To the Editor:

I was greatly interested to read the article by Gross on "Hematologic Studies on Erythropoietic Porphyria," appearing in the June issue of Blood (23:762, 1964). An experience with four cases of classical erythropoietic porphyria (PE) prompts us to mention some of our findings related to the presence of a hemolytic component. The patients were Bengalees residing in and around Calcutta. All were classical cases with red urine, red teeth, characteristic photosensitive skin lesions, excessive growth of hair, fluorescent normoblasts in the marrow and excessive excretion of type 1 copro- and uroporphyrins. Details of three of these cases were published in Indian Journals.1,2,3 In the Indian literature there were some earlier references to PE which have recently been reviewed.1

I am writing this letter particularly to point out briefly our observations related to the hemolytic component and to the status of Hb F (alkali-resistant fraction) in these cases.

In our first case (S. C.), a 4-year-old boy, there was evidence of severe hemolytic anemia: spleen enlarged, 10 cm.; Coombs test, positive; Hb, 4.3 Gm.; PCV, 14 per cent; Ret., 9 per cent; Hb F, 5.4 per cent; plasma bilirubin, 1.25 mg. per cent; platelet count, 120,000 per mm. (Dameshek's method). Our average normal range was 400,000 to 600,000. With a course of prednisolone therapy, there was a significant clinical response and within one month hemoglobin level improved to 10.8 Gm. per cent, PV to 33 per cent and platelet count to 350,000.

In the second case, (C. B.), a 19-year-old boy, there was no significant evidence of overt hemolysis: spleen was not palpable; Hb, 13.6 Gm.; PCV, 36 per cent; Ret., 3 per cent; plasma bilirubin, 0.4 mg. per cent; platelet count, 432,000; glutathione stability test, abnormal; initial level of 54 mg. per cent dropped to 22.0 mg. on incubation with acetyl phenyl hydrazine (APH); glucose-6-phosphate dehydrogenase activity (G-6-PD), 1.87 minute/ml. RBC/minute; serum vitamin B₁₂, 240 µg. per ml; serum iron, 135 µg. per cent; direct Coombs test, negative. With a course of therapy with prednisolone, there was a significant reduction of urinary porphyrins.

The third case, a 9-year-old boy was unique in having a combination of three inherited defects—PE, G-6-PD deficiency (0 unit), and a heterozygous state of Hb E.3 Parenthood was consanguinous. Spleen was enlarged 2.5 cm.; Hb, 9.2 Gm. per cent; PCV, 28 per cent; Ret., 4 per cent; Hb F, 1.1 per cent; plasma bilirubin, 0.6 mg. per cent; platelet count, 610,000; direct Coombs test, negative; serum vitamin B₁₂, 290 µg./ml.; serum iron, 64 µg. per cent. The glutathione stability test was abnormal with an initial level of 45.0 mg. dropping to 14.0 mg. after incubation with APH. In bone marrow material, an interesting feature was the presence of a few reticulum cells containing darkly stained globular masses which represented possibly the ingested nuclear masses, and also some fluorescing material, presumably porphyrins. This finding could be taken as morphologic evidence of a supposed intramedullary hemolysis of porphyrin-laden normoblasts, the role of the phagocytic reticulum cells being that of scavengers.3

With a course of prednisolone therapy the hemoglobin level improved to 11.6 Gm. per cent and the characteristic phagocytic reticulum cells were no longer visible.

The fourth case was an 8-year-old boy: spleen enlarged, 10 cm.; liver enlarged, 5.5 cm.; Hb, 8.7 Gm. per cent; PCV, 27 per cent; Ret., 20 per cent; Hb F, 2.7 per cent; platelets, 310,000; direct Coombs test, negative; serum iron, 64 µg. per cent. Erythrocytic reduced glutathione presented an unstable pattern with normal activity of G-6-PD. A course of prednisolone improved the Hb level to 11.3 Gm. and PCV to 35 per cent.

In only one of our cases was the Hb F content increased. Evidences of an hemolytic component were, however, clearly seen, although the mechanism of hemolysis was by no means clear. The positive direct Coombs test in one case and the significant response...
to steroid therapy in all the cases suggest the possibility of an associated autoimmune mechanism. The presence of thrombocytopenia in the first case and its correction with steroid therapy are compatible with an autoimmune mechanism. A significant degree of intramedullary hemolysis was also likely to contribute to the development and perpetuation of the hemolytic component.

In view of the limited circulation of the journals in which our reports of this rare disease were published, I thought that the brief information in this letter would be of interest to the readers of Blood.

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REFERENCES