CHAPTER VI

CONCLUSIONS

This study has identified the novel heterozygous missense c.721C>T (p.P241S) mutation in *MSX1* in a Thai patient affected with the following conditions: bilateral cleft lip and palate, hypodontia of left and right maxillary permanent lateral incisors, hypodontia of the left maxillary permanent second premolar, hypodontia of the right mandibular permanent third molar, and preaxial-polydactyly of the left thumb. This is the first such report in a human, indicating that *MSX1* mutation may be associated with orofacial clefts, hypodontia and limb anomalies in the one individual. Moreover, the novel heterozygous missense c.589G>A (p.A197T) mutation was also found in *MSX1* in another Thai patient affected with unilateral right cleft lip and palate and hypodontia of the right maxillary permanent lateral incisor. This study suggests that p.P241S and p.A197T mutations may be associated with syndromic orofacial clefts in the Thai population.