# 痣樣基底細胞癌症候群病人 ptch 基因新的點突變之報告

# A novel nonsense mutation at the Human PTCH gene in a patient with nevoid basal cell carcinoma

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**摘要**

痣樣基底細胞癌症候群是一種具有高度滲透性體及多種表現的体染色體顯性遺傳疾病。其主要的表徵有基底細胞上皮癌，成齒的角質囊腫，手掌和腳蹠角化不良的凹痕，顱骨內鈣化和很多種其他骨骼發育異常。這個疾病被認爲和人類的 ptch 基因突變有關。這裡我們提出一位痣樣基底細胞癌症候群的病人在 ptch 基因上去氧核醣核酸第 403 位置有 G 變成 A 的點突變，而導致在 ptch 基因所轉譯的第 131 個氨基酸造成終結。這個結果進一步支持在人類癌症的發生上 ptch 基因具重要腫瘤抑制的活性。

**Abstract**

The nevoid basal cell carcinoma syndrome (NBCCS) is a highly penetrative but variably expressed autosomal-dominant disorder. The major manifestations are basal cell carcinomas, odontogenic keratinocysts, dyskeratotic pitting of the palms and soles, intracranial calcification, and other developmental skeletal abnormalities. The disorder has beensuggested to result from human ptch gene mutation. Here we show a NBCCS patient with a G-to-A substitution at nucleotide 403 position of the ptch gene resulting in premature termination at codon 131. This finding provides further support for the important tumor suppressor activity of the ptch gene in the development of human cancer.