中文題目：頭頸部扁平細胞癌上皮細胞生長因子接受體突變之研究

英文題目：Identification of Epidermal Growth Factor Receptor Mutations in Patients with Head and Neck Squamous Cell Carcinoma

作 者：謝佳訓[1],張文震[1,6],謝佳娟[1,2],許濤[2],黃秀芬[3,4,6],廖俊達[5,6],王宏銘[1,6]

服務單位：[1]林口長庚醫院血液腫瘤科,[2]國立海洋大學生物科技研究所,[3]林口長庚醫院解剖病理科,[4]財團法人國家衛生研究院分子與基因醫學研究組,[5]林口長庚醫院耳鼻喉部,[6]長庚大學

Background: Overexpression of epidermal growth factor receptor (EGFR) exists in up to 90% of head and neck squamous cell carcinoma (HNSCC). The level of EGFR expression correlates with aggressive phenotype, increased resistance to treatment, and poor clinical outcome. In non-small-cell lung cancer, *EGFR* mutation is predictive for responsiveness to EGFR-tyrosine kinase inhibitors (TKIs). To identify the incidence of *EGFR* mutations in HNSCC, we analyzed 56 human HNSCC tissues in this study.

Methods: Archived formalin-fixed paraffin-embedded tissues of HNSCC were retrieved for *EGFR* mutational analysis. The kinase domain of *EGFR* coding sequence, exons 18, 19, 20, and 21 were amplified by nested PCR, and PCR amplicons were subjected to direct sequencing.

Results: Fifty-six HNSCC samples were analyzed, including 21 (37.5%) buccal cancers, 21 (37.5%) tongue cancers, 5 (8.93%) lip cancers, 4 (7.14%) gum cancers, 2 (3.57%) mouth floor cancer, 1 (1.79%) retromolar cancer, 1 (1.79%) hard palate cancer and 1 (1.79%) gingival cancer. No mutations within exon 18 and exon 19 were observed. One S784F (2.5%) mutation in exon 20 of the 40 cases was found. In exon 21, four *EGFR* mutations were identified, including 1 (2.17%) K852R, 1 (2.17%) L862Q, 1 (2.17%) M825T and 1 (2.17%) G857R of the 46 cases. In this study, a total of 5 mutations (8.93%) of the 56 cases were found.

Conclusion: The incidence of *EGFR* mutation in HNSCC is not rare, albeit it is low. Various mutations in a complex pattern were identified. Further correlation of these findings with clinical outcome is warranted in a large-scale study.