Short communication

α-thalassaemia (Hb-Bart’s) in Rajasthan (India)

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Abstract—A total of 1647 cord blood samples (618 of scheduled tribes, 487 of scheduled castes and 542 of general castes) of newborns from the Banswara, Bhilwara, Chittorgarh, Dungarpur, Sirohi and Udaipur districts of the Aravali hilly region of Rajasthan were investigated electrophoretically for evidence of Hb-Bart’s (α-thalassaemia). Hb-Bart’s was encountered in 31 (1.88%) neonates constituting 1.8 to 12.6% of their total haemoglobin. Of these, 24 (1.46%) were of α-thalassaemia 1 and 7 (0.42%) of α-thalassaemia 2. The incidence of α-thalassaemia genes varied from 3.07% in the scheduled tribes, 1.43% in the scheduled castes to 0.77% in the general castes populations giving an overall incidence of 1.88%. Except Hb-Bart’s allele, no other mutant haemoglobins were observed.

Key words: Cord blood; Hb-Bart’s; α-thalassaemia; Rajasthan.

INTRODUCTION

It is well known that in α-thalassaemia there is defective synthesis of the α-chains of haemoglobin (Hb) and consequently production of Hb-A, A2 and F is decreased. In newborn infants, the production of a defective α-chain is indicated by the presence of an abnormal haemoglobin, Hb-Bart’s which is a tetramer of γ-chains. At least two genes are involved for the two forms of the α-thalassaemia carrier state [1], α-thalassaemia 1 (the severe type) and α-thalassaemia 2 (the mild type). Both variants contain varying concentration of Hb-Bart’s [2, 3]. Both genes are responsible for the genesis of three known syndromes of α-thalassaemia; (a) heterozygous α-thalassaemia (α-thalassaemia minor), (b) homozygous α-thalassaemia (the Hb-Bart’s hydrops fetalis syndrome) and (c) double heterozygous state for the α1 and α2 thalassaemia genes which is also known as Hb-H disease.

In India, very limited information is available on the genetics and incidence of α-thalassaemia in different populations. In Eastern and Western India, reports on Hb-H disease indicate the presence of a-thalassaemia genes [4, 5]. The incidence of Hb-Bart’s (α-thalassaemia) in cord blood samples of mixed Indian populations is about 1% [6–8]. However, a very high incidence (23–81%) has also been recorded.
[9–11] in certain tribal populations. From different geographical provinces of India, several sporadic cases of α-thalassaemia have been reported [12–14]. But population studies for the incidence of Hb-Bart’s in cord blood are rare, a few such studies are available only in Bengalees [15, 16], Gujaratis [9], Maharashtrains [10] and Punjabis [12–17]. In the state of Rajasthan, with the exception of the Dungarpur district [18], no extensive survey on α-thalassaemia in scheduled tribes (S.T.), scheduled castes (S.C.) and general castes (G.C.) populations have been conducted.

Therefore, the present preliminary survey was undertaken to observe the incidence of α-thalassaemia genes (α-thalassaemia 1 and α-thalassaemia 2) in three major S.T., S.C. and G.C. populations residing in hilly areas of the Aravali region of Rajasthan where sickle cell and β-thalassaemic genes are also prevalent [14].

MATERIAL AND METHODS

The study was conducted in the State of Rajasthan where the total population is 44,005,990 containing 12.44% (5,474,881) scheduled tribe, 17.33% (7,627,820) scheduled caste and 70.27% (30,923,289) general caste populations.

The subjects of a tribal population are mostly illiterate, economically very poor and reside in isolated hilly areas. Their physiognomy is characterised by short stature, dark skin, coarse black hair and a flat nose. Their staple diet is maize and grain and they live mainly by agricultural labour. The subjects of the scheduled caste population showed some resemblance to tribal subjects. They are also poor and illiterate but reside in both rural and urban areas. They live mainly on agriculture and their occupations involves leather processing. The general caste population is dominant in urban areas and its subjects are mostly literate, relatively affluent and are involved in the business sector and similar occupations.

The study was based on 1647 cord blood samples (618 from S.T., 487 from S.C. and 542 from G.C. populations) which were collected (from 5th January, 1995) in EDTA vials from the Departments of General Hospitals and from Primary Health Centres of Banswara, Bhilwara, Chittorgarh, Dungarpur, Sirohi and Udaipur districts of the Aravali hilly region of Rajasthan. Simultaneously, peripheral blood smears were also prepared and stained by Giemsa stain for detection of morphological red cell abnormalities (anisocytosis, poikilocytosis, microcytosis and target cells). Horizontal paper and cellulose acetate-electrophoreses were performed for the identification and quantitative evaluation of the fast-moving haemoglobin, Hb-Bart’s. An osmotic fragility test was also done on cord blood and in peripheral blood samples from infants having Hb-Bart’s.

RESULTS

Electrophoretic examination of the 1647 cord blood samples of the newborn infants belonging to the sample group resulted in 31 (1.88%) showing the presence of the