

# Introduction to **GWAS**

## Description of Datasets

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OscarGenomics



# Outline

1. Continuous phenotypes: rice dataset
2. Binary phenotype: dogs dataset



# Rice data (*Oryza sativa*)

Continuous phenotypes: **plant height (PH)**



RESEARCH ARTICLE

## Genome-Wide Association Study for Traits Related to Plant and Grain Morphology, and Root Architecture in Temperate Rice Accessions

Filippo Biscarini<sup>1\*</sup>, Paolo Cozzi<sup>2</sup>, Laura Casella<sup>1a</sup>, Paolo Riccardi<sup>1</sup>, Alessandra Vattari<sup>1</sup>, Gabriele Orasen<sup>3</sup>, Rosaria Perrini<sup>3</sup>, Gianni Tacconi<sup>4</sup>, Alessandro Tondelli<sup>4</sup>, Chiara Biselli<sup>3</sup>, Luigi Cattivelli<sup>4</sup>, Jennifer Spindel<sup>5</sup>, Susan McCouch<sup>5</sup>, Pamela Abbruscato<sup>1</sup>, Giampiero Valé<sup>3,4</sup>, Pietro Piffanelli<sup>1</sup>, Raffaella Greco<sup>1</sup>

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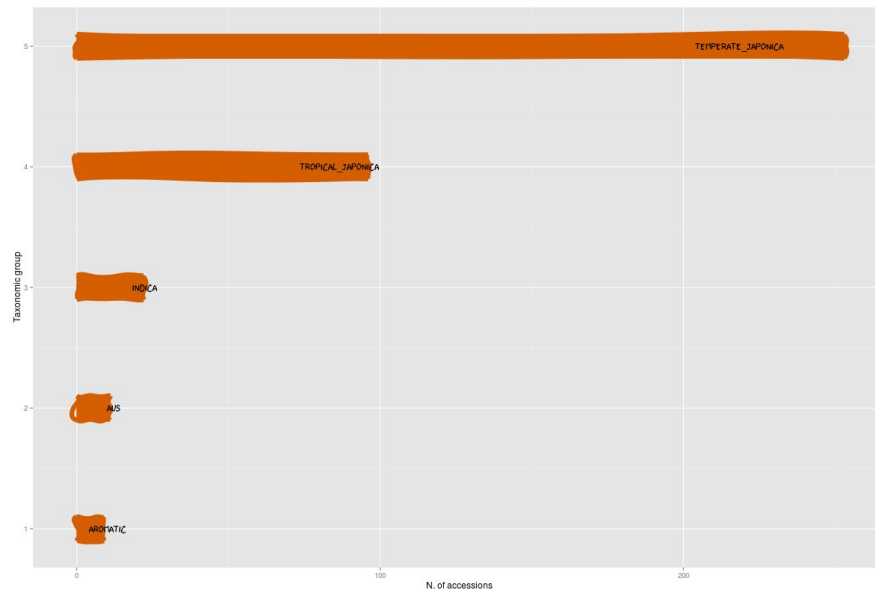


# Rice data (*Oryza sativa*)

Continuous phenotypes: **plant height (PH)**

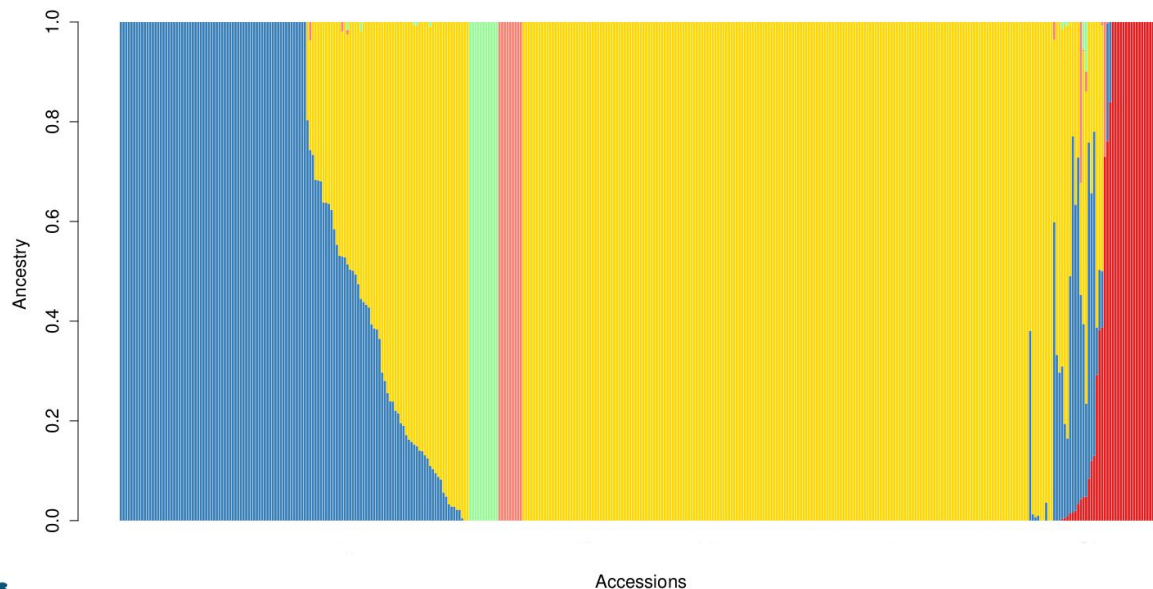
- genotype data from **GBS**
- ~ 400 rice accessions from 5 sub-populations:

- *temperate japonica*
- *tropical japonica*
- *indica*
- *aus*
- *aromatica*



# Rice data (*Oryza sativa*)

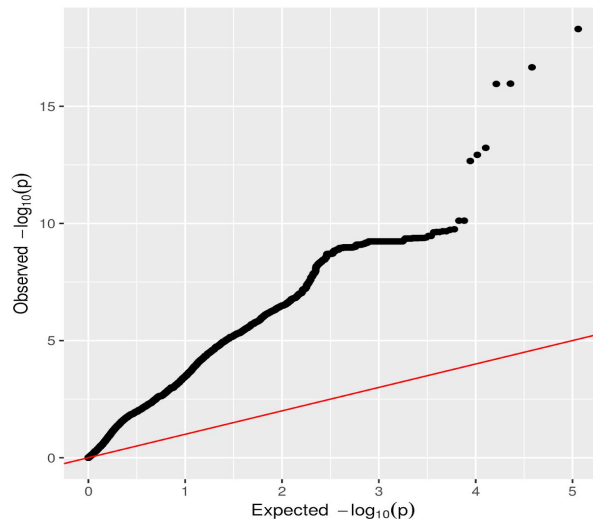
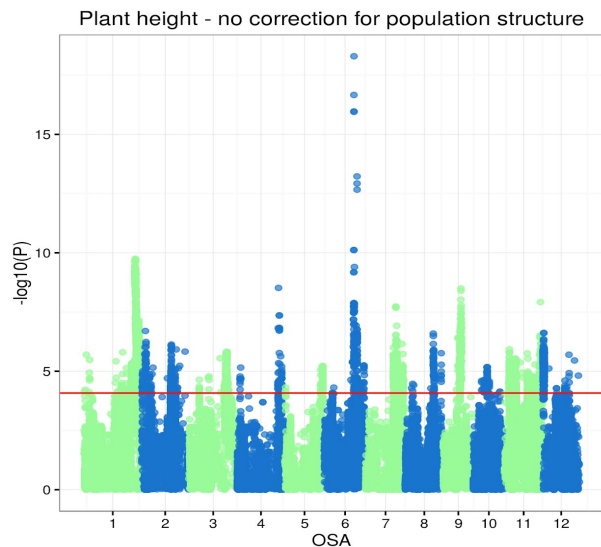
Continuous phenotypes: **plant height (PH)**



- blue = tropical japonica
- yellow = temperate japonica
- green = aus
- pink = aromatic
- red = indica

# Rice data (*Oryza sativa*)

Continuous phenotypes: **plant height (PH)**

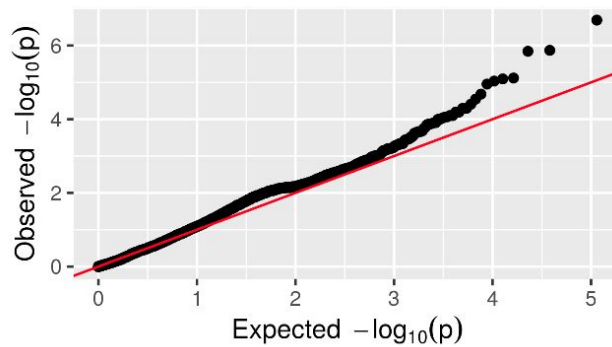
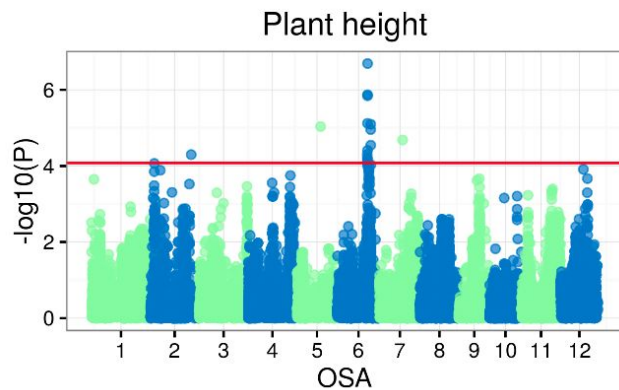


12 chromosomes

No correction for  
population structure

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Continuous phenotypes: **plant height (PH)**

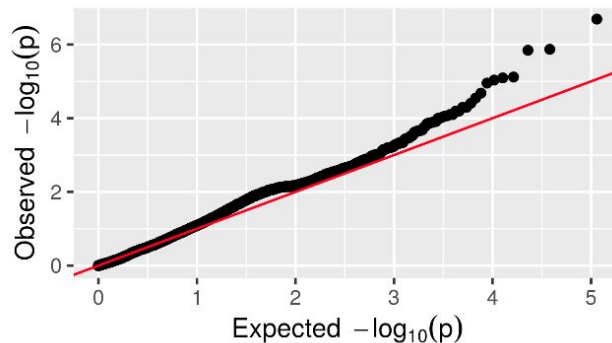
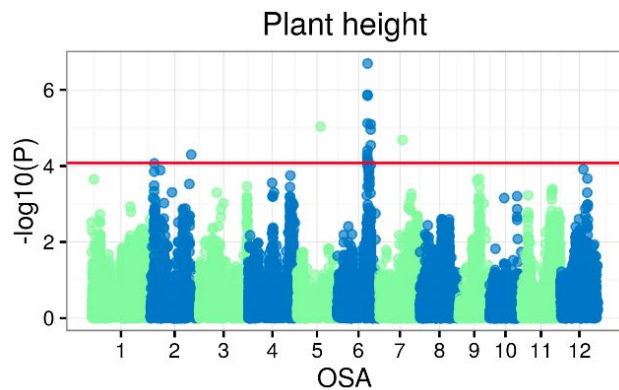


12 chromosomes

Correction for population structure

# Rice data (*Oryza sativa*)

Continuous phenotypes: **plant height (PH)**



12 chromosomes

Correction for population structure

We take chromosomes 1, 2, 6 and 7



# Dogs data (*Canis familiaris*)

Binary phenotype: **cleft lip** (presence/absence)



## RESEARCH ARTICLE

# Genome-Wide Association Studies in Dogs and Humans Identify *ADAMTS20* as a Risk Variant for Cleft Lip and Palate

Zena T. Wolf<sup>1☯</sup>, Harrison A. Brand<sup>2,3☯a</sup>, John R. Shaffer<sup>3☯</sup>, Elizabeth J. Leslie<sup>2</sup>, Boaz Arzi<sup>4</sup>, Cali E. Willet<sup>5</sup>, Timothy C. Cox<sup>6,7,8</sup>, Toby McHenry<sup>2</sup>, Nicole Narayan<sup>9</sup>, Eleanor Feingold<sup>3</sup>, Xiaojing Wang<sup>2ab</sup>, Sandra Sliskovic<sup>1</sup>, Nili Karmi<sup>1</sup>, Noa Safra<sup>1</sup>, Carla Sanchez<sup>2</sup>, Frederic W. B. Deleyiannis<sup>10</sup>, Jeffrey C. Murray<sup>11</sup>, Claire M. Wade<sup>5</sup>, Mary L. Marazita<sup>2,12†\*</sup>, Danika L. Bannasch<sup>1†\*</sup>

# Dogs data (*Canis familiaris*)

Binary phenotype: **cleft lip** (presence/absence)

- Nova Scotia Duck Tolling Retriever (NSDTR)
- 125 dogs:
  - 13 cases
  - 112 controls



# Dogs data (*Canis familiaris*)

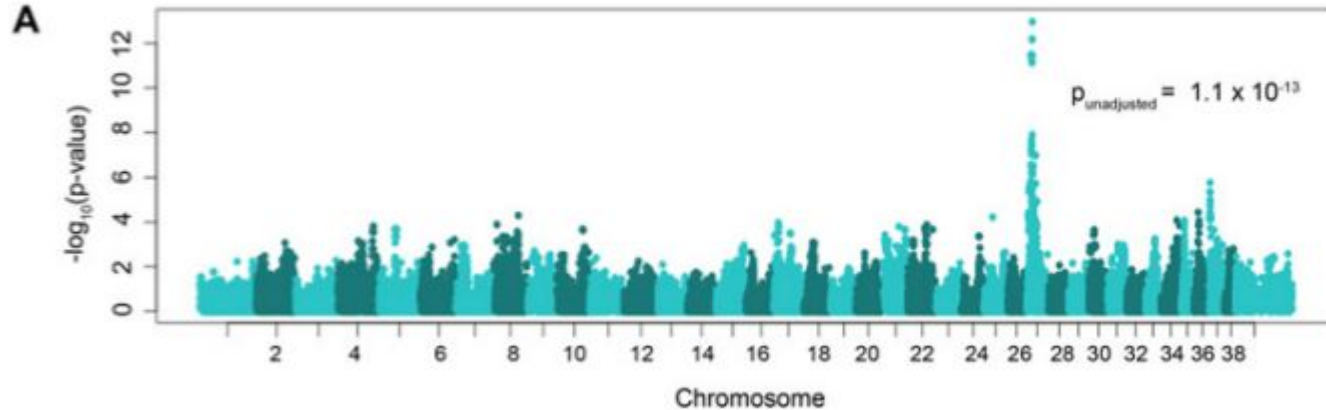
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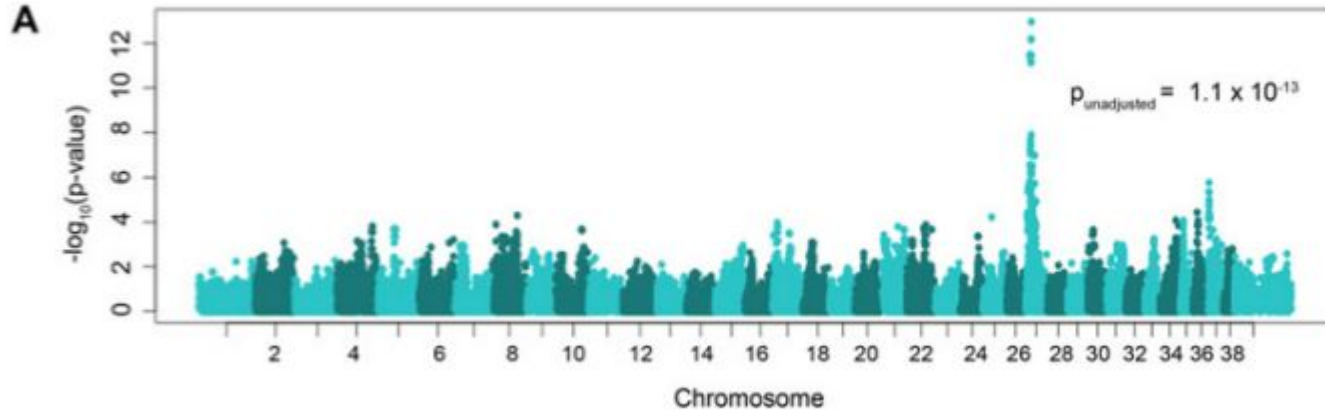


39 chromosomes

Strong signal of  
association on  
chromosome 27

# Dogs data (*Canis familiaris*)

Binary phenotype: **cleft lip** (presence/absence)



39 chromosomes

Strong signal of association on chromosome 27

We take chromosomes 25, 26, 27, 28 and 29