trait and referred for genetic counseling.

As with any chronic illness, the family's needs must be met for optimal adjustment to the stresses imposed by the disorder (see Chapter 19). Sources of information for the family include the Cooley's Anemia Foundation* and the Northern California Comprehensive Thalassemia Center. Genetic counseling for the parents and fertile offspring is mandatory, and both prenatal diagnosis using amniocentesis or fetal blood sampling and screening for thalassemia trait are available.

Aplastic Anemia

Aplastic anemia (AA) is a rare and life-threatening disorder, which can be satisfactorily treated in about 90% of cases (Miano and Dufour, 2015). It refers to a bone marrow failure condition in which the formed elements of the blood are simultaneously depressed. To diagnose AA, the peripheral blood smear demonstrates pancytopenia with at least two of the following present: profound anemia, leukopenia, and thrombocytopenia. Whereas, **hypoplastic anemia** is characterized by a profound depression of RBCs but normal or slightly decreased WBCs and platelets.

Etiology

AA can be primary (**congenital**, or present at birth) or secondary (**acquired**). The best-known congenital disorder of which AA is an outstanding feature is **Fanconi syndrome**, a rare hereditary disorder characterized by pancytopenia, hypoplasia of the bone marrow, and patchy brown discoloration of the skin resulting from the deposit of melanin. It is associated with multiple congenital anomalies of the musculoskeletal and genitourinary systems. The syndrome appears to be inherited as an autosomal recessive trait with varying penetrance; therefore, affected siblings may demonstrate several different combinations of defects.

Several etiologic factors contribute to the development of acquired AA; however, most of the cases are considered idiopathic (Box 24-4). The following discussion focuses on severe acquired AA, which carries a poorer prognosis and follows a more rapidly fatal course than the primary types.