BRIEF NOTE

Hemoglobin J in an American Caucasian Family of Swedish Ancestry

By PRAWASE WASI, JOHN GITHENS AND WILLIAM HATHAWAY

HEMOGLOBIN J was first reported in an American Negro family by Thorup et al.\(^1\) in 1956. Subsequently, it has been found in Indonesians,\(^2,3\) East Indians,\(^4,5\) French-Canadians,\(^6\) Chinese,\(^7\) a Negro-Caucasian,\(^8\) and most recently in a family of Hawaiian-Chinese-Caucasian ancestry.\(^9\) The hemoglobin “Liberian 1” described by Robinson et al.\(^10\) was formerly designated as hemoglobin J but recently has been found to behave differently than J both in electrophoresis and in resin chromatography, and it is now known as hemoglobin N.\(^11\) The hemoglobin originally reported by Cabannes et al.\(^12\) as J was subsequently changed to K.\(^13\) The purpose of this present communication is to report the hemoglobin J trait in an American Caucasian family of Swedish ancestry.

Electrophoretically, at pH 8.6, hemoglobin J moves faster than hemoglobins A and K,\(^13\) but more slowly than hemoglobins H, I, or N.\(^10,11\) At pH 6.5, it moves toward the cathode between hemoglobins A and I. In resin chromatography, hemoglobin J does not separate from hemoglobin A.\(^2\)

METHODS

Hematological examinations were carried out by the currently accepted methods. Alkali resistant hemoglobin was determined according to the method of Singer et al.\(^14\) Quantitative osmotic fragility of red blood cells was performed by the method of Suess et al.\(^15\) Electrophoretic studies of hemoglobin solutions were carried out by flat plate paper electrophoresis described by Smith and Conley\(^16\) using veronal buffer pH 8.6 and phosphate buffer pH 6.5 by starch block electrophoresis with the method of Kunkel\(^17\) as modified by Gerald and Diamond,\(^18\) and by agar electrophoresis modified from the method described by Robinson et al.\(^19\)

The blood containing the fast hemoglobin was one of a few samples that had been drawn from Caucasians among our laboratory personnel for use as normal controls.

CASE REPORT

P. F., the propositus, is a 30 year old white male of Swedish-German ancestry who had been in good health all his life. Physical examination did not reveal any relevant findings. Liver and spleen were not palpable. His hemoglobin solution was subjected to paper electrophoretic study and was found to have a fast-moving component in addition to hemoglobin A at pH 8.6. The same finding was obtained from electrophoresis on starch block (fig. 1).

Blood studies including the hemoglobin, RBC, hematocrit, cell indices, and reticulocyte count, were normal (table 1). Peripheral blood smear and red cell fragility were within normal range. The van den Bergh test showed a conjugated bilirubin of 0.2 mg. per cent,

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and an unconjugated bilirubin of 0.3 mg. per cent. The amount of hemoglobin A₂ was found to be normal at 2.7 per cent as was the fetal hemoglobin of 0.1 per cent.

Further electrophoretic study of the patient's hemoglobin revealed that at pH 6.5 the abnormal component moved toward the cathode more slowly than hemoglobin A. Hemoglobin H could thus be ruled out since at acid pH hemoglobin H moves toward the anode. The red blood cells did not have the inclusion bodies of hemoglobin H when stained with brilliant cresyl blue. Our hemoglobin was then compared on starch block electrophoresis at both acid and alkaline pH with a known sample of hemoglobin H, with an intermediate "fast" hemoglobin, and with a sample of hemoglobin J from one of Thorup's original patients. The mobility of our hemoglobin at pH 8.6 appeared to be identical with that of hemoglobin J (fig. 2). At pH 7.0 the mobility on starch block also was identical with that of the hemoglobin J from Thorup, with both samples showing good separation. The percentage of the fast component was found to be 52 per cent.

The migration of our hemoglobin at a rate more rapid than that of the intermediate "fast" hemoglobin from a patient with thalassemia-hemoglobin H disease (fig. 2) suggests that it is not the same as the hemoglobin of Fessas and Papaspyrou or that found in Thai babies by Tuchinda et al. Hemoglobin K is also ruled out since it should be slower than any of these.

Resin chromatography in amberlite 1CR 50 by the method of Huisman and Prins showed no separation of this fast component from A. This is characteristic of hemoglobin

*This study was performed for us by Miss Virginia Minnich in St. Louis, Mo.
J while hemoglobin N should separate. These findings would appear to confirm this fast hemoglobin as J.

Four members of the immediate family were studied (table 1). All of the hematologic findings were essentially within normal limits except for the presence of hemoglobin J in P. F., the propositus, and his father. The father is of Swedish extraction. The mother, who has German ancestors, was normal as was the sister of the propositus.

**DISCUSSION**

The patient and his father herein reported are believed to be heterozygous carriers of hemoglobin J. Their abnormal hemoglobin showed a mobility that appeared to be identical with the hemoglobin J of Thorup's original patient. However, discrepancies in the electrophoretic behavior of hemoglobin J at pH 6.5 have been noted in the literature and raise the question of whether

**Table 1.—Hematologic Findings in the F. Family**

<table>
<thead>
<tr>
<th>Subjects</th>
<th>Hemoglobin in Gm.%</th>
<th>RBC in million</th>
<th>Hct. %</th>
<th>MCV</th>
<th>MCHC %</th>
<th>Retics %</th>
<th>Hemoglobin types</th>
<th>Fetal Hb. %</th>
</tr>
</thead>
<tbody>
<tr>
<td>Father</td>
<td>16.4</td>
<td>5.65</td>
<td>49</td>
<td>86.7</td>
<td>33.4</td>
<td>0.6</td>
<td>A + J</td>
<td>0.15</td>
</tr>
<tr>
<td>Mother</td>
<td>13.6</td>
<td>4.61</td>
<td>44</td>
<td>95.5</td>
<td>31</td>
<td>0</td>
<td>A</td>
<td>0.3</td>
</tr>
<tr>
<td>Propositus</td>
<td>16.0</td>
<td>5.14</td>
<td>46</td>
<td>89.3</td>
<td>34.8</td>
<td>0.4</td>
<td>A + J</td>
<td>0.1</td>
</tr>
<tr>
<td>Sister</td>
<td>12.9</td>
<td>4.64</td>
<td>41</td>
<td>88.4</td>
<td>31.4</td>
<td>1.2</td>
<td>A</td>
<td>0.4</td>
</tr>
</tbody>
</table>

RBC = red blood cells; HCT = hematocrit; MCV = mean corpuscular volume in cubic microns; MCHC = mean corpuscular hemoglobin concentration in per cent; Retics = reticulocytes.
this is due to variations in the technic employed or actual differences in the hemoglobins.

In the original reports\textsuperscript{1,2} hemoglobin J was found to separate from A at pH 6.5 in moving boundary electrophoresis, and more recently\textsuperscript{9} it was shown to separate from A on paper electrophoresis at this pH. On the other hand, failure of J to separate from A at pH 6.5 on paper has been reported several times.\textsuperscript{4,5,7,11} Details of the electrophoretic behavior are not given in the remaining cases in the literature.\textsuperscript{3,4,8}

Our hemoglobin J separated from A at pH 6.5 on paper, and on starch block electrophoresis at pH 7.0 it separated at a rate similar to that of Thorup’s patient. It was also studied with electrophoresis on agar gel. At pH 8.6 the hemoglobin J separated readily as a fast component similar to the findings on paper and starch. At pH 6.5 it moved more slowly than hemoglobin A but faster than hemoglobin S.

It appears that the genetic defect for hemoglobin J although its incidence is low, is widely distributed throughout the world among people of very different racial origins.

\textbf{Summary}

Two instances of the heterozygous state of hemoglobin J are described in the male members of an American Caucasian family of Swedish ancestry.

\textbf{Summario in Interlingua}

Es describite duo exemplos del stato heterozygotico de hemoglobina J in membros mascule de un familia american de racia caucasian con ancestres svede.

\textbf{Acknowledgments}

We wish to acknowledge with grateful appreciation the assistance of Miss Virginia Minnich at the Washington University Medical School for comparing our hemoglobin with a sample of hemoglobin J from Thorup’s patient. Figure 2 is published with her kind permission.

\textbf{References}

BRIEF NOTE: HEMOGLOBIN J IN AN AMERICAN FAMILY


In heterozygous thalassemia, the value of Hb.A2 as separated on paper electrophoresis was found to be higher than normal. In homozygous thalassemia, the A2 value was distinctly elevated when considered as a percentage of total Hb.A, excluding Hb.F.—J. B. C.
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