Sir,

Thalassemias and hemoglobinopathies are a group of genetic disorders resulting from mutation or deletion of one of the globin genes of hemoglobin. Thalassemias are caused by the defective or absent production of one or more of the globin chains with the deficiency of a chain production leading to α thalassemia and deficiency in β globin production leading to β thalassemia. The hemoglobinopathies are those hemoglobins that are structurally abnormal due to an altered amino acid sequence of the globin chain [1].

Approximately 7% of the world’s populations are carriers of hemoglobinopathies making it as one of the major health problems globally. About 3.5% of the death in children below 5 years is due to hemoglobin disorders and over 330000 infants born with this disorders annually [2]. It is assumed that the global spread of these inheritable diseases might be due to international migration occurring in different locations of the world. One of the reasons for increasing number of hemoglobinopathy related diseases is high number of consanguineous marriages in some under-resource countries and poor public health measures to control this issue. Nepal being located in the south-eastern part of Asia is suspected to have higher number of hemoglobinopathies cases which is also supported by some of the previous studies in different districts of Nepal [2]. It is believed that these disorders occur in particularly high frequency in many of the developing countries as a result of selection by endemic malaria, where they can pose a significant public health problem [3]. So hemoglobinopathies poses a serious health concern in developing country like ours and we need a modern methodological testing like HPLC instead of primitive Hb Electrophoresis for diagnosing the cases early.

Traditionally, electrophoresis has been the method of choice for identification and quantification of variant Hb’s. But it is not possible to differentiate between Hb E and Hb O, and Hb D and Hb G using electrophoretic methods. Moreover, Electrophoresis is slow, labor-intensive, and inaccurate in the quantification of low-concentration Hb variants (e.g., Hb A2) or in the detection of fast Hb variants (Hb H, Hb Barts). HPLC is emerging as the method of choice for the initial screening of Hb variants (including neonatal screening where this is mandated) and for quantification of Hb A2 and Hb F concentrations. The Bio-Rad Variant (Bio-Rad Laboratories) is an automated cation-exchange HPLC instrument that has been used to quantify Hb A2, Hb F, Hb A, Hb S, and Hb C making it an excellent tool for diagnosing thalassemia and hemoglobinopathies, including detection of α-thalassemic genotypes in cord blood [4]. A characteristic retention time (window of detection), together with the relative quantification, permits presumptive identification of all relevant hemoglobin species by HPLC including the Hb Barts. HPLC analysis is very precise and reproducible with an inter-run coefficient of variation (CV) of 1% of total hemoglobin [5].

However, there are some disadvantages of the use of HPLC. Hb variants can falsely increase or decrease reported Hb A1c results. So Hb A1c results in patients who have an Hb variant can be clinically misleading if the variant alters the lifespan of erythrocytes [6]. Furthermore, The measurement of Hb A2 using cation-exchange HPLC is complicated in individuals with Hb S because the Hb A2 is falsely increased by the presence of Hb S adds further implying the need of capillary zone electrophoresis [4].

To conclude, Nepal though being an under-resource country is also an ethnically diverse country with marked regional variation. This diversity is reflected in the presence of different hemoglobin variants in different ethnic groups. So, there is always a need for a screening method which can detect maximum variants. HPLC has the advantage over electrophoresis for quantifying Hb F and Hb A2 along with detecting other variants in a single screening test. Moreover, HPLC is sensitive, specific, reproducible, and less time consuming and requires less man power. Hence, it is ideal for a routine clinical laboratory with high work load. However, one has to be aware of the limitations and problems associated with the diagnostic methods to avoid false negative diagnosis in everyday practice. Further, DNA analysis after presumptive identification of hemoglobinopathies and thalassemia syndromes, and particularly for purposes of genetic counseling, defining the mutation or deletion present may be required. This change in investigation procedure is necessary for early diagnosis and treatment of these disorders.

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1 Name of the registry: none
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3 Hyperlink to your specific registration (must be publicly accessible and will be checked):

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Consent

Studies on patients or volunteers require ethics committee approval and fully informed written consent which should be documented in the paper.

Authors must obtain written and signed consent to publish a case report from the patient (or, where applicable, the patient’s guardian or next of kin) prior to submission. We ask Authors to confirm as part of the submission process that such consent has been obtained, and the manuscript must include a statement to this effect in a consent section at the end of the manuscript, as follows: “Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request”.

Patients have a right to privacy. Patients’ and volunteers’ names, initials, or hospital numbers should not be used. Images of patients or volunteers should not be used unless the information is essential for scientific purposes and explicit permission has been given as part of the consent. If such consent is made subject to any conditions, the Editor in Chief must be made aware of all such conditions.

Even where consent has been given, identifying details should be omitted if they are not essential. If identifying characteristics are altered to protect anonymity, such as in genetic pedigrees, authors should provide assurance that alterations do not distort scientific meaning and editors should so note.

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