The Pelger-Huët Familial Anomaly of Leukocytes

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In 1928, Pelger described an anomaly of granulocytic leukocytes manifested by a lack of normal segmentation of the nucleus. Although the nuclear chromatin was of the mature type, cells with more than two lobes to the nucleus were few and the majority of cells were unsegmented stab forms. Huët, in 1932, disclosed the hereditary nature of the anomaly and subsequent investigators confirmed and described in more detail the genetic features of this disorder.

In 1939, the Pelger anomaly was described in rabbits, and Nachsheim, in 1943, established the dominant character of the disorder by selective breeding in rabbits. Leitner has pointed out that Nachsheim’s work demonstrates that the Pelger gene is pathologic and that in homozygous form in man, a serious hemopathy may result. A recent communication of Begemann and Campagne describes the first case of a possible homozygous human, in which the nuclear anomaly closely resembles that seen in rabbits homozygous for the condition; the condition described is not lethal, the individual having it being 21½ years old.

While there have been comparatively few reports in the literature on Pelger’s anomaly, the disorder is probably more common than realized. The present report concerns the discovery of a case in a Japanese woman and the subsequent investigation of 95 individuals in four generations of her family with the detection of 25 additional cases of Pelger’s anomaly.

Case Report and Results of Investigation of the Family

Case Report

XY, a 50 year old Japanese female, resident of Kure, Hiroshima Prefecture, Japan, was seen as one of a group of controls during an investigation into the late radiation effects of the atomic bomb on hematopoiesis. This patient had no complaints; her past history, family history and system review were noncontributory and the physical examination was within normal limits. Laboratory data is given in table 1.

Family Investigation

The discovery of this peculiar lack of segmentation of the neutrophils prompted an investigation of other members of the family. In all, there were 94 blood relatives in four generations and 25 living spouses. Of these, 74 additional parents...
and siblings and 5 spouses were available for study, and histories, physical examinations and blood studies were carried out in each instance. As shown in figure 1 a total of 26 cases (including the propositus) of Pelger's anomaly were discovered in this group. The occurrence of this anomaly was uninfluenced by age or sex; infection and consanguinuity played no role in the occurrence of this disorder.

Fig. 1.—Inheritance of Pelger-Huët anomaly
A feature of the peripheral blood smears was the presence of eosinophilia and increased monocytes in individuals with and without Pelger's anomaly, findings due, apparently, to the high degree of parasitism among the Japanese population.

<table>
<thead>
<tr>
<th>Blood studies</th>
<th>Differential %</th>
<th>Bone marrow %</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hgb. 13.5 Gm.</td>
<td>Neutrophils 4</td>
<td>Neutrophils 5</td>
</tr>
<tr>
<td>RBC 4.55</td>
<td>seg. 40.5</td>
<td>band 11.5</td>
</tr>
<tr>
<td>Ht. 44%</td>
<td>band 4</td>
<td>Eosinophils 16</td>
</tr>
<tr>
<td>MCV 90.5 cu.m</td>
<td>Basophils 1</td>
<td>Metamyelocyte 16</td>
</tr>
<tr>
<td>MCH 29.8 µg.</td>
<td>Lymphocytes 46.5</td>
<td>Myelocyte 39.5</td>
</tr>
<tr>
<td>MCHC 32.9%</td>
<td>Monocytes 2.5</td>
<td>Promyelocyte 6.5</td>
</tr>
<tr>
<td>WBC 8100</td>
<td>Plasma cells 0.5</td>
<td>Lymphocyte 11.5</td>
</tr>
</tbody>
</table>

* Red cells and platelets normal in appearance. Red cell series and megakaryocytes within normal limits.

The presence of eosinophilia allowed a unique opportunity to confirm the observations of others that the eosinophil takes part in the nuclear anomaly as well as the neutrophil. The striking lack of granulocytic lobulation is demonstrated in figure 2 which
summarizes the findings in the 75 blood relatives examined in this series. The characteristic, poorly segmented cells of the Pelger-Huët anomaly are shown in figure 3. Bone marrow studies were carried out only in the propositus, the findings in this case being similar to those described by Leitner,2 namely an increase in myelocytes, metamyelocytes and band forms with a marked lack of segmentation. No other nuclear abnormalities were observed.

From the genetic standpoint, in the various sibships tested, where either a parent or a sib had the anomaly, a total of 46 individuals were investigated and of these, 26 showed the anomaly. This fits excellently with the expected 1:1 ratio for simple Mendelian inheritance where the heterozygote shows the condition.

**Comments and Conclusions**

In this paper, the fourth case of the Pelger-Huët anomaly occurring in Japan has been reported. Investigation of 80 parents and siblings uncovered 26 cases of the nuclear anomaly in this family. The studies on these individuals confirm the hematologic findings described by Pelger, Leitner and others, and the genetic data support Huët’s concept of the simple Mendelian inheritance.
REFERENCES

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