## R in Systems Biology and Translational Medicine

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## Outline

- BIG and Complex Biological Data
- 2 How do we study diseases?
- 3 How do we use R to study diseases?
- 4 Information



#### Next

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## What is Systems Biology and Translational Medicine?

#### Systems Biology

The study of the interactions between the components of biological systems, and how these interactions give rise to the function and behavior of that system.

-Wikipedia

#### Translational Medicine

- "...translate the remarkable scientific innovations we are witnessing into health gains  $\dots$ "
- Elias A. Zerhouni, NEJM 2005



## Challenges

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- High-quality and large-scale sample collection
- Money
- Big and complex data analysis



## **BIG Biological Data**

- One human genome, 3 GB
- ullet 60X depth sequencing of human genome, 3 imes 60 = 180GB
- A typical study of complex disease need 2K samples in the 1st stage and more than 10K samples for validation.
- $1K \times 60 \times 3GB = 180TB$



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## Complex Biological Data

#### Why?

- Biological system is very complicated
- Biological data is very complicated
- DNA
- RNA
- Protein
- Metabolite
- Microbe
- Virus



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## Diseas<u>es</u>

#### Disease

- Genetic factor
- Environmental factor

#### Examples

- Traffic accident
- Psoriasis
- Tumor
- ......



## Complex Diseases

## Complex Disease

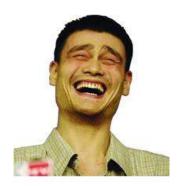
- Genes
- Gene Interactions
- Subtypes
- Environment
- Nation
- .....



## Complex Diseases

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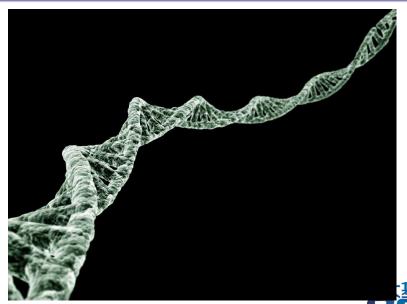




R in Systems Biology and Translational Medicine How do we study diseases?

Could you please help me to find the key I lost in the mountain?



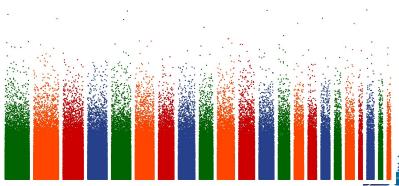


Where is the disease related mutation?

## Genome-Wide Association Study

#### **GWAS**

Check the differences of genotype of each loci in genome between patient and healthy people.

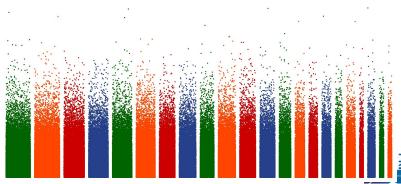




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## Why do we want to use R?

- Rapid implementation
- Active community, COS and CRAN
- Statistics is the fundamental of big data analysis
- Data visualization, ggplot2!
- Integration with other programming language, Rcpp
- Widely supported by commercial products





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- We love R!





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- Bioconductor
- Free and Free
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## Why do we have to use R?

- Most new methods are implemented as R packages
- Bioconductor
- Free and Free
- Training and learning material
- We cannot live without R!





## How do we use R?

#### R in disease study

- Statistical Analysis
- Classification and regression for disease diagnosing and prediction
- Feature selection for disease gene identification
- Visualization for publication and customers





## What we have done?

#### R in BGI

- Thousands of researchers are using R
- R is installed on almost every computer and server
- Most plots in our world-class publications are generated by R
- A novel multi-loci GWAS framework for complex diseases
- A framework for various biological data integration
- An auto report-generation system for personal genome sequencing service



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## **Problems**

#### **Problems**

- Where is R expert?
- R is a statistical tool or a programming language for data analysis?
- How to use R to handle TB data? Hadoop?
- Interfaces for biological data: sequences, interaction, model ...
- Algorithms for extreme high dimension data
- ......



## Expectation

#### **Future**

- R for big data
- Packages for new biological problems
- Soft-engineering methodology and tool-chain for R



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#### **Future**

- R for big data
- Packages for new biological problems
- Soft-engineering methodology and tool-chain for R
- R Geek



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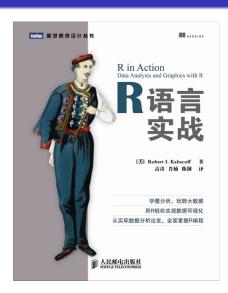
## Conferences

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- ISCB-Asia
   Dec 17 19, Shenzhen
- AYRCOB
   Dec 20 21, Shenzhen
   http://2012.ayrcob.org
- 7th ICG & Bio-IT APAC
   Nov 28 Dec 1, Hong Kong



## Book





## **Thanks**

We are seeking data guru and collaboration! chengangcs@weibo chengang@genomics.cn

http://gossipcoder.com

