A note on the inheritance of the hereditary persistence of fetal haemoglobin and the δ-chain variant Hb-\(\text{A}_2^{\prime}\)

The most common form of the condition known as the hereditary persistence of fetal haemoglobin (HPFH) is characterized by a continued synthesis of Hb-F and an absence of β- and δ-chain synthesis by the chromosome carrying the HPFH determinant. This conclusion is based on observations made in homozygotes who produce Hb-F only; in subjects heterozygous for the β-chain variant Hb-S and for the HPFH condition who produce Hb-S, Hb-F, a decreased amount of Hb-\(\text{A}_2\) and no Hb-\(\text{A}_2^{\prime}\); and in one subject who is heterozygous for the HPFH condition and the δ-chain variant Hb-\(\text{A}_2^{\prime}\), producing Hb-F, Hb-\(\text{A}_2\), Hb-\(\text{A}_2^{\prime}\), and no Hb-\(\text{A}_2^{\prime}\) (Huisman et al, 1971; Huisman, 1972). Recently we were able to study a large family in which the HPFH anomaly and the Hb-\(\text{A}_2^{\prime}\) variant occurred separately and in combination with each other, thus allowing a further evaluation of the inheritance of these two conditions.

The family consisted of the parents, three sons, and five daughters. Blood samples were analysed by routine haematological procedures using a Coulter Counter Model S. Haemoglobin in red cell haemolysates was analysed by starch gel electrophoresis at pH 9.0 (Efremov et al, 1969), by an alkali denaturation procedure to determine the percent Hb-F (% \(\text{F}_{\text{AD}}\) Betke et al, 1959)), and by DEAE-Sephadex chromatography to quanitate the levels of Hb-\(\text{A}_2\) and Hb-\(\text{A}_2^{\prime}\) (Huisman and Dozy, 1965; Dozy et al, 1968). The data are given in the Table. The father appears to have a HPFH heterozygosity and the mother a heterozygosity for the δ-chain variant Hb-\(\text{A}_2^{\prime}\). Three of their children are normal, one has the Hb-\(\text{A}_2^{\prime}\) heterozygosity, two have the HPFH heterozygosity, and two have the HPFH heterozygosity as well as the Hb-\(\text{A}_2^{\prime}\) heterozygosity. The level of Hb-\(\text{A}_2\) in the three normal family members (2.4, 2.7, and 2.7%) is comparable to that of the sum of Hb-\(\text{A}_2\) and Hb-\(\text{A}_2^{\prime}\) in the two Hb-\(\text{A}_2^{\prime}\) heterozygotes (2.4 and 3.0%) and is distinctly more than that of Hb-\(\text{A}_2\) in the HPFH heterozygotes (1.6, 1.7, and 2.2%) and of Hb-\(\text{A}_2^{\prime}\) in the two persons with the Hb-\(\text{A}_2^{\prime}\) HPFH condition (1.8 and 2.0%) who do not produce any Hb-\(\text{A}_2^{\prime}\).

The complete absence of Hb-\(\text{A}_2\) in the two individuals with the Hb-\(\text{A}_2^{\prime}\)–HPFH combination confirms previously published, rather incomplete, data (Huisman et al, 1971) and offers additional evidence that δ-chain production is absent on the chromosome carrying this HPFH determinant.

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**Table**

<table>
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<th></th>
<th>Age</th>
<th>Hb (g/dl)</th>
<th>PCV (%/100)</th>
<th>RBC (×10³/l)</th>
<th>(\text{F}_{\text{AD}}) (%)</th>
<th>(\text{A}_2) (%)</th>
<th>(\text{A}_2^{\prime}) (%)</th>
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<td>58</td>
<td>12.7</td>
<td>38</td>
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<td>4.03</td>
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<td>13.5</td>
<td>40</td>
<td>4.79</td>
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<td>37</td>
<td>4.05</td>
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Short communication

REFERENCES


Announcements

Data on cytogenetic registers

The International Advisory Committee on Cytogenetic Registers to the Standing Committee on Standardization in Human Cytogenetics was charged with the responsibility of compiling, updating, and publishing a list of cytogenetic registers, together with summaries of aims, objectives, and key elements. The committee has developed a form for the collection of such data and invites individuals who operate Cytogenetic Registers (either in isolation or as part of a larger register system) to write for these data forms as soon as possible to Dr James R. Miller, Department of Medical Genetics, The University of British Columbia, Vancouver, B.C., V6T 1W5, Canada.

Eighth Stadler Genetics Symposium

The eighth Stadler Genetics Symposium will be held in the University of Missouri, 9-10 April 1976. Information and detailed programmes can be obtained by writing to: Conferences and Short Courses, University of Missouri, 347 Hearnes Building, Columbia, MO 65201, U.S.A.

Teratology Society: 1976 Annual Meeting

The Teratology Society will hold its Sixteenth Annual Meeting 20-23 June 1976 at Highlands Inn, Carmel, California, U.S.A.

Further information can be obtained by writing to Dr Lucille S. Hurley, Department of Nutrition, University California, Davis, California 95616, U.S.A.
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