台灣腸胃道間質腫瘤之發生率，免疫化學染色及基因突變之分析研究

Gastrointestinal stromal tumors in Taiwan: The incidence, immunohistochemical staining and mutation analysis

中文摘要

腸胃道基質瘤(gastrointestinal stromal tumor, GIST)乃是源自腸胃壁肌肉層的腫瘤，是最常見的間質腫瘤。其診斷的標準在 1997 年被重新定義。因爲新的分子標靶藥物對轉移及某些腸胃道基質瘤具有療效，因此我們想知道目前台灣在新的定義之下，此腫瘤的發生率，以及研究其免疫染色的特性以及基因突變與預後的相互情形。由馬偕醫院 1998 年至 2004 年的 17850 個腸胃道腫瘤中篩選出 108 個符合新定義的腸胃道間質腫瘤進行統計及研究分析，並以 CD117、CD34、S-100 及 SMA，4 種抗體進行免疫染色，以及 KIT 及 PDGFRA 的基因分析，並且將其突變分類，比較不同形式的突變與沒有突變的腫瘤其預後情況是否有統計上的差別。統計所得台灣的腸胃道間質腫瘤的發生率爲約每百萬人有 13.74 人罹病，其盛行率爲每百萬人有 303.60 人。免疫染色研究之結果顯示 CD117 免疫染色研究之結果顯示 CD117 陰性的腸胃道間質腫瘤亦有基因突變的情形發生，而此類腫瘤其組織型態並無特別的。KIT 基因突變情形在 exon 9、11、13、17 分別爲 13/134 (9.7 %)、79/134 (59 %)、0/134 (0 %) and 0/134 (0 %)。PDGFRA 基因突變情形則只在 exon 12 發生 1/134 (0.07 %)，這樣的結果與目前大多數的文獻報告類似。而我們發現不同突變的型態也會有不同的預後情形； 此篇研究奠定重新定義之後，台灣腸胃道間質腫瘤的發生率，及其免疫化學染色之特性及 KIT 和 PDGFRA 基因在此腫瘤的情形。

英文摘要

Gastrointestinal stromal tumors (GIST) are the most common mesencymal tumors in gastrointestinal tract. The diagnostic criteria of GISTs had been redefined. Because of the new target therapy was successfully in unresectable or metastatic GIST patients, We examined the incidence, prevalence, immunohistochemical staining and mutation analysis in Taiwan. All patients (n=17850) who had primary mesenchymal tumors of the gastrointestinal tract in Mackay memorial hospital from 1998 to 2004 were selected to screen; and 108 patients were identified.The incidence and prevalence of GIST in Taiwan were analyzed statistically. Immunohistochemically, each tumor was staining for CD117,S-100, and alpha smooth muscle actin (SMA). KIT (exons 9，11，13and 17) and PDGFRA (exons 10,12,14 and 18) were sequenced to identify the mutation. The annual incidence of GIST in Taiwan was 13.74 per million. The prevalence of GIST in Taiwan was 303.60 per million. Immunophenotypically, tumors expressed CD117 (97/134),alpha-SMA

(8/134), desmin (1/134), S-100 protein (10/134). The KIT gene mutations of exon 9, 11, 13 and 17 were found in 13/134 (9.7%), 79/134 (59%), 0/134 (0%) and 0/134 (0%). The PDGFRA gene mutations only found in exon 12, 1/134 (0.7%). The incidence and prevalence of GIST in Taiwan was determined. The mutation rate of KIT was similar with other published reports but that of PDGFRA was lower than those. This article summarizes the incidence, molecular biology of GIST in Taiwan.