Delta beta thalassemia: a rare hemoglobin variant

TO THE EDITOR: Delta beta (δβ)-thalassemia results from a deletion in both the delta and beta genes on chromosome 11. The gamma genes on the affected chromosome increase their production of gamma globin, thereby increasing the amount of hemoglobin F (HbF). δβ-Thalassemia heterozygotes clinically show characteristics of thalassemia minor. However, homozygous δβ-thalassemia may give a clinical picture of thalassemia intermedia with a mild anemia.

A 12-month-old boy presented to the hematology outpatient department for evaluation of pallor and jaundice that had been for the past 2 months. He had no history of a blood transfusion. His family history was insignificant and, because of the increased synthesis of HbF, may have thalassemia intermedia rather than thalassemia major [1].

However, the phenotype of heterozygotes resembles that of the β-thalassemia trait, but the HbA2 percentage is not increased and is often normal. HbF in such individuals is consistently elevated, varying from 5% to 20%. Peripheral blood film findings are similar to those for the β-thalassemia trait, and the distribution of HbF is heterocellular, which is best observed via flow cytometry. It is necessary to distinguish it from hereditary persistence of fetal hemoglobin (HPFH). The two groups of disorders are distinguished by the phenotype of heterozygous individuals. Heterozygotes of δβ-thalassemia mutations have 5% to 20% HbF, which is heterocelluarly distributed in red cells, whereas heterozygotes of HPFH mutations have 17% to 30% HbF, with a pancellular distribution. In addition, homozygotes of HPFH are asymptomatic, whereas δβ-thalassemic homozygotes have thalassemia intermedia-like features [2].

At least nine mutations can result in δβ-thalassemia. This type of thalassemia is observed in many ethnic groups, including some Mediterranean populations (Italians, Greeks, and Turks). Although the exact diagnosis of δβ-thalassemia requires genetic analysis for mutations, Hb electrophoresis or HPLC findings of markedly elevated HbF may be suggestive. An extensive PubMed search was done to determine the incidence of δβ-thalassemia in different parts of the world, but owing to the rarity of this Hb variant,

| Table 1. Laboratory parameters of the case and parents. |
|--------------------------------------------------------|
| Case | Mother | Father |
| CBC (g/dL) | 8.0 | 12.6 | 13.8 |
| Mean corpuscular volume (fl) | 76.4 | 76.0 | 73.0 |
| Mean corpuscular Hb (pg) | 23.4 | 25.4 | 23.5 |
| HPLC (g/dL) | 8.0 | 12.6 | 13.8 |
| HbA (%) | 0 | 79.8 | 81.7 |
| HbA2 (%) | 0 | 2.2 | 2.3 |
| HbF (%) | 100 | 18 | 16 |

Abbreviations: CBC, complete blood count; Hb, hemoglobin; HPLC, high-performance liquid chromatography.
only a handful of case reports were identified from across the world [3-6].

Huma Mansoori, Sidra Asad, Anila Rashid, Farheen Karim

Department of Haematology, Aga Khan University Hospital, Karachi, Pakistan

Correspondence to: Huma Mansoori
Department of Haematology, Aga Khan University Hospital, Stadium Road P. O. Box 3500 Karachi 74800, Pakistan
E-mail: huma.omair1986@gmail.com

Received on Jul. 23, 2015; Revised on Nov. 26, 2015; Accepted on Jan. 10, 2016
http://dx.doi.org/10.5045/br.2016.51.3.213

Authors’ Disclosures of Potential Conflicts of Interest
No potential conflicts of interest relevant to this article were reported.

REFERENCES
1. Bain BJ. Haemoglobinopathy diagnosis. 2nd ed. Oxford, UK: Blackwell Publishing Ltd, 2006:116-24.
2. Bollekens JA, Forget BG. Delta beta thalassemia and hereditary persistence of fetal hemoglobin. Hematol Oncol Clin North Am 1991;5:399-422.
3. Khunger JM, Gupta M, Singh R, Kapoor R, Pandey HR. Haematological characterisation and molecular basis of asian Indian inversion deletions delta Beta thalassemia: a case report. J Clin Diagn Res 2014;8:FD01-2.
4. Verma S, Bhargava M, Mittal S, Gupta R. Homozygous delta-beta thalassemia in a child: a rare case of elevated fetal hemoglobin. Iran J Ped Hematol Oncol 2013;3:222-7.
5. Ramot B, Ben-Bassat I, Gafni D, Zaanoon R. A family with three beta-delta-thalassemia homozygotes. Blood 1970;35:158-65.
6. Silvestroni E, Bianco I, Reitano G. Three cases of homozygous beta, delta-thalassemia (or microcythaemia) with high haemoglobin F in a Sicilian family. Acta Haematol 1968;40:220-9.