V. CONCLUSION

In this study, the newly modified method for detection of Hb Constant Spring gene was introduced. The method was a double steps detection system, first was the screening for abnormal α_2-termination codon and then followed with the characterization of α^{CS} gene. The core principle of α^{CS} gene detection was the introduction of a restriction site to the α^{CS} semi-nested PCR product, after the sample had been screened for abnormal α_2-termination codon. The RFLP study of this semi-nested PCR product told whether the α_2-globin template possessed the α^{CS} gene or not.

The method was used to screen for the Hb CS gene in northern Thailand populations. The prevalent of Hb CS gene from this study is 2.62%, with the gene frequency of 0.013.

Study in nondeletional Hb H disease patients found that Hb CS gene was account for most of the samples. Although the gene frequency of the Hb CS in population is much lesser than α-thalassemia 2, The proportion of Hb CS-Hb H disease patients who came to treat at the hospital was greater than of the deletional type. This suggested the more severity of the Hb CS gene in the disease. Thus, the genetic counseling and prenatal diagnosis is significant health care issue for the individuals at risk. The method presented here was found to be convenient, reliable, and suitable for population screening or routine diagnosis.