Clinical phenotype of haemoglobin Q-H disease

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Seven patients of Chinese origin who had haemoglobin (Hb) Q-H disease were studied. They were found to have a similar clinical phenotype to that of patients with deletional Hb H disease, who have a near identical genotypic configuration. The complete absence of Hb A in Hb Q-H disease and the similar clinical phenotype to deletional Hb H disease lends support to the observation that Hb Q-Thailand shares similar functional properties with Hb A.

Take home messages

- Seven Chinese patients with haemoglobin (Hb) Q-H disease were found to have a similar clinical phenotype to that of patients with deletional Hb H disease, which has a near identical genotypic configuration.
- The complete absence of Hb A in Hb Q-H disease and the similar clinical phenotype to deletional Hb H disease lends support to the observation that Hb Q-Thailand shares similar functional properties with Hb A.

HAEMATOLOGICAL AND CLINICAL FINDINGS

Table 1 details the clinical and haematological data. All of the study subjects are Hong Kong Chinese, with patient number 7 being of Chinese descent from Thailand. Diagnosis was made on Hb pattern studies in all cases. Archival blood samples were of sufficient quantity for genotype determination in four individuals (patients 3, 4, 5, and 7). Genotyping revealed the Hb Q-Thailand mutation, as confirmed by direct nucleotide sequencing of the z1 globin gene, together with 4.2 kb single z globin gene deletion and SEA deletion, as detected by multiplex polymerase chain reaction. Hb A was absent and Hb Q-Thailand accounted for 93.9–97.9% of the total Hb. There was a high proportion of Hb H inclusion bodies, which were detected in 70–90% of red blood cells in these patients on supravital staining.

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Abbreviations: Hb, haemoglobin; SEA deletion, (–SEA) z thalassaemia deletion
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Table 1 Haematological and clinical profiles of patients with Hb Q-H disease

| Patient | Sex | Age (years) | L | S | History of transfusion | Hb (g/l) | MCV (fl) | MCH (pg) | Hb H (%) | Hb A (%) | Hb A2 (%) | Hb F (%) | Hb Q-Thailand (%) | Retic (%) | Bilirubin (μmol/l) | Iron (μmol/l) | Ferritin (pmol/l) | Transferrin saturation (%) |
|---------|-----|-------------|---|---|------------------------|---------|---------|---------|---------|---------|---------|---------|---------|-----------------|---------|-----------------|-------------|-----------------|-----------------------|
| 1       | M   | 58          | N | N | N                       | 96      | 83.4    | 20.0    | 70      | 0       | 0.3     | <0.3 97.9 | <0.3 97.9 | 92       | 90    | 32              | 1430    | 77              |             |
| 2       | M   | 17          | N | N | N                       | 105     | 60.5    | 18.5    | 80      | 0       | 3.4     | <0.3 93.9 | <0.3 93.9 | 9.2      | 90    | 32              | 1430    | 77              |             |
| 3       | M   | 28          | Y | Y | N                       | 100     | 63.6    | 18.8    | 90      | 2.7     | 95.6    | <0.3 95.6 | 4.7      | 35    | 20              | 965     | 16              |             |
| 4       | M   | 23          | N | Y*| Y                       | 105     | 59.7    | 18.4    | 89      | 2.4     | 9.5     | <0.3 95.8 | 4.7      | 35    | 20              | 965     | 16              |             |
| 5       | M   | 38          | N | N | N                       | 10.9    | 66.6    | 18.1    | 90      | 3.0     | 94.2    | 0.5      | 94.2   | 3.7   | 30              | 1530    | 38              |             |
| 6†      | F   | 68          | N | Y*| Y                       | 79      | 70.2    | 18.9    | 70      | 2.1     | 93.9    | <0.3 93.9 | 6.5     | –     | 25              | 467     | –               |             |
| 7†      | F   | 49          | Y | Y | Y                       | 84      | 77.2    | 21.7    | 85      | 1.2     | 96.9    | 5.0      | 96.9   | 5.0   | 26              | 1907    | –               |             |

HbH disease

5 cases

* Splectomy performed; † gallstones present; ‡ hepatitis C carrier, liver biopsy showed chronic hepatitis and grade 2 iron overload (modified Scheuer grading); mean values quoted for age and laboratory parameters in these 12 patients with deletional HbH disease as a result of α-SEA/α-SEA configuration; ß-splenectomy performed in one patient.

Reference ranges: Hb, 130–180 g/l (men), 115–165 g/l (women); MCV, 80–96 fl; MCH, 27–32 pg; HbA2, 2.3–3%; HbF<0.9%; reticulocytes, 0.2–2%; bilirubin, 7–19 μmol/l; iron, 9–33 μmol/l (men), 5–28 μmol/l (women); ferritin, 115–884 pmol/l (men), 15–331 pmol/l (women); transferrin saturation, 15–45%.

Hb, haemoglobin; Hb H, percentage of red cells with HbH inclusions; L, hepatomegaly; MCH, mean cell haemoglobin; MCV, mean cell volume; N, absent; NA, not applicable; Retic, reticulocytes; S, splenomegaly; Y, present; –, data not available.

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