

# Surveys of Human Genetic Markers in Malaria Endemic Areas

Human blood polymorphic systems are important biochemical markers in anthropological surveys especially in relation to disease distribution. G-6-PD deficiency and certain haemoglobinopathies are known to confer a selective advantage to the subjects against falciparum malaria. However, certain antimalarials such as primaquine and other 8-aminoquinolines increase the oxidant stress in G-6-PD deficient individuals resulting in haemolytic crisis which can be fatal if not checked in time. Therefore, information on the frequency and distribution of these variants would help in the administration of proper drugs.

## Haemoglobinopathies and G-6-PD Deficiency

Studies carried out by us on mapping of these disorders in various tribal/non-tribal groups living in malarious areas of the country have shown variable frequencies of G-6-PD deficiency (0–17.6%) among tribals of Andhra Pradesh, Assam, Gujarat, Madhya Pradesh, Orissa, Uttar Pradesh and Uttarakhand. Similarly frequencies for sickle-cell haemoglobin (HbAS) ranged from 0 to 18.9% in various tribal groups and HbE (16.5%) was observed only among tribals of Assam. Fig. 101 shows the areas from where population samples have been screened and frequencies of G-6-PD deficiency, carriers of sickle-

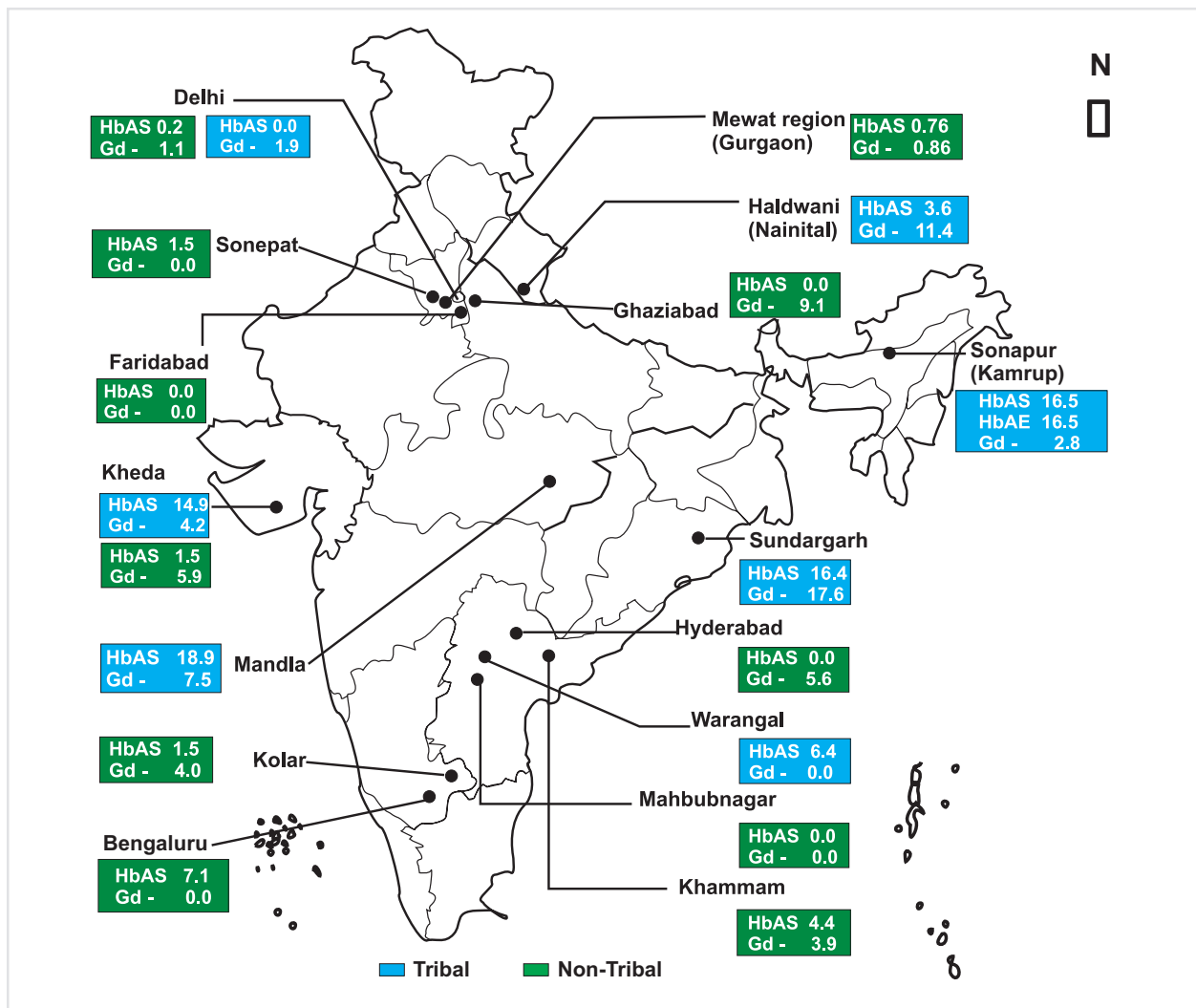


Fig. 101: Surveys of human genetic markers: Frequencies of G-6-PD deficiency (Gd) and haemoglobin (Hb) variants

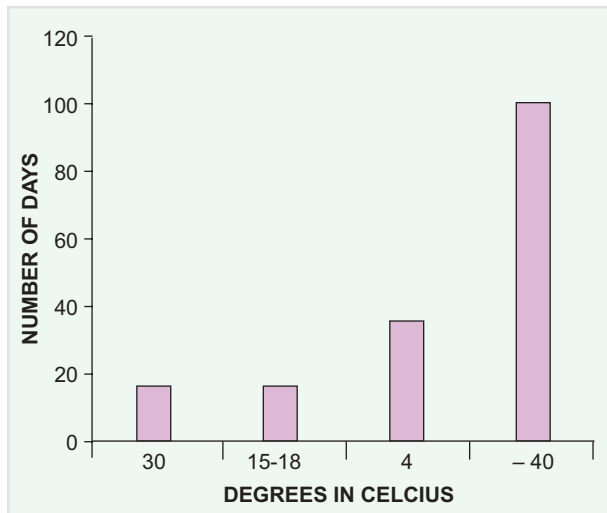


Fig. 102: Stability of the kit stored at different temperatures

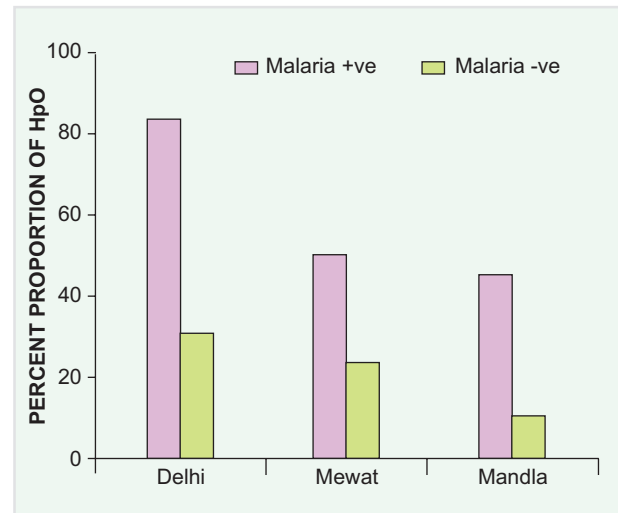


Fig. 103: HpO proportions among malaria positive and malaria negative subjects

cell (HbAS) and haemoglobin E (HbAE) (Joshi *et al* 1985, 1987, 1991, 1998, 1999, 2001). Among non-tribals, G-6-PD deficiency and abnormal haemoglobins occurred in less than 1% of the population with a few exceptions. High incidence of genetic disorders among the tribal groups suggests probable selective role of these genes in the

population in highly malarious areas.

### A Simple Kit for the Detection of G-6-PD Deficiency

Keeping in view the importance of detecting G-6-PD deficiency in malaria chemotherapy, a simple kit was developed based on the principle of

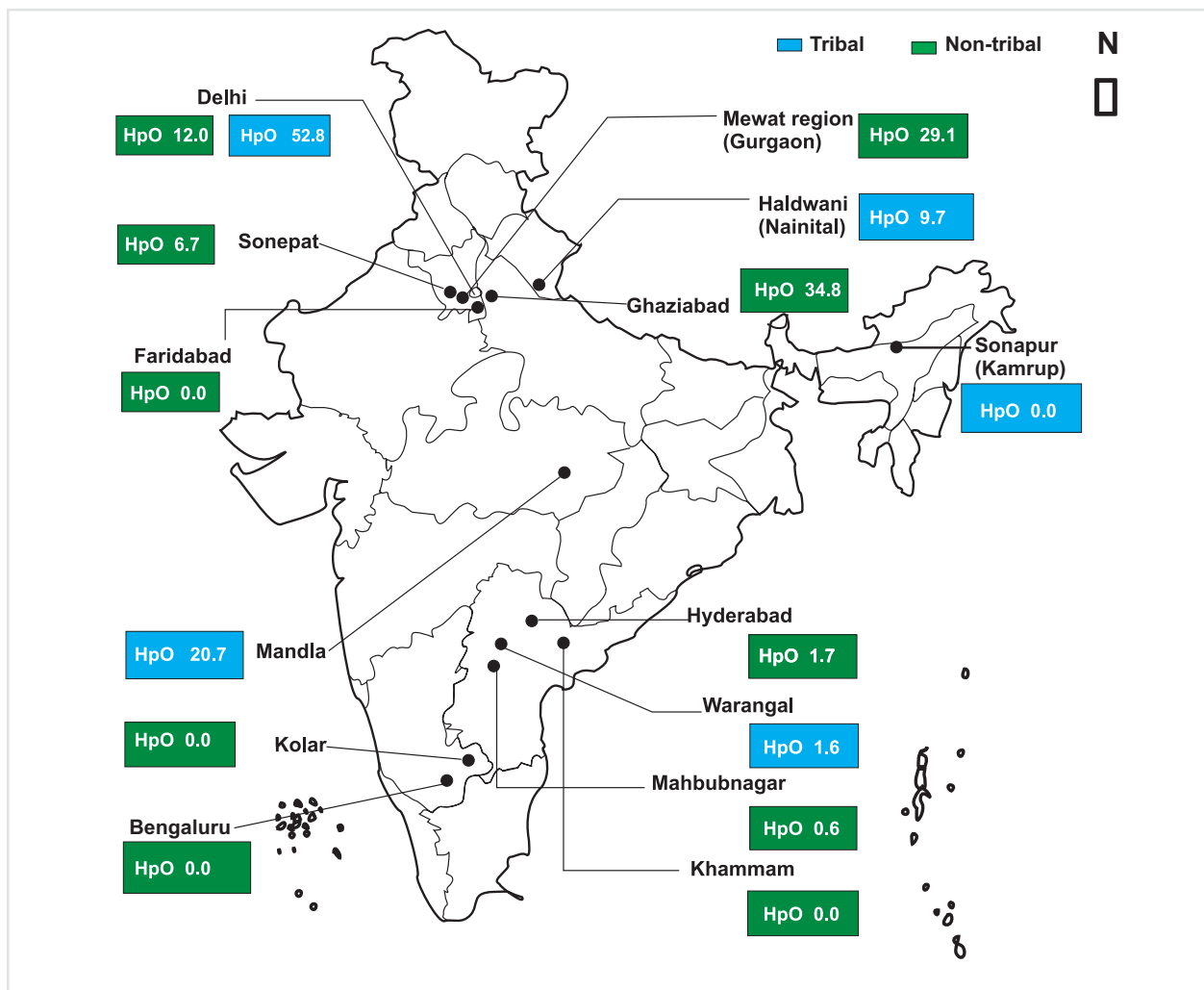


Fig. 104: Surveys of human genetic markers: Frequencies of HpO (ahaptoglobinemia)

fluorescent spot method (Schmidt and Brosions 1978, US Deptt. HEW Pub No. (CDC) 78-8266, p. 77). The kit has been compared with the standard fluorescent spot and electrophoretic method using blood samples collected from Delhi and Sonapur, Assam. This kit has given satisfactory results till 2 weeks (16 days) under field conditions (30°C). Fig. 102 shows the results of evaluation of the kit at different storage conditions. Now the kit is being evaluated at many of the field units of NIMR to test its feasibility under field conditions.

### Ahaptoglobinaemia

A high incidence of ahaptoglobinaemia (nontypable haptoglobin–HpO) was observed among malaria patients (Joshi *et al* 1987, 1998) (Fig. 103)

and incidence increased with the increase in malaria attacks (Joshi *et al* 1991). Higher incidence of HpO was observed in the population during malaria epidemics (Joshi *et al* 1991, 1999). Antimalarial therapy in ahaptoglobinemic patients has shown normal levels of haptoglobins in about 75% of the subjects within 8–9 days of post-treatment. It is concluded from the study that association of HpO with *P. falciparum* and *P. vivax* malaria is present in Indian population. However, HpO can not be used as an index to study malaria positivity because of its low reliability. Fig. 104 shows the areas from where population samples have been screened and incidence of HpO in the population was surveyed (Joshi *et al* 1985, 1987, 1991, 1998, 1999, 2001). □