HEREDITARY SPHEROCYTOSIS IN THE BANTU

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The important diagnostic features of hereditary spherocytosis (congenital spherocytosis, acholuric jaundice) are that it is a congenital and hereditary disease, with spherocytosis and increased osmotic fragility of the red cells, and an excellent response to splenectomy. The disease is rare in Negroes, and the cases recorded in this ethnic group have been discovered in the United States.[1] In the South African Bantu, while acquired haemolytic anaemias are common, the hereditary forms are reported to be rare.[2] The report must however be treated with some reserve, since it is difficult to investigate the families of Bantu hospital patients. Thus, while acceptable examples of hereditary spherocytosis in the Bantu have not been recorded, cases such as that recently reported by Gon[3] might be found to be examples of the disease, were it possible to examine members of the family.

It thus seems worth while recording an example of hereditary spherocytosis in a Bantu patient in whom all the necessary diagnostic criteria were fulfilled.

CASE REPORT

W.M., a Bantu male aged 17, was admitted to Baragwanath Hospital in December 1957 complaining of pain in the right hypochondrium and anorexia of 4 days' duration. He had noticed that his urine was dark and his eyes had become yellow over the previous 2 days. He had not experienced a similar episode previously.

Physical examination showed a thin Bantu male, with slight conjunctival pallor. The spleen was enlarged to 4 cm. below the costal margin. Neither hepatomegaly nor lymphadenopathy was noted.

TABLE I. RELEVANT HAEMATOLOGICAL FINDINGS IN THE PATIENT AND HIS FAMILY

	Jan. 1958	*Jun. 1958*	*17 Jul. 1958**	*19 Jul. 1958*	*25 Jul. 1958*	*15 Aug. 1958*	*Dec. 1958*	*Mother*	*Father*	*Sister*
Haemoglobin g./100 ml.	9·3	12·8		15·8	14·7	15·7	16·3	11·2	17·5	15·3
Packed cell volume %	25	33		43	42	44	45	31	50	47
Mean corpusc. haem. conc. %	37	38		37	35	36	36	36	35	33
Reticulocytes %	13	13		7	5	1	1	9	1	1
Spherocytosis	++	++		+	+	+	+	++	—	—
Serum bilirubin mg./100 ml.										
total	2·5	3·0		1·7	0·8	0·4	0·3	1·0	0·3	0·3
direct	0·4	0·2								

* Splenectomy performed: 1 pint of blood transfused.

Laboratory Investigations (the haematological methods are those described by Dacie[4])

The relevant haematological data are shown in Table I. There was moderate normochromic anaemia with reticulocytosis and hyperbilirubinaemia. Numerous microspherocytes were present in the peripheral blood smears (Fig. 1), but no other morphological abnormalities of the red cells were prominent. The osmotic fragility and autohaemolysis of the red cells was increased (Table II), and the post-incubation increase in fragility was considerably greater than normal. Bone marrow aspirated from the sternum showed normoblastic hyperplasia. The urine contained excessive urobilin, but bilirubin was not present.

The direct and indirect Coombs tests were negative, and antibodies could not be demonstrated with enzyme-treated red cells. Ham's acid-serum test and the Donath-Landsteiner test were negative, and cold agglutinins were not present. Electrophoresis of the haemoglobin in a veronal buffer (pH 8·6) showed a single component with the mobility of haemoglobin A. Alkali-resistant haemoglobin was 0·3%, and solubility in 2·58 M phosphate buffer (pH 6·9, 25°C) was 1·30 g. per litre. Schumm's test was negative, sickling could not be demonstrated, and malaria parasites were not observed. The V.D.R.L., Kahn, Kolmer and treponema-immobilization tests were negative.

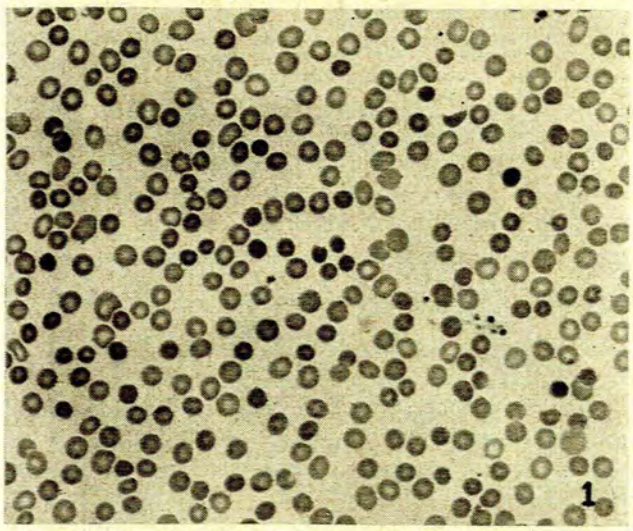

Fig. 1. Peripheral blood film showing numerous spherocytes. × 470.

TABLE II. RESULTS OF TESTS FOR RED-CELL OSMOTIC FRAGILITY AND AUTOHAEMOLYSIS, EXPRESSED AS % HAEMOLYSIS

Osmotic Fragility

% NaCl	*Pre-incubation*		*After incubation at 37°C for 24 hours*	
	Normal range	*Patient*	*Normal range*	*Patient*
0·20		100		100
0·25		100		100
0·30	97–100	100	80–100	100
0·35	90–99	100	72–100	100
0·40	50–90	99	65–100	100
0·45	5–45	90	54–96	100
0·50	0–5	18	36–88	96
0·55	0	5	5–70	72
0·60	0	3	0–40	44
0·65	0	2	0–19	22
0·70	0	1	0–9	12

Autohaemolysis

% NaCl	24 *hours*		48 *hours*	
	Normal range	*Patient*	*Normal range*	*Patient*
0·20	0–0·5	1·2	0·4–3·5	4·8

Course

The condition of the patient was observed over 6 months, during which time anaemia, reticulocytosis, spherocytosis, and hyperbilirubinaemia persisted. A red-cell survival study with radio-active chromium was carried out, and the time taken for half the radio-activity to disappear from the blood ($T\frac{1}{2}Cr$) was 11 days (Fig. 2). There was thus marked shortening of the red-cell life span, $T\frac{1}{2}Cr$ being normally 26 ± 2 days.

Splenectomy was carried out in July 1958. The spleen was enlarged, measuring 15·5 × 11·5 × 8 cm. Histological examination showed congestion of the pulp with dilatation of the sinuses and wide separation of the follicles. Haemosiderin was present in macrophages scattered throughout the section. The post-operative course was uneventful, and 5 weeks after splenectomy the patient was well, with haemoglobin value, reticulocyte count

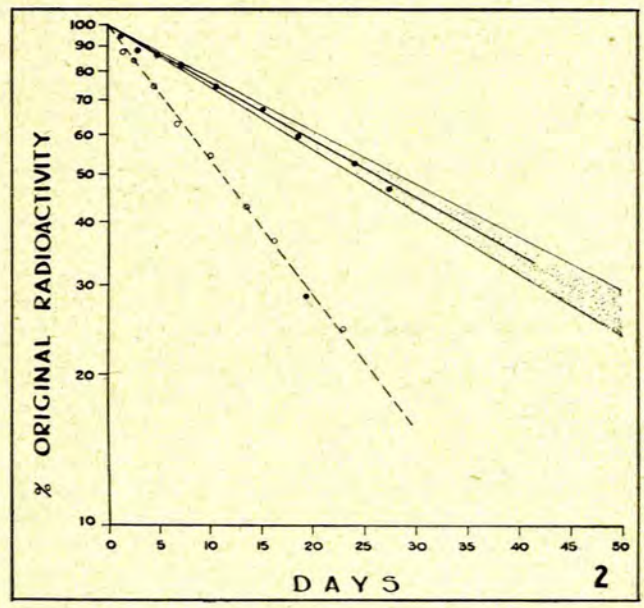

Fig. 2. Red-cell survival before (broken line) and after (continuous line) splenectomy. The shaded area represents the normal range.

and serum bilirubin within normal limits (Table I). Some microspherocytes were still present in the peripheral blood film. The red-cell survival study was repeated, and the red-cell life span was now within normal limits, $T\frac{1}{2}Cr$ being 26 days (Fig. 2). The patient was re-examined 6 months after splenectomy, when he was free of symptoms, with normal haemoglobin value, reticulocyte count and serum bilirubin.

The Family

The family consists of the father (Bechuana), and mother (Zulu), the patient and 2 sisters. The father is in good health; his spleen is not palpable, and there is no haematological abnormality. The mother volunteered a history of 4 or 5 attacks of headache, jaundice and body pains, the last attack dating back to 1945. Physical examination of the mother showed an enlarged spleen palpable 2 cm. below the costal margin. Her blood count (Table I) showed mild anaemia, reticulocytosis, and the presence of numerous microspherocytes in the peripheral blood film. The serum bilirubin was 1·0 mg.%, and there was excessive urobilinuria but no bilirubinuria.

The mother's parents are dead, and she has 4 sisters, none of whom were available for examination. One sister, however, was stated to suffer repeated attacks of jaundice, and to have an enlarged spleen.

Of the patient's 2 siblings, only one was available for examination. The spleen was not palpable, and there was no haematological abnormality (Table I).

DISCUSSION

While hereditary spherocytosis does occur in the South African Bantu, it is evidently rare, and is certainly rarer than in the South African White population, among whom the disease appears to be not uncommon. Although it is difficult to establish the hereditary nature of the disease in Bantu subjects, in the overt case it is often possible to provide evidence of the condition on the haematological findings and the results of splenectomy. During the 3-year period 1956-58, 106 examples of haemolytic anaemia were diagnosed at Baragwanath Hospital. Of this number, the present case, together with 1 other showing very similar features (including the excellent result of splenectomy with return of the red-cell life span to normal) but whose family were not available for examination, were considered to be the only examples of hereditary spherocytosis.

SUMMARY

A case of hereditary spherocytosis in a Bantu family is presented. The disease is apparently quite rare in the South African Bantu.

I wish to thank Dr. V. H. Wilson, Senior Physician, Baragwanath Hospital, for permission to publish this case, which was under his care; Dr. C. G. Anderson for the haemoglobin studies; Dr. V. Bokkenheuser for the treponema-immobilization test, and Dr. S. M. Lewis for helpful criticism.

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