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基本信息:

姓名 : 管敏鑫
 职务 : PI
 职称 : 教授
 学历 : 博士
 专业: 人类遗传学
 所属院系: 遗传学研究所
 研究方向: 线粒体遗传学和母系遗传性疾病基础研究及临床转化
 电话: 0571-88206497
 信箱: gminxin88@zju.edu.cn
 个人主页: <http://mypage.zju.edu.cn/guanmx>

个人简介:

学术团体和社会兼职 :

中国人民解放军总医院/军医进修学院客座教授 (2002-至今)
 美国辛辛那提大学儿童医院医学中心客座教授 (2011.10-至今)
 浙江省医学遗传学重点实验室学术委员会主任 (2005-至今)
 第四届亚洲线粒体研究与医学学会 (ASMRM) 主席 (2011-2014)
 中国遗传学会常务理事 (2014-至今)
 中国遗传学会国际交流委员会主任委员 (2014-至今)

学历和研究经历 :

1979.9-1983.7 杭州大学 (现浙江大学) 生物系本科
 1983.8-1989.9 浙江图书馆 馆员
 1989.10-1993.7 澳大利亚国立大学生物化学与分子生物学博士研究生
 1993.8-1996.7 加州理工学院生物系人类分子遗传学 Research Fellow
 1996.8-1999.7 加州理工学院生物系 Senior Research Fellow
 1999.8-2011.9 辛辛那提大学儿童医院医学中心人类遗传学助理教授、副教授、教授
 2011.1-2013.11 浙江大学生命科学学院 院长
 2011.1-2015.2 浙江大学生命科学学院教授遗传学研究所所长
 2015.3-至今 浙江大学医学院/生命科学学院教授遗传学研究所所长

工作研究领域

国家“千人计划”特聘专家，“973”计划项目首席科学家。长期从事线粒体遗传学和母系遗传性疾病的基因研究和临床转化。作为PI，曾经获得5项美国国立卫生研究院 (NIH) 基金资助。自2011年全职回国后，主持国家重点基础研究发展计划 (973计划) ，“十二五”国家科技支撑计划，国家自然科学基金重点项目等多项课题。已在母系遗传性聋病、高血压和Leber遗传性视神经病变致病机理和tRNA转录后修饰的机制等领域发表论文共174篇，其中在Cell, Am J Hum Genet, Hum Mol Genet, Circulation Research, Ophthalmology等本领域国际权威学术期刊上发表SCI论文100多篇。曾获国家科技进步二等奖、谈家桢生物科学创新奖等多项奖励。主编出版《医学遗传学》等教材。

在研的主要项目 :

1. 国家重点基础研究发展计划 (973 计划) : 单基因遗传性聋病的分子机制研究, 2014.1-2018.12
2. 国家自然科学基金重点项目 : 核修饰基因调控母系遗传性耳聋发病机制及听觉功能重建的策略研究, 2014.1-2017.12
3. 十二五支撑项目 : Leber 遗传性视神经病变的分子诊断和治疗研究, 2012.1-2015.12

专利及代表性论文(*corresponding authors)

国家发明专利 :

用于同时检测线粒体DNA A1555G和C1494T突变的试剂盒及其使用方法.申请号 : 200910223263.8

耳聋遗传

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地址：浙江杭州余杭塘路866号 电话：0571-88208020 传真：0571-88208022
邮箱：zhouzeyong@zju.edu.cn
技术支持：YONCC



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