

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.	课程名称 Course Title	大数据与精准医学 Big data and precision medicine
2.	授课院系 Originating Department	医学院 School of Medicine
3.	课程编号 Course Code	MED224
4.	课程学分 Credit Value	3
5.	课程类别 Course Type	专业选修课 Major Elective Courses
6.	授课学期 Semester	春季 Spring
7.	授课语言 Teaching Language	中英双语 English & Chinese
8.	授课教师、所属学系、联系方式 (如属团队授课, 请列明其他授课教师) Instructor(s), Affiliation & Contact (For team teaching, please list all instructors)	胡宇慧、医学院、huyh@sustech.edu.cn Yuhui Hu, School of Medicine, huyh@sustech.edu.cn
9.	实验员/助教、所属学系、联系方式 Tutor/TA(s), Contact	待公布 To be announced
10.	选课人数限额(可不填) Maximum Enrolment (Optional)	无 None

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

教学大纲及教学日历 SYLLABUS

15. 教学目标 Course Objectives

随着人类基因组计划的完成，生命科学进入“后基因组时代”，迎来以系统生物学和功能基因组学为代表的新兴学科快速发展，也发明了多种尖端的组学工具，特别是高通量 DNA 测序技术、基于单细胞的组学工具和众多的生物信息学工具/数据库。此类“多组学大数据”为系统层面研究人类复杂的生理/病理分子网络提供了机会，而不仅专注于单一的基因/蛋白质/通路。将此类组学工具应用于患者个体和药物干扰的生物模型，可以让我们破译导致对临床治疗产生不同个体反应的遗传和非遗传差异，并通过个性化的医疗方案实现最有效的精准治疗，即“精准医学”（PM）。本课程旨在提供 PM 领域中的两个重要组成模块/部分：1) **精准医疗中多组学大数据组成和组学技术**，包括功能基因组学背景知识，多种前沿组学技术和大数据的组成类型、技术发展和应用特征；2) **多组学技术和大数据在个体化精准医疗的临床运用**。通过本课程学习，学生将熟练掌握多组学的基本概念和知识，基因组/转录组学/蛋白质组学/代谢组学等多领域大数据的产生和相关尖端组学工具，并掌握如何用于研究导致疾病发展和治疗反应个体差异的遗传/非遗传性变异。此外，学生可掌握药物基因组学和精准医学的现状、前景、机遇和挑战、伦理等方面的知识，并发表自己的见解。

With the success of Human Genome Project, life science entered the “post-genomic era”. The rapid advancement in Functional Genomics & Systems Biology, e.g. the high-throughput DNA sequencing technologies, single cell-based omics tools, and the numerous bioinformatics tools/databases, has opened the chance to look at the complex biological networks at global level rather than focusing on single gene/protein/pathway. Applying such omics tools on patient individuals and drug-perturbed biologic models could allow us to decipher the genetic and non-genetic variations responsible for diverse responses to therapeutic drugs, and in long-term goal to achieve the most efficient therapy through personalized medicine, namely “Precision Medicine” (PM). This course delivers two main modules about PM: 1) **Composition of multi-omics big data & omics tools in Precision Medicine**, including concept of multi-omics, technological development & characteristics for big data production and analyses; 2) **Case studies of application of multi-omics tools and big data for PM on clinic**. Within this course, the students are expected to get familiar to basic concepts and knowledge of multi-omics, including cutting-edge omics tools and big data production and analyses in genomics/transcriptomics/proteomics/metabolomics to study genetic/non-genetics variations responsible for disease development and therapeutic responses. Through the course learning, the students are expected to grasp the knowledge on the status, perspective, opportunities, and challenges, as well as ethics of pharmacogenomics and precision medicine, and also to deliver their own opinions.

16. 预达学习成果 Learning Outcomes

通过本课程学习，学生将熟练掌握“后基因组”时代系统生物学领域多种组学大数据的基本概念、数据产生的技术变革，特别是以基因组学/转录组学/表观调控组学/蛋白质组学/代谢组学/表型组学水平的相关尖端组学工具，并掌握如何用于研究导致疾病发展和治疗反应个体差异的遗传/非遗传性变异。此外，学生可学习与临床紧密相关的多种疾病精准医疗实例，掌握药物基因组学和精准医疗的临床现状、前景、机遇和挑战、伦理等方面的知识，并发表自己的见解。

Through the study of this course, students will be proficient in the basic concepts and knowledge of multi-Omics of Systems Biology that are emerging in the post-Genomic era, in particular, the cutting-edge omics tools in the context of genomics/transcriptomics/epigenomics/proteomics/metabolomics/phenomics, and also learn to use them to study disease development and genetic/non-genetic variation of individual differences in therapeutic response. In addition, students can learn multiple clinical cases of precision medicine in treating different types of diseases, and master the status, prospects, opportunities and challenges, ethics, etc. of pharmacogenomics and precision medicine, and express their own insights.

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.)

（教学内容：中英双语）

进入后基因组时代，得益于多组学特别是功能基因组学多种尖端组学工具的出现，医学和生命科学领域获得突飞猛进的发展，产生大量的组学数据，可以在系统生物学层面开展人类疾病产生和治疗过程的研究，并同时揭示个体患者之间的巨大差异。尽管机遇与挑战并存，但实现个性化医疗（即精准医疗）(PM)的重要性已得到广泛认可。

本课程包含以下 2 大核心模块：

模块 1 “精准医疗中多组学大数据组成和组学技术” 涵盖人类基因组的基本概念和范围、功能基因组学（从基因组、表观基因组，到转录组、蛋白质组、代谢组、和表型组）中的关键分子和术语；相应组学工具的历史、概念、技术和应用，包括用于基因组/转录组等分析的第二代和第三代 DNA 高通量测序大数据，以及各组学大数据的特征、应用范围；还涵盖了单细胞技术的最新发展，包括空间位置解析的单细胞技术；在简要介绍大数据分析工具的基础上，重点讲授每类大数据的特征和分析结果诠释。

模块 2 “多组学技术和大数据在个体化精准医疗的临床运用” 涵盖了 PM 的基本概念、研究目标和方法，重点讲授对不同个体患者对药物敏感性和药物代谢至关重要的遗传/基因组、和非遗传变异和多态，并进行应用场景/特点的讲解。此外，本课程还讲授临床组学工具使用、精准医疗相关大数据的管理和医学伦理。

Entering the post-genome era, the rapid advancement in functional genomics have invented multiple cutting-edge omics tools to investigate the human diseases and the therapeutic processes in a systematic way meanwhile disclosing dramatic variations among individual patients. The importance of fulfilling personalized medicine, also namely Precision Medicine (PM), has been widely recognized, albeit possessing both opportunities and challenges.

This course aims to deliver two main modules/parts in PM:

Module-1 “Composition of multi-omics big data & omics tools in Precision Medicine” covers the basic concept and scopes of human genome, key molecular terminologies in functional genomics (from genome, epigenome, to transcriptome, proteome, metabolome, and phenome); the history, concept, technique, and applications of cutting-edge omics tools including next-generation sequencing (2nd & 3rd generation) for genome/transcriptome analyses, as well as characteristics and applications of multi-omics big data. The most recent development on single-cell technologies, including spatially resolved single-cell technologies are also be covered. The analytic tools are briefly introduced with the focuses on the result interpretation and analyses of multi-omics big data.

Module-2 “Case studies of application of multi-omics tools and big data for PM on clinic” covers the basic concept, goals, and approaches of PM, which discovers genetic/genomic and non-genetic variations critical to drug sensitivity and metabolism for individual patient. The key omics tools and analytical methods introduced in Module-1 are also discussed for pharmacogenomic studies. Moreover, the ethics to use omics tools and management of multi-omics big data are also discussed in the course.

（教学日历：中英双语）

Section	Topic	Hours
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1	<p>课程概述</p> <ul style="list-style-type: none"> Overview of Precision Medicine & multi-omics data 精准医学和多组学大数据基本概念及发展简史 Contents of the course 课程内容介绍 	2
Module-1	Composition of big data in Precision Medicine 精准医疗中大数据组成	
2	<p>Lecture-2 Concept & Development of Human Genomics and Systems biology 人类基因组学和系统生物学概述、发展</p> <ol style="list-style-type: none"> Important Molecules in Molecular Biology 分子生物学重要分子和概念 The Human Genome Project 人类基因组项目 The structure of human genome 人类基因组结构组成 From Genes to Phenotypes 从基因到表型 Genomics Nomenclature 基因组学术语 	3
3	<p>Lecture-3 Multi-omics big data 多组学大数据组成</p> <ol style="list-style-type: none"> From Genome to transcriptome to proteome & metabolome 从基因组、转录组、到蛋白组调控 Big data from Genome to Disease 从基因组到疾病相关大数据 <ul style="list-style-type: none"> Sequence variations in coding region affect gene & protein expression 影响转录组和蛋白组的表达谱的基因组编码区序列变异 Sequence variations in non-coding region affect gene expression 影响转录组的表达谱的基因组非编码区序列变异 	3
Module-2	Big data in Genomics 基因组学中的大数据	
Section 2.1	<p>Big data of Nucleic Acids (Genomics tools) 核酸大数据 (基因组研究工具) (Methods for DNA sequences 确定 DNA 序列方法 polymerase chain reaction, Sanger sequencing, microarray for gene expression profiling, next generation sequencing PCR、Sanger 测序、DNA 芯片、新一代 DNA 高通量测序)</p>	
4	<p>Lecture-4: DNA sequence determination methods 确定 DNA 序列方法</p> <ul style="list-style-type: none"> PCR and Sanger sequencing PCR 和 Sanger 测序 Microarray technology 	3
5	<p>Lecture-5: DNA sequence determination methods (continued)—2nd generation sequencing 二代基因组测序技术</p> <ul style="list-style-type: none"> 2nd-generation sequencing techniques 二代基因组测序技术介绍 The type of second-generation sequencing methods 二代基因组测序种类 The principle of second-generation sequencing methods 二代基因组测序原理 	3
6	<p>Lecture-6: DNA sequence determination methods (continued)—3rd generation sequencing 三代基因组测序技术</p> <ul style="list-style-type: none"> 3rd-generation sequencing techniques 三代基因组测序技术介绍 Nanopore, PacBio, 	3
Section 2.2	<p>Applications of Next-Generation Sequencing (NGS) & Big data analyses DNA 高通量测序技术的应用、大数据分析 Application in Genome/Chromosome DNA, Transcriptome, Epigenomics, Protein/DNA/RNA binding & Translation, etc. 基因组/染色体 DNA、转录组、表观基因组、DNA/RNA 结合蛋白</p>	

	&翻译调控等领域应用	
7	<p>Lecture-7:</p> <ol style="list-style-type: none"> 1) Whole-genome sequencing; Exosome sequencing 全基因组测序、外显子测序 2) Transcriptome sequencing (RNA-seq) 转录组测序 <ul style="list-style-type: none"> • Techniques of RNA-seq: 转录组测序方法分类 poly(A) mRNA-seq; 3' mRNA-seq; rRNA-depleted RNA-seq • Definition, advantage/disadvantage of each 各技术的定义和优劣势 • Applications of RNA-seq 转录组测序应用 	3
8	<p>Lecture-8:</p> <ol style="list-style-type: none"> 1) Application in Epigenomic regulation 表观基因组调控应用 <ul style="list-style-type: none"> • DNA/RNA modifications e.g. methylation –Methylome DNA/RNA 修饰（甲基化） • Histone modifications and binding • Nucleosome positioning and occupancy • Chromatin accessibility/openness • 3D Chromatin structures 2) Protein/DNA/RNA binding & Translation, etc. DNA/RNA 结合蛋白&翻译调控等领域应用 <ul style="list-style-type: none"> • ChIP-seq, CLIP & PAR-CLIP-seq, Polysome profiling, GRO-seq 	3
Section 2.3	Single-cell technology—Detection of cell heterogeneity at diverse level. 单细胞技术—多层次检测单细胞异质性	
9	<p>Lecture-9: single cell Omics (scRNA-seq, scATAC-seq, scRNA proteomics) 单细胞多组学（单细胞转录组、染色质开放、蛋白组）</p> <ol style="list-style-type: none"> 1) Single-cell transcriptomics 单细胞转录组 <ul style="list-style-type: none"> • Aim & history of single-cell technology development 单细胞技术发展历史和目的 • Techniques and applications of single-cell RNA-seq 单细胞转录组技术和应用 2) Single-cell multi-omics 单细胞多组学 <ul style="list-style-type: none"> • Principles & Techniques for single-cell epigenomic & proteomic information 单细胞表观组学和蛋白组学原理和技术 	3
10	<p>Lecture-10: Spatially resolved single-cell transcriptomics (3 hrs) 单细胞空间转录组学</p> <ul style="list-style-type: none"> • Principle & aim of spatial transcriptomics 空间组学原理和目的 • Techniques to gain spatial gene expression 基因表达空间信息技术 • Applications of spatial single-cell transcriptomics 单细胞空间组学应用 	3
11	<p>Mid-term summary & assessment: 期中总结和考查</p> <p>Lecture-11: Teamwork & Presentation: literature oral presentation “the omics data technology & application in life science” 文献分组讨论和口头汇报 “组学数据在生命科学中的运用”</p>	3
Module-3	Big data in Proteomics & Metabolomics & Phenomics 蛋白组学/代谢组学/表型组大数据	
12	<p>Lecture-12: Mass Spectrometry-Based Proteomics & Metabolomics 组学技术—基于质谱的蛋白组、代谢组定量技术</p> <ul style="list-style-type: none"> • Basic principle of Proteomics, Techniques & applications, Results Analysis 蛋白组学基本概念、原理、应用、结果分析 • Basic principle of Metabolomics, Techniques & applications, Result analysis 代谢组学 	3

	<p>基本概念、原理、应用、结果分析</p> <ul style="list-style-type: none"> Basic principle of Phenomics, Data collection and application cases 表型组学基本概念、数据收集和应用实例 	
Module-4	Clinical application of multi-omics big data in Precision Medicine 多组学大数据在精准医疗的临床应用	
13	<p>Lecture-13: Genomics & diseases; Applicable Omics tools 基因组和疾病；组学技术应用 Genetic/genomic and non-genetic variations in Disease development 基因（组）遗传和非遗传变异与疾病发生</p> <ul style="list-style-type: none"> Monogenic diseases 单基因疾病 Complex diseases 复杂疾病 Cancer 癌症 <p>Analytical methods in Clinical Functional Genomics & Population genetics 群体遗传和临床功能基因组分析方法</p>	3
14	<p>Lecture-14: Pharmacogenomics and Precision Medicine 药物基因组和精准医疗</p> <ul style="list-style-type: none"> Introduction of Pharmacogenomics 药物基因组背景介绍 Drug metabolism & Variability in drug response 药物效应和代谢差异 Genetic/genomic and non-genetic variations in drug response and metabolism 药物效应和代谢差异中的遗传和非遗传变异 	3
15	<p>Lecture-15: Big data & Precision Medicine—Case study 大数据和精准医疗实例分析 Big data & Precision Medicine in Oncology, Neurodegenerative Disease, Cardiovascular drugs, etc. 肿瘤、神经退行性疾病、心血管疾病等中的大数据和精准医疗</p>	3
16	<p>Lecture-16: Ethics in Precision Medicine 精准医学伦理 Big omics data and Personalized Medicine: Opportunities and Challenges 多组学大数据与精准个体化医疗：机遇与挑战</p>	2
17	<p>Lecture-17: End-term assessment 期末考查 Summary of the course 课程总结 Guidance for writing a review about “Big Omics Data & Precision Medicine (Opportunities and Challenges in your own opinion)” 综述《组学大数据与精准医疗的机遇与挑战》撰写指导</p>	2

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

<p>1) Clinical Genomics, 1st Edition, 2014, Shashikant Kulkarni, John Pfeifer, ISBN: 9780124047488</p> <p>2) Human Genetics and Genomics, 4th Edition, Bruce R. Korf, Mira B. Irons, ISBN: 978-0-470-65447-7</p> <p>3) Pharmacogenomics, Challenges and Opportunities in Therapeutic Implementation, 2nd Edition, 2018, Yui-Wing Francis Lam, Stuart Scott ISBN: 9780128126271</p> <p>Supplementary materials: assigned literatures along with lectures. 以及课堂添加的最新文献进展。 Teaching courseware, video, & lecture powerpoints prepared by teacher. 老师准备的教学课件，视频和课堂幻灯</p>
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课程评估 **ASSESSMENT**

19. 评估形式 Type of Assessment	评估时间 Time	占考试总成绩百分比 % of final score	违纪处罚 Penalty	备注 Notes
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出勤 Attendance		10		
课堂表现 Class Performance		10		
小测验 Quiz		20		
课程项目 Projects		30		Teamwork & Presentation: literature oral presentation “the omics data technology & application in life science” 文献分组讨论和口头汇报 “组学数据在生命科学中的运用”
平时作业 Assignments				
期中考试 Mid-Term Test				
期末考试 Final Exam				
期末报告 Final Presentation		30		Write a review with 1500 words about “Big Omics Data & Precision Medicine (Opportunities and Challenges in your own opinion)” 撰写 1500 字英文综述《组学大数据与精准医疗的机遇与挑战》。查重率 <10%。
其它（可根据需要 改写以上评估方式） Others (The above may be modified as necessary)				

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