TREATMENT

Hemochromatosis

The therapy of hemochromatosis involves removal of the excess body iron and supportive treatment of damaged organs. Iron removal is best accomplished by weekly or, with gross iron loading, twice-weekly phlebotomy of 500 mL. Although there is an initial modest decline in the volume of packed red blood cells to about 35 mL/dL, the level stabilizes after several weeks. The plasma transferrin saturation remains increased until the available iron stores are depleted. In contrast, the plasma ferritin concentration falls progressively, reflecting the gradual decrease in body-iron stores. One 500-mL unit of blood contains 200–250 mg of iron, and ≥25 g of iron may have to be removed. Therefore, in patients with advanced disease, weekly phlebotomy may be required for 1–2 years, and it should be continued until the serum ferritin level is ≤100 μg/L. Thereafter, phlebotomies are performed at appropriate intervals to maintain ferritin levels at ≤100 μg/L. The transferrin saturation fluctuates and may still be elevated but should not dictate further therapy unless it is persistently at 100% when free unbound iron may circulate. Usually one phlebotomy every 3 months will suffice. It is important, however, not to overtreat and render the patient iron deficient.

Chelating agents such as deferoxamine, when given parenterally, remove 10–20 mg of iron per day, which is much less than that mobilized by once-weekly phlebotomy. Phlebotomy is also less expensive, more convenient, and safer for most patients. However, chelating agents are indicated when anemia or hypoproteinemia is severe enough to preclude phlebotomy. Subcutaneous infusion of deferoxamine using a portable pump is the most effective means of its administration.

Effective oral iron chelating agents, deferasirox (Exjade) and deferiprone, are now available. These agents are effective in thalassemia and secondary iron overload, but are expensive and carry the risk of significant side effects.

Alcohol consumption should be severely curtailed or eliminated because it increases the risk of cirrhosis in hereditary