Hereditary hemochromatosis is characterized by the abnormal progressive absorption of dietary iron in the intestines, which is then stored in the liver, heart, and other organs. Inherited as an autosomal recessive disorder, the most common genotype that causes hereditary hemochromatosis is HFE C282Y/C282Y (Adams & Barton, 2010). Signs and symptoms often appear in adults aged 50–69. Complications in the liver include fibrosis, cirrhosis, complete liver failure, or hepatocellular cancer. Damage to other organs from iron overload may result in diabetes mellitus, cardiomyopathy, gonadal dysfunction, arthritis, and dementia. Excess iron storage is thought to be toxic to cells. Increased amounts of free radicals are generated that can damage cellular and subcellular membranes. In this disease, the goal of treatment with phlebotomy is to deplete enough iron to normalize the body’s iron stores and prevent or minimize organ dysfunction (McLaren, 2002).

Diagnosis of hereditary hemochromatosis is based on symptoms and laboratory studies. If the patient is asymptomatic, suspicion of iron overload may first be raised when laboratory tests reveal elevated hemoglobin, hematocrit, and ferritin. Normal ferritin in men is about 18–464 mg/dl; for women it ranges from 12–262 mg/dl. Patients with this disorder may present with readings as elevated as 1,000–5,000 mg/dl. Iron saturation also is typically elevated (normal is 15%–50%). A definitive diagnosis can be made on the basis of genotyping for the HFE mutation. A positive result suggests testing of first-degree relatives (particularly siblings and children) (Schrier & Bacon, 2009). A patient suspected of having iron overload but who is negative for the common genetic variants associated with the disease may undergo a specialized liver magnetic resonance imaging scan to visualize liver iron stores. If this imaging procedure is not available, a liver biopsy may be performed.

Treatment of iron overload is based on phenotype (high ferritin and evidence of elevated iron stores), not genotype (Adams & Barton, 2010). Genotype is a pair of genes for a particular characteristic or protein, whereas phenotype is the observable expression of a trait or characteristic that is visible or biochemically detectable (Lashey, 1998). Phlebotomy is the most effective method to deplete iron stores in a patient that is not anemic. Each phlebotomy removes about 500 ml of blood, which contains 200–250 mg of iron. This process then depletes the body’s iron stores by mobilizing approximately the same amount of iron out of the liver. The goal of treatment is to remove the circulating iron as quickly and safely as possible. Depending on the size and overall condition of the patient, phlebotomy can be performed once or twice weekly. The end-point goal of treatment is a serum ferritin in the range of 50–100 mg/dl. Therefore, the length of treatment depends on the baseline level. To prevent anemia, a hemoglobin and hematocrit usually is drawn before each phlebotomy. Parameters ordered by the provider to hold the treatment will be compared with the most recent hemoglobin and hematocrit usually is drawn before each phlebotomy. Parameters ordered by the provider to hold the treatment will be compared with the most recent hemoglobin and hematocrit usually is drawn before each phlebotomy. Parameters ordered by the provider to hold the treatment will be compared with the most recent hemoglobin and hematocrit usually is drawn before each phlebotomy. Parameters ordered by the provider to hold the treatment will be compared with the most recent hemoglobin and hematocrit usually is drawn before each phlebotomy. Parameters ordered by the provider to hold the treatment will be compared with the most recent hemoglobin and hematocrit usually is drawn before each phlebotomy. Parameters ordered by the provider to hold the treatment will be compared with the most recent hemoglobin and hematocrit usually is drawn before each phlebotomy. Parameters ordered by the provider to hold the treatment will be compared with the most recent hemoglobin and hematocrit usually is drawn before each phlebotomy. Parameters ordered by the provider to hold the treatment will be compared with the most recent hemoglobin and hematocrit usually is drawn before each phlebotomy. Parameters ordered by the provider to hold the treatment will be compared with the most recent hemoglobin and hematocrit usually is drawn before each phlebotomy. Parameters ordered by the provider to hold the treatment will be compared with the most recent hemoglobin and hematocrit usually is drawn before each phlebotomy. Parameters ordered by the provider to hold the treatment will be compared with the most recent hemoglobin and hematocrit usually is drawn before each phlebotomy. Parameters ordered by the provider to hold the treatment will be compared with the most recent hemoglobin and hematocrit usually is drawn before each phlebotomy. Parameters ordered by the provider to hold the treatment will be compared with the most recent hemoglobin and hematocrit usually is drawn before each phlebotomy. Parameters ordered by the provider to hold the treatment will be compared with the most recent hemoglobin and hematocrit usually is drawn before each phlebotomy. Parameters ordered by the provider to hold the treatment will be compared with the most recent hemoglobin and hematocrit usually is drawn before each phlebotomy. Parameters ordered by the provider to hold the treatment will be compared with the most recent hemoglobin and hematocrit usually is drawn before each phlebotomy. Parameters ordered by the provider to hold the treatment will be compared with the most recent hemoglobin and hematocrit usually is drawn before each phlebotomy.

In addition to phlebotomy, some practitioners recommend specific dietary restrictions to reduce dietary absorption of iron. The restrictions include limiting alcohol intake and avoiding vitamin C supplements, oral iron replacement,