



The 67th ASH Annual Meeting Abstracts

POSTER

904. OUTCOMES RESEARCH: HEMOGLOBINOPATHIES

Characteristics of women who have beta thalassemia discovered during pregnancySteven Fein¹, Dayne Alonso¹, Adam Lewkowitz², Gina Peralta¹, Nicole Peralta¹, Mariana Alvarado¹¹Heme on Call, telemedicine hematology practice, Miami, United States²Brown University, Obstetrics and Gynecology, Providence, United States

Abstract Background: Anemia is common during pregnancy, usually attributed to iron deficiency. Some pregnant women with anemia are found to have normal iron or microcytosis out of proportion to iron deficiency, leading to more testing and occasional diagnosis of beta thalassemia minor (i.e., subclinical non-transfusion-dependent beta thalassemia). Most women diagnosed in this way are unaware of their hematologic disorder, the challenges it may pose during their pregnancy (e.g., peripartum anemia or RBC transfusions) and the potential sequelae (e.g., future iron overload or early osteoporosis). Also, some patients referred for iron infusions may be mistakenly assumed to have iron deficiency anemia and prescribed iron infusions. In this study, we describe a cohort of pregnant women with newly diagnosed or rediscovered beta thalassemia minor.

Methods: In this single center case series, we reviewed the charts of pregnant women who were found to have diagnostic tests consistent with beta thalassemia minor (elevated hemoglobin A2 or beta globin mutation). We sought to describe the frequency of newly discovered or re-discovered beta thalassemia minor among pregnant women referred to a women's health-focused hematology practice. Those who were found to have iron deficiency (Ferritin <30ng/dL or iron saturation <20%) received iron infusions totaling 1000mg. We summarized the frequency of iron deficiency and associated symptoms, and we compared changes in hemoglobin and ferritin in those who received iron infusions versus those who did not.

Results: Seventy-six (1.3%) pregnant women out of 5,833 who were referred for anemia, iron deficiency, or microcytosis during 2021-2025 were diagnosed or with beta thalassemia. Most were referred during their 2nd or 3rd trimester. Almost all had elevated hemoglobin A2. One patient was diagnosed based on having a beta globin mutation with normal hemoglobin electrophoresis. 65% were newly diagnosed, whereas 35% were aware of possibly having beta thalassemia from prior testing, prior RBC transfusions (six patients), or known family history of beta thalassemia.

Sixty-seven (88%) of these 76 pregnant beta thalassemia patients had hemoglobin <11g/dL at the time of referral, with average hemoglobin 9.9g/dL (7.5-10.8g/dL). Twenty-nine of 76 (38%) had iron deficiency based on ferritin <30ng/mL or iron saturation <20%, with average hemoglobin 9.7 g/dL (7.5-14.7 g/dL). Among these, only 12 (16%) had iron saturation <20%. At the time of referral, 82% of the entire cohort reported fatigue and 30% reported shortness of breath, including many who did not have iron deficiency.

Among 37 patients who received iron infusions for iron deficiency, follow-up iron and blood counts were measured about two weeks after completing iron infusions. Ferritin increased by an average of 62ng/mL, though there was an average hemoglobin change of only +0.1. Among those who did not have iron deficiency, ferritin decreased by an average of 35ng/mL over about one month, with an average hemoglobin change of 0.

Conclusion: Our findings highlight the prevalence of beta thalassemia among pregnant women referred for anemia or iron deficiency, and the diagnostic challenges of identifying those for whom iron deficiency during pregnancy needs treatment. Consultation with a hematology expert and diagnostic testing can help differentiate between iron deficiency and newly discovered beta thalassemia, ensuring that women receive the most appropriate care. Pregnancy-related diagnostic testing provides a unique opportunity to identify these patients and to improve their long-term health outcomes.

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