

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^{1*}, Paolo Cozzi², Laura Casella^{1a}, Paolo Riccardi¹, Alessandra Vattari¹, Gabriele Orasen³, Rosaria Perrini³, Gianni Tacconi⁴, Alessandro Tondelli⁴, Chiara Biselli³, Luigi Cattivelli⁴, Jennifer Spindel⁵, Susan McCouch⁵, Pamela Abbruscato¹, Giampiero Valé^{3,4}, Pietro Piffanelli¹, Raffaella Greco¹

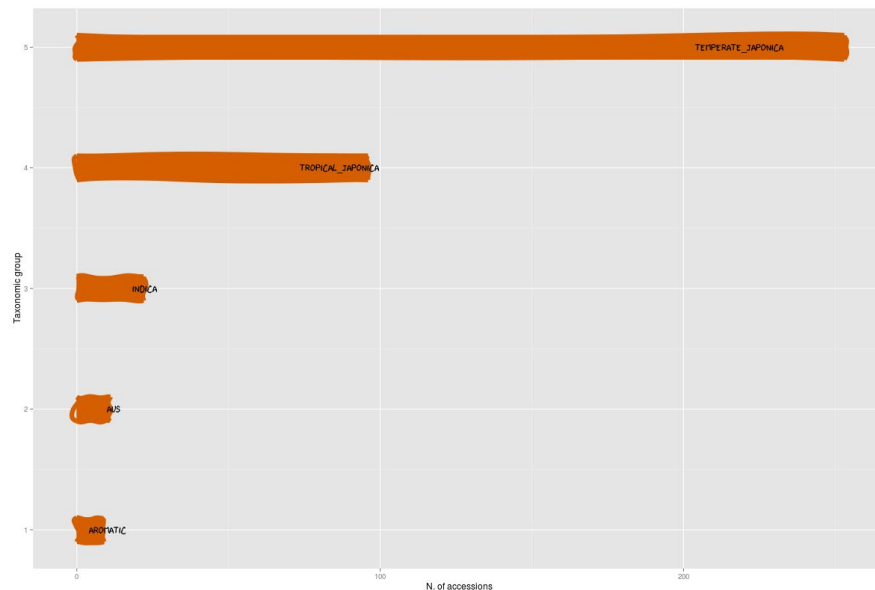
¹ PTP Science Park, 26900 Lodi, Italy, ² IBBA-CNR, Lodi, Italy, ³ CREA-Council for Agricultural Research and Economics, Rice Research Unit, 13100 Vercelli, Italy, ⁴ CREA-Council for Agricultural Research and Economics, Genomics Research Centre, 29017 Fiorenzuola d'Arda (Piacenza), Italy, ⁵ Department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



Rice data (*Oryza sativa*)

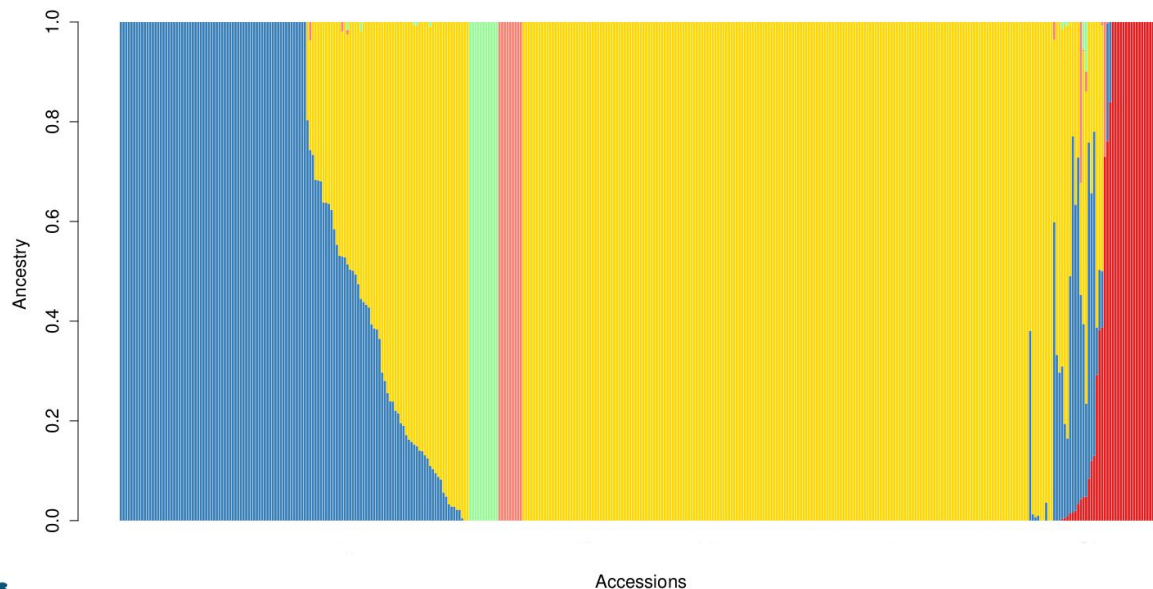
Continuous phenotypes: **plant height (PH)**

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



Rice data (*Oryza sativa*)

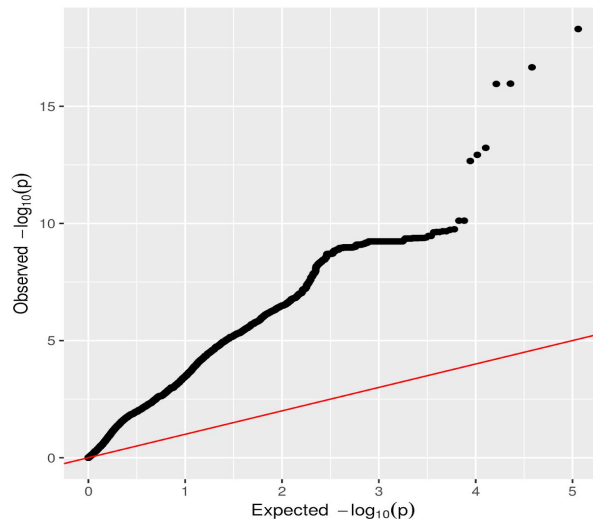
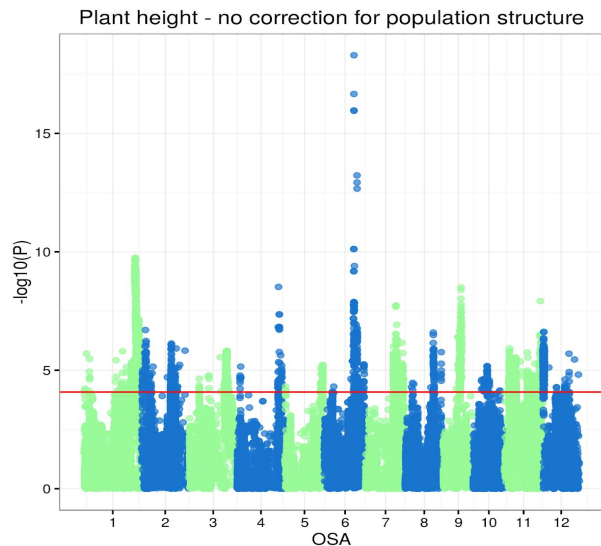
Continuous phenotypes: **plant height (PH)**



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

Rice data (*Oryza sativa*)

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

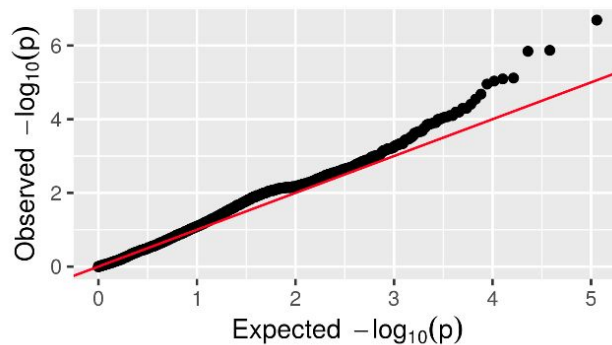
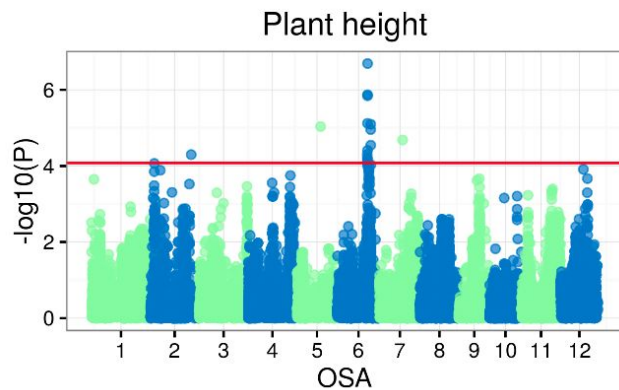


12 chromosomes

No correction for
population structure

Rice data (*Oryza sativa*)

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

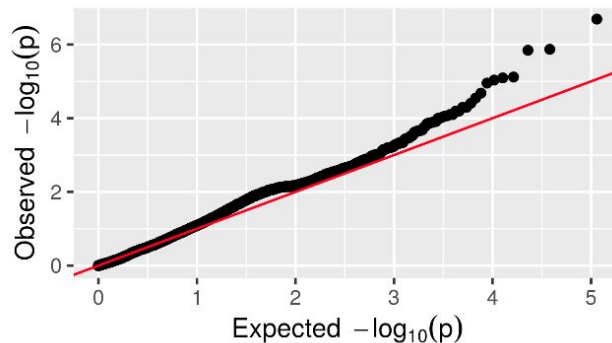
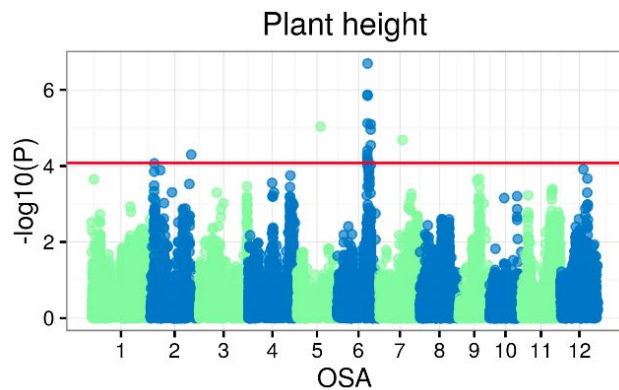


12 chromosomes

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)



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. Leslie², Boaz Arzi⁴, Cali E. Willet⁵, Timothy C. Cox^{6,7,8}, Toby McHenry², Nicole Narayan⁹, Eleanor Feingold³, Xiaojing Wang^{2ab}, Sandra Sliskovic¹, Nili Karmi¹, Noa Safra¹, Carla Sanchez², Frederic W. B. Deleyiannis¹⁰, Jeffrey C. Murray¹¹, Claire M. Wade⁵, 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*)

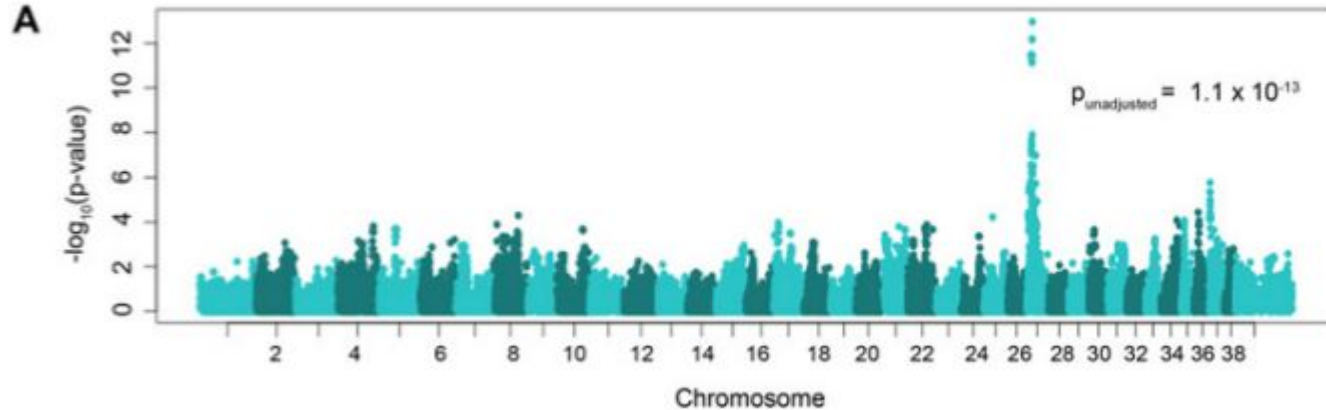
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Dogs data (*Canis familiaris*)

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

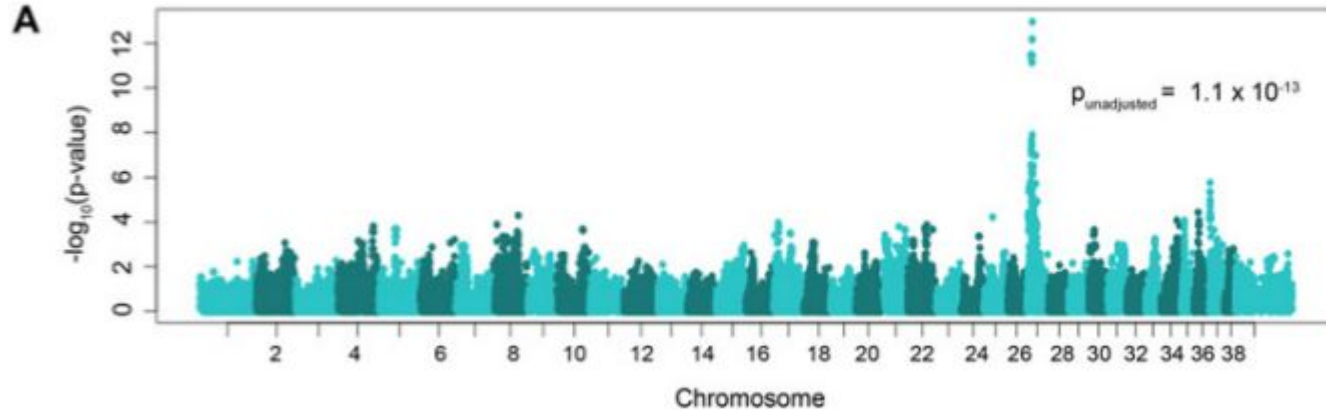


39 chromosomes

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