

## 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。

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1. 课程名称 Course Title	医学遗传学 Medical Genetics
2. 授课院系 Originating Department	医学院 School of Medicine
3. 课程编号 Course Code	MED404
4. 课程学分 Credit Value	3
5. 课程类别 Course Type	专业基础课 Major Foundational Courses
6. 授课学期 Semester	秋季 / Fall
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)	董金堂, 教授, 南方科技大学医学院 dongjt@sustech.edu.cn 0755-88018032 DONG Jintang, Professor, School of Medicine, Southern University of Science and Technology Email: dongjt@sustech.edu.cn 0755-88018032
9. 实验员/助教、所属学系、联系方式 Tutor/TA(s), Contact	无 NA/
10. 选课人数限额(可不填) Maximum Enrolment (Optional)	

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	0	0	0	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

医学遗传学是将遗传学理论应用于医学实践，研究人类疾病与遗传关系的一门学科。本课程的教学目标为：

1. 培养学生掌握遗传学的基本概念和原理、掌握常见遗传疾病的遗传学机制，提高学生探讨医学遗传问题的兴趣。
2. 适当培养学生能结合医学实际，对临床上常见遗传病的诊断、检验、预防、治疗及遗传咨询有初步的分析判断能力。
3. 介绍医学遗传学科的新成就、新技术，为进行有关的研究工作和进一步学习医学知识打下基础。

Medical genetics is a new subject which applies genetic theory to medical practice and studies the relationship between human diseases and heredity. The teaching objectives of this course are:

- (1) Training students to master the basic concepts and principles of genetics. Training students to grasp the mechanisms of common genetic diseases. Enhancing students' interest in medical genetic research.
- (2) Properly training students to have the ability of critical analysis and judgment for the common genetic diseases in the diagnosis, testing, prevention, treatment and genetic counseling.
- (3) Introducing the new achievements and new technologies in the field of medical genetics. Thus, it lays a solid foundation for related research work and further study of medical knowledge.

#### 16. 预达学习成果 Learning Outcomes

通过本门课程的学习使学生掌握医学遗传学的基础知识，包括基因的结构、功能与突变，正常人类染色体基础知识，临床常见的单基因病与多基因病，线粒体疾病，染色体疾病，遗传与肿瘤发生的关系等。要求学生能够：

1. 掌握五大类遗传性疾病的遗传缺陷和基本特点；
2. 掌握和熟悉常见的遗传性疾病的发病机理、遗传学改变及主要临床特征；
3. 初步做到理论联系实际，能识别常见的遗传病，并能估计它在患者亲属中的发生风险。

Through the course, students can master the basic knowledge of medical genetics, including the structure, function and mutation of genes, basic knowledge of normal human chromosomes, common clinical monogenic diseases and polygenic diseases, mitochondrial diseases, chromosomal diseases, the relationship between heredity and tumor genesis, etc.

- (1) To grasp the genetic defects and basic characteristics of five kinds of hereditary diseases;

- (2) To grasp and familiarize with the pathogenesis, genetic changes and main clinical features of common hereditary diseases.
- (3) To have the ability of combining theory with practice and can identify common genetic diseases and estimate their recurrence risk in patients' relatives.

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.)

第一章 Chapter 1	<b>遗传学与医学</b> <b>Genetics and Medicine</b> 健康与疾病的遗传基础 Genetic basis of health and disease 医学遗传学简史 A brief history of medical genetics 遗传病的分类及特征 Classification and characteristics of genetic diseases
第二章 Chapter 2	<b>人类基因组与基因</b> <b>Human Genome and Gene</b> 人类基因组组成与遗传规律 Human Genome Composition and Heredity 基因及其表达与调控 Gene and its expression and regulation 基因突变及其生物学效应 Gene mutation and its biological effects
第三章 Chapter 3	<b>表观遗传</b> <b>Epigenetics</b> 表观遗传修饰 Epigenetic modification 表观遗传与疾病 Epigenetics and disease 表观遗传与肿瘤 Epigenetics and tumor 表观遗传与衰老 Epigenetics and aging 表观遗传的生物学意义 Biological significance of epigenetics
第四章 Chapter 4	<b>医学遗传学研究技术</b> <b>Medical Genetics Research Technology</b> 染色体分析技术 Chromosome analysis technique 核酸分析技术 Nucleic acid analysis technology 基因功能分析技术 Gene function analysis
第五章	人类染色体和染色体病

<b>Chapter 5</b>	<b>Human Chromosome and Chromosomal Diseases</b> 人类染色体的基本特征 Basic characteristics of human chromosomes 染色体畸变 Chromosomal aberration 染色体病 Chromosomal disease
<b>第六章</b> <b>Chapter 6</b>	<b>单基因遗传病</b> <b>Monogenous hereditary diseases</b> 常染色体显性遗传 Autosomal dominant inheritance 常染色体隐性遗传 Autosomal recessive inheritance X 连锁显性遗传 X-linked dominant inheritance X 连锁隐性遗传 X-linked recessive inheritance Y 连锁遗传病 Y-linked genetic disease 非经典性孟德尔遗传 Non-classical Mendelian inheritance
<b>第七章</b> <b>Chapter 7</b>	<b>多基因遗传病</b> <b>Polygenic hereditary diseases</b> 多基因遗传病发病机制的相关学说 Related theory of the pathogenesis of polygenic genetic diseases 多基因遗传病致病基因的研究方法 Polygenic genetic disease research method 几种常见的多基因遗传病 Several common polygenic genetic diseases
<b>第八章</b> <b>Chapter 8</b>	<b>群体遗传</b> <b>Population genetics</b> 什么是群体遗传学? What is population genetics? 群体遗传组成 - 基因频率和基因型频率 Population genetic composition - gene frequency and genotype frequency 影响基因频率的因素 Factors affecting gene frequency 近亲婚配及其对群体遗传素质的影响 Close relatives marriage and its influence on the genetic quality of the population
<b>第九章</b> <b>Chapter 9</b>	<b>生化遗传病</b> <b>Biochemical hereditary diseases</b> 生化遗传学 Biochemical Genetics 生化遗传疾病 Biochemical genetic disorders

第十章 Chapter 10	<b>线粒体遗传病</b> <b>Mitochondrial hereditary diseases</b> 线粒体 DNA 的遗传学特点 Genetic characteristics of mitochondrial DNA 线粒体遗传病 Mitochondrial genetic disease
第十一章 Chapter 11	<b>药物反应的遗传基础</b> <b>The genetic basis of drug response</b> 药物遗传学 Pharmacogenetics 药物代谢的遗传变异 Genetic variation of drug metabolism
第十二章 Chapter 12	<b>免疫遗传</b> <b>Immunological inheritance</b> 人类红细胞抗原遗传 Human erythrocyte antigen inheritance 人类白细胞抗原遗传 Human leukocyte antigen inheritance
第十三章 Chapter 13	<b>遗传与肿瘤</b> <b>Genetics and Tumors</b> 染色体异常与肿瘤 Chromosomal abnormalities and tumors 癌基因 Cancer gene 肿瘤抑制基因 Tumor suppressor gene 遗传型恶性肿瘤 Hereditary malignant tumor 肿瘤发生的遗传学理论 Genetic theory of tumorigenesis
第十四章 Chapter 14	<b>临床遗传</b> <b>Clinical genetics</b> 遗传病的诊断 Diagnosis of genetic diseases 遗传病的治疗 Treatment of genetic diseases 遗传病的预防 Prevention of genetic diseases
第十五章 Chapter 15	<b>遗传服务的伦理问题</b> <b>Ethical issues of genetic services</b> 遗传服务 Genetic services 遗传咨询中的伦理问题 Ethical issues in genetic counseling 基因治疗中的伦理问题

Ethical issues in gene therapy  
辅助生殖中的伦理问题  
Ethical issues in assisted reproduction

第十六章  
Chapter 16

复习和学生报告  
**Review and Student Report**

章节 Chapter	教 学 内 容 Teaching Contents	学 时 Teaching Hours
Chapter 1	遗传学与医学 Genetics and Medicine	3
Chapter 2	人类基因组与基因 Human Genome and Gene	3
Chapter 3	表观遗传 Epigenetics	3
Chapter 4	医学遗传学研究技术 Medical Genetics Research Technology	3
Chapter 5	人类染色体和染色体病 Human Chromosome and Chromosomal Diseases	3
Chapter 6	单基因遗传病 Monogenous hereditary diseases	3
Chapter 7	多基因遗传病 Polygenic hereditary diseases	3
Chapter 8	群体遗传 Population genetics	3
Chapter 9	生化遗传病 Biochemical hereditary diseases	3
Chapter 10	线粒体遗传病 Mitochondrial hereditary diseases	3
Chapter 11	药物反应的遗传基础 The genetic basis of drug response	3
Chapter 12	免疫遗传 Immunological inheritance	3
Chapter 13	遗传与肿瘤 Genetics and Tumors	3
Chapter 14	临床遗传 Clinical genetics	3
Chapter 15	遗传服务的伦理问题 Ethical issues of genetic services	3
	复习和学生报告 Review and Student Report	3

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

教材 **Text book:**

- [1] 陈竺. 医学遗传学 (第3版) [M]. 北京: 人民卫生出版社, 2015.  
[2] 林恩·乔德. Medical Genetics 医学遗传学[M].北京: 人民卫生出版社, 2017.

参考书 **Reference books:**

- [1] 左伋. 医学遗传学 (第 7 版) [M]. 北京: 人民卫生出版社, 2018.
- [2] Lynn B. Jorde. Medical Genetics, 5e[M]. Amsterdam: Elsevier, 2015.
- [3] G. Bradley Schaefer. Medical Genetics[M]. United States: McGraw-Hill Education, 2017.
- [4] Peter D Turnpenny, Sian Ellard. Emery's Elements of Medical Genetics[M]. Amsterdam: Elsevier, 2017.

### 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
出勤与课堂表现 Attendance and Class Performance		5		出勤与课堂表现综合评价占 5%。 Attendance and overall classroom performance accounted for 5%.
小测验 Quiz				
课程项目 Projects				
平时作业 Assignments				
平时考试 Monthly Test		80		共 4 次考试 (平均每个月一次考试) 每次考试占总成绩 20%, 4 次合计占总成绩 80%。 Totally four tests (average monthly). Each exam accounted for 20%. Four exams accounted for 80%.
期中考试 Mid-Term Test				
期末考试 Final Exam				
期末报告 Final Presentation		15		写一篇与遗传性疾病有关的论文 Write a paper on medical genetic disease
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

本课程已经医学院教学副主任张文勇教授审核通过。

