

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

Yi-Tong Ma



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Whispering the Strategies of Language: An Emotional Journey through **Sistemokvanty Fiziologicheskikh Protsebov**

In a digitally-driven world where displays reign supreme and quick interaction drowns out the subtleties of language, the profound secrets and psychological nuances hidden within words often go unheard. However, situated within the pages of **Sistemokvanty Fiziologicheskikh Protsebov** a captivating fictional prize sporting with fresh feelings, lies an extraordinary journey waiting to be undertaken. Written by a skilled wordsmith, this marvelous opus invites visitors on an introspective journey, gently unraveling the veiled truths and profound impact resonating within the cloth of each and every word. Within the mental depths with this touching review, we shall embark upon a heartfelt exploration of the book's key subjects, dissect its interesting publishing model, and yield to the effective resonance it evokes strong within the recesses of readers' hearts.

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