

Alpha Thalassaemia: Experience of Referral Cases in Kolkata, India

Tapash Rudra, Sila Chakrabarti and Bani Sengupta

*Thalassaemia Counselling Unit, Vivekananda Institute of Medical Sciences, 99 Sarat Bose
Road, Kolkata 700 026, West Bengal, India*

KEYWORDS Haemoglobinopathies; Thalassaemia; anaemia, mutation

ABSTRACT Haemoglobinopathies and different forms of thalassaemias including alpha thalassaemia has been found to be as high as 10% in Eastern India. The alpha globin disorders are less commonly reported because the diagnosis of alpha thalassaemia is usually missed unless in the severe homozygous form or as Hb Barts. But presence of alpha gene has been found in cases of unexplained anaemias and also in several suspected cases having α mutations and other common haemoglobinopathies. Two of the common α mutations ($-\alpha^{3.7}$, $-\alpha^{4.2}$) have been detected either in heterozygous or homozygous states in significant frequencies, so patients coming for Hb Electrophoresis and carrier detection should be also checked the alpha status through specific DNA studies.