遗传 2005 27 (3): 466-472 ISSN: 0379-4172 CN: 11-5450/R

专论与综述

表观遗传学与人类疾病的研究进展

张永彪, 褚嘉祐

中国医学科学院 中国协和医科大学 医学生物学研究所 遗传室，昆明 650118

收稿日期 2004-6-30 修回日期 2004-8-4 网络版发布日期 接受日期

摘要 在过去的几年里，人们对表观遗传疾病的机理有了新的认识，这些疾病与染色质重塑、基因组印记、X染色体失活以及非编码RNA调控这4个表观遗传过程相关。这4个过程通过调节染色质结构，在染色体或基因簇水平上对基因表达进行调控；异常调控导致复杂的突变且表现为出生前后生长发育和神经功能的异常。对这些疾病的探讨为表观遗传机制的研究提供了很好的模型，进而有助于生物医学的研究。文章就表观遗传学和表观遗传疾病机制的研究进展做一综述。

关键词 表观遗传；组蛋白修饰；DNA甲基化；基因组印记；X染色体失活；非编码RNA

分类号 R394

Progress of Research on Epigenetic and Human Disease

ZHANG Yong-Biao,CHU Jia-You

Department of Genetics, Institute of Medical Biology, Peking Union Medical College, Kunming 650118, China

Abstract

In the past few fears, there has been a nascent convergence of scientific understanding of human disease with epigenetic. Identified epigenetic processes involved in human disease include chromatin remodeling, genomic imprinting, X chromosome inactivation, and noncoding RNAs regulation. These processes influence chromatin structure and thereby regulate gene expression on the chromosome level or a cluster of linked genes level. Deregulation of these processes result in lots of disease which are characterized by complex patterns of mutations and associated phenotypes affecting pre- and postnatal growth, development, and neurological function. Epigenetic diseases are illustrated by the array of multi-system disorders and neoplasias and investigations of these diseases have an impact on biomedical research and provide interesting models for functions and mechanisms of epigenetic gene control.

Key words epigenetic；histone modification；DNA methylation；genomic imprinting；X chromosome inactivation；noncoding RNA

DOI:

通讯作者 褚嘉祐 chujy@public.km.yn