Carrier detection of α thalassaemia: An emerging role of HPLC


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Cord blood; HPLC; Beta thalassemia.

ABSTRACT
Beta thalassemia is one of the commonest recessive genetic diseases. It results in a severe anemia needing monthly transfusion. Our aim was to evaluate hematological profile of cord blood HPLC for prenatal diagnosis of beta thalassemia. Cord blood samples at 18-24 weeks gestation from 113 mothers suspected for beta thalassemia was studied. Out of them 34 was maternal contamination. HbF & HbA0 were determined by cation exchange high performance liquid chromatography. In HPLC studies for beta thalassemia, 13 fetuses were affected, 32 were carriers and 34 were normal.

INTRODUCTION
Thalassaemia is a heterogeneous group of autosomal recessive disorders due to mutations causing decreased or absent production of the polypeptide globin chains. The distribution of beta thalassemia gene is not uniform in the Indian subcontinent. The highest frequency of beta thalassemia trait is reported in Gujarat, followed by Sindh, Punjab, Tamil Nadu, South India and Maharashtra (1.9%) so prenatal diagnosis of beta thalassemia is very useful for prevention of affected child birth[1]. The introduction of a new simple HPLC method allowing precise quantification of the various Hb fractions prompted us to evaluate its reliability in prenatal diagnosis of thalassemia and hemoglobinopathies[2]. This method, which utilizes a commercial column designed for hemoglobin fractions measurement, allows much more rapid and precise evaluation of fetal blood samples in the mid trimester of pregnancy[3].

MATERIALS AND METHOD
Total 113 cord blood sample were collected for the study. They were come from Obstetric & Gynecology OPD for screening of beta thalassemia in hematology department AIIMS. Blood samples were collected in 2 ml vacutainer containing EDTA as an anticoagulant. Complete blood count and red cell indices were measured by automated Analyzer (SYSMEX K-4500, Kobe Japan). Quantitative assessment of hemoglobin, HbF, HbA0 were performed by HPLC (Bio-Rad-VariantTM Bio Rad, CA, USA)
RESULT AND DISCUSSION

Total 113 cord blood were included in the study out of them 34 were maternal contaminated while 34 (mean age 27.3±3.9) were normal, 32 (mean age 26.7±3.7) were carrier and 13 (mean age 27.7±3) were affected. Hematological features are summarized in TABLE 1. HbF were 100% in affected cord blood and HbA0 level were <8; however mean Hb (11.5±0.9) and red cell indices were elevated. Beta thalassemia is common in Asian Indians with carrier frequency of 3-7% (8). Many thalassemia major children are born every year and require regular transfusions to sustain life

<table>
<thead>
<tr>
<th>Type</th>
<th>HbF</th>
<th>HbA0</th>
<th>WBC</th>
<th>RBC</th>
<th>HGB</th>
<th>HCT</th>
<th>MCV</th>
<th>MCHC PLT</th>
</tr>
</thead>
<tbody>
<tr>
<td>Affected</td>
<td>100±2.2</td>
<td>0.5±0.7</td>
<td>15.71±19.10</td>
<td>2.7±0.2</td>
<td>11.5±0.9</td>
<td>35.5±2.3</td>
<td>127.4±7.9</td>
<td>41.5±2.3</td>
</tr>
<tr>
<td>Carrier</td>
<td>96.9±3.6</td>
<td>2±0.4</td>
<td>12.46±11.32</td>
<td>2.7±0.3</td>
<td>11.37±1.27</td>
<td>35.6±3.9</td>
<td>130.9±8.9</td>
<td>41.62±3.33</td>
</tr>
<tr>
<td>Normal</td>
<td>94.3±2.8</td>
<td>5±1.7</td>
<td>15.3±19.7</td>
<td>2.5±0.6</td>
<td>10.81±2.60</td>
<td>33.9±7.6</td>
<td>131.5±10.4</td>
<td>42.14±3.70</td>
</tr>
</tbody>
</table>

REFERENCES

