Automated blood counts and identification of thalassemia carriers

Sir,
I read the article by Kakkar et al., with great excitement. The authors emphasize that clinicians overlook the red cell abnormalities on automated blood counts and also misinterpret the abnormal red cell parameters (they either do not carry out further evaluation for excluding thalassemia or misinterpret microcytosis as iron deficiency). This situation was also brought to the attention of the medical community by other authors.

The most important measure for the control and prevention of thalassemia is early identification and counseling of its carriers, as is also stated by the authors. Over the years a great deal has been achieved towards the management of homozygous thalassemia. However, preventing the birth of a baby with homozygous thalassemia is of primary importance. Complete blood counts (CBCs) provided by a routine automated blood counter are major contributors for extensive screening and appropriate detection of carriers. They are inexpensive and easily accessible by individuals who are not professional in
hematolgy. In the event that automated blood counts are well appreciated, most of the carriers would be easily detected. Therefore, it would be beneficial to make the most use of the cues put forward by CBCs.

Turkey is a Mediterranean country in which thalassemia occurs rather frequently. Carrier frequency is 2.1% in the population and up to 17% in some regions. In accordance with our national health policies, early detection and counseling of at risk population are of primary importance. Our hospital is a tertiary care teaching institution in which approximately 500 new outpatients per year are followed up in hematology and 35-40% of them are referred for differential diagnosis of microcytosis. Therefore, discrimination of these two most common forms of microcytic anemia, which are iron deficiency and thalassemia minor, by the simplest methods such as CBC is very important. The red cell parameters that we use for differentiating these two anemias are Hb (hemoglobin), mean corpuscular volume (MCV), red blood cell (RBC), and red cell distribution width (RDW). Very low Hb (<9 g/dL) is suggestive of iron deficiency (ID), whereas low normal or normal Hb (9.0-9.5 g/dL) of either carrier state or ID; very low MCV (<50 fl) is suggestive of ID, while MCV values of 50-70 fl of either carrier state or ID; RBC >5.0 x 10^{12} / L is suggestive of carrier state; RDW >15% is suggestive of ID. Borderline values for RBC (=5 x 10^{12} /L) and RDW (=15%) might be suggestive of both ID and carrier state. The condition which is maintained more strongly by these indices is the appropriate diagnosis. In case both conditions are maintained equally, we also assess mean corpuscular hemoglobin (MCH); MCH values of ≤22 pg are suggestive of carrier state. In addition, microcytosis without anemia or erythrocytosis with very low MCV, irrespective of Hb is interpreted as carrier state. As such, most of the carriers are detected and either parental studies and hemoglobin electrophoresis as further laboratory evaluation, or estimation of ferritin levels are ordered. However, I would like to emphasize by sharing my personal experience that there might be some confusing conditions while interpreting the red cell parameters. In those conditions, which are summarized below, thalassemia minor can be masked or some other conditions may present as heterozygous thalassemia. First, in the event that a patient with thalassemia trait has concomitant ID, both RBC and RDW are elevated, second, in the event of iron deficient individual receiving iron, both RBC and RDW are elevated. Thus, it is important to ask the patients if they are taking iron when microcytosis is detected, third, normal RBC might be present in thalassemia trait, especially in children or due to the type of mutation, and fourth, presence of high RBC in pure iron deficiency anemia has been recently shown. Therefore, it is my personal opinion that, considering those conditions are of importance before making any differential diagnostic and/or therapeutic decisions. In addition, the presence of elevated RBC and RDW in an individual with microcytosis who is not receiving iron should prompt consideration of delta-beta thalassemia as the underlying condition.

In conclusion, in countries located in the thalassemia belt, it is important to first educate the involved parties about the significance of thalassemia, then to emphasize over and over again the priority of the automated blood counts in detecting the carriers, then educate people regarding the meaning of the abnormal red cell parameters on automated blood counts. Also one has to keep in mind that there can be a number of misleading and confusing factors while interpreting the red cell parameters in order to help detect the carriers and consequently prevent thalassemia. Hence, I would like to thank Kakkar and coworkers for re-emphasizing the importance of automated blood counts in detecting thalassemia minor.

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References