Introduction to GWAS
Description of Datasets

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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[1]*, Paolo Cozzi[2], Laura Casella[1¤], Paolo Riccardi[1], Alessandra Vattari[1], Gabriele Orasen[3], Rosaria Perrini[3], Gianni Tacconi[4], Alessandro Tondelli[4], Chiara Biselli[3], Luigi Cattivelli[4], Jennifer Spindel[5], Susan McCouch[5], Pamela Abbruscato[1], Giampiero Valé[3,4], Pietro Piffanelli[1], Raffaella Greco[1]

1 PTP Science Park, 26900 Lodi, Italy, 2 IBBA-CNR, Lodi, Italy, 3 CREA-Council for Agricultural Research and Economics, Rice Research Unit, 13100 Vercelli, Italy, 4 CREA-Council for Agricultural Research and Economics, Genomics Research Centre, 29017 Fiorenzuola d'Arda (Piacenza), Italy, 5 Department of Plant Breeding & Genetics, Cornell University, Ithaca, NY, United States of America
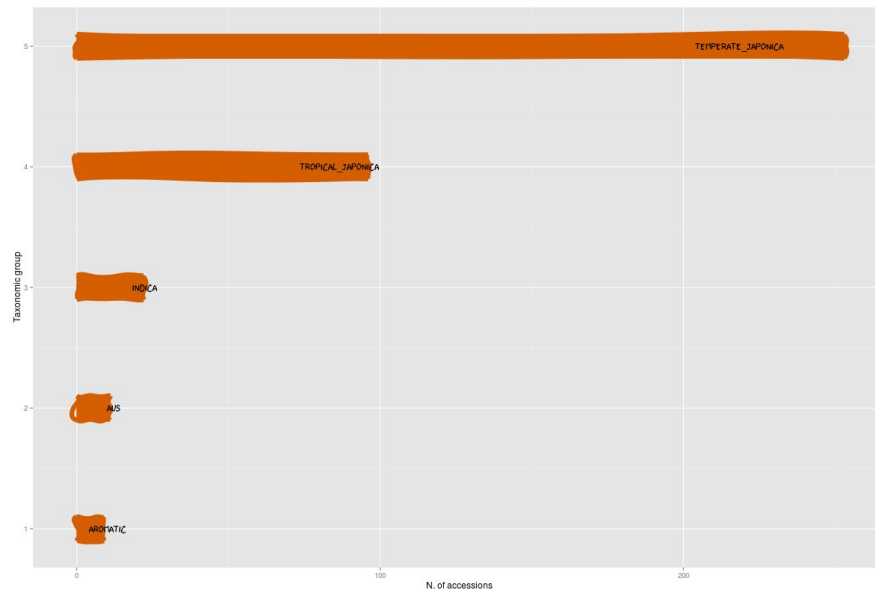
Rice data (*Oryza sativa*)

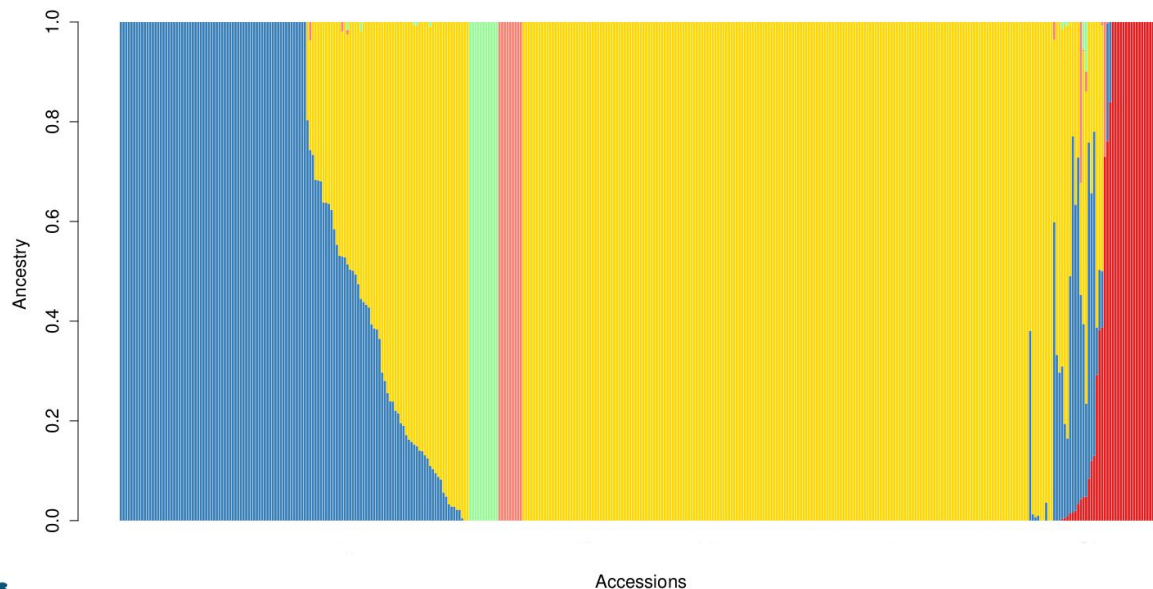Continuous phenotypes: **plant height** (PH)

- genotype data from **GBS**
- ~ 400 rice accessions from
  5 sub-populations:
  - *temperate japonica*
  - *tropical japonica*
  - *indica*
  - *aus*
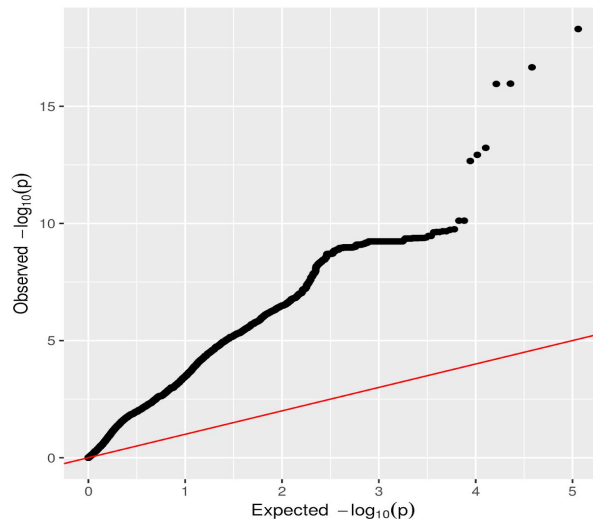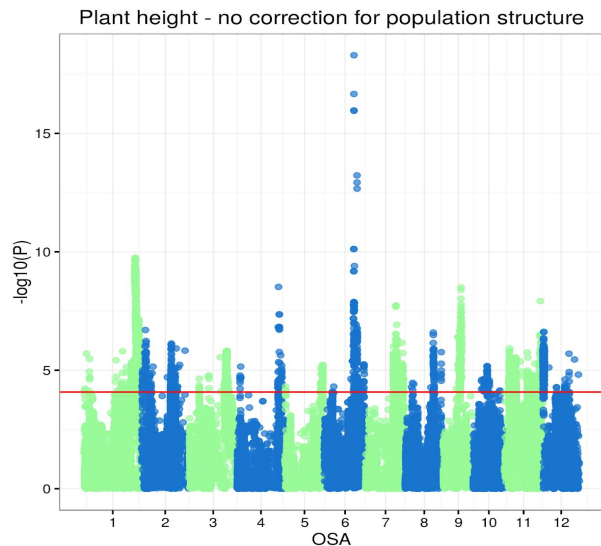 - *aromatica*

Rice data (*Oryza sativa*)

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

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

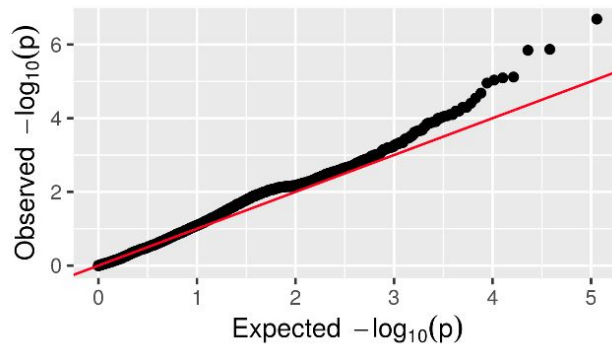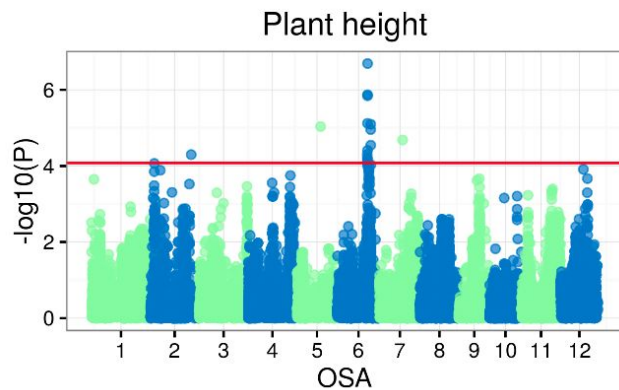# Rice data (*Oryza sativa*)

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

Plant height - no correction for population structure

12 chromosomes

No correction for population structure

Rice data (*Oryza sativa*)

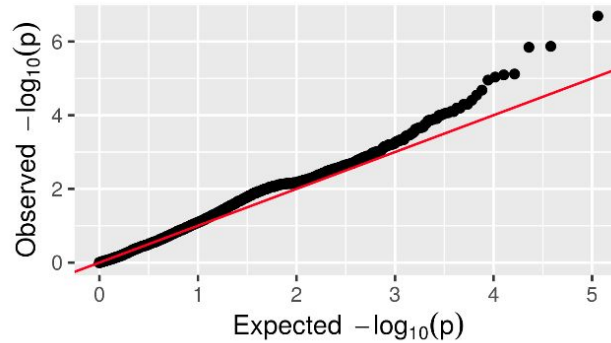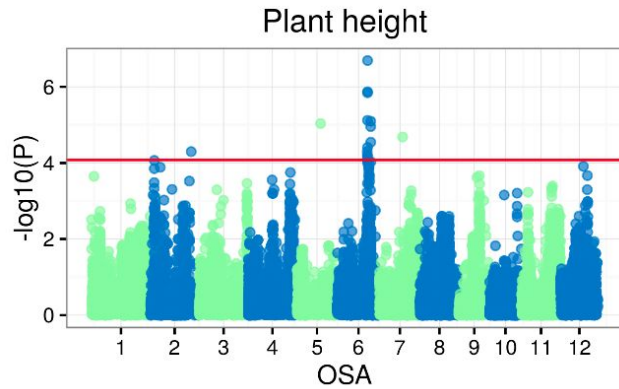Continuous phenotypes: **plant height** (PH)

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)

PLOS | GENETICS

RESEARCH ARTICLE

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

Zena T. Wolf[1], Harrison A. Brand[2,3¤a], John R. Shaffer[3], Elizabeth J. Leslie[2], Boaz Arzi[4], Cali E. Willet[5], Timothy C. Cox[6,7,8], Toby McHenry[2], Nicole Narayan[9], Eleanor Feingold[3], Xioajing Wang[2¤b], Saundra Sliskovic[1], Nili Karmi[1], Noa Safra[1], Carla Sanchez[2], Frederic W. B. Deleyiannis[10], Jeffrey C. Murray[11], Claire M. Wade[5], Mary L. Marazita[2,12‡*], Danika L. Bannasch[1‡*]

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*)
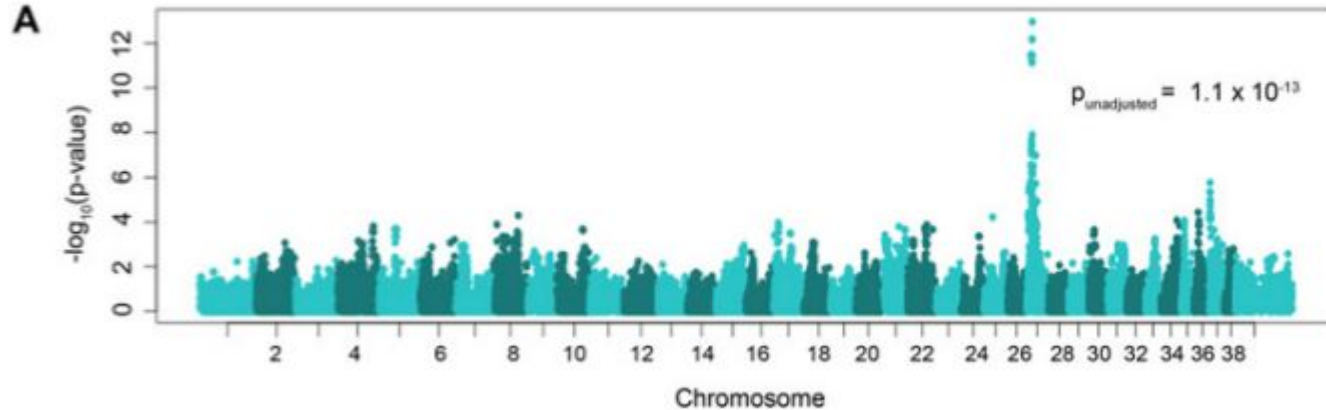
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Dogs data (*Canis familiaris*)

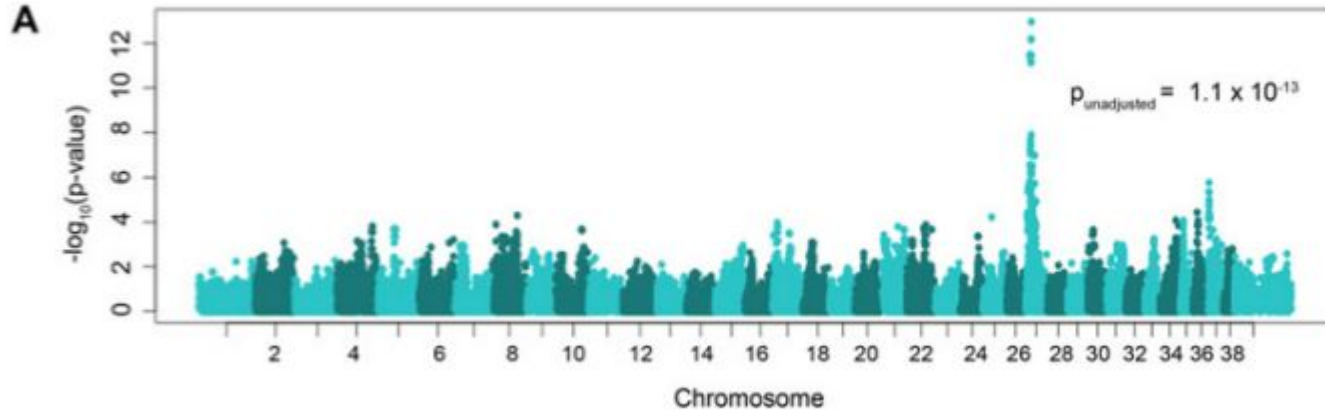Binary phenotype: **cleft lip** (presence/absence)

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