Letter to the Editor

[The Editor is not responsible for the views expressed by the correspondents]

Imperative Mandate for Thalassemia Screening in Pregnancy

Sir, — We write to emphasise the compelling need for mandatory and indispensable Thalassemia screening during pregnancy, with a specific emphasis on perinatal diagnosis. Current standards for thalassemia screening in pregnancy involve prenatal screening using various methods such as full blood count, hemoglobin electrophoresis, and thalassemia mutation tests[1]. These screening methods aim to identify pregnant women who are carriers of thalassemia genes, which can help in reducing the incidence of thalassemia[2]. Haematological parameters, such as the Mentzer formula (MCV/RBC) and HbA2 levels, have shown promise in differentiating between different types of thalassemia genotypes in pregnant women[3]. Additionally, raising awareness among pregnant women and their families about thalassemia is crucial to encourage early screening and detection[4]. Laboratories performing antenatal screening should utilize methods capable of detecting significant variants and quantitating haemoglobins A2 and F at the required cut-off points [5]. Overall, the current standards for thalassemia screening in pregnancy involve a combination of different screening methods and raising awareness among pregnant women. Thalassemia, a hereditary blood disorder, requires heightened scrutiny, particularly within distinct communities such as Lohana, Khoja, Agarwal, Bhanushalli, Bania, Neobuddhist, Sindhi, and Jain.

Individuals with chronic microcytosis and normal serum ferritin levels should undergo thorough evaluation for Thalassemia diagnosis. Hemoglobin electrophoresis is crucial for confirming the presence of Thalassemia, while computed parameters such as MCV/RBC ratio and RDW can indicate Beta-Thalassemia Minor[6]. Borderline HbA2 values should be reevaluated after correcting anemia to avoid false diagnoses, as elevated HbA2 levels can be observed in Megaloblastic anemia[7]. Microcytic hypochromic anemia can be caused by various conditions, including IDA and Thalassemia[8]. Thalassemia screening and Level-Three ultrasound during pregnancy are essential for ruling out congenital and hereditary diagnoses[9]. Thalassemia screening should be mandatory for every pregnancy[10].

Individuals manifesting chronic microcytosis and maintaining normal serum ferritin levels within these communities necessitate particular vigilance. An unequivocal Thalassemia diagnosis becomes imperative, making the conduct of hemoglobin electrophoresis paramount. Additionally, a meticulous examination of the Red Blood Cell (RBC) count assumes critical importance; an RBC count surpassing 5 x 10^9/L indicates a predisposition to Thalassemia minor. Various computed parameters, including the MCV/RBC ratio and Red Cell Distribution Width (RDW), serve as valuable indicators for suspecting Beta-Thalassemia Minor.

The gold standard for confirming a Thalassemia diagnosis remains hemoglobin electrophoresis. While normal results are anticipated in cases of Alfa Thalassemia trait, abnormalities signify the presence of Beta-Thalassemia trait or other Thalassemic syndromes. It is imperative to acknowledge that concurrent conditions, such as Iron Deficiency Anemia (IDA) or Megaloblastic anemia, may obscure the typical abnormalities associated with the Beta-Thalassemia trait.

To mitigate the risk of false diagnoses, it is crucial to reevaluate borderline Hemoglobin A2 (HbA2) values after correcting anemia. This is particularly relevant, given that elevated HbA2 levels may be observed in cases of Megaloblastic anemia, potentially leading to an erroneous diagnosis of Beta-Thalassemia minor. Microcytic hypochromic anemia, a frequently encountered clinical scenario, primarily originates from IDA, followed by Thalassemia, anemia of chronic origin, lead poisoning, sideroblastic anemia, and the ingestion of copper or zinc.

In light of these considerations, we underscore the absolute necessity of Thalassemia screening and Level-Three ultrasound during pregnancy as intrinsic components for ruling out congenital and hereditary diagnoses. The unequivocal stance is that every pregnancy should undergo mandatory Thalassemia screening as an indispensable and non-negotiable measure.

Disseminating this information through this esteemed platform will undoubtedly contribute significantly to the collective understanding of this crucial facet of maternal and child healthcare.

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