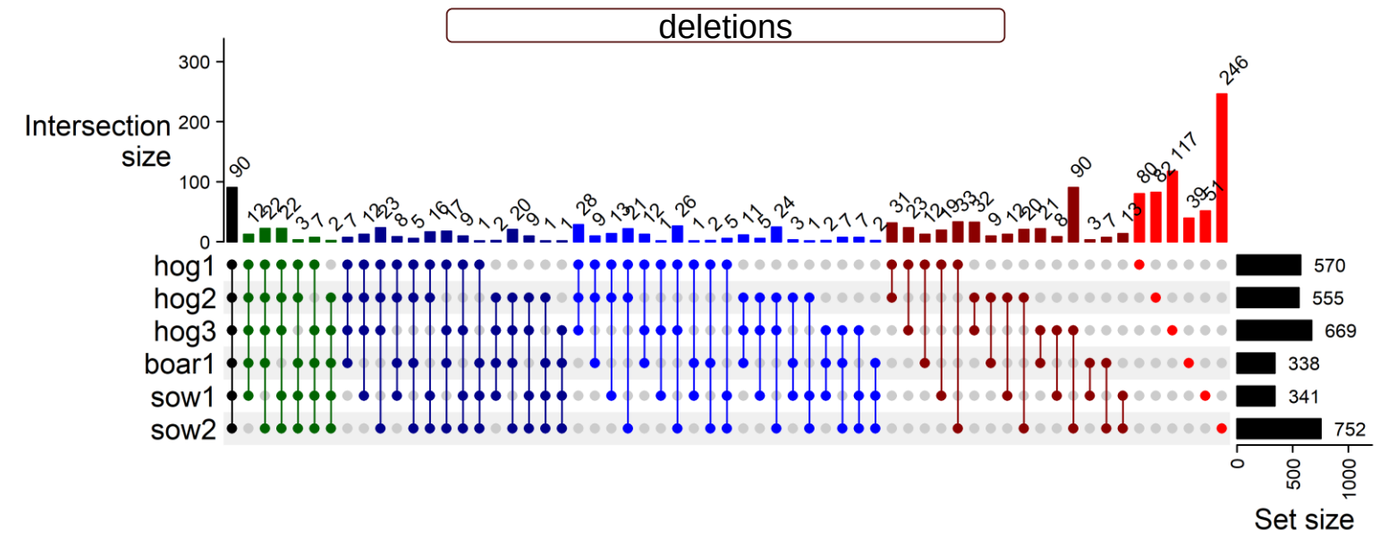
De novo deletions are shorter than inherited in all animals

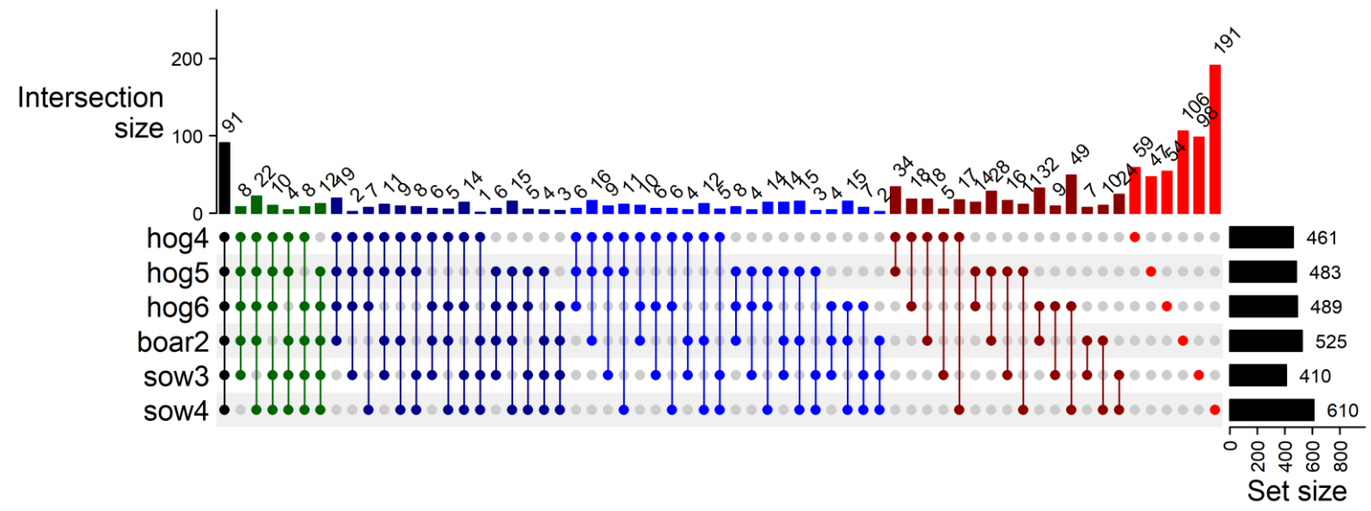
	hog 1	hog 2	hog 3	hog 4	hog 5	hog 6
P-value	$1.0 \cdot 10^{-10}$	$3.8 \cdot 10^{-10}$	$1.6 \cdot 10^{-7}$	$2.7 \cdot 10^{-3}$	$6.7 \cdot 10^{-4}$	$6.2 \cdot 10^{-6}$

Enrichment analysis:

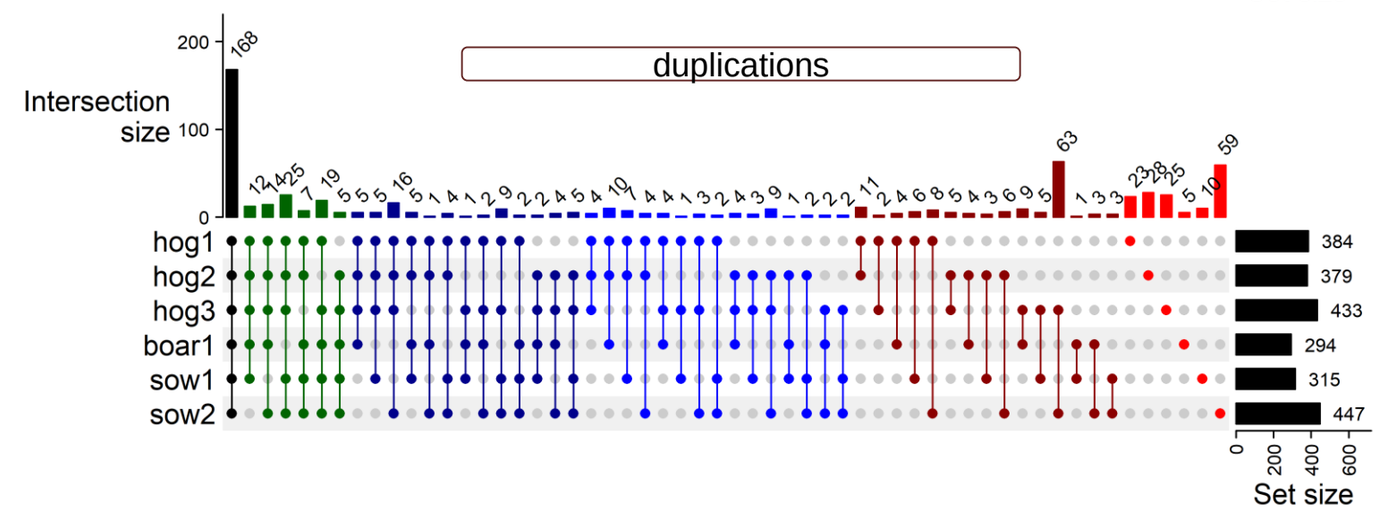
- Common duplications in all 12 pigs are related to the regulation of basic biological processes (metabolic and olfactory receptor activity, as well as G protein-coupled receptor signaling pathway)
- No significant results found for common deletions
- No significant results found for *de novo* CNVs

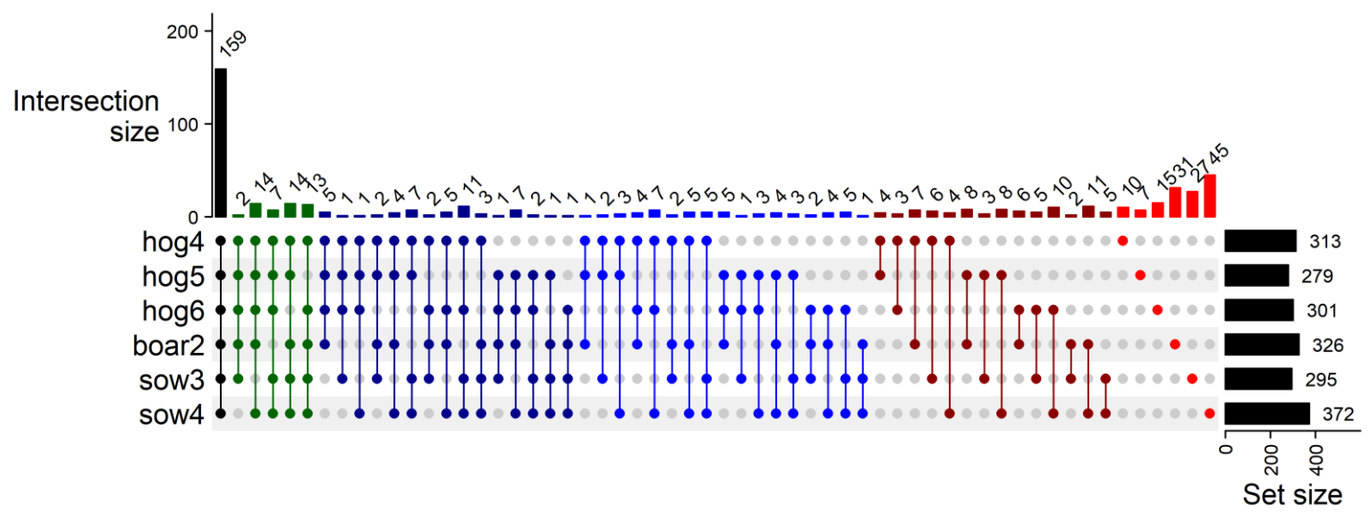
deletions





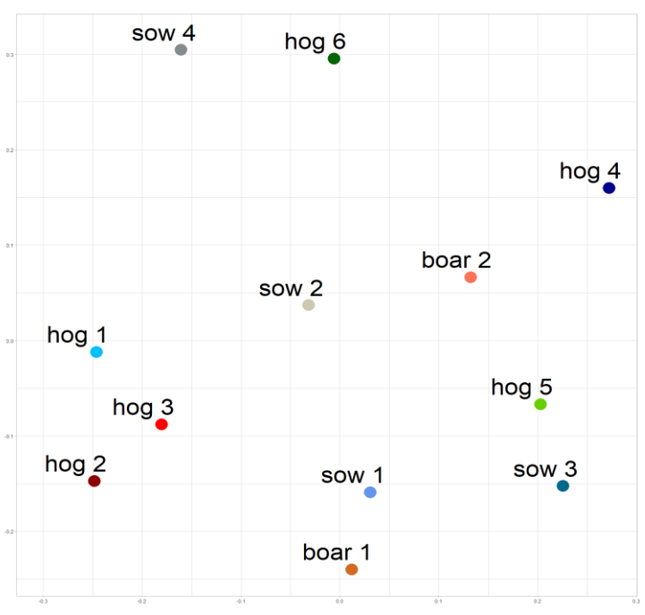
duplications



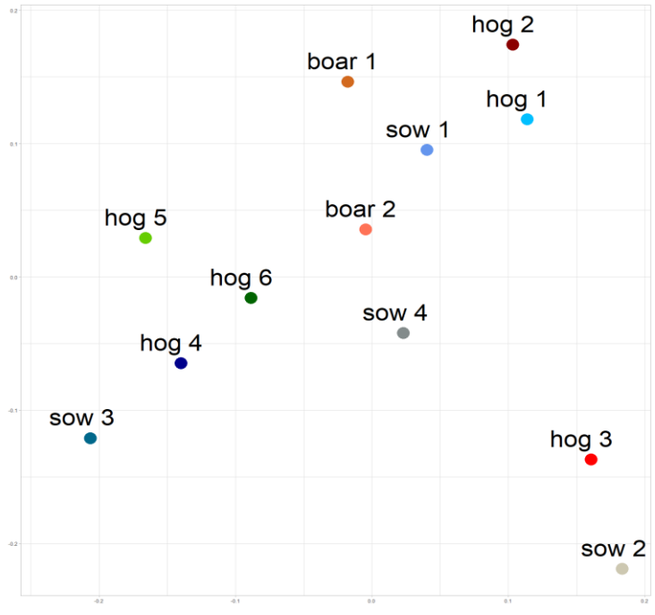


Multidimensional scalling

deletions



duplications



Acknowledgements: