

中文題目：特定基因rs855791對生育期婦女罹患缺鐵性貧血的影響

英文題目：**TMPRSS6 rs855791 Polymorphism Influences the Susceptibility to Iron Deficiency Anemia in Women at Reproductive**

### **Age**

作者：裴松南<sup>1,2,3</sup> 馬銘君<sup>1,2</sup> 尤慧玲<sup>4</sup> 傅宏鈞<sup>2,3,5</sup> 郭景元<sup>1,2</sup> 饒坤銘<sup>1,2</sup> 王銘崇<sup>1,2</sup> 李建德<sup>2,6</sup>

服務單位：高雄長庚醫院內科部血液腫瘤科<sup>1</sup> 長庚大學<sup>2</sup> 長庚大學臨床醫學研究所<sup>3</sup> 高雄長庚醫院檢驗醫學科<sup>4</sup> 高雄長庚醫院婦產部<sup>5</sup> 高雄長庚醫院內科部腎臟科<sup>6</sup>

### **Abstract**

**Background:** Genome-wide-association studies have identified the *TMPRSS6* polymorphism rs855791 has the strongest association with red blood cell indices or iron parameters in general population. Whether this genetic variant influences the susceptibility of iron deficiency anemia (IDA) in women with menstruation has not been well studied.

**Methods:** In this case-control study, we enrolled 67 women with IDA and 107 healthy volunteers, and analyzed their complete blood counts, rs855791 genotypes, and menstrual amounts. Menstrual blood loss was evaluated with a pictorial blood-loss assessment chart.

**Results:** There were significantly fewer rs855791 *C* homozygotes in the IDA group

than in the healthy group (11.9% vs. 25.2%,  $p = 0.03$ ). The odds ratio (OR) of *C* homozygotes having IDA versus non-*CC* subjects having IDA was 0.4 (95% CI, 0.17 – 0.95,  $p = 0.04$ ). When the analysis was confined to study subjects with menorrhagia, this difference became more prominent (9.6% vs. 28.6%,  $p = 0.01$ ; OR, 0.27, 95% CI, 0.09 – 0.77,  $p = 0.01$ ). For women with non-*CC* genotypes, there was an inverse correlation between hemoglobin levels and menstrual loss ( $p < 0.001$ ); however, this association was not found for those with genotypes *CC* ( $p = 0.15$ ).

**Conclusions:** Our study suggests homozygosity for *TMPRSS6* rs855791 *C* genotype has a protective role against IDA in women at reproductive age, especially in those with menorrhagia.

Key words: iron deficiency anemia, menorrhagia, single nucleotide polymorphism, rs855791, transmembrane protease serine 6 (*TMPRSS6*).