Editorial

Micro mapping the frequencies of beta thalassemia and sickle cell anemia in India: A way forward to plan control strategies

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Abnormalities of hemoglobin (Hb) synthesis are the most common inherited disorders in human and results from *quantitative* reduction of globin chain synthesis (thalassemia syndromes) or *qualitative* defect in the globins (hemoglobinopathies). Sickle cell anemia (SCA) is the commonest symptomatic hemoglobinopathy found in India and the frequency of sickle cell trait (SCT) is 4.3% based on 308 populations studied. The average frequency of beta thalassemia trait (BTT) is 3-4%. Since the inheritance is autosomal recessive in both the conditions, the asymptomatic carriers or traits constitute the reservoir of the disease. The burden of these inherited disorders is not uniform throughout the country. SCT is more common in the central belt of India covering states of Maharashtra, Madhya Pradesh, Chhattisgarh and Orissa with some spillover in the adjacent areas. Some communities and castes have higher frequency of both these disorders than others. India has many geographical, ethnic, religious and language divisions. As the peoples of India have traditionally married and reproduced within these sub-divisions, major problems are encountered in estimating the impact of genetic disease at national, regional, state or even local levels. The reason is due to the majority of the marriages being arranged within the castes and certain communities such as the tribal populations are strictly endogamous.[1-3]

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In this issue of the *Journal*, Bhukhanvala *et al*., report the prevalence of BTT and SCA in four communities of Surat city. Their finding of different prevalence rates of both BTT and SCT in the four different communities reiterates the fact that it is important to perform micro mapping so that when resources are limited it will be possible to implement screening programs on communities at higher risk. This data will also help to accurately estimate the expected annual births of children with beta thalassemia and SCA. A total of 9,447 individuals were studied systematically in the four communities which included Muslim, Dhodia Patel, Kacchiya Patel, and Modh Bania. Of them the only tribal community was Dhodia Patels and they showed the highest prevalence of SCT of 14%. Also 80% of the Dhodia Patels showed an MCV of < 76 fL and 80.3% showed the MCH of < 26 pg which are the cutoff of values used in community screening programs. Though not included in this study, based on the data of automated blood cell counts it is expected many of the individuals would have iron deficiency anemia or iron deficient stores. The study detected the highest prevalence of BTT in the Modh Bania community. The data of the diagnostic tests in the four communities has been compared with 24,917 randomly selected unrelated individuals who are in included all major castes from the blood donation camps. This comparison would not be correct since they

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strictly would not constitute a "control" group based on non-segregation of castes and communities.[4]

However, it is important to realize that values of automated blood cell counters is useful for screening purposes and identifying BTT with reasonable certainty, the same does not hold true while screening for hemoglobinopathies such as SCT/ SCA. Detection of SCT requires showing the presence of both adult Hb (HbA) and HbS in appropriate amounts. Tests available from whole blood and or hemolysate include sickling test, Hb S solubility test, Hb electrophoresis at alkaline and acid pH, HPLC, and isoelectric focussing. DNA based methods can detect the defect either at the heterozygous or homozygous state. Therefore any screening program which includes detecting hemoglobinopathies such as SCA needs to incorporate a method which detects the abnormal protein product or gene. At present some of the centres in India have initiated newborn screening programs for SCA.[5]

The uneven distribution of the frequencies of BTT and SCT in the various studies on different districts and states is expected based on the caste and community structure and marriage practices in India. Studies such as by Bhukhanvala *et al.*, will help to understand the heterogeneity.

References

1. Rao VR, Gorakshakar AC. Sickle cell hemoglobin, β-thalassemia and G6PD deficiency in tribes of Maharashtra, India. Gene Geogr 1990;4:131-4.
2. Bittles AH. Endogamy, consanguinity and community genetics. J Genet 2002;81:91-8.
3. Gupta RB. Sickle cell disease in Central India– Need for Micro level Planning. In Tribal Health: Proc. National Symp. Oct 2006 RMRC (ICMR) Jabalpur; 2006. p. 109-15.
4. Bhukhanvala DS, Sorathiya SM, Shah AP, Patel AG, Gupte SC. Prevalence and hematological profile of β-thalassemia and sickle cell anemia in four communities of Surat city. Indian J Hum Genet 2012;18:167-71.
5. Jain DL, Sarathi V, Upadhye D, Gulhane R, Nadkarni AH, Ghosh K, *et al*. Newborn screening shows a high incidence of sickle cell anemia in central India. Hemoglobin 2012;36:316-22.