PREVALENCE OF HISTONE FAMILY E 1 (HFE) MUTANT ALLELES IN A TAMILIAN COHORT FROM VELLORE, SOUTH INDIA

Sir,

Hereditary hemochromatosis (HH) is considered to be a disease of the Caucasian population.\(^1\) C282Y mutation is associated with most of the HH manifesting individuals in the European population. H63D causes predisposition to iron overload only when associated with C282Y. In the European population, about 10% carry the C282Y mutation while more than 20% carry the H63D mutation. S65C mutation is another mutation causing mild HH. The prevalence of S65C is much lower in the European population as compared to the first two mutations.\(^2\) So far, all studies from India have failed to associate HFE mutations with HH. The C282Y mutation is reported very low in the north and absent in the south Indians.\(^3,4\) H63D mutation is more frequently distributed in the Indian subcontinent. Dominant forms of HH caused by other iron overload genes have been implicated for Asian Indians.\(^5\) We report here the prevalence of HFE mutations in the local Tamilian population from Vellore.

Cord blood samples from 50 neonates born to Tamilian parents in the Obstetric department of Christian Medical College, Vellore, were collected after approval of the Institute’s Research Committee and consent from the families. Qiagen mini kits were used for DNA extraction. The samples were tested for HFE gene mutations C282Y, H63D, and
Polymerase chain reaction (PCR) was carried out using specific primers for the HFE gene, and the PCR products were digested using restriction digestion with specific enzymes, namely; SnaB1 for C282Y, MboI for H63D, and HinfI for S65C, using standard protocols. The products were run on 3% agarose gels to detect the presence or absence of mutations along with positive and negative controls. Allele frequency was carried out using Hardy-Weinberg equations. In the 50 neonatal samples, there was a pair of twins. Of the 49 families, consanguinity was present in 13 families giving an overall consanguinity rate of 26.5%. Of the samples tested for C282Y and S65C, all showed wild genotype, five samples were heterozygous, and two were homozygous for the H63D mutation. Of the twins’ sample, one was a homozygote and the other a heterozygote. The overall frequency for the H63D mutant allele was 0.09 and that of wild allele was 0.91. Our study showed the absence of C282Y and S65C in this population and a frequency of 9% of H63D mutation.

The present study showed a low prevalence of HFE mutations in the local Tamilian population. The study supports the very low prevalence of HFE-related hemochromatosis in the Indian population. This study looks at the S65C mutation prevalence in the Tamilian population so far not studied in the Indian subcontinent. This prevalence study would have future implications in determining genetic factors of iron metabolism that might have a role in iron deficiency, a common problem in Tamil Nadu.

ACKNOWLEDGMENT

The authors would like to thank the Fluid Research Committee, Department of Obstetric and Gynecology, Christian Medical College, Vellore.

SUMITA DANDA, C. E. EAPEN, K. A. KURUVILLA, S. SANTHOSH, G. KURIAN, G. M. CHANDY

Departments of Gastrointestinal Sciences and Neonatology, Christian Medical College, Vellore, Tamil Nadu, India

Address for correspondence:
Dr. Sumita Danda,
Department of Gastrointestinal Sciences,
Christian Medical College, Dr. Ida Scudder Road,
Vellore – 632 004, Tamil Nadu, India.
E-mail: sdanda@cmcvellore.ac.in

REFERENCES


Table 1: Detection of H63D mutation in 50 Tamilian neonatal samples

<table>
<thead>
<tr>
<th>Number</th>
<th>wild (+/+</th>
<th>H63D heterozygote (+/-)</th>
<th>H63D homozygote (-/-)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Total</td>
<td>43</td>
<td>5</td>
<td>2</td>
</tr>
<tr>
<td>Allele</td>
<td>Wild allele</td>
<td>Mutant allele</td>
<td></td>
</tr>
<tr>
<td>frequency</td>
<td>0.91</td>
<td>0.09</td>
<td></td>
</tr>
</tbody>
</table>

Indian Journal of Medical Sciences, Vol. 64, No. 5, May 2010