FACULTY OF AGRICULTURAL SCIENCES
AARHUS UNIVERSITY

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Assessing porcine structural variation with array-CGH and massively parallel sequencing

João Fadista, Jakob Hedegaard, Lars-Erik Holm, Bo Thomsen, Christian Bendixen

Aarhus University, Blichers Allé 20, DK-8830 Tjele Denmark. Tel. +4589991280, Email: joao.fadista@agrsci.dk

Objectives

Our aim is to describe the structural genetic variation (including CNVs and indels) in the porcine genome with array comparative hybridization (aCGH) and massively parallel sequencing (MPS)

Background

CNVs are:

- known to be abundant in primates and rodents

- underlying factor in many disease/traits

- abundance/distribution unknown in farm animals

Methods

aCGH – 16 * 2.1M Nimblegen arrays of BoarX **vs.** 4 piglets and their 4 mothers

MPS – 12 Illumina sequencing runs from BoarX **vs.** Illumina data from sow used for pig assembly

CNV calling pipeline – figure 1

Genome assembly - Sus scrofa 9 (May 2009)

Results

Table 1 – Results for aCGH and CNV-seq platforms

	aCGH	CNV-seq
probes ... reads mapped	2.1M	497M / 501M
Probe spacing ... Read depth	474 bp	8X coverage
CNV resolution	2.4 kb	2.8 kb
Number of CNVs	86	122
False positive rate (based on Chr X)	5.0E-05	3.8E-03
False negative rate (based on Chr X)	0.76	0.26
indel detection	No	11 016
SNP detection	No	3.5 M
Cost	12 000 €	60 000 €

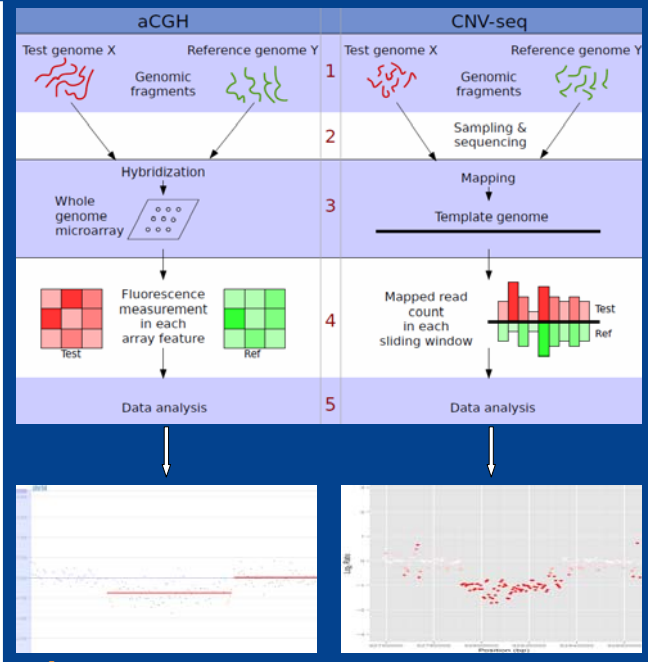

Figure 1 - Conceptual steps in aCGH and CNV-seq methods, with a CNV example

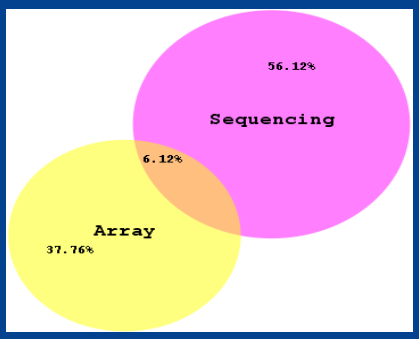

Figure 2 - CNVs from sequence and array data

Conclusions

- At 8X coverage depth, CNV-seq has smaller FPR and FNR, and provides broader view of genetic variation (indels and SNPs detection)

- At ≤ 8X coverage, aCGH is cheaper and detects smaller CNVs

- aCGH and CNV-seq are complementary techniques to assess structural variation