

## 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>	遗传咨询 Genetic counseling
2.	<b>授课院系 Originating Department</b>	医学院 School of Medicine
3.	<b>课程编号 Course Code</b>	MED323
4.	<b>课程学分 Credit Value</b>	2
5.	<b>课程类别 Course Type</b>	专业选修课/ Major Elective Courses
6.	<b>授课学期 Semester</b>	春季/ Spring
7.	<b>授课语言 Teaching Language</b>	英文 English
8.	<b>授课教师、所属学系、联系方式 Instructor(s), Affiliation &amp; Contact</b> (For team teaching, please list all instructors)	孟焯, 医学院, mengx@mail.sustech.edu.cn; Xuan Meng, School of Medicine, mengx@mail.sustech.edu.cn
9.	<b>实验员/助教、所属学系、联系方式 Tutor/TA(s), Contact</b>	
10.	<b>选课人数限额(可不填) Maximum Enrolment (Optional)</b>	

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			64		64
12. 先修课程、其它学习要求 Pre-requisites or Other Academic Requirements	无/NA				
13. 后续课程、其它学习规划 Courses for which this course is a pre-requisite	待定/To be determined				
14. 其它要求修读本课程的学系 Cross-listing Dept.	待定/To be determined				

### 教学大纲及教学日历 SYLLABUS

#### 15. 教学目标 Course Objectives

遗传咨询是发达国家新兴的交叉学科，通过对患者遗传信息的处理和解码，为患者提供疾病的遗传支持与相应的解决方案。鉴于临床中遗传疾病的重要性，遗传咨询是很多临床医师需要掌握的必备技能。本课程介绍各类遗传疾病的原理与检测方法，通过实践的教学方式使学生对各种检测原理和技术充分了解，并掌握遗传咨询的数据分析方法和患者沟通技巧。

Genetic counseling is a newly popular crossing course in developed countries. Through the detailed analysis and interpretation of the genetic information of patients, genetic counselor offers the genetic solution for patients. Genetic counseling is a basic technology that a clinician should master. This course offers the introduction to various types of genetic diseases, in a combination of theory teaching and clinical practice, students can know various types of genetic counseling tests and master the basic data analysis of genetic information and patients communication skills.

#### 16. 预达学习成果 Learning Outcomes

本课程结束后，学生应该掌握遗传咨询的基本概念和流程，熟悉常见遗传疾病的表征与检测方法，精通遗传基因组学技术和遗传变异报告的解读分析，并能够与病人沟通遗传报告。

At the end of this course, students should master the basic concepts and processes of genetic counseling, be familiar with the characterization and detection methods of common genetic diseases, be proficient in genetic genomics technology and interpretation and analysis of genetic variation reports, and be able to communicate genetic reports with patients.

#### 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	遗传咨询概论与遗传原理 Introduction to genetic counseling and genetic principle	4
2	染色体水平遗传病极其诊断方法 Chromosome level genetic diseases and their diagnosis	4
3	遗传变异的分类标准与指南	4

	The classification of genetics variants and genetic guidelines	
4	产前检测 Prenatal testing	4
5	遗传代谢疾病及诊断方法 Metabolic disease and their diagnosis	4
6	单基因遗传病及诊断方法 Mitochondrial disease and their diagnosis	4
7	复杂疾病的遗传咨询策略 Genetic counseling of complicated diseases	4
8	个体化用药与遗传咨询 Personalized medicine and genetic counseling	4
9	散发性肿瘤和靶向治疗遗传咨询 Non hereditary cancer and targeted therapy genetic counseling	4
10	遗传性肿瘤与诊断治疗 Hereditary cancer genetic counseling	4
11	NGS 在遗传咨询中应用与报告解读 NGS in genetic counseling	4
12	临床遗传咨询：头颈部疾病 Clinical genetic counseling: head and neck disease	4
13	临床遗传咨询：心血管系统疾病 Clinical genetic counseling: vascular disease	4
14	临床遗传咨询：神经肌肉系统疾病 Clinical genetic counseling: nervous system and muscle disease	4
15	临床遗传咨询流程与实例讨论 Clinical genetic counseling process and panel discussion	4
16	学生汇报 Student reports	4

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

教材 Textbook:

Practical genetic counseling, Peter S. Harper. 7<sup>th</sup> Edition, 2019

**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
出勤 Attendance				
课堂表现 Class Performance		10		
小测验 Quiz				

课程项目 <b>Projects</b>		30		Lab Notebook
平时作业 <b>Assignments</b>		30		Lab Report
期中考试 <b>Mid-Term Test</b>				
期末考试 <b>Final Exam</b>				
期末报告 <b>Final Presentation</b>		30		Group Oral Presentation
其它（可根据需要 改写以上评估方 式） <b>Others (The above may be modified as necessary)</b>				

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**

本课程已经医学院教学副院长张文勇教授审核通过。

