## Genomics of Human Diseases

2019 Dragon Star Bioinformatics Course

### Housekeeping issues

- Course date: July 29 August 2, 2019
- Theme: Genomics of Human Diseases
- Morning (9am-12pm with break): lecture session on basic concepts and bioinformatics methods to study the genomics of human diseases
- Afternoon (2pm-4pm): computing exercise to practice software tools discussed in the morning
- Lecture notes and computing exercises in English
- The lecture slides and exercises are shared to students.
  The computing exercise instructions are also available at GitHub: <a href="https://github.com/WGLab/dragonstar2019">https://github.com/WGLab/dragonstar2019</a>

# Housekeeping issues (continued)

- The afternoon computing exercise session will be split into four rooms, with maximum capacity of 110, 110, 55 and 55 students, respectively.
- We will have teaching assistants in each room to address questions and troubleshoot problems.
- Each student will be assigned a separate IP address to connect to a cloud server by SSH.
  - The username and password are biouser and biouser, respectively.
  - You should change your password using passwd command upon logging into the server.
- Each cloud server is only used by one student, and is not shared with the rest of the class.

#### Acknowledgements

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## Outline of the five days

- Day 1: genomic technologies, typical file formats
- Day 2: alignment of sequencing data and genome assembly
- Day 3: detection of structural variants, phenotypedriven variant annotation/interpretation
- Day 4: SNP and sequencing based association studies, rare or de novo variants in human diseases
- Day 5: RNA-Seq and other advanced topics (artificial intelligence, somatic mutation, etc)