P1516 SCREENING FOR BETA-THALASSEMA MINOR IN PREGNANT FEMALES PREVIOUSLY DIAGNOSED
WITH IRON DEFICIENCY ANEMIA: WHAT ARE THE RATES OF MISDIAGNOSIS AND THE NEED FOR AN
INCREASED UPTAKE OF CARRIER SCREENING

Topic: 27. Thalassemias

Sasmith Menakuru1, 2, Sruti Kalla3

1 Internal Medicine, Indiana University Health, Muncie, United States;2 Internal Medicine, Narayana Medical College, Nellore, India;3 Internal Medicine, Maharajah’s Institute Of Medical Sciences, Vizinagaram, India

Background: In rural India, iron deficiency anemia is a common diagnosis in pregnant women; however, several females are possibly misdiagnosed due to lack of follow-up. There are high rates of consanguineous marriages, which may be a cause for the higher rates of beta-thalassemia major in these populations. The rates of beta-thalassemia major may be prevented by proper screening for beta-thalassemia minor during the prenatal period. Uptake of premarital screening in India is poor, and therefore the effectiveness of a prenatal screen was evaluated in those with a prior history of iron deficiency anemia (IDA). The reason for choosing patients with a history of IDA was that the patients already had a diagnosis of microcytic anemia, and the authors wanted to evaluate for possible rates of misdiagnosis.

Aims: To study the prevalence and the need to screen for beta-thalassemia minor in India’s pregnant rural population, and to help prevent couples at risk from producing a fetus with beta-thalassemia major. In addition, an assessment of the rates of misdiagnosis of IDA was done to check the accuracy of the current estimates of beta-thalassemia in India.

Methods:

800 pregnant women, previously diagnosed with IDA, in their first trimester from various rural villages around Andhra Pradesh, India were selected. These 800 females were screened for beta-thalassemia minor during their prenatal screening period. The mean gestational age for screening was 12 ± 4 weeks. Screening was done utilizing a complete iron profile with complete blood count and the Mentzer index, defined as the mean corpuscular volume divided by the red blood cell count. Greater than 13 on the Mentzer index indicated that iron deficiency anemia was more likely, while less than 13 indicated probable beta-thalassemia. Those individuals with a Mentzer index less than 13 were then screened with the Naked Eye Single Tube Red cell Osmotic Fragility Test (NESTROFT) and then confirmed with High-Performance Liquid Chromatography (HPLC). The individuals who were positive for beta-thalassemia minor then had their husbands screened.

Results:

Of the 800 pregnant women screened 47 came back positive for beta-thalassemia minor, equating to 5.87% of the sample size. At-risk couples, defined as both husband and wife being positive for the beta-thalassemia minor was found in 7 couples. The 7 couples were informed about the risk of development of beta-thalassemia major in their fetus, and a prenatal diagnosis was offered to them. Of the 7 patients, 5 elected to terminate the pregnancy after a positive prenatal diagnosis of beta-thalassemia major, and the remaining 2 couples did not respond and were lost to follow-up.

Summary/Conclusion:

There was a moderately high prevalence of undiagnosed beta-thalassemia minor, and presently the uptake of the current voluntary screening program is limited in India. The ultimate goal would be to detect carrier couples with a
premarital screening program; however, it has been challenging to implement thus far. Therefore the authors propose the utilization of a prenatal carrier screening method ideally as early as possible as this may help prevent cases of beta-thalassemia major. There should be more campaigns to increase awareness about the need to screen pregnant females, and if they are positive, then their husbands should be screened as well to detect at-risk couples. The authors also conclude that the rates of misdiagnosis of IDA were equally alarming as there may be more cases of beta-thalassemia minor in India than previously suspected.