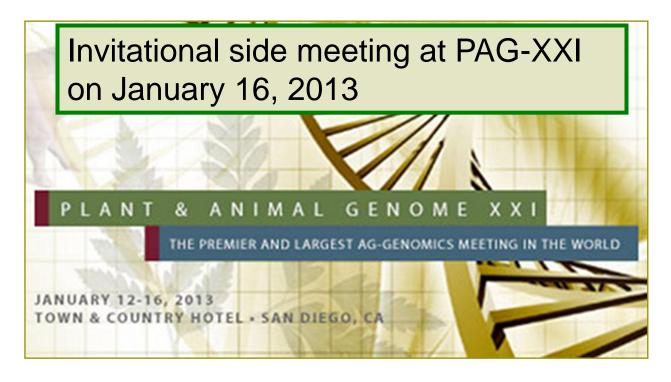


# 3K genome and International Rice Informatics Consortium, Concept and Operation

Nickolai Alexandrov

T.T.Chang Genetic Resources Center

# **IRIC Kick-off Meeting**

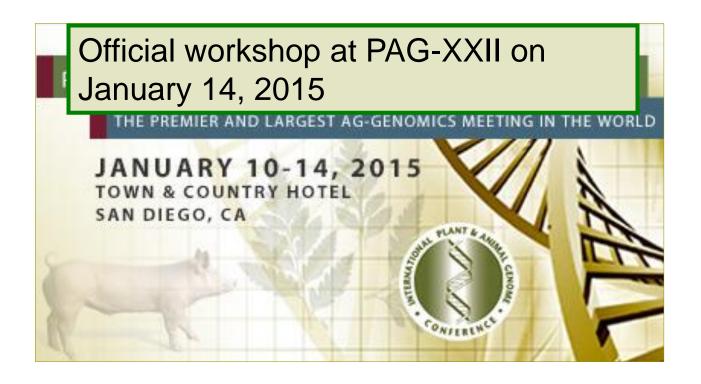


56 people from 38 organizations

Provide access to well organized information about rice, facilitate communication and collaboration for rice community having germplasm diversity as a focal entry point



# **IRIC PAG Workshop**



#### **IRIC** progress

Pierre Larmande, IRD, UMR DIADE, Institut de Biologie Rod A. Wing, University of Arizona, AGI Ruaraidh Hamilton, International Rice Research Institute



# **IRIC** objectives

- 1. Organize available genotyping, phenotyping, expression and other information about rice into a linked, consistent and reliable source of knowledge
- 2. Provide user-friendly portal to browse, search and analyze the data
- 3. Support information sharing, public awareness and capacity building



# **The 3000 Rice Genome Project**









#### **Our Project Team & Donors**

**BGI IRRI** CAAS Shuai-Shuai Tai Ma. Elizabeth B. Naredo Wen-Sheng Wang Xin Liu Sheila M. Q. Mercado Yong-Ming Gao Jun Li Myla C. Rellosa Xiu-Qing Zhao Renato A. Reaño **Guo-Jie Zhang** Jian-Long Xu **Bo Wang** Grace Lee S. Capilit Fan Zhang Xun Xu Flora C. de Guzman Yong-Li Zhou **Gengyun Zhang** N. R. Sackville Hamilton Bin-Ying Fu Jauhar Ali **Zhikang Li** Ramil P. Mauleon Nickolai N. Alexandrov Hei Leung Kenneth L. McNally



**GRISP & CAAS** 





MOST: 科学技术部



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Abbreviations

Competing

Authors' contributions

Acknowledgements

References

1,000,000 antibodies ELISA kits proteins Data Note

Highly accessed

Open Access

Author Affiliations

#### The 3,000 rice genomes project

The 3,000 rice genomes project

Correspondence: The 3,000 rice genomes project

† Equal contributors

Institute of Crop Sciences/National Key Facilities for Crop Gene Resources and Genetic Improvement, Chinese Academy of Agricultural Sciences, 12 S. Zhong-Guan-Cun St, Beijing 100081, China

BGI, Bei Shan Industrial Zone, Yantian District, Shenzhen 518083, China

International Rice Research Institute, DAPO 7777, Metro Manila 1301, Philippines

GigaScience 2014, 3:7 doi:10.1186/2047-217X-3-7

Correspondence: lizhikang@caas.cn; zhanggengyun@genomics.cn; k.mcnally@irri.org; † The list of project participants and their affiliations is given at the end of this paper.

The electronic version of this article is the complete one and can be found online at: http://www.gigasciencejournal.com/content/3/1/7

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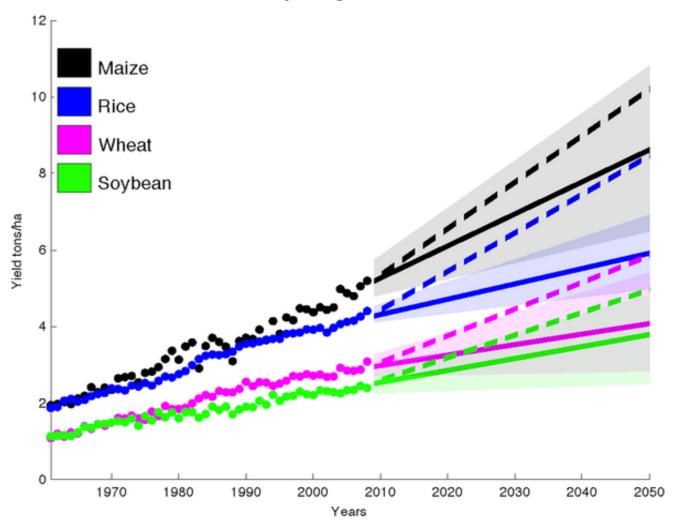
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#### "Terrifying Chart"



Ray DK, Mueller ND, West PC, Foley JA (2013) Yield Trends Are Insufficient to Double Global Crop Production by 2050. PLoS ONE 8(6): e66428. doi:10.1371/journal.pone.0066428

http://www.plosone.org/article/info:doi/10.1371/journal.pone.0066428





#### **Rice and Health**

India, with 41 million obese people, ranks third after the US and China in having the highest number of overweight people in the world. Together, India and China represent 15% of the world's obese population.

The Lancet May 29, 2014

Replace white rice with brown to cut obesity and diabetes

Dr. V. Mohan World Diabetes Conference 2013

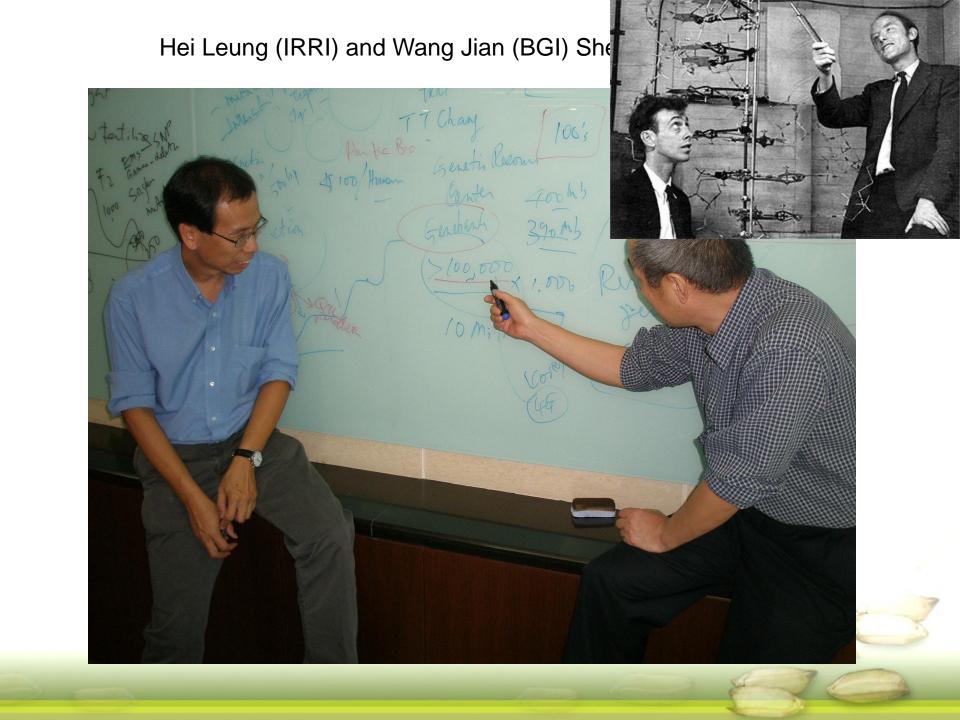
IRRI's mission: make rice healthier



# How can knowledge of 3,000 rice genomes help?

- 1. By discovering new locus-trait associations
- 2. By finding causative genome variations
- 3. By introducing new varieties to breeding programs





#### IRIC deliverables for 2014

- ✓ Initial version of IRIC portal on-line
- ✓ Advisory Board is formed
- ✓ IRIC guidelines documents are finalized
- ✓ Established communication with IRIC members and supporters



# **IRIC Progress for 2014**

- IRRI hired an AXA Chair on Genome Biology and Evolutionary Genomics, two postdoctoral fellows and two senior software developers
- IRIC Guidelines for Partnership has been finalized. Information page is now available at <a href="http://iric.irri.org/">http://iric.irri.org/</a>
- IRRI purchased hardware, licensed Oracle database, has been granted access to iPlant computational infrastructures
- Data from the 3,000 rice genome project (CAAS, BGI, IRRI): QCed, aligned with Nipponbare 1.0 assembly, SNPs called, admixture analysis done, phylogenetic tree constructed. The data has been published at GigaScience on May 28, 2014
- Database schema has been populated with SNPs from the 3,000 project data, 700K chip data, other data
- The first version of IRIC portal (SNP-Seek database) is online <a href="http://oryzasnp.org/iric-portal/">http://oryzasnp.org/iric-portal/</a> and is published in 2015 NAR database issue
- Collaborations are being established



# **IRIC Advisory Committee**

Mario Caccamo, Director, The Genome Analysis Center
Dave Edwards, Professor, The University of Western Australia
Harish Gandhi, Head – Rice Genetics and Traits Project, Syngenta
Hei Leung, Principal Scientist – Genetics, Program Leader – Genetic Diversity, IRRI
Takashi Matsumoto, Director – Agronomics Research Center, NIAS
Susan McCouch, Professor, Cornell University
Debashis Rana, Team Leader – Rice Genetics, Bayer
Rod Wing, Arizona Genomics Institute, University of Arizona
Gengyun Zhang, Vice President, Beijing Genomics Institute



# **IRIC** Information page

http://iric.irri.org/



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#### Navigation

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#### **Important dates**

**14 January 2015** <u>IRIC Workshop</u> at **PAG XXIII** (Pacific Salon 2)

#### Interact with us









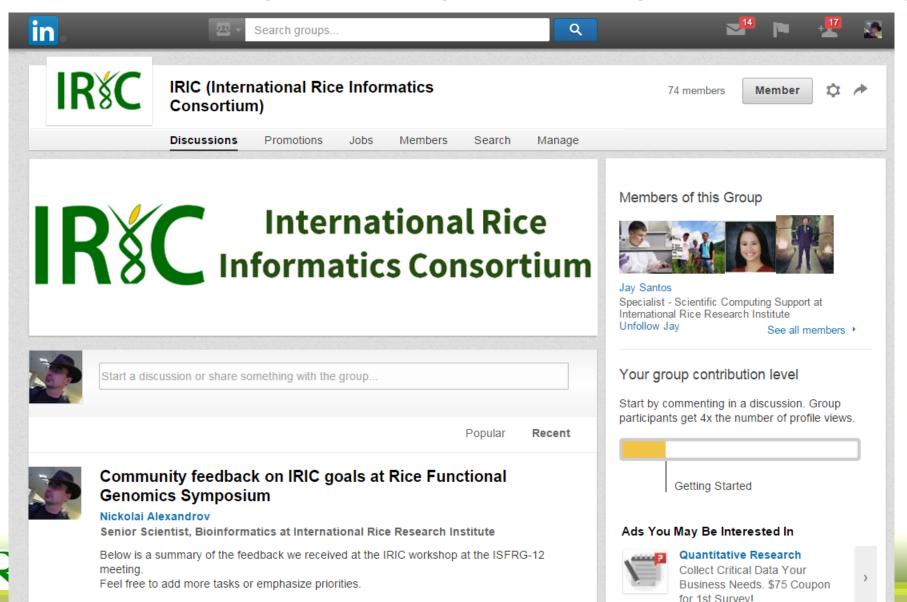






# IRIC LinkedIn group

https://www.linkedin.com/groups?home=&gid=6722493&trk=groups most recent-h-log



# IRIC portal: target users

#### **Breeders**

find germplasm with blast resistance find germplasm with good Pi5 allele

#### Molecular biologists

show genes associated with grain width show all SNPs in Hd1 gene in given lines

#### **Bioinformatics scientists**

give me all SNP calls in tropical japonica API



#### Initial sets of the IRIC data

#### IRRI Genebank genetic stocks for trait discovery research

>25,000 purified genetic stocks, RILs, CSSLs, MAGIC lines Annotated with standard unique names, origins, and identifiers Purified seed of all lines can be requested via IRRI seed request forms

#### Genotypic data

>10M SNPs from 3000 rice genomes (BGI/CAAS/IRRI)

700K SNP chip for ~1800 lines (Cornell/IRRI)

44K SNP chip for ~400 lines (Cornell/IRRI)

All SNPs will be mapped into annotated genome assemblies

#### Phenotypic data

- ~60 morphological traits observed for genetic stock lines from the IRRI Genebank
- ~30 traits for ~400 lines genotyped with 44K SNPs
- ~60 traits for ~350 indica lines genotyped with 700K chip
- ~25 traits for ~300 tropical japonica and ~250 aus lines genotyped with 700K chip grain quality traits for ~1600 lines genotyped with 700K chip

#### Gene expression data

Basic developmental and selected stress experiments from NIAS, Japan RNA-Seq data from C4 consortium



#### IRIC portal: architecture

**JBrowse** HTML5 CSS3 BitBucket JavaScript ZK JIRA **JSON** Apache Tomcat7 Java **JDBC** Oracle11g Chado HDF5



#### **Developers framework**

Oracle: capable to store and quickly query 60B SNPs JBrowse: interactive, easily customizable cool browser

ZK: enables smooth navigation in 1Mx1M table

HDF5: amazingly quick data retrieval

https://bitbucket.org/irridev/iric\_portal

https://riceinfo.atlassian.net/browse/IRIC-220?filter=-4&jql=project%20%3D%20IRIC%

Providing RESTful web APIs and web services for communications with other systems



# **Existing rice portals**

MSU Rice Genome Annotation Project http://rice.plantbiology.msu.edu/



Rice Genome Annotation Project Funded by the NSF



RAP-DB http://rapdb.dna.affrc.go.jp/



**BGI-RIS** Rice Information System http://rice.genomics.org.cn/rice/index2.jsp





**PlantGDB** 





Gramene http://www.gramene.org/





#### Initial version of IRIC Portal



#### Rice SNP-Seek Database





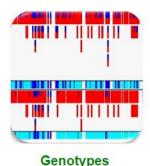








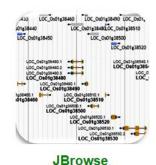
This site provides Genotype, Phenotype, and Variety Information for rice (Oryza sativa L.). SNP genotyping data (called against Nipponbare reference Os-Nipponbare-Reference-IRGSP-1.0) came from 3,000 Rice Genomes Project . Phenotype and passport data for the 3,000 rice varieties came from the International Rice Information System (IRIS). We are a part of an ongoing effort by the International Rice Informatics Consortium (IRIC) to centralize information access to rice research data and provide computational tools to facilitate rice improvement via discovery of new gene-trait associations and accelerated breeding.



Query for SNPs from the 3000 genome project



**Varieties** Query for Variety passport and phenotypes



Rice Genome Browser



Help Help and documentation



Nucleic Acids Research, 2014 1 doi: 10.1093/nar/gku1039

# SNP-Seek database of SNPs derived from 3000 rice genomes

Nickolai Alexandrov<sup>1,\*,†</sup>, Shuaishuai Tai<sup>2,†</sup>, Wensheng Wang<sup>3,†</sup>, Locedie Mansueto<sup>1</sup>, Kevin Palis<sup>1</sup>, Roven Rommel Fuentes<sup>1</sup>, Victor Jun Ulat<sup>1</sup>, Dmytro Chebotarov<sup>1</sup>, Gengyun Zhang<sup>2,\*</sup>, Zhikang Li<sup>3,\*</sup>, Ramil Mauleon<sup>1</sup>, Ruaraidh Sackville Hamilton<sup>1</sup> and Kenneth L. McNally<sup>1</sup>

<sup>1</sup>T.T.Chang Genetic Resources Center, IRRI, Los Baños, Laguna 4031, Philippines, <sup>2</sup>BGI, Shenzhen 518083, China and <sup>3</sup>CAAS, Beijing 100081, China

Received September 08, 2014; Revised October 10, 2014; Accepted October 10, 2014

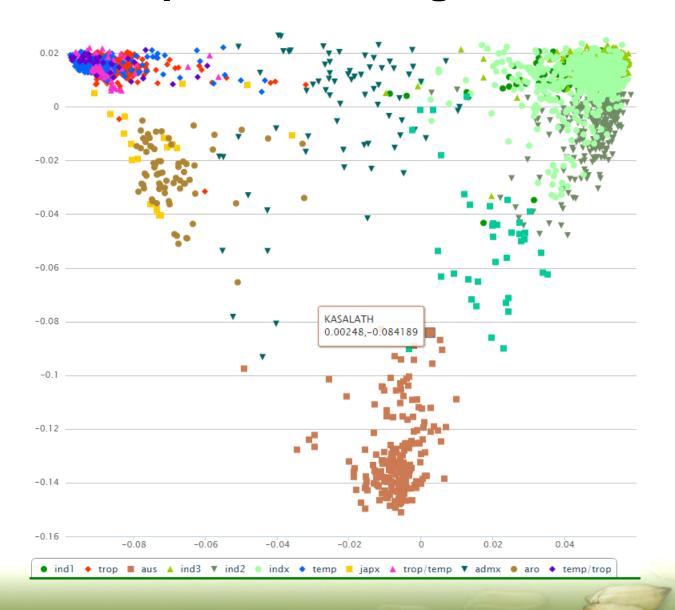
#### **ABSTRACT**

We have identified about 20 million rice SNPs by aligning reads from the 3000 rice genomes project with the Nipponbare genome. The SNPs and allele information are organized into a SNP-Seek system (http://www.oryzasnp.org/iric-portal/), which consists of Oracle database having a total number of rows with SNP genotypes close to 60 billion (20 M SNPs  $\times$  3 K rice lines) and web interface for convenient querying. The database allows quick retrieving of SNP alleles for all varieties in a given genome

information, is an important step toward establishing genetrait associations, building predictive models and applying these models in the breeding process. The 3000 rice genome project (6) produced millions of genomic reads for a diverse set of rice varieties. SNP-Seek database is designed to provide a user-friendly access to the single nucleotide polymorphisms, or SNPs, identified from this data. Short, 83 bp pair-ended Illumina reads were aligned using the BWA program (7) to the Nipponbare temperate japonica genome assembly (8), resulting in average of 14× coverage of rice genome among all the varieties. SNP calls were made using GATK pipeline (9) as described in (6).



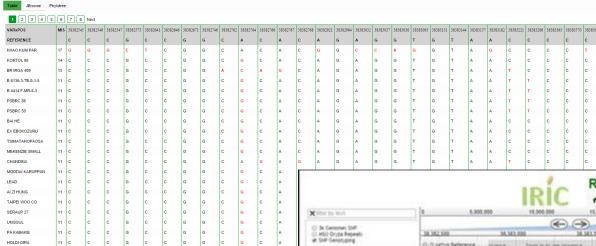
# MDS plot for 3000 genomes







· Reference organice W. Octativa Reference



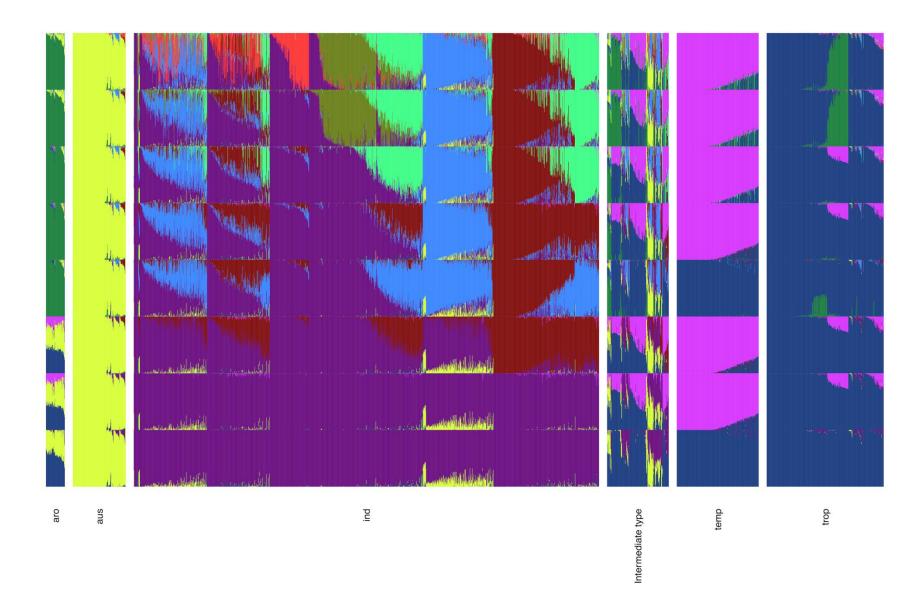
PA KABAKIE HOLDI GIRA

**SNP-Seek online** 





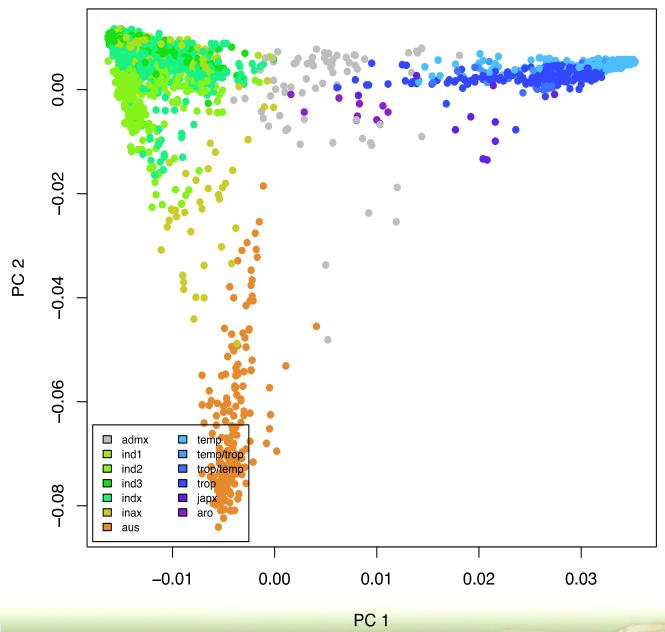
# Admixture analysis of the 3K genomes



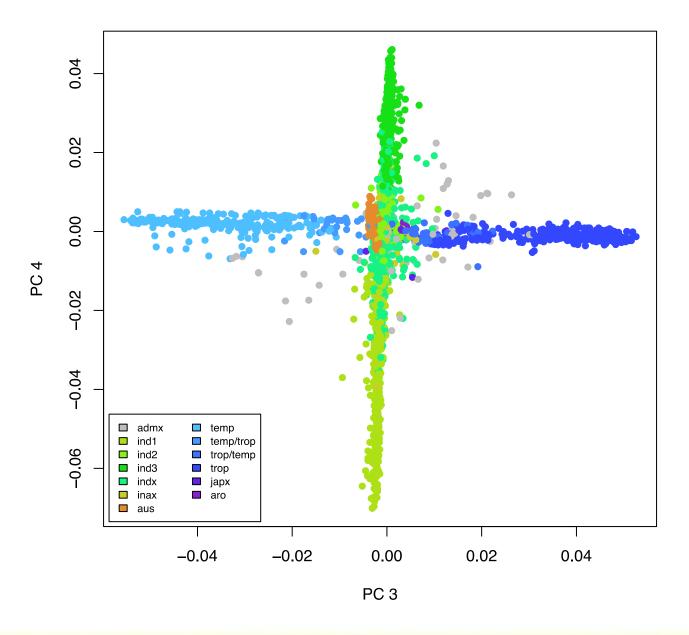
# **Principal Component Analysis**

- PCA was run on 3k core SNP dataset using software EIGENSOFT
- 10 PCs were extracted
- The PCs suggest finer population structure
   ( both within indica and japonica)

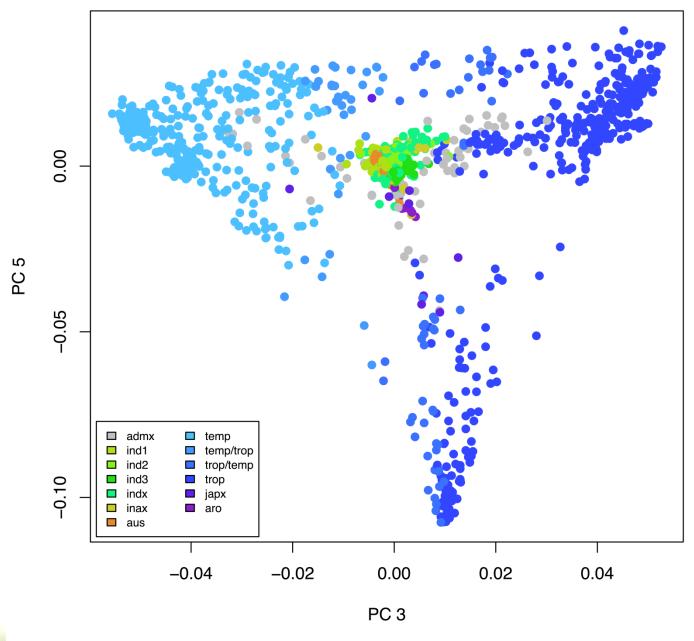




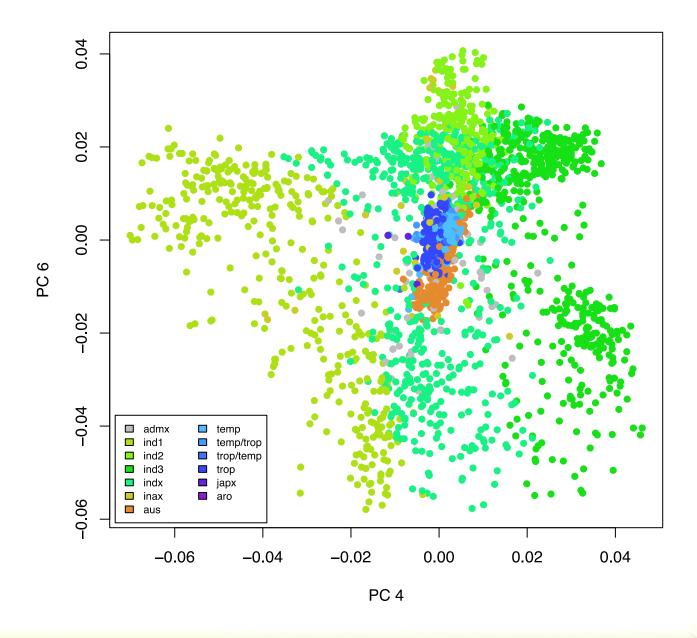




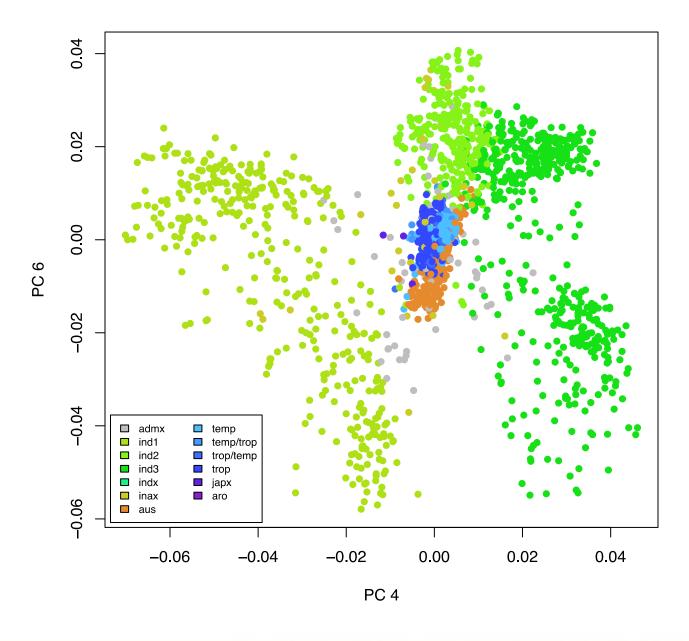




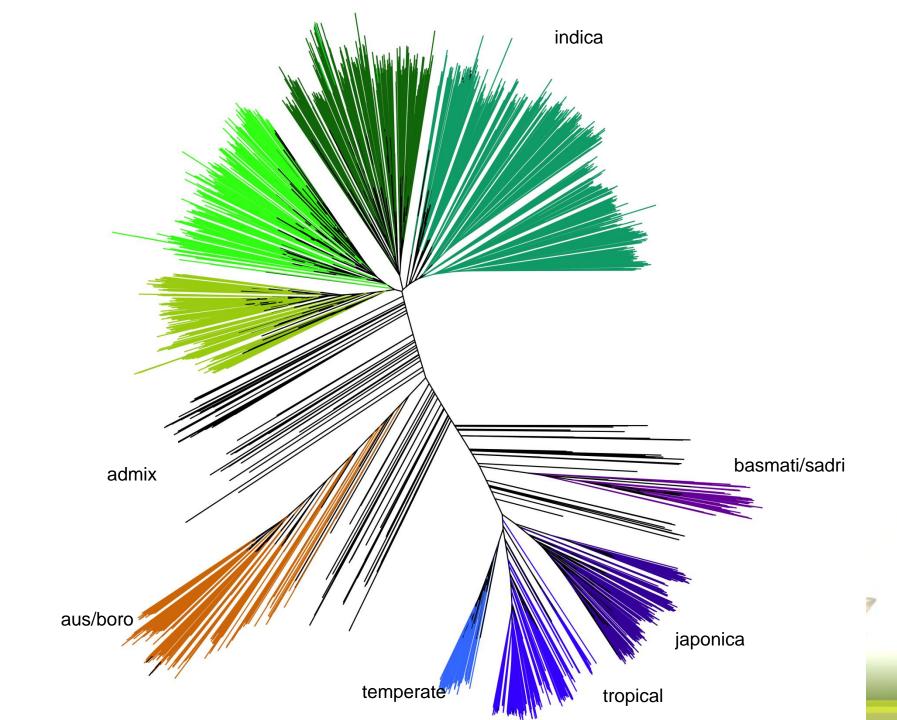






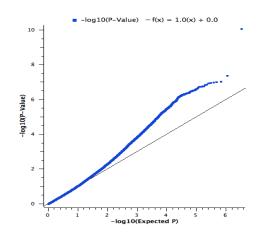


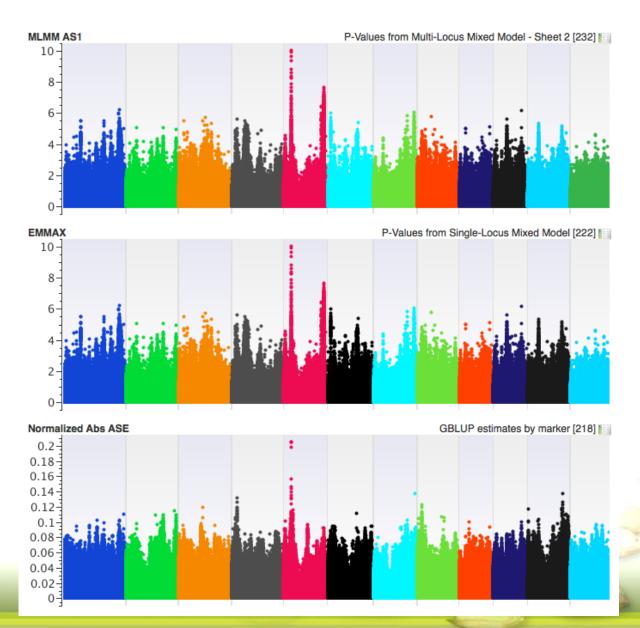




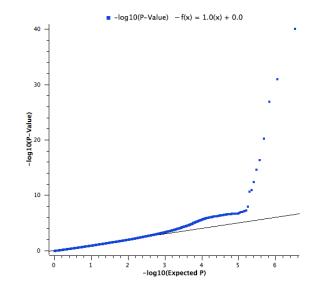


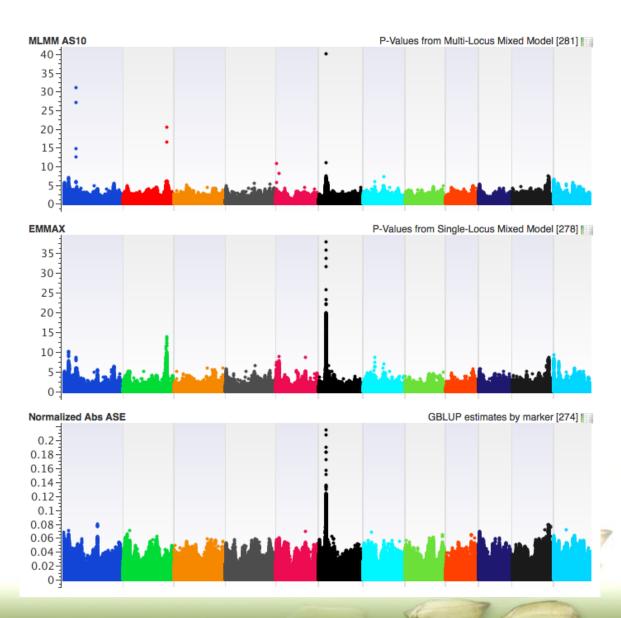
# Historical data for grain weight of 100 seeds from 1621 lines of 3K RGP (1.65M SNPS, CR>0.8, MAF<0.05)



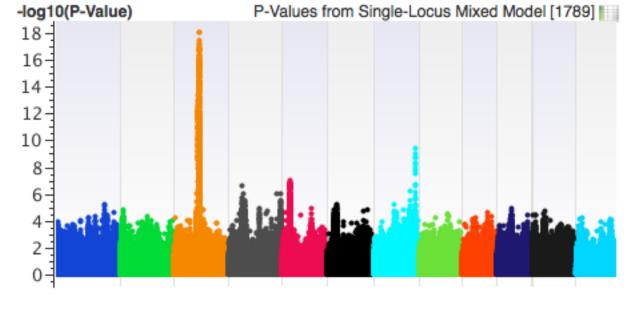


# Historical data for apiculus color from 1621 lines of 3K RGP (1.65M SNPS, CR>0.8, MAF<0.05)

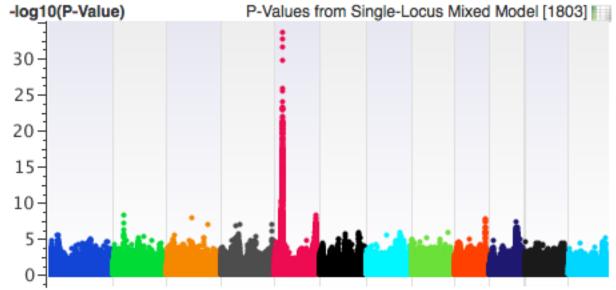




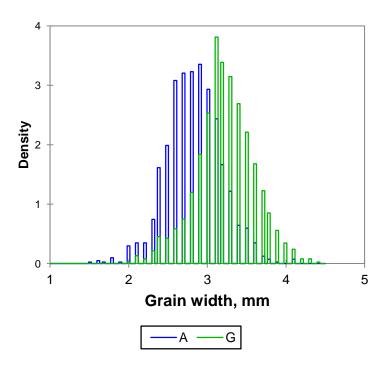
#### Grain length



#### Grain width



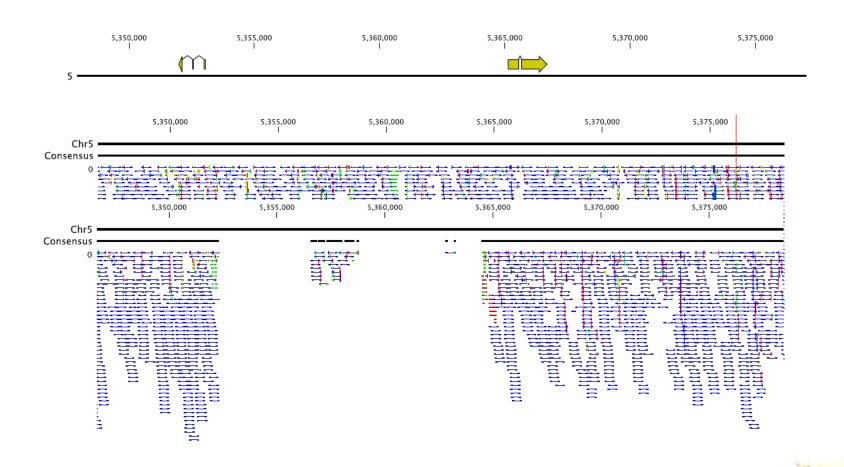
## Grain width distribution



IRIS 313-8431 : G : 4.3 mm : trop : BOCAO

IRIS 313-12151 : A : 1.6 mm : ind3 : DAMNOEUB KRACHAK SESS

# Genome region at the GWAS peak



#### **GW5** rediscovered

Cell Research (2008) 18:1199–1209. doi: 10.1038/cr.2008.307; published online 18 November 2008

# Isolation and initial characterization of *GW5*, a major QTL associated with rice grain width and weight

Jianfeng Weng<sup>1</sup>, Suhai Gu<sup>2</sup>, Xiangyuan Wan<sup>1</sup>, He Gao<sup>1</sup>, Tao Guo<sup>1</sup>, Ning Su<sup>2</sup>, Cailin Lei<sup>2</sup>, Xin Zhang<sup>2</sup>, Zhijun Cheng<sup>2</sup>, Xiuping Guo<sup>2</sup>, Jiulin Wang<sup>2</sup>, Ling Jiang<sup>1</sup>, Huqu Zhai<sup>2</sup> and Jianmin Wan<sup>2</sup>

<sup>1</sup>National Key Laboratory for Crop Genetics and Germplasm Enhancement, Nanjing Agricultural University, Nanjing 210095, China

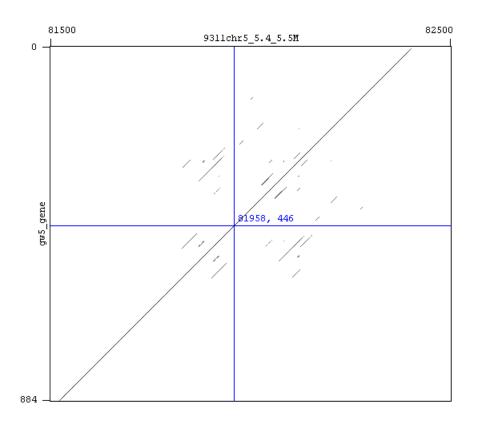
<sup>2</sup>Institute of Crop Science and the National Key Facility for Crop Gene Resources and Genetic Improvement, Chinese Academy of Agricultural Sciences, Beijing 100081, China

Correspondence: Jianmin Wan, Tel/Fax: +86-10-62186628 E-mail: wanjm@caas.net.cn

Received 3 September 2008; Revised 6 October 2008; Accepted 6 October 2008; Published online 18 November 2008.



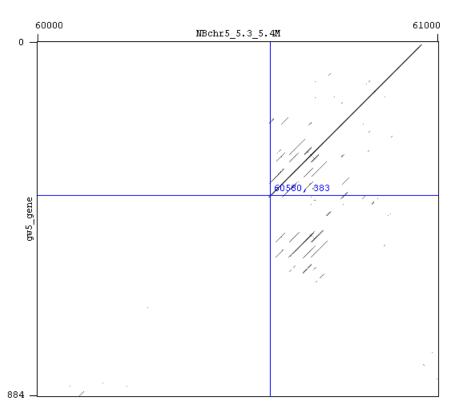
#### Gw5 vs. 9311







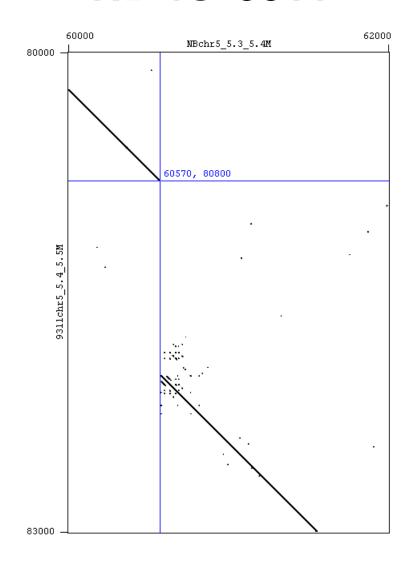
## Gw5 vs. NB



60486	60580 60673
I .	1
${\tt TCCATCTATCCGTCCACACCACGCTGCAAGTTTAAATTCCCATGCAGTTACATGAACAGTACTGCTGTCAAGTTTAATTTAATTTCGCGCTAGTCAACAGTACTGCACACACA$	CGCTCCTCCTTCTGCCTACTCCCTCCCGGCACCGCTCCTCCTTCTGCCTACTCCCCCCGGCACCACTCCGTCCTCCTCCGCTTGCTCCCT
GTCCTCCTCTCTCTCGCCGCCGCCAAGTTGCCGGCTGACCGCCCGC	CGCTCCTCCTTCTGCCTACTCCCTCCCCGGCACCGCTCCTCCTTCTGCCTACTCCCTCC
1	1
477	383 290



## NB vs. 9311





# Feedback from Rice Functional Genomics, Tucson AZ

Have a central

Site for listing major

genes/QTLs of interest

to the commenty and

adding information

"Rice gene cleaning house"

for community annotation

and prioritization for breeders

mike Thomson

Set up Working

Group for "Plant Breeding

applications" to gather

input from breeder on how

to apply dota as breeding

(including limking with IBP

and Breeding 4 rice databases

and tools)

mire Thanson

Sorge Duitama (2)

I think that information
of itructural variation
our sample is very imour tant to design good
narkers. This information
hould be discovered and
made available in the
serie dat abase

D Get broodens involved

(2) Make data sacily and and understandible by and for broaders!

3) Integrate other sequence

Tracking of germplasm beyond inst "accession" seed source and generation are important when distributing seeds for phenotyping.

International support

International support

John Gorts of Develope

Coventrie to support

Genome Sequencing

Projects

(5)

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### Feedback from Rice Functional Genomics, Tucson AZ

- Germplasm, mutants
  - Tracking
  - Access to seeds
  - Metadata (location, breeding history,
  - Mutants, knockouts and overexpression lines
  - Define subset(s) for phenotyping
- Phenotyping
  - Controlled vocabulary
  - Trait ontology, Crop ontology, Environment ontology, Method ontology
  - Global phenotyping



### Feedback from Rice Functional Genomics, Tucson AZ

#### Sequencing

- Information on the planned/ongoing projects
- Version control
- Correspondence between different assemblies (unified coordinates)

#### Genes and loci

- Versions of genome annotation
- Gene names, Gene ontology
- SNP
- Location in different assemblies
- Trait-loci associations
- Gene expression
- Gene networks



#### Feedback from Rice Functional Genomics, Tucson AZ

#### Tools

- Web portal with restful queries
- Mobile application
- GWAS: population structure
- Causative mutations, assay design
- Predictive models, validation of trait-loci associations
- Comparative genomics (e.g. for C4/C3 analysis)
- Structural variant detection
- Promoter analysis
- Breeder assistance on selection of new lines, crosses

#### Organizational

- Monthly letters
- Interest groups
- IP management
- Coordinating with other projects



#### **Priorities**

- ✓ Use other reference genome sequences with good annotation.
- ✓ Find important SNPs (merging with current GWAS/QTL results)
  - in CDSs
  - in promoters and other regulatory motifs
- ✓ Reconstruct large deletions/insertions/inversions in genome
- ✓ Find correlated SNPs
- ✓ Organize available GWAS/QTL data
- ✓ Create new genome assemblies
- ✓ Rice metagenome analysis
- ✓ Focus on known genes associated with traits
- ✓ Find conserved genome regions selected by breeders
- ✓ Population genetics analysis and predictions



#### **IRIC Goals for 2015**

- 1. Develop a mechanism for unified germplasm tracking
- 2. Select a subset(s) of germplasm for detailed studies and organize corresponding metadata using existing ontologies
- 3. Create a curated database of functional loci annotation
- 4. Establish communication with similar plant consortia (Arabidopsis, wheat, soy, etc)
- 5. Approach funding agencies



## IRIC portal development team

Rolando Jay Santos
Victor Jun Ulat
Frances Nikki Borja
Venice Margarette Juanillas
Jeffrey Detras
Roven Rommel Fuentes
Dmytro Chebatorov
Millicent Sanciangco
Kevin Palis
Locedie Mansueto

**Technological advice**Marco van den Berg

Visionary input
Achim Dobermann
Matthew Morell

**Administrative support** Vina Alvarez

Management team
Hei Leung
Ruaraidh Sackville Hamilton
Kenneth McNally
Ramil Mauleon
Nickolai Alexandrov
Rod Wing



