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• 計畫英文名稱	Protein Interaction with the Human c21 Gene Promoter.
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• 研究人員	張淑芬 Chang, Shu-Fen
• 中文關鍵字	腎上腺增生症；起動子活性；類固醇水解酵素
• 英文關鍵字	Adrenal hyperplasia；Promoter activity；Steroid hydroxylase
• 中文摘要	類固醇 21-hydroxylase(CYP21)酵素活性的缺失是造成先天性遺傳疾病,腎上腺增生症的主要原因。此酵素活性的缺失是起因於此酵素基因 CYP21A2 發生突變。在 CYP21A2(c21B)基因上突變的發生主要是由於基因的轉換變成其鄰近具高相似性但無活性之假基因的核酸序列。我們早期的實驗結果顯示在 CYP21A2(c21B)基因上游約 170bp 之區域決定了此基因基礎起動子活性的強度。而此活性基因與其假基因在此區域內,僅有四個核酸序列的差異。在此篇報告中,我們主要是針對各別核酸序列,對其與核蛋白質因子之間結合情形加以研究。結果顯示那四個核酸序列均對此區域 DNA 蛋白質之結合非常重要。雖然影響的程度不同。另外,我們亦發現一蛋白質存在腎上腺皮質的萃取液可能與此活性基因的表現有關。而存在另一蛋白質可能與此基因在腎上腺皮質的特異性表現有關。
• 英文摘要	Deficiency in steroid P450c21-hydroxylase (P450c21; CYP21) is the major cause of congenital adrenal hyperplasia, a common genetic disease due to steroid imbalance. The main cause for mutation occurred on the CYP21A2 (c21B) gene is conversion of its nucleotide sequence to the neighboring homologous but nonfunctional c21A gene. Our previous results have shown that determinant for basal promoter activity of the c21 gene was located at the upstream 170bp region (Chang and Chung, 1995). Within that region, there are only four nucleotide differences between c21A and c21B genes. In this report, we have analyzed the importance of those individual nucleotides in their ability to interact with nuclear protein. Gel mobility shift assay showed that those four nucleotides at position -117, -104, -101, and -94 were all required for interaction with nuclear protein though to different degree. A nuclear protein with molecular weight about 45Kd might be an adrenal-specific factor that activate the c21B gene expression but not c21A gene expression. Another 70Kd protein was responsible for the adrenal-specific expression of c21 gene.