1. GENETIC DISORDERS

1.1 Prevalence of alpha thalassaemia among the SC & ST population of Nimar area of Madhya Pradesh

The study is being carried out in Nimar area comprising East Nimar, West Nimar and Badwani districts. About 3 ml of blood sample was drawn in EDTA vial from apparently healthy individuals of various castes/ tribes, after obtaining consent. Family was taken as a sampling unit. Complete blood count was performed using automatic blood cell counter. Common deletional form of α-thalassaemia type II i.e. 4.2 kb and 3.7kb deletion were typed by PCR.

One hundred samples from each population group are targeted to be studied. The samples analysed till date indicate that Korku, Bhil and Barela tribes have high prevalence of α-thalassaemia II. The gene frequency of α gene was higher in Korku (0.33), Bhil (0.35), Barela (0.28), while it was lower in Bhilala (0.14) and Balai - a Scheduled tribe (0.17). The Bhil, Bhilala and Korku tribes have lower prevalence of HbS as compared to Barela tribe and have higher prevalence of α-thalassaemia type II than Barela. The study is in progress.



Map of Madhya Pradesh Showing the study area

1.2 Morbidity profile of sickle cell disease in central India

The centre is involved in the study on morbidity profile of sickle cell disease (SCD) patents attending sickle cell clinic run by RMRCT at NSCB Medical College, Jabalpur. Three hundred and forty three SCD patients participated in the study. Common symptoms for which patient sought medical intervention are painful crises of bones and joints with fever. The patients were given folic acid, vitamin B-complex and antipyretic/anti-inflammatory tablets. Treatment was also given for other common ailments. Genetic counseling was given to the patients or their guardians. This included education regarding avoiding the precipitating factors.

There was a marked reduction in the clinical severity of the disease after intervention. The CBC profile of sickle cell disease patients is given in Table 1.2.1. The severity of disease was assessed by converting the clinical observations into the numerical score.

A reduction in the severity of the disease was observed in the patients after intervention for a period of 2 to 3 years. Deletional form of α-thalassaemia type II was studied in 71 SCD patients. Nine SCD persons were homozygous for -thalassaemia type II and 17 were heterozygotes. The main form of deletion was- $\alpha^{3.7}$ kb deletion. The haematological and clinical profile of these patients will be correlated with the number of functional α -globin genes.

Table 1.2.1: Haematological parameters in sickle cell disease patients

Group	N	Hb (g/dl)	PCV (l/l)	TRBC $10X^{12}/L$	MCV (fl)	MCH (pg)	MCHC g/dl	HbF (%)	HbA_2 (%)
Male	91	8.1 ± 2.5	0.24 ±0.07	3.2 ± 1.0	78.8 ± 12.8	26.6 ± 5.5	33.7 ± 3.5	12.8 ± 5.7	2.8 ± 1.4
Female	40	7.9 ± 2.2	0.23 ±0.06	2.9 ± 0.8	79.9 ± 10.7	27.0 ± 5.0	33.6 ± 3.7	14.1 ± 5.8	2.3 ± 1.0
Children	212	7.3 ±1.9	0.22 ±0.06	2.9 ± 1.0	78.1 ± 13.5	26.2 ± 5.1	33.4 ± 4.8	12.8 ± 5.2	2.5 ± 1.0



A child having splenomegaly

1.3 Community control programme of haemoglobinopathies

This is a collaborative study between RMRCT and Department of Obstetrics and Gynaecology, NSCB Medical College Jabalpur. A total of 321 pregnant women from antenatal clinic were screened for common haemoglobinopathic disorders. Majority of the women (58%) were in third trimester and about one third were in second trimester. Twenty women (6.2%) were having sickle cell trait and 2.2% -thalassaemia trait. They were counselled for risk of pregnancy for haemoglobinopathies and advised for screening of their husband for haemoglobinopathies. Overall anaemia (Hb <11 g/dl) was rampant (71.6%) in these women. About 7% of women were severely anaemic (Hb <7g/dl) and 22.4% moderately anaemic. Iron deficiency was detected by estimating free erythrocyte protoprophyrin level in the blood. About 24% of women had iron deficiency.



An anaemic lady