Medical Memoranda

Thalassaemia Major in the Sudan

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Thalassaemia was not known in the Sudan until three years ago, when one case was described in a Sudanese family in which haematological investigations and family study proved the presence of thalassaemia major in the child and the trait in the parents (Vella and Hassan, 1961). This case drew attention to the existence of thalassaemia in Sudanese people. Two cases of thalassaemia minor in adult patients presenting with mild recurrent haemolytic anaemia were recently seen in Khartum Hospital (Daoud Mustafa, unpublished report). Another case of thalassaemia major was recently diagnosed in Omdurman Hospital (Abdulla and Ziada, unpublished report).

This paper deals with a case of thalassaemia major in a Sudanese family that is in no way related to people of Mediterranean stock.

CASE REPORT

A girl aged 3 was brought to hospital because of continuous fever, watery diarrhoea, and occasional vomiting for seven days before admission. For the last six months her mother had noticed an abdominal swelling which was increasing in size. There was nothing significant in the prenatal, natal, and post-natal history. The child's growth and development were normal during the first year of life, but subsequently she failed to thrive.

On examination she was severely anaemic. Her intelligence was normal, but she was physically retarded. She weighed 21 lb. 3 oz. (9.6 kg.), her height was 34 in. (86.3 cm.), her head circumference 18 in. (45.7 cm.), and her chest circumference 17½ in. (45 cm.).

Girl, aged 3, with thalassaemia major. The facies is characteristic.

She had marked bossing of the forehead and prominent malar eminences, and a depression of the nasal bridge that gave her the characteristic mongoloid appearance (see Fig.). The spleen was grossly enlarged, palpable 10 cm. below the left costal margin, firm, and not tender. The liver was palpable 6 cm. below the right costal margin. The pulse was 128 per minute, blood-pressure 84/54 mm. Hg. There was no cardiac enlargement, but a haemic murmur could be heard at the base. There was generalized lymphadenopathy, the glands were discrete and painless.

Various tests were carried out to detect the endemic diseases, and malaria, schistosomiasis, kala-azar, typhoid and paratyphoid fevers, melitensis, and ankylostomiasis were all excluded. Haematological findings were: haemoglobin 5 g./100 ml.; P.C.V. 21%; M.C.H.C. 24%; white cells 16,500/c.mm. (neutrophils 70%, lymphocytes 30%). A peripheral blood film showed marked hypochromia with numerous target cells and nucleated red cells in addition to anisocytosis and poikilocytosis. The reticulocyte count was 7.4%; serum bilirubin 3.2 mg./100 ml., sickling test negative (sodium metabisulphite). Direct Coombs test negative; osmotic fragility was markedly decreased. Foetal haemoglobin 79% (alkali denaturation method). A sample of blood was sent to Professor J. A. P. Jonxis, who also found foetal haemoglobin 79% and haemoglobin A₂ 3.4%. Radiographs of the bones showed early changes in the metacarpal bones, mainly widening of the medullary spaces and osteoporosis, and thickening of the skull.

Family study showed that the patient's father, aged 28, a cotton farmer in Gezira, and his mother, aged 20, were first cousins. They belonged to a well-known Sudanese tribe in which close inter-marriage is common, but there was no evidence of foreign ancestry. They had another younger daughter, aged 2 years, who was healthy. Both parents were examined and their blood investigated. They both had a mild hypochromic microcytic anaemia, and osmotic fragility was decreased. Samples of the parents' blood were sent to Professor Jonxis, who found that both the father and mother had a raised haemoglobin A₂, 5.2% and 5.9% respectively. No foetal haemoglobin was detected in their blood.

COMMENT

There is evidence suggesting that thalassaemia major occurs more often in the Sudan than has been previously supposed. In addition to the case reported here, we have had two other children under our care who were referred from district hospitals because of resistant anaemia, and in whom haematological investigations revealed an abnormally high foetal haemoglobin. Complete family studies were not possible because their fathers were not available.

It is interesting to note that the Sudan shares with countries in the Far East—for example, Thailand, Malaysia, Indonesia—where the incidence of thalassaemia is high the same geographical zone in the peri-equatorial belt. The close correlation between the incidence of malaria and thalassaemia further supports this view, since malaria is endemic in many parts of the Sudan.

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