

## 课程详述

### COURSE SPECIFICATION

以下课程信息可能根据实际授课需要或在课程检讨之后产生变动。如对课程有任何疑问，请联系授课教师。

The course information as follows may be subject to change, either during the session because of unforeseen circumstances, or following review of the course at the end of the session. Queries about the course should be directed to the course instructor.

|     |  |   |
|-----|--|---|
| 1.  | <b>课程名称 Course Title</b>   | 医学基因组学——从理论到实践<br>Medical Genomics - From Theory to Practice                          |
| 2.  | <b>授课院系<br/>Originating Department</b>   | 医学院 School of Medicine  |
| 3.  | <b>课程编号<br/>Course Code</b>  | MED235  |
| 4.  | <b>课程学分 Credit Value</b>   | 3   |
| 5.  | <b>课程类别<br/>Course Type</b>  | 专业选修课 Major Elective Courses  |
| 6.  | <b>授课学期<br/>Semester</b>   | 春季 Spring   |
| 7.  | <b>授课语言<br/>Teaching Language</b>  | 中英双语 English & Chinese  |
| 8.  | <b>授课教师、所属学系、联系方式<br/>Instructor(s), Affiliation &amp; Contact</b><br>(For team teaching, please list all instructors) | 李职秀, 医学院, lizx3@sustech.edu.cn<br>Zhixiu Li, School of Medicine, lizx3@sustech.edu.cn |
| 9.  | <b>实验员/助教、所属学系、联系方式<br/>Tutor/TA(s), Contact</b>   | 待公布 To be announced   |
| 10. | <b>选课人数限额(可不填)<br/>Maximum Enrolment (Optional)</b>  | 无 None  |

|   |                |                       |                        |                                     |              |
|---|----------------|-----------------------|------------------------|-------------------------------------|--------------|
| 11. 授课方式<br>Delivery Method   | 讲授<br>Lectures | 习题/辅导/讨论<br>Tutorials | 实验/实习<br>Lab/Practical | 其它(请具体注明)<br>Other (Please specify) | 总学时<br>Total |
|   | 48             |                       |                        |                                     | 48           |
| 12. 先修课程、其它学习要求<br>Pre-requisites or Other Academic Requirements    | 无 None         |                       |                        |                                     |              |
| 13. 后续课程、其它学习规划<br>Courses for which this course is a pre-requisite | 无 None         |                       |                        |                                     |              |
| 14. 其它要求修读本课程的学系<br>Cross-listing Dept.                             | 无 None         |                       |                        |                                     |              |

### 教学大纲及教学日历 SYLLABUS

#### 15. 教学目标 Course Objectives

让学生了解与掌握医学基因组学的基本知识，原理和方法，深入了解与熟悉医学基因组学领域的最新进展及其对医学和生命科学各领域的影响，从本质上理解生命的起源，变异与进化，理解基因组学与人类健康的关系，理论联系实际，为学生理解生命遗传物质的奥秘、进入医学科学前沿领域的科研实践打下必要基础。

To guide students understand and master the basic knowledge, principles, and methods of medical genomics, become familiar with the latest developments in the field of genomics and their impact on various areas of medical and life sciences, fundamentally understand the origin, variation and evolution of life, comprehend the relationship between genomics and human health, from the theory to practice, and lay the necessary foundation for students to understand the mysteries of the genetic material of life and engage in cutting-edge scientific research in medical sciences.

#### 16. 预达学习成果 Learning Outcomes

使学生熟悉医学基因组学的基本知识，原理和方法。预期学习本课程之后，学生将掌握当前基因组学的基本知识，了解相关的方法学、数据库、分析软件，会使用基本的相关数据库进行搜索等。

By the end of the course, students will have a knowledge of current medical genomics technology and approaches as well as the types of databases and computational tools available.

#### 17. 课程内容及教学日历（如授课语言以英文为主，则课程内容介绍可以用英文；如团队教学或模块教学，教学日历须注明主讲人）

**Course Contents (in Parts/Chapters/Sections/Weeks. Please notify name of instructor for course section(s), if this is a team teaching or module course.)**

| Section | Topic  | Hours |
|---------|--|-------|
| 1       | Overview 绪论 <ul style="list-style-type: none"> <li>● Course introduction, introduction to genomics 课程概况介绍</li> <li>● Introduction of Human Genomics Project 人类基因组计划介绍</li> </ul>   | 3     |
| 2       | Gene and Genome 基因和基因组 <ul style="list-style-type: none"> <li>● Basic concept of gene</li> <li>● Genome and Genomics</li> <li>● Structure of Genome</li> </ul>   | 3     |
| 3       | Genome mapping 基因组绘图 <ul style="list-style-type: none"> <li>● Genetic mapping 遗传图绘制</li> <li>● Physical mapping 物理图绘制</li> </ul>   | 3     |
| 4       | Genome Sequencing 基因组测序 <ul style="list-style-type: none"> <li>● First generation sequencing 一代测序</li> <li>● Next generation sequencing 二代测序</li> <li>● Third generation sequencing 三代测序</li> <li>● Applications 在医学和生命科学中的应用</li> </ul> | 3     |
| 5       | Genome annotation <ul style="list-style-type: none"> <li>● Gene annotation</li> <li>● Function annotation</li> <li>● Gene structure and function</li> </ul>  | 3     |
| 6       | Databases 数据库 <ul style="list-style-type: none"> <li>● Database resources (GenBank, Ensembl, UCSC) 常见数据库介绍</li> <li>● Database search strategy 数据库检索策略</li> </ul>  | 3     |
| 7       | Comparative genomics <ul style="list-style-type: none"> <li>● Multiple sequence alignment 多序列比对</li> </ul>   | 3     |

|    |  |   |
|----|--|---|
|    | <ul style="list-style-type: none"> <li>● Phylogenetic tree estimation 系统发育树构建</li> <li>● Ortholog prediction 直系同源基因预测</li> <li>● Applications 相关应用</li> </ul>  |   |
| 8  | <p>Transcriptomics 转录组学</p> <ul style="list-style-type: none"> <li>● Transcriptome 转录组</li> <li>● Transcriptomics techniques 转录组学技术</li> <li>● Applications 相关医学应用</li> </ul>  | 3 |
| 9  | <p>Population Genomics 群体基因组学</p> <ul style="list-style-type: none"> <li>● Genetic variation in populations 群体中的基因变异</li> <li>● Linkage disequilibrium 连锁不平衡</li> <li>● Natural selection and adaptation 中性选择和适应</li> </ul>      | 3 |
| 10 | Presentation and discussion PPT 汇报讨论   | 3 |
| 11 | <p>Genome wide association studies (GWAS) 全基因组关联分析</p> <ul style="list-style-type: none"> <li>● Association analysis 关联分析</li> <li>● GWAS and diseases GWAS 和疾病</li> </ul>   | 3 |
| 12 | <p>Databases and algorithms 数据库与算法</p> <ul style="list-style-type: none"> <li>● GWAS catalog etc. 主要数据库介绍</li> <li>● introduction to Linux Linux 入门</li> </ul>   | 3 |
| 13 | GWAS analysis practice GWAS 分析练习   | 3 |
| 14 | <p>Epigenomics 表观基因组学</p> <ul style="list-style-type: none"> <li>● Chromatin structure 染色质结构</li> <li>● Histone modifications 组蛋白修饰</li> <li>● Regulatory mechanisms 调控机理</li> <li>● Epigenomics and health 表观基因组学与健康</li> </ul> | 3 |
| 15 | Genome Editing 基因组编辑   | 3 |

|    |  |   |
|----|--|---|
|    | <ul style="list-style-type: none"> <li>● DNA-repair, knock-out, knock-in DNA 修复, 敲除, 敲进</li> <li>● Applications of genome editing 基因编辑的应用</li> <li>● Ethical considerations 伦理的考虑</li> </ul> |   |
| 16 | <p>Final assessment</p> <ul style="list-style-type: none"> <li>● Review of the course 课程总结</li> <li>● Presentation and discussion 汇报讨论</li> </ul>  | 3 |

18. 教材及其它参考资料 **Textbook and Supplementary Readings**

杨金水主编, 高等教育出版社, 基因组学(第4版), 2019

Terry A. Brown, Genomics, CRC Press, 5th Edition, 2013



课程评估 ASSESSMENT

| 19. 评估形式<br>Type of Assessment   | 评估时间<br>Time | 占考试总成绩百分比<br>% of final score | 违纪处罚<br>Penalty | 备注<br>Notes |
|--|--------------|-------------------------------|-----------------|-------------|
| 出勤 Attendance  | 每次上课         | 10%                           |                 |             |
| 课堂表现<br>Class Performance  | 每次上课         | 20%                           |                 |             |
| 小测验<br>Quiz  |              |                               |                 |             |
| 课程项目 Projects  | 第十四次课        | 30%                           |                 |             |
| 平时作业<br>Assignments  |              |                               |                 |             |
| 期中考核<br>Mid-Term Assessment  |              |                               |                 |             |
| 期末考试<br>Final Exam   |              |                               |                 |             |
| 期末报告<br>Final Presentation   | 第十六次课        | 40%                           |                 |             |
| 其它（可根据需要<br>改写以上评估方式）<br>Others (The above may be modified as necessary) |              |                               |                 |             |

Southern University of Science and Technology

20. 记分方式 GRADING SYSTEM

- A. 十三级等级制 Letter Grading  
 B. 二级记分制（通过/不通过） Pass/Fail Grading

课程审批 REVIEW AND APPROVAL

21. 本课程设置已经过以下责任人/委员会审议通过  
 This Course has been approved by the following person or committee of authority