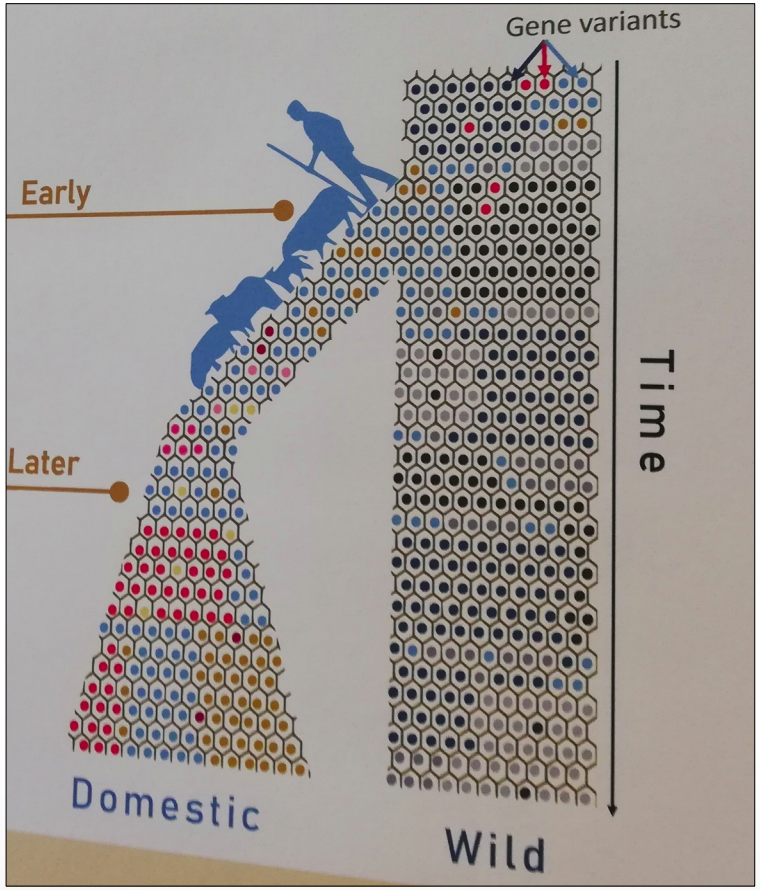
Which is the best sequencing option for the identification of deleterious alleles?

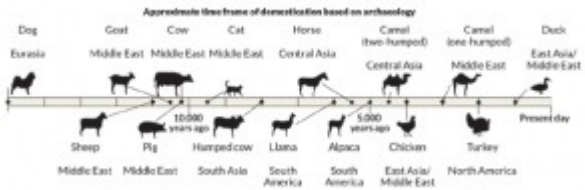
This question makes sens

Duhyadi Oliva García

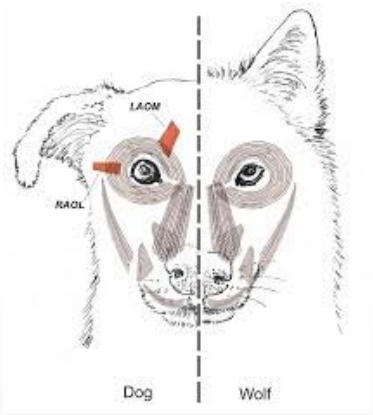
What is domestication?



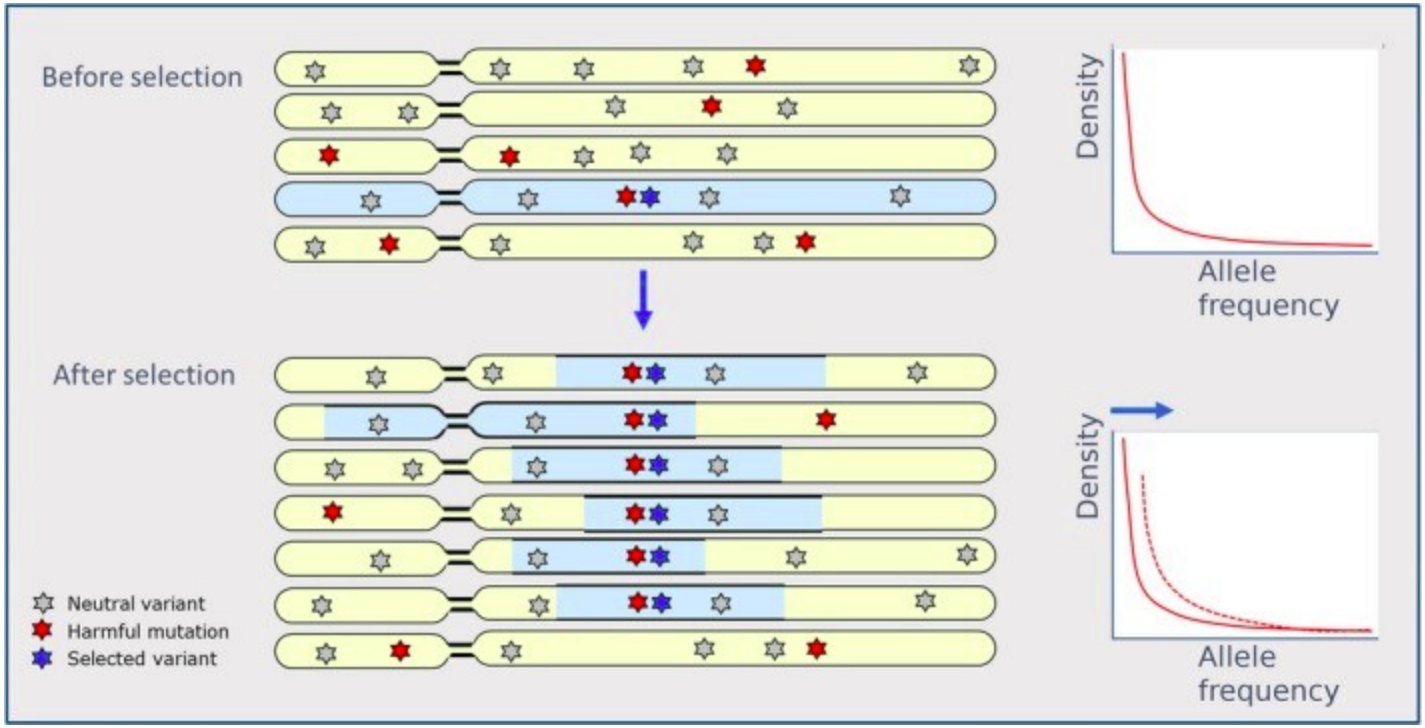
Charlote Her, Harlan 2019



It is acs continum of demographic and selective procesesses leading to organisms adapted to human need



Domestication cost: harmful mutation

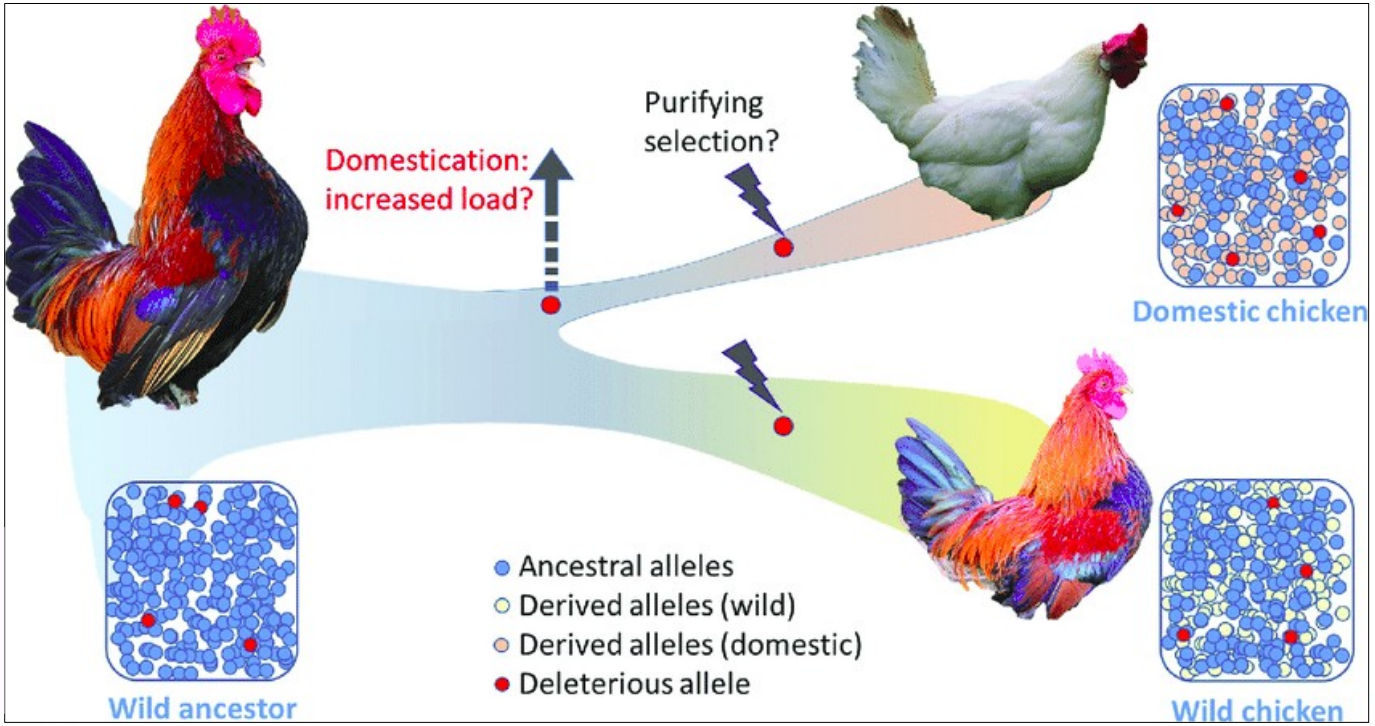


Boose *et al.* 2018 - online

Many mutations with a putative deleterious effect ***seem to be desired*** in the domestic setting

Example

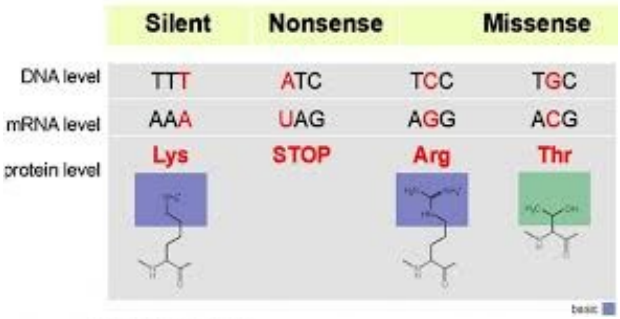
*20 million SNPs derived from **whole-genome sequences** from 127 animals*

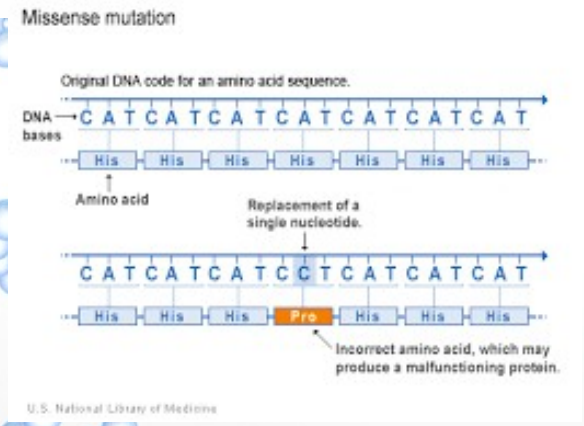


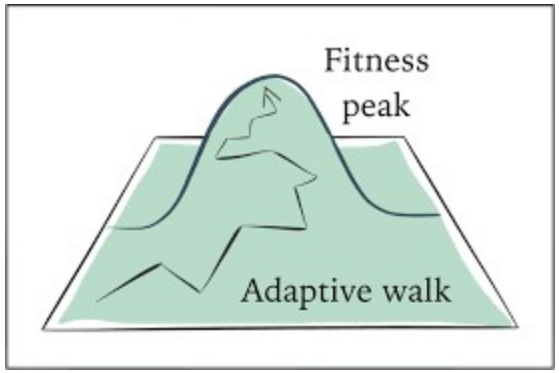
In this figure we can see that the loci are highly differentiated between wild and domestic chickens significantly lack missense mutations, which is indicative of purifying selection (Boose, M. 2019)

Definitions: missense mutations and purifying selection

Look for a needle in a haystack...

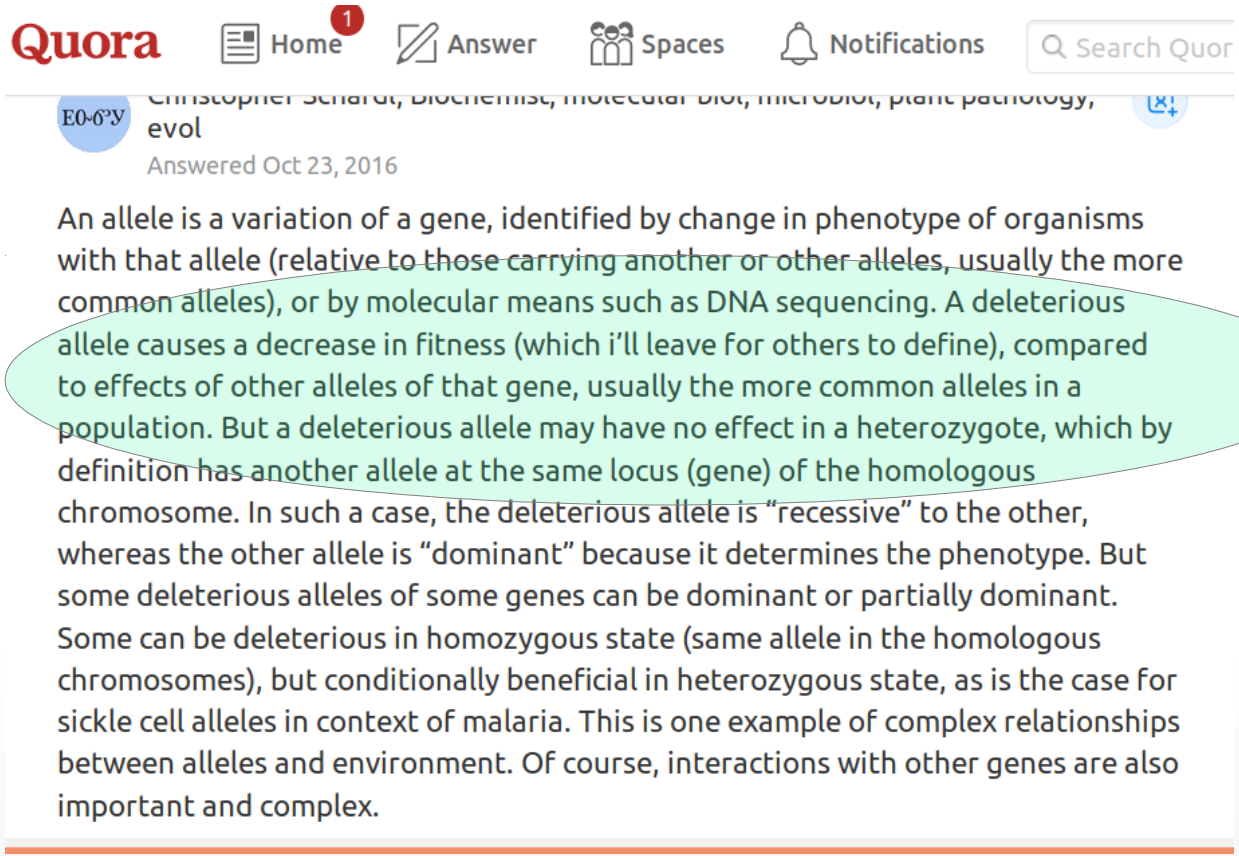






What is deleterious allele?

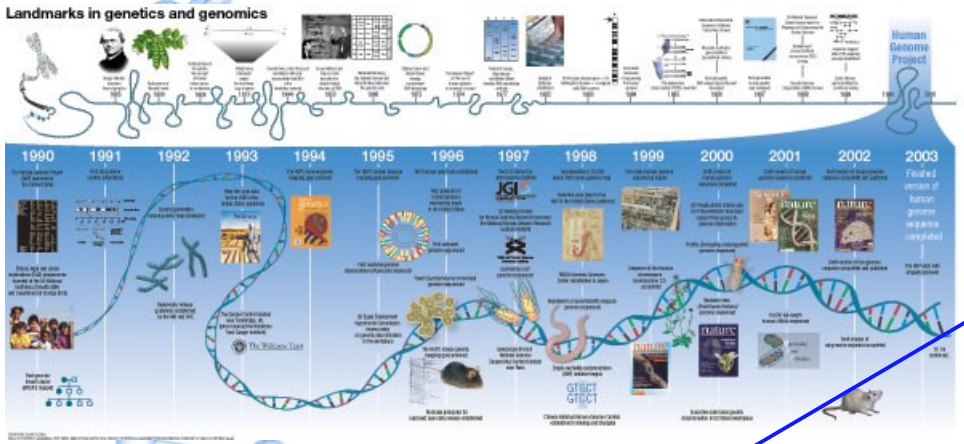
In very small populations, deleting alleles can be fixed



Questions?

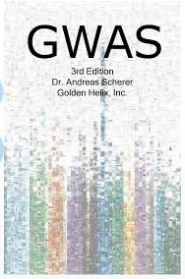
Which is the best option for the identification of deletereos alleles?

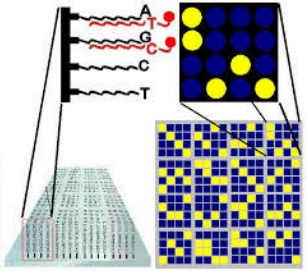
Currently the best option would be WGS?



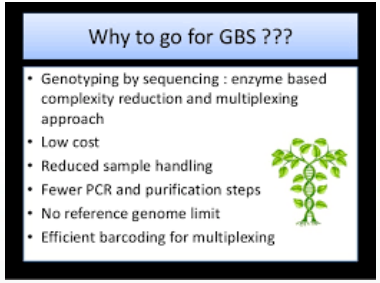
2009 — **2014** — **Today**

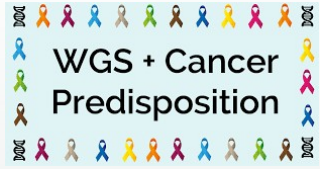
What slows to use it would be the high cost and bioinformatic work time



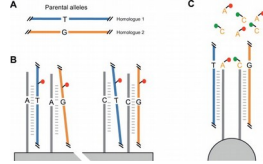


SNP array





Today SNP array



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SNP array data analysis

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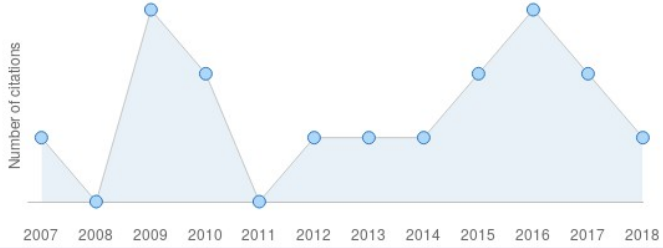
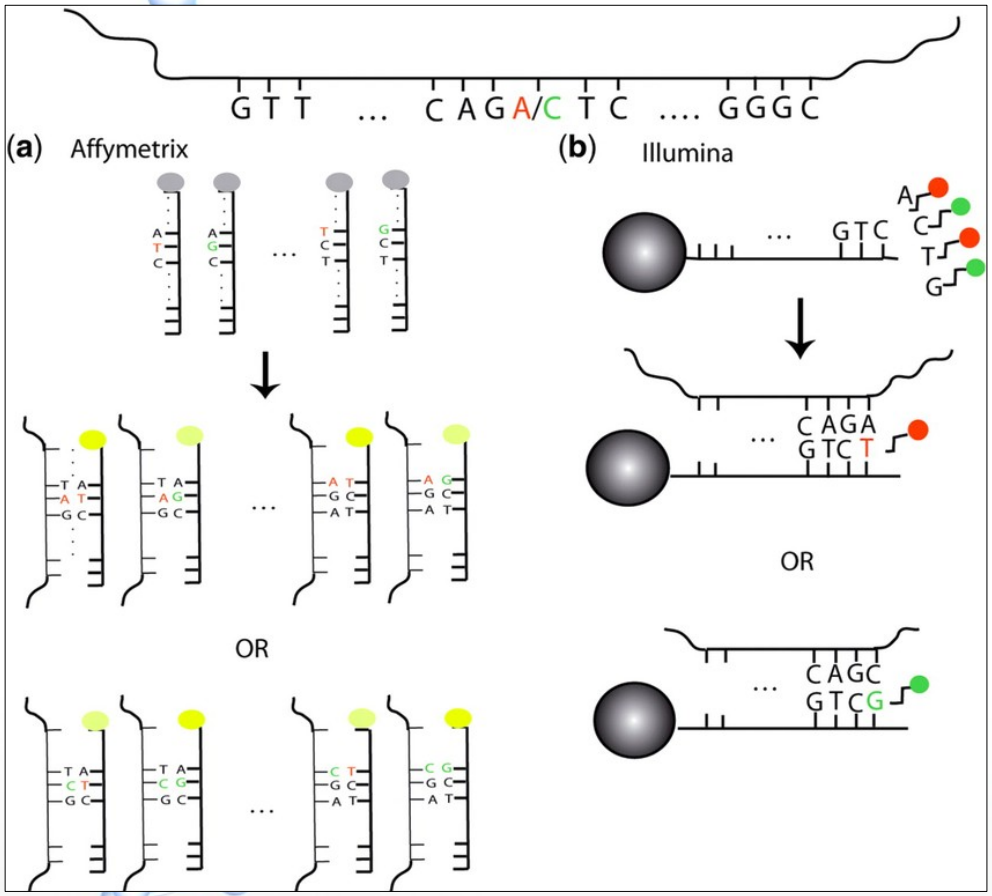
Full analysis of SNP array data analysis citations



The latest datasets generated from SNP array data analysis

Dataset title	Year
Copy number variation in fetal al...	11-2018
CSMD1 shows complex somatic ...	10-2018
CSMD1 shows complex somatic ...	10-2018

SNP array



LaFramboise T, 2019

Overview of SNP array technology. At the top is the fragment of DNA harboring an A/C SNP to be interrogated by the probes shown. (a) In the Affymetrix assay, there are 25-mer probes for both alleles, and the location of the SNP locus varies from probe to probe. The DNA binds to both probes regardless of the allele it carries, but it does so more efficiently when it is complementary to all 25 bases (bright yellow) rather than mismatching the SNP site (dimmer yellow). This impeded binding manifests itself in a dimmer signal. (b) Attached to each Illumina bead is a 50-mer sequence complementary to the sequence adjacent to the SNP site. The single-base extension (T or G) that is complementary to the allele carried by the DNA (A or C, respectively) then binds and results in the appropriately-colored signal (red or green, respectively). For both platforms, the computational algorithms convert the raw signals into inferences regarding the presence or absence of each of the two alleles.

More questions...There are significant differences between GBS and SNP array?

Negro *et al. BMC Plant Biology* (2019) 19:318
https://doi.org/10.1186/s12870-019-1926-4

BMC Plant Biology

RESEARCH ARTICLE Open Access

Genotyping-by-sequencing and SNP-arrays are complementary for detecting quantitative trait loci by tagging different haplotypes in association studies

Sandra S. Negro[1], Emilie J. Millet[2,3], Delphine Madur[1], Cyril Bauland[1], Valérie Combes[1], Claude Welcker[2], François Tardieu[2], Alain Charcosset[1] and Stéphane D. Nicolas[1*]

In this article they analyze the impact of marker density and genotyping technologies (sequencing vs array) on (i) the estimates of relatedness and population structure, and (ii) the detection of QTLs

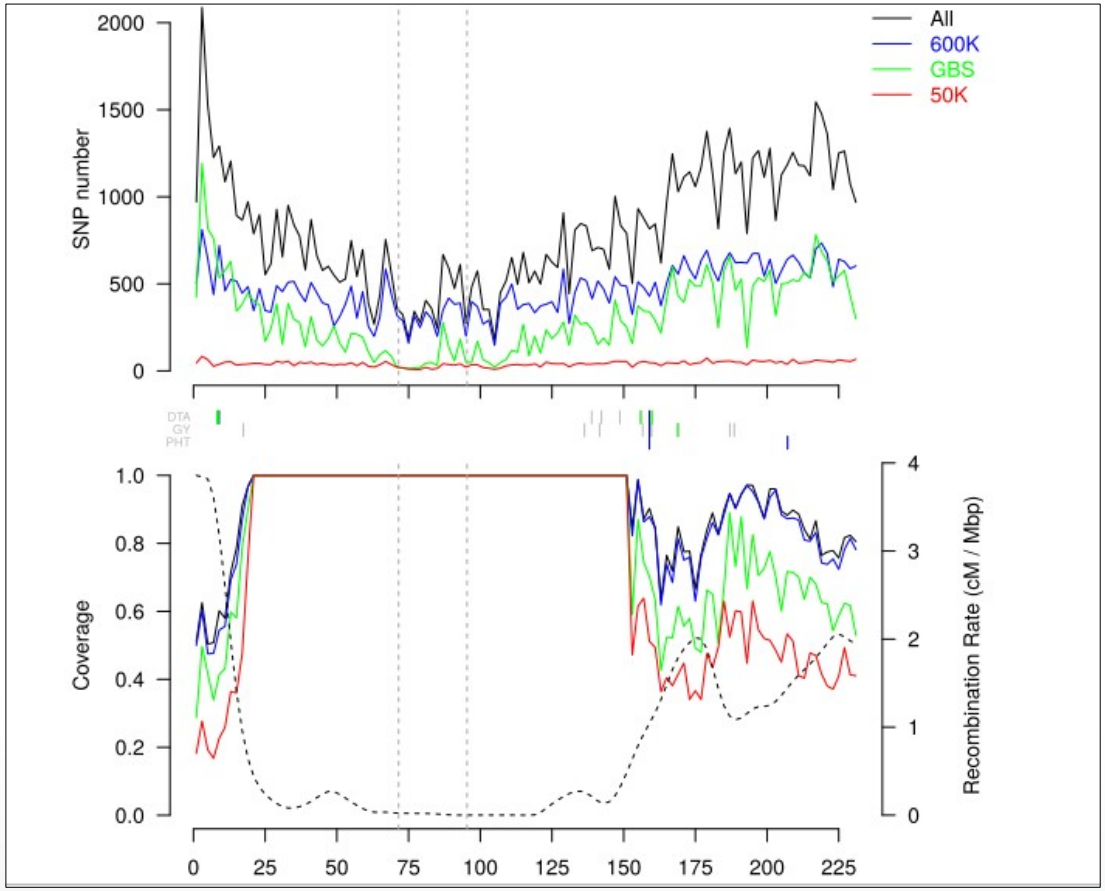
SNP array & GBS

- Consistent with MAF distribution, the average gene diversity (He) was lower for GBS(0.27)than for arrays (0.35 and 0.34 for the 50 K and 600 K arrays, respectively).
- The distribution of SNP residual heterozygosity of inbred lines was similar for the three technologies, with a mean of 0.80, 0.89 and 0.22% for the 50 K, 600 K and GBS, respectively.
- The distribution of the SNPs along the genome was denser in the telomeres for the GBS and in the peri-centromeric regions for the 600 K, whereas the 50 K exhibited a more uniform distribution .

Independent of the sequencing technique

- Furthermore, minor allele frequencies (MAF), population stratification and cryptic relatedness are three other important param- eters affecting power and false positive detection.

Results from article



Variation of the markers density

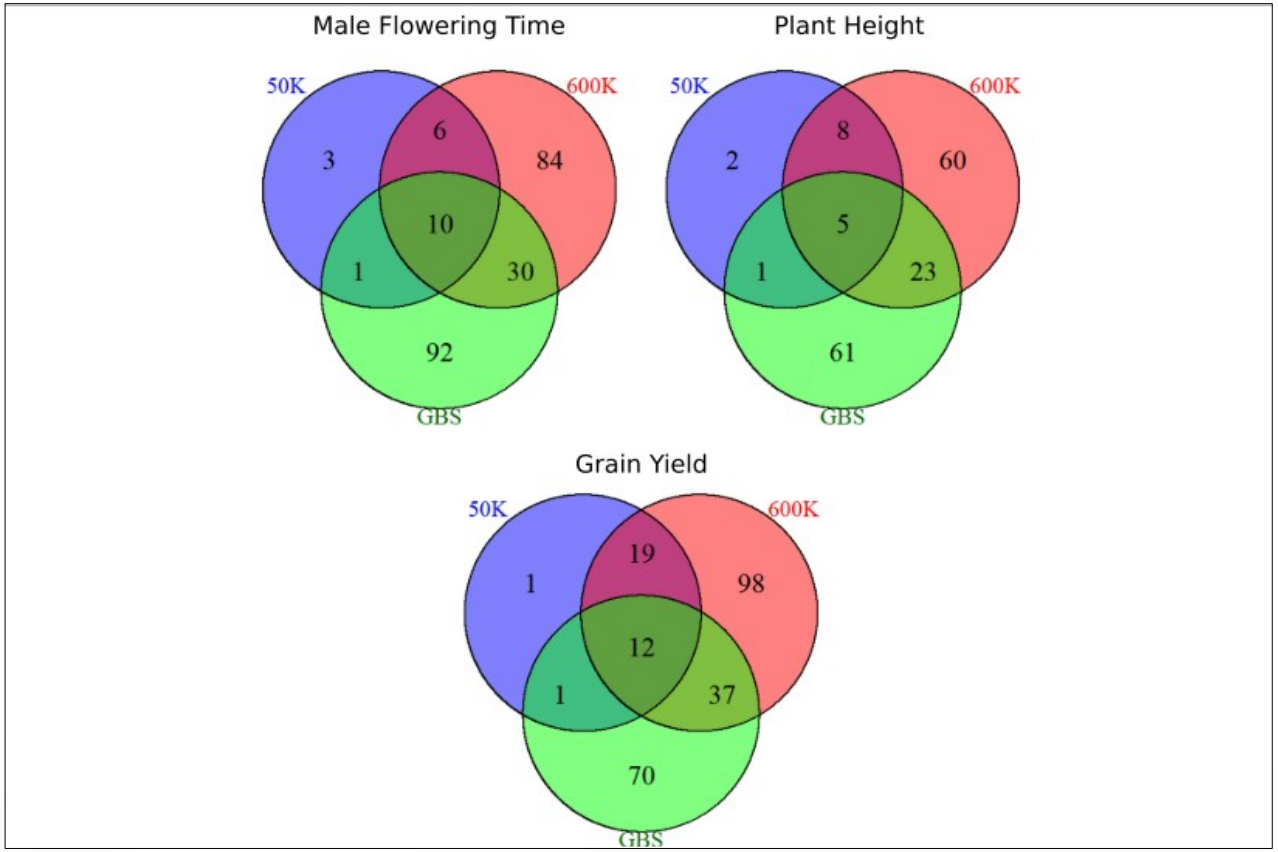
Results from article

Table 4 Comparison of associated SNPs and QTLs detected between traits and three technologies

		Significant SNPs				QTLs			
Technology		50 K	600 K	GBS	ALL	50 K	600 K	GBS	ALL
Marker Nb		42046	459191	308929	810580	42046	459191	308929	810580
Total Nb	DTA	52	759	345	1115	20	130	133	226
	plantHT	68	778	299	1061	16	96	90	160
	GY	123	1416	538	1941	33	166	120	238
	Per trait	81	984	394	1372	23	131	114	208
Average per envir.	DTA	2.4	34.5	15.7	50.7	0.9	5.9	6.0	10.3
	plantHT	3.1	35.4	13.6	48.2	0.7	4.4	4.1	7.3
	GY	5.6	64.4	24.5	88.2	1.5	7.5	5.5	10.8
	Per trait	3.7	44.7	17.9	62.4	1.0	5.9	5.2	9.5

QTLs were obtained by grouping associated SNPs with overlapping LD windows (LD_win) for the three traits (*DTA* male flowering time, *PlantHT* plant height, *GY* grain yield). "Marker Nb" indicates the number of markers tested in GWAS. "Total number": is the sum of associated SNPs or QTLs across environments. "Average per envir" indicates the average number of QTLs obtained in 22 environments for three traits (66 trait-environments combinations)

Results from article



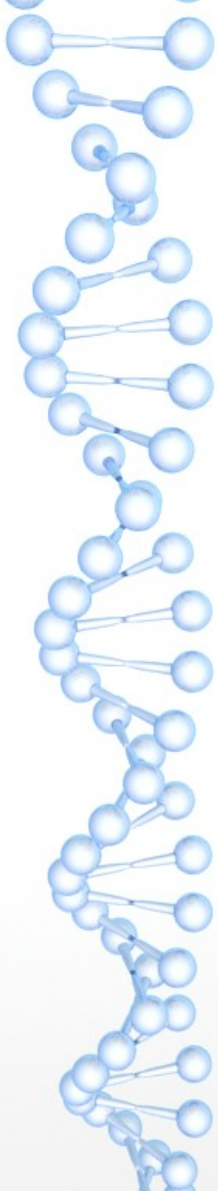
Complementarity of the three technologies to detect QTLs

SNP array as option a low cost

Check for updates

Development and Applications of a High Throughput Genotyping Tool for Polyploid Crops: Single Nucleotide Polymorphism (SNP) Array

Qian You[1,2], Xiping Yang[2], Ze Peng[2], Liping Xu[1] and Jianping Wang[2,3,4]**



Thanks