To the Editor:

Erythropoietic protoporphyrin (EPP) is a rare genetic disease, the symptoms of which are itching or burning sensations in light-exposed skin.\(^1\) In EPP, ferrochelatase, the enzyme that inserts iron into protoporphyrin to form heme, is defective. This defect leads to excess accumulation of protoporphyrin in erythroid cells and plasma and excretion of the excess protoporphyrin in the bile and feces. If a smear of peripheral blood is examined under a fluorescence microscope using 405 nm excitation light, 5% to 20% of the erythrocytes have a red fluorescence.\(^2\)

FIG 1. (A) BFUe from an EPP patient, light microscope (original magnification \(+\) 100). (B) BFUe from (A), fluorescense microscope, 405 nm excitation light. (C) BFUe from a normal individual, light microscope (original magnification \(+\) 100). (D) BFUe from (C), 405 nm excitation light.
We have found that, when peripheral blood mononuclear cells are cultured in methyl cellulose medium fortified with interleukin-3, interleukin-6, granulocyte-macrophage colony-stimulating factor, Steel factor, and erythropoietin, all burst-forming unit-erythroid (BFUe) colonies grown from the blood of EPP patients fluoresced when viewed under 405 nm light, whereas BFUe from the blood of normal individuals did not fluoresce. Figure 1A shows a BFUe from an EPP patient viewed under the light microscope. Figure 1B shows the same BFUe viewed under the fluorescence microscope, using 405 nm excitation light. Figure 1C shows a BFUe from a normal individual viewed under the light microscope. Figure 1D shows the same BFUe viewed under 405 nm excitation light.

Although β-carotene is effective in preventing the symptoms of EPP, only genetic therapy will cure this disease. Because the presence of fluorescence in BFUe from EPP patients is an indication of defective ferrochelatase activity, we suggest that the presence or absence of fluorescence in EPP BFUe derived from stem or progenitor cells transfected in vitro with the normal ferrochelatase gene can be used to determine whether normal ferrochelatase enzyme activity has been restored.

REFERENCES
Burst-forming units-erythroid from erythropoietic protoporphyria patients fluoresce under 405 nm light [letter]

MM Mathews-Roth, RJ Wise and BA Miller