

Thalassemia Tests

Mayo Medical Laboratories provides, through the Metabolic Hematology and Molecular Genetics Laboratories, tests for the detection of alpha-thalassemias, beta-thalassemias, delta-beta-thalassemia, and of hemoglobin variants that are commonly accompanied by thalassemias: hemoglobin H, hemoglobin Lepore, hemoglobin Barts, unstable hemoglobin, hemolytic anemias, hemoglobin E, hereditary persistence of high fetal hemoglobin (several varieties), and combinations of hemoglobin S with alpha- or beta-thalassemia, hemoglobin E/beta-O-thalassemia, and many other complex thalassemic disorders. In addition, the Molecular Genetics Laboratory offers tests for alpha-thalassemia by DNA probe analysis. Some of the alpha-thalassemias (eg, hemoglobin H disease) can be reliably identified by hemoglobin electrophoresis alone; some require DNA probe studies. DNA probe studies reveal deletional mutations that include most but not all alpha-thalassemias. The hemoglobin tests and the DNA probe studies are often complementary. Therefore, we offer our clients a consultative evaluation to assist in identifying thalassemia disorders. See #84158 “Thalassemia and Hemoglobinopathy Evaluation.”

Since iron deficiency can mimic thalassemias, we include a plasma ferritin test as an option to evaluate this possibility.

Specimens must be accompanied by a completed copy of the “Thalassemia/Hemoglobinopathy Information Sheet” (Supply T358).