

Genome Wide Association Studies- Case Study

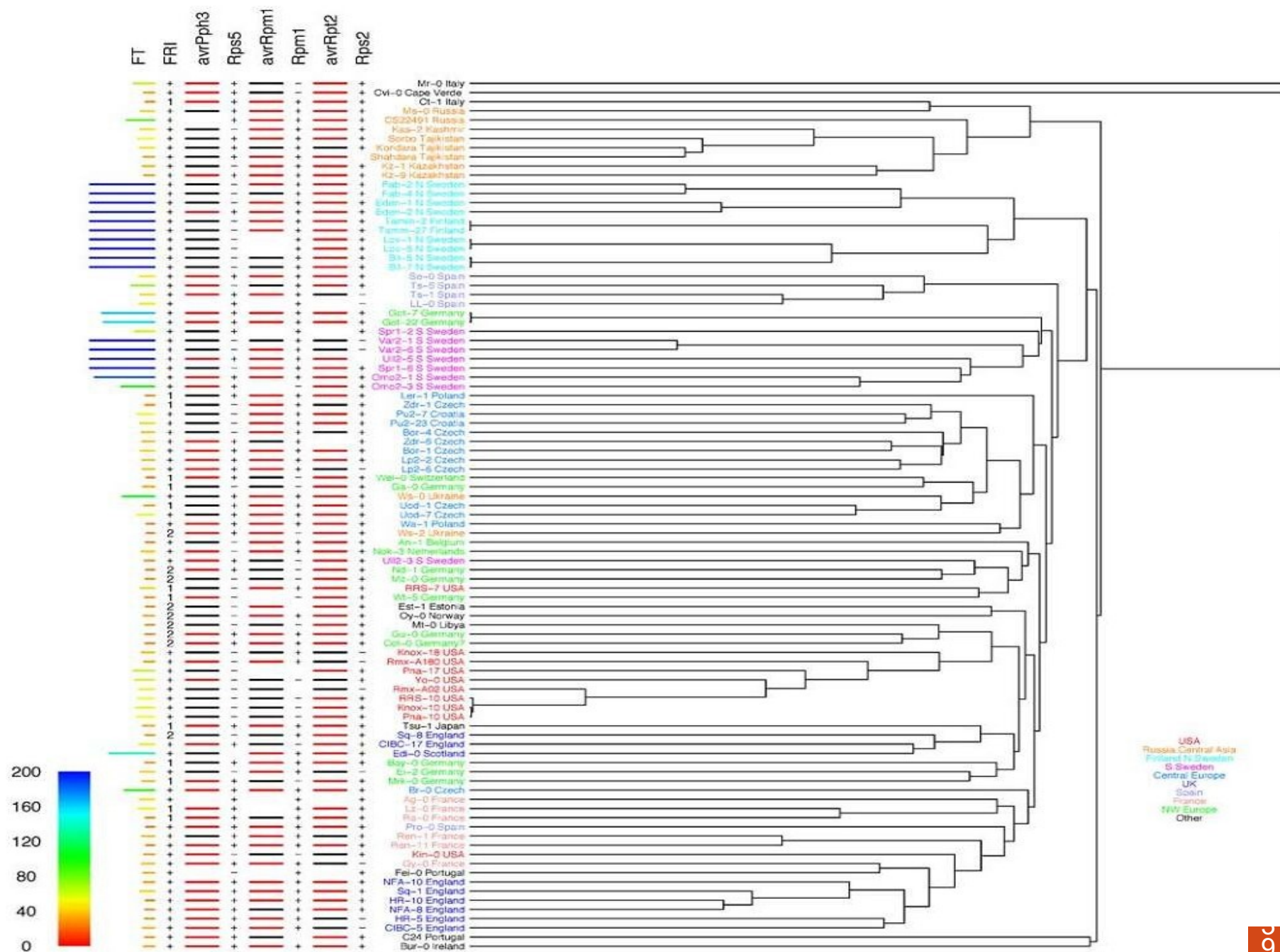
Genome Wide Association Study

- ❑ Genomic mapping is a useful approach in plant breeding to locate regions that are important for particular traits (Grotewold *et al.* 2015).
- ❑ The more recently developed genome-wide association studies (GWASs) can identify precise chromosomal locations with the resolution to define specific genes or even polymorphisms within coding regions.
- ❑ In general, these approaches employ statistical formalisms to determine the strength of the association between a genotype and phenotype and as such help inform breeding by providing molecular markers or by identifying genes and alleles that contribute to specific traits.
- ❑ In association mapping the genetic markers usually lie

Genome-Wide Association Mapping in *Arabidopsis* Identifies Previously Known Flowering Time and Pathogen Resistance Genes

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- They considered 4 phenotypes for which the major loci were known.
- Vernalization response locus-FRI.
- 3 pathogen resistant loci-Rpm1, Rps2, Rps5.



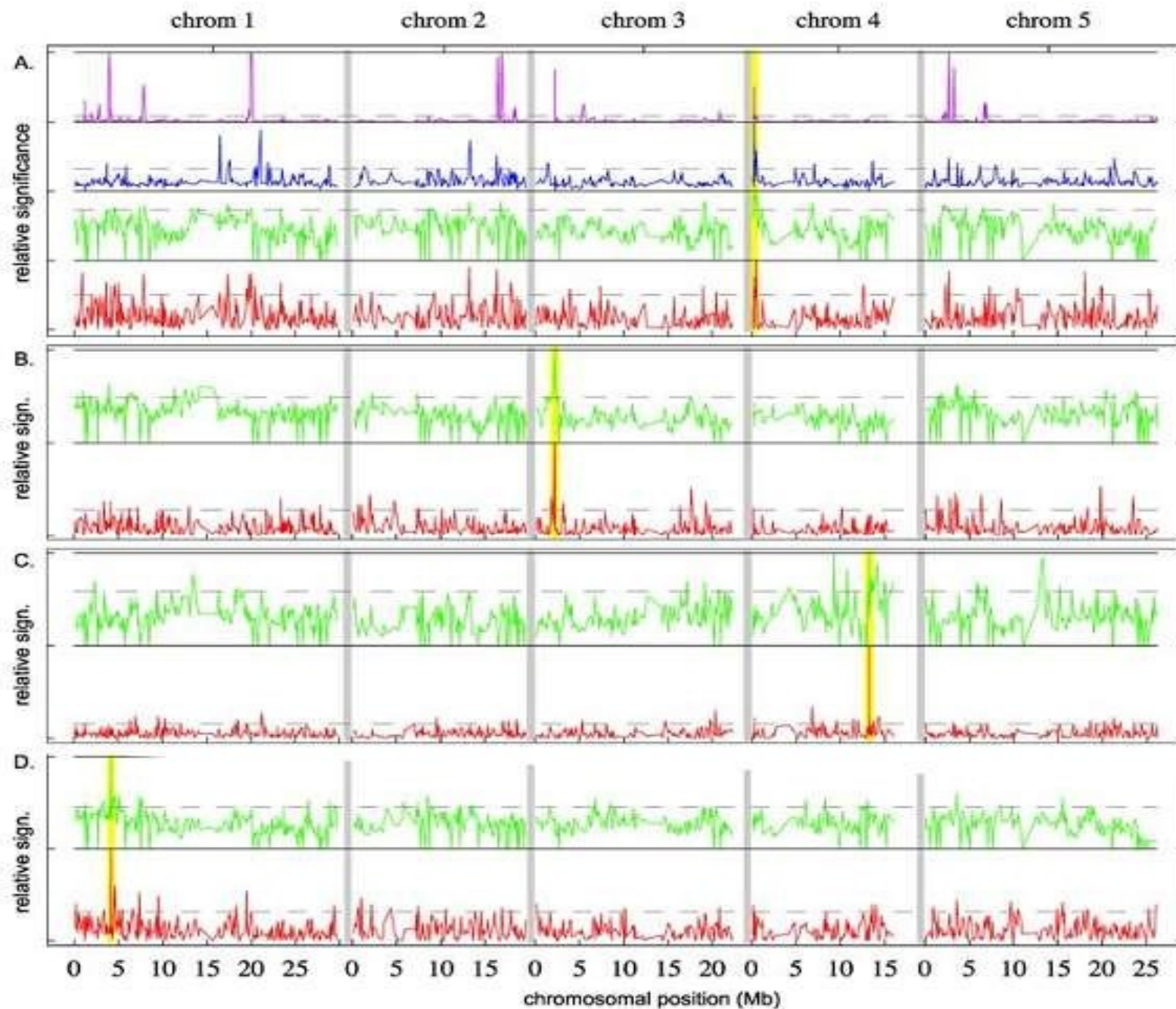
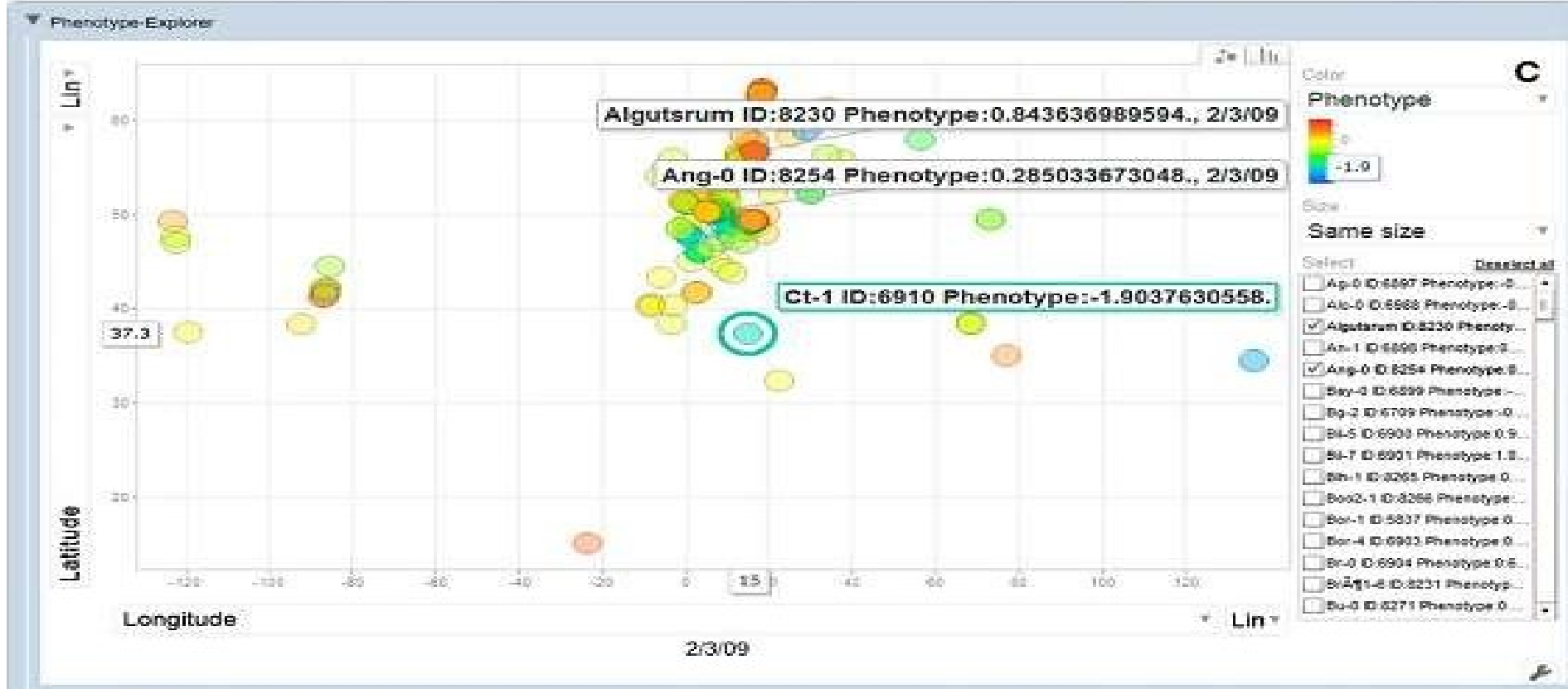
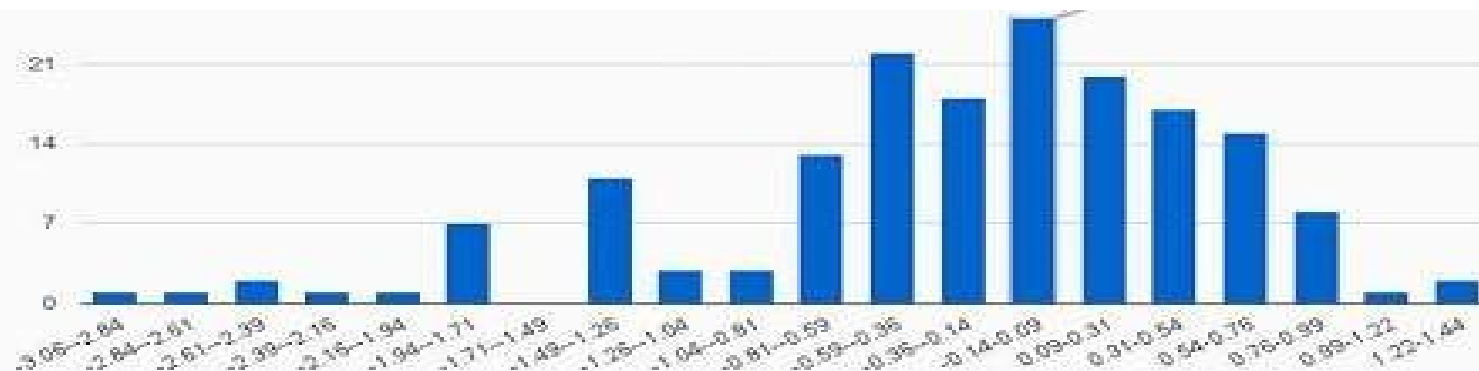


Figure 3. Genome-Wide Scans for Association with Flowering Time and Pathogen Resistance

Figure 1. Summary of the Data Used in the Study



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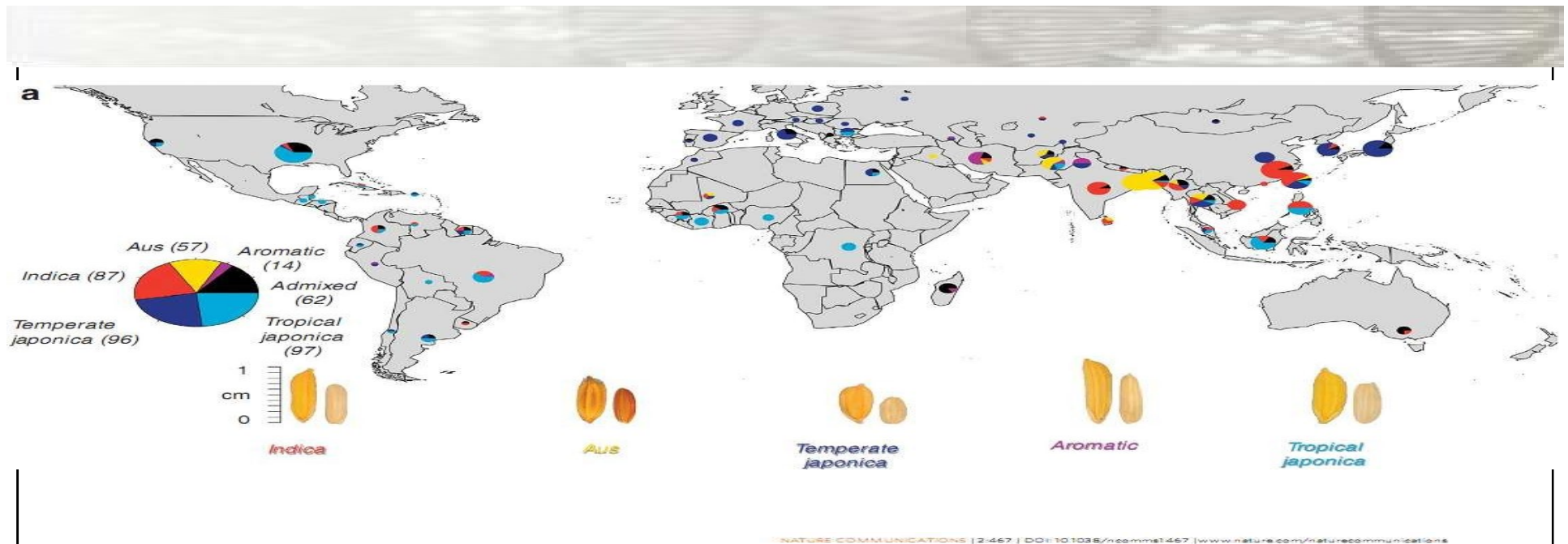
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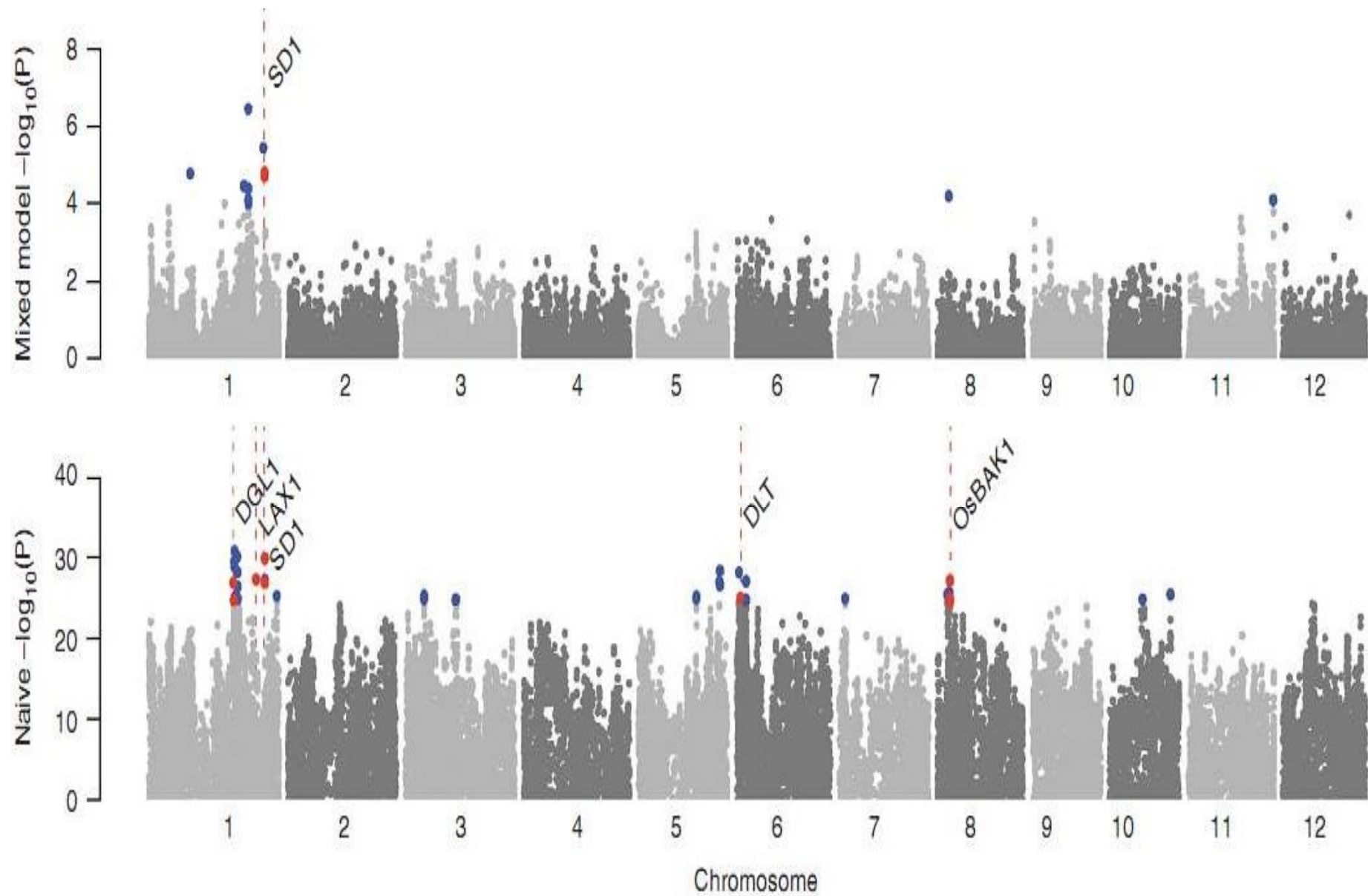
Genome-wide association mapping reveals a rich genetic architecture of complex traits in *Oryza sativa*

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Results

- Correlation analysis between different phenotypes.
- Correlation ranged from -0.41 to 0.9 (seed width and length).



Genome-wide association study (GWAS) of resistance to head smut in maize

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- Smut (*Sphacelotheca reiliana*) study using Illumina maize SNP 50 array.
- 45868 SNPs in the panel of 144 inbred lines.
- Classified candidate genes as resistant genes, disease response genes and other genes.
- **Outcome:**
- 50K SNP offers highest array.
- Chromosomal region and specific SNP that affect resistance level in commercial build up population.
- Assessed the extent of LD in target population.
- Identified genes or QTLs that significantly affect smut resistance.
- Characterized those genes based on known function and co-location.

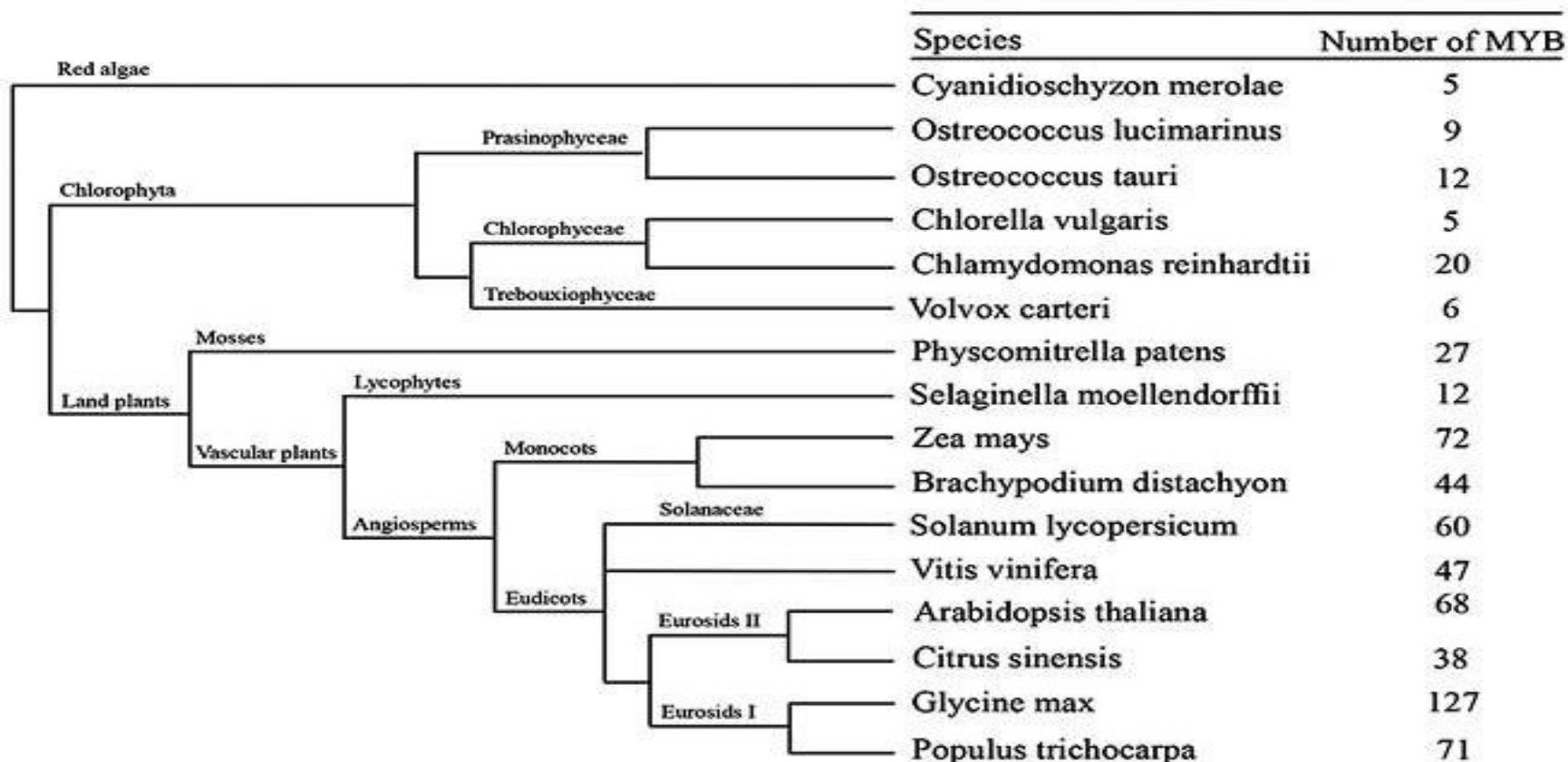
Physical positions of 19 SNPs significantly associated with head smut resistance and the predicted function or homology of adjacent candidate genes.

SNP	Chr.	Physical position (AGPv1 bp)	bin	r ²	P	Candidate genes	Function	Class	Previous studies
1	1	278884507	1.09	0.046	3.84E–07 ^a	GRMZM2G300990	Serine/threonine protein kinase	Plant defense response	
2	2	56673949	2.04	0.043	5.54E–17 ^a	GRMZM2G434669	Antifreeze	Plant defense response	
3	2	201359446	2.08	0.051	6.71E–06	GRMZM2G140231	Serine/threonine protein kinase	Plant defense response	Gao (2005)
4	2	219834173	2.09	0.093	4.05E–06	GRMZM2G166566	Basic-leucine zipper transcription factor	Plant defense response	Gao (2005), Chen et al. (2008), Li et al. (2008)
5	3	124139795	3.05	0.074	1.52E–08 ^a	GRMZM2G137289	MADS-box	Plant defense response	Lübberstedt et al. (1999), Li et al. (2008)
6	4	220145226	4.08	0.036	6.31E–41 ^a	GRMZM2G046816	Tubby-like	Plant defense response	
7	5	187810205	5.05	0.043	6.87E–06	GRMZM2G139858	Antifreeze	Plant defense response	
8	5	188373740	5.05	0.035	5.97E–07 ^a	GRMZM2G312274	Auxin	Plant defense response	
9	6	163988192	6.06	0.047	2.25E–08 ^a	AC195860.3.FG002	Unknown	Unknown	
10	7	3672618	7.01	0.038	9.00E–06	GRMZM2G465226	Pathogenesis-related protein	Disease resistance	Liet al. (2008)
11	8	103237278	8.03	0.036	1.39E–06	GRMZM2G047152	NB-ARC	Disease resistance	Lübberstedt et al. (1999)
12	8	131978501	8.05	0.045	4.86E–06	GRMZM2G075000	Actin cross link	Plant defense response	
13	8	136910174	8.05	0.039	3.74E–06	GRMZM2G164341	Antifreeze protein	Plant defense response	
14	8	136948482	8.05	0.039	1.16E–10 ^a	GRMZM2G164341	Antifreeze protein	Plant defense response	
15	8	139906939	8.05	0.041	1.05E–05	GRMZM2G017603	NB-ARC	disease resistance	
16	9	84924940	9.03	0.037	4.58E–06	GRMZM2G443953	WD40 repeat	Plant defense response	Lübberstedt et al. (1999), Lu and Brewbaker (1999)
17	9	85140137	9.03	0.035	2.45E–07 ^a	GRMZM2G383607	Unknown	Unknown	Lübberstedt et al. (1999), Lu and Brewbaker (2000)
18	9	90505475	9.03	0.050	4.24E–06	GRMZM2G348666	aminoacyl-tRNA ligase activity	Plant defense response	Lübberstedt et al. (1999), Lu and Brewbaker (2001)
19	10	137723733	10.05	0.037	1.01E–05	GRMZM2G117667	Lipase	Disease resistance	

r², proportion of phenotypic variance explained by SNP.^a Significant SNP-trait associations with a stringent threshold of $-\log(0.05/45,868)$.

Genome-Wide Identification and Evolutionary and Expression Analyses of MYB-Related Genes in Land Plants

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ORIGINAL ARTICLE

Open Access

Genome wide association studies for *japonica* rice resistance to blast in field and controlled conditions

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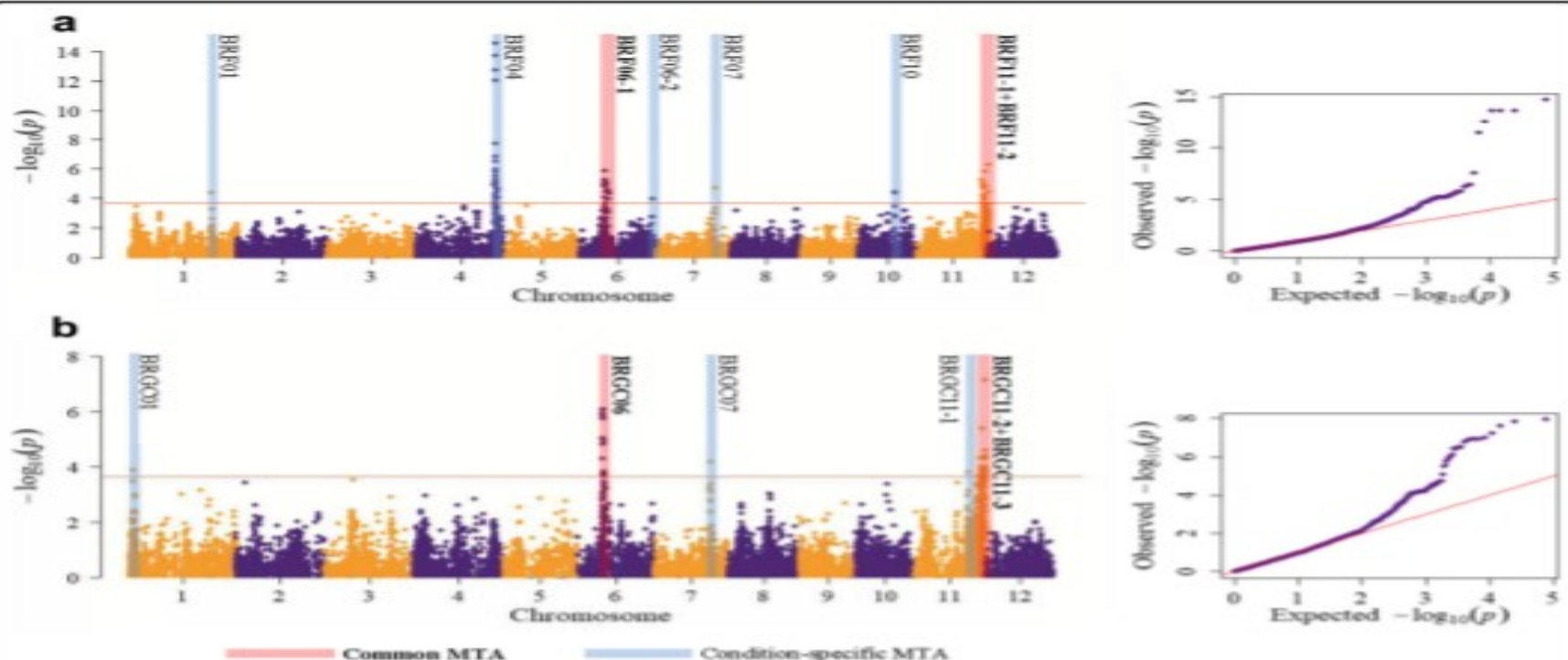
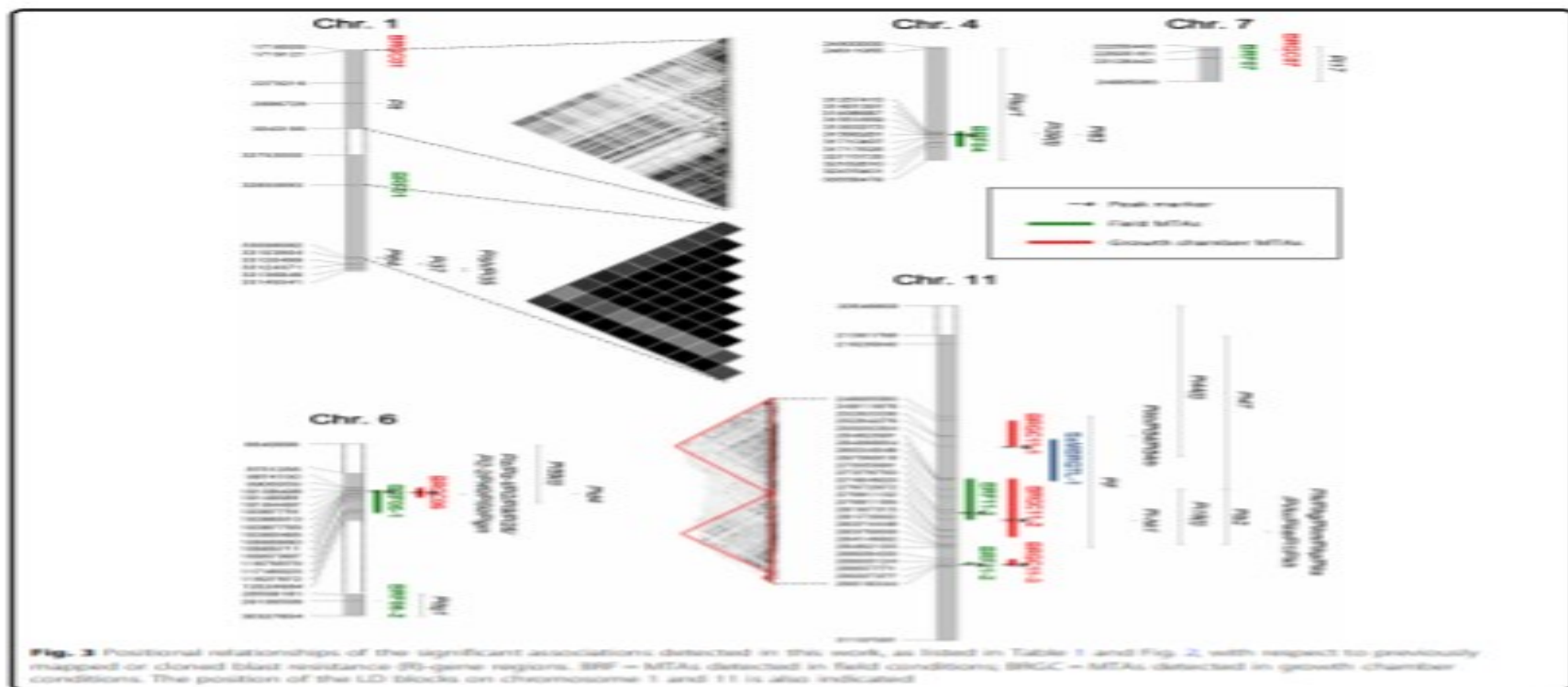


Fig. 2 Results of the association analysis for field (**a**) and growth chamber (**b**) evaluations. Left panel: Manhattan plots of the markers associated with rice blast disease resistance. X axis shows markers along the 12 rice chromosomes and Y axis shows the negative log10-transformed p-values for each association. MTAs detected in both conditions are red-shaded and named in bold; conditions-specific MTAs are blue shaded. Full redlines indicate the 0.05 FDR threshold. Right panel: Q-Q plots of the $-\log_{10}(p)$ of the markers from association analyses



Outcome:

- ✓The GWAS approach identified 14 MTAs associated to blast resistance. Five of them were detected only under field conditions, while three were discovered only for the growth chamber experiment.
- ✓The screening procedures identified the largest number of resistant accessions in the cluster of tropical japonica and out-group varieties, though highly resistant temperate japonica genotypes were also identified.

Thank you!

