课程大纲
COURSE SYLLABUS

1.	**课程代码/名称** **Course Code/Title**	**医学遗传学与精准医学前沿 Frontiers in Medical Genetics and Precision Medicine**
2.	**课程性质** **Compulsory/Elective**	专业选修/Elective
3.	**开课单位** **Offering Dept.**	医学院/ School of Medicine
4.	**课程学分/学时** **Course Credit/Hours**	3
5.	**授课语言** **Teaching Language**	中英双语 English/Chinese
6.	**授课教师** **Instructor(s)**	李职秀
7.	**开课学期** **Semester**	春/秋季 Spring/ Autumn
8.	**是否面向本科生开放** **Open to undergraduates or not**	否 No
9.	**先修要求** **Pre-requisites**	（如面向本科生开放，请注明区分内容。 If the course is open to undergraduates, please indicate the difference.） 无

10. 教学目标
Course Objectives

（如面向本科生开放，请注明区分内容。 If the course is open to undergraduates, please indicate the difference.）

本课程拟面向研究生系统介绍医学遗传学方面的前沿研究进展，着重介绍人类疾病与遗传的关系、遗传病的发生机制、遗传特点，提供诊断、治疗和预防遗传病的依据及方法，以及医学遗传学在精准医学中的前沿应用，使学生了解医学遗传学与人类健康的基本理念以及最前沿的科学研究和发展趋势，为从事医学研究以及公共卫生体系建设贡献力量。

This course aims to systematically introduce graduate students to the latest advances in medical genetics research. It focuses on illustrating the relationship between human diseases and genetics, the pathogenesis and genetic characteristics of genetic disorders, and providing diagnostic, therapeutic, and preventive principles and methods for genetic diseases. The course enables students to understand the fundamental concepts of medical genetics and human health, as well as cutting-edge scientific research and development trends. It strives to equip students with essential knowledge and skills to contribute to medical research and public health system building.

11. 教学方法
Teaching Methods

（如面向本科生开放，请注明区分内容。 If the course is open to undergraduates, please indicate the difference.）

1. 导师课程讲授为主：用多媒体教学方式授课，促进学生了解课程前沿进展；
2. 采用引入式教学，同时理论联系实际，增强课程的互动性及趣味性；
3. 学生文献阅读，讨论和演讲为辅：培养学生的自主阅读及演讲交流能力。

12. 教学内容
Course Contents
（如面向本科生开放，请注明区分内容。 If the course is open to undergraduates, please indicate the difference.）

Section 1	1. 导论 Introduction 1.1 课程介绍 Course Introduction 1.2 遗传学历史 History of Genetics 1.3 遗传与医学 Genetics and Medicine
Section 2	2. 人类基因和基因组 Human Genes and Genome 2.1 基因及其结构 Genes and Structure 2.2 基因组的组成 Composition of the Genome 2.3 基因复制与表达 Gene Replication and Expression
Section 3	3. 基因突变 Gene Mutations 3.1 基因突变的特性与诱发因素 Characteristics and Inducing Factors of Mutations 3.2 基因突变的形式 Types of Mutations
Section 4	4. 单基因疾病的遗传 Genetics of Monogenic Diseases 4.1 单基因遗传的基本概念与研究方法 Basic Concepts and Methods of Monogenic Inheritance 4.2 常染色体显性遗传病的遗传 Genetics of Autosomal Dominant Diseases 4.3 常染色体隐性遗传病 Genetics of Autosomal Recessive Diseases 4.4 X 连锁显性遗传病的遗传 Genetics of X-linked Dominant Diseases 4.5 Y 连锁遗传病的遗传 Genetics of Y-linked Diseases 4.6 影响单基因遗传的因素 Factors Affecting Monogenic Inheritance
Section 5	5. 多基因疾病的遗传 Genetics of Multifactorial Diseases 5.1 质量性状与数量性状 Qualitative and Quantitative Traits 5.2 多基因遗传及特点 Multifactorial Inheritance and Its Characteristics 5.3 多基因疾病的遗传 Genetics of Multifactorial Diseases 5.4 遗传度 Heritability
Section 6	6. 群体遗传学 Population Genetics 6.1 群体中的基因和基因型频率 Gene and Genotype Frequencies in Populations 6.2 遗传平衡定律 Hardy-Weinberg Equilibrium 6.3 影响遗传平衡的因素 Factors Affecting Genetic Equilibrium
Section 7	7. 线粒体疾病的遗传 Mitochondrial Disease Inheritance 7.1 线粒体概述 Overview of Mitochondria 7.2 线粒体的遗传特征 Inheritance Characteristics of Mitochondria 7.3 线粒体突变与疾病 Mitochondrial Mutations and Diseases
Section 8	8. 人类染色体与染色体病 Human Chromosomes and Chromosomal Disorders 8.1 染色质与染色体 Chromatin and Chromosomes 8.2 染色体核型与显带技术 Chromosomal Karyotype and Banding Techniques 8.3 染色体数目异常及其产生机制 Aneuploidy and Its Mechanisms 8.5 常染色体病 Autosomal Disorders
Section 9	9. 分子病与先天性代谢缺陷 Molecular Diseases and Congenital Metabolic Defects 9.1 珠蛋白基因及其表达 Globin Genes and Their Expression 9.2 血红蛋白病 Hemoglobinopathies 9.3 地中海贫血 Thalassemia 9.4 先天性代谢缺陷 Congenital Metabolic Disorders
Section 10	10. 肿瘤遗传学 Cancer Genetics 10.1 肿瘤发生的遗传现象 Genetic Events in Tumorigenesis 10.2 基因组不稳定性与肿瘤发生 Genomic Instability and Tumorigenesis 10.3 肿瘤发生的遗传理论 Genetic Theories of Tumorigenesis

		10.4 肿瘤的分子诊断与靶向治疗 Molecular Diagnosis and Targeted Therapy of Tumors
	Section 11	11. 遗传病的诊断，治疗和预防 Diagnosis, Treatment and Prevention of Genetic Diseases 11.1 Diagnosis of Genetic Diseases 11.2 Treatment of Genetic Diseases 11.3 Prevention of Genetic Diseases
	Section 12	12. 药物基因组学 Pharmacogenomics 12.1 药物基因组学介绍 – Pharmacogenomics 12.2 药物基因组学与药物反应 – Pharmacogenomics and drug response
	Section 13	13. 基因编辑与伦理 Genome Editing and Ethics 13.1 基因编辑和应用 - Genome Editing and Applications 13.2 伦理的考虑 - Ethical Considerations
	Section 14	13 遗传学与精准医学 Genetics and Precision Medicine 13.1 疾病的遗传学大数据 Genetic Big Data in Disease 13.2 精准医学实例分析 Precision Medicine Case study
	Section 15	小组汇报讨论总结 Presentation, Discussion and Review
	Section 16	小组汇报讨论总结 Presentation, Discussion and Review

13. 课程考核
Course Assessment

（①考核形式 Form of examination；②.分数构成 grading policy；③如面向本科生开放，请注明区分内容。If the course is open to undergraduates, please indicate the difference.）

十三级等级制 Letter Grading

出勤 Attendance: 10%

课堂表现 Class Performance: 20%

平时作业 Assignments: 30%

期末大作业 Projects:40%

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

1. 杨保胜,李刚.医学遗传学,2 版.高等教育出版社，2019
2. 傅松滨..医学遗传学,4 版.北京：北京大学医学出版社,2018
3. 王宗霞,杨保胜. 医学细胞生物与遗传学.高等教育出版社，2018
4. Krebs J S, Goldstein E S, Kilpatrick S T. 基因Ⅻ Lewin’s Genes Ⅻ.高等教育出版社.2018
5. Pyeritz R E, Korf B R, Grody W W.Emery and Rimoin's Principles and Practice of Medical Genetics and Genomics. Foundations. 7th ed. Elsevier. 2019