發育再生研究論文評介

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Epistatic Adaptive Evolution of Human Color Vision

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Abstract

Establishing genotype-phenotype relationship is the key to understand the molecular mechanism of phenotypic adaptation. This initial step may be untangled by analyzing appropriate ancestral molecules, but it is a daunting task to recapitulate the evolution of non-additive (epistatic) interactions of amino acids and function of a protein separately. To adapt to the ultraviolet (UV)-free retinal environment, the short wavelength-sensitive (SWS1) visual pigment in human (human S1) switched from detecting UV to absorbing blue light during the last 90 million years. Mutagenesis experiments of the UV-sensitive pigment in the Boreoeutherian ancestor show that the blue-sensitivity was achieved by seven mutations. The experimental and quantum chemical analyses show that 4,008 of all 5,040 possible evolutionary trajectories are terminated prematurely by containing a dehydrated nonfunctional pigment. Phylogenetic analysis further suggests that human ancestors achieved the blue-sensitivity gradually and almost exclusively by epistasis. When the final stage of spectral tuning of human S1 was underway 45–30 million years ago, the middle and long wavelength-sensitive (MWS/LWS) pigments appeared and so-called trichromatic color vision was established by interprotein epistasis.

The adaptive evolution of human S1 differs dramatically from orthologous pigments with a major mutational effect used in achieving blue-sensitivity in a fish and several mammalian species and in regaining UV vision in birds. These observations imply that the mechanisms of epistatic interactions must be understood by studying various orthologues in different species that have adapted to various ecological and physiological environments.

作者提要

檢定基因型－表現型的相互關係是瞭解不同的表現型在自然界如何演化出來的必要工作。

人類對藍色敏感的視覺色素(human S1)是從其Boreoeutherian先祖的對紫外線敏感色素（AncBoreotheria S1)經過七輪突變變異實驗顯示連結AncBoreotheria S1和human S1之間可能的5040演化路徑，有4008（約80％）會提前中斷。

量子化學分析顯示：這些會提前中斷的演化路徑都是因為含有一個脫水的色素所致。物種遺傳分析更顯示：人眼對藍色的敏感度一定要七個藉由互不相加的相互作用之氨基酸才能逐步達成。

在4500萬到3000萬年前human S1進入發展出藍色敏感度的最後階段，而此時也是人類對紅色敏感基因複製成兩個，而其中之一變成對綠色敏感。

最後，人類的三原色色覺在約3000萬年前，藉由三種視覺色素的蛋白質相互壓制(interprotein epistasis)而達成。

操弄受到遺傳修飾的先祖蛋白質是分別重演表現型適應的演化過程以及蛋白質壓制性互動的關鍵。