

INTRODUCTION

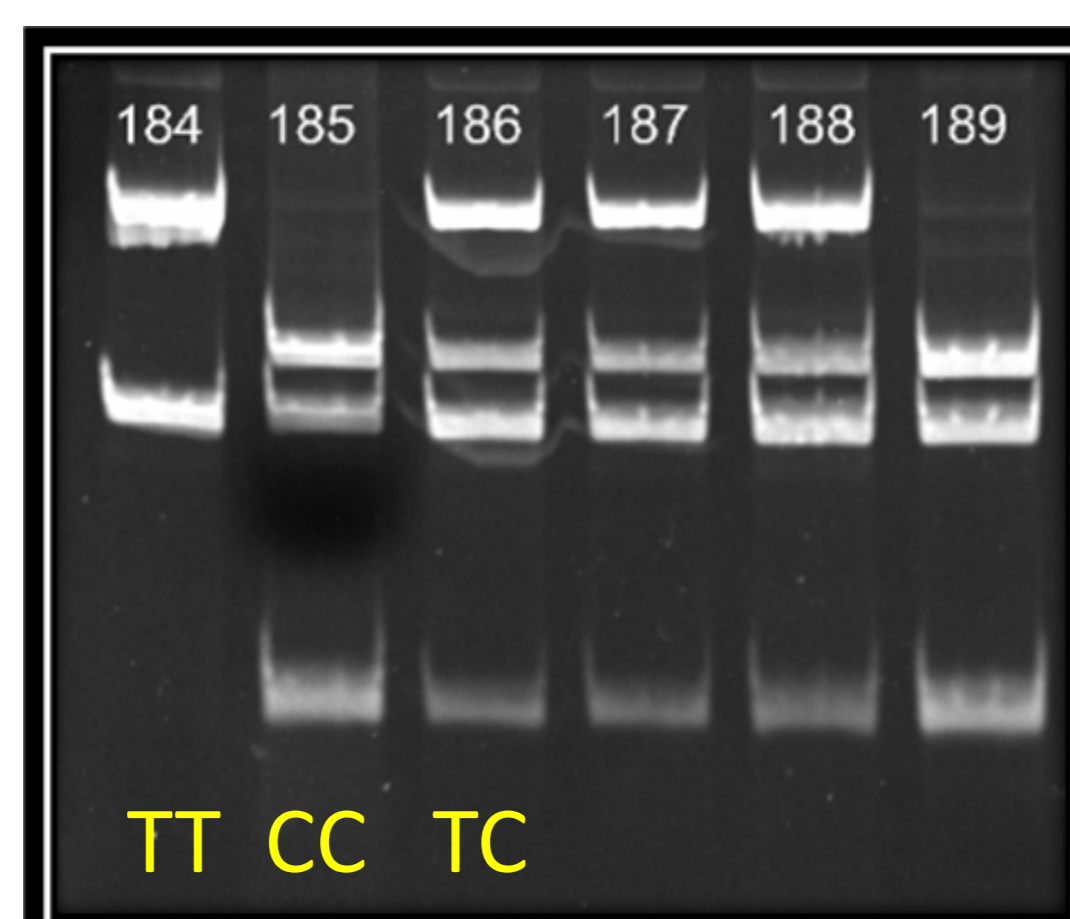
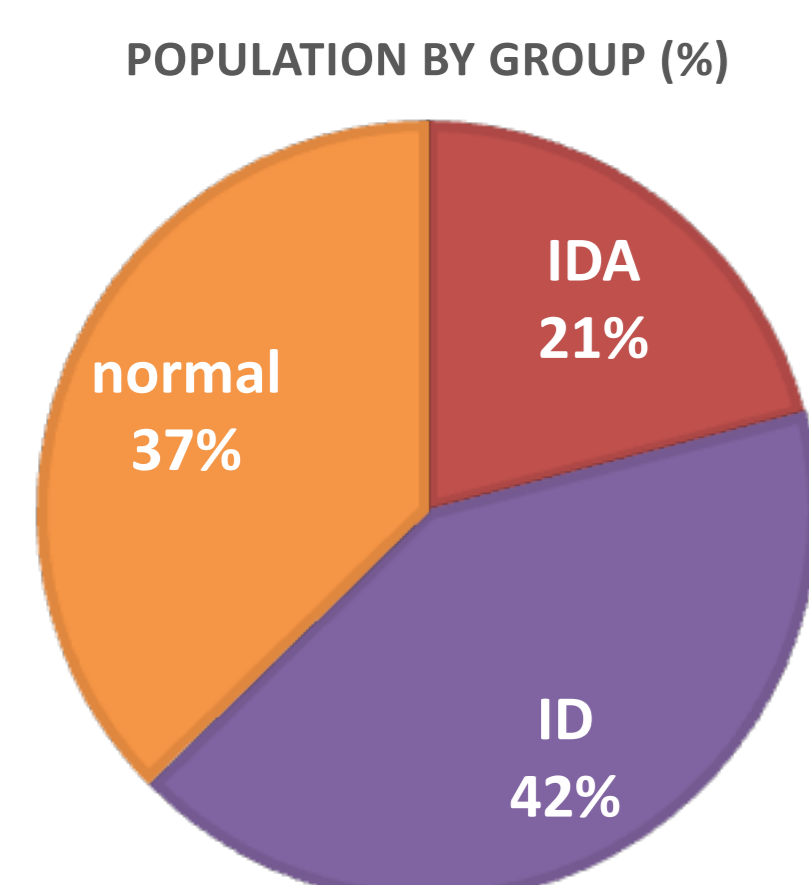
In Thailand, the prevalence of iron deficiency anemia (IDA) in children is around 10-16%. Common etiologies of IDA include inadequate iron intake and chronic blood loss. Other than those, mutations in *TMPRSS6* gene have been reported. The *TMPRSS6* gene regulates hepcidin expression by encoding Matriptase-2, an enzyme which cleaves a co-factor promoting hepcidin transcription. With mutations to such gene, the patient has an elevated hepcidin level despite the iron deficiency status and is described as having iron refractory IDA (IRIDA). Several studies showed that the *TMPRSS6* gene polymorphisms, for example rs855791 (p.V736A) and rs2235324 (p.K253E), associate with lower iron status. Whether these single nucleotide polymorphisms are associated with an increased risk of developing IDA in our population, especially children, is an enticing question.

OBJECTIVE(S)

- To identify the prevalence of the *TMPRSS6* gene polymorphism (p.V736A) in the study population
- To identify the association between the *TMPRSS6* polymorphism (p.V736A) and the risk of IDA in Thai children

METHOD(S)

- A total of 415 children, aged 9-15 years, were recruited
- They were classified into 3 groups
 - Normal iron status – normal Hb, ferritin ≥ 30 ng/L
 - Iron depletion (ID) – normal Hb, ferritin < 30 ng/L
 - Iron deficiency anaemia (IDA) – low Hb, ferritin < 30 ng/L or $T_{sat} < 16\%$
- DNA was extracted and PCR was performed for all participants
- RFLP was used to identify the genotype as TT (wild type), CT and CC
- Chi-square test was used to determine the difference in iron status with respect to genotypes
- Logistic regression analysis was used to determine the association between the genotype and serum iron, ferritin, as well as hepcidin levels.



RESULT(S)

- The T allele frequency in this population was 0.6.
- The TT genotype was highest in IDA group (48%) compared to ID (36.6%) and normal group (31.9%), but no significant difference was found, p -value = 0.226
- Compared to the CT genotype, the TT genotype had significantly lower serum iron with a mean difference of -7.9 [Standard Error, S.E. 3.2] mmol/L, p -value = 0.043 while the difference was not significant when compared to the CC genotype (mean difference -7.4 mmol/L [S.E. 4.8] mmol/L, p -value = 0.354)
- No significant difference in hepcidin level was demonstrated in different genotypes.

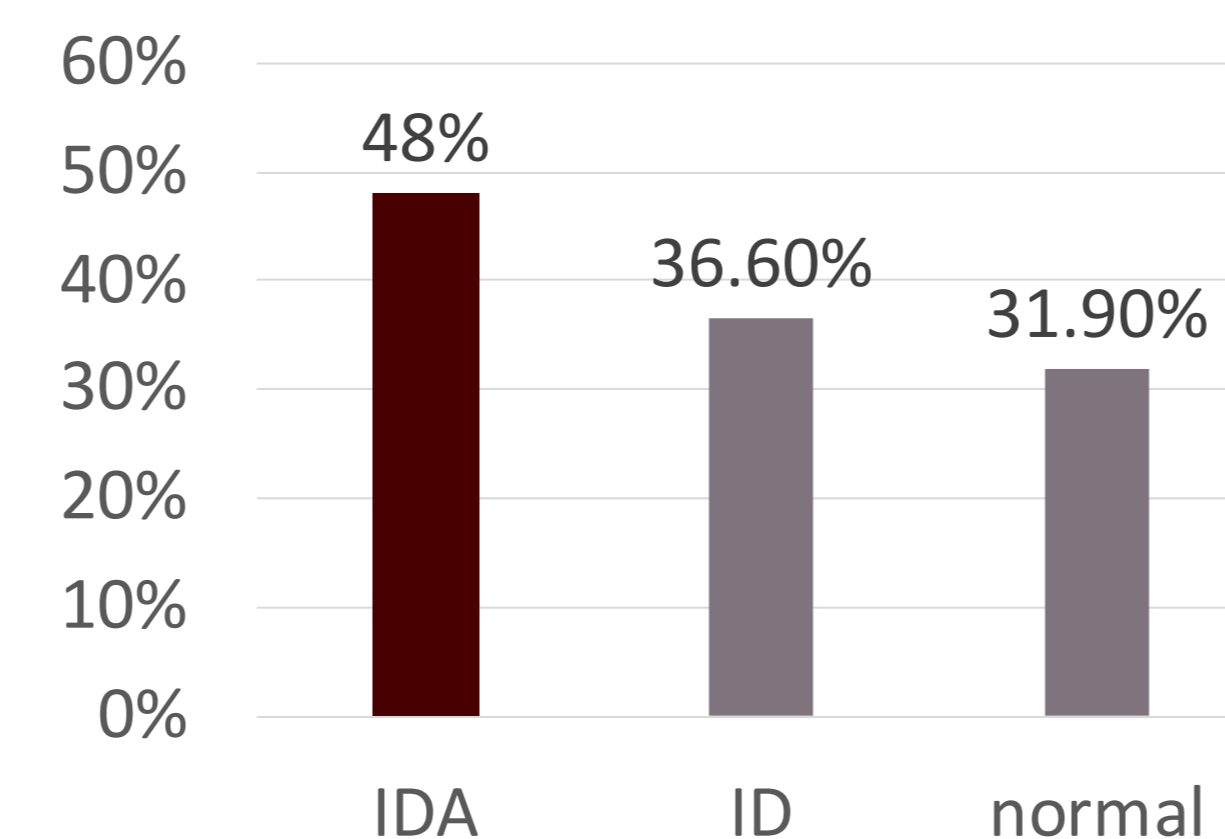


Figure 1. Demonstrate the percentage of TT in normal, ID and IDA

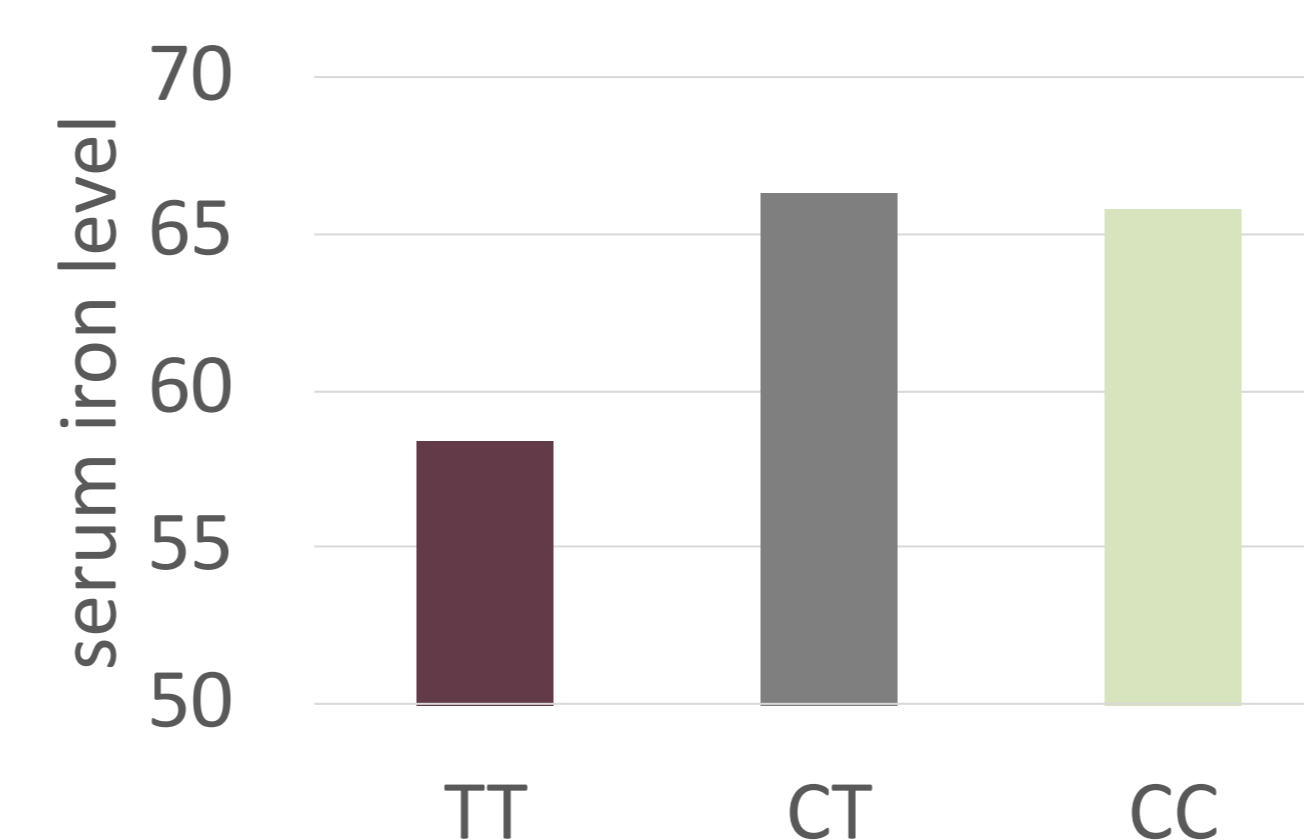


Figure 2. Demonstrate serum iron in TT, CT and CC genotypes

CONCLUSION(S)

The TT polymorphism of c.T2321C is common among Thai children with IDA and associates with a lower level of serum iron when compared to the other genotypes. Other polymorphisms of the *TMPRSS6* gene will be further studied to determine additional effects of combined polymorphisms to the iron status.





ACKNOWLEDGEMENTS

- Ramathibodi Hospital
- Thai Medical Association Fund

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