



## 1. GENETIC DISORDERS

### 1.1 Prevalence of deletional form of alpha thalassaemia type in Scheduled Caste and Scheduled Tribes of Nimar area of Madhya Pradesh

Haemoglobinopathies in the form of sickle haemoglobin and  $\alpha$ -thalassaemia is very common in Nimar area with a prevalence of 18.5% and 1.6% respectively in the Scheduled tribe and Scheduled caste population. The co-existence of  $\alpha$ -thalassaemia type II in sickle cell disease and  $\alpha$ -thalassaemia is stated to reduce the disease severity.  $\alpha$ -thalassaemia is also stated to provide protection against severe form of malaria. The status of  $\alpha$ -thalassaemia is not known for Nimar area. The typing of the deletional form of  $\alpha$ -thalassaemia type II was done by PCR using allele specific primers.

In Nimar area  $\alpha$ -<sup>3.7</sup> deletional allele dominate the  $\alpha$ <sup>+</sup>-thalassaemia type II contributing about 93% of allele. Bhil showed the highest prevalence of  $\alpha$ <sup>+</sup>-thalassaemia i.e. 68% had at least one  $\alpha$ -gene deleted. Surprisingly, the Bhil group or tribes i.e. Bhil, Bhilala and Barela did not have  $\alpha$ -<sup>4.2</sup> allele but it was found in Korku tribe. Least prevalence of  $\alpha$ <sup>+</sup>-thalassaemia type II was in Balai Scheduled caste (Table 1.1.1).

**Table 1.1.1 : Prevalence of deletional form of  $\alpha$ <sup>+</sup>-thalassaemia in Scheduled Caste and Scheduled Tribes of Nimar area of Madhya Pradesh**

Population	N	/	- /	- /-
Korku	26	6	10	10
Bhil	50	11	10	29
Barela	91	33	9	49
Bhilala	71	43	9	19
Balai	79	51	18	10
Total	317	144	56	117

\*-thalassaemia type II did show the typical mild microcytic, hypochromic mild anaemic effect on RBC in its homozygous state. As expected it also lowered the HbA<sub>2</sub> and HbF level slightly (Table 1.1.2). These differences were statistically insignificant.

**Table 1.1.2 : CBC profile and alpha thalassaemia in Nimar area of Madhya Pradesh**

No. of -gene.	N	Hb	PCV	TRBC	MCV	MCH	MCHC	HbF	HbA <sub>2</sub>
/	144	11.6 1.9	31.2 5.5	4.3 0.7	73.1 10.9	27.4 4.5	37.4 2.5	1.2 1.0	2.4 0.7
- /	56	12.0 1.6	33.9 5.1	4.5 0.6	75.4 8.1	26.7 3.6	35.5 3.0	1.1 0.3	2.4 0.8
- /-	117	10.8 1.8	29.6 4.6	4.6 0.7	65.1 7.4	23.7 3.4	36.3 2.8	1.1 0.3	2.1 0.7



## 1.2 Prevalence of common haemoglobinopathies and G6PD deficiency in Scheduled caste and Scheduled tribes of district Damoh of Madhya Pradesh

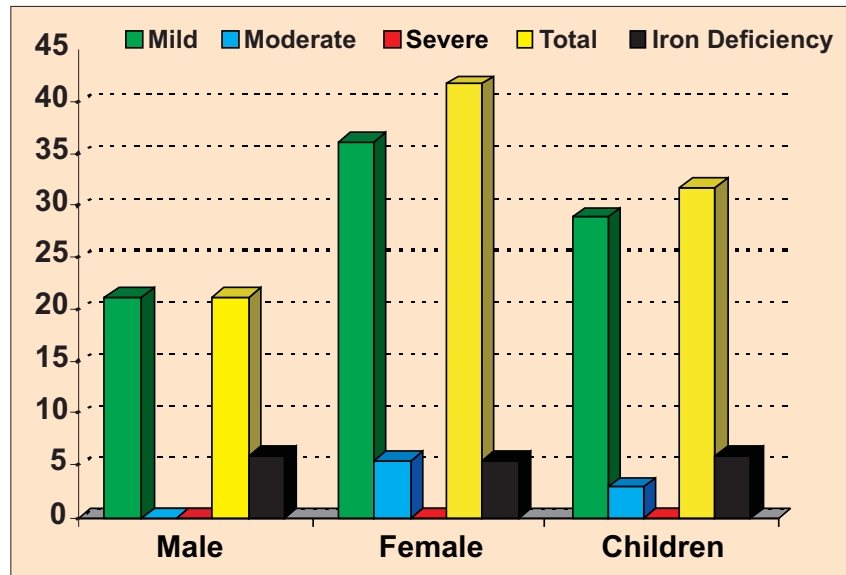
It is a part of ongoing activity of the centre to map the prevalence of common haemoglobinopathies in various Scheduled castes and Scheduled tribes populations at micro level. The total population of district Damoh (Census 2001) is about 10.8 lacs and the Scheduled tribe and Scheduled caste population is 12.6% and 19.5% respectively. The main tribal population is Gond and the main Scheduled caste is Chaudhary (Chamar). Both these populations practice strict non-consanguineous caste/ tribe endogamy but gotra exogamy marriage practices. The study population was drawn from two Tehsils i.e. Jabera and Tendukheda which has high concentration of Scheduled tribes. Gond and Chaudhary dominated villages were selected randomly from the blocks of Jabera, Sangrampur, Nahata, Tendukheda, Taradehi. The blood sample are being analysed for common haemoglobinopathies. The data generated so far suggest that prevalence of G6PD deficiency is higher among Gonds (9.3%) and it was 1.8% in Chaudhary (Table 1.2.1).

**Table 1.2.1 Prevalence of G-6-PD deficiency in Damoh district**

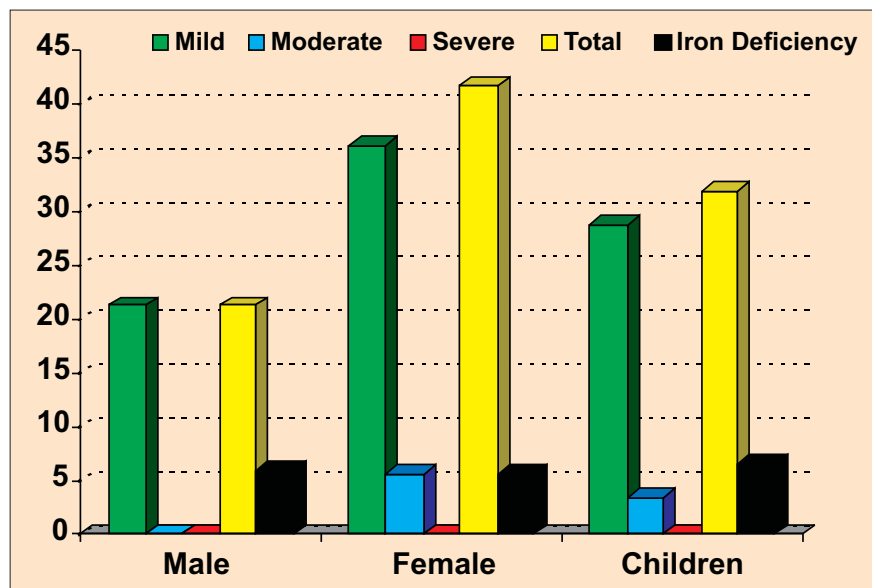
Population	N	G6PD deficiency
Gonds	321	30 (9.3)
Chaudhary	339	6 (1.8)

Anaemia was more common in Gond tribe (71.9%) as compared to Chaudhary (31.8%). Most of these anaemic cases were of mild category. Females and children (<12 years) were more prone to anaemia as compared to males in both the population groups (Fig. 1.2.1 & 1.2.2). Prevalence of iron deficiency, as judged by estimation of free erythrocyte protoporphyrin level, was less in comparison to prevalence of anemia. All severe anaemic and about half of moderate anaemic persons were having deficiency of iron. Anaemia in the study area is caused by many other factors like common infections, worm infestation and  $\alpha$ -thalassaemia type II etc. The analysis of samples for sickle haemoglobin,  $\alpha$ -thalassaemia and  $\beta$ -thalassaemia type II is in progress.

**Fig 1.2.1 Percent prevalence of anaemia and iron deficiency in Gond tribe of district Damoh, M.P.**



**Fig 1.2.2 Percent prevalence of anaemia and iron deficiency in Chaudhary Scheduled caste of district Damoh, M.P.**

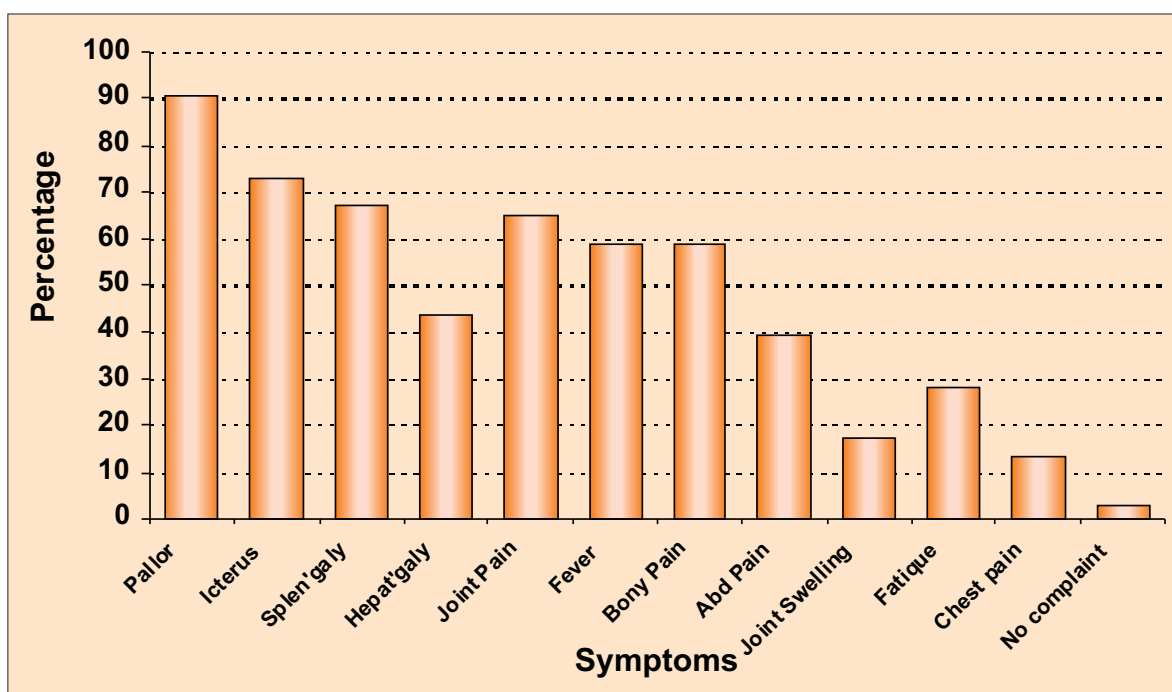


### 1.3 Morbidity profile of sickle cell disease in Central India

In the year 2006-2007, 41 new patients of sickle cell disease (SCD) were enrolled in the clinic making a total of 395 patients. Till date we have followed the 42 SCD patients for 3 years and 31 patients for 4 years and 243 patients for more than a year. These patients were evaluated clinically as per structured proforma after obtaining written consent.

The common signs and symptoms of sickle cell disease in 41 patients are given in Fig. 1.3.1. About three-fourth of these patients are below 15 years and belong to Scheduled castes, tribes and OBCs groups. Beside clinical anaemia, splenomegaly, joint and bony pain with fever is the most common symptoms. About 3% of the patient did not reveal any symptom of typical sickle cell disease other than clinical anaemia. Most of the patients with abdominal pain reported pain at splenic site.

Fig 1.3.1 Common signs and symptoms in sickle cell disease



Patients/ and their parents were advised to avoid disease precipitating/ or aggravating factors like exposure to extreme climate, excessive exercise, dehydration etc. They were given 5 mg of folic acid daily and anti-pyretic and anti-inflammatory drugs on SOS basis. They were told to seek appropriate medical intervention quickly upon any minor ailment. Patients were advised to take enough water/ fluids. They were asked to re-visit for clinical examination every three months. Patients were given symptomatic treatment as outdoor patients and were referred to respective clinic in case of emergency. Most of the patients responded well with simple antipyretic and anti-inflammatory drugs during ordinary painful crisis at home. There was marked reduction ( $p < 0.001$ ) in clinical severity of the disease by providing simple intervention (Fig. 1.3.2).

**Fig. 1.3.2: Effect of intervention on sickle cell disease**

