DOI: 10.7860/JCDR/2016/21099.8149

Letter to Editor

Internal Medicine Section

Delta-Beta Thalassaemia Screening

BEUY JOOB[1], VIROJ WIWANITKIT[2]

Keywords: Fetal haemoglobin, Haematological finding, Hypochromic microcytic, Thalassaemia intermedia

Dear Editor,

We read the recent report by Kumar BV et al., with great interest [1]. They have reported an interesting case of elevated haemoglobin F and mentioned it as a possible diagnosis of heterozygous delta-beta–thalassaemia [1]. In fact, the diagnosis of delta-beta thalassaemia can be difficult. In the case of delta-beta thalassaemia, the clinical appearance is usually of thalassaemia intermedia with only abnormal haematological findings and hypochromic microcytic anaemia [2]. Based on a study from Brazil, the incidence of these abnormalities is less than 1:10,000 [3]. However, in a report from Thailand [4], the prevalence of carrier of this disorder is as high as 4%. The main haematological finding of delta beta thalassaemia is “absent or reduced synthesis of adult haemoglobin (HbA) and increased synthesis of fetal haemoglobin” [5]. Nevertheless, this finding can also be seen in another haematological disorder namely hereditary persistence of fetal haemoglobin. To differentiate delta beta thalassaemia from hereditary persistence of fetal haemoglobin it requires good laboratory testing. The main difference between hereditary persistence of fetal haemoglobin and delta beta-thalassaemia can be confirmed by alpha-beta-globin chain synthesis ratio and DNA analysis. However, this is not easy for simple screening [5]. The use of haemoglobin electrophoresis might be useful for detection of fetal haemoglobin and applied in screening. However, the cost for performing haemoglobin electrophoresis is not cheap. In our setting, Thailand, where thalassaemia is very common, the screening is proposed as national policies. The use of simple screening such as DCIP (Dichlorophenol-indophenol) and Osmotic Fragility (OF) test is implemented for screening of common disorders such as alpha thalassaemia, beta thalassaemia and haemoglobin E disorder [6]. For the delta beta thalassaemia, the combination between DCIP and OF tests might help to screen but not definitively diagnose the case [6]. According to the local reports from Thailand, the screening test had a good sensitivity. The sensitivity of the technique to detect the cases with abnormal high haemoglobin F level including delta beta thalassaemia is 100% [7,8]. However, the positive predictive value is extremely low (about 0.02% –1%) due to the fact that the test can also help screen other kinds of more common haemoglobin disorders [6]. The definitive diagnosis for any test positive case usually requires “molecular characterization and PCR detection” [2]. For a case of “hereditary persistence of fetal haemoglobin”, a more complex situation “gamma delta beta” or “zero thalassaemia is also to be ruled out [9]. The validation and tests on possible usefulness of screening tests for the cases with abnormal high fetal haemoglobin levels in different settings should be done. Finally, for any case with final confirmation for abnormal fetal haemoglobin level, family studies on both parents must be done.

REFERENCES

[1] Kumar BV, Choccalingam C, Samuel P. Incidental identification of possible delta-beta thalassaemia trait in a family: a rare cause of elevated Hb F. *J Clin Diagn Res*. 2016;10(3):BD01-02.
[2] Trachoo O, Sura T, Sakuntabhai A, Singhasivanon P, Krudsood S, Phimpraphi W, et al. Molecular characterization of hereditary persistence of fetal haemoglobin in the Karen people of Thailand. *Haemoglobin*. 2003;27(2):97-104.
[3] Zago MA, Costa FF, Tone LG, Bottura C. Hereditary haemoglobin disorders in a Brazilian population. *Hum Hered*. 1983;33(2):125-29.
[4] Panyasai S, Fucharoen S, Surapot S, Fucharoen G, Sanchaisuriya K. Molecular basis and haematologic characterization of delta beta thalassaemia and hereditary persistence of fetal haemoglobin in Thailand. *Haematologica*. 2004;89:777-81.
[5] Bollekens JA, Forget BG. Delta beta thalassaemia and hereditary persistence of fetal haemoglobin. *Haematol Oncol Clin North Am*. 1991;5(3):399-422.
[6] Joob B, Wiwanitkit V. Genetic counseling for thalassaemia screening in resource limited setting: a case study. *Clin Lab*. 2014;60(8):1421.
[7] Panyasai S, KeowkarnkahT, Jattawan N. The detection of high HbF determinants at Lampang Hospital Center, Lampang Province: Preliminary report. *Bull Dept Med Sci*. 2008;50(3):207-17 (in Thai).
[8] Panyasai S. The prevalence of Sβ-thalassaemia and hereditary persistence of fetal haemoglobin (HPFH) among population in the South of Thailand. *J Med Tech Assoc Thai*. 2006;34:1729-40 (in Thai).
[9] Winichagoon P, Fucharoen S, Thonglairoam V, Wasi P. Thai G gamma (A gamma delta beta) zero-thalassaemia and its interaction with a single gamma-globin gene on a chromosome carrying beta zero-thalassaemia. *Haemoglobin*. 1990;14(2):185-97.

PARTICULARS OF CONTRIBUTORS:
1. Medical Center Sanitation 1 Medical Academic Center, Bangkok, Thailand.
2. Professor, Department of Tropical Medicine, Hainan Medical University, China.

NAME, ADDRESS, E-MAIL ID OF THE CORRESPONDING AUTHOR:
Dr. Beuy Joob,
Sanitation 1 Medical Academic Center, Bangkok, Thailand.
E-mail: beuyjoob@hotmail.com

FINANCIAL OR OTHER COMPETING INTERESTS: None.

Date of Submission: **May 03, 2016**
Date of Peer Review: **May 26, 2016**
Date of Acceptance: **Jun 11, 2016**
Date of Publishing: **Jul 01, 2016**