Thalassemia Syndromes in Pregnancy

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Imagine this scenario: A 20-year-old woman reports to the obstetric triage unit complaining of fatigue and light-headedness, with very pale skin. She has had no prenatal care, she states that this is her first pregnancy, and her dates are consistent with a 22-week gestation. The fetal heart rate is in the normal range, and ultrasonography results indicate that the dates are accurate. Both of her parents are of Asian descent. Her hemoglobin and hematocrit levels are 8.2 g/dl and 24.7%, respectively, and mean corpuscular volume is 70 μm^3. The obstetrician on call suspects a thalassemia and orders a hemoglobin electrophoresis and iron studies.

What Are Thalassemia Syndromes?

Thalassemia syndromes are the most common genetic blood disorders worldwide, and

Abstract Thalassemia syndromes are becoming more common in the United States as the population becomes more diverse. To provide appropriate care to this patient population, nurses must know that thalassemia syndromes are classified into two main types, α-thalassemia and β-thalassemia. α-Thalassemia is further delineated into four clinical patterns: silent carrier state, mild α-thalassemia, hemoglobin H disease, and hydrops fetalis. Understanding each of these complex anemias and their potential effects on a pregnant woman and her fetus will enable nurses to interpret these women's unique laboratory test results. Intervention when necessary with appropriate treatment may lead to optimal outcomes for women and newborns. http://dx.doi.org/10.1016/j.nwh.2016.07.008

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they are found predominately in the Mediterranean, Southeast Asia, India, and the Middle East. Characterized by anemia, these autosomal recessive disorders result from hemoglobin defects or structural abnormalities that impair globin synthesis (Sheiner, Levy, Yerushalmi, & Katz, 2004). Hemoglobin molecules are carried by erythrocytes and are responsible for transporting oxygen to the body's tissues. Erythrocytes originate from bone marrow stem cells, and their production is regulated by the hormone erythropoietin, which is controlled by the kidneys. Erythrocyte formation, or erythropoiesis, is triggered when oxygen sensors in the kidneys detect increasing tissue oxygen requirements (Higgs, Engel, & Stamatoyannopoulos, 2012).

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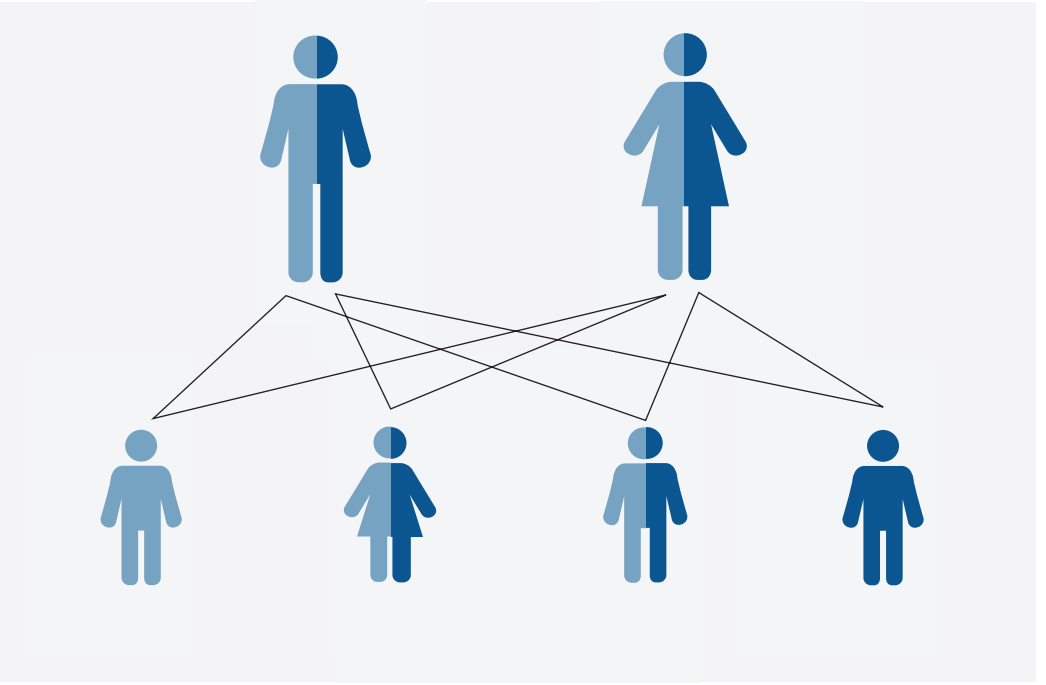
Hemoglobin's ability to bind with oxygen and release oxygen into tissues is dependent on its 2,3-diphosphoglycerate structure, affinity, and concentration. These molecules consist of a nonprotein central heme complex surrounded by four protein globin chains (Leung & Lao, 2012). The globin chains are composed of two α-globin and two non-α–globin chains such as two β subunits (α2β2) or two δ (α2δ2) subunits.

How Are Thalassemia Syndromes Classified?

Thalassemia syndromes are classified into two main types, α-thalassemia and β-thalassemia, based on the subunits or chains that are affected (see Table 1). α-Thalassemia is further delineated into four clinical patterns: silent carrier state, mild α-thalassemia, hemoglobin H disease (Hb H), and Bart's hemoglobin (Hb Bart's). In people with α-thalassemia, the larger the number of alleles affected, the greater the severity of clinical manifestations (Leung & Lao, 2012). For example, if only one of the four alleles is abnormal, patients may have a normal or slightly reduced hemoglobin cellular size and be asymptomatic. Having two altered alleles is classified as mild microcytic hypochromic anemia, and, if symptomatic, patients will commonly complain of increasing fatigue or light-headedness. When three alleles are altered, patients will present with mild to moderate microcytic hypochromic anemia, or Hb H. Hb H has the highest survivable severity, but because of its instability, it can result in an increase in red blood cell (RBC) destruction, causing the patient to require perpetual blood transfusions. If all four alleles are missing, it results in Hb Bart's, a type of hydrops fetalis, a condition incompatible with life. Hb



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