中文題目：*GATA3* 基因上三種新型突變引發家族性副甲狀腺功能不全合併耳聾

英文題目： Identification of three novel mutations in the *GATA3* gene responsible for familial hypoparathyroidism and deafness in the Chinese population

作　　者：邱偉益　陳煥文　陳惠文　李宗諺　蔡克嵩

服務單位：台大醫院檢驗醫學部及內科部

羅東博愛醫院內科部

前　　言：Familial hypoparathyroidism may be caused by mutations of several genes. The CaSR and GATA3 genes are the two candidates most commonly responsible for this condition.

材料及方法：We collected 5 unrelated families with familial hypoparathyroidism, and examined their CaSR and GATA3 genes. Blood samples from these 5 families and 50 ethnically matched unrelated controls were acquired. Biochemistry screening and formal audiogram were performed to evaluate the affected individuals. All the exons and exon-intron boundaries of the GATA3 and CaSR genes were sequenced.

結　　果：We identified three novel mutations in the GATA3 gene responsible for familial hypoparathyroidism and deafness. (1) A frameshift deletion occurring in codon 160 (478delG) was hypothesized to disrupt dual zinc fingers as well as one transactivating domain. (2) A donor splice site mutation at exon 4/intron 4 boundary (IVS4+2 T to GCTTACTTCCC) was predicted to lead to truncated GATA3 proteins that lack both N-terminal and C-terminal zinc-containing fingers. (3) A missense mutation R353S was predicted to disrupt the helical turn and thus changed the angle between the C-terminal zinc finger and the adjacent C-terminal tail. Except for a previously described polymorphism, G990R, we did not find any genetic variants in CaSR gene

結　　論：This is the first article presenting mutations of the GATA3 gene responsible for familial hypoparathyroidism and deafness in the Chinese population. Our results expand the spectrum of mutations and report the first splice donor site mutation of GATA3 gene. In contrast, we do not find causal sequence variants of the CaSR gene from our collection of familial hypoparathyroidism