

РОЛЬ ИНТРОНОВ В ЭВОЛЮЦИИ

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Федеральное государственное бюджетное учреждение науки Института биологии и генетики Уфимского научного центра РАН.

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В статье описана роль интронов в эволюции эукариот. Накапливающиеся в интронных областях полиморфизмы, входящие для сплайсинговых изоформ в рамки считывания, могут приводить к изменению свойства альтернативного продукта экспрессии гена, что дает основу адаптации к меняющимся условиям существования, закрепляясь эволюционно на уровне популяции и вида. Описаны основные механизмы обеспечения динамичности генома эукариот и роль ассоциативных комбинаций в генезе мультифакториальных заболеваний.

Ключевые слова: Альтернативный сплайсинг, избыточная ДНК, мультифакториальные заболевания, мобильные элементы генома, нуклеотидные последовательности, сателлитная ДНК.

ВВЕДЕНИЕ

Описана адаптирующая роль альтернативного сплайсинга к внутренней и внешней среде. В отношении внутренней среды организма регуляция экспрессии определенных сплайсинговых изоформ белков способствует тканеспецифической активности данного белка. В отношении внешней среды альтернативный сплайсинг способствует эволюционным преобразованиям. Сопоставляя накапливающиеся полиморфизмы в интронных областях и возможность экспрессии сплайсинговых изоформ жизненно-важных белков с расширением рамки считывания, можно предположить существование особого способа эволюции у эукариот. Данный способ, в отличие от изменения нормальных изоформ белков под действием мутаций, является более безопасным, что может служить одним из объяснений целесообразности мозаичной структуры генов эукариот. Интронные полиморфизмы, для определенных сплайсинговых изоформ белка могут входить в рамку считывания и влиять на функционирование продукта гена, в том числе в отношении лучшей приспособляемости к изменениям среды, в связи с чем в ряду поколений возможен отбор в пользу экспрессии данных изоформ.

Представлена схема приобретения геномом эукариот свойств динамичности в виде кооперации четырех важных процессов – внедрения транспозонов в геном, появления высокомутабельной сателлитной ДНК, межгенной компенсации и альтернативного сплайсинга. Ключевым моментом

для образования эукариот послужила адаптирующая роль внедрения мобильных генетических элементов в геном прокариот. Внедрение мобильных генетических элементов имеет свои пределы для каждого вида, у организмов в процессе эволюции сформировались механизмы, «сдерживающие» интеграцию транспозонов, ретротранспозонов и ретровирусов в геном. Внедрение транспозонов, ретротранспозонов и ретровирусов послужило основным фактором эволюции эукариот и причиной огромного разнообразия живой природы. Динамичность генома – главное отличительное свойство эукариот от прокариот.

ОСОБЕННОСТИ ГЕНОМА ЧЕЛОВЕКА И ЭУКАРИОТ

Геном человека содержит около 25 000 ядерных генов, при этом кодирующая часть генов составляет менее 3% [7], по последним данным – 1,5% всего генома [25]. Остальная часть ДНК не несет кодирующей функции, при этом 75% нуклеотидных последовательностей (НП) находится между генами [7]. Недавние исследования показали, что геном человека содержит 69% повторяющихся последовательностей, главным образом копий транспозонов [16]. Соотношение ДНК человека в зависимости от повторяемости представлено в табл. 1. Повторы разделены на *LINE*-, *SINE*-, *ALU*-элементы, сателлиты и другие. *LINE*- и *ALU*-элементы распределены по всему геному. Сателлиты расположены, в основном,

Sistemokvanty Fiziologicheskikh Protsebov

Ensheng Dong



Sistemokvanty Fiziologicheskikh Protsebov:

The book delves into Sistemokvanty Fiziologicheskikh Protsebov. Sistemokvanty Fiziologicheskikh Protsebov is an essential topic that must be grasped by everyone, ranging from students and scholars to the general public. The book will furnish comprehensive and in-depth insights into Sistemokvanty Fiziologicheskikh Protsebov, encompassing both the fundamentals and more intricate discussions.

1. The book is structured into several chapters, namely:
 - Chapter 1: Introduction to Sistemokvanty Fiziologicheskikh Protsebov
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 - Chapter 4: Sistemokvanty Fiziologicheskikh Protsebov in Specific Contexts
 - Chapter 5: Conclusion
 2. In chapter 1, the author will provide an overview of Sistemokvanty Fiziologicheskikh Protsebov. The first chapter will explore what Sistemokvanty Fiziologicheskikh Protsebov is, why Sistemokvanty Fiziologicheskikh Protsebov is vital, and how to effectively learn about Sistemokvanty Fiziologicheskikh Protsebov.
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- The book is crafted in an easy-to-understand language and is complemented by engaging illustrations. It is highly recommended for anyone seeking to gain a comprehensive understanding of Sistemokvanty Fiziologicheskikh Protsebov.

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