Atlas of Haemoglobinopathies in India



Malay Mukherjee

Pallavi Thaker

Roshan Colah

Namrata Mahajan

INDIAN COUNCIL OF MEDICAL RESEARCH

NATIONAL INSTITUTE OF IMMUNOHAEMATOLOGY

ATLAS OF HAEMOGLOBINOPATHIES IN INDIA

Malay Mukherjee
Former Scientist F and Consultant
ICMR-National Institute of Immunohaematology
Mumbai

Roshan Colah
Former Scientist G & Director In-Charge
ICMR-National Institute of Immunohaematology
Mumbai

Pallavi Thaker
Technician C
ICMR-National Institute of Immunohaematology
Mumbai

Namrata Mahajan
Senior Research Fellow
ICMR-National Institute of Immunohaematology
Mumbai

ICMR-NATIONAL INSTITUTE OF IMMUNOHAEMATOLOGY
13th Floor, New Multistoreyed Building, K.E.M. Hospital Campus,
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महानिदेशक, आई सी एम आर

Prof. (Dr.) Balram Bhargava, Padma Shri
MD, DM, FRCP (Glasg.), FRCP (Edin.),
FACC, FAHA, FAMS, FNASc, FASc, FNA, DSc
Secretary to the Government of India
Department of Health Research
Ministry of Health & Family Welfare &
Director-General, ICMR

भारतीय आयुर्विज्ञान अनुसंधान परिषद
स्वास्थ्य अनुसंधान विभाग
स्वास्थ्य एवं परिवार कल्याण मंत्रालय
भारत सरकार
वी. रामलिंगस्वामी भवन, अंसारी नगर
नई दिल्ली - 110 029

Indian Council of Medical Research
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V. Ramalingaswami Bhawan, Ansari Nagar
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Foreword

India is an ethnically and culturally diverse country with a significant burden of the inherited disorders of haemoglobin. The prevalence of thalassemia syndromes and sickle cell disorders varies widely in different regions and selected caste and tribal populations. Considerable amount of work has been undertaken on the distribution of haemoglobinopathies in the country, however it has not been collated to give a comprehensive picture.

The ICMR-National Institute of Immunohaematology (ICMR-NIIH) in Mumbai has been at the forefront and served as a National Referral Centre for co-ordinating epidemiological studies, resolving diagnostic problems and developing control programmes for haemoglobinopathies for about 40 years. With this vast experience, this institute is undoubtedly the right one to bring out an " Atlas of Hemoglobinopathies in India" on the occasion of the inauguration of the 'Centre for Research, Management and Control of Hemoglobinopathies' under ICMR-NIIH at Chandrapur.

The authors have collated all the published data from different states in the country to bring out this atlas, which includes data on population screening, screening of antenatal women, newborn screening, hospital-based screening as well as spectrum of β and α thalassemia mutations in the country along with the rare haemoglobin variants identified. This atlas will serve as reference to plan the education, awareness, and control programmes at the state and the national level.

I congratulate the authors for painstakingly undertaking this immense task.

Balram Bhargava

(Balram Bhargava)

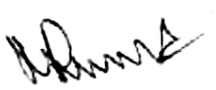
Tele.: 26588204, 26589620, Fax (Off.) : 91-11-26588662, E-mail: secy-dg@icmr.gov.in

PREFACE

Haemoglobinopathies, including the thalassaemia syndromes and sickle cell disorders (SCD), are among the most common treatable inherited diseases and pose a significant health care burden in India. The prevalence of these disorders varies significantly depending on the ethnic background. Though multiple studies have been undertaken to understand the distribution of haemoglobinopathies in India, a systematic compilation of this data is not available.

ICMR-NIIH has been working in haemoglobinopathies for more than 40 years and has made pioneering contributions, including understanding population prevalence, molecular mechanisms, developing technologies for control and coordinating important national mission mode programmes for India. A dedicated 'Centre for Research, Management and Control of Haemoglobinopathies' is being developed at Chandrapur in Maharashtra under the ICMR- National Institute of Immunohaematology. This centre will support the efforts of the government of India for the control and management of haemoglobinopathies. On this occasion, Dr Malay Mukherjee Former Scientist F and Consultant and Dr Roshan Colah, Former Director In-Charge have painstakingly tried to collate the published data from different regions in the country and analysed it to understand the region-wise distribution based on the published literature. They have also enlisted the different haemoglobinopathies described in India including the rare variants and the spectrum of α and β thalassaemia mutations in the country.

This has been a much-needed exercise that will not only help the clinicians and researchers but will also serve as a reference guide for the planning of haemoglobinopathies control and management programmes in India. Kudos to all those who have helped in getting this information together.



Dr Manisha Madkaikar,
Director,
ICMR-National Institute of Immunohaematology, Mumbai.

FROM THE AUTHORS

The inherited disorders of haemoglobin, particularly the β thalassaemia syndromes and sickle cell disease are the most common monogenic disorders in India that lead to a significant health, economic and social burden in the country. The cost of lifelong quality care for these patients is enormous and the only way to reduce the birth of affected children is by implementing a successful control programme by effective screening and counselling and adequate antenatal diagnosis facilities in different states. Knowledge of the distribution and prevalence of these abnormal genes is important to focus attention in regions and communities where the incidence is highest as well as to initiate studies where data is inadequate or lacking. Over the last several years, there have been innumerable studies on the epidemiology and molecular genetics of the β and α thalassaemias, haemoglobin S, haemoglobin E and haemoglobin D disorders as well as on the identification of rare and novel haemoglobin variants.

In this "Atlas of Haemoglobinopathies in India" we have attempted to undertake this seemingly daunting task of collating all the available data from published reports from different regions of the country on screening of various groups of individuals among both non-tribal and tribal communities along with the molecular abnormalities identified in different globin genes.

It is hoped that this information would be of immense value for researchers, administrators, stake holders, policy makers and funding bodies to know the extent of the burden of different haemoglobin disorders state wise and plan and execute future activities where they are most needed.

We sincerely thank Professor Balram Bhargava, Secretary, Dept of Health Research and Director General, Indian Council of Medical Research, New Delhi for his encouragement to undertake this work and to Dr Manisha Madkaikar, Director, ICMR-National Institute of Immunohaematology, Mumbai for giving us this opportunity and providing the necessary facilities.

We gratefully acknowledge Dr Bipin Kulkarni, Scientist D and Head, Dept of Hemostasis & Thrombosis for designing the Cover page, Mr Vijay Padwal, Senior Technical Officer (Library and IT) for support from the library, Ms Rakshanda Nalawade and Ms Shivali Kate for their help with the data entry and Mr Manoj Das and Ms Aparna Vaidya for printing this book.

Malay Mukherjee
Roshan Colah
Pallavi Thaker
Namrata Mahajan

Abbreviations

AD	HbD Punjab trait
AE	HbE trait
AS	HbS trait
SS	Sickle cell homozygous
HbSE	Hb S- Hb E
HbD- β Thal	HbD Punjab - β Thalassaemia
DD	HbD Punjab homozygous
HbE- β Thal	Hb E- β Thalassaemia
EE	Hb E homozygous
HbH	Haemoglobin H
S-β thal	Hb S- β Thalassaemia
βTM	β Thalassaemia Major
βTT	β Thalassaemia Trait
δβ thal trait	δβ thalassaemia trait
HPFH trait	Hereditary Persistance of Fetal Haemoglobin Trait
IVS	Intervening sequence
Hb	Haemoglobin
Del	Deletion
Fig.	Figure
No.	Number
Ref.	References
Sr.No.	Serial number
bp.	Base pair
CD	Codon
SA	South African
SEA	South East Asian
UT	Union Territory

Introduction

Indians represent one-sixth of the world population and consist of ethnically, geographically and genetically diverse populations with several thousand endogamous groups. Historically, the Indian populations have a multi cultural and evolutionary history. The evolutionary antiquity of Indian ethnic groups and subsequent migration from central Asia, west Asia and southern China has resulted in a rich socio-cultural, linguistic and biological diversity. Broadly, Indians belong to Austro–Asiatic (AA), Tibeto–Burman (TB), Indo–European (IE) and Dravidian (DR) language families. Distinct religious communities, hierarchical castes and sub castes, and several isolated tribal groups that comprise the people of India remain largely endogamous. Most of these groups have strict social rules governing mating patterns. India comprises of 28 states and 8 union territories. These states and the union territories are divided into 752 districts. These districts are further divided into sub-districts, which are known differently in different parts in the country (e.g., tahsil, taluka, community development (CD) block, police station, mandal, revenue circle, etc.). The ethnic composition of the Indian population is complex, with the coexistence of more than 2000 ethnic groups. Each ethnic group is classified into one of four official social designations. These are: (i) Scheduled Tribes (ST), (ii) Scheduled Castes (SC), (iii) Other Backward Classes (OBCs), and (iv) General Classes (GCs).

Indian populations are different in their genetic makeup, which may either predispose them to a disease or protect them from a disease, depending on the environment they are exposed to. In some communities the load of genetic disorders is relatively high due to consanguineous marriages practiced in the community. Genetically isolated populations are considered to be important in dissecting complex diseases and mapping underlying genes. It is possible that populations living in close geographic proximity are more likely to exchange genes, thereby enhancing genetic similarity, despite the fact that these populations may not belong to the same socio-cultural stratum.

Thalassaemias and other haemoglobinopathies are the most common monogenic disorders globally. They have an autosomal recessive inheritance and result from point mutations or deletions in one or more globin gene(s), The thalassaemias are due to a reduction or absence of synthesis of one of the globin chains altering the normal α : non α synthesis ratios (α: non-α) while the abnormal haemoglobin variants are due to production of structurally defective genes. Ultimately these disorders cause varying degrees of anaemia that can range from insignificant to life threatening. β-thalassaemias and their co-inheritance with haemoglobin E (HbE) or haemoglobin S (HbS) result in considerable health problems in India and contribute significantly to morbidity and mortality. Sickle Cell Disease has much diversity in its occurrence among the tribal populations; having an alarmingly higher prevalence among the tribes from Central and Western region of India. Interestingly, though the incidence of Malaria has a role behind the evolution of the HbS mutation; not all high endemic zones for malaria have been found to have a higher prevalence of the HbS mutation. Tribes of Assam and Tripura show a strikingly higher prevalence of HbE. In India, the cumulative gene frequency of haemoglobinopathies is around 4.2%. With a population of over one billion and a birth rate of 28 per thousand, it has been estimated that there would be around 42 million carriers and over 12000 babies are born each year with a major and clinically significant haemoglobinopathy.

ICMR-National Institute of Immunohaematology is the leader in the field of Haemoglobinopathies in

the country and has undertaken large multicentre studies in tribal and non tribal regions in several collaborations with other medical colleges, government agencies and NGOs. Several years back in 1986 the Institute had compiled a Genetic Atlas of Indian Tribes showing the distribution and prevalence of Blood Groups, Haemoglobinopathies and some Enzyme Polymorphisms in tribal populations in different states in India. Since then, a huge amount of work has been reported from different states on the distribution and prevalence of the thalassaemias and other haemoglobinopathies in tribal as well as non-tribal population groups. With a Centre for Research, Management and Control of Haemoglobinopathies coming up at Chandrapur under the ICMR-National Institute of Immunohaematology, it was felt that for any future planning of Haemoglobinopathy research, there was an urgent need for an updated Atlas of Haemoglobinopathies in India. We therefore collated the published data on distribution of haemoglobinopathies from the country.

We created an up-to-date database of haemoglobinopathy studies in India taking into account the population group (tribal and non-tribal) along with data on antenatal and newborn screening. We also included hospital based data and case reports of rare haemoglobin variants as well as the spectrum of mutations causing β-thalassaemias and α-thalassaemias in Indian populations. An electronic search was done in databases including Scopus, PubMed, and Google Scholar for articles from 1985 to 2021. The following terms and keywords were used to conduct a comprehensive literature search: haemoglobinopathies or sickle cell disease or SCD or α and β thalassaemias or δβ thalassaemia or HbH disease or abnormal haemglobinopathies or haemoglobin variants. Furthermore, we conducted a manual review of the reference lists of the relevant articles and the previously published Genetic Atlas for additional pertinent studies. To avoid repetition of the data, review articles on haemoglobinopathies were excluded. We also excluded the articles where only solubility test and Naked Eye Single Tube Red Cell Osmotic Fragility (NESTROFT) were done. Many studies only reported the prevalence of β thalassaemia, or HbS or HbE The required data were extracted from the articles using sheet form of Excel. The following data were extracted: Name of the state or union territory, communities, study places, total number of samples tested along with phenotypes and mutations identified. We have also included the community wise frequency of different haemoglobinopathies wherever available. However, in many studies the communities and the districts from where samples were collected have not been mentioned. Therefore, the present work is an attempt at compilation of data to the best of our ability, being fully aware that it may be far from complete.

The published data on haemoglobinopathies are categorized state/union territory wise in 3 sections as follows:

Section I:

1. Population screening in non-tribals.
2. Population screening in tribals.
3. Antenatal screening
4. Newborn screening.
5. Referred cases to the hospital.

Section II:

1. β-thalassaemia mutations in different States and Union Territories.
2. α-thalassaemia mutations in different States and Union Territories.

Section III:

Case Reports

1) Hb H Disease.
2) HPFH and δβ Thalassaemia.
3) Common Hb Variants.
4) Novel and Rare Hb Variants.

The Atlas is broadly divided into 7 geographical regions;

1. Eastern Region: The states of Bihar, Jharkhand, Odisha and West Bengal.
2. North Eastern Region: The states of Arunachal Pradesh, Assam, Tripura, Manipur, Meghalaya, Mizoram and Nagaland.
3. Northern Region: The states of Haryana, Himachal Pradesh, Punjab, Uttarakhand, Uttar Pradesh and union territories of Delhi and Jammu & Kashmir.
4. Western Region: The states Gujarat, Maharashtra, Rajasthan, Goa and union territories of Dadra and Nagar Haveli.
5. Central Region: The states of Chattisgarh and Madhya Pradesh.
6. Southern Region: The states of Andhra Pradesh, Karnataka, Kerala, Tamil Nadu and Telangana.
7. Island Region: Union territories of Andaman and Nicobar Islands.

In each region, maps of the states indicating the place (District) from where individuals were screened, followed by list of districts and communities studied along with tables of the respective data as reported in original studies are presented. After exclusion, a total of 460 studies from 27 States and 7 Union Territories of India were compiled. Approximately one-third of the studies were carried out in the eastern and north eastern region particularly in Orissa, West Bengal and Assam followed by western, southern, central, northern and island regions.

Compilation of the available data showed that β-thalassaemia was seen in high frequencies in the North-West and Eastern states in widely varying frequencies while α-thalassaemia was highly prevalent in some of the tribal groups in Western and Central India. δβ thalassaemias and hereditary persistence of fetal haemoglobin (HPFH) were sporadically reported from different regions. Among the abnormal haemoglobins, HbS was predominantly found among the tribal populations of Central, Western and Southern states of India while HbE and HbD Punjab were widely distributed in the North-Eastern states and Northern states of India respectively. Several other rare α and β chain haemoglobin variants have also been sporadically reported from India. The profile of β thalassaemia mutations have been reported from different states, many of them as a part of prenatal diagnosis programmes. The prevalence of α thalassaemias has largely been studied in populations where the sickle gene is predominantly seen. Besides this, cases of HbH disease have been reported sporadically.

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Western Region

The Western region includes four states, Maharashtra, Gujarat, Rajasthan, Goa and the union territory of Dadra & Nagar Haveli. A total of 86 articles were compiled which included population, antenatal, newborn and hospital based studies. Table 1 shows the number of districts which were covered and studies that were available for compilation in each state.

Table 1 - Studies available from the Western Region

States/Union Territory	Districts	No of Studies
Maharashtra	32	42
Gujarat	25	28
Rajasthan	6	10
Goa	1	2
Dadra & Nagar Haveli	2	4
Total	**66**	**86**

Approximately 50% of the studies were carried out in Maharashtra followed by Gujarat, Rajasthan, Dadra & Nagar Haveli and Goa. Majority of the studies were on prevalence of β thalassaemia and other haemoglobinopathies among the tribal and non-tribal populations. In many studies in this region either the districts or specific population groups studied have not been mentioned hence this information may be incomplete. The states/union territory (shaded areas) covered in the Western region are shown in Fig. 1.



Fig 1: Map of India showing the states / union territory covered.

MAHARASHTRA

Maharashtra is the richest and second most populous state in India with a population of 112.3 million among whom 11.8% and 8.9% belong to scheduled castes and scheduled tribes respectively (Census 2011). It is bordered by the Arabian Sea to the west, Karnataka and Goa to the south, Telangana to the southeast and Chhattisgarh to the east, Gujarat and Madhya Pradesh to the north, and the union territory of Dadra and Nagar Haveli and Daman and Diu to the northwest. Mumbai, the capital being the financial and commercial hub houses many population groups from different states who have migrated here for a better livelihood.

A total of 42 studies were available. Of these, 27 were population based, 5 were on antenatal screening, 3 were on newborn screening and 8 were hospital based studies. Fig 2 shows that studies have been undertaken in most of the districts (shaded areas) in the state. The names of the districts covered have not been mentioned in two studies.

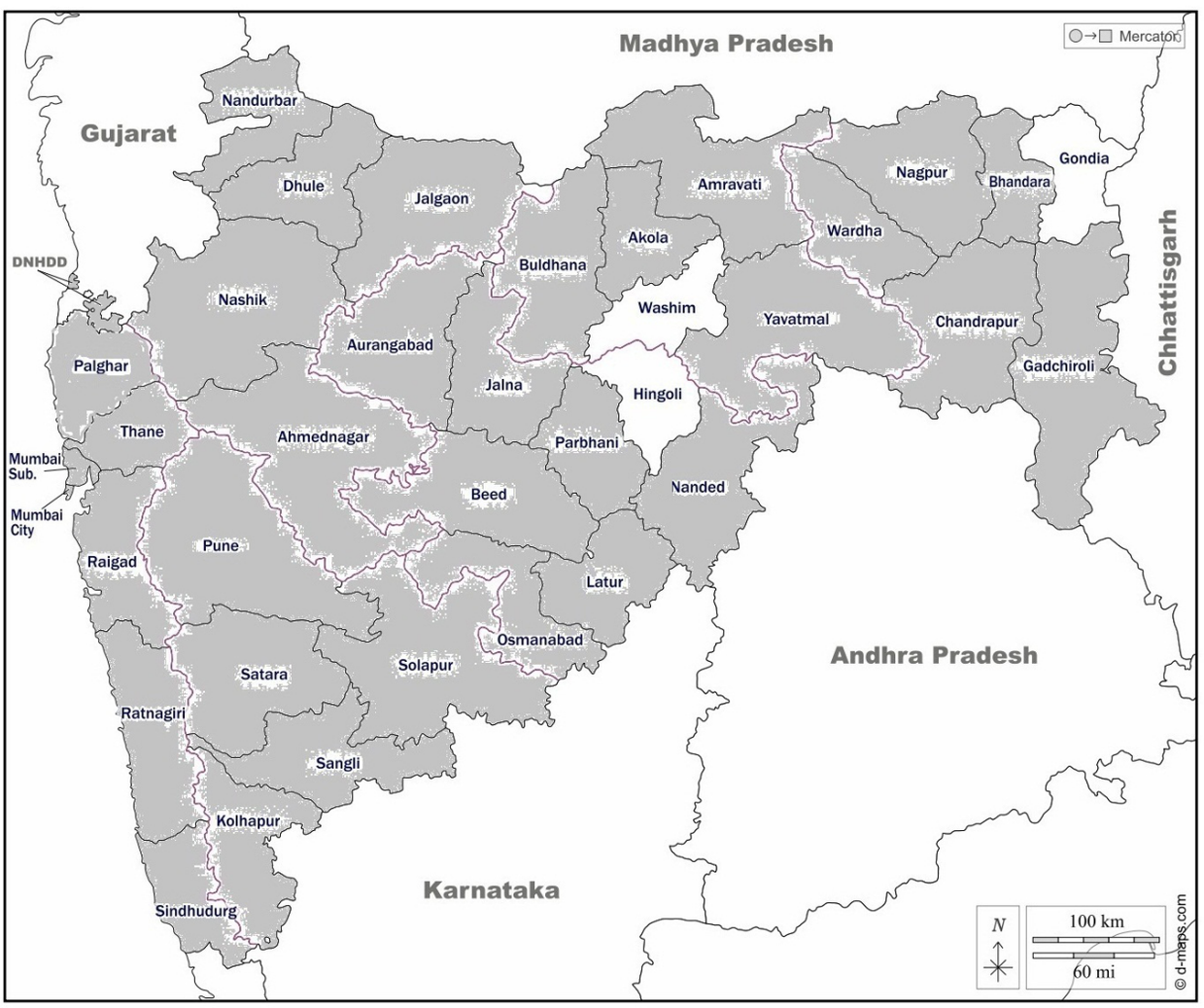

Fig 2: Map of Maharashtra showing the studied areas

The number of districts covered along with the population groups (non-tribal and tribal) studied are shown in Tables 2-4. A total of 40 non-tribal and 30 tribal groups were studied from 32 districts.

Table 2: Districts covered in Maharashtra

Ahmednagar	Akola	Amravati	Aurangabad	Beed
Bhandara	Buldana	Chandrapur	Dhule	Gadchiroli
Jalgaon	Jalna	Kolhapur	Latur	Mumbai
Nagpur	Nanded	Nandurbar	Nashik	Osmanabad
Palghar	Parbhani	Pune	Raigad	Ratnagiri
Sangli	Satara	Sindhudurg	Solapur	Thane
Wardha	Yavatmal			

Table 3: Non-Tribal populations studied

Artisan	Bari	Bhandari	Bhang	Bhoi
Bhoyar	Brahmin	Chammar	Christian	Dhangar
Dhiwar	Dhobi	Jain	Cutchhi Bhanusali	Kallar
Khatri	Komti	Kosti	Kunbi	Lohana
Lohar	Mahar	Maheshwari	Mali	Maratha
Matang	Mehetar	Mehta	Muslim	Nabhi
Oza	Pathare Prabhu	Rajput	Sahu	Saraswat
Shimpi	Sindhi	Sonar	Sutar	Teli

Table 4: Trbal populations studied

Andha	Balai	Banjara	Bhil	Dhor
Gawalan	Gond	Gowari	Halba	Katkari
Kokna	Kolam	Koli	Korku	Madia Gond
Madgi	Mana	Mahadev Koli	Mannewar	Naik Gond
Nihaal	Pardhan	Pawara	Powa	Raj Gond
Tadavi	Thakur	Thakkar	Vanjara	Warli

Population based studies indicated the presence of HbS and β thalassaemia gene in both the tribal and non-tribal populations. HbS was mainly found in the Vidharbha region (Akola, Amravati, Bhandara, Chandrapur, Gadchiroli, Nagpur, Wardha and Yavatmal) and north Maharashtra (Dhule and Nandurbar).

The prevalence of β thalassaemia trait (βTT) varied from 0.10 to 21.20 % among the non-tribal populations in different studies with sickle cell trait (AS) ranging from 0.16 to 16.10 % and HbD Punjab trait (AD) from 0.35 to 2.73 % in a few studies (Table 5). Occasional studies have reported the presence

of HbE trait (AE), δβ thalassaemia trait, HPFH trait and some rare Hb variants. HbS has been predominantly reported among the tribal populations with the prevalence of HbS carriers (AS) varying from 0.55 to 55.00% % and sickle homozygotes (SS) from 0.18 to 30.00% %. Few studies have also screened for β thalassaemia with the prevalence of (βTT) ranging from 0.43 to 5.88 % (Table 6). Antenatal screening has been reported mainly from Mumbai where apart from βTT, individuals with AS, AE and AD were also encountered (Table 7). Data on newborn screening is reported only from Nagpur where a large number of AS and SS babies were identified as a targeted screening approach was used where only babies of sickle heterozygous mothers were screened (Table 8). In the hospital based reports (Table 9) where large numbers were studied, β thalassaemia and all other haemoglobinopathies were picked up including many rare Hb variants. Unusual compound heterozygous combinations like HbS-Q India, HbS-HPFH and HbSC disease were also reported from Maharashtra although the HbSC cases in one of the studies were among individuals of African origin.

Table 5: Population studies in Non- Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Mumbai	Cuttchi Bhanushali	296	44 (14.90)	-	-	-	-	-	-	-	-	-	-	-	-	-	1
2	Mumbai	Saraswat	587	22 (3.77)	-	-	-	-	-	-	-	7(1.19)	-	-	-	-	-	2
3	Mumbai	Lohana	564	51 (9.00)	-	-	-	-	-	-	-	-	-	-	-	-	HbJ trait--1, HbL trait--2	2
4	Mumbai	Sindhi	70	5 (7.10)	-	-	-	-	-	-	-	-	-	-	-	-	HbL trait--1	2
5	Ratnagiri	Brahmin	72	0	-	0	0	-	-	-	-	-	-	-	-	-	-	3
6	Ratnagiri	Artisan	117	1 (0.85)	-	1 (0.85)	0	-	-	-	-	-	-	-	-	-	-	3
7	Ratnagiri	Maratha	131	0	-	0	0	-	-	-	-	-	-	-	-	-	-	3
8	Ratnagiri	Bhandari	28	0	-	0	0	-	-	-	-	-	-	-	-	-	-	3
9	Ratnagiri	Kunbi	144	0	-	0	0	-	-	-	-	-	-	-	-	-	-	3
10	Ratnagiri	Mahar	38	0	-	0	0	-	-	-	-	-	-	-	-	-	-	3
11	Ratnagiri	Muslim	30	0	-	0	0	-	-	-	-	-	-	-	-	-	-	3
12	Sindhudurg	Brahmin	70	0	-	1 (1.42)	0	-	-	-	-	-	-	-	-	-	-	3
13	Sindhudurg	Artisan	156	1 (0.64)	-	0	0	-	2 (1.28)	-	-	-	-	-	-	-	-	3
14	Sindhudurg	Maratha	447	3 (0.67)	-	0	0	-	-	-	-	-	-	-	-	-	-	3
15	Sindhudurg	Bhandari	96	0	-	0	0	-	-	-	-	-	-	-	-	-	-	3
16	Sindhudurg	Mahar	35	0	-	0	0	-	-	-	-	-	-	-	-	-	-	3
17	Sindhudurg	Christians	10	0	-	0	0	-	-	-	-	-	-	-	-	-	-	3
18	Sindhudurg	others	7	0	-	0	0	-	-	-	-	-	-	-	-	-	-	3
19	Wardha	Bari, Bhoi,Chambar, Dhangar, Halba, Jain, Kalar, Khatri, Komti, Koshti, Maheshwari, Mana, Muslim, Navi, Oza, Maratha, Shimpi, Sonar, Sutar, Thakur, Matang, Pradhan, Gowari, Mahar, Kunbi, Teli	5561	-	-	151 (3.70)	9 (0.20)	-	-	-	-	-	-	-	-	-	-	4

(contd.)

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
20	Nagpur	Larkana Sindhis*	415	73 (17.60)	13 (3.10)	-	-	-	-	-	-	-	-	-	-	-	-	5
21	Nagpur	Ghoti Sindhi*	267	46 (17.50)	8 (3.00)	-	-	-	-	-	-	-	-	-	-	-	-	5
22	Nagpur	Sahiti Sindhi*	227	19 (8.70)	1 (0.07)	-	-	-	-	-	-	-	-	-	-	-	-	5
23	Nagpur	Sakhru Sindhi*	127	22 (17.20)	3 (2.90)	-	-	-	-	-	-	-	-	-	-	-	-	5
24	Nagpur	Dadu Sindhi*	86	7 (8.00)	1 (0.60)	-	-	-	-	-	-	-	-	-	-	-	-	5
25	Nagpur	Others Sindhi*	215	45 (21.20)	9 (4.50)	-	-	-	-	-	-	-	-	-	-	-	-	5
26	Nagpur	Unknown*	226	27 (12.10)	3 (2.20)	-	-	-	-	-	-	-	-	-	-	-	-	5
27	Mumbai	Not Mentioned	5004	122 (2.44)	0	25 (0.50)	0	0	7 (0.14)	0	0	18(0.36)	0	0	2 (0.04)	11(0.22)	7	6
28	Mumbai	Not Mentioned	547	22 (4.00)	-	-	-	-	-	-	-	-	-	-	-	-	-	7
29	Raigad	Not Mentioned	578	30 (5.20)	-	-	-	-	-	-	-	-	-	-	-	-	-	7
30	Thane	Not Mentioned	233	11 (4.70)	-	-	-	-	-	-	-	-	-	-	-	-	-	7
31	Ratnagiri	Not Mentioned	1696	35 (2.10)	-	-	-	-	-	-	-	-	-	-	-	-	-	7
32	Sidhudurg	Not Mentioned	1070	11 (1.00)	-	-	-	-	-	-	-	-	-	-	-	-	-	7
33	Aurangabad	Not Mentioned	168	4 (2.40)	-	-	-	-	-	-	-	-	-	-	-	-	-	7
34	Beed	Not Mentioned	255	11 (4.90)	-	-	-	-	-	-	-	-	-	-	-	-	-	7
35	Jalna	Not Mentioned	77	1 (1.30)	-	-	-	-	-	-	-	-	-	-	-	-	-	7
36	Latur	Not Mentioned	112	3 (2.70)	-	-	-	-	-	-	-	-	-	-	-	-	-	7
37	Nanded	Not Mentioned	101	2 (2.00)	-	-	-	-	-	-	-	-	-	-	-	-	-	7
38	Osmanabad	Not Mentioned	86	1 (1.20)	-	-	-	-	-	-	-	-	-	-	-	-	-	7
39	Parbhani	Not Mentioned	38	1 (2.60)	-	-	-	-	-	-	-	-	-	-	-	-	-	7
40	Ahmednagar	Not Mentioned	503	30 (6.00)	-	-	-	-	-	-	-	-	-	-	-	-	-	7
41	Dhule	Not Mentioned	138	5 (3.90)	-	-	-	-	-	-	-	-	-	-	-	-	-	7
42	Jalgaon	Not Mentioned	173	3 (1.70)	-	-	-	-	-	-	-	-	-	-	-	-	-	7
43	Nashik	Not Mentioned	316	13 (4.10)	-	-	-	-	-	-	-	-	-	-	-	-	-	7
44	Kolhapur	Not Mentioned	452	11 (2.40)	-	-	-	-	-	-	-	-	-	-	-	-	-	7
45	Solapur	Not Mentioned	400	7 (1.80)	-	-	-	-	-	-	-	-	-	-	-	-	-	7
46	Pune	Not Mentioned	807	11 (1.40)	-	-	-	-	-	-	-	-	-	-	-	-	-	7
47	Sangli	Not Mentioned	301	7 (2.30)	-	-	-	-	-	-	-	-	-	-	-	-	-	7

(contd.)

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
48	Satara	Not Mentioned	823	36 (4.40)	-	-	-	-	-	-	-	-	-	-	-	-	-	7
49	Chandrapur	Not Mentioned	489	14 (2.90)	-	-	-	-	-	-	-	-	-	-	-	-	-	7
50	Gadchiroli	Not Mentioned	360	4 (1.10)	-	-	-	-	-	-	-	-	-	-	-	-	-	7
51	Bhandara	Not Mentioned	157	3 (1.90)	-	-	-	-	-	-	-	-	-	-	-	-	-	7
52	Akola	Not Mentioned	132	2 (1.50)	-	-	-	-	-	-	-	-	-	-	-	-	-	7
53	Amravati	Not Mentioned	54	1 (1.90)	-	-	-	-	-	-	-	-	-	-	-	-	-	7
54	Buldhana	Not Mentioned	72	2 (2.80)	-	-	-	-	-	-	-	-	-	-	-	-	-	7
55	Yavtamal	Not Mentioned	540	9 (1.70)	-	-	-	-	-	-	-	-	-	-	-	-	-	7
56	Mumbai	Not Mentioned	5682	152 (2.68)	0	9 (0.20)	0	0	2 (0.04)	0	0	38 (0.70)	0	0	12(0.20)	0	0	8
57	Nagpur, Yavatmal, Chandrapur, Bhandara	Mahar	1651	0	-	195(11.80)	8 (4.48)	-	0	0	-	0	0	-	-	4 (0.24)	-	9
58	Nagpur, Yavatmal, Chandrapur, Bhandara	Kunbi	666	1 (0.10)	-	47 (7.00)	0	-	-	-	-	0	0	-	-	0	-	9
59	Nagpur, Yavatmal, Chandrapur, Bhandara	Teli	329	0	-	12 (3.65)	0	-	-	-	-	0	0	-	-	0	-	9
60	Nagpur, Yavatmal, Chandrapur, Bhandara	Mali	118	0	-	19 (16.10)	0	-	-	-	-	0	0	-	-	0	-	9
61	Nagpur, Yavatmal, Chandrapur, Bhandara	Kalar	111	0	-	6 (5.41)	0	-	-	-	-	0	0	-	-	0	-	9
62	Nagpur, Yavatmal, Chandrapur, Bhandara	Brahmin	144	0	-	6 (4.17)	0	-	-	-	-	0	0	-	-	0	-	9

(contd.)

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
63	Nagpur, Yavatmal, Chandrpur, Bhandara	Muslim	161	0	-	6 (3.73)	0	-	-	-	-	0	0	-	-	0	-	9
64	Nagpur, Yavatmal, Chandrapur, Bhandara	Sindhi	1241	115 (9.27)	-	2 (0.16)	0	-	-	-	-	9 (2.73)	4 (0.32)	-	-	0	-	9
65	Nagpur, Yavatmal, Chandrapur, Bhandara	Chamar	54	0	-	1 (1.85)	0	-	-	-	-	0	0	-	-	0	-	9
66	Nagpur, Yavatmal, Chandrapur, Bhandara	Dhangar	43	1 (2.33)	-	1 (2.33)	0	-	-	-	-	0	0	-	-	1 (2.33)	-	9
67	Nagpur, Yavatmal, Chandrapur, Bhandara	Dhiwar	55	0	-	5 (9.00)	0	-	-	-	-	0	0	-	-	0	-	9
68	Nagpur, Yavatmal, Chandrapur, Bhandara	Lohar	59	0	-	1 (1.69)	0	-	-	-	-	0	0	-	-	0	-	9
69	Nagpur, Yavatmal, Chandrapur, Bhandara	Mehetar, Madgi, Dhobi, Rajput	194	3 (1.50)	-	8 (4.10)	0	-	-	-	-	0	1 (0.50)	-	-	0	-	9
70	Nagpur, Yavatmal, Chandrapur, Bhandara	Others	836	1 (0.10)	-	8(0.90)	0	-	-	-	-	3 (0.35)	0	-	-	0	-	9
71	Nagpur	Sindhi	1498	217 (14.49)	-	-	-	-	-	-	-	28 (1.87)	-	-	-	-	HbQ trait - -6, HbQ-β thal--1 , HbD/β-thal--3	10

(contd.)

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
72	Nagpur, Amravati, Gadhchiroli, Chandrapur	Bhang, Chamar, Mehta	173	-	-	4 (2.33)	0	0	-	-	-	-	-	-	-	-	-	11
73	Nagpur, Amravati, Gadhchiroli, Chandrapur	Mahar	4058	-	-	541(13.30)	10 (0.25)	0	-	-	-	-	-	-	-	-	-	11
74	Nagpur, Amravati, Gadhchiroli, Chandrapur	Kunbi	1953	-	-	89 (4.36)	1 (0.05)	0	-	-	-	-	-	-	-	-	-	11
75	Nagpur, Amravati, Gadhchiroli, Chandrapur	Navi	105	-	-	2 (1.90)	0	0	-	-	-	-	-	-	-	-	-	11
76	Nagpur, Amravati, Gadhchiroli, Chandrapur	Dhobi	129	-	-	3 (2.33)	0	0	-	-	-	-	-	-	-	-	-	11
77	Nagpur, Amravati, Gadhchiroli, Chandrapur	Sonar	174	-	-	2 (1.15)	0	0	-	-	-	-	-	-	-	-	-	11
78	Nagpur, Amravati, Gadhchiroli, Chandrapur	Sutar	119	-	-	5 (4.20)	0	0	-	-	-	-	-	-	-	-	-	11
79	Nagpur, Amravati, Gadhchiroli, Chandrapur	Sahu,Shahu,Teli	1105	-	-	32 (2.90)	0	0	-	-	-	-	-	-	-	-	-	11
80	Nagpur, Amravati, Gadhchiroli, Chandrapur	Mali	502	-	-	16 (3.19)	0	0	-	-	-	-	-	-	-	-	-	11

(contd.)

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
81	Nagpur, Amravati, Gadhchiroli, Chandrapur	Thakur	100	-	-	8 (8.00)	0	0	-	-	-	-	-	-	-	-	-	11
82	Nagpur, Amravati, Gadhchiroli, Chandrapur	Pawar, Powar, Bhoyar	156	-	-	1 (0.64)	0	0	-	-	-	-	-	-	-	-	-	11
83	Nagpur, Amravati, Gadhchiroli, Chandrapur	Kalar	158	-	-	4 (2.53)	0	0	-	-	-	-	-	-	-	-	-	11
84	Mumbai	Pathare Prabhus	257	10 (3.89)	0	0	0	0	0	0	0	2 (0.77)	0	0	0	0	0	12

*Eastimated Prevalence

Table 6: Population studies in Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Dhule	Bhil	82	-	-	13(15.85)	0	-	-	-	-	-	-	-	-	-	-	13
2	Aurangabad	Bhil	102	-	-	2 (1.96)	0	-	-	-	-	-	-	-	-	-	-	13
3	Ahmednagar	Bhil	33	-	-	0	0	-	-	-	-	-	-	-	-	-	-	13
4	Pune	Koli	181	-	-	1 (0.55)	0	-	-	-	-	-	-	-	-	-	-	13
5	Pune	Katkari	61	-	-	0	0	-	-	-	-	-	-	-	-	-	-	13
6	Raigad	Katkari	109	-	-	6 (5.50)	0	-	-	-	-	-	-	-	-	-	-	13
7	Thane/ Palghar	Katkari	70	-	-	1 (1.43)	0	-	-	-	-	-	-	-	-	-	-	13
8	Thane/ Palghar	Dhor koli	67	-	-	5 (7.46)	0	-	-	-	-	-	-	-	-	-	-	13
9	Thane/ Palghar	Konkana	86	-	-	2 (2.33)	0	-	-	-	-	-	-	-	-	-	-	13
10	Thane/ Palghar	Warli	145	-	-	1 (6.90)	0	-	-	-	-	-	-	-	-	-	-	13

(contd.)

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
11	Nashik	Mahadev Koli	50	-	-	0	0	-	-	-	-	-	-	-	-	-	-	13
12	Amravati	Gond	29	-	-	5 (17.24)	0	-	-	-	-	-	-	-	-	-	-	13
13	Amravati	Korku	172	-	-	16 (9.30)	0	-	-	-	-	-	-	-	-	-	-	13
14	Amravati	Balai	11	-	-	5 (45.45)	0	-	-	-	-	-	-	-	-	-	-	13
15	Dhule	Pawara	110	-	-	27 (24.55)	0	-	-	-	-	-	-	-	-	-	-	13
16	Dhule	Bhil	186	-	-	16 (8.60)	0	-	-	-	-	-	-	-	-	-	-	14
17	Dhule	Pawara	122	-	-	27 (22.13)	0	-	-	-	-	-	-	-	-	-	-	14
18	Dhule	Katkari	131	-	-	8 (6.11)	0	-	-	-	-	-	-	-	-	-	-	14
19	Nanded & Yavatmal	Pardhan	146	-	-	48 (33.50)	0	-	-	-	-	-	-	-	-	-	-	15
20	Nanded& Yavatmal	Gond	257	-	-	17 (6.60)	0	-	-	-	-	-	-	-	-	-	-	15
21	Dhulia	Bhil	673	-	-	125(18.57)	0	-	-	-	-	-	-	-	-	-	-	15
22	Yavatmal	Kolam	36	-	-	3 (8.30)	0	-	-	-	-	-	-	-	-	-	-	15
23	Jalgaon	Pawara	219	-	-	39 (17.80)	0	-	-	-	-	-	-	-	-	-	-	15
24	Jalgaon	Tadavi	72	-	-	6 (8.30)	0	-	-	-	-	-	-	-	-	-	-	15
25	Jalgaon	Vanjara	413	-	-	23 (5.60)	0	-	-	-	-	-	-	-	-	-	-	15
26	Nanded	Andha	152	-	-	3 (1.90)	0	-	-	-	-	-	-	-	-	-	-	15
27	Thane, Pune	Mahadev Koli	155	-	-	4 (2.90)	0	-	-	-	-	-	-	-	-	-	-	15
28	Thane, Pune, Ahmednagar	Thakur	358	-	-	0	0	-	-	-	-	-	-	-	-	-	-	15
29	Chandrapur	Raj Gond	265	14 (5.28)	-	36 (13.58)	1 (0.38)	-	-	-	-	-	-	-	-	-	-	16
30	Chandrapur	Naik Gond	106	6 (5.66)	-	12 (10.81)	0	-	-	-	-	-	-	-	-	-	-	16
31	Chandrapur	Pardhan	133	2 (1.59)	-	42(31.58)	3 (2.26)	-	-	-	-	-	-	-	-	-	-	16
32	Yavatmal	Raj Gond	283	5 (1.90)	-	28 (9.89)	1 (0.35)	-	-	-	-	-	-	-	-	-	-	16
33	Yavatmal	Kolam	144	4 (2.77)	-	27 (18.85)	0	-	-	-	-	-	-	-	-	-	-	16
34	Ahmednagar	Bhil	216	7 (3.24)	-	4 (1.85)	1 (0.46)	0	0	0	-	0	0	-	-	-	-	17
35	Ahmednagar	Mahadeo Koli	102	0	-	1 (0.98)	0	0	0	0	-	0	0	-	-	-	-	17

(contd.)

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
36	Nashik	Bhil	261	5 (2.00)*	-	22 (8.40)	0	0	0	0	-	0	0	-	-	-	-	17
37	Nashik	Mahadeo Koli	71	2 (2.82)	-	1 (1.40)	0	0	0	0	-	0	0	-	-	-	-	17
38	Chandrapur	Madia Gond	113	-	-	25(26.80)	3 (2.10)	-	-	-	-	-	-	-	-	-	-	18
39	Chandrapur	Raj Gond	102	-	-	9 (10.40)	1 (0.30)	-	-	-	-	-	-	-	-	-	-	18
40	Not Mentioned	Bhil & Pawar	314	12 (3.80)	-	112(35.60)	2 (0.63)	-	-	-	-	-	-	-	-	-	-	19
41	Amravati	Korku	865	-	-	38 (4.40)	8 (0.9)	-	-	-	-	-	-	-	-	-	-	20
42	Amravati	Korku	212	-	-	19 (8.96)	4 (1.89)	-	-	-	-	-	-	-	-	-	-	21
43	Amravati	Bhil	53	-	-	13(24.52)	1 (1.88)	-	-	-	-	-	-	-	-	-	-	21
44	Amravati	Gawalan	54	-	-	6 (11.11)	3 (5.55)	-	-	-	-	-	-	-	-	-	-	21
45	Amravati	Gowari	54	-	-	2 (3.70)	2 (3.70)	-	-	-	-	-	-	-	-	-	-	21
46	Amravati	Nihaal	63	-	-	3 (4.76)	2 (3.17)	-	-	-	-	-	-	-	-	-	-	21
47	Nagpur	Not Mentioned	908	-	-	55 (6.05)	2 (0.22)	-	-	-	-	-	-	-	-	-	-	22
48	Nagpur, Yavatmal, Chandrapur, Bhandara	Gond	230	1 (0.43)	-	27(11.74)	0	-	-	-	-	-	-	-	-	-	-	9
49	Nagpur, Yavatmal, Chandrapur, Bhandara	Gowari	55	0	-	4 (7.27)	0	-	-	-	-	-	-	-	-	-	-	9
50	Nagpur, Yavatmal, Chandrapur, Bhandara	Pardhan	64	0	-	13(20.31)	1 (1.56)	-	-	-	-	-	-	-	-	-	-	9
51	Nagpur, Yavatmal, Chandrapur, Bhandara	Banjara	17	1 (5.88)	-	1 (5.88)	1 (5.88)	-	-	-	-	-	-	-	-	-	-	9
52	Nagpur, Yavatmal, Chandrapur, Bhandara	Halba	139	1 (0.72)	-	2 (1.44)	0	-	-	-	-	1 (0.72)	-	-	-	-	-	9

(contd.)

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
53	Nagpur, Yavatmal, Chandrapur, Bhandara	Powar	113	4 (3.54)	-	2(1.77)	0	-	-	-	-	-	-	-	-	-	-	9
54	Yavatmal	Gond	33	-	-	16(48.48)	6 (18.18)	-	-	-	-	-	-	-	-	-	-	23
55	Yavatmal	Kolam, Banjara, Gowari, Madgi	22	-	-	8 (36.36)	2 (9.09)	-	-	-	-	-	-	-	-	-	-	23
56	Yavatmal	Pardhan	60	-	-	33(55.00)	18(30.00)	-	-	-	-	-	-	-	-	-	-	23
57	Yavatmal	Gond,Kolam,Pardhan	1365	-	-	13 (0.95)	15 (1.09)	-	-	-	-	-	-	-	-	-	-	24
58	Yavatmal	Gond	495	-	-	4 (0.80)	5 (1.08)	-	-	-	-	-	-	-	-	-	-	25
59	Yavatmal	Kolam	291	-	-	5 (1.71)	8 (2.70)	-	-	-	-	-	-	-	-	-	-	25
60	Yavatmal	Pardhan	292	-	-	3 (1.02)	6 (2.05)	-	-	-	-	-	-	-	-	-	-	25
61	Chandrapur, Nagpur, Gadchiroli	Halba	216	-	-	29(13.40)	0	-	-	-	-	-	-	-	-	-	-	11
62	Amravati	Korku	883	-	-	76 (8.61)	4 (0.45)	-	-	-	-	-	-	-	-	-	-	11
63	Nagpur, Amravati, Gadchiroli, Chandrapur	Gond, Gowari, Mannewar, Madiya, Mana	5992	-	-	754(12.58)	11 (0.18)	-	-	-	-	-	-	-	-	-	-	11
64	Raigad	Katkari, Thakkar, Kokana	51	1 (1.96)	-	2 (3.92)	0	-	-	-	-	1 (1.96)	0	-	-	-	-	26
65	Gadchiroli	Madia	2137	30 (1.40)	-	413(19.32)	24 (1.12)	0	0	0	0	0	0	0	52(2.43)	0	0	27
66	Yavatmal	Kolam	1848	57 (3.08)	-	275(14.88)	12 (0.64)	0	0	0	0	0	0	0	28(1.51)	0	0	27
67	Raigad	Katkari	1941	74 (3.81)	-	199(10.25)	6 (0.30)	0	0	0	0	12 (0.61)	0	0	8 (0.41)	0	0	27

*5 out of 250 tested

Table 7: Antenatal screening

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Mumbai	Not Mentioned	14086	270(1.92)	-	-	-	-	-	-	-	-	-	-	-	-	-	28
2	Mumbai	Not Mentioned	61935	1020(1.60)	-	77 (0.12)	0	-	34(0.05)	0	-	51 (0.08)	0	-	-	-	HbQ India trait--5 HbD Iran trait--5 δβ thal/HPFH trait-- 46	29
3	Mumbai	Not Mentioned	5001	133(2.70)	0	28 (0.60)	0	0	6 (0.10)	0	0	3 (0.5)	0	0	5 (0.10)	9 (0.18)	2	6
4	Sangli	Not Mentioned	1279	38 (3.10)	-	-	-	-	-	-	-	-	-	-	-	-	-	30
5	Pune	Not Mentioned	4335*	458(10.56)	24 (0.55)	54 (1.25)	5 (0.12)	21(0.48)	37(0.85)	20(0.46)	3 (0.07)	9 (0.21)	4 (0.10)	3 (0.07)	0	29(0.66)	β-thal intermedi–20, HbQ India trait--4	31

* Includes antenatal woman & microcytic, hypochromic anemic individuals.

Table 8: Newborn screening

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	BTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Nagpur*	Not Mentioned	1162	0	0	536(46.10)	88 (7.50)	4 (0.39)	0	0	0	0	0	0	0	0	HbSD Punjab--2	32
2	Nagpur*	Not Mentioned	1534	-	-	704(45.80)	88 (5.73)	-	-	-	-	-	-	-	-	-	α Chain variant--3 (Hb Koya Dora, Hb Fonatineble-au and HbO Indonesia), HbS-β thal/ HbSD Punjab --8	33

(contd.)

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
3	Nagpur*	Not Mentioned	2134	4 (0.18)	0	978(45.80)	104(4.80)	7 (0.32)	5 (0.23)	0	0	0	0	0	1 (0.04)	0	HbS-δβ--1 , HbH disease --1, α chain variants-3 (Hb Koya Dora, Hb Fonatineble-au and HbO Indonesia), HbSD Punjab--2	34

*Targeted Newborn screening

Table 9: Hospital based studies

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Pune	Navbudha, Maratha, Muslim, Brahmin, Gujarati, Dhangar, Kunbi, Mang, Madari, Pawara, Koshthi, Bengali,Lingayat,Gargi	1291	91 (7.00)	76 (5.90)	0	30 (2.30)	0	0	8 (0.60)	0	0	2 (0.20)	0	0	0	0	35
2	Not Mentioned	Not Mentioned	900	-	-	-	-	-	0	14(1.20)	-	2 (0.20)	0	-	-	-	HbSE disease - 1	36
3	Dhule, Nashik, Nandurbar, Jalgaon	Not Mentioned	24732	183(0.74)	3 (0.01)	2253(9.11)	270(1.09)	10 (0.04)	3 (0.01)	0	1 (0.01)	3 (0.01)	0	0	0	0	0	37
4	Dhule, Nashik, Nandurbar, Jalgaon	Not Mentioned	10081	228(2.30)	25 (0.20)	6364(63.10)	667(6.60)	56(0.60)	5 (0.05)	0	3 (0.03)	4 (0.04)	0	0	0	2 (0.02)	β-thal intermedia--5, HbS-HbQ India--3,HbS-HPFH-3,HbSD Punjab--2 ,	38
5	Pune	Not Mentioned	3465	129(3.70)	1 (0.02)	8 (0.20)	7 (0.20)	10(0.28)	1 (0.02)	0	0	0	1 (0.02)	0	0	3 (0.08)	HbSD Punjab -- 1, HbSC- 1	39

(contd.)

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
6	Akola	Not Mentioned	91	-	-	-	49(53.80)	36(39.56)	-	-	-	-	-	-	-	-	HbSD Punjab--6,α-deletion-8	40
7	Mumbai	Not Mentioned	65779	7377 (11.21)	527 (0.80)	1324 (2.01)	1049 (1.59)	0	521 (0.79)	221 (0.34)	0	317 (0.48)	62 (0.09)	0	-	-	HbQ India trait- 50 HbJ trait-46,HPFH /δβ-thal trait –98*HbC trait–36, *HbC disease--6, *HbSC disease--12, HbSD Punjab --32, HbSE disease--2, HbH disease--24	41
8	Not Mentioned	Not Mentioned	31075	8512 (27.39)	1484 (4.77)	2156 (6.93)	676 (2.17)	510 (1.64)	385 (1.23)	63 (0.20)	165 (0.53)	129 (0.41)	10 (0.03)	23 (0.07)	-	-	HbSE disease -- 4, HbED Punjab dise-ase--3, HbD Iran trait--15 HbD Iran- β-thal --5,HbQ India trait-15 HbQ India-β thal--12,HbQ India - HbD Punjab--1,β-δβ-thal--7,δβ thal trait / HPFH trait–209,Hb Lepore trait--11,Hb Lepore-β thal --5, HbE-Lep-ore-1, HbSD Punjab--19 δβ-thal ho-mozygous-3	42

*All the cases showing HbC were of African origin.

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Gujarat

Gujarat is a state on the western coast of India and having a population of 60.4 million (Census 2011) with scheduled tribes accounting for 14.75 % of the population mainly inhabiting districts in South Gujarat. Gujarat is bordered by Rajasthan to the northeast, Dadra and Nagar Haveli and Daman and Diu to the south, Maharashtra to the southeast, Madhya Pradesh to the east, and the Arabian Sea and the Pakistani province of Sindh to the west . Many Sindhis settled here following the Partition of India in 1947. The Kolis are also a predominant group making up 24% of the total population of the state. A significant number of people have migrated to Mumbai and Dadra and Nagar Haveli as well as to many other countries, a large population of Gujarat being industrialists, businessmen and traders.

Of the 28 studies available, 18 were population based, 3 were on antenatal screening, one was on population and antenatal screening, one was on newborn screening and 6 were hospital based studies. Fig 3 shows that studies have been reported from majority of the districts (shaded areas) in the state. Three studies did not mention the names of the districts covered.

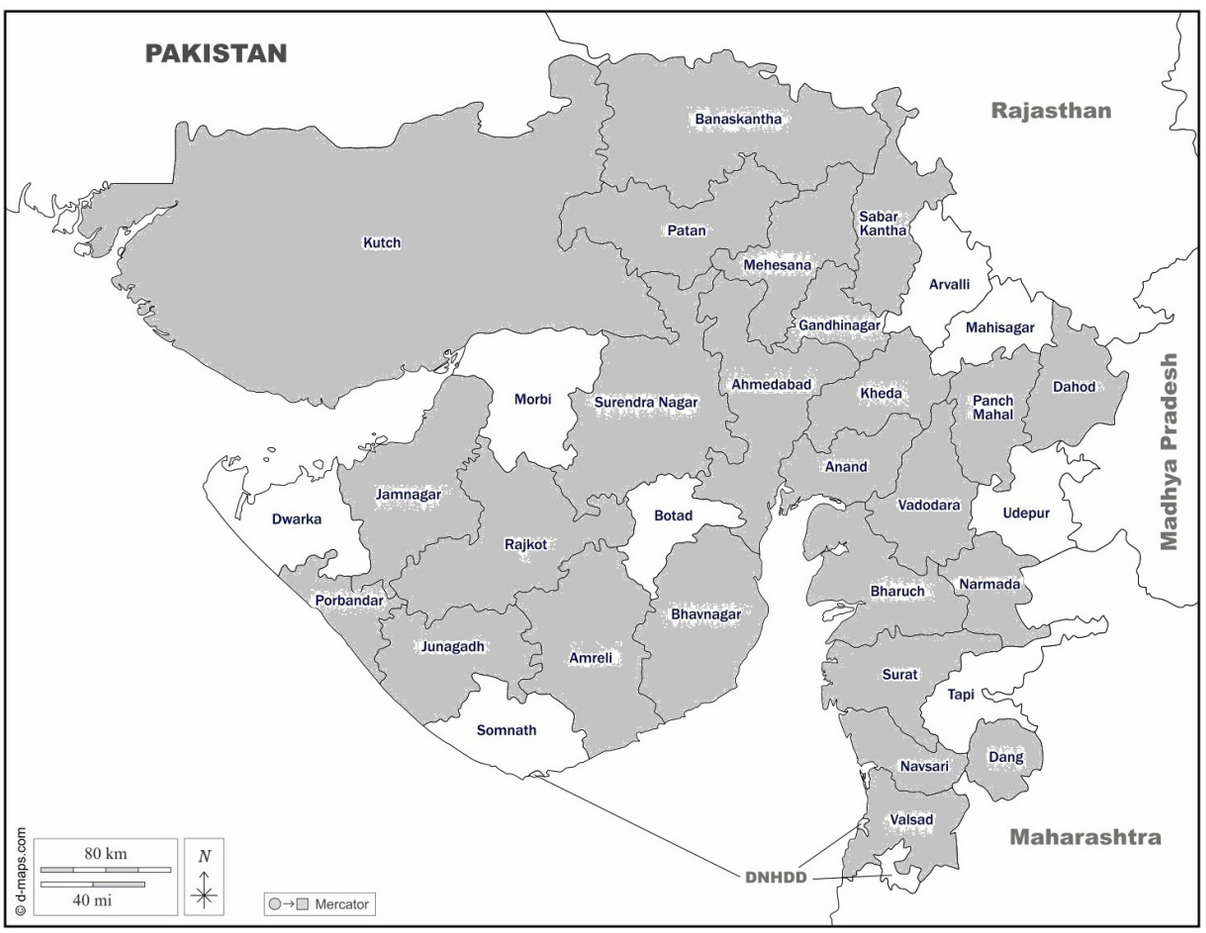

Fig 3: Map of Gujarat showing the studied areas

The number of districts covered along with the population groups (non-tribal and tribal) studied are shown in Tables 10 -12. A total of 27 non-tribal and 28 tribal groups were studied from 25 districts.

Table 10: Districts covered in Gujarat

Ahmedabad	Amrelli	Anand	Banaskantha	Bharuch
Bhavnagar	Dahod	Dang	Gandhinagar	Jamnagar
Junagadh	Kheda	Kutch	Mehsana	Narmada
Navsari	Pachmahal	Patan	Porbandar	Rajkot
Sabarkantha	Surat	Surendranagar	Vadodara	Valsad

Table 11: Non-Tribal populations studied

Barod	Bhakta	Bhanusali	Bhavasar	Brahmin
Chamar	Chouhan	Ghanchi	Golarana	Harijan
Kachhia	Khatri	Lohana	Mahiya Vanshi	Maratha
Modh Baniya	Muslim	Parmar	Patel	Patil
Prajapati	Rabari	Rajput	Rohit	Sindhi
Solanki	Vankar			

Table 12: Trbal populations studied

Bamaniya	Baria	Bhagat	Bhil	Bhilala
Chaudhuri	Dhanka	Dhodia Patel	Dubla	Gamit
Gharasiya	Halpati	Kadva	Kathodi	Kokana
Kolcha	Koli	Kotvadia	Leva	Naik
Naika	Paradhi	Patelia	Rathva	Tadvi
Vanjara	Vasava	Warli		

There are reports of some very large screening programmes from Gujarat among non-tribal and tribal populations as well as among antenatal women. The presence of HbS and β thalassaemia gene was observed in both the tribal and non tribal populations. HbS was mainly found in South Gujarat (Vadodara, Bharuch, Surat, Valsad, Dang).

Among the non-tribal populations, the β thalassaemia trait (βTT) frequency varied from 0.70 to 15.59 % in different studies while the prevalence of sickle cell trait ranged from 0.15 to 6.30 % and HbD trait from 0.10 to 1.70 % in a few studies. Occasionally, HbE, δβ thalassaemia trait, HPFH trait and some rare Hb variants have been reported (Table 13). HbS has been mainly observed among the tribal populations with the prevalence of HbS carriers and HbS homozygotes varying from 0.10 to 46.66% and 0.04 to 10.00% respectively. Few reports on β thalassaemia among tribal groups showed a very variable prevalence of βTT ranging from 1.29 to 15.90 % showing that β thalassemia is also prevalent in some tribal populations. Sporadic cases of HbD trait and δβ thalassaemia trait have also been reported during population screening (Table 14). Antenatal screening identified individuals with βTT, AS, SS, AE, AD, E-β thalassaemia and δβ-thalassaemia trait along with rare Hb variants (Table 15). There is only one study on newborn screening targeting largely tribal mothers from south Gujarat where the prevalence of AS babies varied from 7.30 to 22.90% and SS babies from 0.20 to 1.80% in different tribal groups (Table 16). In the hospital based studies, apart from β thalassaemia and the common Hb variants HbS, HbE and HbD, few rare Hb variants like Hb Q India, Hb Lepore, Hb SD disease and HbSE disease were also identified (Table 17).

Table 13: Population studies in Non- Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Vadodara	Not Mentioned	4996	133 (2.66)	0	125 (2.50)	2 (0.24)	0	5 (0.10)	0	0	19 (0.38)	1 (0.02)	0	3 (0.06)	1 (0.03)	7	1
2	Valsad	Not Mentioned	200	7 (3.50)	0	0	0	0	0	0	0	0	0	0	0	0	0	2
3	Dang	Not Mentioned	141	4 (2.80)	0	0	0	0	0	0	0	0	0	0	0	0	0	2
4	Navasari	Not Mentioned	137	3 (2.20)	0	0	0	0	0	0	0	0	0	0	0	0	0	2
5	Surat	Not Mentioned	552	12 (2.20)	0	0	0	0	0	0	0	0	0	0	0	0	0	2
6	Bharuch	Not Mentioned	432	9 (2.10)	0	0	0	0	0	0	0	0	0	0	0	0	0	2
7	Narmada	Not Mentioned	41	0	0	0	0	0	0	0	0	0	0	0	0	0	0	2
8	Anand	Not Mentioned	300	10 (3.30)	0	0	0	0	0	0	0	0	0	0	0	0	0	2
9	Ahmedabad	Not Mentioned	504	18 (3.60)	0	0	0	0	0	0	0	0	0	0	0	0	0	2
10	Vadodara	Not Mentioned	2194	67 (3.10)	0	0	0	0	0	0	0	0	0	0	0	0	0	2
11	Kheda	Not Mentioned	353	13 (3.70)	0	0	0	0	0	0	0	0	0	0	0	0	0	2
12	Gandhinagar	Not Mentioned	51	1 (2.00)	0	0	0	0	0	0	0	0	0	0	0	0	0	2
13	Panchmahal	Not Mentioned	258	10 (3.90)	0	0	0	0	0	0	0	0	0	0	0	0	0	2
14	Dahod	Not Mentioned	119	4 (3.70)	0	0	0	0	0	0	0	0	0	0	0	0	0	2
15	Porbandar	Not Mentioned	105	10 (9.50)	0	0	0	0	0	0	0	0	0	0	0	0	0	2
16	Jamnagar	Not Mentioned	185	6 (3.30)	0	0	0	0	0	0	0	0	0	0	0	0	0	2
17	Rajkot	Not Mentioned	309	16 (5.20)	0	0	0	0	0	0	0	0	0	0	0	0	0	2
18	Surendranagar	Not Mentioned	92	5 (5.40)	0	0	0	0	0	0	0	0	0	0	0	0	0	2
19	Amrelli	Not Mentioned	144	9 (6.30)	0	0	0	0	0	0	0	0	0	0	0	0	0	2
20	Bhavnagar	Not Mentioned	379	17 (4.50)	0	0	0	0	0	0	0	0	0	0	0	0	0	2
21	Junagad	Not Mentioned	304	25 (8.20)	0	0	0	0	0	0	0	0	0	0	0	0	0	2
22	kutch	Not Mentioned	603	27 (4.50)	0	0	0	0	0	0	0	0	0	0	0	0	0	2
23	Patan	Not Mentioned	74	2 (2.70)	0	0	0	0	0	0	0	0	0	0	0	0	0	2
24	Banaskantha	Not Mentioned	74	0	0	0	0	0	0	0	0	0	0	0	0	0	0	2
25	Mehshana	Not Mentioned	307	4 (1.30)	0	0	0	0	0	0	0	0	0	0	0	0	0	2
26	Sabarkantha	Not Mentioned	146	1 (0.70)	0	0	0	0	0	0	0	0	0	0	0	0	0	2

(contd.)

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
27	Ahmedabad, Anand, Gandhinagar, Bhavnagar, Amreli, Jamnagar, Kheda, Kutch, Navsari, Mehsana, Patan, Rajkot, Surat, Porbandar, Surendran-agar, Valsad	Barot, Brahmin, Bhanusali, Bhakta, Bhavsar, Lohana, Patel, Parmar, Patil, Rajput, Rabari, Chamar, Chauhan, Harijan, Solanki, Vankar	149044	3261(2.18)	0	1604(1.10)	0	0	0	0	0	0	0	0	0	0	0	3
28	Surat	Kachhia	1173	71 (6.00)	0	2 (0.17)	0	0	-	-	-	-	-	-	-	-	-	4
29	Surat	Modh Baniya	2249	140 (6.20)	0	5 (0.22)	0	0	-	-	-	-	-	-	-	-	-	4
30	Surat	Prajapati	1283	81 (6.30)	-	3 (0.15)	0	0	0	0	-	0	0	-	-	-	-	5
31	Surat	Khatri	1239	15 (1.20)	0	2 (0.16)	0	0	0	0	-	0	0	-	-	-	-	5
32	Surat	Maratha	1016	25 (2.50)	0	11 (1.20)	0	0	0	0	-	0	0	-	-	-	-	5
33	Surat	Rajput	988	44 (4.50)	0	11 (1.10)	0	0	0	0	-	0	0	-	-	-	-	5
34	Surat	Ghanchi	930	58 (6.20)	0	2 (0.20)	0	0	0	0	-	0	0	-	-	-	-	5
35	Surat	Gola-Rana	632	20 (3.20)	0	0	0	0	0	0	-	0	0	-	-	-	-	5
36	Surat	Mahiyavanshi	609	42 (6.90)	0	26 (4.30)	0	0	0	0	-	0	0	-	-	-	-	5
37	Surat	Lohana	185	20(10.80)	0	1 (0.50)	0	0	0	0	-	0	0	-	-	-	-	5
38	Surat	Sindhi	127	13 (10.20)	0	1 (0.80)	0	0	0	0	-	0	0	-	-	-	-	5
39	Surat	Rohit	127	8 (6.30)	0	8 (6.30)	0	0	0	0	-	0	0	-	-	-	-	5
40	Surat	Muslim	4870	139(2.80)	0	59 (1.20)	2 (0.04)	2 (0.04)	41(0.85)	0	-	6 (0.12)	0	-	2 (0.04)	3 (0.06)	HbD Iran trait--2, HbSD disease--1	6
41	Bharuch	Ganchi	120	8 (6.70)	-	0	0	0	-	-	-	2 (1.70)	0	-	-	-	-	7
42	Jamnagar	Sindhi	109	17(15.59)	-	4 (3.66)	0	0	-	-	-	0	0	-	-	-	-	8

(contd.)

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
43	Not Mentioned	Not Mentioned	*1000	41 (4.10)	0	3 (0.30)	0	0	0	0	0	1 (0.10)	0	0	-	-	-	9
44	Rajkot	Not Mentioned	1000	78 (7.80)	0	4 (0.40)	0	0	1 (0.10)	0	0	6 (0.60)	0	0	-	-	δβ thal trait/ HPFH trait - 1	10

* Medical students

Table 14: Population studies in Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Surat	Naik	174	-	-	28(16.00)	0	-	-	-	-	-	-	-	-	-	-	11
2	Panchmahal	Bhil	206	-	-	32(15.53)	0	-	-	-	-	-	-	-	-	-	-	11
3	Surat	Gamit	207	-	-	65(31.40)	0	-	-	-	-	-	-	-	-	-	-	11
4	Bharuch	Dhanka (Bhil)	215	-	-	44(20.47)	0	-	-	-	-	-	-	-	-	-	-	11
5	Surat	Dhodia	213	-	-	38(17.84)	0	-	-	-	-	-	-	-	-	-	-	11
6	Surat	Dubla	211	-	-	20 (9.48)	0	-	-	-	-	-	-	-	-	-	-	11
7	Surat	Koli	182	-	-	8 (4.39)	0	-	-	-	-	-	-	-	-	-	-	11
8	Valsad	Naik	12	-	-	3 (25.00)	0	-	-	-	-	-	-	-	-	-	-	12
9	Panchmahal	Bhil	200	-	-	27(13.50)	0	-	-	-	-	-	-	-	-	-	-	12
10	Kutch	Bhil	32	-	-	0	0	-	-	-	-	-	-	-	-	-	-	12
11	Panchmahal	Patelia	15	-	-	4 (26.66)	0	-	-	-	-	-	-	-	-	-	-	12
12	Junagadh	Koli	78	-	-	1 (1.28)	0	-	-	-	-	-	-	-	-	-	-	12
13	Kutch	Paradhi	14	-	-	0	0	-	-	-	-	-	-	-	-	-	-	12
14	Kutch	Koli	45	-	-	0	0	-	-	-	-	-	-	-	-	-	-	12
15	Bharuch	Koli	13	-	-	0	0	-	-	-	-	-	-	-	-	-	-	12
16	Bharuch	Rohit	14	-	-	1 (7.14)	0	-	-	-	-	-	-	-	-	-	-	12
17	Surat	Koli	12	-	-	0	0	-	-	-	-	-	-	-	-	-	-	12
18	Surat	Rohit	31	-	-	1 (3.22)	0	-	-	-	-	-	-	-	-	-	-	12
19	Surat	Kotwalia	71	-	-	13(18.31)	0	-	-	-	-	-	-	-	-	-	-	12

(contd.)

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
20	Surendra Nagar	Vanjara	23	-	-	1 (4.35)	0	-	-	-	-	-	-	-	-	-	-	12
21	Valsad	Kotwalia	8	-	-	0	0	-	-	-	-	-	-	-	-	-	-	12
22	Valsad	Kokana	58	-	-	5 (8.62)	0	-	-	-	-	-	-	-	-	-	-	12
23	Valsad	Koli	36	-	-	5 (13.89)	0	-	-	-	-	-	-	-	-	-	-	12
24	Valsad	Rohit	8	-	-	1 (12.50)	0	-	-	-	-	-	-	-	-	-	-	12
25	Dang	Kokana	128	-	-	10 (7.81)	0	-	-	-	-	-	-	-	-	-	-	12
26	Valsad	Dhodia Patel	789	17 (2.10)	-	179(22.70)	6 (0.70)	-	-	-	-	-	-	-	-	-	-	13
27	Valsad	Kokna	132	3 (2.20)	-	7 (5.30)	1 (0.70)	-	-	-	-	-	-	-	-	-	-	13
28	Valsad	Koli	103	2 (1.90)	-	9 (8.70)	0	-	-	-	-	-	-	-	-	-	-	13
29	Valsad	Kunkana, Warli, Kolcha, Kotwadia, Nayka, Dhodia	456	-	-	0	46(10.00)	-	-	-	-	-	-	-	-	-	-	14
30	Surat	Vasava	759	-	-	201(26.48)	17 (2.23)	-	-	-	-	-	-	-	-	-	-	15
31	Surat	Chaudhari	49	-	-	15(30.61)	2 (4.08)	-	-	-	-	-	-	-	-	-	-	15
32	Surat	Gamit, Dhodia	15	-	-	7 (46.66)	0	-	-	-	-	-	-	-	-	-	-	15
33	Surat	Dhodia Patel	1155	25 (2.10)	-	162(14.00)	8 (0.69)	-	-	-	-	-	-	-	-	-	-	4
34	Banaskantha, Vadodara, Bharuch, Dahod, Narmada, Panchmahal, Sabarkantha	Adivasi, Bamaniya, Bariya, Gamit, Koli, Nayka,Rathva, Tadvi	168495	2935(1.74)	2 (0.01)	19154 (11.37)	966(0.57)	0	0	0	0	0	0	0	0	0	HBD, HBE - 167	3
35	Surat	Gamit	176	28(15.90)	0	40(22.70)	0	0	0	0	0	0	0	-	-	-	-	5
36	Surat	Chaudhary	372	47(12.60)	0	83(22.30)	0	0	0	0	0	0	0	-	-	-	-	5
37	Surat	Vasava	125	17(13.60)	0	19(15.20)	0	0	0	0	0	0	0	-	-	-	-	5
38	Surat	Leva	8530	416(4.90)	0	7 (0.10)	0	0	0	0	0	5 (0.60)	0	-	-	-	-	5
39	Surat	Koli	2531	97 (3.80)	0	58 (2.30)	1 (0.04)	0	0	0	0	0	0	-	-	-	-	5
40	Surat	Kadva	1695	28 (1.70)	0	5 (2.30)	0	0	0	0	0	0	0	-	-	-	-	5
41	Surat	Dhodia	325	12 (3.70)	0	53(16.30)	1 (0.30)	0	0	0	0	0	0	-	-	-	-	5

(contd.)

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
42	Surat	Kukana	34	5 (14.70)	0	3 (8.80)	0	0	0	0	0	0	0	-	-	-	-	5
43	Surat	Others	85	13 (15.30)	0	3 (3.50)	0	0	0	0	0	0	0	-	-	-	-	5
44	Surat	Not Mentioned	7307	-	-	1740 (23.70)	112(1.53)	-	-	-	-	-	-	-	-	-	-	16
45	Not Mentioned	Kotvadia	838	21 (2.50)	0	86 (10.26)	8 (0.95)	0	0	0	0	0	0	0	2 (0.23)	0	0	17
46	Not Mentioned	Kolcha	1831	56 (3.05)	0	97 (5.29)	0	0	0	0	0	2 (0.10)	0	0	8 (0.43)	0	0	17
47	Not Mentioned	Kathodi	464	6 (1.29)	0	49 (10.56)	1 (0.21)	0	0	0	0	0	0	0	0	0	0	17
48	Vadodara	Bhil, Dhodiya Patel, Vasava, Rathva, Chaudhari, Gamit, Kukna, Gharasiya, Bhagat,Patelia	77	1 (1.29)	-	15 (19.4)	1 (1.29)	-	-	-	-	-	-	-	-	-	-	18

Table 15: Antenatal screening

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Vadodara	Not Mentioned	4995	135(2.70)	0	169 (3.40)	12 (0.20)	0	7 (0.10)	0	1 (0.02)	15 (0.30)	0	1 (0.02)	3 (0.10)	8 (0.20)	2	1
2	Surat	Hindu, Jain, Muslim	3009	102(3.38)	0	46 (1.50)	0	0	7(0.23)	0	0	11 (0.36)	0	0	0	0	HbD Iran trait--2 HbJ Paris trait--3 HbQ India trait-- 2 HbO Indonesia trait --1	19
3	Ahmedabad	*SC,ST,OBC	111426	515(0.46)	0	30 (0.02)	0	0	0	0	0	0	0	0	6 (0.01)	0	0	20
4	Bharuch	Not Mentioned	10519	-	-	1645(15.60)	131(1.20)	-	-	-	-	-	-	-	-	-	-	21

*Scheduled Caste, Scheduled Tribe, Other Backward Classes.

Table 16: Newborn screening

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Valsad, Navsari, Dang, Surat,	Warli	109	-	-	25(22.90)	1 (0.90)	0	-	-	-	-	-	-	-	-	-	22
2	Valsad, Navsari, Dang, Surat,	Bhil	23	-	-	5 (21.70)	0	0	-	-	-	-	-	-	-	-	-	22
3	Valsad, Navsari, Dang, Surat,	Chaudhry	85	-	-	17 (20.00)	0	0	-	-	-	-	-	-	-	-	-	22
4	Valsad, Navsari, Dang, Surat,	Dhodia Patel	1623	-	-	301(18.50)	19 (1.10)	6 (0.30)	-	-	-	-	-	-	-	-	-	22
5	Valsad, Navsari, Dang, Surat,	Naika	554	-	-	82 (14.80)	4 (0.70)	2 (0.30)	-	-	-	-	-	-	-	-	-	22
6	Valsad, Navsari, Dang, Surat,	Koli	55	-	-	7 (12.70)	1 (1.80)	0	-	-	-	-	-	-	-	-	-	22
7	Valsad, Navsari, Dang, Surat,	Kukna	1216	-	-	139(11.40)	3 (0.20)	2 (0.10)	-	-	-	-	-	-	-	-	-	22
8	Valsad, Navsari, Dang, Surat,	Kolcha	109	-	-	8 (7.30)	0	0	-	-	-	-	-	-	-	-	-	22
9	Valsad, Navsari, Dang, Surat,	Halpati	716	-	-	56 (7.80)	5 (0.70)	1 (0.10)	-	-	-	-	-	-	-	-	-	22

Table 17: Hospital based studies

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Ahmedabad	Not Mentioned	428	70 (16.35)	32 (7.48)	12 (2.80)	22 (5.14)	3 (0.70)	2 (0.46)	1(0.23)	2 (0.46)	2 (0.46)	1 (0.23)	0	3 (0.70)	0	HbSD Punjab - 1, β-thal intermedia-2	23
2	Not Mentioned	Not Mentioned	7261	839(11.50)	292(4.02)	214(2.95)	85 (1.17)	52(0.72)	11(0.15)	10(0.14)	4 (0.06)	47 (0.65)	11(0.15)	9 (0.12)	0	8 (0.11)	β-thal intermedia - 16, HbQ India trait-4, Hb Lepore-2, HbDE-3, HbSD Punjab- 8	24
3	Bharuch	Scheduled tribe	7832	-	-	-	182(2.32)	-	-	-	-	-	-	-	-	-	-	25
4	Mixed states	Not Mentioned	*75	49(65.33)	9 (12.00)	-	-	-	6 (8.00)	2 (2.66)	1 (1.34)	-	-	1 (1.34)	-	-	-	26
5	Vadodara	Bariya, Rathva, Parmar, Adivasi, Bhilala	#1360	-	-	1160 (85.20)	173 (12.72)	26(1.95)	-	-	-	-	-	-	-	-	HbSE disease - 1	27
6	Vadodara	Not Mentioned	5624	20 (0.36)	5 (0.09)	681(12.10)	272(4.80)	38(0.68)	0	0	-	2 (0.03)	0	-	-	-	HbSD Punjab - 2, HbSE-1, β-thal intermedia-1	28

*Samples include individuals from Gujarat, Rajasthan, Maharashtra, Assam, West Bengal.
#Solubility positive cases.

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Rajasthan

Rajasthan is the largest Indian state by area having a population of 68.5 million with scheduled tribes constituting 13.48% of the population (Census 2011). It is on the northwestern side of the country and covers a large part of the Thar Desert. It shares a border with the Pakistani provinces of Punjab to the northwest and Sindh to the west, along the Sutlej-Indus River valley. It is bordered by Punjab to the north; Haryana and Uttar Pradesh to the northeast; Madhya Pradesh to the southeast; and Gujarat to the southwest. The native Rajasthani people make up the majority of the state's population, however, a large number of Sindhis also came here in 1947 from the Sindh province (now in Pakistan) during the India-Pakistan separation. The population comprises predominantly of Hindus, although a significant number of Muslims, Sikhs and Jains also reside here.

A total of 10 studies were available of which 8 were population based, one was on antenatal screening and one was a hospital based study. Fig 4 shows that only six districts (shaded areas) in the state have been covered in these studies. One study did not mention the names of the districts covered.



Fig 4: Map of Rajasthan showing the studied areas

The number of districts covered along with the population groups (non-tribal and tribal) studied are shown in Tables 18 - 20. A total of 15 non-tribal and 6 tribal groups were studied from 6 districts.

Table 18: Districts covered in Rajasthan

Banaskhanta (Banaswara)	Chittorgarh	Dungarpur	Jodhpur	Sirohi
Udaipur				

Table 19: Non-Tribal populations studied

Baniya	Bhoi	Brahmin	Garg	Lambana
Meghwal	Mochi	Muslim	Panchal	Patel
Patidhar	Rajput	Sompura	Suthar	Yadaw

Table 20: Tribal populations studied

Bhil	Damor	Gamit	Garasia	Koli Thakurda
Mina				

Some studies have only screened for HbS and few have also screened for β thalasemia. The presence of sickle cell gene was observed in both the non-tribal and tribal populations (Tables 21 and 22). The prevalence of sickle cell carriers (AS) in non-tribal groups was 3.18% while it varied from 0.58 to 31.14% in tribal groups. Sickle cell homozygous (SS) cases were also encountered. Few reports on β thalassemia in tribal populations showed the prevalence of βTT ranging from 2.50 to 7.60%. Antenatal screening and hospital based studies have identified βTT and HbS carriers and HbS homozygous cases. HbE, HbD and any other rare Hb variants have not been reported in any of these studies (Tables 23 and 24).

Table 21: Population studies in Non-Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Dungarpur	*Scheduled Caste General Caste, Scheduled Tribe	2922	0	0	93 (3.18)	14 (0.47)	0	2 (0.06)	0	0	12 (0.41)	0	0	0	0	HbSC--1	1
2	Not Mentioned	Not Mentioned	251	0	0	0	0	0	0	0	0	0	0	0	0	0	0	2

*Scheduled Castes include Meghwal,Lambana,Mochi,Garg,Yadaw
*General Castes include Baniya, Brahmin, Muslim, Patel, Suthar, Patidhar, Rajput, Bhoi, Sompura, Panchal.
*Scheduled Tribes include Bhil,Damor,Garasia,Mina.

Table 22: Population studies in Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Udaipur	Garasiya	35	-	-	10 (31.14)	0	-	-	-	-	-	-	-	-	-	-	3
2	Sirohi	Garasiya	75	-	-	15 (20.00)	0	-	-	-	-	-	-	-	-	-	-	3
3	Udaipur	Meena	56	-	-	0	0	-	-	-	-	-	-	-	-	-	-	3
4	Udaipur	Gamit	75	-	-	11 (14.67)	0	-	-	-	-	-	-	-	-	-	-	3
5	Sirohi	Gamit	58	-	-	2 (3.45)	0	-	-	-	-	-	-	-	-	-	-	3
6	Bamaswara	Bhil	100	-	-	18 (18.00)	0	-	-	-	-	-	-	-	-	-	-	3
7	Banaskantha (Bamaswara)	Bhil (Dumgari)	176	-	-	19 (10.80)	0	-	-	-	-	-	-	-	-	-	-	3
8	Banaskantha (Bamaswara)	Bhil (Dumgari)	69	-	-	1 (1.45)	0	-	-	-	-	-	-	-	-	-	-	3
9	Banaskantha (Bamaswara)	Koli Thakurda	14	-	-	1 (7.14)	0	-	-	-	-	-	-	-	-	-	-	3
10	Udaipur	Bhil	280	7 (2.50)	-	12 (4.28)	0	-	-	-	-	-	-	-	-	-	-	4
11	Udaipur	Not Mentioned	1198	0	0	7 (0.58)	0	0	0	0	0	3 (0.25)	0	0	0	0	0	5
12	Dungarpur	Bhil	474	27 (5.70)	-	8 (1.69)	0	0	0	0	0	0	0	0	-	-	-	6

(contd.)

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
13	Chittorgarh	Mina	481	16 (3.33)	-	39 (8.11)	0	0	0	0	0	0	0	0	-	-	-	6
14	Sirohi	Garasiya	368	28 (7.60)	1 (0.01)	22 (5.97)	1 (0.27)	2 (0.54)	0	0	0	0	0	0	-	-	-	7
15	Sirohi, Udaipur	Garasiya	1090	-	-	84 (7.70)	9 (0.77)	-	-	-	-	-	-	-	-	-	-	8

Table 23: Antenatal screening

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Jodhpur	Not Mentioned	1500	88 (5.90)	-	-	-	-	-	-	-	-	-	-	-	-	-	9

Table 24: Hospital based studies

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Sirohi	Garasiya	1676	-	-	144(8.40)	14 (0.83)	-	-	-	-	-	-	-	-	-	-	10

References:

1. Choubisa SL. Abnormal haemoglobins, thalassaemia and G-6-PD enzyme deficiency in Rajasthan (western-India). Haematologia (Budap). 1991;24:153-165.
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7. Choubisa SL. Sickle cell haemoglobin, thalassemia and G-6-PD enzyme deficiency genes in Garasiya tribe inhabited malaria endemic areas of Sirohi district, Rajasthan (India). J Community Dis. 2009; 41:13-18.
8. Mandot S, Ameta G. Prevalence, clinical, and hematological profile of sickle cell disease in south Rajasthan. Ind J Child Health. 2016; 3:248-250.
9. Gupta V , Sharma P, Jora R, Amandeep M, Kumar A. Screening for thalassemia carrier status in pregnancy and pre-natal diagnosis. Indian Pediatr. 2015; 52:808-809.
10. Mandot S, Khurana VL, Sonesh JK. Sickle cell anemia in Garasia tribals of Rajasthan. Indian Pediatr. 2009;46 : 239-240.

Goa

Goa is one of the smallest states by area with a population of 1.45 million (Census 2011). It is situated on the southwestern coast of India within the Konkan region, geographically separated from the Deccan highlands by the Western Ghats. It is surrounded by Maharashtra to the north and Karnataka to the east and south, with the Arabian Sea forming its western coast. Although in the past, the native Goans were predominantly Christians however, at present 66.1 % of the population are Hindus, 25.1 % are Christians and 8.3 % are Muslims.(Census 2011).

Only two studies, one population screening and one hospital based study are available. Fig 5 shows that individuals from only the Northern district (shaded area) in the state have been studied.

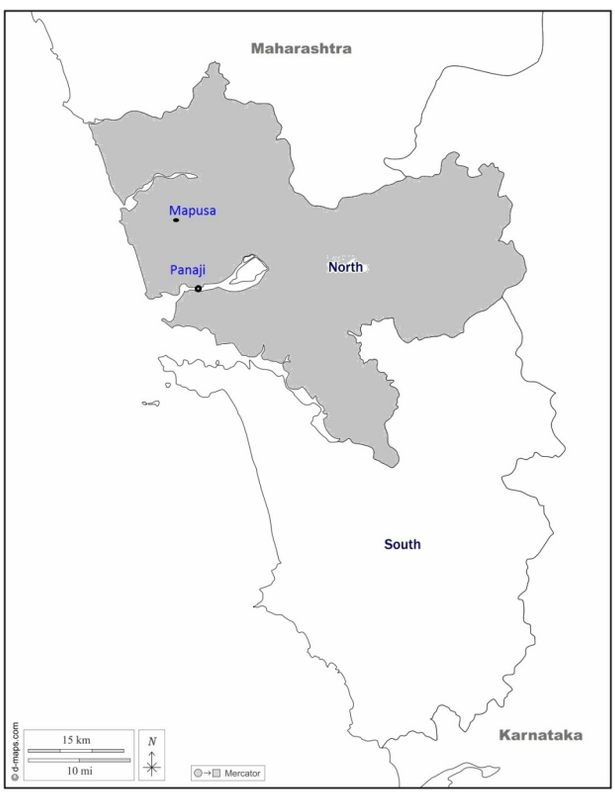


Fig 5: Map of Goa showing the studied areas

A total of five non tribal groups (Table 25) were studied from one district.

Table 25: Non-Tribal populations studied

Artisan	Brahmin	Christian	Maratha	Muslim

There are only two reports from Goa, one on population screening among non-tribals where no abnormalities were detected (Table 26) and the second being a hospital based study from North Goa where β thalassaemia, HbS, HbE, HbD, δβ thalassaemia and HPFH cases were identified (Table 27).

References:

1. Gorakshakar AC, Sathe MS, Shirsat RS, Bhatia HM. Genetic markers (A1,A2 BO, Rh D), Ina, G6PD and abnormal hemoglobins. Indian Anthropol. 1987;17:1-5.

2. Munj VS, Dias M, Gomes N. A retrospective laboratory study of the spectrum of hemoglobin disorders in north Goa district hospital. J Med Sci Clin Res. 2019;7:262-267.

Table 26: Population studies in Non- Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	North Goa (Panaji and Mapuca)	Christian	94	0	0	0	0	0	0	0	0	0	0	0	-	-	-	1
2	North Goa (Panaji and Mapuca)	Brahmin	36	0	0	0	0	0	0	0	0	0	0	0	-	-	-	1
3	North Goa (Panaji and Mapuca)	Maratha	94	0	0	0	0	0	0	0	0	0	0	0	-	-	-	1
4	North Goa (Panaji and Mapuca)	Artisan	32	0	0	0	0	0	0	0	0	0	0	0	-	-	-	1
5	North Goa (Panaji and Mapuca)	Others	18	0	0	0	0	0	0	0	0	0	0	0	-	-	-	1

Table 27: Hospital based studies

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	North Goa	Muslim, Hindu, Christians & Tribals	1084	89(8.21)	4(0.37)	7(0.68)	3 (0.28)	0	10(0.92)	0	4(0.37)	3(0.28)	0	-	3(0.28)	2(0.18)	-	2

Dadra and Nagar Haveli

Dadra and Nagar Haveli is a union territory in northwestern India. The territory was constituted through the merger of the former territories of Dadra, Nagar, Haveli, Daman and the island of Diu. The population is 0.34 million of which around 62 % are tribal communities locally known as Adivasis.

Only 4 population based studies were available. Fig 6 shows that two districts (shaded area) in the Union Territory have been studied.



Fig 6: Map of Dadra & Nagar Haveli showing the studied areas

A total of one non-tribal (Rajput) and 3 tribal groups (Table 28) were studied from two districts.

Table 28: Tribal populations studied

Dhodia	Kokna	Warli		

The presence of HbS gene was observed in both the tribal and non-tribal populations (Tables 29 and 30) and the prevalence of HbS carriers (AS) ranged from 2.98% to 19.10%. Five sickle homozygous cases were also reported in the Dhodia tribe.

Table 29: Population studies in Non- Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Dadra & Nagar Haveli	Rajput	101	-	-	3 (2.98)	0	-	-	-	-	-	-	-	-	-	-	1

Table 30: Population studies in Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Dadra & Nagar Haveli	Dhodia	183	2 (1.09)	0	35 (19.10)	0	0	0	0	0	0	0	-	-	-	-	2
2	Dadra & Nagar Haveli	Warli	490	6 (1.22)	0	36 (7.30)	0	0	0	0	0	0	0	-	-	-	-	2
3	Dadra & Nagar Haveli	Kokana	381	9 (2.36)	0	12 (3.10)	0	0	0	0	0	0	0	-	-	-	-	2
4	Dadra & Nagar Haveli	Warli	102	-	-	13(12.70)	0	0	-	-	-	-	-	-	-	-	-	3
5	Dadra & Nagar Haveli	Dhodia	149	-	-	22(14.76)	5 (3.34)	0	-	-	-	-	-	-	-	-	-	4

References:

1. Devi NA and Sachdeva MP. Sickle cell haemoglobin and Glucose-6- Phosphate Dehydrogenase deficiency among the Rajputs of Dadra and Nagar Haveli . Anthropologist. 2009;11:45-47.
2. Vasantha K, Gorakshakar AC, Pavri RS and Bhatia HM. Genetic markers in malarial endemic tribal populations of Dadra & Nagar Haveli. Proc Recent Trends in Immunohaematology. 1982:162-168.
3. Samtani R, Mondal PR, Saraswathy KN. Glucose-6-Phosphate Dehydrogenase deficiency and sickle cell hemoglobin among the Warli tribe of Dadra and Nagar Haveli. Anthropologist. 2008; 10:301-303.
4. Asghar M, Sachdeva MP. Glucose-6-Phosphate Dehydrogenase deficiency and sickle cell hemoglobin among the Dhodias of Dadra and Nagar Haveli. Anthropologist. 2009;11:61-62.

Central Region

The Central region includes two states, Chhattisgarh and Madhya Pradesh. A total of 33 articles comprising population, antenatal, newborn and hospital based studies were compiled. The number of districts covered and articles that were available for compilation in each state are shown in Table 1.

Table 1 : Studies available from the Central Region

States	Districts	No of articles
Chhattisgarh	14	13
Madhya Pradesh	12	20
Total	**26**	**33**

Almost 60% of the studies were from Madhya Pradesh and majority of the studies in both the states were on prevalence of β thalassaemia and other haemoglobinopathies among the tribal and non-tribal populations. Fig 1 shows the map with the states (shaded areas) covered in the Central region.

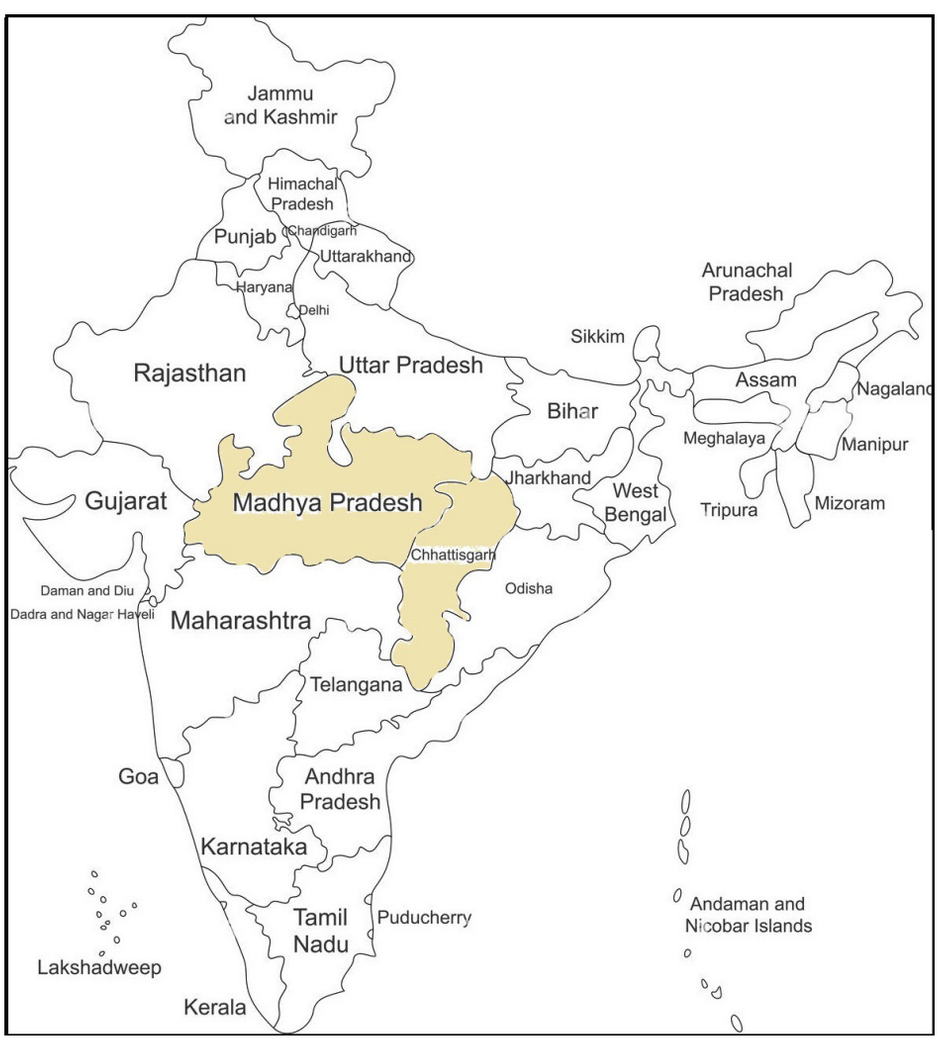
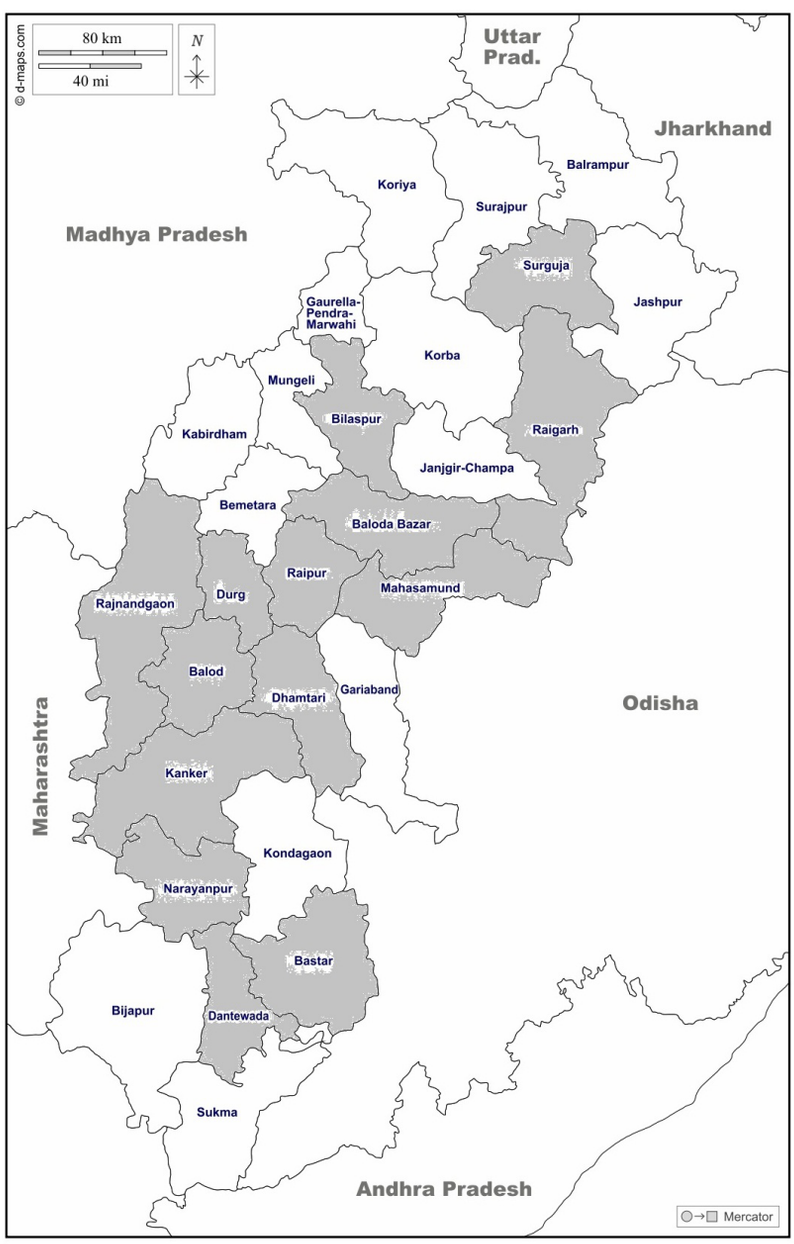


Fig 1: Map of India showing the states covered

Chhattisgarh

Chhattisgarh, formerly part of Madhya Pradesh is a heavily forested state located in Central India. It borders Uttar Pradesh to the north, Madhya Pradesh to the northwest, Maharashtra to the southwest, Jharkhand to the northeast, Odisha to the east and Telangana and Andhra Pradesh to the south. As of 2021, the state has a population of roughly 30 million, making it the 17th most populated state in the country. Chhattisgarh has one of highest percentage of scheduled tribe (ST) populations accounting for about 10% of the total scheduled tribes in India. Scheduled tribes make up 30.62% of the population and mainly inhabit the dense forests of Bastar and other districts of south Chhattisgarh. The Scheduled caste (SC) population of Chhattisgarh is 11.6 percent of the total population.

A total of 13 studies were available, of which 10 were population based and remaining three were each on antenatal screening, newborn screening and hospital based studies. Fig 2 shows that studies have been conducted in 50% of the districts (shaded areas) in the state. One study did not mention the name of the districts covered.



The number of districts covered along with the population groups (non-tribal and tribal) studied are shown in Tables 2-4. A total of 9 non tribal and 23 tribal groups were studied from 14 districts.

Table 2: Districts covered in Chhattisgarh

Balod	Baster	Bilaspur	Baloda Bazar	Dhantewada
Dhamtari	Durg	Kankar	Mahasamund	Narayanpur
Raigarh	Raipur	Rajnadgaon	Surguja	

Table 3: Non-Tribal populations studied

Agharia	Ganda	General caste	Ghasia	Kolta
Kumhar	Kurmi	Mahar	Sahu	

Table 4: Trbal populations studied

Abhuj Maria	Bada Bhatra	Bhatra	Bhinjwar	Bishonhorn Maria
Dhanwar	Dorla	Gana	Gahira	Gharsia
Gond	Halba	Hill Maria	Kanwar	Kol
Manjhwar	Manjole Bhatra	Muria	Oraon	Pando
Panka	Raj Gond	San Bhatra		

Population based studies indicated the presence of HbS gene in most of the tribal and non-tribal populations studied (Tables 5 and 6). The prevalence of sickle cell trait varied from 5.65 to 10.70% among the non-tribal populations and in the tribal populations, sickle cell trait ranged from 0.90 to 27.70 % while sickle homozygotes (Hb SS) from 0.14 to 2.40 % in different studies. Two studies reported the presence of β thalassaemia trait, HbSE and HbSD cases. Antenatal and newborn screening along with hospital based studies have picked up HbAS and HbSS cases (Tables 7 – 9).

Table 5: Population studies in Non- Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D β-thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Raipur	Agharia, Kolta, Kumhar	65177	-	-	4996 (7.67)	138 (0.21)	-	-	-	-	-	-	-	-	-	-	1
2	Raipur	Ghasia Ganda, Mahar	220041	-	-	22595 (10.27)	475 (0.22)	-	-	-	-	-	-	-	-	-	-	1
3	Raipur	General Caste	29152	-	-	1648 (5.65)	40 (0.14)	-	-	-	-	-	-	-	-	-	-	1
4	Not mentioned	Mixed Population	261	10 (3.80)	-	34 (13.02)		-	-	-	-	-	-	-	-	-	*α- thal--79	2
5	Durg	Sahu&Kurmi	982	-	-	75 (7.63)	16 (1.62)	-	-	-	-	-	-	-	-	-	-	3
6	Rajnandgaon	Sahu&Kurmi	767	-	-	44 (5.73)	8(1.04)	-	-	-	-	-	-	-	-	-	-	3

* Total 230 samples with low indices have been screened.

Table 6: Population studies in Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D β-thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Bilaspur	Kanwar	91	-	-	4(4.40)	0	-	-	-	-	-	-	-	-	-	-	4
2	Bilaspur	Gond	129	-	-	25(19.38)	0	-	-	-	-	-	-	-	-	-	-	4
3	Bastar	Dhanwar	348	-	-	12(3.45)	0	-	-	-	-	-	-	-	-	-	-	5
4	Bastar	Dhurwar (N)	60	-	-	10(16.67)	0	-	-	-	-	-	-	-	-	-	-	5
5	Bastar	Hill Maria	85	-	-	17(20.00)	0	-	-	-	-	-	-	-	-	-	-	5
6	Bastar	Bisonhorn Maria	442	-	-	82(18.55)	0	-	-	-	-	-	-	-	-	-	-	5
7	Bastar	Manjole Bhatra	64	-	-	7(10.94)	0	-	-	-	-	-	-	-	-	-	-	5
8	Bastar	Bade Bhatra	153	-	-	25(16.34)	0	-	-	-	-	-	-	-	-	-	-	5
9	Bastar	Muria (W)	169	-	-	27(15.98)	0	-	-	-	-	-	-	-	-	-	-	5
10	Bastar	San Bhatra	88	-	-	17(19.32)	0	-	-	-	-	-	-	-	-	-	-	5
11	Bastar	RajGond	70	-	-	10(14.29)	0	-	-	-	-	-	-	-	-	-	-	5

(contd.)

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D β-thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
12	Bastar	Dorla	200	-	-	26(137.00)	07	-	-	-	-	-	-	-	-	-	-	5
13	Bastar	Muria (E)	143	-	-	15(10.49)	0	-	-	-	-	-	-	-	-	-	-	5
14	Bastar	Halba	77	-	-	21(27.27)	0	-	-	-	-	-	-	-	-	-	-	5
15	Surguja	Kanwar	100	-	-	0	0	-	-	-	-	-	-	-	-	-	-	5
16	Bilaspur	Oraon	6	-	-	0	0	-	-	-	-	-	-	-	-	-	-	5
17	Bilaspur	Kol	10	-	-	0	0	-	-	-	-	-	-	-	-	-	-	5
18	Bilaspur	Pando	10	-	-	0	0	-	-	-	-	-	-	-	-	-	-	5
19	Bilaspur	Panka (mixed)	35	-	-	1(2.85)	0	-	-	-	-	-	-	-	-	-	-	5
20	Bilaspur	Dhanwar	31	-	-	0	0	-	-	-	-	-	-	-	-	-	-	5
21	Bilaspur	Manjhwar	17	-	-	0	0	-	-	-	-	-	-	-	-	-	-	5
22	Bilaspur	Bhinjwar	13	-	-	1(7.69)	0	-	-	-	-	-	-	-	-	-	-	5
23	Surguja	Gond	127	-	-	26(20.47)	1 (0.80)	-	-	-	-	-	-	-	-	-	-	6
24	Surguja	Oraon	422	-	-	9(2.19)	2 (0.50)	-	-	-	-	-	-	-	-	-	-	6
25	Surguja	Kanwar	114	-	-	1(0.90)	0	-	-	-	-	-	-	-	-	-	-	6
26	Raipur	Gond	157	-	-	25 (15.90)	1 (0.60)	-	-	-	-	-	-	-	-	-	-	6
27	Raipur	Kawar	72	-	-	4 (5.50)	0	-	-	-	-	-	-	-	-	-	-	6
28	Raipur	Halba	122	-	-	17(13.90)	0	-	-	-	-	-	-	-	-	-	-	6
29	Narayanpur, Kanker	Gond	433	-	-	43(9.90)	0	-	-	-	-	-	-	-	-	-	-	7
30	Narayanpur, Kanker	Halba	116	-	-	12(10.30)	2 (1.70)	-	-	-	-	-	-	-	-	-	-	7
31	Narayanpur, Kanker	Abhuj Maria	147	-	-	35(23.80)	1 (0.70)	-	-	-	-	-	-	-	-	-	-	7
32	Narayanpur, Balod,Kanker	Not mentioned	1054	-	-	213(20.20)	13(1.23)	-	-	-	-	-	-	-	-	-	-	8
33	Bastar	Muria	101	-	-	15(14.80)	1 (0.90)	-	-	-	-	-	-	-	-	-	-	9
34	Bastar	Bhatra	102	-	-	10(9.80)	2 (1.90)	-	-	-	-	-	-	-	-	-	-	9
35	Bastar	Maria	94	-	-	19(20.20)	2 (2.10)	-	-	-	-	-	-	-	-	-	-	9
36	Bastar	Halba	99	-	-	12(12.10)	2 (2.00)	-	-	-	-	-	-	-	-	-	-	9
37	Bastar	Dhurwar	81	-	-	5 (6.10)	2 (2.40)	-	-	-	-	-	-	-	-	-	-	97

(contd.)

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D β-thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
38	Raipur	Halba,Gond,Binjhwar	45453	-	-	4228(9.30)	94 (0.21)	-	-	-	-	-	-	-	-	-	-	1
39	Kanker, Dantewada Raigarh	Not mentioned	15701*	-	-	1672(10.60)	22 (0.14)	12(0.07)	-	-	-	-	-	-	-	-	HbSE disease --2, HbSD Punjab--1	10

*Includes Scheduled Tribe, Scheduled Caste, Other Backward Classes & General Caste

Table 7: Antenatal screening

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D β-thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Bastar	Not Mentioned	75*	-	-	53(70.66)	20(26.66)	2 (2.60)	-	-	-	-	-	-	-	-	-	11

*Sickling Positive Cases

Table 8: Newborn screening

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D β-thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Raipur	General	112	-	-	2 (2.00)	0	-	-	-	-	-	-	-	-	-	-	12
2	Raipur	Sahu	242	-	-	20 (8.20)	1(0.50)	-	-	-	-	-	-	-	-	-	-	12
3	Raipur	Kurmi	117	-	-	11 (9.40)	1(0.80)	-	-	-	-	-	-	-	-	-	-	12
4	Raipur	Gahira	84	-	-	9 (10.70)	0	-	-	-	-	-	-	-	-	-	-	12
5	Raipur	Gharsia	101	-	-	2 (2.00)	0	-	-	-	-	-	-	-	-	-	-	12
6	Raipur	Satnami	152	-	-	8 (5.20)	0	-	-	-	-	-	-	-	-	-	-	12
7	Raipur	Mahar	114	-	-	3 (2.70)	0	-	-	-	-	-	-	-	-	-	-	12
8	Raipur	Gana	116	-	-	11 (9.50)	0	-	-	-	-	-	-	-	-	-	-	12
9	Raipur	Gond	120	-	-	2(1.70)	1(0.80)	-	-	-	-	-	-	-	-	-	-	12

Table 9: Hospital based studies

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D β-thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Bilaspur	Not Mentioned	1260	-	-	441(35.00)	56(4.40)	-	-	-	-	-	-	-	-	-	-	13

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Madhya Pradesh is the second largest Indian state by area and the fifth largest state by population with over 72 million residents. It borders the states of Uttar Pradesh to the northeast, Chhattisgarh to the southeast, Maharashtra to the south, Gujarat to the west, and Rajasthan to the northwest. The population of Madhya Pradesh consists of a number of ethnic groups and tribes, castes and communities. The scheduled castes (15.6%) and the scheduled tribes (21.1%) constitute a significant proportion of the population of the State. According to the 2011 Census, the tribal population in Madhya Pradesh was 15.34 million.

A total of 20 studies which included, 16 on population screening and of the remaining four studies, two on antenatal screening and two hospital based studies were available for compilation. Fig 3 shows that studies have been conducted in 1/4th of the districts in the state. Two studies did not mention the names of the districts covered.

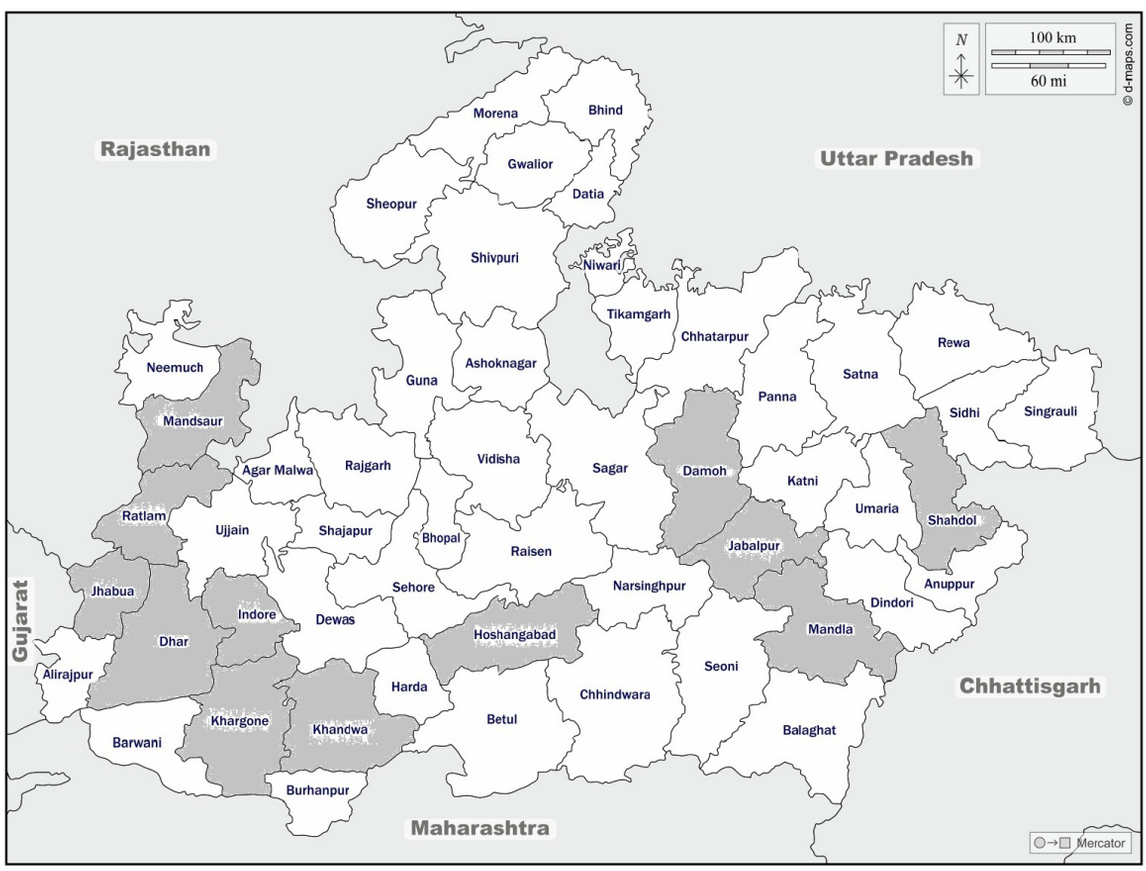

Fig 3: Map of Madhya Pradesh showing the studied areas

The number of districts covered along with the population groups (non-tribal and tribal) studied are shown in Tables 10-12. A total of 8 non-tribal and 10 tribal groups were studied from 12 districts.

Table 10: Districts covered in Madhya Pradesh

Damoh	Dhar	Hoshangabad	Indore	Jabalpur
Jhabua	Khandwa	Khargone	Mandla	Mandsaur
Ratlam	Shahdol			

Table 11: Non-Tribal populations studied

Balai	Brahmin	Chaudhury	Gujarati	Kunbi
Marwari	Punjabi	Sindhi		

Table 12 : Trbal populations studied

Baiga	Barela	Bhil	Bhilala	Gond
Korku	Panika	Pardhan	Patelia	Raj Gond

Population based studies indicated the presence of HbS and β thalassaemia gene in both the tribal and non tribal populations. HbS was found in all the districts studied. The prevalence of β thalassaemia trait (βTT) varied from 1.80 to 20.50% among the non-tribal populations with the highest frequency in Sindhis while sickle cell trait ranged from 1.40 to 14.10 % in different studies. HbD trait and HbD-β thalassaemia were also reported among the Sindhis (Table 13). Among the tribal populations, the prevalence of HbS carriers and sickle homozygotes varied from 0.98 to 31.10 % and from 0.20 to 17.5% respectively. Few studies have also screened for β thalassaemia and the prevalence of βTT ranged from 0.57 to 4.44 % (Table 14). Antenatal screening identified βTT along with Hb AS, Hb AE and Hb AD and other rare variants like Hb J and HbQ India (Table 15). Data on newborn screening was not available while hospital based studies have picked up β thalassaemia and sickle cell disease cases (Table 16).

Table 13: Population studies in Non- Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D β-thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Jabalpur	Sindhi	508	104(20.50)	1 (0.20)	0	0	0	0	0	0	12(2.40)	0	1 (0.20)	0	0	0	1
2	Shahdol	Choudhary	195	7 (3.60)	-	10 (5.10)	0	-	-	-	-	-	-	-	-	-	-	2
3	Khandwa	Balai	276	7(2.50)	-	39(14.10)	1(0.40)	-	-	-	-	-	-	-	-	-	-	2
4	Damoh	Chaudhary	168	5 (1.80)	-	4 (1.40)	-	-	-	-	-	-	-	-	-	-	-	3

Table 14: Population studies in Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D β-thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Ratlam, Mandsaur	Bhil	84	-	-	5 (5.90)	0	-	-	-	-	-	-	-	-	-	-	4
2	Indore	Bhil	46	10(1.12)	-	11(23.91)	0	-	-	-	-	-	-	-	-	-	-	5
3	Dhar	Bhil	44	-	-	5 (11.36)	0	-	-	-	-	-	-	-	-	-	-	6
4	Dhar	Bhilala	139	-	-	39(28.05)	0	-	-	-	-	-	-	-	-	-	-	6
5	Jhabua	Bhil	145	-	-	25(17.24)	0	-	-	-	-	-	-	-	-	-	-	7
6	Mandla	Pardhan	100	-	-	9(9.00)	0	-	-	-	-	-	-	-	-	-	-	8
7	Jhabua	Bhilala	116	-	-	33 (28.50)	0	-	-	-	-	-	-	-	-	-	-	8
8	Khandwa	Barela	120	-	-	27 (22.50)	0	-	-	-	-	-	-	-	-	-	-	9
9	Jhabua	Bhil	105	-	-	8 (7.62)	0	-	-	-	-	-	-	-	-	-	-	9
10	Jhabua	Patelia	103	-	-	32(31.10)	-	-	-	-	-	-	-	-	-	-	-	9
11	Hoshangabad	Korku	102	-	-	1 (0.98)	-	-	-	-	-	-	-	-	-	-	-	10
12	Jhabua	Bhils	904	-	-	183 (20.00)	8 (0.90)	-	-	-	-	-	-	-	-	-	-	11

(contd.)

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D β-thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
13	Jhabua	Bhilalas	403	-	-	123(30.50)	7 (1.70)	-	-	-	-	-	-	-	-	-	-	11
14	Jhabua	Patelias	166	-	-	34(20.50)	3 (1.80)	-	-	-	-	-	-	-	-	-	-	11
15	Ratlam	Bhils	433	-	-	51(11.50)	1 (0.20)	-	-	-	-	-	-	-	-	-	-	11
16	Khargone	Bhilalas	345	-	-	88(25.50)	3 (0.80)	-	-	-	-	-	-	-	-	-	-	11
17	Jhabua	Bhils	1106	15(1.36)*	-	209(18.90)	10 (0.90)	0	0	0	-	0	0	-	-	-	-	12
18	Jhabua	Bhilalas	429	0	-	130(30.30)	9 (2.10)	0	0	0	-	0	0	-	-	-	-	12
19	Jhabua	Patelias	176	0	-	40(22.73)	4 (2.27)	0	0	0	-	0	0	-	-	-	-	12
20	Ratlam	Bhils	421	5 (2.76)@	-	50(11.88)	1 (0.24)	0	0	0	-	0	0	-	-	-	-	12
21	Khargone	Barelas	354	9(2.33)#	-	93(26.20)	3 (0.85)	0	0	0	-	0	0	-	-	-	-	12
22	Khargone	Bhilalas	68	2 (4.44)$	-	15(22.06)	0	0	0	0	-	0	0	-	-	-	-	12
23	Mandla and Jabalpur	Gond	308	0	1 (0.32)	87(28.20)	54(17.5)	0	1(0.32)	0	1 (0.32)	1 (0.32)	0	-	-	-	-	13
24	Jabalpur	Baiga	1566	9 (0.57)	-	244(15.58)	7(0.44)	-	-	-	-	-	-	-	-	-	-	14
25	Mandla	Baiga	624	-	-	94(15.06)	2(0.34)	-	-	-	-	-	-	-	-	-	-	15
26	Mandla	Baiga	547	-	-	87(15.90)	2(0.36)	-	-	-	-	-	-	-	-	-	-	15
27	Shahdol	Panika	210	3 (1.40)	-	60 (28.6)	7(3.30)	-	-	-	-	-	-	-	-	-	-	2
28	Khandwa	Korku	301	7 (2.30)	-	50(16.90)	2(0.70)	-	-	-	-	-	-	-	-	-	-	2
29	Mandla	Gond	6190	-	-	732(11.90)	0	-	-	-	-	-	-	-	-	-	-	16
30	Damoh	RajGond	267	9 (3.40)	-	27(10.10)	1(0.40)	-	-	-	-	-	-	-	-	-	-	3

* Total Tested 1104 @ Total Tested 181 # Total Tested 345 $ Total Tested 45

Table 15: Antenatal screening

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D β-thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Indore	Sindhi, Gujarati, Marwari, Kunbi, Punjabi, Bhilal, Brahmin	1006	28 (2.70)	0	1 (0.09)	0	0	1 (0.09)	0	0	3 (0.29)	0	0	0	0	HbJ trait --1, HbQ India trait-- 1	17
2	Jabalpur	Not Mentioned	416	12 (2.89)	0	31 (7.45)	7 (1.68)	0	1 (0.24)	0	0	0	0	0	0	0	0	18

Table 16: Hospital based studies

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D β-thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Indore	Not Mentioned	510	9 (1.67)	5(0.98)	7(1.37)	5(0.98)	23(4.50)	0	0	0	0	0	0	0	0	0	19
2	Jabalpur, Damoh	General Population	1251	151(12.10)	0	57 (4.60)	0	0	0	0	0	0	0	0	0	0	0	20

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Southern Region

The Southern region includes the states of Andhra Pradesh, Karnataka, Kerala, Tamil Nadu and Telangana. A total of 57 articles which include population, antenatal, newborn and hospital based studies were compiled. The number of districts along with the articles that were available for compilation in each state is shown in Table 1.

Table 1: Studies available from the Southern Region

States	Districts	No of articles
Andhra Pradesh	8	14
Telangana	8	10
Karnataka	5	7
Tamil Nadu	15	16
Kerala	6	10
Total	**42**	**57**

Majority of the studies in all the states were on population based prevalence in β thalassaemia and other haemoglobinopathies among the tribal and non tribal populations. Fig 1 shows the map with the states (shaded areas) covered in the Southern region.



Fig 1: Map of India showing the states covered

Andhra Pradesh

Andhra Pradesh is in the south-eastern coastal region of India which comprises of two major regions, namely Rayalaseema in the south-west and Coastal Andhra bordering the Bay of Bengal in the east and north-east. It is bordered by Telangana to the north-west, Chhattisgarh to the north, Odisha to the north-east, Tamil Nadu to the south, Karnataka to the west and the Bay of Bengal to the east. The total population is 49.6 million of which 17.1% and 5.3% are scheduled caste and scheduled tribe populations respectively (Census 2011). The majority of the people of Andhra Pradesh are Hindus while Muslims constitute a sizeable minority.

Of the 14 studies that were available, 12 were population based and remaining two were hospital based studies. Fig 2 shows the districts (shaded areas) where the studies have been conducted. The number of districts covered have not been mentioned in 4 studies.

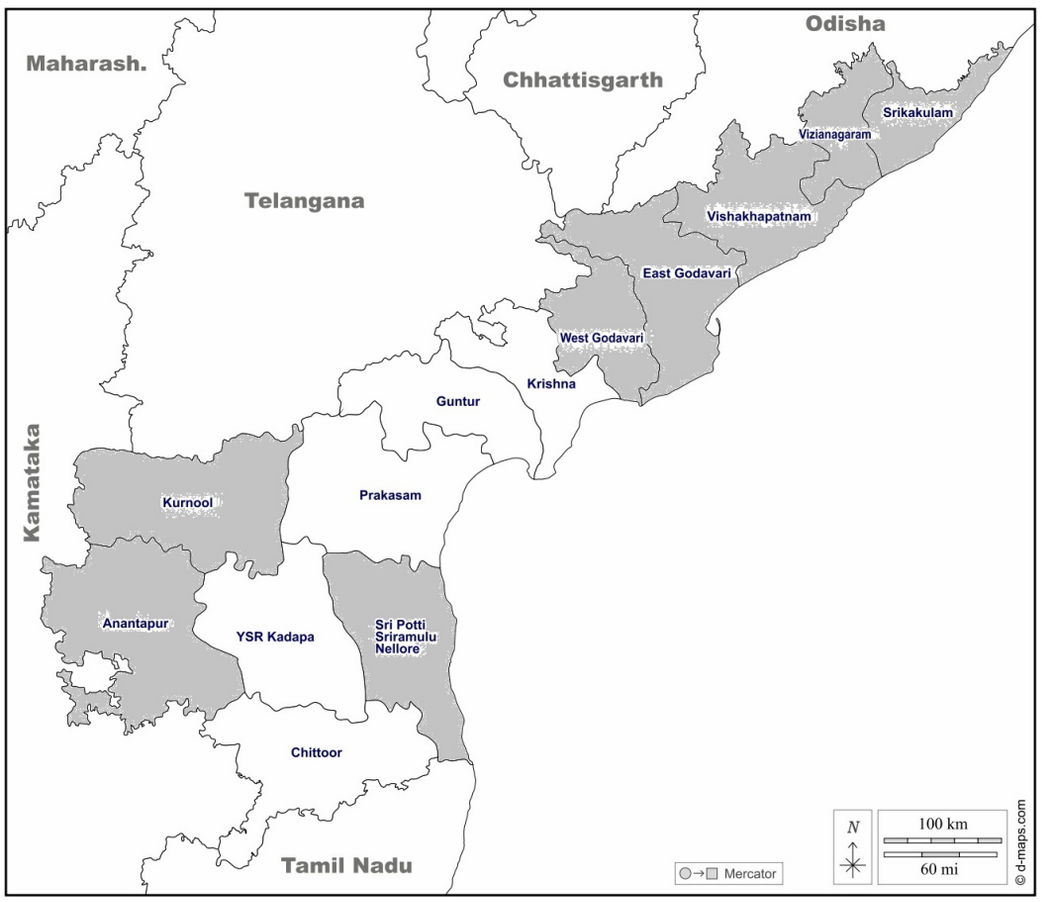

Fig 2: Map of Andhra Pradesh showing the studied areas

Tables 2- 4 show the number of districts covered along with the population groups (non-tribal and tribal) studied. A total of 15 non-tribal and 11 tribal groups were studied from 8 districts.

Table 2: Districts covered in Andhra Pradesh

Anantapur	East Godavari	Kurnool	Nelore	Srikakulam
Visakhapatnam	Vizanagaram	West Godavari		

Table 3: Non-Tribal populations studied

Brahmin	Dhobi	Gavara	Galaris	Jalari
Kaliga	Kapu	Madiga	Mala	Muslim
Relli	Sishtakarnam	Telegu	Turpu Kapu	Vysya

Table 4: Tribal populations studied

Chenchu	Jataput	Konda Kammara	Konda Reddy	Koya Dora
Koya Kammara	Savara	Sugali	Valmiki	Yanadi
Yerukula				

Population based studies indicated the presence of the HbS gene in the tribal and non-tribal populations studied. The prevalence of sickle cell trait varied from 0.40 to 19.30 % among the non-tribal populations (Table 5) and from 1.27 to 24.24 % among the tribal populations (Table 6). None of the studies reported the presence of sickle homozygous (SS) cases. One study reported the presence of β thalassemia trait in a tribal population group. Sporadic cases of βTT, AS, SS and S-β thalassemia were identified among the hospital based studies (Table 7).

Table 5: Population studies in Non- Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Not Mentioned	Telagu	109	-	-	0	0	-	-	-	-	-	-	-	-	-	-	1
2	Vishakha-patnam	Relli	215	-	-	35 (19.30)	0	-	-	-	-	-	-	-	-	-	-	2
3	Not Mentioned	Vysya	152	-	-	0	0	-	-	-	-	-	-	-	-	-	-	3
4	Not Mentioned	Brahmin	203	-	-	0	0	-	-	-	-	-	-	-	-	-	-	4
5	Not Mentioned	Vysya	211	-	-	0	0	-	-	-	-	-	-	-	-	-	-	4
6	Not Mentioned	Kappu	205	-	-	2 (0.98)	0	-	-	-	-	-	-	-	-	-	-	4
7	Not Mentioned	Mala	107	-	-	1 (0.94)	0	-	-	-	-	-	-	-	-	-	-	4
8	Not Mentioned	Madiga	504	-	-	2 (0.40)	0	-	-	-	-	-	-	-	-	-	-	4
9	Not Mentioned	Muslim	200	-	-	2 (1.00)	0	-	-	-	-	-	-	-	-	-	-	4
10	Not Mentioned	Jalari	104	-	-	2 (1.92)	0	-	-	-	-	-	-	-	-	-	-	5
11	Not Mentioned	Brahmin	85	-	-	0	0	-	-	-	-	-	-	-	-	-	-	5
12	Vishakha-patnam	Dhobi	200	0	0	2 (1.00)	0	0	0	0	0	0	0	0	0	0	0	6
13	Vishakha-patnam, Vizianagaram, Shrikakulam	Turpu Kapu	250	-	-	15 (6.00)	0	-	-	-	-	-	-	-	-	-	-	7
14	Vishakha-patnam, Vizianagaram, Shrikakulam	Sishta Karnam	100	-	-	8 (8.00)	0	-	-	-	-	-	-	-	-	-	-	7

(contd.)

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
15	Vishakha-patnam, Vizianagaram, Shrikakulam	Gavara	103	-	-	4 (3.90)	0	-	-	-	-	-	-	-	-	-	-	7
16	Vishakha-patnam, Vizianagaram, Shrikakulam	Kaliga	150	-	-	0	0	-	-	-	-	-	-	-	-	-	-	7

Table 6: Population studies in Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Nellore	Yanadi	187	-	-	0	0	-	-	-	-	-	-	-	-	-	-	8
2	East Godavari	Konda Reddy	129	-	-	3 (2.33)	0	-	-	-	-	-	-	-	-	-	-	8
3	East Godavari	Valmiki	99	-	-	20(20.20)	0	-	-	-	-	-	-	-	-	-	-	8
4	Nellore	Yerukula	38	-	-	0	0	-	-	-	-	-	-	-	-	-	-	8
5	Anantapur	Yerukula	20	-	-	0	0	-	-	-	-	-	-	-	-	-	-	8
6	Kurnool	Sugali	7	-	-	1 (14.28)	0	-	-	-	-	-	-	-	-	-	-	8
7	Kurnool	Chenchu	154	-	-	0	0	-	-	-	-	-	-	-	-	-	-	8
8	Anantpur	Sugali	110	-	-	3 (2.73)	0	-	-	-	-	-	-	-	-	-	-	8
9	East Godavari	Koya Dora	112	-	-	9 (8.04)	0	-	-	-	-	-	-	-	-	-	-	8
10	East Godavari	Koya Kammara	59	-	-	10(16.95)	0	-	-	-	-	-	-	-	-	-	-	8
11	SrikaKulam	Savara	132	-	-	2 (1.52)	0	-	-	-	-	-	-	-	-	-	-	9
12	SrikaKulam	Jataput	157	-	-	2 (1.27)	0	-	-	-	-	-	-	-	-	-	-	9
13	Kurnool	Chenchu	62	-	-	0	0	-	-	-	-	-	-	-	-	-	-	10
14	East Godavari (Maredumalli), West Godavari	Koya Dora	99	-	-	24(24.24)	-	-	-	-	-	-	-	-	-	-	-	11
15	Vishaka-patnam	Konda Kammara	103	1 (0.90)	-	14(13.59)	0	0	-	-	-	-	-	-	-	-	-	12

Table 7: Hospital based studies

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Vishakha-patnam	Not mentioned	100*	-	-	4 (4.00)	0	-	-	-	-	-	-	-	-	-	-	13
2	Vishakha-patnam	Not mentioned	151	8 (5.30)	0	36 (23.80)	15 (9.90)	3 (0.20)	0	0	0	0	0	0	0	0	0	14

* Mentally Challenged Cases.

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Telangana

Telangana is situated on the south-central stretch of the Indian peninsula on the high Deccan Plateau and consists mostly of hills, mountain ranges, and thick dense forests. It is bordered by the states of Maharashtra to the north, Chhattisgarh to the northeast, Karnataka to the west, and Andhra Pradesh to the east and south. As per the Census 2011, the total population of Telangana is 35 million of whom 20% and 10% belong to scheduled castes and scheduled tribes respectively.

A total of 10 studies were available of which 8 were population based covering tribal groups and the remaining two were each on newborn screening and hospital based studies. The districts covered (shaded areas) in the state are shown in Fig 3. Majority of the studies were conducted in the south eastern districts of the state.



Fig 3: Map of Telangana showing the studied areas

The number of districts covered along with the population groups (tribal) studied are shown in Tables 8 and 9. The data on population screening among non-tribal groups were not available. A total of 11 tribal groups were studied from 8 districts.

Table 8: Districts covered in Telangana

Adilabad	Bhadradri-Kothagudem	Hyderabad	Khammam	Mahabubnagar
Mahabubabad	Mulugu	Warangal		

Table 9: Tribal populations studied

Chenchu	Kolam	Konda Reddy	Koya	Koya Dora
Lambadi	Naik Pod	Pardhan	Raj Gond	Sugali
Yerukula				

Population based studies have only reported the presence of the HbS gene in all most all the tribal groups studied and the prevalence of sickle cell trait varied from 0.72 to 34.65 % while sickle homozygotes ranged from 0.46 to 0.99 % in different studies (Table 10). Universal newborn screening identified Hb AS cases (Table 11) while hospital based studies have picked up few cases of βTT, βTM, AS, SS, S-β thalassemia, EE, E-β thalassemia, HPFH, SD and β-thalassemia intermedia (Table 12).

Table 10: Population studies in Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Khamman	Koya	70	-	-	1 (1.43)	0	-	-	-	-	-	-	-	-	-	-	1
2	Khamman	Koya	123	-	-	9 (7.32)	0	-	-	-	-	-	-	-	-	-	-	1
3	Warangal	Yerukula	125	-	-	1 (0.80)	0	-	-	-	-	-	-	-	-	-	-	2
4	Khamman, Warangal (Otai), Mulugu, Mahabuba-bdad	Koya Dora	547	-	-	42 (7.86)	0	-	-	-	-	-	-	-	-	-	-	3
5	Adilabad	Naikpod	90	-	-	4 (4.61)	0	-	-	-	-	-	-	-	-	-	-	3
6	Mahabuba-bad & Warangal	Lambadi	154	-	-	4 (2.64)	0	-	-	-	-	-	-	-	-	-	-	3
7	Adilabad	Pardhan	122	-	-	38 (31.71)	0	-	-	-	-	-	-	-	-	-	-	3
8	Adilabad	Raj Gond	197	-	-	22 (11.34)	0	-	-	-	-	-	-	-	-	-	-	3
9	Adilabad	Kolam	215	-	-	3 (14.42)	1 (0.46)	-	-	-	-	-	-	-	-	-	-	4
10	Mahabub-nagar	Chenchu	139	-	-	1 (0.72)	0	-	-	-	-	-	-	-	-	-	-	5
11	Mahabub-nagar	Sugali	*61	-	-	3 (4.92)	0	-	-	-	-	-	-	-	-	-	-	6
12	Mahabub-nagar	Yerukula	*40	-	-	0	0	-	-	-	-	-	-	-	-	-	-	6
13	Khammam	Konda Reddy	92	-	-	2 (2.17)	0	-	-	-	-	-	-	-	-	-	-	6
14	Khammam	Koya	159	-	-	20 (12.58)	0	-	-	-	-	-	-	-	-	-	-	6
15	Adilabad	Raj Gond	133	-	-	19 (14.28)	1 (0.75)	-	-	-	-	-	-	-	-	-	-	6
16	Adilabad	Pardhan	101	-	-	34 (34.65)	1 (0.99)	-	-	-	-	-	-	-	-	-	-	6
17	khammam	Koya Dora	132	-	-	6 (4.50)	0	-	-	-	-	-	-	-	-	-	-	7
18	khammam	Nayakpod	40	-	-	0	0	-	-	-	-	-	-	-	-	-	-	7
19	Bhadradri kothagudem	Koya Dora	105	-	-	34 (32.80)	0	-	-	-	-	-	-	-	-	-	-	8

*Samples included from Kurnool district, Andhra Pradesh.

Table 11: Newborn screening

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Hyderabad	Not mentioned	7100	-	-	3 (0.04)	0	-	-	-	-	-	-	-	-	-	-	9

Table 12: Hospital based studies

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Hyderabad	Not mentioned	1592	347(21.70)	119(7.47)	39 (2.44)	53 (3.32)	24(1.50)	0	0	28(1.75)	0	0	9(0.50)	0	5 (0.31)	HbSD Punjab disease -1, β-thalassaemia intermedia-81	10

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Karnataka originally known as the State of Mysore lies in the south-western region of the Indian peninsula and it is the only southern state to have land borders with the other four southern Indian states. It is bordered by the Arabian Sea to the west, Goa to the northwest, Maharashtra to the north, Telangana to the northeast, Andhra Pradesh to the east, Tamil Nadu to the southeast, and Kerala to the south. The state has three principal geographical zones; the coastal region of Karavali and Tulu Nadu, the hilly Malenadu region comprising the Western Ghats and the Bayaluseeme region comprising the plains of the Deccan Plateau. According to the Census 2011, the total population of Karnataka is 61.1 million of which 17.15% belong to the scheduled castes and 6.95% to the scheduled tribes.

Of the 7 studies available, 4 were population based of which one study also reported antenatal screening and the remaining three were hospital based studies. Fig 4 shows the districts (shaded areas) where studies have been conducted. One study did not mention the names of the districts.

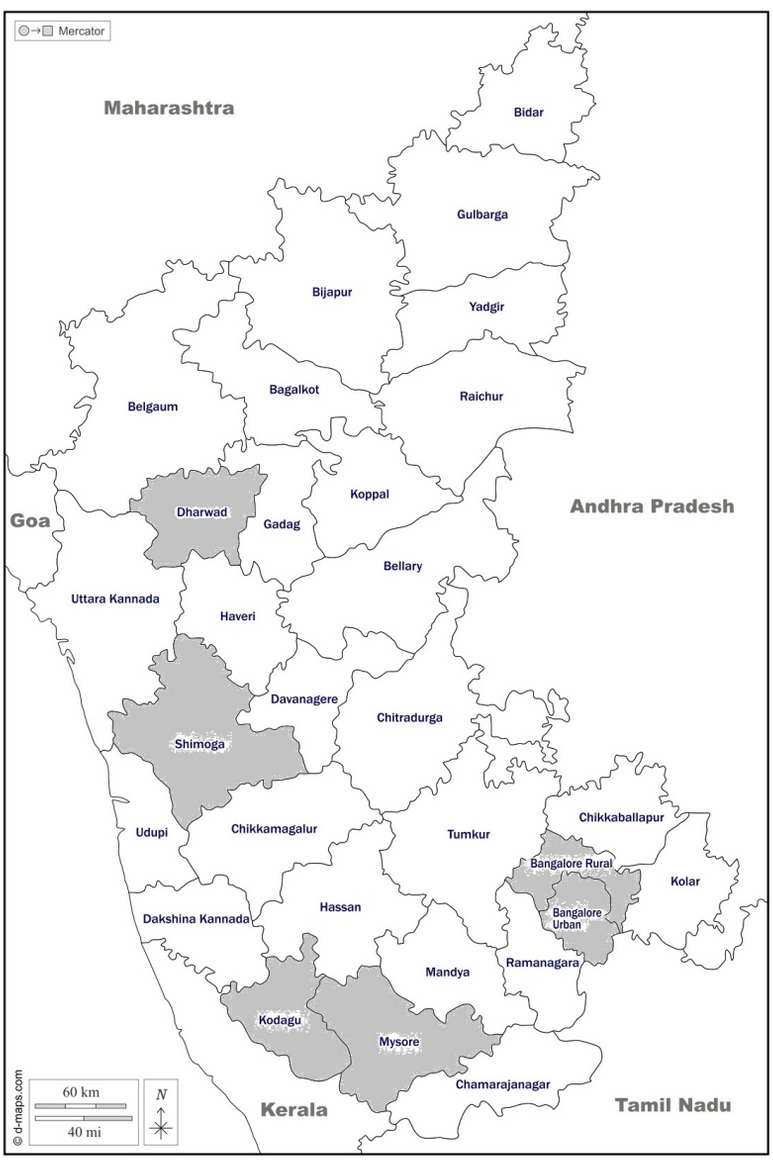

Fig 4: Map of Karnataka showing the studied areas

The number of districts covered along with the population groups (tribal) studied are shown in Tables 13 and 14. Many districts in Karnataka have not been covered. The names of the non tribal groups studied have not been mentioned. A total of 7 tribal groups were studied from 5 districts.

Table 13: Districts covered in Karnataka

Bangalore	Kodagu	Dharwad	Mysore	Shimoga

Table 14 : Tribal populations studied

Adi Karnataka	Betta Kurumba	Hasalaru	Iruliga	Jenu Kurumba
Soliga	Yerava			

Of the two studies available among non-tribal populations, one study has identified cases of β thalassemia and other haemoglobinopathies with a variable frequency while another study reported only the presence of βTT (Table 15). On the other hand, all the studies in tribal populations showed the presence of sickle cell gene and the prevalence of sickle cell trait varied from 1.27 to 25.00%. None of the studies picked up sickle homozygous cases (Table 16). Antenatal screening has been reported where apart from βTT, AS and SS, individuals with other haemoglobinopathies were also encountered (Table 17). In the hospital based studies besides βTT and other common Hb variants like HbS, Hb E, HbD, a rare variant HbQ India disease has also been identified (Table 18).

Table 15: Population studies in Non- Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Bangalore	Not mentioned	4992	122 (2.44)	0	13 (0.26)	6 (0.12)	6 (0.06)	7 (0.14)	2(0.04)	1 (0.01)	12(0.24)	1 (0.02)	0	5 (0.10)	24(0.48)	1	1
2	Not mentioned	Not mentioned	261	17 (6.50)	0	0	0	0	0	0	0	0	0	0	0	0	0	2

Table 16: Population studies in Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Kodagu (Coorg)	Jenu Kurumba	107	-	-	3 (2.80)	0	-	-	-	-	-	-	-	-	-	-	3
2	Kodagu (Coorg)	Betta Kurumba	48	-	-	4 (8.33)	0	-	-	-	-	-	-	-	-	-	-	3
3	Kodagu (Coorg)	Yerava	131	-	-	31(23.66)	0	-	-	-	-	-	-	-	-	-	-	3
4	Kodagu (Coorg)	Adi Karnatka	5	-	-	0	0	-	-	-	-	-	-	-	-	-	-	3
5	Mysore	Jenu Kurumba	20	-	-	3 (15.00)	0	-	-	-	-	-	-	-	-	-	-	3
6	Mysore	Yerava	4	-	-	1 (25.00)	0	-	-	-	-	-	-	-	-	-	-	3
7	Bangalore	Iruliga	79	-	-	1 (1.27)	0	-	-	-	-	-	-	-	-	-	-	3
8	Shimoga	Hasalaru	52	-	-	0	0	-	-	-	-	-	-	-	-	-	-	3
9	Mysore	Jenu Kuruba	175	-	-	17 (9.71)	0	-	-	-	-	-	-	-	-	-	-	4
10	Mysore	Soliga	78	-	-	5 (6.40)	0	-	-	-	-	-	-	-	-	-	-	4
11	Mysore	Betta Kurumba	99	-	-	10 (10.10)	0	-	-	-	-	-	-	-	-	-	-	4

Table 17: Antenatal screening

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Bangalore	Not mentioned	4989	94 (1.90)	0	7 (0.10)	1 (0.02)	0	6 (0.10)	1 (0.02)	0	7 (0.10)	1 (0.02)	0	8 (0.20)	15(0.30)	0	1

Table 18: Hospital based studies

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Dharwad	Not Mentioned	50	20 (40.00)	15(30.00)	2 (4.00)	0	1 (2.00)	0	0	0	0	0	0	0	0	5	5
2	Banglore	Sikh	154	8 (5.20)	1 (0.60)	1 (0.60)	0	0	0	0	0	3 (1.90)	0	1 (0.60)	0	0	HbQ India Disease-1	6
3	Banglore	Mixed Population	106	35 (33.00)	4 (3.80)	4 (3.80)	2 (1.90)	3 (2.80)	15(14.10)	18(16.70)	24(22.60)	0	0	-	-	1 (0.90)	-	7

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Tamil Nadu is the seventh most populous state in India and 48.4% of the state's population lives in urban areas. The state is surrounded by the Western Ghats in the west, the Eastern Ghats in the north, the Bay of Bengal in the east, the Gulf of Mannar and Palk Strait to the south-east, and the Indian Ocean in the south. It is bordered by the Union Territory of Puducherry and the states of Kerala, Karnataka, and Andhra Pradesh, as well as an international border with Sri Lanka. As per the Census 2011, the total population of Tamil Nadu is 72.1 million of which 20.01% and 1.10% belonged to scheduled castes and scheduled tribes respectively.

A total of 16 studies were available, 13 were population based and the remaining three were newborn screening and hospital based studies. The districts (shaded areas) where studies have been conducted in the state are shown in Fig 5. The name of the district has not been mentioned in one study.

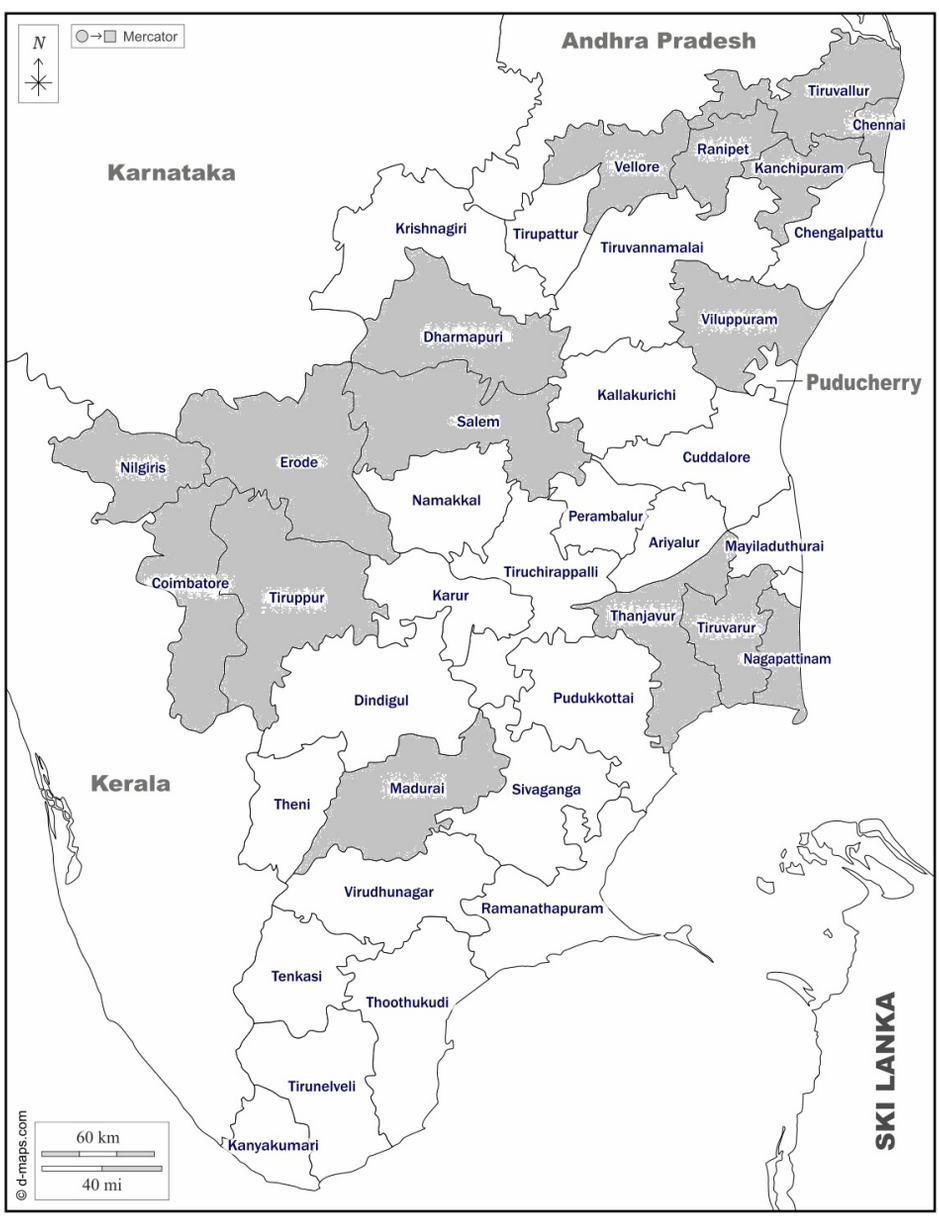

Fig 5: Map of Tamil Nadu showing the studied areas

Tables 19 and 20 show the number of districts covered along with the population groups (tribal) studied. The names of the non-tribal groups studied were not mentioned. A total of 17 tribal groups were studied from 15 districts.

Table 19: Districts covered in Tamilnadu

Chennai	Coimbatore	Dharampuri	Erode	Kancheepuram
Madurai	Nagapattinam	Nilgiris	Salem	Thanjavur
Thiruvallur	Thiruvarur	Tiruppur	Vellore	Viluppuram

Table 20: Tribal populations studied

Adi Dravida	Badaga	Chakkiliyan	Irula	Kadar
Kattu Nayakan	Kota	Kurumba	Malasar	Maliyali
Mannadiyar	Moolu Kurumba	Palliyan	Paniyan	Pulayan
Soliga	Toda			

Among the non tribal groups, the presence of βTT was only reported (Table 21). Population based studies indicated the presence of the HbS gene in most of the tribal groups studied. The prevalence of sickle cell trait varied from 0.94 to 40.00 % while the prevalence of sickle homozygotes ranged from 1.10 to 1.90 % in different studies. One study reported the presence of βTT, AD, δβ-thal trait and HPFH trait apart from AS and SS in the tribal groups (Table 22). Newborn screening identified few cases of βTT, AS, SS, S-β thalassemia and AE (Table 23). Hospital based studies reported β thalassaemia and all other haemoglobinopathies including many rare Hb variants and unusual compound heterozygous combinations like Hb Lepore Trait, HbH, HbSE, and HbJ Trait (Table 24)

Table 21: Population studies in Non- Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Not mentioned	Not mentioned	39	2 (5.10)	0	0	0	0	0	0	0	0	0	0	0	0	0	1

Table 22: Population studies in Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Nilgiri Hills	Kurumba	16	-	-	3 (18.75)	0	-	-	-	-	-	-	-	-	-	-	2
2	Nilgiri Hills	Irula	124	-	-	39 (31.45)	0	-	-	-	-	-	-	-	-	-	-	2
3	Nilgiri Hills	Paniyan	61	-	-	21 (34.43)	0	-	-	-	-	-	-	-	-	-	-	2
4	Nilgiri Hills	Badaga	191	-	-	16 (8.38)	0	-	-	-	-	-	-	-	-	-	-	2
5	Nilgiri Hills	Toda	84	-	-	3 (3.57)	0	-	-	-	-	-	-	-	-	-	-	2
6	Nilgiri Hills	Kota	86	-	-	0	0	-	-	-	-	-	-	-	-	-	-	2
7	Nilgiri Hills	Badaga	30	-	-	2 (6.67)	0	-	-	-	-	-	-	-	-	-	-	3
8	Nilgiri Hills	Irula	18	-	-	4 (22.20)	0	-	-	-	-	-	-	-	-	-	-	3
9	Nilgiri Hills	Kota	22	-	-	0	0	-	-	-	-	-	-	-	-	-	-	3
10	Nilgiri Hills	Toda	50	-	-	1 (2.00)	0	-	-	-	-	-	-	-	-	-	-	3
11	Nilgiri Hills	Kurumba	26	-	-	7 (26.90)	0	-	-	-	-	-	-	-	-	-	-	3
12	Nilgiri Hills	Kurumba	83*	-	-	14 (16.80)	0	-	-	-	-	-	-	-	-	-	-	4
13	Nilgiri Hills	Irula	15	-	-	6 (40.00)	0	-	-	-	-	-	-	-	-	-	-	5
14	Nilgiri Hills	Kurumba	43	-	-	10(23.26)	0	-	-	-	-	-	-	-	-	-	-	5

(contd.)

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
15	Nilgiri Hills	Toda	60	-	-	2 (3.33)	0	-	-	-	-	-	-	-	-	-	-	5
16	Coimbatore	Kadar	16	-	-	0	0	-	-	-	-	-	-	-	-	-	-	6
17	Coimbatore	Pulayan	15	-	-	0	0	-	-	-	-	-	-	-	-	-	-	6
18	Coimbatore	Malasar	41	-	-	3 (7.30)	0	-	-	-	-	-	-	-	-	-	-	6
19	Coimbatore	Soliga (Vrali)	115	-	-	30(26.00)	0	-	-	-	-	-	-	-	-	-	-	6
20	Coimbatore	Chakkiliyan	40	-	-	0	0	-	-	-	-	-	-	-	-	-	-	6
21	Madurai	Chakkiliyan	11	-	-	0	0	-	-	-	-	-	-	-	-	-	-	6
22	Madurai	Mannadiyar	51	-	-	0	0	-	-	-	-	-	-	-	-	-	-	6
23	Madurai	Pulayan	62	-	-	0	0	-	-	-	-	-	-	-	-	-	-	6
24	Madurai	Palliyan	51	-	-	0	0	-	-	-	-	-	-	-	-	-	-	6
25	Salem	Malayali (Kolli Hilla)	100	-	-	8 (8.00)	0	-	-	-	-	-	-	-	-	-	-	6
26	Salem	Malayali (Yarcand)	133	-	-	10 (7.52)	0	-	-	-	-	-	-	-	-	-	-	6
27	Vellore	Irula	120	-	-	0	0	-	-	-	-	-	-	-	-	-	-	6
28	Vellore	Adi Dravida	107	-	-	0	0	-	-	-	-	-	-	-	-	-	-	6
29	Dharmapuri	Adi Dravida	70	-	-	0	0	-	-	-	-	-	-	-	-	-	-	6
30	Dharmapuri	Panayan	41	-	-	0	0	-	-	-	-	-	-	-	-	-	-	6
31	Dharmapuri	Panchi Malyali	132	-	-	0	0	-	-	-	-	-	-	-	-	-	-	6
32	Ranipet (Arcot) & Dharmapuri	Panch Malyali	132	-	-	0	0	-	-	-	-	-	-	-	-	-	-	6
33	Coimbatore and Tiruppur (Annamalai Hills)	Kadar	213	-	-	2 (0.94)	0	-	-	-	-	-	-	-	-	-	-	7
34	Nilgiri Hills	Irula	175	-	-	45 (26.20)	2 (1.10)	-	-	-	-	-	-	-	-	-	-	8
35	Nilgiri Hills	Kurumba	43	-	-	9 (20.90)	0	-	-	-	-	-	-	-	-	-	-	8
36	Nilgiri Hills	Toda	98	-	-	1 (1.00)	0	-	-	-	-	-	-	-	-	-	-	8
37	Nilgiri Hills	Kota	549	-	-	0	0	-	-	-	-	-	-	-	-	-	-	9

(contd.)

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
38	Coimbatore	Irula	130	-	-	46(35.39)	0	-	-	-	-	-	-	-	-	-	-	10
39	Nilgiri Hills	Irula	536	-	-	162(30.30)	7 (1.30)	-	-	-	-	-	-	-	-	-	-	11
40	Nilgiri Hills	Paniya	196	-	-	72(36.70)	3 (1.60)	-	-	-	-	-	-	-	-	-	-	11
41	Nilgiri Hills	Kurumba	87	-	-	17(19.50)	1 (1.20)	-	-	-	-	-	-	-	-	-	-	11
42	Nilgiri Hills	Mullukurumba	156	-	-	59(37.80)	3 (1.90)	-	-	-	-	-	-	-	-	-	-	11
43	Nilgiri Hills	Soliga	402	-	-	120(30.00)	8 (1.90)	-	-	-	-	-	-	-	-	-	-	11
44	Nilgiri Hills	Kurumba	87	-	-	17(19.54)	0	-	-	-	-	-	-	-	-	-	-	11
45	Nilgiri Hills	Not Mentioned	9646	-	-	1089(11.30)	137(1.40)	-	-	-	-	-	-	-	-	-	-	12
46	Nilgiri Hills	Irula	967	8 (0.82)	0	208(21.50)	18 (1.86)	0	0	0	0	0	0	0	3 (0.31)	1 (0.10)	Unknown Variant--1	13
47	Nilgiri Hills	Kurumba	681	6 (0.88)	0	150(22.02)	10 (1.46)	0	0	0	0	2 (0.29)	0	0	3 (0.44)	2 (0.29)	0	13
48	Nilgiri Hills	Moolu Kurumba	860	2 (0.23)	0	230(26.70)	17 (1.90)	0	0	0	0	0	0	0	0	0	0	13
49	Nilgiri Hills	Paniya	877	1 (0.11)	0	231(26.30)	16 (1.80)	0	0	0	0	0	0	0	4 (0.45)	0	0	13

* Samples included from Wyanad district, Kerala.

Table 23: Newborn screening

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Chennai Madurai, Coimbatore, Vellore, Salem, Villuppuram, Erode, Kanc-heepuram, Thanjavur, Thiruvarur, Thiruvallur	Not mentioned	474	3 (0.63)	0	3 (0.63)	1 (0.21)	1 (0.21)	8 (1.68)	0	0	0	0	0	0	0	α-Thal--30	14

Table 24: Hospital based studies

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Chennai	Not mentioned	543	207 (37.90)	13 (2.30)	29 (5.30)	8 (1.40)	14 (2.57)	126 (23.20)	103 (18.90)	25 (4.60)	4 (0.70)	1 (0.18)	0	0	1 (0.10)	Hb Lepore trait- 1 , HbH disease -9, HbSE disease--1, HbJ trait--1	15
2	Chennai	Not mentioned	996	221 (22.10)	6 (0.60)	4 (0.40)	0	1 (0.10)	13 (1.31)	7(0.70)	2 (0.20)	0	3 (0.30)	0	0	0	Hb Lepore trait-- 1, HbH disease --1, βthal intermedia-3	16

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Kerala

Kerala is a state on the Malabar Coast of India and bordered by Karnataka to the north and northeast, Tamil Nadu to the east and south, and the Lakshadweep Sea to the west. Total population is 33.3 millions of which 1.0% belong to scheduled tribes (Census 2011). The population comprises predominantly of Hindus with a significant number of Muslims and Christians.

A total of 10 population based studies were only available. Fig 6 shows that studies have been conducted in 6 districts (shaded areas) in the state.

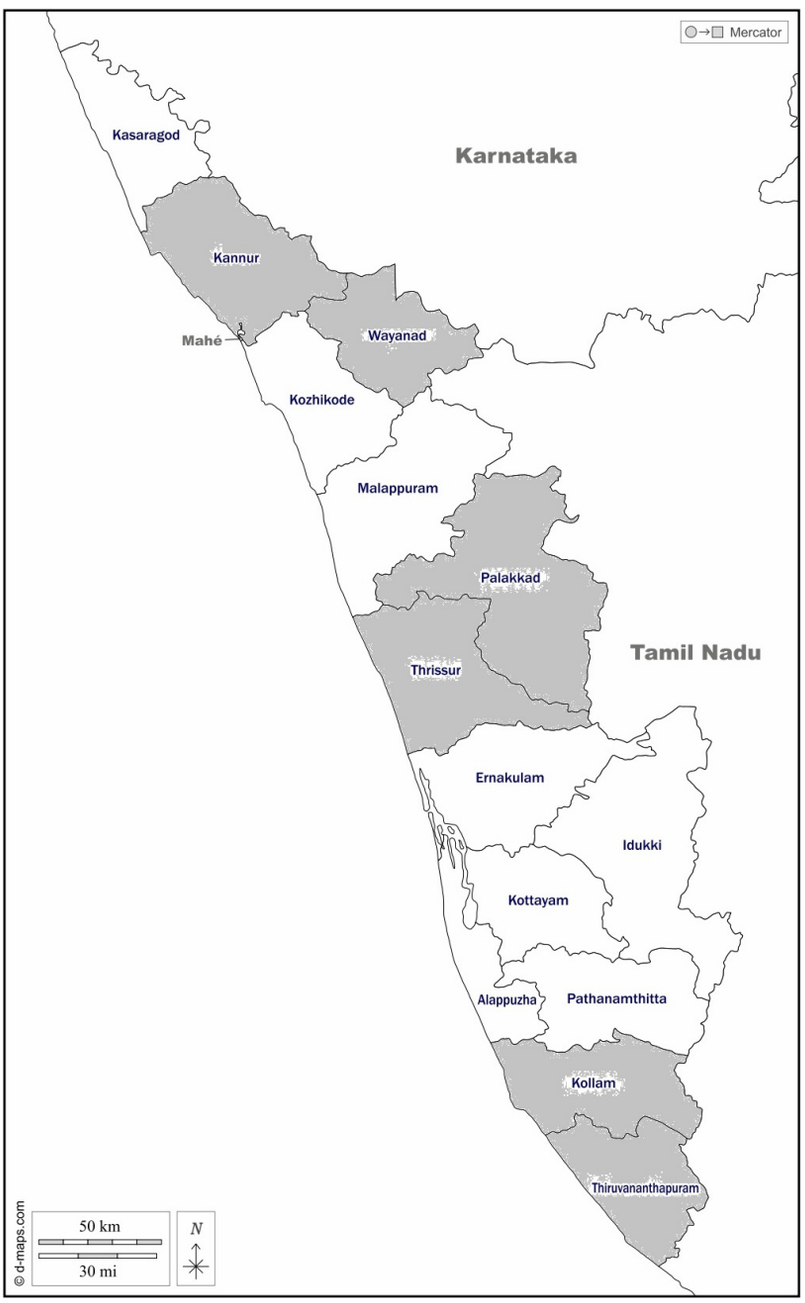

Fig 6: Map of Kerala showing the studied areas

The number of districts covered along with the population groups (non-tribal and tribal) studied are shown in Tables 25 and 26. Only one non tribal group (Chetty) along with 22 tribal groups were studied from 6 districts.

Table 25: Districts covered in Kerala

Kannur	Kollam	Palakkad	Thiruvanatapuram	Thrissur
Wayanad				

Table 26 : Tribal populations studied

Adiya	Adiyan	Irula	Jenu Kurumba	Kadar
Kannikar	Kattanayakan	Kurichan	Kurmar	Kurucha
Kuruma	Kurumba	Kuruvan	Malavedan	Malampandaram
Miss	Muduga	Oorali	Paniyan	Pulayan
Ulladan	Yerukula			

Population based studies indicated a high prevalence of sickle cell trait in both the tribal and non tribal populations studied. The prevalence of sickle cell trait varied from 28.97 to 29.60 % among the non-tribal populations and in the tribal populations, it ranged from 3.80 to 32.30 % while the prevalence of sickle homozygotes (Hb SS) varied from 0.70 to 16.20 % in different studies (Tables 27 and 28).

Table 27: Population studies in Non- Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Wayanad	Chetti	240	-	-	71(29.60)	39(16.20)	-	-	-	-	-	-	-	-	-	-	1
2	Wayanad	Chetti	214	-	-	62 (28.97)	11 (5.14)	-	-	-	-	-	-	-	-	-	-	2

Table 28: Population studies in Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Wayanad	Paniyan	74	-	-	22 (29.73)	0	-	-	-	-	-	-	-	-	-	-	3
2	Kollam (Quilon)	Malampandaram	116	-	-	0	0	-	-	-	-	-	-	-	-	-	-	4
3	Kollam (Quilon)	Ulladan	142	-	-	0	0	-	-	-	-	-	-	-	-	-	-	5
4	Kollam (Quilon)	Kuruvan	36	-	-	0	0	-	-	-	-	-	-	-	-	-	-	6
5	Thrissur (Quilon)	Kadar	167	-	-	0	0	-	-	-	-	-	-	-	-	-	-	7
6	Kollam (Quilon)	Malavedan	69	-	-	0	0	-	-	-	-	-	-	-	-	-	-	8
7	Palakkad (Palghat)	Irula	184	-	-	37 (20.27)	0	-	-	-	-	-	-	-	-	-	-	9
8	Wayanad	Paniyan	955	-	-	54 (26.70)	0	-	-	-	-	-	-	-	-	-	-	9
9	Kannur (Cannanore)	Jenu Kurumba	6	-	-	0	0	-	-	-	-	-	-	-	-	-	-	9
10	Thiruvana-nthapuram (Trivandrum)	Kannikar	102	-	-	0	0	-	-	-	-	-	-	-	-	-	-	9
11	Thiruvana-nthapuram (Trivandrum)	Pulaya	36	-	-	0	0	-	-	-	-	-	-	-	-	-	-	9

(contd.)

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
12	Thiruvana-nthapuram (Trivandrum)	Paraya	224	-	-	0	0	-	-	-	-	-	-	-	-	-	-	9
13	Palakkad (Palghat)	Yerukula	148	-	-	30(20.27)	0	-	-	-	-	-	-	-	-	-	-	9
14	Palakkad (Palghat)	Muduga	100	-	-	6 (6.00)	0	-	-	-	-	-	-	-	-	-	-	9
15	Palakkad (Palghat)	Kurumba	14	-	-	2 (14.29)	0	-	-	-	-	-	-	-	-	-	-	9
16	Kannur (Cannanore)	Adiyan	75	-	-	24(32.00)	0	-	-	-	-	-	-	-	-	-	-	9
17	Kannur (Cannanore)	Kurumba (Vrali)	42	-	-	8 (19.05)	0	-	-	-	-	-	-	-	-	-	-	9
18	Kannur (Cannanore)	Paniyan	43	-	-	8 (18.60)	0	-	-	-	-	-	-	-	-	-	-	9
19	Kannur (Cannanore)	Kurucha	52	-	-	0	0	-	-	-	-	-	-	-	-	-	-	9
20	Thiruvana-nthapuram (Trivandrum)	Malayarayan	59	-	-	0	0	-	-	-	-	-	-	-	-	-	-	10
21	Wayanad	Paniya	230	-	-	39(16.90)	2 (0.87)	-	-	-	-	-	-	-	-	-	-	1
22	Wayanad	Kurmar	210	-	-	68(32.30)	28(13.30)	-	-	-	-	-	-	-	-	-	-	1
23	Wayanad	Kattanayakan	193	-	-	20 (10.36)	2 (1.00)	-	-	-	-	-	-	-	-	-	-	1
24	Wayanad	Miss	100	-	-	19 (19.00)	5 (5.00)	-	-	-	-	-	-	-	-	-	-	1
25	Wayanad	Paniya	279	-	-	49 (17.60)	2 (0.70)	-	-	-	-	-	-	-	-	-	-	2
26	Wayanad	Kuruma	256	-	-	80 (31.30)	6 (2.30)	-	-	-	-	-	-	-	-	-	-	2
27	Wayanad	Adiya	91	-	-	25 (27.50)	4 (4.40)	-	-	-	-	-	-	-	-	-	-	2
28	Wayanad	Kattunayakan	52	-	-	2 (3.80)	0	-	-	-	-	-	-	-	-	-	-	2
29	Wayanad	Oorali	11	-	-	1 (9.10)	0	-	-	-	-	-	-	-	-	-	-	2
30	Wayanad	Kurichan	68	-	-	0	0	-	-	-	-	-	-	-	-	-	-	2

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Eastern Region

The eastern region includes the states of West Bengal, Odisha, Bihar and Jharkhand. A total of 65 articles which includes population, antenatal and hospital based studies were compiled. The number of districts along with the articles that were available for compilation in each state are shown in the Table 1.

Table 1: Studies available from the Eastern Region

States	Districts	No of articles
West Bengal	20	28
Odisha	18	21
Jharkhand	5	12
Bihar	1	4
Total	**44**	**65**

Majority of the studies in all the states were on population based prevalence of β thalassaemia and other haemoglobinopathies and hospital based studies among the non tribal populations. Reports on antenatal and newborn screening were available only from West Bengal and Odisha respectively. Fig 1 shows the map of India with the states (shaded areas) covered in the Eastern region.

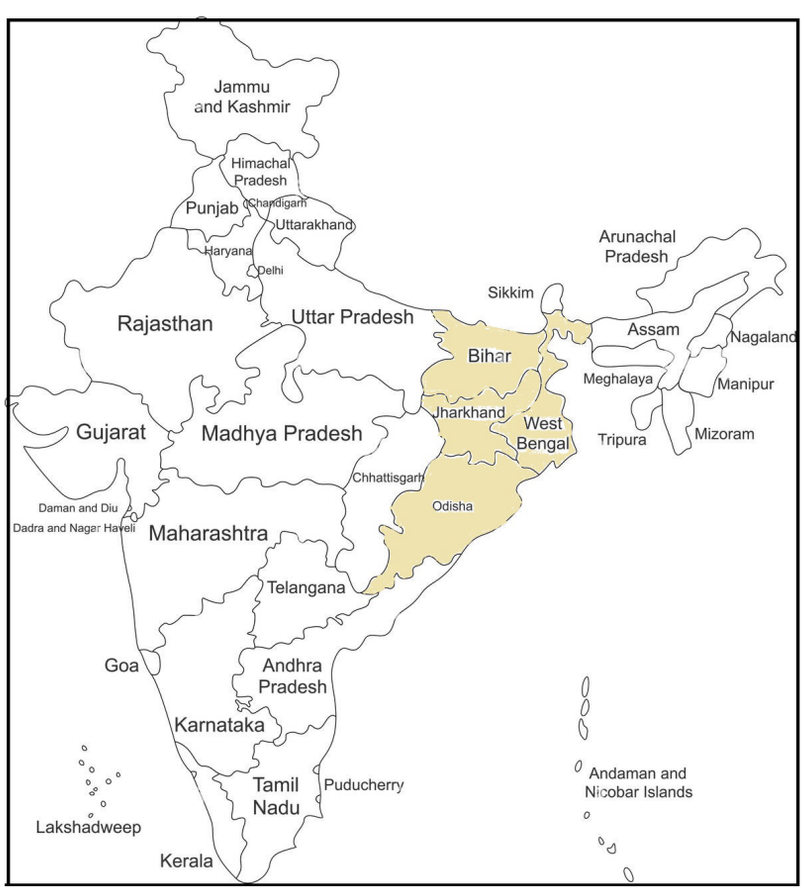

Fig 1: Map of India showing the states covered in the Eastern region

West Bengal

West Bengal is along the Bay of Bengal and it borders Bangladesh in the east, and Nepal and Bhutan in the north along with the states of Odisha, Jharkhand, Bihar, Sikkim and Assam. It is the fourth-most populous state with a population of over 91 million with 28.6% of people belonging to scheduled castes and 5.8% belonging to the scheduled tribes(Census 2011). Majority of people in the state are Bengalis while various Gorkha communities are mainly inhabitants of the Darjeeling Hills.

A total of 28 studies were available, of which 16 were population based prevalence studies mainly on β thalassemia trait and HbE trait, 5 were on antenatal screening and 7 on hospital based data. Fig 2 shows that studies have been conducted in majority of the districts (shaded areas) in the state. The names of the districts covered have not been mentioned in two studies.

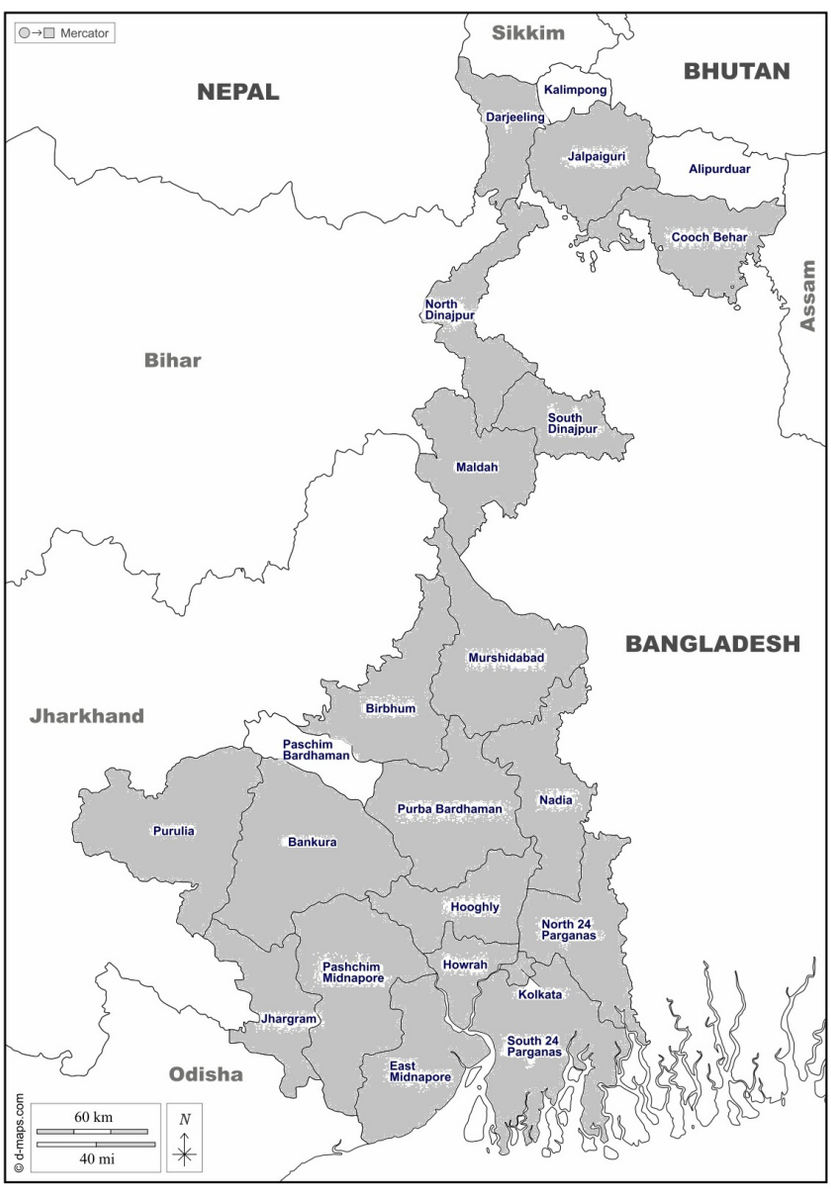

Fig 2: Map of West Bengal showing the studied areas

The number of districts covered along with the non-tribal and tribal population groups studied are shown in Tables 2-4. A total of 5 non-tribal and 12 tribal groups were studied from 20 districts.

Table 2: Districts covered in West Bengal

Bankura	Birbhum	Burdwan	Cooch Behar	Darjeeling
East Midnapur	Hoogly	Howrah	Jalpaiguri	Jhargram
Kolkata	Malda	Murshidabad	Nadia	North Dinajpur
North 24 Parganas	Puruliya	South Dinajpur	South 24 Parganas	West Midnapur

Table 3: Non Tribal populations studied

Bagdi	Jalia Kaibarta	Rajbanshi	Rahri Brahmin	Vaidya

Table 4: Tribal populations studied

Bhumiz	Deshi	Garo	Lepcha	Lodha
Mech	Munda	Poliya	Rabha	Santal
Tiyor	Toto			

Population based studies indicated that apart from the presence of βTT, βTM, HbAS, HbSS, HbS-β thal, HbAE, HbEE, HbE-β thal, HbAD Punjab and HbDD Punjab, individuals with other haemoglobinopathies like D- βthal, δβ-thal trait and HPFH trait were also encountered. Both β halassamia trait and Hb E trait were prevalent among the non-tribal populations with fewer numbers of individuals with Hb S and HbD Punjab (Table 5) while the tribal populations showed mainly Hb E (Table 6). Antenatal screening (Table 7) in different studies showed that the prevalence of βTT varied from 2.59 to 8.34% and HbAE from 1.56 to 23.37%. Other less frequent or rare haemoglobinopathies like Hb Lepore trait, HbQ India trait, HbJ Meerut trait, HbD Iran trait, HbJ trait, HbJ Bangkok, Hb Midnapore, HbSD Punjab, and HbSE were also identified during hospital based (Table 8) along with other studies .

Table 5: Population studies in Non-Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Cooch Behar	Rajbanshi	63	-	-	-	-	-	13(20.60)	0	-	-	-	-	-	-	-	1
2	Cooch Behar	Rahri Brahmin	42	-	-	-	-	-	3 (7.14)	0	-	-	-	-	-	-	-	1
3	Cooch Behar	Vaidya	29	-	-	-	-	-	2 (6.89)	0	-	-	-	-	-	-	-	1
4	Cooch Behar	Jalia Kaibarta	89	-	-	-	-	-	5 (5.60)	0	-	-	-	-	-	-	-	1
5	Cooch Behar	Bagdi	180	-	-	-	-	-	9 (5.00)	0	-	-	-	-	-	-	-	1
6	Kolkata	Not Mentioned	4989	153 (3.07)	0	5 (0.10)	0	0	205(4.11)	6(0.12)	1 (0.02)	11(0.22)	1 (0.02)	1 (0.02)	1 (0.02)	2 (0.04)	5	2
7	West Midnapur, East Midnapur, Puruliya, Hoogly, Burdwan	Not Mentioned	35413	3676(10.38)	0	399 (1.12)	0	3 (0.01)	1526 (4.30)	5(0.01)	7 (0.01)	134 (0.37)	0	0	0	2(0.01)	Hb Lepore trait-- 2, HbQ India trait--1, HbJ Meerut trait -- 1, HbD Iran trait--1	3
8	Medinipur (West Midnapore)	Not Mentioned	50487	3339(6.61)	368(0.73)	284(0.56)	0	75(0.15)	1403 (2.78)	26 (0.05)	213 (0.42)	106(0.21)	0	0	0	34(0.07)	Hb-Lepore-trait--6	4
9	Kolakata	Not Mentioned	3097	299 (9.60)	0	5 (0.10)	0	0	82(2.60)	0	0	4 (0.10)	0	0	0	0	0	*5
10	North 24 Parganas	Not Mentioned	1708	124 (7.20)	0	1 (0.05)	0	0	34 (1.99)	0	0	1 (0.05)	0	0	0	0	0	*5
11	South 24 Parganas	Not Mentioned	236	60(25.40)	0	1 (0.40)	0	0	16(6.70)	0	0	0	0	0	0	0	0	*5
12	Howrah	Not Mentioned	2359	181 (7.67)	0	5 (0.20)	0	0	24(1.01)	0	0	6 (0.25)	0	0	0	0	0	*5
13	Hoogly	Not Mentioned	2325	146 (6.20)	0	2 (0.08)	0	0	52(2.20)	0	0	2 (0.08)	0	0	0	0	0	*5
14	Medinipur (West Midnapore)	Not Mentioned	6117	577 (9.40)	0	5 (0.08)	0	0	144(2.30)	0	0	0	0	0	0	0	0	*5
15	Birbhum	Not Mentioned	1179	92 (7.80)	0	4 (0.30)	0	0	17(1.40)	0	0	3 (0.25)	0	0	0	0	0	*5

(contd.)

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
16	Burdwan (Purba Bardhaman)	Not Mentioned	604	72(11.90)	0	0	0	0	5 (0.80)	0	0	0	0	0	0	0	0	*5
17	Nadia	Not Mentioned	209	42(20.00)	0	0	0	0	15(7.10)	0	0	0	0	0	0	0	0	*5
18	Maldah	Not Mentioned	111	12(10.80)	0	0	0	0	5 (4.50)	0	0	0	0	0	0	0	0	*5
19	Darjeeling	Not Mentioned	150	17(11.30)	0	0	0	0	13(8.60)	0	0	0	0	0	0	0	0	*5
20	Murshidabad	Not Mentioned	71	14(19.70)	0	0	0	0	10(14.00)	0	0	0	0	0	0	0	0	*5
21	Hoogly	Not Mentioned	#21137	1137 (5.37)	59 (0.28)	49 (0.23)	0	0	729 (3.50)	0	104 (0.49)	0	0	0			@81	6
22	Darjeeling	Not Mentioned	1792	175(9.80)	0	0	27 (1.50)	0	406 (22.60)	44 (2.40)	131 (7.30)	12 (0.60)	12 (0.60)	-	-	-	HbJ trait--2	7
23	West Midnapur, Jhargram	Not Mentioned	278258	20774 (7.23)	345 (0.12)	2203(0.78)	125(0.04)	155 (0.05)	7963 (2.77)	102 (0.03)	412 (0.14)	592 (0.21)	0	-	-	171 (0.06)	Hb Lepore trait--3, HbQ India trait--3, HbJ Meerut trait--2, HbJ Bangkok --2 Hb Midnapore--1 Other variants--39	8

*Includes school children, pre-marriage cases, pre-pregancy cases, affected family members & preganant women
#Includes children, pre-marriage cases, post maritial cases, affected family members,family members of carriers & preganant women
@Includes HbAD Punjab, HPFH, δβ-thal trait, HbSD Punjab, HbSE, HbEE, HbSS etc.

Table 6: Population studies in Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Midnapore	Santal	336	-	-	3 (1.19)	0	-	-	-	-	-	-	-	-	-	-	9
2	Midnapore	Santal	164	-	-	0	0	-	-	-	-	-	-	-	-	-	-	10
3	Midnapore	Santal	102	4 (3.92)	-	0	0	-	1 (0.99)	0	-	-	-	-	-	-	-	11
4	Midnapore	Bhumiz	95	8 (8.42)	-	0	0	-	0	0	-	-	-	-	-	-	-	11

(contd.)

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
5	Cooch Behar	Rava	90	0	-	0	0	-	11(12.22)	0	-	-	-	-	-	-	-	1
6	Cooch Behar	Garo	21	0	-	0	0	-	2 (9.52)	0	-	-	-	-	-	-	-	1
7	Cooch Behar	Mech	26	0	-	0	0	-	9(34.61)	0	-	-	-	-	-	-	-	1
8	Midnapore	Munda	161	0	-	0	0	-	8 (4.97)	0	-	-	-	-	-	-	-	1
9	Midnapore	Lodha	197	0	-	0	0	-	20(10.16)	0	-	-	-	-	-	-	-	1
10	Darjeeling	Lepchas	215	0	-	0	0	-	9 (4.18)	0	-	-	-	-	-	-	-	12
11	Malda	Poliya	85	0	-	0	0	-	52 (61.20)	16 (18.80)	-	-	-	-	-	-	-	13
12	Malda	Deshi	103	0	-	0	0	-	46 (44.50)	40 (38.90)	-	-	-	-	-	-	-	13
13	Malda	Tiyor	95	0	-	0	0	-	20(21.00)	2 (2.10)	-	-	-	-	-	-	-	13
14	Birbhum, Hoogly	Santal	89	0	0	1 (1.12)	0	0	0	0	0	0	0	0	0	0	0	14
15	Jalpaiguri	Toto	443	-	-	-	-	-	218 (49.21)	85 (19.19)	-	-	-	-	-	-	-	15
16	Jalpaiguri, Cooch Behar	Rabha	234	-	-	-	-	-	101 (43.16)	99 (42.31)	-	-	-	-	-	-	-	16

Table 7: Antenatal screening

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Kolkata	Not Mentioned	5001	211 (4.2)	0	9 (0.20)	1 (0.02)	0	187(3.70)	6(0.10)	8 (0.20)	9 (0.20)	0	0	1 (0.02)	11(0.20)	9	2
2	Darjeeling	Rajbanshi	78	2 (2.59)	0	1 (1.29)	0	0	18 (23.37)	10 (12.98)	1 (1.29)	0	0	0	0	0	HbJ Meerut trait -1	17
3	Darjeeling	Hill tribes & Adivasis	39	0	0	0	0	0	7(17.94)	2(5.12)	0	0	0	0	0	0	0	17
4	Darjeeling	General Caste	71	2 (2.80)	0	1 (1.40)	0	0	4 (5.63)	1(1.40)	0	0	0	0	0	0	0	17
5	Kolkata	Not Mentioned	20883	856 (4.09)	0	48 (0.23)	0	0	1192 (5.70)	0	20(0.09)	0	0	0	0	-	77	18
6	Not Mentioned	Not Mentioned	10407	579 (5.60)	11 (0.10)	37 (0.40)	7 (0.10)	4 (0.04)	522(5.00)	32(0.30)	92(0.90)	7 (0.10)	3 (0.03)	0	0	9 (0.10)	HbSD Punjab-1	19
7	Bankura	Not Mentioned	875	51 (5.80)	0	4 (0.45)	0	0	27(3.08)	0	0	0	0	0	0	0	0	20
8	Bankura	Not Mentioned	2625	107(4.00)	0	7 (0.20)	0	0	72(2.70)	3(0.10)	4(0.10)	0	0	0	0	0	0	20
9	Medinipur, Jhargram	Not Mentioned	899	75 (8.34)	0	20 (2.22)	3 (0.33)	1 (0.11)	14(1.56)	3(0.33)	3(0.33)	1 (0.11)	0	0	0	3 (0.33)	0	21

Table 8: Hospital based studies

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Birbhum	Santal	100	3 (3.00)	0	1 (1.00)	0	0	1 (1.00)	0	0	0	0	0	0	0	0	22
2	Burdwan	Not Mentioned	3823	626(16.37)	1 (0.03)	24 (0.63)	6 (0.15)	11(0.30)	176(4.60)	7(0.18)	134(3.50)	4 (0.10)	0	0	0	8 (0.21)	122	23
3	Kolkata	Not Mentioned	14145	1362 (9.62)	105 (0.74)	39 (0.27)	3 (0.02)	135 (0.95)	696 (4.92)	18 (0.28)	570 (4.02)	12(0.08)	0	120 (0.84)	0	0	Hb Lepore--trait 1,HbSD Punjab- 1,β-thal interm-edia -36	24
4	Kolkata	Bengali	660	176(26.67)	44(6.67)	6 (0.91)	0	6 (0.91)	42(6.36)	16(2.40)	102 (15.45)	0	0	0	0	12(1.80)	0	25
5	Darjeeling, Jalpaiguri, Coochbehar, North Dinajpur, South Dinajpur and Malda	Not Mentioned	1872	158 (17.80)	13 (1.50)	10 (1.10)	2 (0.20)	30(3.40)	644 (34.40)	474 (25.30)	283 (15.10)	7 (0.80)	0	0	2 (0.20)	0	HbJ Trait-2	26
6	Malda	Not Mentioned	5156	157 (3.04)	1 (0.02)	18 (0.35)	0	0	465 (9.02)	101 (1.96)	32 (0.62)	0	0	0	0	0	24	27
7	Not Mentioned	Not Mentioned	119336	5488 (4.60)	1981 (1.66)	453 (0.32)	382(0.32)	310 (0.26)	3604 (3.02)	406 (0.34)	1384 (1.16)	107(0.09)	0	-	24(0.02)	143 (0.12)	Hb lepore trait -4, HbQ India trait-48, HbH disease - 12, HbJ Meerut trait -- 36, α-thal trait- 143, HbSE disease -6, HbSD Punjab- 1	28

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Odisha

Odisha, formerly known as Orissa shares a border with West Bengal and Jharkhand to the north, Chhattisgarh to the west, Andhra Pradesh to the south, and a very negligible border with Telangana to the southwest. According to the 2011 Census, the total population of Odisha is 42 million with 17.13% being scheduled caste and 22.85% being scheduled tribe populations. The state has the third largest population of Scheduled Tribes in India.

Of the 21 available studies, 12 were population based and one was on newborn screening while the remaining 8 were hospital based studies. Fig 3 shows the districts (shaded areas) where studies have been undertaken. In 5 studies the names of the districts covered were not mentioned.

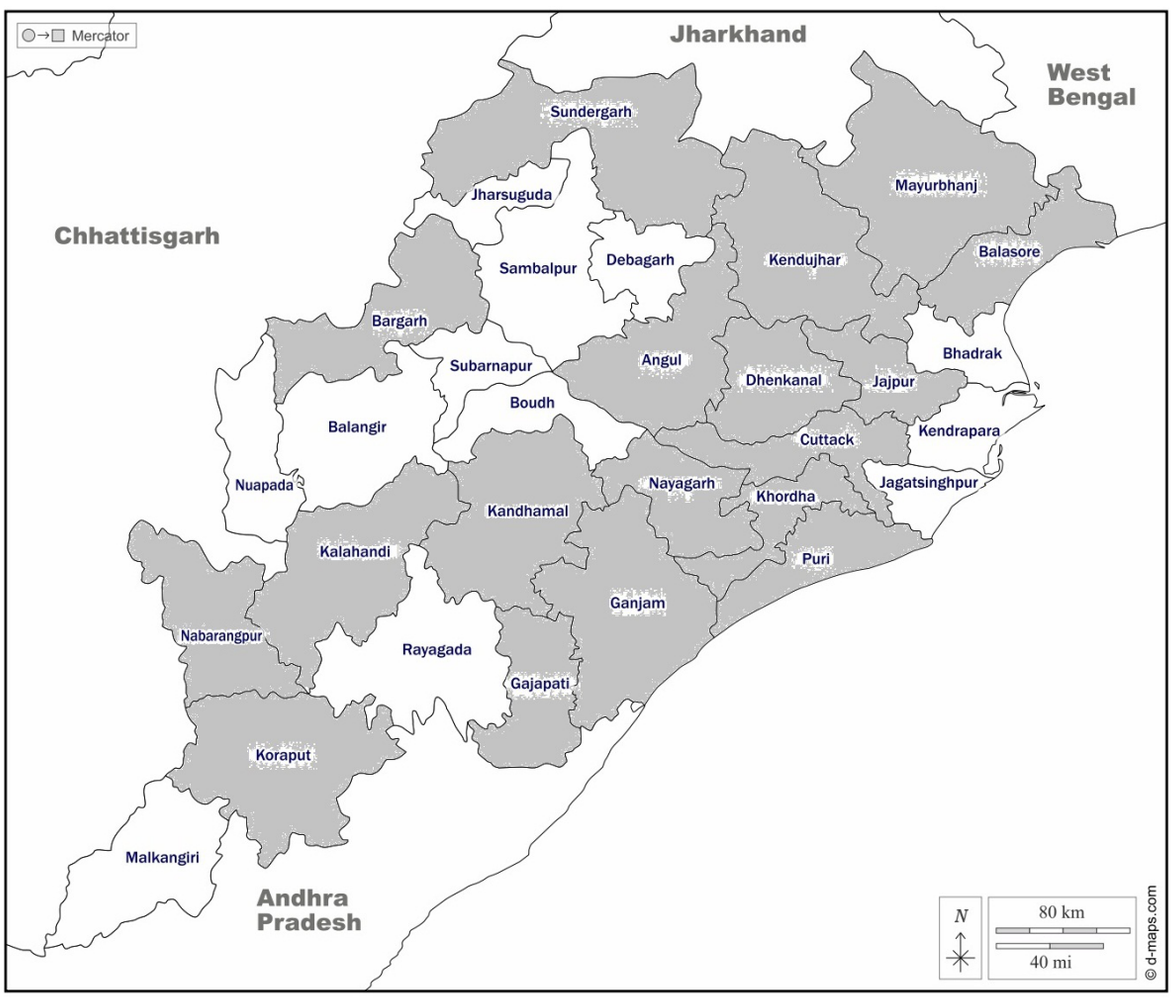

Fig 3: Map of Odisha showing the studied areas

The districts covered and the number of tribal groups studied is shown in Tables, 9 and 10. The names of the non-tribal groups studied were not mentioned. A total of 36 tribal groups were studied from 18 districts.

Table9: Districts covered in Odisha

Andul	Balassore	Bargarh	Cuttack	Dhenkanal
Gajapati	Ganjam	Jajpur	Kalahandi	Kandhamal
Kendujhar	Khordha	Koraput	Mayurbhanj	Nabarangpur
Nayagarh	Puri	Sundargarh		

Table10: Tribal populations studied

Bada Gadaba	Bhatra	Bhatudi	Bhumiz	Bhuyan
Binjhal	Bondo	Deshia Khond	Dhelki Kharia	Didayi
Dudh Kharia	Gond	Juanga	Kharia Kissan	Kharia Sahara
Khond Gadaba	Kondha	Konda Dora	Konda Paroja	Kolha
Kutia	Kutia Khond	Kuvi Khond	Lodha	Munda
Nuka Dora	Ollaro Gadaba	Oraon	Paraja	Parang Gadaba
Parang Paroja	Poroja	Raj Gond	Saora	Santhal
Savara Bhuyan				

A high prevalence of the HbS gene was reported both among non tribal and tribal groups. Among the non tribal groups (Table 11), the prevalence of sickle cell trait and sickle homozygotes was as high as 40.40% and 17.40% respectively. Population based studies indicated the presence of HbS gene in all most all the tribal groups studied and the prevalence of sickle cell trait varied from 0.90 to 21.00 % and sickle homozygotes from 0.13 to 8.80% in different studies. Few studies also reported the presence of βTT with a prevalence rate of 0.50 to 8.50% along with sporadic cases of S-β thal, AE, EE, AD, δβ-thal trait and HPFH trait in tribal populations (Table 12). Newborn screening has identified AS, SS and AD cases (Table 13). Hospital based studies reported β-thalassaemia and all other haemoglobinopathies including unusual variants like HbSD, HbSE, HbLepore and homozygous HPFH (Table 14).

Table 11: Population studies in Non-Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Angul	Scheduled Caste	46	0	-	14 (30.40)	8 (17.40)	-	0	0	-	-	-	-	-	-	-	1
2	Angul	General Caste	94	0	-	38 (40.40)	16(17.00)	-	0	0	-	-	-	-	-	-	-	1

Table 12: Population studies in Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Koraput	Kondh	100	-	-	0	0	-	-	-	-	-	-	-	-	-	-	2
2	Koraput	Nuka Dora	111	-	-	10 (9.09)	0	-	-	-	-	-	-	-	-	-	-	2
3	Cuttack, Puri & Balassore	Santal	51	-	-	0	0	-	-	-	-	-	-	-	-	-	-	3
4	Kalahandi	Kondh	116	-	-	13 (11.10)	0	-	-	-	-	-	-	-	-	-	-	4
5	Kandhamal (Udigiri, Phulbani)	Kondh	901	-	-	180(19.90)	79 (8.80)	-	-	-	-	-	-	-	-	-	-	5
6	Mayurbhanj	Others	52	2 (3.80)	-	0	0	-	-	-	-	-	-	-	-	-	-	5
7	Not Mentioned	Bada Gadaba	99	-	-	0	0	-	-	-	-	-	-	-	-	-	-	7
8	Not Mentioned	Binjhal	103	-	-	7 (6.80)	3 (2.91)	-	-	-	-	-	-	-	-	-	-	7
9	Not Mentioned	Deshia Khond	107	-	-	9 (8.41)	0	-	-	-	-	-	-	-	-	-	-	7
10	Not Mentioned	Gadaba	104	-	-	8 (7.69)	1 (0.96)	-	-	-	-	-	-	-	-	-	-	7
11	Not Mentioned	Khond	116	-	-	13 (11.21)	0	-	-	-	-	-	-	-	-	-	-	7
12	Not Mentioned	Kissan	108	-	-	1 (0.93)	3 (2.78)	-	-	-	-	-	-	-	-	-	-	7
13	Not Mentioned	Konda Dora	86	-	-	13 (15.12)	1 (1.16)	-	-	-	-	-	-	-	-	-	-	7

(contd.)

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
14	Not Mentioned	Konda Paroja	225	-	-	0	0	-	-	-	-	-	-	-	-	-	-	7
15	Not Mentioned	Kutia Khond	330	-	-	54 (16.36)	0	-	-	-	-	-	-	-	-	-	-	7
16	Not Mentioned	Kuvi Khond	94	-	-	2 (2.13)	1 (1.06)	-	-	-	-	-	-	-	-	-	-	7
17	Not Mentioned	Nuka Dora	111	-	-	10 (9.09)	0	-	-	-	-	-	-	-	-	-	-	7
18	Not Mentioned	Ollaro Gadaba	225	-	-	6 (2.67)	0	-	-	-	-	-	-	-	-	-	-	7
19	Not Mentioned	Parang Gadaba	225	-	-	28 (12.44)	0	-	-	-	-	-	-	-	-	-	-	7
20	Not Mentioned	Parang Paroja	104	-	-	0	0	-	-	-	-	-	-	-	-	-	-	7
21	Not Mentioned	Paroja	94	-	-	6 (6.38)	2 (2.13)	-	-	-	-	-	-	-	-	-	-	7
22	Not Mentioned	Raj Gond	104	-	-	6 (5.77)	0	-	-	-	-	-	-	-	-	-	-	7
23	Not Mentioned	Savara	102	-	-	0	0	-	-	-	-	-	-	-	-	-	-	7
24	Sundargarh	Bhuyan	244	16 (6.60)	-	13 (5.30)	0	3 (1.20)	0	0	-	4 (1.60)	0	-	-	-	-	8
25	Sundargarh	Dhelki Kharia	337	15 (4.50)	-	40 (11.90)	2 (0.60)	0	9 (2.70)	1(0.30)	-	0	0	-	-	-	-	8
26	Sundargarh	Dudh Kharia	421	32 (7.70)	-	0	0	0	0	0	-	0	0	-	-	-	-	8
27	Mayurbhanj	Bhatudi	95	0	-	1 (1.05)	0	1 (1.05)	0	0	-	0	0	-	-	-	-	9
28	Nawarangpur (Nabarangpur)	Bhatra	166	11 (6.60)	-	25(15.10)	5 (3.00)	0	0	0	-	0	0	-	-	-	-	9
29	Mayurbhanj	Bhumiz	116	2 (1.70)	-	1 (0.90)	0	0	0	0	-	0	0	-	-	-	-	9
30	Sundargarh	Bhuyan	92	0	-	0	0	0	0	0	-	0	0	-	-	-	-	9
31	Kalahandi	Gond	219	1 (0.50)	-	46 (21.00)	3 (1.40)	0	0	0	-	0	0	-	-	-	-	9
32	Sundargarh	Kharia	54	1 (1.90)	-	4 (7.40)	0	0	0	0	-	0	0	-	-	-	-	9
33	Sundargarh	Kissan	130	2 (1.50)	-	0	0	0	0	0	-	0	0	-	-	-	-	9
34	Mayurbhanj	Kolha	102	2 (2.00)	-	0	0	0	0	0	-	0	0	-	-	-	-	9

(contd.)

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
35	Kandhamal	Kondh	254	16 (6.30)	-	8 (3.10)	0	0	0	0	-	0	0	-	-	-	-	9
36	Mayurbhanj	Lodha	78	6 (6.70)	-	0	0	0	0	0	-	0	0	-	-	-	-	9
37	Sundargarh	Munda	96	5 (5.20)	-	3 (3.10)	0	0	0	0	-	0	0	-	-	-	-	9
38	Sundargarh	Oraon	104	2 (1.90)	-	0	0	0	0	0	-	0	0	-	-	-	-	9
39	Koraput	Paraja	176	15 (8.50)	-	23(13.10)	3 (1.70)	0	0	0	-	0	0	-	-	-	-	9
40	Mayurbhanj	Santhal	100	8 (8.00)	-	1 (1.00)	0	0	0	0	-	0	0	-	-	-	-	9
41	Ganjam & Gajapati	Saora	177	11 (6.20)	-	13 (7.30)	0	0	0	0	-	0	0	-	-	-	-	9
42	Sundargarh	Bhuyan	836	53 (6.33)	-	20 (2.20)	0	-	0	0	-	4 (0.40)	0	-	-	4 (0.40)	-	10
43	Sundargarh	Kharia	767	48 (6.20)	-	41 (5.40)	2 (0.20)	-	10 (1.30)	1 (0.10)	-	0	0	-	-	0	-	10
44	Bargarh, Kalahandi	Sahara, Kuda, Kutia, Oraon, Gond	594	20 (3.37)	0	60(10.10)	18 (3.03)	0	0	0	0	0	0	0	0	0	α -thal-trait 302	11
45	Not Mentioned	Bondo	569	19 (3.33)	0	9 (1.67)	0	0	0	0	0	0	0	0	2 (0.35)	1 (0.17)	0	12
46	Not Mentioned	Didayi	748	21 (2.80)	0	65 (8.60)	1 (0.13)	0	0	0	0	0	0	0	13(1.73)	0	Unknown variant-2	12
47	Not Mentioned	Juanga	769	44 (5.72)	0	8 (1.04)	2 (0.26)	0	0	0	0	0	0	0	5 (0.65)	4 (0.52)	0	12
48	Not Mentioned	Kondha	670	21 (3.13)	0	68(10.14)	3 (0.44)	0	0	0	0	0	0	0	12(1.79)	1 (0.14)	0	12

Table 13: Newborn screening

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Kalahandi	Not Mentioned	761	-	-	112(14.71)	13 (1.70)	-	-	-	-	1 (0.13)	-	-	-	-	-	13

Table 14: Hospital based studies

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Not Mentioned	Not Mentioned	442	-	-	-	-	-	-	-	14(3.16)	0	0	-	-	-	-	14
2	Khordha (Bhubaneswar)	Scheduled Caste, Scheduled Tribe, General Caste	520	103(19.80)	46(8.80)	131(25.20)	49 (9.4)	17(3.20)	6 (1.10)	0	17(3.20)	0	0	0	0	0	0	15
3	Andul,Khordha, Nayagah, Kandhamahal, Cuttack,Jajpur, Dhenkanal, Ganjam, Kendujhar, Mayurbhanj	Not Mentioned	1015	185(18.20)	54(5.30)	302(29.80)	77 (7.60)	17(1.70)	9 (0.90)	3(0.30)	7 (0.70)	2 (0.20)	0	0	9 (0.90)	0	HbSD Punjab-- 2	16
4	Khordha (Bhubaneswar)	Hindu, Muslim	877	265(30.20)	64 (7.3)	148 (16.90)	63 (7.20)	55(6.30)	26(3.00)	0	47 (5.50)	5 (0.60)	0	0	2 (0.20)	3 (0.30)	0	17
5	Not Mentioned	Not Mentioned	820	89 (10.80)	10 (1.20)	153 (18.60)	71 (8.70)	11 (1.40)	10 (1.20)	0	1 (0.10)	2 (0.20)	3 (0.30)	4 (0.40)	4 (0.40)	0	α-thal trait -- 14, HbSE disease --2, β- thal intermedia--1	18
6	Not Mentioned	Not Mentioned	331	66(19.93)	7 (2.10)	51(15.40)	34 (10.20)	43 (13.00)	5 (1.50)	3(0.91)	9 (2.70)	4 (1.20)	0	0	0	0	α-thal trait-4	19
7	Ganjam (Berhampur)	Not Mentioned	435	29 (6.66)	13 (2.98)	78(17.93)	101 (23.21)	23(5.28)	0	0	1 (0.22)	0	0	0	1 (0.40)	0	Hb Lepore-trait-1,α-thal trait--3,β-thal intermedia-3 HPFH hom-ozygous --4	20
8	Ganjam (Berhampur)	Not Mentioned	2332	156 (6.60)	25 (1.07)	535 (22.90)	517 (22.10)	75 (3.20)	11 (0.47)	0	4 (0.17)	1 (0.04)	0	0	0	28(1.20)	Hb Lepore / β thal-- 4 HbS-Lepore -- 6, HbSE disease--2 Hb Lepore trait--8,Others --5, HPFH homozygous -3	21

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Jharkhand

Jharkhand has been called 'the land of forests' and shares the border with Bihar to the north, Uttar Pradesh to the northwest, Chhattisgarh to the west, Odisha to the south and West Bengal to the east. The total population is 32.96 million of whom 12.08% and 26.21% belong to scheduled castes and scheduled tribes respectively (Census 2011.)

Of the 12 studies which were available, 9 were on population screening in both non tribal and tribal groups and the remaining 3 studies were hospital based.The districts where the studies have been conducted (shaded areas) are shown in Fig 4. The names of the districts were not mentioned in three studies.

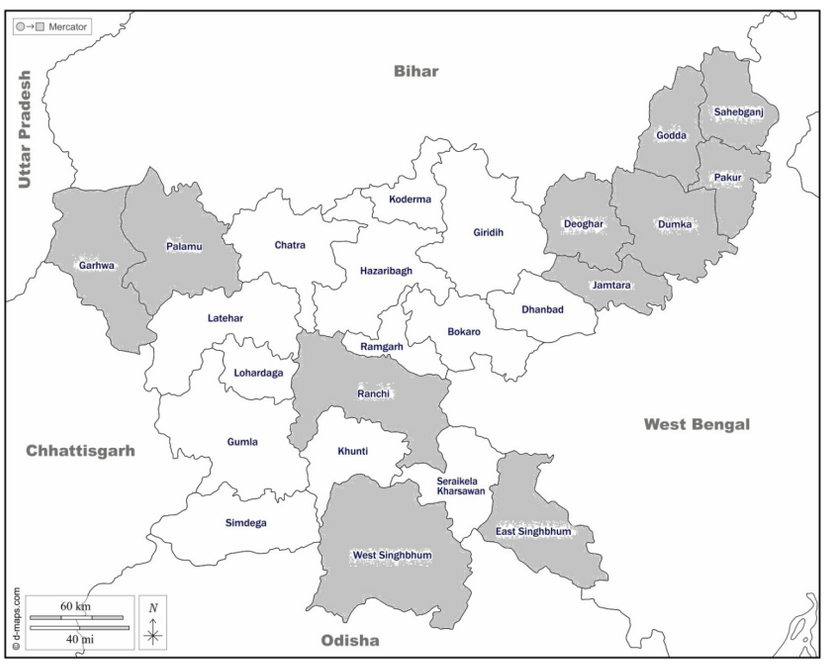

Fig 4: Map of Jharkhand showing the studied areas

Tables15 and 16 show the number of districts covered along with the tribal population groups studied. The names of the Non-tribal groups studied were not mentioned. Totally 10 tribal groups from 5 districts were studied.

Table 15: Districts covered in Jharkhand

Garahwa	Palamau	Ranchi	Sahebganj	East-Singhbhaum

Table 16: Tribal populations studied

Bhuiya	Chero	Desi Bhumij	Dudh Kharia	Ho
Khairwar	Munda	Oraon	Santhal	Saura Paharia

Population based studies indicated the presence of the HbS gene in both Non-tribal and tribal groups (Tables 17 and 18). The prevalence of sickle cell trait varied from 1.30 to 15.38% and 0.50 to 16.66% in Non-tribal and tribal groups respectively. None of the studies reported the presence of SS cases. Hospital based studies reported β thalassaemia and all other haemoglobinopathies like HbS, HbE and HbD Punjab as well as HbJ triat, HbJ Merut and HbSD Punjab (Table 19).

Table 17: Population studies in Non-Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Garahwa	Not Mentioned	153	-	-	2 (1.30)	0	-	-	-	-	-	-	-	-	-	-	1
2	Tatanagar(East Singhbhum)	Not Mentioned	52	-	-	8 (15.38)	0	-	-	-	-	-	-	-	-	-	-	1
3	Ranchi	Not Mentioned	97	-	-	0	0	-	-	-	-	-	-	-	-	-	-	1
4	Sahebganj	Not Mentioned	23	-	-	0	0	-	-	-	-	-	-	-	-	-	-	1

Table 18: Population studies in Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Not Mentioned	Saura Paharia	126	-	-	0	0	-	-	-	-	-	-	-	-	-	-	2
2	Not Mentioned	Santal	126	-	-	0	0	-	-	-	-	-	-	-	-	-	-	3
3	Ranchi	Dudh Kharia	119	-	-	0	0	-	-	-	-	-	-	-	-	-	-	4
4	Ranchi	Munda	105	-	-	0	0	-	-	-	-	-	-	-	-	-	-	4
5	Singhbhum	Desi Bhumij	168	-	-	1 (0.60)	0	-	-	-	-	-	-	-	-	-	-	5
6	Singhbhum	Ho	200	-	-	1 (0.50)	0	-	-	-	-	-	-	-	-	-	-	6
7	Ranchi	Oraon	100	-	-	0	0	-	-	-	-	-	-	-	-	-	-	7
8	Palamau	Khairwar	67	-	-	0	0	-	-	-	-	-	-	-	-	-	-	8
9	Palamau	Chero	57	-	-	0	0	-	-	-	-	-	-	-	-	-	-	8
10	Palamau	Bhuiya	41	-	-	0	0	-	-	-	-	-	-	-	-	-	-	8
11	Not Mentioned	Scheduled Tribe	544	18 (3.30)	0	18 (3.30)	0	0	0	0	0	0	0	0	0	0	α-thal trait -64	9
12	Garahwa	Not Mentioned	24	-	-	2 (8.33)	0	-	-	-	-	-	-	-	-	-	-	1
13	Tatanagar(East Singhbhum)	Santhal	48	-	-	8 (16.66)	0	-	-	-	-	-	-	-	-	-	-	1
14	Ranchi	Not Mentioned	16	-	-	0	0	-	-	-	-	-	-	-	-	-	-	1
15	Sahebganj	Santhal	99	-	-	0	0	-	-	-	-	-	-	-	-	-	-	1

Table 19: Hospital based studies

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Ranchi	Not Mentioned	107	13 (12.10)	5 (4.60)	10 (9.30)	12(11.20)	7 (6.50)	0	0	0	0	0	0	9 (8.40)	7 (6.50)	HbJ Triat-1	10
2	Ranchi	Not Mentioned	1044	156 (14.94)	*112 (10.70)	55 (5.27)	128 (12.26)	128 (12.26)	2 (0.20)	0	13 (1.25)	3 (0.30)	0	0	0	0	HbSD Punjab --2,HbJ Merrut trait --1	11
3	Ranchi	Tibals, Non-Tribals	100	10 (10.00)	7 (7.00)	8 (8.00)	12 (12.00)	11 (11.00)	0	0	5 (5.00)	0	0	0	0	0	0	12

*β-thal major/intermedia

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Bihar

Bihar is the third-largest state by population and borders Uttar Pradesh to its west, Nepal to the north, the northern part of West Bengal to the east, and Jharkhand to the south. The Bihar plain is split by the river Ganges, which flows from west to east. As per the 2011 Census, the total population is 104 million with 15.91% belonging to scheduled castes and 1.28% to scheduled tribes. Hindus constitute the major religious group followed by Muslims, Christians, Buddhists and Sikhs.

A total of 4 studies were available, of which 3 were population based prevalence studies and one was a hospital based study. Fig.5 shows that only one district (shaded areas) has been covered in the state. The names of the districts covered were not mentioned in three studies.



Fig 5: Map of Bihar showing the studied areas

Only 2 tribal groups were studied from 1 district (Tables 20 and 21). The names of the non-tribal groups studied were not mentioned.

Table20: Districts covered in Bihar

Purnia				

Table 21: Tribal populations studied

Lohra	Santhal			

The single study among non tribal populations (Table 22) reported only the presence of βTT(4.81%). On the other hand, population studies in the tribal groups encountered βTT, AS, AE and α-thalassemia (Table 23). Hospital based studies picked up cases of β-thal major, β-thal intermedia and HbEE (Table 24).

Table 22: Population studies in Non-Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Not Mentioned	Not Mentioned	83	4 (4.81)	0	0	0	0	0	0	0	0	0	0	0	0	0	1

Table 23: Population studies in Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Purnia	Santal	214	0	0	2 (0.93)	0	0	0	0	0	0	0	0	0	0	0	2
2	Purnia	Lohra	111	0	0	1 (0.90)	0	0	0	0	0	0	0	0	0	0	0	2
3	Not Mentioned	Not Mentioned	231	7 (3.03)	0	0	0	0	2 (0.86)	0	0	0	0	0	0	0	α-thal trait -22	3

Table 24: Hospital based studies

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Not Mentioned	Not Mentioned	100		52 (52.00)	-	-	-		31 (31.00)	-	-	-	-	-	-	β-thal intermedia --12,*5	4

*Includes β-thal trait/AE & other haemoglobinopathies

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North Eastern Region

The North Eastern region includes the states of Assam, Arunachal Pradesh, Mizoram, Meghalaya, Manipur, Nagaland and Tripura. A total of 47 articles which included population, antenatal and hospital based studies were compiled. The number of districts along with the articles that were available for compilation in each state are shown in Table 1.

Table 1: Studies available from the North Eastern Region

States/Union Territory	Districts	No of Studies
Assam	10	23
Arunachal Pradesh	3	5
Mizoram	Not mentioned	1
Meghalaya	2	6
Manipur	4	3
Nagaland	1	2
Tripura	1	7
Total	**21**	**47**

Population based prevalence studies on β thalassaemia and other haemoglobinopathies were available from all the states. Reports on antenatal screening and hospital based studies were available from Assam, Meghalaya and Tripura while newborn screening was done only in Tripura. The states (shaded areas) covered in the North Eastern region are shown in Fig. 1.



Fig 1 –Map of India showing the state covered.

Assam

Assam is in the southern part of the eastern Himalayas along the Brahmaputra and Barak river valleys. It is bordered by Bhutan and Arunachal Pradesh to the north, Nagaland and Manipur to the east, Meghalaya, Tripura, Mizoram and Bangladesh to the south, and West Bengal to the west. As per the 2011 Census, the total population is 31.2 million of which 12.45% belong to scheduled castes and 7.45% are scheduled tribes. Assamese is the official language of the state, while Bengali is the co-official language in Assam and the official one in the Bodo land Territorial Region.

A total of 23 studies were available of which 16 were population based prevalence studies mainly on β thalassemia trait and HbE trait, one was on antenatal screening and one included both population and antenatal screening while 6 were hospital based studies. Fig 2 shows that studies have been conducted among 10 districts (shaded areas) in the state. Five studies did not mention the names of the districts covered.

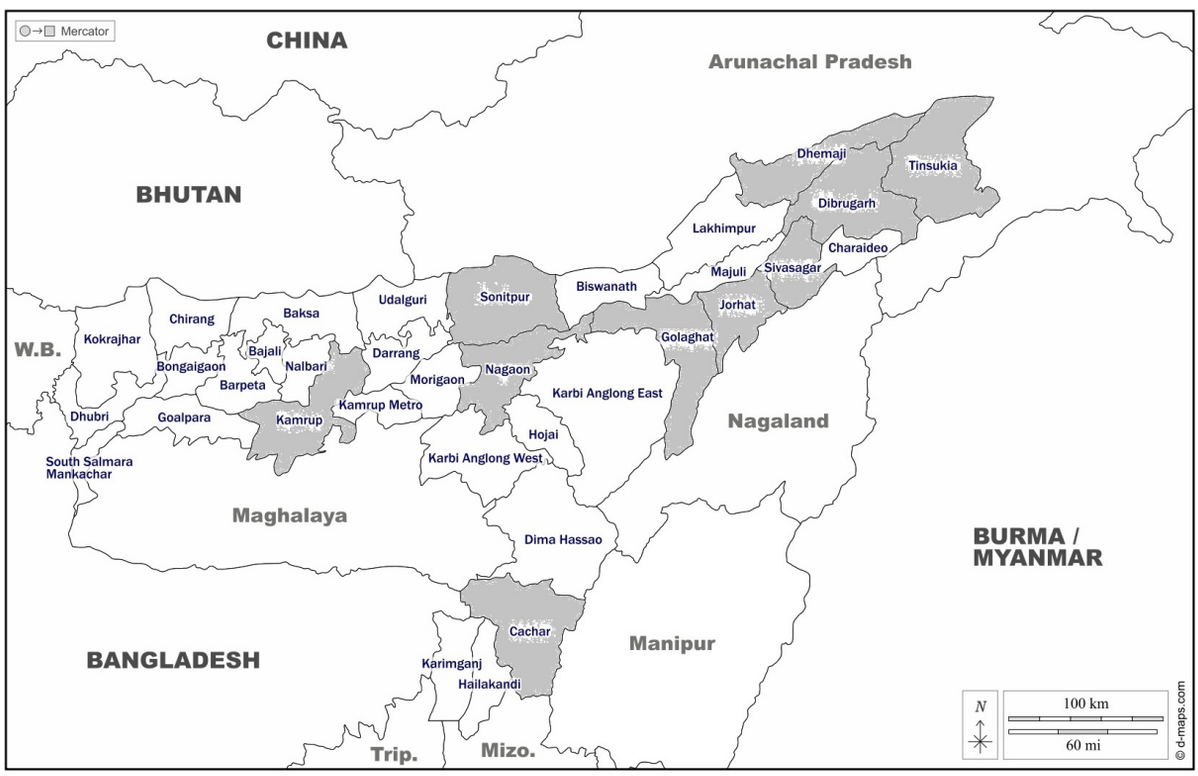

Fig 2: Map of Assam showing the studied areas

The number of districts covered along with the population groups (non-tribal and tribal) studied are shown in Tables 2-4. A total of 13 non-tribal and 18 tribal groups were studied from 10 districts.

Table 2: Districts covered in Assam

Cachar	Dhemaji	Dibrugarh	Golaghat	Jorhat
Kamrup	Nagaon	Sivasagar	Sonitpur	Tinsukia

Table 3: Non-Tribal populations studied

Assamese	Brahamin	Kaibatra	Kalita	Keot
Koch	Mech	Muslim	Muttok	Rajbanshi
Shut	Sikh	Sunni		

Table 4: Tribal populations studied

Ahom	Bodo	Chutiya	Deori	Garo
Hmar	Kachari	Karbi	Lalung	Mikir
Mishing	Munda	Naga	Rabha	Rengma Naga
Santhal	Sonowal	Totos		

Population based studies (Tables 5 and 6) indicated the presence of β thalassaemia trait in both non-tribal and tribal groups and the prevalence varied from 0.42 to 8.64%. Among the non-tribal populations, the prevalence of HbE trait varied from 13.20 to 36.27% and HbE homozygous from 0.13 to 5.60% while in tribal groups the prevalence of HbE trait and homozygous HbE ranged from 0.85 to 55.47% and 0.33 to 45.45% respectively. Few population based studies picked up HbSE disease cases in tribal populations. The presence of the HbS gene with a high prevalence of the trait (AS) and homozygous condition(SS) was mainly reported among the tea garden tribal groups. Antenatal screening (Table 7) and hospital based studies (Table 8) reported β thalassaemia and all other haemoglobinopathies including unusual variants like HbJ Meerut, HbSD Punjab, HbED and HbSE disease.

Table 5: Population studies in Non- Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Lower Assam	Asamese	112	-	-	-	-	-	18(15.90)	3(2.80)	-	-	-	-	-	-	-	1
2	Upper Assam	Asamese	133	-	-	-	-	-	26(19.50)	5(3.80)	-	-	-	-	-	-	-	1
3	Kamrup (Guwahati)	Rajbanshi	102	-	-	-	-	-	37(36.27)	5(4.90)	-	-	-	-	-	-	-	2
4	Kamrup (Guwahati)	Kalita	104	-	-	-	-	-	20(19.20)	2(1.90)	-	-	-	-	-	-	-	2
5	Kamrup (Guwahati)	Kaibarta	101	-	-	-	-	-	25(24.80)	1(0.13)	-	-	-	-	-	-	-	2
6	Kamrup (Guwahati)	Muslim	104	-	-	-	-	-	20(19.20)	1(1.00)	-	-	-	-	-	-	-	2
7	Dibrugarh	Muslim (Garia)	205	-	-	-	-	-	27(13.20)	1(0.50)	-	-	-	-	-	-	-	3
8	Dibrugarh	Muslim (Maria)	155	-	-	-	-	-	37(23.80)	6(3.80)	-	-	-	-	-	-	-	3
9	Dibrugarh	Not Mentioned	*4899	76 (1.55)	0	0	0	0	1235 (25.21)	261 (5.33)	74 (1.51)	1 (0.02)	0	0	5 (0.10)	4 (0.08)	0	4
10	Not Mentioned	Sikh	107	-	-	-	-	-	35(32.70)	6(5.60)	-	-	-	-	-	-	-	5

*Includes tribal/non tribals.

Table 6: Population studies in Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Not Mentioned	Ahom	82	-	-	-	-	-	37 (45.10)	11 (13.40)	-	-	-	-	-	-	-	6
2	Not Mentioned	Ahom	129	-	-	-	-	-	60 (46.50)	15 (11.60)	-	-	-	-	-	-	-	1
3	Dibrugarh	Kachari	555	-	-	-	-	-	276 (49.73)	145 (26.12)	-	-	-	-	-	-	-	7
4	Dibrugarh	Ahom	82	-	-	-	-	-	37 (45.12)	11 (13.41)	-	-	-	-	-	-	-	7
5	Dibrugarh	Ahom	129	-	-	-	-	-	60 (46.51)	15 (11.62)	-	-	-	-	-	-	-	7

(contd.)

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
6	Kamrup	Garo	135	-	-	-	-	-	50(37.04)	42(31.11)	-	-	-	-	-	-	-	8
7	Kamrup	Rabha	128	-	-	-	-	-	71(55.47)	33(25.78)	-	-	-	-	-	-	-	8
8	Kamrup	Kachari	131	-	-	-	-	-	62(47.33)	41(31.30)	-	-	-	-	-	-	-	8
9	Nagaon	Lalung	114	-	-	-	-	-	58(50.88)	22(19.30)	-	-	-	-	-	-	-	8
10	Kamrup	Mikir	131	-	-	-	-	-	36(27.48)	8(6.11)	-	-	-	-	-	-	-	8
11	Dibrugarh	Ahom	119	-	-	-	-	-	56(47.05)	20(16.80)	-	-	-	-	-	-	-	3
12	Dibrugarh	Sonowal	106	-	-	-	-	-	48(45.28)	18(16.70)	-	-	-	-	-	-	-	3
13	Dibrugarh	Chutiya	62	-	-	-	-	-	29(46.77)	4(6.45)	-	-	-	-	-	-	-	3
14	Kamrup (Guwahati)	Bodo Kachari	110	-	-	-	-	-	42 (38.18)	50 (45.45)	-	-	-	-	-	-	-	3
15	Kamrup (Guwahati)	Karbi	110	-	-	-	-	-	32 (29.09)	9(8.18)	-	-	-	-	-	-	-	3
16	Cachar	Rengma Naga	148	-	-	-	-	-	*0.01-0.02		-	-	-	-	-	-	-	9
17	Cachar	Hamar	81	-	-	-	-	-			-	-	-	-	-	-	-	9
18	Cachar	Naga	148	0	-	0	0	-	2 (1.35)	0	-	-	-	-	-	-	-	10
19	Cachar	Hamar	81	0	-	0	0	-	2 (2.46)	0	-	-	-	-	-	-	-	10
20	Dibrugarh (Sivasagar)	Ahom,Garo	234	1 (0.43)	0	1 (0.43)	0	-	81 (34.61)	36 (15.38)	-	-	-	-	-	-	HbSE disease - 1	11
21	Dibrugarh	Mishing	52	-	-	-	-	-	*0.403		-	-	-	-	-	-	-	12
22	Dibrugarh	Devri	29	-	-	-	-	-	*0.569		-	-	-	-	-	-	-	12
23	Not Mentioned	Mishing	44	0	-	-	-	-	16 (36.36)	6 (13.63)	0	-	-	-	-	-	HbSE disease - 1	13
24	Not Mentioned	Sonowal	100	0	-	-	-	-	36 (36.00)	24 (24.00)	2 (2.00)	-	-	-	-	-	0	13
25	Not Mentioned	Deori	45	0	-	-	-	-	18 (40.00)	17 (37.78)	0	-	-	-	-	-	0	13
26	Not Mentioned	Ahom	54	0	-	-	-	-	28 (51.85)	9 (16.67)	0	-	-	-	-	-	0	13
27	Not Mentioned	Munda	118	5 (4.24)	-	-	-	-	1 (0.85)	0	0	-	-	-	-	-	0	13

(contd.)

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
28	Not Mentioned	Santhal	89	0	-	-	-	-	1 (1.12)	0	0	-	-	-	-	-	0	13
29	Dibrugarh (Sivasagar)	Ahom	238	1 (0.42)	-	-	-	-	119 (50.00)	39 (16.40)	0	-	-	-	-	-	-	14
30	Not Mentioned	Totos	443	-	-	-	-	-	218 (49.21)	85 (19.20)	-	-	-	-	-	-	-	15
31	Tinsukia, Dibrugarh (Sivasagar) Jorhat, Golaghat, Dhemaji	Tea Garden Tribals	1204	104 (8.64)	15 (1.25)	199(16.53)	59 (4.90)	16(1.33)	14(1.16)	4(0.33)	4 (0.33)	0	0	-	0	0	HPFH with S window-2, HbSE disease - 2	16

* HbE gene frequency

Table 7: Antenatal screening

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Dibrugarh	Ahom,Kachari	190	-	-	-	-	-	60(31.57)	18(9.47)	-	-	-	-	-	-	-	17
2	Dibrugarh	Not Mentioned	1917	25 (1.30)	0	9 (0.50)	0	3 (0.20)	394 (20.50)	102 (5.30)	24 (1.30)	0	0	0	0	8 (0.40)	-	4

Table 8: Hospital based studies

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Kamrup (Guwahati)	Not Mentioned	800	145 (18.10)	17(2.10)	26 (3.20)	16 (2.00)	1 (0.12)	188 (23.50)	52 (6.50)	72(9.00)	0	0	0	0	0	0	18
2	Upper Assam	Not Mentioned	9000	313(3.48)	32 (0.36)	189(2.10)	203(2.26)	53(0.59)	2294 (25.48)	1892 (21.02)	114 (1.26)	6 (0.07)	0	0	33(0.37)	5 (0.06)	HbJ Meerut trait-8,HbSD Punjab -2, HbED Punjab -2, HbSE disease -13	19

(contd.)

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
3	Cachar	Not Mentioned	100	10 (10.00)	0	5 (5.00)	2 (2.00)	0	0	10 (10.00)	8 (8.00)	0	0	0	0	0	0	20
4	Upper Assam	Not Mentioned	1200	70 (5.80)	16(1.33)	0	0	5 (0.42)	0	0	28(0.33)	0	0	0	0	0	0	21
5	Jorhat	Brahim, Kalita, Koch, Keot, Muttok, Sunni, Ahom, Chutya, Shut, Mech, Kaibarta, Bodo, Kachari, Deori, Mishing	800	1 (0.12)	2 (0.25)	12 (1.50)	17 (2.12)	12(1.50)	243 (30.37)	100 (12.50)	30 (3.75)	0	0	0	0	0	0	22
6	Sonitapur (Tezpur)	Not Mentioned	75	16 (21.30)	5 (6.70)	14(18.70)	6 (8.00)	1 (1.30)	20 (26.60)	6(8.00)	7 (9.30)	0	0	0	0	0	0	23

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Arunachal Pradesh

Arunachal Pradesh was formed from the erstwhile North-East Frontier agency region, and shares a border with Assam and Nagaland to the south, international borders with Bhutan in the west, Myanmar in the east, and a disputed border with China in the north. Of the total 1.3 million population, 68.79% belong to scheduled tribes (Census 2011). Christians are the major religious group followed by Hindus, Buddhists and Muslims.

All the 5 available studies were population based screening on hemoglobinopathies in tribal groups. Fig 3 shows that studies have been conducted only in three districts (shaded areas) in the state. The names of the districts have not been mentioned in two studies.

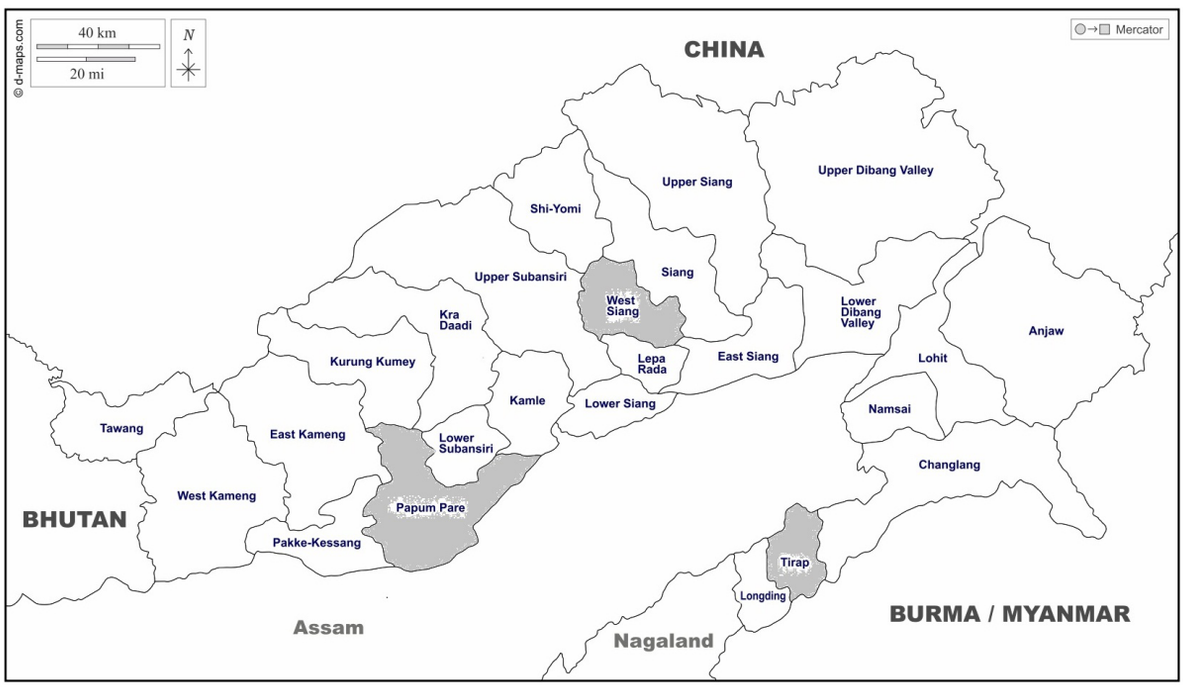

Fig 3: Map of Arunachal Pradesh showing the studied areas

A total of 10 tribal groups were studied from 3 districts (Tables 9 and 10).

Table 9: Districts covered in Arunachal Pradesh

Itanagar	Tirap	West Siang		

Table 10: Tribal populations studied

Adi	Apatani	Bokar	Bori	Galong
Hillmiri	Miniyoung	Nishi	Pallibo	Tagin

Population based studies indicated the presence of βTT, AE, EE and E-β thal. A high prevalence of HbE gene was encountered and the prevalence of HbE trait varied from 5.61 to 35.13% while the prevalence of HbE homozygous ranged from 1.51 to 3.70% (Table 11).

Table 11: Population studies in Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Not Mentioned	Adi, Nishi, Aptani	568	-	-	-	-	-	89(15.7)	10(1.76)	-	-	-	-	-	-	-	1
2	Not Mentioned	Not Mentioned	89	0	0	0	0	0	5 (5.61)	0	1 (1.12)	0	0	0	0	0	0	2
3	Itanagar	Adi, Galong	179	1 (0.55)	0	0	0	0	25(13.96)	4(2.23)	0	0	0	0	0	0	0	3
4	West Siang, Papum Pare (Itanagar), Tirap	Gallong, Apatani, Miniyoung, Bokar, Tagin, Bori, Nishi, Hillmiri, Pallibo	331	1 (0.30)	-	1 (0.30)	0	-	61 (18.43)	5(1.51)	-	-	-	-	-	-	-	4
5	West Siang	Gallong	108	0	0	1 (0.98)	0	0	38 (35.19)	4(3.70)	0	0	0	0	0	0	0	5

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Mizoram

Mizo-ram means "land of the Mizos". This state shares borders with three of the seven sister states of the North Eastern Region, namely Tripura, Assam and Manipur. The state also shares a border with two neighboring countries, Bangladesh and Myanmar. According to the 2011 Census, Mizoram with a total population of 1 million is the second least populous state in the country. Among all the states of India, Mizoram has the highest concentration of scheduled tribe populations (94.43%) while scheduled castes constitute only 0.11%. It is one of three states of India with a Christian majority (87%).

Only one population based study was available where Mizos were studied however, the name of the study place (district) was not mentioned. The presence of only HbE trait (1.50%) was reported in this study.(Table 12).

References:

1. Dash S, Chhanhimi L ,Chhakchhuak L, Zomawaia E. Screening for haemoglobinopathies and G6PD deficiency among the Mizos of Mizoram: a preliminary study. Indian J Pathol Microbiol. 2005;48:17-18.

Table 12 - Population studies in Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Not Mentioned	Mizos	490	0	0	0	0	0	7 (1.50)	0	0	0	0	0	0	0	0	1

Meghalaya

Meghalaya meaning "abode of clouds" was formed by carving out the two hills from Assam. It is bordered to the south and west by Bangladesh and to the north and east by Assam. As per the 2011 Census, the total population is estimated to be 2.9 million of which 86.15% belong to scheduled tribes and 0.58% are scheduled castes. About 75% of the population pratice Christianity.

A total of 6 studies were available of which 5 were on population based prevalence mainly on HbE and one was on antenatal screening. The districts where the studies have been conducted (shaded areas) are shown in Fig 4.

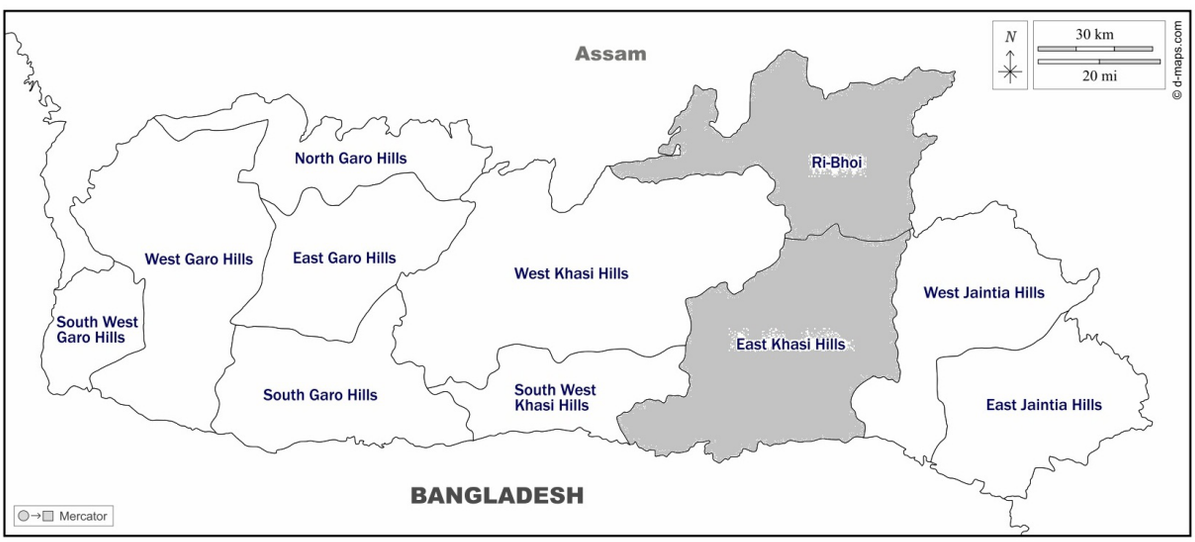

Fig 4: Map of Meghalaya showing the studied areas

The districts covered and the tribal groups studied are shown in Tables 13 and 14. A total of three tribal groups were studied from two districts.

Table 13: Districts covered in Meghalaya

East Khasi Hills	Ri- bhoi			

Table 14: Tribal populations studied

Bodo	Khasi	Khynriem Khasi		

Population based studies have reported only the presence of the HbE gene among the tribal groups and the prevalence of HbE trait (AE) varied from 4.50 to 41.30% while the prevalence of HbE homozygotes (EE) varied from 2.50% to 29.10% (Table 15). Antenatal screening identified cases of βTT, AE, EE, E-β thal and the rare variant HbD Iran (Table 16).

Table 15: Population studies in Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Ri- bhoi	Khasi	80	0	-	-	-	-	33(41.30)	2(2.50)	0	-	-	-	-	-	-	1
2	Ri- bhoi	Khasi	120	0	-	-	-	-	44(36.43)	5(4.16)	0	-	-	-	-	-	-	2
3	East Khasi Hills	Khasi	157	0	-	-	-	-	7(4.50)	0	0	-	-	-	-	-	-	3
4	East Khasi Hills	Bodo	24	0	-	-	-	-	4(16.80)	7(29.10)	0	-	-	-	-	-	-	3
5	East Khasi Hills	Khynriem Khasi	215	0	-	-	-	-	10(4.65)	0	0	-	-	-	-	-	-	4
6	East Khasi Hills	Khasi	302	-	-	-	-	-	0.008*		-	-	-	-	-	-	-	5

*HbE gene frequency

Table 16: Antenatal screening

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	East Khasi	Not Mentioned	467	9 (1.93)	0	0	0	0	46(9.85)	12(2.57)	2 (0.43)	0	0	0	0	0	HbD Iran trait--1	6

References:

1. Das BM, Charavarthy MR, Del Bruck H, Flatz G. High prevalence of HbE in two populations in Assam. Hum Genetik. 1971;12:264-266.
2. Flatz G, Chakaravarti MR, Das BM, Del Bruck H. Genetic survey in the population of Assam. ABO blood groups, glucose-6-phosphate dehydrogenase and hemoglobin type. Hum Hered. 1972;22:323-330.
3. Saha N. Distribution of HbE in several Mongoloid populations of North-East India. Hum Biol. 1990;62:535-544.
4. Das A, Paddaiah G, Sarker BN, Rao VR, Ramesh M. Presence of heamoglobin E in Khynriem Khasi of Meghalaya, India. Indian J Multi Res. 2009;5:337-340.
5. Das B, Sengupta S. Hemoglobin E among the Khasi of east Khasi hill, Meghalaya. In: Baruah T, (Ed). People of contemporery north east India. Pratishruti Publication, Guwahati. 2011:pp26-37.
6. Shah N, Khonglah Y, Raphael V, Swer B, Nath C, Singh AS. Antenatal screening for hemoglobinopathies with HPLC. Rec Adv Path Lab Med. 2018;4:1-8.

Manipur

Manipur is bordered by Nagaland to the north, Mizoram to the south, Assam to the west and Myanmar to the east. The total population is around 3 million and scheduled castes constitute 3.8 % and scheduled tribes 35.1%. Of the total population, 57.2% live in the valley districts and the remaining 42.8% in the hill districts.

All the 3 available studies were population based. Fig 5 shows that studies have been conducted in 4 districts (shaded areas) in the state. One study did not mention the names of the districts covered.

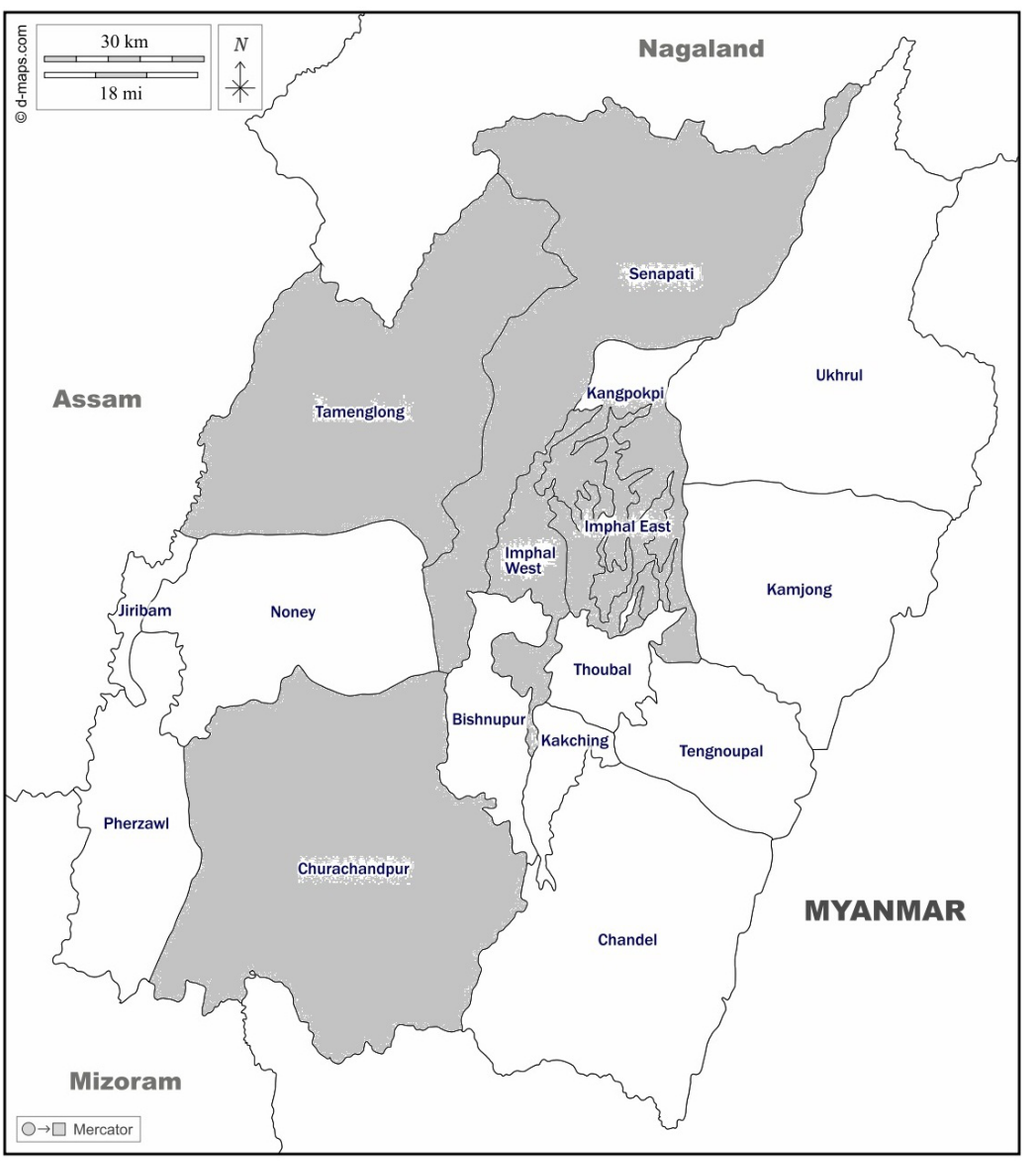

Fig 5: Map of Manipur showing the studied areas

Tables 17-19 show the districts covered and the non-tribal and tribal groups studied. A total of 2 non-tribal and 4 tribal groups were studied from 4 districts.

Table 17: Districts covered in Manipur

Churachandpur	Imphal	Senapati	Tamenglong	

Table 18: Non-Tribal populations studied

Brahmin	Muslim			

Table 19: Tribal populations studied

Hill Kabui	Koireng	Meitei	Simte	

The presence of the HbE gene was reported from both non-tribal and tribal groups. Among the non-tribal groups, the prevalence of HbE trait (AE) and HbE homozygotes (EE) ranged from 0.99 to 4.33% and 0.66 to 2.00% respectively (Table 20). The presence of the HbE gene was seen in most of the tribal groups studied and the prevalence of HbE trait (AE) varied from 2.44 to 15.81 % and HbE homozygotes (EE) from 0.37 to 1.92%. Few cases of E-β thal were also reported (Table 21).

References:

1. Thiyam U, Hoobam S, Singh YM, Sharma DC. A study of haemoglobin patterns among the students of RIAM. J Evol Med Dent Sci. 2015;4:15979-15981.
2. Achoubi N, Asghar M, Saraswathy KN, Murry B. Prevalence of β-thalassemia and hemoglobin E in two migrant populations of Manipur, north east India. Genet Test Mol Biomarkers. 2012; 16: 1195-2000.
3. Singh MR, Choudhury B, Singh TS. Hemoglobin E distribution in four endogamous populations of Manipur India. Eurasian J Anthropol. 2010;1:109–117.

Table 20: Population studies on Non-Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Not Mentioned	Not Mentioned	100*	0	-	-	-	-	0	2(2.00)	-	-	-	-	-	-	-	1
2	Not Mentioned	Brahmin	300	1 (0.33)	0	0	0	0	13 (4.33)	0	0	0	0	0	0	0	0	2
3	Not Mentioned	Muslim	302	0	0	0	0	0	3 (0.99)	2 (0.66)	0	0	0	0	0	0	0	2

*Students belonging to different ethnic groups.

Table 21: Population studies on Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Tamenglong	Hill Kabui	270	0	0	0	0	0	17(6.30)	1(0.37)	0	0	0	0	0	0	0	3
2	Senapati	Koireng	174	0	0	0	0	0	10(5.75)	0	0	0	0	0	0	0	0	3
3	Churachandpur	Simte	164	0	0	0	0	0	4(2.44)	0	0	0	0	0	0	0	0	3
4	Imphal	Meitei	626	2 (0.32)	0	0	0	0	99(15.81)	12(1.92)	3 (0.48)	0	0	0	0	0	Unknown-1	3

Nagaland

Nagaland is bordered by Arunachal Pradesh to the north, Assam to the west, Manipur to the south and the Sagaing Region of Myanmar to the east. The total population is almost 1.9 million of which 86.48% are scheduled tribes (Census 2011). The Naga people constitute over 90% of the population.

Only two population based prevalence studies mainly on HbE were available. Fig.6 shows that only one district (shaded area) has been covered in the state. The names of the districts have not been mentioned in one study.



Fig 6: Map of Nagaland showing the studied areas

Only two tribal groups were studied from one district (Tables 22 and 23).

Table 22: Districts covered in Nagaland

Mokokchung				

Table 23: Tribal populations studied

Ao Nagas	Rangma Naga			

Only two studies were available from tribal populations and they only reported the presence of HbE trait (AE) and HbE homozygotes (EE) (Table 24).

References:

1. Saha N. Dsitribution of HbE In several Mongoloid populations of northeast india. Hum Biol. 1990;62:535-544.
2. Kar S, Seth S, Seth PK. Prevalence of malaria in Ao Nagas and its association with G6PD and HbE. Hum Biol. 1992;64:187-197.

Table 24: Population studies on Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Not Mentioned	Rangma Naga	148	-	-	-	-	-	2 (1.40)	0	-	-	-	-	-	-	-	1
2	Mokokchung	Ao Naga	708	-	-	-	-	-	92(12.90)	12(1.70)	-	-	-	-	-	-	-	2

Tripura

Tripura is the third-smallest state in the country and is bordered by Bangladesh to the north, south, and west, and Assam and Mizoram to the east. According to the Census 2011, the total population is 3.6 million, constituting 0.3% of the country's population. The scheduled castes and scheduled tribes constitute 17.83% and 31.76% respectively of the total population. Bengalis make up almost 70 % of the population while the remaining 30 % are Tripuris.

A total of 7 studies were available of which 5 were on population based prevalence on β thalassemia and HbE while one was on newborn screening and one was a hospital based study. Fig 7 shows that studies have been conducted only in one district (shaded area) in the state. Six studies did not mention the names of the districts.

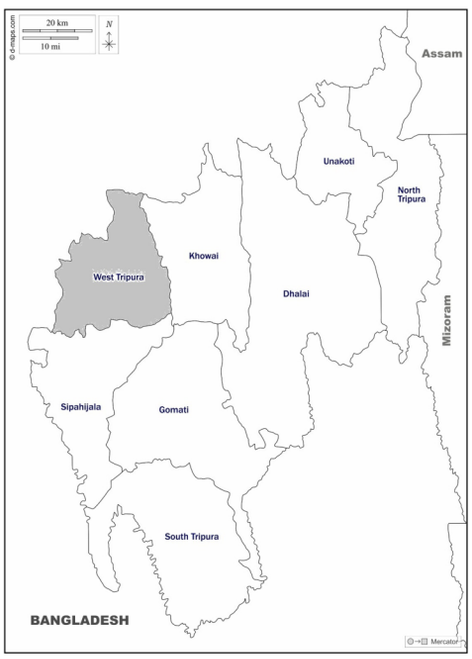

Fig 7: Map of Tripura showing the studied areas

Tables 25-26 show the districts covered and the tribal groups studied. A total of 12 tribal groups were studied from one district.

Table 25: Districts covered in Tripura

Agartala				

Table 26: Tribal populations studied

Adi	Chakma	Darlong	Halam	Jamatia
Marek	Mog	Mora Singh	Noatia	Riang
Tripuri	Uchai			

Population based studies among the tribal groups indicated that apart from the presence of βTT, βTM, HbAS, HbAE, HbEE and HbE β thal individuals with other hemoglobinopathies like HbSE were also encountered (Table 27). Newborn screening among the non-tribal and tribal groups have identified few cases of βTT, AS, AE, EE, Eβ thal, AD, D-β thal and HbD Punjab/HbE disease (Table 28). Apart from β-thalassemia and other common hemoglobinopathies, hospital based studies have identified cases of HbD Punjab trait, δβ thal, HPFH and HbSE disease (Table 29).

Table 27: Population studies in Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Not Mentioned	Riang	51	0	-	-	-	-	0	0	-	-	-	-	-	-	-	1
2	Not Mentioned	Tripuri	310	26 (8.38)	6 (1.93)	0	0	0	142 (45.80)	46 (14.83)	12(3.87)	0	0	0	0	0	0	2
3	Not Mentioned	Not Mentioned	889	1 (0.11)	0	1 (0.11)	0	0	425 (47.80)	147 (16.50)	0	0	0	0	0	0	HbSE disease - 4	3
4	Not Mentioned	Not Mentioned	205	0	0	0	0	0	85 (41.46)	32 (15.61)	0	0	0	0	0	0	0	4
5	Not Mentioned	Chakma	162	-	-	-	-	-	84 (51.80)	16 (9.80)	-	-	-	-	-	-	-	5
6	Not Mentioned	Darlong	57	-	-	-	-	-	28 (49.10)	15 (26.30)	-	-	-	-	-	-	-	5
7	Not Mentioned	Debbarman (Tripuri)	104	-	-	-	-	-	51 (49.00)	17 (16.30)	-	-	-	-	-	-	1*	5
8	Not Mentioned	Halam	60	-	-	-	-	-	30 (50.00)	11 (18.30)	-	-	-	-	-	-	-	5
9	Not Mentioned	Jamatia	65	-	-	-	-	-	25 (38.40)	9 (13.80)	-	-	-	-	-	-	-	5
10	Not Mentioned	Marek	64	-	-	-	-	-	32 (50.00)	20 (31.20)	-	-	-	-	-	-	-	5
11	Not Mentioned	Mog	74	-	-	-	-	-	31(41.20)	4(5.40)	-	-	-	-	-	-	-	5
12	Not Mentioned	Mora Singh	20	-	-	-	-	-	0	3 (15.00)	-	-	-	-	-	-	-	5
13	Not Mentioned	Noatia	27	-	-	-	-	-	3(11.10)	4 (14.80)	-	-	-	-	-	-	-	5
14	Not Mentioned	Riang	48	-	-	-	-	-	24 (50.00)	10 (20.80)	-	-	-	-	-	-	-	5
15	Not Mentioned	Tripuri	89	-	-	-	-	-	48 (53.90)	11 (12.40)	-	-	-	-	-	-	5*	5
16	Not Mentioned	Uchi	70	-	-	-	-	-	42 (60.00)	15 (21.40)	-	-	-	-	-	-	-	5

* Includes -βTT-1, AS-1 & HbSE disease - 4

Table 28: Newborn screening

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Agartala	Non tribal	2014	2 (0.09)	0	11 (0.54)	0	0	84(4.17)	4(0.19)	1 (0.04)	2 (0.09)	0	0	0	0	0	6
2	Agartala	Tribal	386	2 (0.51)	0	4 (1.03)	0	0	141 (36.52)	76 (19.68)	0	0	0	2 (0.51)	0	0	HbD Punjab /HbE disease -2	6

Table 29: Hospital based studies

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Not Mentioned	Not Mentioned	1661	656(39.49)	87 (5.23)	15 (0.90)	4 (0.24)	6 (0.36)	199 (11.98)	20 (1.20)	134 (8.06)	0	0	-			HbD Punjab trait, δβ thal/HPFH trait, α-thal trait -7	3
2	Not Mentioned	Debbarman, Chakma, Jamatia, Adi, Tripuri, Riyang, Naotia	698	25 (3.58)	5 (0.72)	0	0	0	286 (40.90)	107 (15.33)	10 (1.43)	0	0	0	0	0	HbSE disease-3	7

References:

1. Kumar N, Sastry DB. A genetic study among the Riang : a Mongoloid tribe of Tripura. (north east India). Z Morph Anth. 1961;15.
2. De M, Chakraborty G, Das SK, Bhattacharya DK, Talukder G. Molecular studies of haemoglobin-E in tribal populations of Tripura. The Lancet. 1997.349:1297.
3. Das SK, De M, Bhattacharya DK, Sengupta B, Das N, Talukder G. Interaction of different hemoglobinopathies in eastern India with a view to establish gnotype–phenotype correlation. Am J Hum Biol. 2000;12:454-459.
4. Sengupta B, De M, Dasgupta I, Poddar S, Chakrabarti S, Talukder G, Kukreti R, Brahmachari S. Comparative study of haemoglobinopathies in tribal populations of Arunachal Pradesh and Tripura, north east India. Int J Hum Genet. 2002;2:169-172.
5. Das SK, De M, Sengupta B, Das N, Bhattacharya DK, Talukdar G. High incidence of hemoglobin E in tribal populations of Tripura, north east, India. The Antropologist. (special issue) 2002;1:105-108.
6. Upadhye D, Das RS, Ray J, Acharjee A, Ghosh K, Colah RB, Mukherjee MB. Newborn screening for hemoglobinopathies and red cell enzymopathies in Tripura state: a malaria-endemic state in north east India. Hemoglobin. 2018;42:43-46.
7. De M, Halder A, Podder S, Sen R, Chakrabarty S, Sengupta B, Chakraborty T, Das U, Talukder G. Anemia and hemoglobinopathies in tribal population of eastern and north-eastern India. Hematology. 2006;11:371-373.

Northern Region

The Northern region includes the Union Territory of Delhi and the states of Punjab, Haryana, Uttar Pradesh, Uttarakhand, Himachal Pradesh and Jammu & Kashmir. A total of 34 articles which included population, antenatal and hospital based studies were compiled. Newborn screening studies were not found in this region. The number of districts along with the articles that were available for compilation in each state are shown in Table 1.

Table 1: Studies available from the Northern Region

States/Union Territory	Districts	No of Studies
Delhi	Not Mentioned	12
Punjab	2	3
Haryana	2	3
Uttar Pradesh	3	6
Uttarakhand	7	4
Himachal Pradesh	Not Mentioned	2
Jammu & Kashmir	1	4
Total	**15**	**34**

Majority of the studies in all the states/UTs were on population based prevalence on β thalassaemia and other haemoglobinopathies and hospital based studies among the non-tribal populations. Antenatal screening was reported only from Delhi and Punjab. The states/UTs (shaded areas) covered in the Northern region are shown in Fig. 1.



Fig 1: Map of India showing the states/UTs covered

Delhi

Delhi officially the National Capital Territory (NCT) of Delhi, is a city containing New Delhi, the capital of India. Delhi shares borders with the state of Uttar Pradesh in the east and Haryana in the remaining directions. According to the 2011 Census, Delhi's city population is over 11 million while the NCT's population is about 16.8 million. Hindus are the major religious group followed by Muslim, Sikh, Jain, Christian and Buddhist.

Of the 12 studies which were available, 2 were on population screening, 3 on antenatal screening and remaining 7 were hospital based studies. None of the studies mentioned the names of the districts and population groups covered.

Population based studies (Table 2) indicated that apart from the presence of βTT, Hb AD Punjab and Hb DD Punjab, individuals with other hemoglobinopathies like δβ-thal trait/ HPFH trait and Hb SD Punjab were also encountered. Antenatal screening (Table 3) and Hospital based studies (Table 4) reported β thalassaemia and all other haemoglobinopathies including many unusual Hb variants and compound heterozygous combinations like Hb Lepore trait, HbH Disease, HbQ India trait, HbJ Meerut trait, HbD Iran trait, α-thal trait, HbSD Punjab, Hb Hope and Hb J Oxford.

Table 2: Population studies in Non-Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Not mentioned	Not mentioned	5408	297 (5.50)	0	0	0	0	0	0	0	59(1.10)	0	0			δβ-thal trait / HPFH trait --14	1
2	Not mentioned	Not mentioned	484*	-	-	-	-	-	-	-	-	23(4.75)	9 (1.85)	4 (0.82)	-	-	HbSD Punjab--2	2

*Total number of structural haemoglobin variants

Table 3: Antenatal screening

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Not mentioned	Not mentioned	1000*	79 (7.90)	-	-	-	-	-	-	-	-	-	-	-	-	-	3
2	Not mentioned	Hindu, Muslim	7737	388(5.01)	0	4 (0.05)	0	0	45(0.58)	0	0	28 (0.36)	0	0	0	0	Uncommon Hb variants--3	4
3	Not mentioned	Not mentioned	2000#	59 (2.90)	0	1 (0.05)	0	0	1 (0.05)	0	1 (0.05)	1 (0.05)	0	0	0	0	0	5

*Includes Delhi, Uttar Pradesh, Haryana, Punjab, Bihar
Includes only women with microcytic hypocromic anemia.

Table 4: Hospital based studies

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Not mentioned	Not mentioned	230	-	-	-	-	-	4 (1.70)	3(1.30)	4 (1.73)	-	-	-	-	-	-	6
2	Not mentioned	Not mentioned	1032	176(17.00)	4 (0.40)	24 (2.30)	18 (1.70)	6(0.50)	8 (0.80)	6(0.60)	6 (0.60)	10(1.00)	0	0	0	0	0	7

(contd.)

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
3	Not mentioned	Not mentioned	800	145(18.10)	23 (2.90)	11 (1.40)	4 (0.50)	6 (0.80)	9 (1.10)	1(0.10)	10(1.30)	7 (0.90)	0	1 (0.10)	6 (0.80)	1 (0.10)	Hb Lepore trait-- 1 HbH disease--1 HbQ India trait--3 HbJ Meerut trait --1 HbD Iran trait--3 α-thal trait–11,Hb-SD Punjab--1	8
4	Not mentioned	Not mentioned	2600*	232(8.90)	15 (0.60)	0	1 (0.03)	2 (0.07)	5 (0.19)	2 (0.07)	6 (0.23)	13 (0.50)	0	0	0	25(0.96)	Hb Lepore--trait 1 ,HbQ India trait--5, HbQ-India and β thal trait-- 2,HbD Iran trait--1 , HbJ Meerut trait-- 1, β-thal inter-media --16	9
5	Not mentioned	Not mentioned	4800@	216 (4.50)	9 (0.20)	9 (0.20)	0	0	28 (0.60)	2(0.04)	2 (0.04)	15(0.31)	0	4 (0.08)	3 (0.06)	0	HbJ Meerut trait --1,Hb Hope--1	10
6	Not mentioned	Not mentioned	110	62(56.36)	6 (5.45)	1 (0.90)	1 (0.90)	2 (1.81)	7 (6.36)	1 (0.90)	8 (7.27)	8 (7.27)	1 (0.90)	0	0	0	HbH disease --4,HbD Iran trait --1, HbJ Oxford–2 , HbSE disease --1, β-Thal intermedia--5	11
7	Not mentioned	Not mentioned	126#	-	-	-	78(62.00)	-	-	-	-	-	-	-	-	-	HbS/HPFH--2 HbS/Gϒ $(Aγδβ)^0$ thal --1	12

* Cases are from New Delhi, Haryana,Uttar Pradesh & Jammu & Kashmir

@ Includes cases of microcytic hypocromic anemia, not responding to conventional treatment, clinicaly suspected cases of hemoglobinopathies,

Includes only sickle cell disease cases.

References:

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Punjab

Punjab is a state in northern India bordered by Himachal Pradesh to the north and northeast, Haryana to the south and southeast, and Rajasthan to the southwest, by the union territory of Chandigarh to the east, and by Jammu and Kashmir to the north. It is bordered by a province of Pakistan to the west. The total population is 27.7 million of which 31.9% are scheduled castes and 31.3% are other backward class populations. Punjabis are the main ethnic group, with Sikhs and Hindus as the major religious groups.

A total of 3 population based prevalence studies were available out of which one study also reported both population and antenatal screening among the non tribal populations. Fig.2 shows that only two districts (shaded areas) have been covered in the state. One study did not mention the names of the districts covered.



Fig 2: Map of Punjab showing the studied areas

A total of 6 non-tribal groups were studied from 2 districts (Tables 5 and 6).

Table 5: Districts covered in Punjab

Hoshiarpur	Ludhiana			

Table 6: Non-Tribal populations studied

Bania	Brahmin	Jain	Jat Sikh	Kayastha
Khatri				

Population based studies have reported the presence of βTT, AS and AD along with compound heterozygous D- β Thalassemia (Table 7). Antenatal screening picked up few cases of βTT, AS, SS, AE, EE, AD Punjab, DD and δβ-thalassemia (Table 8).

Table 7: Population studies in Non-Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Hosiarpur	Khatri	271	-	-	1 (0.36)	0	-	-	-	-	5 (1.84)	0	-	-	-	-	1
2	Hosiarpur	Jat sikh	467	-	-	0	0	-	-	-	-	1 (0.21)	0	-	-	-	-	1
3	Hosiarpur	Brahmin	355	-	-	0	0	-	-	-	-	1 (0.28)	0	-	-	-	-	1
4	Hosiarpur	Bania	219	-	-	0	0	-	-	-	-	0	0	-	-	-	-	1
5	Ludhiana	Mixed Populations	4997	168 (3.36)	0	3 (0.06)	0	0	3 (0.06)	1 (0.02)	0	62 (1.24)	0	2 (0.04)	0	0	13	2
6	Not Mentioned	Jat Sikh, Jain, Brahmin, Baniya, Khatri, Jat, Kayastha	975*	32 (3.30)	0	1 (0.10)	0	-	0	0	-	8 (0.80)	0	-	-	-	-	3

* Blood donors from Punjab, Haryana, Himachal Pradesh & Jammu & Kashmir.

Table 8: Antenatal screening

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Ludhiana	Not Mentioned	4994	207(4.10)	0	3 (0.10)	0	0	1 (0.02)	1(0.02)	0	47 (0.90)	3 (0.10)	0	2 (0.04)	0	12 (0.20)	2

References:

1. Kaur R, Kaur H. Hemoglobin variants in some ethnic groups of Punjab (north-India). Indian J Hum Genet. 1999;5:57-60.
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Haryana

Haryana was carved out of the former state of East Punjab located in the northern-part of the country. It is bordered by Himachal Pradesh to the north-east, by river Yamuna along its eastern border with Uttar Pradesh, by Rajasthan to the west and south, and the Ghaggar-Hakra River flows along its northern border with Punjab. According to the 2011 Census, the total population of Haryana is 25.35 million of which Hindus constitute the majority followed by Muslims and Sikhs. Haryana has the second largest Sikh population in India after Punjab, and they mostly live in the districts adjoining Punjab,

Of the 3 available studies, 2 were population based and one was a hospital based study. Fig 3 shows the districts (shaded areas) where studies have been conducted in the state. The names of the districts covered have not been mentioned in one study.



Fig 3: Map of Haryana showing the studied areas

The two districts covered are shown in Table 9. The names of the non tribal groups studied have not been mentioned.

Table 9: Districts covered in Haryana

Faridabad	Rothak			

Only two studies were available from non-tribal populations and both the studies reported only the presence of BTT (Table 10). On the other hand, hospital based studies besides reporting βTT, BTM and other common Hb variants like HbS, HbE and HbD, also identified β-thalassemia intermedia, homozygous HPFH, HbD Iran and HbC (Table 11).

Table 10: Population studies in Non-Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Faridabad Dayalpur	Not Mentioned	150	7 (4.66)	-	-	-	-	-	-	-	-	-	-	-	-	-	1
2	Not Mentioned	Not Mentioned	397	14 (3.50)	-	-	-	-	-	-	-	-	-	-	-	-	-	2

Table 11: Hospital based studies

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Rothak	Not Mentioned	4275	318 (7.40)	19 (0.40)	2 (0.05)	2 (0.05)	2 (0.05)	11 (0.26)	6 (0.14)	2 (0.05)	11 (0.26)	0	0	0	1 (0.02)	β- thal intermedia--24, HPFH homozyg-ouse--1,HbD Iran trait--2, HbC trait--1	3

References:

1. Verma AK, Kokkat AJ, Verma IC. Prevalence of beta thalassemia trait in school children in rural Haryana. Indian J Hematol Blood transf. 1996;14: 86-87.
2. Madan N, Sharma S, Sood SK, Colah RB, (Late) Bhatia HM. Frequency of β-thalassemia trait and other hemoglobinopathies in northern and western India. Indian J Hum Genet. 2010;16:16-25.
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Uttar Pradesh

Uttar Pradesh is bordered by Rajasthan to the west, Haryana, Himachal Pradesh and Delhi to the northwest, Uttarakhand and an international border with Nepal to the north, Bihar to the east, Madhya Pradesh to the south, and Jharkhand and Chhattisgarh to the south east. It is the most populous state in India and contributes to 16.2% of the total Indian population. As per the Census 2011, the total population is over 200 million and has the largest scheduled caste population whereas scheduled tribes are less than 1 %.

Of the 6 studies which were available, 5 were on population screening in non-tribal or tribal groups including antenatal screening and one was a hospital based study. The districts where studies were reported (shaded areas) are shown in Fig 4.



Fig 4: Map of Uttar Pradesh showing the studied areas

Tables 12 and 13 show the number of districts covered along with the tribal population groups studied. The names of the non-tribal groups studied were not mentioned. A total of 9 tribal groups were studied from Mirjapur district.

Table 12: Districts covered in Uttar Pradesh

Lucknow	Mirjapur	Varanasi		

Table 13: Tribal populations studied

Bhiya	Chero	Gond	Khairwar	Kol
Panika	Pathari	Raj Gond	Ranatharu	

Among the studies in non-tribal groups (Table 14), the presence of βTT, AS, SS, AE and AD Punjab along with the less common variant Hb Q India were reported. Population based studies indicated the presence of HbS gene in most of the tribal groups studied. The prevalence of sickle cell trait varied from 0.40 to 32.70 % in different studies (Table 15). None of the studies reported the presence of SS cases. Hospital based studies reported β-thalassaemia and all other haemoglobinopathies including β-thalassaemia intermedia, HbSD disease and HbSE disease (Table 16).

Table 14: Population studies in Non- Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Lucknow	Not mentioned	939*	27 (2.80)	0	1 (0.10)	0	0	3 (0.30)	0	0	6 (0.60)	0	0	0	0	HbQ India trait--1	1
2	Varanasi	Mixed Populations	606	21 (3.40)	0	0	0	0	2 (0.30)	0	0	0	0	0	0	0	α-thal trait-40	2

* College students

Table 15: Population studies in Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Mirjapur	Raj Gond	270	-	-	4 (1.40)	-	-	-	-	-	-	-	-	-	-	-	3
2	Mirjapur	Ranatharu	207	-	-	1 (0.40)	-	-	-	-	-	-	-	-	-	-	-	4
3	Mirjapur	Bhiya	19	-	-	0	-	-	-	-	-	-	-	-	-	-	-	4
4	Mirjapur	Chero	19	-	-	0	-	-	-	-	-	-	-	-	-	-	-	4
5	Mirjapur	Khairwar	46	-	-	1 (2.17)	-	-	-	-	-	-	-	-	-	-	-	4
6	Mirjapur	Kol	64	-	-	0	-	-	-	-	-	-	-	-	-	-	-	4
7	Mirjapur	Panika	55	-	-	18(32.70)	-	-	-	-	-	-	-	-	-	-	-	4
8	Mirjapur	Pathari	7	-	-	2 (28.57)	-	-	-	-	-	-	-	-	-	-	-	4
9	Mirjapur	Gond	108	-	-	20(18.50)	-	-	-	-	-	-	-	-	-	-	-	5

Table 16: Hospital based studies

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Lucknow	Not mentioned	1348*	216(16.00)	15 (1.11)	17 (1.26)	8 (0.50)	23(1.70)	20(1.50)	4(0.30)	28(2.10)	10(0.70)	1 (0.10)	5 (0.40)	0	0	β-thal intermedia–33	1
2	Lucknow	Not mentioned	394#	231(58.60)	0	29 (7.30)	0	0	36(9.10)	0	0	12(3.00)	0	0	0	0	0	1
3	Lucknow	Not mentioned	17047	1185(6.90)	579(3.39)	187 (1.09)	136(0.79)	85(0.49)	204 (1.19)	68 (0.39)	51(0.29)	85(0.49)	51(0.29)	0	0	0	HbSD Punjab --34 HbSE disease --17, Others-511	6

*Cases of anemia #Includes extended family members of affected chlidren

References:

1. Tamhankar PM, Agarwal S, Arya V, Kumar R, Gupta UR, Agarwal SS. Prevention of homozygous beta thalassemia by premarital screening and prenatal diagnosis in India. Prenat Diagn. 2009; 29:83–88.
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Uttarakhand

Uttarakhand formerly known as Uttaranchal is a state which is divided into two divisions, Garhwal and Kumaon. It borders the Tibet Autonomous Region of China to the north; the Province of Nepal to the east; Uttar Pradesh to the south and Himachal Pradesh to the west and north-west. The native people of Uttarakhand are generally called Uttarakhandi and sometimes specifically either Garhwali or Kumaoni depending on their place of origin. The total population is 10 millions and among them 18.76% belong to scheduled castes and 2.89% to scheduled tribes (Census 2011).

Of the 4 reported studies, one was population based and 3 were hospital based studies. Fig 5 shows the 7 districts (shaded areas) where studies were undertaken in the state.



Fig 5: Map of Uttarakhand showing the studied areas

The seven districts covered are shown in Table 17. The names of the non-tribal groups studied have not been mentioned.

Table 17: Districts covered in Uttarakhand

Chamoli	Dehradun	Nainital	Pauri	Rudraprayag
Tehri	Uttarkashi			

Population based study among the non-tribals from Garhwal region district encountered only few βTT cases (Table 18). Hospital based studies reported β thalassaemia and all other haemoglobinopathies including many rare variants as well as unusual compound heterozygous combinations like HbH disease, homozygous HPFH, HbQ India trait, HbD Iran trait, HbJ Meerut trait, HbSD Punjab and HPFH/ δβ thal (Table 19).

Table 18: Population studies in Non- Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Chamoli, Pauri, Rudraparyag, Tehri, Uttarkashi (Garhwal region)	Not Mentioned	920	14 (1.50)	-	-	-	-	-	-	-	-	-	-	-	-	-	1

Table 19: Hospital based studies

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Nainital	Not Mentioned	38	0	0	0	0	1 (2.60)	0	0	0	0	0	0	0	0	0	2
2	Dehradun	Not Mentioned	8144	230(2.80)	12(0.15)	12 (0.15)	0	0	34 (0.42)	5(0.06)	5 (0.06)	45 (0.55)	0	4 (0.05)	0	4 (0.05)	HbH disease --2, HPFH homozygouse --3, HbQ India trait--2, HbD Iran trait--3, HbJ Merrut trait--1	3
3	Dehradun	Not Mentioned	933	30 (3.21)	3 (0.32)	6 (0.64)	0	0	0	0	0	0	0	0	0	0	HbSD Punjab --1, HPFH/ δβthal--1	4

References:

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3. Nair S, Acharya S, Acharya R, Kishor S, Thakur B. Spectrum of hemoglobinopathies: A hospital based study in Uttarakhand. J Clin Diagn Res. 2017;11:18-21.
4. Malik AK, Shukla ML, Seena AN. Prevalence of gene frequency in hemoglobinopathies & blood groups in Uttarakhand. Int J Physiol. 2008;6:103-108.

Himachal Pradesh

Himachal Pradesh is the northern most state of India and one of the eleven mountain states situated in Western Himalayas. It shares borders with Jammu and Kashmir and Ladakh to the north, and Punjab to the west, Haryana to the southwest, Uttarakhand to the southeast and a very narrow border with Uttar Pradesh to the South. The state also shares an international border to the east with the Tibet region in China. According to the Census 2011, the total population of the state is 6.8 million, the scheduled castes and scheduled tribes accounting for 25.19 % and 5.71 % respectively.

Only two population based studies (Table 20) were available and neither of them have mentioned the names of the districts where the studies were done nor the communities screened. Of the two studies, one reported the presence of βTT while the other study, apart from βTT, encountered few cases of HbD Punjab trait and the rare variant HbD Iran trait.

References:

1. Madan N, Sharma S, Sood SK, Colah RB,(Late) Bhatia HM. Frequency of β-thalassemia trait and other hemoglobinopathies in northern and western India. Indian J Hum Genet. 2010;16:16-25.
2. Bharti OK, Sood RK, Sharma HR, Kaur H, Minhis W, Chauhan R, Nigam N, Phull A. Mean corpuscular volume/mean corpuscular hemoglobin values are not reliable predictors of the β-thalasemmia carrier status among healthy diverse populations of Himachal Pradesh , India. Asian J Transf Sci. 2020;14:172-178.

Table 20: Population studies in Non- Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Not mentioned	Not mentioned	69	4 (5.80)	-	-	-	-	-	-	-	-	-	-	-	-	-	1
2	Not mentioned	General Caste, Other Backward Class, Scheduled Caste, Scheduled Tribe	*2220	47 (2.10)	0	0	0	0	0	0	0	8 (0.30)	0	0	0	0	HbD Iran trait--2	2

*College students

Jammu and Kashmir is a region administered by India as a union territory and consisting of the southern portion of the larger Kashmir region. The Line of Control separates Jammu and Kashmir from the Pakistani territories. It lies to the north of Himachal Pradesh and Punjab and to the west of Ladakh. As per the 2011 Census, the total population is 1.2 million. Of this, around 7.6% of the population is comprised of the scheduled castes and 10.9% belong to the scheduled tribes.

Among the 4 reported studies two were on population screening and two were hospital based studies. Fig 6 shows the district (shaded area) where the study was done.



Fig 6: Map of Jammu and Kashmir showing the studied areas

The district and the number of non tribal groups studied are shown in Tables 21 and 22. A total of 6 non- tribal groups were studied.

Table 21: Districts covered in Jammu & Kashmir

Jammu				

Table 22: Non tribal groups studied

Gujjar Bakarwal	Khan	Malik	Mir	Mughal
Syed				

Population screening showed the presence of β thalassemia and HbS gene in the non tribal populations and the prevalence of sickle cell trait varied from 5.40 to 18.40% (Table 23). Hospital based studies reported β thalassaemia and all other haemoglobinopathies like HbS, Hb E and HbD Punjab as well as α-Thal Trait, β-Thal Intermedia, HbSD disease and HbQ India (Table 24).

Table 23: Population studies in Non- Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Jammu	Not mentioned	31	2 (6.40)	0	0	0	0	0	0	0	0	0	-	-	-	-	1
2	Jammu	Mughal	103	-	-	19(18.40)	-	-	-	-	-	-	-	-	-	-	-	2
3	Jammu	Khan	128	-	-	7 (5.40)	-	-	-	-	-	-	-	-	-	-	-	2
4	Jammu	Malik	97	-	-	10(10.30)	-	-	-	-	-	-	-	-	-	-	-	2
5	Jammu	Mir	95	-	-	11 (11.50)	-	-	-	-	-	-	-	-	-	-	-	2
6	Jammu	Syed	117	-	-	9 (7.60)	-	-	-	-	-	-	-	-	-	-	-	2
7	Jammu	Gujjar Bakarwal	136	-	-	10 (7.35)	-	-	-	-	-	-	-	-	-	-	-	2

Table 24: Hospital based studies

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Jammu	Not mentioned	543	76(13.90)	7 (1.29)	1 (0.18)	5 (0.93)	0	2 (0.37)	0	0	0	0	0	0		α-thal trait–34, β-thal intermedia–1, HbSD Punjab--1, High HbF-25	3
2	Jammu	Not mentioned	2356*	72 (3.05)	4 (0.21)	3 (0.12)	0	0	6 (0.25)	0	0	14 (0.59)	0	2 (0.08)	0	2 (0.08)	HbQ India trait-2, α thal trait-20	4

*Includes antenatal cases, spouses of carrier women, family members of index cases & cases refered for HbA1c testing.

References:

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Island Region

Andaman and Nicobar Islands

Andaman and Nicobar Islands is a Union Territory comprising of two island groups, the Andaman and the Nicobar Islands. It consists of 572 islands, of which 38 are inhabited. According to the 2011 Census, the total population is nearly 0.38 million of which 7.5% belong to scheduled tribes. Hindus constitute the major religious group followed by Christians and Muslims.

A total of 6 population based studies among the tribal populations were available. Fig 1 shows the regions where studies were conducted in the islands.

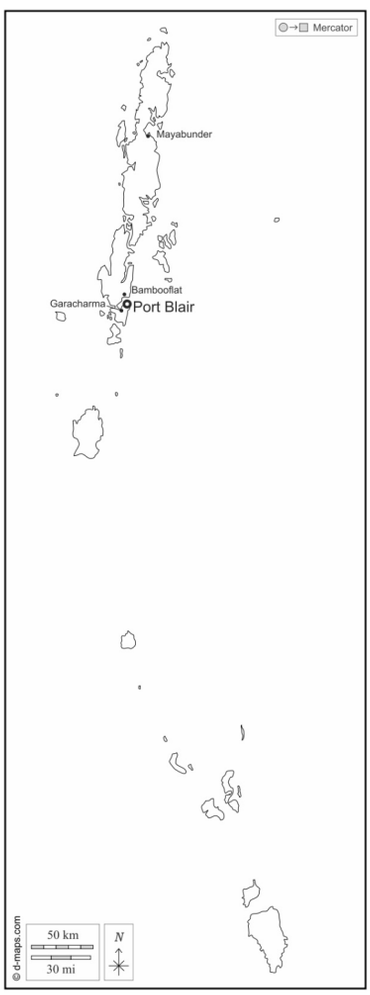

Fig 1: Map of Andaman and Nicobar Islands showing the studied areas

Table 1 shows the 6 tribal groups studied.

Table 1: Tribal populations studied

Bhantu	Great Andamanese	Jarawa	Nicobarese	Onge
Shompen				

Population based screening in the tribal populations indicated the presence of βTT and HbE trait (AE) and the prevalence varied from 3.44 to 7.80% and 10.52 to 17.20% respectively. A complete absence of thalassemia and hemoglobinopathies was reported among the Jarawas (Table 2).

Table 2: Population studies in Tribal groups

Sr. No.	Districts	Communities	Total Tested	βTT No (%)	βTM No (%)	AS No (%)	SS No (%)	S-β thal No (%)	AE No (%)	EE No (%)	E-β thal No (%)	AD No (%)	DD No (%)	D-β thal No (%)	δβ-thal Trait No (%)	HPFH Trait No (%)	Others No	Ref.
1	Andaman & Nicobar	Bhantu	122	-	-	0	0	-	-	-	-	-	-	-	-	-	-	1
2	Andaman & Nicobar	Shompen	55	-	-	0	0	-	-	-	-	-	-	-	-	-	-	2
3	Andaman & Nicobar	Great Andamanese	29	1 (3.44)	0	0	0	0	5(17.20)	0	0	0	0	0	0	0	0	3
4	Andaman & Nicobar	Jarawa	54	0	0	0	0	0	0	0	0	0	0	0	0	0	0	4
5	Andaman & Nicobar	Nicobarese	229	1 (7.80)	-	-	-	-	-	-	-	-	-	-	-	-	-	5
6	Andaman & Nicobar	Great Andamanese, Onge, Shompen	19	-	-	-	-	-	2(10.52)	0	-	-	-	-	-	-	-	6

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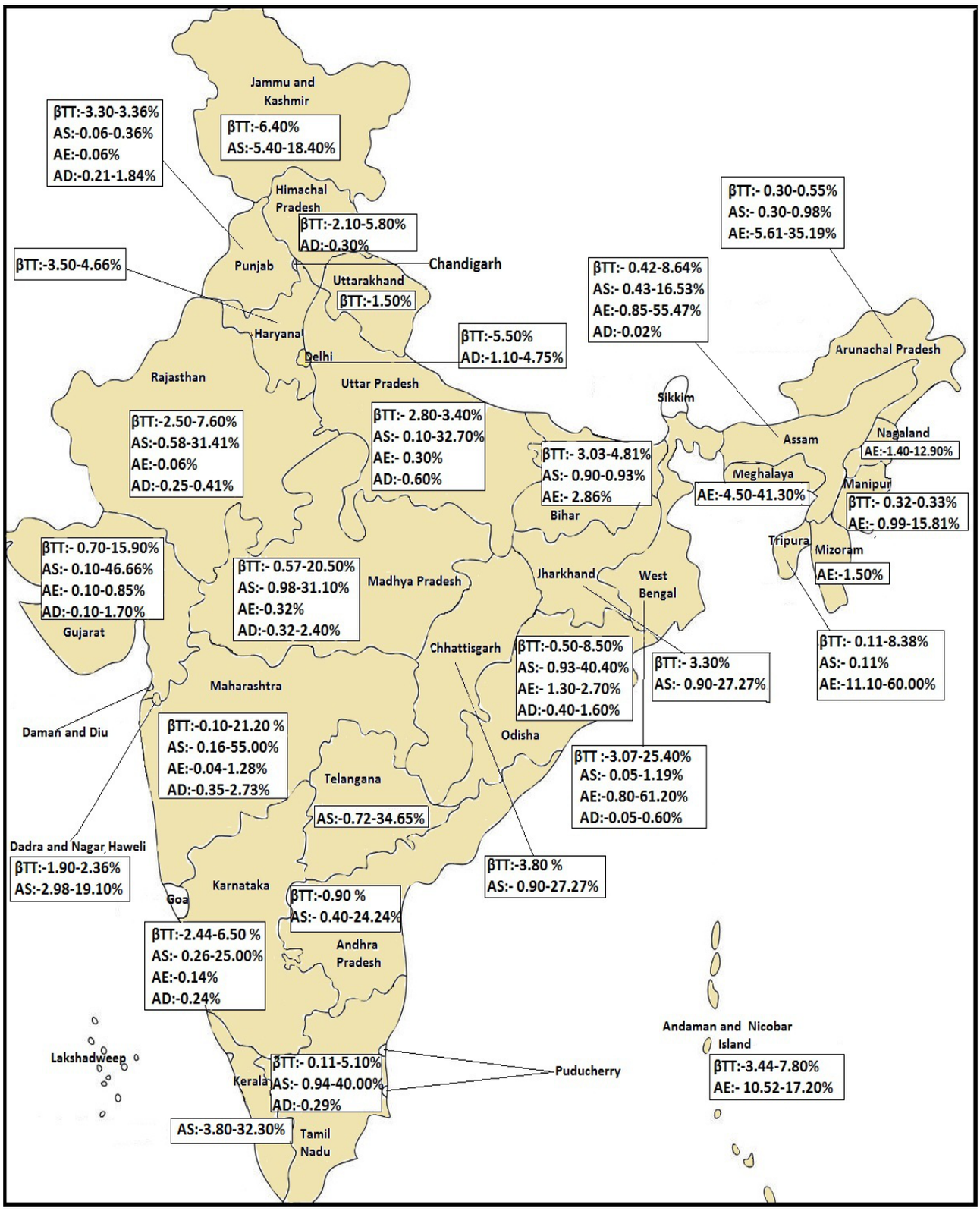

Map of India showing the overall prevalence of carriers of β-thalassaemia and the three common haemoglobin variants in India

The number of individuals screened in some of the studies in few of the states/union territories are very small. Hence some of the very high frequencies reported in these studies may not be reliable. Only the population based studies have been included in the map.

Prevalence of Mutations

β Thalassaemia mutations

β thalassaemia is seen all over the country and there are several studies reporting the prevalence of the common and rare β thalassaemia mutations from different regions. There were 20 states and 4 Union Territories from where these mutations have been reported (Fig 1).



Fig 1: States and Union Territories from where β thalassaemia mutations have been reported

Many of these studies have not mentioned the ethnic groups or communities, hence the distribution of β thalassaemia mutations is presented according to the states and union territories from where they are reported which is indicated by * or from where the individuals originated as in most of the studies. Majority of the mutations have been reported in β thalassaemia heterozygotes while in some studies β thalassaemia homozygotes have also been included as well as cases of Hb E-β thalassaemia and HbS- β thalassaemia. In a couple of studies either the mutations have only been reported in individuals with borderline HbA2 levels or individuals having only rare mutations are reported. In all such instances the percentages would not be indicative of the actual prevalence of different mutations. The maximum number of studies have been reported from Maharashtra and Gujarat followed by West Bengal, Uttar Pradesh and Punjab. In most of the regions IVS 1-5 (G→C) was the most common severe β+ thalassaemia mutation. The other common mutations, IVS I-1 (G→T), CD 8/9 (+G), CD 41/42 (-TCTT), 619 bp deletion, CD 15 (G→A) and CD 30 (G→C) were reported in variable frequencies in different studies. Many rare mutations have been reported particularly in studies where large numbers have been tested in different states (Tables 1 - 22). In a few studies the numbers tested are only a handful, yet these have also been included.

Table 1: β-thalassaemia mutations in Andhra Pradesh

Sr. No.	Total Tested	IVS-I-5 (G→C)	IVS-I-1 (G→T)	CD 8/9 (+G)	CD 41/42 (-TCTT)	619 bp deletion	CD 15 (G→A)	CD 30 (G→C)	CD 30 (G→A)	IVS I-1 (G→A)	CD 16 (-C)	Capsite +1 (A→C)	Others	Uncharacterized	Ref
1	8	4 (50.00)	0	0	0	0	4 (50.00)	0	0	0	0	0	0	0	1
2	77[a]	58 (75.32)	0	2 (2.59)	3 (3.89)	0	7 (9.09)	0	0	0	0	0	CD 5(-CT)→ 1 (1.29) CD 6 (A→T)→ 5 (6.49) CD 26 (G→A)→ 1 (1.29)	0	2
3	136	90 (66.17)	0	0	1 (0.73)	0	22 (16.17)	9 (6.61)	2 (1.47)	0	1 (0.73)	0	CD 5 (-CT)→ 2 (1.47) -80 (C→T)→1 (0.74) -28 (A→G)→ 2 (1.47) IVS I-110 (G→A)→2 (1.47) Poly A (T→C)→ 4 (2.94)	0	3
4	39	31 (79.48)	0	0	0	0	2 (5.12)	2 (5.12)	0	2 (5.12)	0	1 (2.63)	Poly A (T→C)→ 1 (2.63)	0	4
5	200[a]	159 (79.50)	2 (1.00)	2 (1.00)	2 (1.00)	15 (7.50)	1 (0.50)	0	0	0	0	0	CD 6 (A→T)→ 6 (3.00) CD 26 (G→A)→ 4 (2.00)	9 (4.50)	5

Table 2: β-thalassaemia mutations in Assam

Sr. No.	Total Tested	IVS-I-5 (G→C)	IVS-I-1 (G→T)	CD 8/9 (+G)	CD 41/42 (-TCTT)	619 bp deletion	CD 15 (G→A)	CD 30 (G→C)	CD 30 (G→A)	IVS I-1 (G→A)	CD 16 (-C)	Capsite +1 (A→C)	Others	Uncharacterized	Ref
1	6[b]	3 (50.00)	0	0	0	0	0	0	0	0	0	0	CD 26 (G→A) → 3 (50.00)	0	1

Table 3: β-thalassaemia mutations in Bihar

Sr. No.	Total Tested	IVS-I-5 (G→C)	IVS-I-1 (G→T)	CD 8/9 (+G)	CD 41/42 (-TCTT)	619 bp deletion	CD 15 (G→A)	CD 30 (G→C)	CD 30 (G→A)	IVS I-1 (G→A)	CD 16 (-C)	Capsite +1 (A→C)	Others	Uncharacterized	Ref
1	33	22 (66.70)	0	5 (15.15)	1 (3.03)	0	0	0	2 (6.06)	0	1 (3.03)	0	0	2 (6.06)	6
2	8	7 (87.50)	0	0	0	0	1 (12.50)	0	0	0	0	0	0	0	1
3	111	83 (74.77)	1 (0.90)	4 (3.60)	4 (3.60)	2 (1.80)	5 (4.50)	8 (7.21)	0	0	2 (1.80)	2 (1.80)	0	0	3
4	23	17 (73.91)	0	0	3 (13.04)	0	0	3 (13.04)	0	0	0	0	0	0	4
5	11	5 (45.45)	0	6 (54.54)	0	0	0	0	0	0	0	0	0	0	7

Table 4: β-thalassaemia mutations in Chhattisgarh

Sr. No.	Total Tested	IVS-I-5 (G→C)	IVS-I-1 (G→T)	CD 8/9 (+G)	CD 41/42 (-TCTT)	619 bp deletion	CD 15 (G→A)	CD 30 (G→C)	CD 30 (G→A)	IVS I-1 (G→A)	CD 16 (-C)	Capsite +1 (A→C)	Others	Uncharacterized	Ref
1	17	6 (35.29)	4 (23.5)	1 (5.88)	0	4 (23.52)	0	0	0	0	1 (5.88)	1(5.88)	0	0	4

Table 5: β-thalassaemia mutations in Delhi

Sr. No.	Total Tested	IVS-I-5 (G→C)	IVS-I-1 (G→T)	CD 8/9 (+G)	CD 41/42 (-TCTT)	619 bp deletion	CD 15 (G→A)	CD 30 (G→C)	CD 30 (G→A)	IVS I-1 (G→A)	CD 16 (-C)	Capsite +1 (A→C)	Others	Uncharacterized	Ref
1	46	11 (22.80)	9 (19.60)	6 (13.00)	4 (8.69)	16(34.78)	0	0	0	0	0	0	0	0	8
2	2	2 (100.00)	0	0	0	0	0	0	0	0	0	0	0	0	1
3	25c*	3 (12.00)	0	0	1 (4.00)	2 (8.00)	0	0	0	0	0	2 (8.00)	0	0	9

Table 6: β-thalassaemia mutations in Goa

Sr. No.	Total Tested	IVS-I-5 (G→C)	IVS-I-1 (G→T)	CD 8/9 (+G)	CD 41/42 (-TCTT)	619 bp deletion	CD 15 (G→A)	CD 30 (G→C)	CD 30 (G→A)	IVS I-1 (G→A)	CD 16 (-C)	Capsite +1 (A→C)	Others	Uncharacterized	Ref
1	13	2 (15.40)	0	0	0	0	0	3 (23.10)	1 (7.70)	0	0	0	IVS II-837 (T→G)→ 7(53.80)	0	4

Table 7: β-thalassaemia mutations in Gujarat

Sr. No.	Total Tested	IVS-I-5 (G→C)	IVS-I-1 (G→T)	CD 8/9 (+G)	CD 41/42 (-TCTT)	619 bp deletion	CD 15 (G→A)	CD 30 (G→C)	CD 30 (G→A)	IVS I-1 (G→A)	CD 16 (-C)	Capsite +1 (A→C)	Others	Uncharacterized	Ref
1	197	81 (41.11)	33 (16.75)	8 (4.06)	17 (8.62)	51(25.88)	3 (1.52)	4 (2.03)	0	0	0	0	0	0	10
2	396	198 (50.00)	0	17 (4.29)	31 (7.82)	74(18.68)	20 (5.05)	4 (1.01)	2 (0.50)	32 (8.08)	2 (0.50)	4 (1.01)	CD 5 (-CT)→5 (1.26) IVS I-3'end(-25 bp del)→2(0.50) IVS II-I (G→A)→1 (0.25) CD 44 (-C)→1 (0.25)	3 (0.75)	1
3	47	34 (72.34)	0	3 (6.38)	6 (12.76)	3 (6.38)	1 (2.12)	0	0	0	0	0	0	0	11
4	3	1 (33.30)	0	0	0	0	0	0	0	0	0	0	c.366_494 del→ 2 (66.7)#	0	2

(contd.)

Sr. No.	Total Tested	IVS-I-5 (G→C)	IVS-I-1 (G→T)	CD 8/9 (+G)	CD 41/42 (-TCTT)	619 bp deletion	CD 15 (G→A)	CD 30 (G→C)	CD 30 (G→A)	IVS I-1 (G→A)	CD 16 (-C)	Capsite +1 (A→C)	Others	Uncharacterized	Ref
5	248	104 (41.94)	13 (5.24)	15 (6.05)	16 (6.45)	46 (18.54)	14 (5.65)	4 (1.61)	2 (0.80)	6 (2.42)	5 (2.02)	1 (0.40)	CD 5 (-CT)→15 (6.05) IVS I-1 (G→C)→1 (0.40) IVS I-5 (G→T)→1 (0.40) IVS I-128 (T→G)→1 (0.40)	4 (1.61)	12
6	45	19 (42.22)	4 (8.89)	2 (4.44)	3 (6.67)	10 (22.22)	4 (8.89)	0	1 (2.22)	0	0	1 (2.22)	CD 44 (-C)→ 1 (2.22)	0	3
7	618	301 (48.70)	38 (6.14)	30 (4.85)	40 (6.47)	74 (11.97)	50 (8.09)	11 (1.77)	3 (0.48)	3 (0.48)	2 (0.32)	13 (2.10)	Poly A (T→C)→ 3 (0.48) CD 47/48 (+ATCT)→1(0.16) CD 5 (-CT)→ 43 (6.95) IVS II-654 (C→T)→ 1 (0.16) IVS I-130 (G→C)→ 1 (0.16)	4 (0.64)	4
8	175	42 (24.00)	2 (1.14)	1 (0.57)	16 (9.14)	7 (4.00)	26 (14.85)	3 (1.71)	66 (37.71)	0	1 (0.57)	2 (1.14)	CD 5 (-CT)→ 2 (1.14) CD 8 (-AA)→ 1 (0.57) CD 15 (-T)→2 (1.14) -88 (C→A)→ 1 (0.57)	3 (1.71)	13
9	477	265 (55.55)	23 (4.82)	27 (5.66)	32 (6.70)	44 (9.22)	16 (3.35)	16 (3.35)	0	5 (1.04)	12 (2.51)	0	CD 5 (-CT)→ 16 (3.35)	21 (4.40)	14
10	17	10 (58.82)	0	0	1 (5.88)	4 (23.52)	2 (11.76)	0	0	0	0	0	0	0	15
11	348	167 (47.98)	20 (5.74)	16 (4.59)	28 (8.04)	43 (12.35)	22 (6.32)	5 (1.43)	0	6 (1.72)	6 (1.72)	2 (0.60)	CD 5 (-CT)→ 15 (4.31) IVS I-130 (G→A)→2 (0.57)	16 (4.59)	16

Table 8: β-thalassaemia mutations in Haryana

Sr. No.	Total Tested	IVS-I-5 (G→C)	IVS-I-1 (G→T)	CD 8/9 (+G)	CD 41/42 (-TCTT)	619 bp deletion	CD 15 (G→A)	CD 30 (G→C)	CD 30 (G→A)	IVS I-1 (G→A)	CD 16 (-C)	Capsite +1 (A→C)	Others	Uncharacterized	Ref
1	24	11 (45.83)	5 (20.83)	0	1 (4.17)	6 (25.00)	0	0	0	0	0	0	-88 (C→T)→ 1 (4.17)	0	3

Table 9: β-thalassaemia mutations in Himachal Pradesh/Jammu & Kashmir

Sr. No.	Total Tested	IVS-I-5 (G→C)	IVS-I-1 (G→T)	CD 8/9 (+G)	CD 41/42 (-TCTT)	619 bp deletion	CD 15 (G→A)	CD 30 (G→C)	CD 30 (G→A)	IVS I-1 (G→A)	CD 16 (-C)	Capsite +1 (A→C)	Others	Uncharacterized	Ref
1	8	4 (50.00)	0	0	3 (37.50)	0	0	0	0	0	0	1 (12.50)	0	0	4

Table 10: β-thalassaemia mutations in Jharkhand

Sr. No.	Total Tested	IVS-I-5 (G→C)	IVS-I-1 (G→T)	CD 8/9 (+G)	CD 41/42 (-TCTT)	619 bp deletion	CD 15 (G→A)	CD 30 (G→C)	CD 30 (G→A)	IVS I-1 (G→A)	CD 16 (-C)	Capsite +1 (A→C)	Others	Uncharacterized	Ref
1	12	9 (75.00)	0	0	0	0	1 (8.33)	2 (16.66)	0	0	0	0	0	0	4

Table 11: β-thalassaemia mutations in Karnataka

Sr. No.	Total Tested	IVS-I-5 (G→C)	IVS-I-1 (G→T)	CD 8/9 (+G)	CD 41/42 (-TCTT)	619 bp deletion	CD 15 (G→A)	CD 30 (G→C)	CD 30 (G→A)	IVS I-1 (G→A)	CD 16 (-C)	Capsite +1 (A→C)	Others	Uncharacterized	Ref
1	17	6 (35.29)	0	0	1 (5.88)	0	3 (17.64)	0	0	0	0	0	IVS II-837→5 (29.41)	2 (11.76)	1
2	15	15 (100.00)	0	0	0	0	0	0	0	0	0	0	0	0	11
3	28	20 (71.42)	0	2 (7.14)	0	0	5 (17.85)	0	0	0	0	0	IVS II-14 (T→G)→1 (3.57)	0	2
4	118	54 (45.76)	4 (3.39)	1 (0.85)	2 (1.69)	0	16 (13.56)	1 (0.85)	0	3 (2.54)	1 (0.85)	0	IVS II-837→9 (7.63) IVS II-I (G→A) →1 (0.85) Poly A (T→C)→ 17 (14.41) -25 (A→G)→ 1 (0.85) -28 (A→G)→ 3 (2.54) CD 26 (G→T)→ 1 (0.85) IVS I-130 (G→A) → 1 (0.85) CD 36/37 (-T) → 1 (0.85)	2 (1.69)	3
5	140	96 (68.57)	0	2 (1.42)	1 (0.71)	1 (0.71)	14 (10.00)	3 (2.14)	0	0	0	0	Poly A (T→C)→ 3 (2.14) CD 8 (-AA)→2 (1.42) IVS II-837→5 (3.57) IVS II-I (G→A) →3 (2.14) CD 15 (-T)→2 (1.42) CD 121 (G→T)→3 (2.14) -88(C→T)→1 (0.71) CD 39 (C→T)→1 (0.71) CD 8 (-AA)→1 (0.71) IVS II-613 (C→T)→1 (0.71)	1 (1.14)	4

Table 12: β-thalassaemia mutations in Kerala

Sr. No.	Total Tested	IVS-I-5 (G→C)	IVS-I-1 (G→T)	CD 8/9 (+G)	CD 41/42 (-TCTT)	619 bp deletion	CD 15 (G→A)	CD 30 (G→C)	CD 30 (G→A)	IVS I-1 (G→A)	CD 16 (-C)	Capsite +1 (A→C)	Others	Uncharacterized	Ref
1	67	42 (62.68)	0	0	3 (4.48)	0	0	0	0	0	5 (7.46)	2 (2.91)	CD 5 (-CT)→ 2 (2.91) IVS II-837→ 4 (5.97) -90 (C→T)→1 (1.49) CD22/23/24 (7 bp del)→1(1.49) CD 106/107(+G)→1 (1.49) IVS I-110 (G→A)→ 1 (1.49) Poly A (T→C)→ 4 (5.97) IVS I-3'end(-25 bp del)→1 (1.47)	0	3

Table 13: β-thalassaemia mutations in Madhya Pradesh

Sr. No.	Total Tested	IVS-I-5 (G→C)	IVS-I-1 (G→T)	CD 8/9 (+G)	CD 41/42 (-TCTT)	619 bp deletion	CD 15 (G→A)	CD 30 (G→C)	CD 30 (G→A)	IVS I-1 (G→A)	CD 16 (-C)	Capsite +1 (A→C)	Others	Uncharacterized	Ref
1	38	36 (94.73)	0	0	1 (2.63)	0	1 (2.63)	0	0	0	0	0	0	0	1
2	65	35 (53.90)	6 (20.83)	2 (3.08)	4 (6.15)	11 (16.9)	1 (1.54)	1 (1.54)	0	1 (1.54)	0	1 (1.54)	-88(C→T)→1 (1.54) CD 15 (-T)→1 (1.54) CD 126-131 (-17 bp deletion)→1 (1.54)	0	3
3	145	56 (38.62)	15 (10.34)	16 (11.03)	8 (5.51)	28 (19.31)	7 (4.82)	6 (4.13)	1 (0.68)	1 (0.68)	3 (2.06)	2 (1.37)	CD 126-131 (-17 bp deletion)→ 1 (0.68)	1 (0.68)	4
4	83	30 (36.10)	14 (16.80)	4 (4.80)	21 (25.30)	10 (12.04)	0	0	0	0	0	0	0	4 (5.00)	17
5	676[d,e*]	252 (37.28)	23 (3.41)	53 (7.84)	69 (10.20)	36 (5.33)	0	26 (3.85)	0	3 (0.45)	36 (5.32)	0	c.-50(A→T)→ 17 (2.51)# -88(C→T)→4 (0.60) c.48 (C→T)→ 21 (3.10)# CD 26 (G→A)→ 61 (9.02) CD 6 (A→T)→ 75 (11.09)	0	18
6	60[f]	48 (80.00)	0	4 (6.70)	4 (6.70)	0	4 (6.70)	0	0	0	0	0	0	0	19

Table 14: β-thalassaemia mutations in Maharashtra

Sr. No.	Total Tested	IVS-I-5 (G→C)	IVS-I-1 (G→T)	CD 8/9 (+G)	CD 41/42 (-TCTT)	619 bp deletion	CD 15 (G→A)	CD 30 (G→C)	CD 30 (G→A)	IVS I-1 (G→A)	CD 16 (-C)	Capsite +1 (A→C)	Others	Uncharacterized	Ref
1	13	7 (53.84)	0	0	1 (7.69)	0	0	0	0	0	0	0	IVS II-I (G→A)→ 1 (7.69)	4 (30.76)	10
2	45	27 (60.00)	1 (2.22)	0	1 (2.22)	3 (6.66)	8 (17.77)	0	0	2 (4.44)	0	0	CD 5 (-CT)→1 (2.22) CD 30→1 (2.22)[$]	1 (2.22)	20
3	356*	174 (48.87)	23 (6.46)	29 (8.14)	23 (6.46)	54 (15.16)	12 (3.37)	7 (1.96)	9 (2.52)	2 (0.56)	0	9 (2.52)	CD 5 (-CT)→1 (0.28) -88 (C→T)→ 1 (0.28) CD 121 (G→T)→ 2 (0.56) CD 10 (C→A)→ 1 (0.28) -28 (A→G)→ 1 (0.28)	8 (2.24)	21
4	233[b]	177 (75.96)	1 (0.42)	1 (0.42)	1 (0.42)	3 (1.28)	25 (10.72)	3 (1.28)	1 (0.42)	1 (0.42)	1 (0.42)	0	CD 5 (-CT)→1 (0.42) IVS II-837 (T→G)→ 3 (1.28) CD 26 (G →A)→1 (0.42)	14 (6.00)	1
5	89	55 (61.8)	4 (4.49)	3 (3.37)	4 (4.49)	12 (13.48)	2 (2.25)	3 (3.37)	0	1 (1.12)	2 (2.25)	1 (1.12)	Poly A (T→C)→ 2 (2.25)	0	3
6	805	525 (65.61)	13 (1.61)	5 (0.62)	17 (2.11)	13 (1.61)	153 (19.00)	31 (3.85)	6 (0.74)	7 (0.86)	2 (0.24)	11 (1.36)	IVS I-3'end(-25 bp del)→2(0.24) CD 55 (-A)→ 2 (0.24) Poly A (T→C)→ 1 (0.12) CD 5 (-CT)→ 1 (0.12) IVS II-837→2 (0.24) CD 111 (-G)→2 (0.24) CD 110 (T→C)→4 (0.49) IVS 1-130 (G→A)→ 2 (0.24) CD 26 (C→T)→1 (0.12) CD 16 (C→T)→1 (0.12) CD 15 (-T) →1 (0.12)	3 (0.37)	4
7	126	82 (65.07)	12 (9.52)	8 (6.34)	5 (3.96)	3 (2.38)	8 (6.34)	0	0	0	0	0	0	8 (6.34)	22
8	93	31 (33.33)	10 (10.78)	13 (13.97)	14 (15.05)	4 (4.30)	7 (7.52)	0	0	0	0	0	0	14 (15.05)	23
9	50	4 (8.00)	3 (6.00)	3 (6.00)	5 (10.00)	0	4 (8.00)	0	0	0	0	0	CD 14/15 (+G)→ 1 (2.00) CD 27/28 (+C)→ 1 (2.00) CD 71/72 (+A)→ 11 (22.00) CD 17 (A→T)→4 (8.00) CD 19 (A→G)→ 7 (14.00) CD 26 (G→T)→ 1 (2.00) CD 41(-C)→ 5 (10.00) CD 43 (G→T)→1 (2.00)	0	24

(contd.)

Sr. No.	Total Tested	IVS-I-5 (G→C)	IVS-I-1 (G→T)	CD 8/9 (+G)	CD 41/42 (-TCTT)	619 bp deletion	CD 15 (G→A)	CD 30 (G→C)	CD 30 (G→A)	IVS I-1 (G→A)	CD 16 (-C)	Capsite +1 (A→C)	Others	Uncharacterized	Ref
10	21[g]	15 (71.42)	0	1 (4.76)	2 (9.52)	0	0	0	0	0	1 (4.76)	0	CD 15 (-T)→ 2 (9.52)	0	7
11	7[g]	2 (28.57)	0	0	1 (5.88)	2 (28.57)	1 (5.88)	0	0	0	0	0	CD 26 (G→A)→ 1 (5.88)	0	15
12	98[g]	65 (60.32)	0	2 (2.04)	4 (4.08)	4 (4.08)	7 (7.14)	6 (6.12)	0	0	0	0	IVS II-837 (T→G)→ 2 (2.04) CD 15 (-T)→ 1 (1.02) IVS I-130 (G→C)→ 1 (1.02) IVS II-848 (C→A)→ 2 (2.04) -90(C→T)→ 1 (1.02) -88 (C→T)→ 1 (1.02) IVS I-129 (A→C)→ 1 (1.02) IVS II-I (G→A)→ 1 (1.02)	0	25
13	10	2 (20.00)	0	0	0	0	0	0	0	0	0	0	Initiation CD (T→C)→8 (80.00)	0	26
14	18[h]*	2 (11.11)	0	4 (22.22)	0	9 (50.00)	0	0	0	0	0	0	CD 6 (A→T) → 3 (16.66)	0	27
15	5615*	3110 (55.38)	277 (4.93)	279 (4.96)	248 (4.41)	409(7.28)	535 (9.52)	188(3.34)	43 (0.76)	35 (0.62)	76(1.35)	104 (1.85)	IVS I-3'end(-25bp del)→8(0.14) CD 55 (-A)→ 1 (0.01) Poly A (T→C)→ 54 (0.96) CD 5 (-CT)→ 74 (1.31) IVS II-837→32 (0.56) CD 15 (-T)→ 17 (0.30) IVS I-110 (T→C)→ 27 (0.48) IVS I-130 (G→A)→ 2 (0.33) IVS I-130 (G→C)→ 8 (0.14) IVS II-848 (C→A)→ 1 (0.01) −86 (C→G)→3 (0.05) -90(C→T)→ 5 (0.08) -88 (C→T)→ 13 (0.23) Initiation CD (T→C)→2 (0.03) CD 121 (G→T)→ 15 (0.26) CD 16 (C→T)→ 1 (0.01) IVS II-I (G→A)→ 7 (0.12) IVS I-110 (G→A)→ 5 (0.08) Poly A (-AA)→ 5 (0.08) 5'UTR + 20 (-C)→ 4 (0.07) CD 39 (C→T)→ 3 (0.05) CD 37 (-T)→ 3 (0.05) CD 47/48 (+ATCT)→ 2 (0.03) CD 90 (G→A)→ 2 (0.03)	0	28

(contd.)

Sr. No.	Total Tested	IVS-I-5 (G→C)	IVS-I-1 (G→T)	CD 8/9 (+G)	CD 41/42 (-TCTT)	619 bp deletion	CD 15 (G→A)	CD 30 (G→C)	CD 30 (G→A)	IVS I-1 (G→A)	CD 16 (-C)	Capsite +1 (A→C)	Others	Uncharacterized	Ref
													-28 (A→G)→ 2 (0.03) CD 22 (A→C)→ 2 (0.03) -87 (C→T)→ 1 (0.01) CD 7/8 (+G)→ 1 (0.01) CD 8 (-AA)→ 1 (0.01) CD 22-24 (7 bp del)→ 1 (0.01) CD 26 (G→T)→ 1 (0.01) IVS II-654 (C→T)→ 1 (0.01) IVS II-745 (C→G)→ 1 (0.01) IVS 1-5 (G→T)→ 1 (0.01) CD 39 (-A)→1 (0.01) CD 13 (C→A)→1 (0.01) CD 126/131 (7 bp del) →1 (0.01) CD 22 (G→ C)→ 2 (0.03)		
16	458[d]*	275 (60.11)	28 (6.11)	18 (3.93)	17 (3.71)	33 (7.20)	37 (8.07)	12 (2.62)	3 (0.65)	4 (0.87)	2 (0.43)	2 (0.43)	CD 55 (-A)→ 1 (0.21) Poly A (T→C)→ 10 (2.18) CD 5 (-CT)→ 4 (0.87) IVS I-1 (G→A)→4 (0.87) IVS II-837→1 (0.21) CD 15 (-T)→1 (0.21) IVS 1-110 (T→C)→ 1 (0.21) IVS II-848 (C→A)→ 2 (0.43) −86 (C→G)→1 (0.21) -90(C→T)→ 1 (0.21) CD 26 (G→T)→ 1 (0.21)	0	29

Table 15: β-thalassaemia mutations in Odisha

Sr. No.	Total Tested	IVS-I-5 (G→C)	IVS-I-1 (G→T)	CD 8/9 (+G)	CD 41/42 (-TCTT)	619 bp deletion	CD 15 (G→A)	CD 30 (G→C)	CD 30 (G→A)	IVS I-1 (G→A)	CD 16 (-C)	Capsite +1 (A→C)	Others	Uncharacterized	Ref
1	47	39 (83.00)	0	0	2 (4.26)	0	3 (6.38)	1 (2.13)	0	0	0	0	-87 (G→C)→ 1 (2.13) Poly A (T→C)→ 1 (2.13)	0	3
2	78	69 (88.6)	1(1.28)	0	2 (2.56)	0	0	1(1.28)	1(1.28)	0	2 (2.56)	0	0	2 (2.56)	4

(contd.)

Sr. No.	Total Tested	IVS-I-5 (G→C)	IVS-I-1 (G→T)	CD 8/9 (+G)	CD 41/42 (-TCTT)	619 bp deletion	CD 15 (G→A)	CD 30 (G→C)	CD 30 (G→A)	IVS I-1 (G→A)	CD 16 (-C)	Capsite +1 (A→C)	Others	Uncharacterized	Ref
3	431	306 (71.00)	9 (2.08)	13 (3.01)	52 (12.06)	0	30 (6.96)	21 (6.86)	0	0	0	0	0	0	30
4	25	10 (40.00)	1 (4.00)	1 (4.00)	0	0	0	0	0	0	0	1 (4.00)	IVS I-5 (G→A)→ 7 (28.00) CD 19 (A→G)→5 (20.00)	0	31
5	3[g]	2 (66.66)	0 (0.0)	0	0	0	0	1 (33.33)	0	0	0	0	0	0	7

Table 16: β-thalassaemia mutations in Punjab

Sr. No.	Total Tested	IVS-I-5 (G→C)	IVS-I-1 (G→T)	CD 8/9 (+G)	CD 41/42 (-TCTT)	619 bp deletion	CD 15 (G→A)	CD 30 (G→C)	CD 30 (G→A)	IVS I-1 (G→A)	CD 16 (-C)	Capsite +1 (A→C)	Others	Uncharacterized	Ref
1	142	54 (38.02)	15 (10.56)	19 (13.38)	20 (14.08)	22 (15.49)	1 (0.70)	1 (0.70)	0	0	4 (2.81)	2 (1.40)	-88 (C→T)→ 1 (0.70) IVS II-I (G→A)→ 1 (0.70)	2 (1.40)	10
2	153[d,g]	36 (23.52)	35 (22.87)	19 (12.41)	19 (12.41)	27 (17.64)	0	0	0	0	1 (0.65)	9 (5.88)	-88 (C→T)→ 2 (1.30) CD5 (-CT)→ 2 (1.30) CD 47/48 (-ATCT)→ 1 (0.65) IVS 1-3 (T→G) → 1 (0.65) CD 26 (G→A) → 1 (0.65)	0	20
3	139	72 (51.79)	12 (8.63)	10 (7.19)	0	20 (14.38)	0	0	14(10.07)	0	3 (2.15)	4 (2.87)	-88 (C→T)→ 4 (2.87)	0	6
4	71	11 (15.49)	0	6 (8.45)	4 (5.63)	21 (29.57)	0	0	0	20 (28.16)	1 (1.40)	2 (2.81)	-88 (C→T)→ 1 (1.40) CD5 (-CT)→ 1 (1.40) CD 47/48 (-ATCT)→ 2 (2.81)	2 (2.81)	1
5	41	21 (51.21)	0	10 (24.39)	1 (2.43)	0	1 (2.43)	0	4 (9.75)	0	2 (4.87)	0	0	2 (4.87)	32
6	11	3 (27.27)	1 (9.09)	0	0	6 (54.54)	0	0	0	0	1 (9.09)	0	0	0	11
7	352[d]	112 (31.8)	0	52 (14.8)	54 (15.3)	66 (18.7)	0	0	0	45 (12.8)	0	9 (2.50)	-88 (C→T)→ 3 (0.85)	11 (3.10)	33
8	26	9 (34.62)	6 (23.08)	3 (11.54)	3 (11.54)	0	0	0	0	1 (3.85)	0	3 (11.54)	-88 (C→T)→ 1 (3.85)	0	3
9	52	14 (26.92)	5 (9.61)	9 (17.30)	7(13.46)	4 (7.69)	0	1 (1.92)	3 (5.76)	1 (1.92)	1 (1.92)	3 (5.76)	-88 (C→T)→ 1 (1.9) CD 5 (-CT)→ 1 (1.9) -87(C→T)→1 (1.9) IVS I-110 (T→C)→ 1 (1.9)	0	4

Table 17: β-thalassaemia mutations in Rajasthan

Sr. No.	Total Tested	IVS-I-5 (G→C)	IVS-I-1 (G→T)	CD 8/9 (+G)	CD 41/42 (-TCTT)	619 bp deletion	CD 15 (G→A)	CD 30 (G→C)	CD 30 (G→A)	IVS I-1 (G→A)	CD 16 (-C)	Capsite +1 (A→C)	Others	Uncharacterized	Ref
1	50	33 (66.00)	1 (2.00)	5 (10.00)	6 (12.00)	2 (4.00)	0	1 (2.00)	0	0	2 (4.00)	0	0	0	6
2	51	33 (64.70)	4 (7.84)	0	5 (9.80)	2 (3.92)	0	1 (1.96)	0	1 (1.96)	0	3 (5.88)	-88 (C→T)→ 1 (1.96)	1 (1.96)	1
3	34	6 (17.64)	11 (32.35)	0	1 (2.94)	14 (41.17)	0	2 (5.88)	0	0	0	0	0	0	3
4	90	65 (72.22)	1 (1.11)	3 (3.33)	9 (9.99)	2 (2.22)	3(3.33)	1(1.11)	0	0	2(2.22)	3(3.33)	0	1 (1.11)	4
5	40[g,i]	26 (65.00)	0	2 (5.00)	2 (5.00)	3 (7.5)	2 (5.00)	0	0	0	2 (5.00)	0	CD 26 (G→A) → 2 (5.00) CD 121 (G→C) → 1 (2.50)	0	15

Table 18: β-thalassaemia mutations in Tamil Nadu

Sr. No.	Total Tested	IVS-I-5 (G→C)	IVS-I-1 (G→T)	CD 8/9 (+G)	CD 41/42 (-TCTT)	619 bp deletion	CD 15 (G→A)	CD 30 (G→C)	CD 30 (G→A)	IVS I-1 (G→A)	CD 16 (-C)	Capsite +1 (A→C)	Others	Uncharacterized	Ref
1	59	48 (81.35)	0	2 (3.38)	6 (10.16)	1 (1.69)	0	0	0	0	0	0	0	2 (3.38)	10
2	1	1 (100.00)	0	0	0	0	0	0	0	0	0	0	0	0	1
3	212	121 (57.14)	1 (0.47)	0	5 (2.36)	0	28 (13.21)	0	4 (1.89)	1 (0.47)	0	0	IVS II-837 (T→G)→ 2 (0.94) CD 5 (-CT)→ 2 (0.94) -28 (A→G)→ 19 (8.96) -29 (A→G)→ 1 (0.47) Poly A (T→C)→ 23 (10.85) IVS 1-130 (G→C) → 2 (0.94) CD 44(-C)→2 (0.94) IVS II-I (G→A)→ 1 (0.47)	0	3
4	16	9 (56.25)	0	0	1 (6.31)	0	0	0	0	0	0	0	IVS I-3'end (-25bp del) →2(12.50) IVS I-30 (G→C)→ 2 (12.50) IVS II-613 (C→T) → 1 (6.25) IVS II-745 (C→G)→ 1 (6.25)	0	4

Table 19: β-thalassaemia mutations in Telangana

Sr. No.	Total Tested	IVS-I-5 (G→C)	IVS-I-1 (G→T)	CD 8/9 (+G)	CD 41/42 (-TCTT)	619 bp deletion	CD 15 (G→A)	CD 30 (G→C)	CD 30 (G→A)	IVS I-1 (G→A)	CD 16 (-C)	Capsite +1 (A→C)	Others	Uncharacterized	Ref
1	406	297 (73.15)	0	14 (3.44)	31 (7.63)	1 (0.24)	53 (13.05)	0	0	0	10 (2.46)	0	0	0	34

Table 20: β-thalassaemia mutations in Uttar Pradesh

Sr. No.	Total Tested	IVS-I-5 (G→C)	IVS-I-1 (G→T)	CD 8/9 (+G)	CD 41/42 (-TCTT)	619 bp deletion	CD 15 (G→A)	CD 30 (G→C)	CD 30 (G→A)	IVS I-1 (G→A)	CD 16 (-C)	Capsite +1 (A→C)	Others	Uncharacterized	Ref
1	61	30 (49.18)	0	7 (11.47)	8 (13.11)	2 (3.27)	3 (4.91)	1 (1.63)	0	0	3 (4.91)	2 (3.27)	-88 (C→T)→ 3 (4.91)	2 (3.27)	6
2	238	155 (65.12)	6 (2.52)	10 (4.20)	30 (12.60)	6 (2.52)	9 (3.78)	4 (1.68)	0	0	8 (3.36)	3 (1.26)	0	7 (2.94)	32
3	44[a]	16 (36.36)	1 (2.27)	3 (6.81)	5 (11.36)	2 (4.54)	1 (2.27)	8 (18.18)	0	0	3 (6.81)	0	CD 26 (G→A)→ 1 (2.27) CD 6 (A→T)→ 3 (6.81)	1 (2.3)	1
4	98[b]	54 (55.10)	3 (3.06)	5 (5.10)	6 (6.12)	12 (12.24)	2 (2.04)	2 (2.04)	0	0	2 (2.04)	1 (1.02)	-88 (C→T)→ 1 (1.02) CD5 (-CT)→ 2 (2.04) CD 26 (G→A)→ 3 (3.06)	5 (5.20)	35
5	218	161 (73.85)	1 (0.45)	9 (4.12)	18 (8.25)	3 (1.37)	0	5 (2.29)	0	0	6 (2.75)	8 (3.66)	CD 5 (-CT)→ 2 (0.91) CD 47/48 (-ATCT)→ 1 (0.45) 8 bp del (CD 36/37/38/39) →2 (0.91)	2 (0.91)	11
6	125	81 (64.8)	1 (0.80)	7 (5.60)	12 (9.60)	2 (1.60)	6 (4.80)	3 (2.40)	0	1 (0.80)	2(1.60)	2(1.60)	-88 (C→T)→ 2 (1.60) Poly A(T→C)→ 2 (1.60) IVS I-130 (G→C)→ 1 (0.80) CD 22-24 (-7 bp del)→ 1 (0.80)	2 (1.60)	4
7	300[e,i]	140 (46.66)	14 (4.66)	25 (8.33)	28 (9.33)	18 (6.00)	17 (5.66)	14 (4.66)	0	0	25(8.33)	5 (1.66)	0	14 (4.60)	36
8	56	29 (51.78)	0	2 (3.57)	3 (5.35)	0	11 (19.64)	0	0	0	2 (3.57)	3 (5.35)	-88 (C→T)→ 4 (7.14) CD 5 (-CT)→ 2 (3.57)	0	37
9	13[e]	7 (53.84)	0	0	3 (23.07)	0	0	1 (7.69)	0	0	2 (15.38)	0	0	0	7

Table 21: β-thalassaemia mutations in West Bengal

Sr. No.	Total Tested	IVS-I-5 (G→C)	IVS-I-1 (G→T)	CD 8/9 (+G)	CD 41/42 (-TCTT)	619 bp deletion	CD 15 (G→A)	CD 30 (G→C)	CD 30 (G→A)	IVS I-1 (G→A)	CD 16 (-C)	Capsite +1 (A→C)	Others	Uncharacterized	Ref
1	125	95 (76.00)	0	4 (3.20)	12 (9.60)	1 (0.80)	0	1 (0.80)	0	0	0	0	0	12 (9.60)	6
2	324[d,g]	149 (45.98)	0	3 (0.92)	0	3 (0.92)	3 (0.92)	17 (5.24)	0	0	0	0	CD 26 (G→A)→ 107 (33.00)	42 (12.96)	38
3	25[b]	11 (44.00)	0	1 (4.00)	1 (4.00)	0	1 (4.00)	1 (4.00)	0	0	0	0	CD 26 (G→A)→ 5 (20.00)	5 (20.00)	1
4	13	10 (76.90)	2 (15.40)	0	0	0	1 (7.70)	0	0	0	0	0	0	0	11
5	650[b,d,j*]	317 (48.76)	0	7 (1.07)	9 (1.38)	5 (0.76)	15 (2.30)	30 (4.61)	0	0	0	6 (0.92)	IVS-I-130 (G→A)→ 3 (0.46) -88 (C→T)→6 (0.92) Indian inv/del Gγ (Aγ δβ)^{o}→1 (0.15) CD 26 (G→A)→ 207(31.84)	44 (6.76)	39

(contd.)

Sr. No.	Total Tested	IVS-I-5 (G→C)	IVS-I-1 (G→T)	CD 8/9 (+G)	CD 41/42 (-TCTT)	619 bp deletion	CD 15 (G→A)	CD 30 (G→C)	CD 30 (G→A)	IVS I-1 (G→A)	CD 16 (-C)	Capsite +1 (A→C)	Others	Uncharacterized	Ref
6	11[a]	7 (63.63)	0	0	0	0	1 (9.09)	0	0	0	0	0	CD26 (G→A)→ 2 (18.18) CD 6 (A→T)→ 1 (9.09)	0	2
7	529	384 (72.58)	0	7 (1.32)	19 (3.59)	1 (0.19)	40 (7.56)	41 (7.75)	0	0	6 (1.13)	0	-90 (C→T)→11 (2.08) CD 15 (-C)→ 5 (0.95) IVS I-129 (A→C)→ 1 (0.19) CD 41 (-C)→ 2 (0.38) CD 62/64(-7 bp del)→2(0.38) CD 81/87(-22 bp del)→1(0.19) CD 110 (T→C)→ 4 (0.76) Poly A (T→C)→ 4 (0.76)	1 (0.19)	3
8	101	78 (77.20)	1(1.00)	4 (3.96)	4 (3.96)	0	3(2.90)	4 (3.96)	0	0	2 (1.98)	2 (1.98)	CD 15 (-T)→ 2 (1.98) -90 (C→T)→ 1 (0.96)	0	4
9	660[e,k]	308 (46.66)	0	7 (1.07)	23 (3.58)	0	33 (5.01)	50 (7.53)	0	0	0	0	HPFH 3→12 (1.80) CD 26 (G→A)→ 208 (31.54)	19 (2.87)	40
10	30[g]	21 (70.00)	1 (3.33)	1 (3.33)	1 (3.33)	0	0	4 (13.33)	0	0	1 (3.33)	0	CD 15 (-T)→ 1 (3.33)	0	7
11	6[e]	3 (50.00)	0	1 (16.66)	0	0	0	0	0	0	0	0	CD 26 (G→A)→ 2 (33.33)	0	15

Table 22: β-thalassaemia mutations in other States/Union territories

Sr. No.	State	Total Tested	IVS-I-5 (G→C)	IVS-I-1 (G→T)	CD 8/9 (+G)	CD 41/42 (-TCTT)	619 bp deletion	CD 15 (G→A)	CD 30 (G→C)	CD 30 (G→A)	IVS I-1 (G→A)	CD 16 (-C)	Capsite +1 (A→C)	Others	Uncharacterized	Ref
1	UK##	102[d]	23 (22.54)	14 (13.72)	20 (19.60)	12 (11.76)	21 (20.58)	5 (4.90)	0	0	0	1 (0.98)	2 (1.96)	-88 (C→T)→2 (1.96)	2 (1.96)	41
2	Sindh Migrants (Maharashtra)	325*	18 (5.53)	0	49 (15.07)	7 (2.15)	160 (49.23)	0	0	0	83(25.53)	5 (1.53)	0	0	3 (0.92)	1
3	Daman	1*	0	0	0	0	0	1 (100.00)	0	0	0	0	0	0	0	1
4	Lakshadeep	2*	2 (100.00)	0	0	0	0	0	0	0	0	0	0	0	0	1
5	Sindh Migrants (Pakistan)	24*	3 (12.50)	10 (41.66)	1 (4.16)	1 (4.16)	9 (37.50)	0	0	0	0	0	0	0	0	11
6	Immigrants (Pakistan)	291*	44(15.12)	73 (25.08)	45 (15.46)	13 (4.46)	108 (37.11)	1 (0.34)	1(0.34)	5(1.71)	0	0	1 (0.34)	0	0	4

The figure shown for each mutation indicate No(%) in Tables 1 to 22

* Reported from respective states/UTs, # HGVS nomenclature, ## Asian Indian origin residing in UK, $ Amino acid change not mentioned, a: HbS trait and HbE trait, b: HbE trait, c: Borderline A2 samples screened- Mutation identified only in 8 cases, d: No of chromosones, e: HbS-β thalassaemia and HbE- β thalassaemia cases, f: HbS-β thalassaemia cases, g: HbE-β thalassaemia cases, h: HbS trait, i: HbD- β thalassaemia, j: Includes 1 δβ-thalassaemia mutation, k: 12 cases of HPFH 3 mutation.

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α Thalassaemia mutations

Alpha thalassaemia is particularly prevalent among tribal populations in India but is also seen in variable frequencies among non-tribal populations from different regions. There were 29 studies reporting the prevalence of α thalassaemia where α genotyping has been done. In some of these studies, α genotyping was done in cases of β thalassaemia, sickle cell disorders or Hb E-β thalassaemia. Studies based only on phenotypic analysis (presence of inclusion bodies/a fast moving band on alkaline Hb electrophoresis) have not been included.

The states and union territories (shaded areas) from where these studies have been reported are shown in Fig 1.



Fig 1 : Map of India showing the states and union territories where individuals with α thalassaimia originated or were reported

In many of the studies the names of the tribal and non-tribal groups screened have not been mentioned. Majority of these studies have only looked for the prevalence of deletional α thalassaemia and a single α gene deletion (- $\alpha^{3.7}$) was the commonest α gene defect. Some of the studies have not distinguished between the - $\alpha^{3.7}$ rightward deletion and the - $\alpha^{4.2}$ leftward deletion (Table 1). The highest prevalence of a single α gene deletion (-α) was reported from Madhya Pradesh (73.60 %) while the highest prevalence of 2 α gene deletions (- α / - α) was from Gujarat (72.97 %). A couple of studies have also reported uncommon mutations like the South African deletion, the South East Asian deletion, the Poly A deletion, Hb Constant Spring, Hb Koya Dora, Hb Rampa and Hb Pakse. α gene triplication was also reported in a few studies with the prevalence varying from 0.85 to 5.1 % with a single case of quadruplicated α genes in one study.

Table 1: Prevalence of mutation causing α-thalassaemia

Sr. No.	States/UTs	Districts	Communities	Total Tested	α-Thalassemia cases No (%)	Ref
1	Andhra Pradesh*	Not Mentioned	Valmiki (Non-tribal)	90[a]	-α/αα→31(34.44) -α/-α→42(46.66)	1
2	Assam*	Not Mentioned	Kachari (Tribal)	102	$-\alpha^{3.7}/\alpha\alpha$→ 51 (50.00) $-\alpha^{4.2}/\alpha\alpha$→1 (0.98) ααα/αα→1 (0.98)	2
3	Odisha*	Not Mentioned	Not Mentioned	282	-α/αα and -α/-α→82 (29.00)	3
4	Madhya Pradesh*	Mandala, Jabalpur	Non-tribal	54[b]	$-\alpha^{3.7}/\alpha\alpha$→ 0.037 $-\alpha^{4.2}/\alpha\alpha$→ 0.37	4
5	Madhya Pradesh*	Mandala, Jabalpur	Gond	308[b]	$-\alpha^{3.7}/\alpha\alpha$→ 0.292 $-\alpha^{4.2}/\alpha\alpha$→ 0.224 αα Koya Dora→ 0.023	4
6	Madhya Pradesh*	Jabalpur	Baiga	125[a,c]	-α/αα→92 (73.60) -α/-α→ 29 (23.20)	5
7	Andhra Pradesh*	Adilabad, Vishakhapatnam, East & West Godavari	Konda Reddy	17	$-\alpha^{3.7}/\alpha\alpha$→ 6 (35.29)	6
8	Andhra Pradesh*	Adilabad,Vishakhapatnam, East & West Godavari	Valmiki	50	$-\alpha^{3.7}/\alpha\alpha$→13 (26.00) $-\alpha^{3.7}/-\alpha^{3.7}$→ 6 (12.00) $-\alpha^{4.2}/\alpha\alpha$→ 4 (8.00)	6
9	Andhra Pradesh*	Adilabad,Vishakhapatnam, East & West Godavari	Konda Dora	22	$-\alpha^{3.7}$ /αα→4 (18.18) $-\alpha^{4.2}/\alpha\alpha$→7 (31.81) αα Koya Dora→1 (4.54)	6
10	Andhra Pradesh*	Adilabad,Vishakhapatnam, East & West Godavari	Konda Kammari	24	$-\alpha^{3.7}/\alpha\alpha$→7 (29.16) $-\alpha^{3.7}/-\alpha^{3.7}$→2 (8.33) $-\alpha^{4.2}/\alpha\alpha$→6 (25.00)	6
11	Andhra Pradesh*	Adilabad,Vishakhapatnam, East & West Godavari	Kotiya	10	$-\alpha^{3.7}/\alpha\alpha$→5 (50.00) $-\alpha^{4.2}/\alpha\alpha$→2 (20.00)	6
12	Andhra Pradesh*	Adilabad,Vishakhapatnam, East & West Godavari	Bhaghatha	27	$-\alpha^{3.7}/\alpha\alpha$→12 (44.44) $-\alpha^{4.2}/\alpha\alpha$→7 (25.92)	6
13	Andhra Pradesh*	Adilabad,Vishakhapatnam, East & West Godavari	Koya Dora	30	$-\alpha^{3.7}/\alpha\alpha$→9 (30.00), $-\alpha^{3.7}/-\alpha^{3.7}$→2 (6.66) $-\alpha^{4.2}/\alpha\alpha$→10 (33.33) αα Koya Dora→2 (6.66) $-\alpha^{3.7}$ Rampa→2 (6.66)	6
14	Andhra Pradesh*	Adilabad,Vishakhapatnam, East & West Godavari	Kolam	13	$-\alpha^{3.7}/\alpha\alpha$→7 (53.84) $-\alpha^{3.7}/\alpha^{3.7}$→2 (15.38) $-\alpha^{4.2}/\alpha\alpha$→2 (15.38)	6
15	Andhra Pradesh*	Adilabad,Vishakhapatnam, East & West Godavari	Nooka Dora	8	$-\alpha^{3.7}/\alpha\alpha$→ 4 (50.00) $-\alpha^{3.7}/-\alpha^{3.7}$→1 (12.50) $-\alpha^{4.2}/\alpha\alpha$→1 (12.50)	6
16	Andhra Pradesh*	Not Mentioned	Non-tribals	170	$-\alpha^{3.7}/\alpha\alpha$→ 15 (8.82) $-\alpha^{3.7}$ /$-\alpha^{3.7}$→2 (1.17) $-\alpha^{4.2}/\alpha\alpha$→3 (1.76)	6
17	Maharashtra*	Not Mentioned	Non-tribal	93	-α/αα→ 14 (15.05) -α/-α→1 (1.07) ααα/αα→ 4 (4.30)	7
18	Maharashtra*	Not Mentioned	Tribals	17	-α/αα→2 (11.76) -α/-α→0 ααα/αα→0	7

Sr. No.	State	Districts	Community	Total Tested	α-Thalassemia cases No. (%)	Ref
19	West Bengal*	Not Mentioned	Not Mentioned	12	-α/αα→1 (8.33) -α/-α→0 ααα/αα→0	7
20	Punjab*	Not Mentioned	Not Mentioned	19	-α/αα→ 4 (21.05) -α/-α→ 1 (5.26) ααα/αα→ 0	7
21	Rajasthan*	Not Mentioned	Tribals	17	-α/αα→2 (11.76) -α/-α→0 ααα/αα→0	7
22	Maharashtra*	Nagpur	Scheduled caste	117[a,c]	-α/αα→10 (8.54) -α/-α→1 (0.85) ααα/αα→1 (0.85)	8
23	Gujarat*	Valsad	Tribal	67[a,c]	-α/-α→ 50 (74.62) -α/αα→15 (22.38)	8
24	Rajasthan*	Banswar,Bhilara,Chittorgarh, Dungarpur, Sirohi,Udaipur	Scheduled cast, General caste	1129[d]	-α/αα→10 (0.88) -α/-α →2 (0.17)	9
25	Rajasthan*	Banswar, Bhilara, Chittorgarh, Dungarpur, Sirohi,Udaipur	Scheduled tribe	618[d]	-α/αα →14 (2.26) -α/-α→5 (0.32)	9
26	Chandigarh, Punjab, Haryana, Himachal Pradesh*	Not Mentioned	Not Mentioned	419[e]	-α/αα or -α/-α→52 (12.41) ααα/αα →13 (3.10) αααα/αα → 1 (0.23)	10
27	New Delhi#	Not Mentioned	Not Mentioned	118[f]	$-\alpha^{3.7}/\alpha\alpha$ or $-\alpha^{3.7}/-\alpha^{3.7}$ →18 (15.25) $\alpha\alpha/\alpha\alpha\alpha^{anti3.7}$→3 (2.54)	11
28	West Bengal*	Not Mentioned	Not Mentioned	100	$-\alpha^{3.7}$ or $-\alpha^{4.2}$→18 (18.00)	12
29	Arunachal Pradesh*	Itanagar	Adi, AdiGalong	179	$-\alpha^{3.7}$or $-\alpha^{4.2}$→ 7 (3.91)	12
30	Assam*	Dibrugarh	Ahom,Garo, Kalita ,Matta Kachari, Michty, Chutia, Miri	234	$-\alpha^{3.7}$ or $-\alpha^{4.2}$→ 9 (3.84)	12
31	Uttar Pradesh*	Not Mentioned	Not Mentioned	276	-α/αα→33(11.95) -α/-α→2 (0.72) ααα/αα→14 (5.07)	13
32	West Bengal*	Not Mentioned	Bengali	133	$-\alpha^{3.7}/\alpha\alpha$→2 (1.50) $-\alpha^{4.2}/\alpha\alpha$→2 (1.50) $-\alpha^{4.2}/-\alpha^{4.2}$→1 (0.75) $-\alpha^{3.7}/-\alpha^{4.2}$ →1 (0.75)	14
33	Punjab*	Not Mentioned	Mixed group	38	-α/αα→ 9 (23.68) -α/-α→ 1 (2.63) ααα/αα→ 1 (2.63)	15
34	Uttar Pradesh*	Not Mentioned	Mixed group	35	-α/αα→ 6 (17.14)	15
35	Karnataka*	Not Mentioned	Mixed group	31	-α/αα→ 5 (16.12) -α/-α→ 1 (3.22)	15
36	Rajasthan*	Not Mentioned	Mixed group	42	-α/αα→ 6 (14.00) ααα/αα→ 2 (5.00)	15
37	Madhya Pradesh*	Not Mentioned	Mixed group	89	-α/αα→ 11 (12.35) -α/-α→ 3 (3.37)	15
38	Maharashtra*	Not Mentioned	Mixed group	510	-α/αα→ 61 (11.96) -α/-α→ 17 (3.33) ααα/αα→ 5 (0.98) -α/ααα→1 (0.19)	15
39	West Bengal*	Not Mentioned	Mixed group	24	-α/αα→ 5 (20.83)	15

Sr. No.	State	Districts	Community	Total Tested	α-Thalassemia cases No. (%)	Ref
40	Gujarat*	Not Mentioned	Mixed group	311	-α/αα→ 10 (3.21) -α/-α→ 5 (1.60) ααα/αα→ 5 (1.60)	15
41	Miscellaneous*	Not Mentioned	Mixed group	89	-α/αα→ 7 (7.86) -α/-α→ 1 (1.12)	15
42	Immigrants from Pakistan*	Not Mentioned	Not Mentioned	84	-α/αα→ 12 (14.28) -α/-α→ 1 (1.19) ααα/αα→ 2 (2.38)	15
43	New Delhi#	Not Mentioned	Not Mentioned	240[h]	$-\alpha^{3.7}/\alpha\alpha$→19 (7.91) $-\alpha^{3.7}/-\alpha^{3.7}$→4 (1.66) $--^{SA}/\alpha\alpha$→2 (0.83) $\alpha\alpha^{CS}/\alpha\alpha$→8 (3.33) $\alpha\alpha^{PS}/\alpha\alpha$→6 (2.50) $\alpha^{CS}\alpha^{PS}/\alpha\alpha$→1 (0.41) $\alpha\alpha/\alpha\alpha\alpha^{anti3.7}$→7 (2.91) $-\alpha^{3.7}/\alpha\alpha^{CS}$→1 (0.41)	16
44	New Delhi#	Not Mentioned	Not Mentioned	60[c]	$-\alpha^{3.7}/\alpha\alpha$→ 9 (15.00) $-\alpha^{3.7}/-\alpha$ → 7 (11.66) $-\alpha^{4.2}/\alpha\alpha$→ 2 (3.33) ααα/αα→3 (5.00)	17
45	Odisha*	Kalahandi, Bargrah	Sahara, Kandha, Oraon, Gond, Kuda (Tribal)	594	$-\alpha^{3.7}/\alpha\alpha$→ 148 (24.91) $-\alpha^{4.2}/\alpha\alpha$→ 180 (30.30) $-\alpha^{3.7}/-\alpha^{3.7}$→ 106 (17.84) $-\alpha^{4.2}/-\alpha^{4.2}$→ 2(0.34) $-\alpha^{3.7}/-\alpha^{4.2}$→ 28 (4.71)	18
46	Maharashtra*	Raigad	Thakkar	33	$-\alpha^{3.7}/\alpha\alpha$→ 7 (21.21) $-\alpha^{3.7}/-\alpha^{3.7}$→ 21 (63.63)	19
47	Maharashtra*	Raigad	Katkari, Kokana	18	$-\alpha^{3.7}/\alpha\alpha$→ 3 (16.66) $-\alpha^{3.7}/-\alpha^{3.7}$ → 13 (72.22) $-\alpha^{3.7}/-\alpha^{4.2}$→ 1 (5.55)	19
48	Uttar Pradesh, Chhattisgarh, Bihar, Jharkhand*	Varanasi	Mixed group	939	$-\alpha^{3.7}/\alpha\alpha$→ 90 (9.58) $-\alpha^{4.2}/\alpha\alpha$→37 (3.94) $-\alpha^{3.7}/-\alpha^{3.7}$→ 17 (1.81) $-\alpha^{4.2}/-\alpha^{4.2}$→11 (1.17) $-\alpha^{3.7}/-\alpha^{4.2}$→ 27 (2.87) $\alpha\alpha\alpha^{anti3.7}$→ 19 (2.02) $\alpha\alpha\alpha^{anti4.2}$→ 4 (0.42)	20
49	Delhi#	New Delhi	Not Mentioned	54[i]	$-\alpha^{3.7}/\alpha\alpha$→15 (27.77) $-\alpha^{3.7}/-\alpha^{3.7}$→ 3 (5.55) $\alpha\alpha/--^{SA}$→ 1 (1.85)	21
50	Gujarat*	Valsad, Navsari, Dang, and Surat	Tribal	37[j]	$-\alpha^{3.7}/\alpha\alpha$→4 (10.81) $-\alpha^{3.7}/-\alpha^{3.7}$→27 (72.97) $-\alpha^{4.2}/\alpha\alpha$→1 (2.70) $-\alpha^{3.7}/-\alpha^{4.2}$→2 (5.40)	22
51	Madhya Pradesh*	Damoh	Chaudhary (Non-tribal)	168	$-\alpha^{3.7}/\alpha\alpha$→ 7 (4.16) $-\alpha^{4.2}/\alpha\alpha$→ 8 (4.76)	23
52	Madhya Pradesh*	Damoh	Raj Gond (Tribal)	267	$-\alpha^{3.7}/\alpha\alpha$→ 47 (17.60) $-\alpha^{4.2}/\alpha\alpha$→ 54 (20.22) $-\alpha^{3.7}/-\alpha^{3.7}$→106 (39.70) $-\alpha^{3.7}/-\alpha^{4.2}$→ 1 (0.37)	23
53	Tripura*	Agartala	Tribal/ Non-tribal	162	-α/αα→42 (25.92) -α/-α→7 (4.32)	24
54	Maharashtra*	Mumbai	Pathare Prabhus	257	$-\alpha^{3.7}/\alpha\alpha$→ 14 (5.44)	25

Sr. No.	State	Districts	Community	Total Tested	α-thalassaemia cases No. (%)	Ref
55	Maharashtra*	Nagpur	Not Mentioned	265	-α/αα→75 (28.30) -α/-α→ 57 (21.50)	26
56	Maharashtra#	Not Mentioned	Mixed group	1275	$-\alpha^{3.7}/\alpha\alpha$→131 (10.27) $-\alpha^{3.7}/-\alpha^{3.7}$→50 (3.92) $-\alpha^{4.2}/\alpha\alpha$→22 (1.72) $-\alpha^{4.2}/-\alpha^{4.2}$→11 (0.86) $-\alpha^{3.7}/-\alpha^{4.2}$→2 (0.15) Poly A (AATAAA→AATA)trait→8(0.62) $--^{SEA}/\alpha\alpha$→ 4 (0.31) $--^{SA}/\ \alpha\alpha$→2 (0.15) αα/αααα→53 (4.15)	27
57	Madhya Pradesh*	Chhindwara, Dindori, Mandla	Tribal	3353	$-\alpha^{4.2}/\alpha\alpha$→547(16.31) $-\alpha^{4.2}/-\alpha^{4.2}$→ 367 (10.94) $-\alpha^{3.7}/\alpha\alpha$→ 629 (18.75) $-\alpha^{3.7}/-\alpha^{3.7}$→570 (16.99) $-\alpha^{3.7}/-\alpha^{4.2}$→715 (21.32)	28
58	Maharashtra#	Not Mentioned	Mixed group	580^{k}	-α/αα→100 (17.24) -α/-α→ 29 (5.00)	29

a: Sickle cell trait cases; b: Number of chromosomes studied; c: Sickle cell anaemia cases; d: α-genotypes are based on % of Hb Bart's; e: β-thalassaemia trait and normal individuals; f: Normal, β thalassaemia trait, β-thalassaemia intermedia and β-thalassaemia major cases. g: β-thalassaemia trait, β-thalassaemia major, Sickle cell anaemia, HbS-β-thalassaemia, HbE-β-thalassaemia, Iron deficiency anaemia, other hemolytic anaemia, and undiagnosed cases; h: HbE-β thalassaemia cases; i: Microcytic anaemia cases, Normal, j: Sickle cell anaemia and HbS- β-thalassaemia cases; k: Normal, β-thalassaemia trait, β-thalassaemia major, Sickle cell trait, Sickle cell anaemia, HbE trait cases; CS: Constant Spring; PS: Hb Pakse; SA: South Africa; SEA: South East Asia * State of origin; # State where diagnosis has been done.

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Case Reports

This section on Case Reports has been divided into 4 groups as shown in Table 1.

Table 1 : Four sub-sections of Case Reports

1	Hb H Disease
2	HPFH and δβ Thalassaemia
3	Common Hb Variants
4	Novel and Rare Hb Variants

Some of them are single case reports while there were several studies where a series of cases were reported.

1.HbH Disease cases

There were 10 reports on Hb H disease cases from 11 States and one Union Territory. Fig 1 shows the regions from where these cases originated or were reported (shaded areas).



Fig 1: Map of India showing the States and Union Territories where cases of Hb H Disease originated or were reported

Only those cases where molecular analysis was done for confirmation of the genotypes have been included. The maximum number of cases were from Chandigarh and Maharashtra. Deletional as well as non-deletional α gene mutations in different combinations have been reported. The non-deletional Hb Sallanches and the Poly A(-AA) mutation were quite common (Table 2).

Occasional cases of Hb H disease were also picked up during population screening, antenatal screening or hospital based screening in different states but these were mainly based on phenotypic analysis.

Table 2: HbH disease cases

Sr. No.	States/UTs	Communities	HbH Disease	Mutations	Ref.
1	Assam*	Not Mentioned	$-\alpha^{3.7}/^{--Kol}\rightarrow 1$	3.7 kb (type I) deletion α2+ 33.3 kb deletion	1
2	Chandigarh#	North Indian	Hb Sallanches homozygous→ 6 $\alpha^{Poly\ A}\ \alpha/\alpha^{Poly\ A}\ \alpha\rightarrow 3$ $-\alpha^{3.7}/\alpha^{\ Hb\ Sallanches}\ \alpha\rightarrow 1,$ $\alpha^{76\ +T}\alpha/\alpha^{Hb\ Sallanches}\ \alpha\rightarrow 1$ $\alpha^{Poly\ A}\ \alpha/\ \alpha^{Hb\ Sun\ Prairie}\ \alpha\rightarrow 1$ $\alpha^{Poly\ A}\ \alpha/\ \alpha^{76\ +T}\alpha\rightarrow 1$ α2 Codon 19 (-G) homozygous→1 $-\alpha^{3.7}/^{--SA}\rightarrow 3,$ $\alpha^{Hb\ Seal\ Rock}\ \alpha/--\rightarrow 1$ $-\alpha^{3.7}/\alpha^{Hb\ Seal\ Rock}\ \alpha\rightarrow 1$ $-\alpha^{3.7}/--\ \rightarrow 15$	α2 CD104 (G→A) homozygous α2 Poly A (AATAAA> AATA--) homozygous 3.7 kb (type I) deletion α2 + α2 CD 104 (G→A) α CD 76 (+T) + CD 104 (G→A) α2 Poly A (AATAAA> AATA--)+ α2 CD 130 (G→C) α2 Poly A (AATAAA> AATA--) + α CD 76 (+T) α2 CD 19 (-G) homozygous 3.7 kb (type I) deletion α2+ α 23601 bp deletion α2 142 (T→G)+ Large deletion involving α1 and α2 3.7 kb (type I) deletion α2+ α2 142 (T→G) 3.7 kb (type I) deletion α2+ Large deletion involving α1 and α2	2
3	Chandigarh#	Sikh	αα/α Sallanches α Zürich-Albisrieden→1	α2 CD 104 (G→A) + CD 59 (G→C)	3
4	Karnataka*	Not Mentioned	$-\alpha^{3.7}/^{--SA}\rightarrow 3$	α 23601 bp deletion+ 3.7 kb (type I) deletion α2	4
5	Kerala*	Not Mentioned	$-\alpha^{3.7}/^{--SEA}\rightarrow 1$	α 19301 bp deletion+ 3.7 kb (type I) deletion α2	4
6	Madhya Pradesh*	Not Mentioned	$-\alpha^{4.2}/^{--SA}\rightarrow 1$	4.2 kb deletion + α 23601 bp deletion	5
7	Maharashtra*	Not Mentioned	$-\alpha^{3.7}/-\alpha^{3.7\ Sallanches}\rightarrow 1$	3.7 kb (type I) deletion α2 + α2 CD 104 (G→A)	4
8	Maharashtra#	Muslim	Hb Sallanches Homozygous→1	α2 CD104 (G→A) homozygous	6
9	Maharashtra*	Maratha, Sunni	Poly A(-AA) homozygous→3	α2 Poly A (AATAAA→ AATA--) homozygous	7
10	Maharashtra#	Mixed group	Poly A (-AA) homozygous→ 8 Hb Sallanches homozygous→1 $-\alpha^{3.7}/^{--MED}\rightarrow 2$ $-\alpha^{3.7}/^{--SEA}\rightarrow 9$ $-\alpha^{3.7}/^{--SA}\rightarrow 4$ $-\alpha^{3.7}/\alpha^{Sallanches}\alpha\rightarrow 4$ $--SA/-\alpha^{3.7Sallanches}\rightarrow 2$ $-\alpha^{3.7}/-\ \alpha^{3.7Sallanches}\ \rightarrow 1$ $--^{SA}/\alpha^{Sallanches}\alpha\rightarrow 1$ $--^{SEA}/\alpha^{Sallanches}\alpha\rightarrow 1$	α2 Poly A (AATAAA→AATA--) homozygous α2 CD 104 (G→A) homozygous 3.7 kb (type I) deletion α2+ α 16401 bp deletion 3.7 kb (type I) deletion α2+ α 19301 bp deletion 3.7 kb (type I) deletion α2+ α 23601 bp deletion 3.7 kb (type I) deletion α2+ α2 CD 104 (G→A) α 23601 bp deletion + 3.7 kb (type I) deletion α2 +α2 CD 104 (G→A) 3.7 kb (type I) deletion α2 + 3.7 kb (type I) deletion α2+ α2 CD 104 (G→A) α 23601 bp deletion+ α2 CD 104 (G→A) α 19301 bp deletion+ α2 CD 104 (G→A)	8
11	Manipur*	Not Mentioned	$-\alpha^{3.7}/--^{SEA}/\rightarrow 1$	3.7 kb (type I) deletion α2+ α 19301 bp deletion	4

(contd.)

Sr. No.	States/UTs	Communities	HbH Disease	Mutations	Ref.
12	Odisha*	Not Mentioned	$-\alpha^{3.7}/--^{SA}\rightarrow 2$	3.7 kb (type I) deletion α2+ α 23601 bp deletion	5
13	Punjab*	Not Mentioned	$-\alpha^{3.7}/--^{SA}\rightarrow 1$	3.7 kb (type I) deletion α2+ α 23601 bp deletion	5
14	Punjab*	Punjabi	Hb Sallanches homozygous→1	α2 CD104 (G→A) homozygous	9
15	Tamil Nadu*	Not Mentioned	Poly A (-AA) homozygous→ 2 $-\alpha^{3.7}/--^{MED}\rightarrow 1$ $-\alpha^{3.7}/--^{SEA}\rightarrow 1$	α2 Poly A (AATAAA> AATA--) homozygous 3.7 kb (type I) deletion α2+ α 16401 bp deletion 3.7 kb (type I) deletion α2+ α 19301 bp deletion	5
16	Uttar Pradesh*	Khan	Poly A (-AA) homozygous→1	α2 Poly A (AATAAA> AATA--) homozygous	7
17	West Bengal*	Not Mentioned	$--^{SA}/-\alpha^{3.7Sallanches}\rightarrow 1$	α 23601 bp deletion +3.7 kb (type I) deletion α2 + α2 CD 104 (G→A)	4
18	West Bengal*	Not Mentioned	$\alpha\alpha/--^{Kol}$ trait→2[a]	33.3 kb deletion	1
19	West Bengal*	Not Mentioned	$-\alpha^{3.7}/--^{SA}\rightarrow 4$ $-\alpha^{4.2}/--^{SA}\rightarrow 2$ $\alpha^{IVS\ 1\text{-}1\ (G>A)}\alpha/--^{SA}\rightarrow 3$	3.7 kb (type I) deletion α2+ α 23601 bp deletion 4.2 kb deletion + α23601 bp deletion α IVS I - I (G→A) + α23601 bp deletion	10

a: Non deletional mutation was not available; SA: South African; SEA: South East Asian, MED: Mediterranean, Kol: Kolkata

* States/UTs of Origin; # States/UTs where diagnosis has been done

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2. HPFH and δβ Thalassaemia

There were 8 reports from 3 States and one Union Territory where molecular characterization was done to differentiate cases of HPFH from δβ thalassaemia. Fig 2 shows the regions from where these cases originated or were reported.

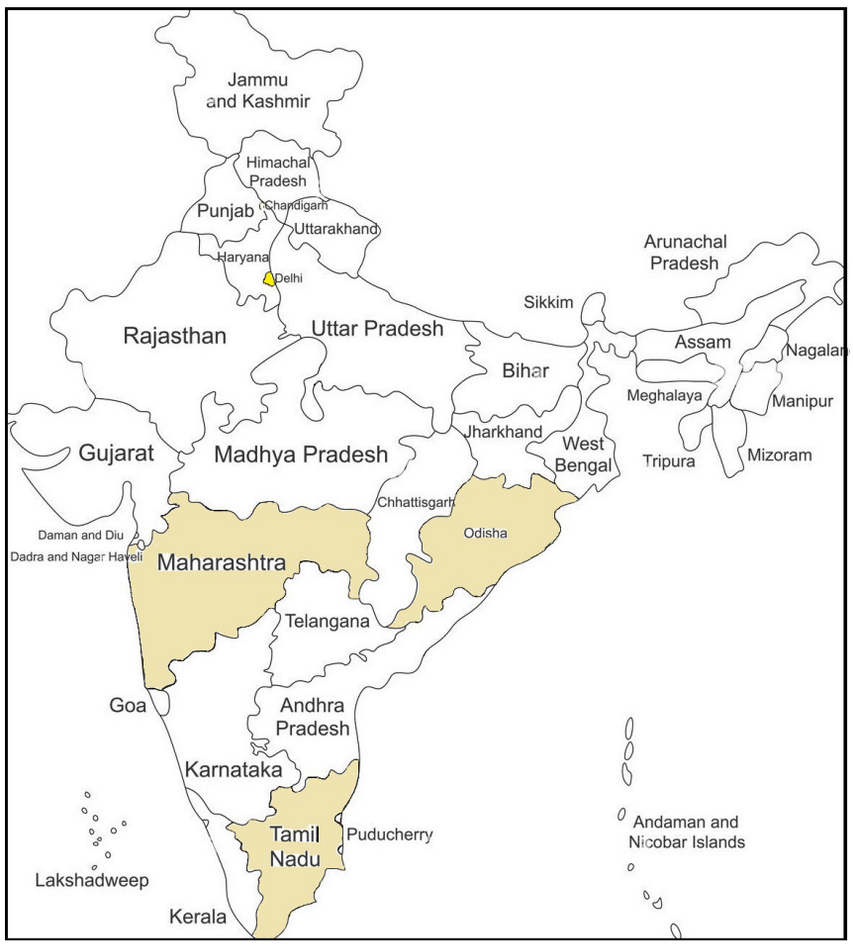

Fig 2 - Map of India showing the States and Union Territories where cases of HPFH and δβ thalassaemia originated or were reported.

Several large deletions have been reported. Four of the studies include a series of a large number of cases with increased Hb F levels. The most common δβ thalassaemia mutation reported was the Asian Indian Inversion Gγ(Aγδβ)0 thalassaemia while the most common HPFH mutation reported was the 48.5 Kb deletion (HPFH - 3) (Table 3).

Few cases of HPFH and δβ thalassaemia have also been picked up during population screening, antenatal screening and hospital based studies from different states, although they were not all confirmed by molecular analysis and often the 2 groups were not differentiated between each other.

Table 3: HPFH and δβ thalassaemia cases

Sr. No.	States/UTs	Communities	HPFH and δβ Thalassaemia	Mutations	Ref.
1	Maharashtra#	Not Mentioned	δβ thalassaemia trait→20 HPFH trait →26 Uncharacterized →9	Asian Indian Inversion Gγ(Aγδβ)0 thal deletion heterozygous→15 Vietnamese/Chinese 27 kb deletion heterozygous →5 48.5 kb deletion (HPFH 3) heterozygous → 26	1
2	Maharashtra#	Not Mentioned	δβ thalassaemia trait→ 122 HPFH trait→ 57 Uncharacterized →13	Asian Indian Inversion Gγ (Aγδβ)0 thal deletion heterozygous →73 49.3 Kb Gγ (Aγδβ)0 deletion heterozygous→39 32.6 kb Gγ (Aγδβ)0 deletion heterozygous→ 10 48.5 kb deletion (HPFH 3) heterozygous→57	2
3	New Delhi#	Not Mentioned	δβ thalassaemia homozygous→ 1	Asian Indian Inversion Gγ(Aγδβ)0 thal deletion homozygous	3
4	New Delhi#	Not Mentioned	δβ thalassaemia trait→7 δβ thalassaemia homozygous →3 δβ thalassaemia + -α3.7 →5 β-δβ thalassaemia→3 HPFH trait→28	Asian Indian Inversion Gγ(Aγδβ)0 thal deletion hetrozygous→7 Asian Indian Inversion Gγ(Aγδβ)0 thal deletion homozygous→3 Asian Indian Inversion Gγ(Aγδβ)0 + 3.7 kb (type I) deletion α2 →5 β IVS 1-5 (G→C) + Asian Indian Inversion Gγ(Aγδβ)0 thal deletion →3 48.5 kb deletion (HPFH 3) heterozygous →28	4
5	New Delhi#	Not Mentioned	HbS/δβ thalassaemia→ 4 HbS/HPFH→2	β CD 6 (A→T) + Asian Indian Inversion Gγ(Aγδβ)0 thal deletion →4 β CD 6 (A→T) + 48.5 kb deletion (HPFH 3) →2	5
6	Odisha*	Burla	HbS-δβ thalassaemia→3	β CD 6 (A→T) + Asian Indian Inversion Gγ (Aγδβ)0 thal deletion → 3	6
7	Odisha*	Chasa	HPFH trait→1	48.5 kb deletion (HPFH 3) heterozygous→1	7
8	Tamil Nadu#	Not Mentioned	δβ thalassaemia trait → 27 HPFH trait→14	Indian Inversion Gγ (Aγδβ)0 thal deletion heterozygous→15 32.6Kb Gγ (Aγδβ)0 deletion heterozygous→ 5 49.3 Kb Gγ (Aγδβ)0 deletion heterozygous→ 7 48.5 kb deletion (HPFH 3) heterozygous→9 49.98 Kb deletion (HPFH 9) heterozygous→4 86.7 Kb deletion (HPFH 10) heterozygous→1	8

*States/UTs of origin, #States/UTs where diagnosis has been done

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3. Common Hb Variants

In this group we have included Hb variants which are not extremely rare and yet are not seen as frequently as the 3 most common Hb variants in India, Hb S trait, Hb E trait and Hb D Punjab trait. Thus, case reports of HbQ India and the relatively uncommon co-inherited disorder, HbSD Punjab disease are included. Case reports of the more common compound heterozygotes, HbD Punjab-β thalassaemia and HbS-β thalassaemia reported from different states have been included only where the mutations were analysed. The states from where these cases originated or have been reported are shown in Fig 3



Fig 3: Map of India showing the states where cases of the common Hb variants originated or were reported

Majority of the studies include single case reports with only one large series of 64 cases of HbQ India either in heterozygous or homozygous state or co-inherited with β thalassaemia (Table 4).

Table 4: Common Hb variants

Sr. No.	States/UTs	Communities	Common Hb variants	Mutations	Ref.
1	Andhra Pradesh*	Sunni	HbSD Punjab disease→1	β CD 6 (A→T) + β CD 121 (G→C)	1
2	Assam*	Not Mentioned	HbS-β-thalassaemia→1	β CD 6 (A→T) + β IVS 1-5 (G→C)	2
3	Assam*	Not Mentioned	HbS-β-thalassaemia→1	β CD 6 (A→T) + β IVS 1-5 (G→C)	3
4	Gujarat#	Sindhi	HbQ india trait→1	α1 CD 64 (G→C) heterozygous	4
5	Gujarat*	Tribal	HbS-β-thalassaemia→5	β CD 6 (A→T) + β CD 15 (G→C)	5
6	Gujarat*	Non-Tribal	HbS-β-thalassaemia→3	β CD 6 (A→T) + β CD 30 (G→C) β CD 6 (A→T) + β IVS 1-5 (G→C)	5
7	Gujarat*	Prajapati, SC, Naika, Ghachi	HbSD Punjab disease→4	β CD 6 (A→T) + β CD 121 (G→C)	1
8	Jharkhand*	Ansari	HbSD Punjab disease→1	β CD 6 (A→T) + β CD 121 (G>C)	1
9	Madhya Pradesh*	Bilala	HbSD Punjab disease→1	β CD 6 (A→T) + β CD 121 (G→C)	1
10	Maharashtra#	Sindhi	HbQ India -β- thalassemia[a]→3	α1 CD 64 (G→C) + β thalassaemia	6
11	Maharashtra#	Kutchi Lohana	HbQ India Disease→1	α1 CD 64 (G→C) homozygous	7
12	Maharashtra#	Not Mentioned	HbSD Punjab disease→1	β CD 6 (A→T) + β CD 121 (G→C)	8
13	Maharashtra#	Mixed	HbQ India Trait→36 HbQ India Disease→3 HbQ India -β thalassaemia trait→22 HbQ India -β thalassaemia major→ 3	α1 CD 64 (G→C) heterozygous α1 CD 64 (G→C) homozygous α1 CD 64 (G→C) + β 619 bp deletion, CD 8/9 (+G), IVS 1-1 (G→T), IVS 1-5 (G→C), CD 30 (G→C) α1 CD 64 (G→C) + β 619 bp deletion, IVS 1-1 (G→T), IVS 1-5 (G→C)	9
14	Maharashtra*	Non-Tribal	HbS-β-thalassaemia→11	β CD 6 (A→T) + β IVS 1-5 (G→C) β CD 6 (A→T) + β CD 30 (G→C) β CD 6 (A→T) + β CD 15 (G→A) β CD 6 (A→T) + β CD 8/9 (+G)	5
15	Maharashtra*	Adivasi, Shaikh, Kunbi, Punjabi, Mahar	HbSD Punjab disease→6	β CD 6 (A→T) + β CD 121 (G→C)	1

(contd.)

Sr. No.	States/UTs	Communities	Common Hb variants	Mutations	Ref.
16	Odisha*	Brahmin, Telugu	HbSD Punjab disease→2	β CD 6 (A→T) + β CD 121 (G→C)	1
17	Punjab*	Not Mentioned	HbD Punjab -β-thalassaemia→1	β CD121 (G→C) + β CD 8/9 (+G)	10
18	Punjab*	Not Mentioned	HbQ India trait → 3	α1 CD 64 (G→C) heterozygous	11
19	Punjab*	Not Mentioned	HbD Punjab -β-thalassaemia→3	β CD121 (G→C) + CD 8/9 (+G) β CD121 (G→C)+ IVS 1-5 (G→C) β-thalassaemia mutation in one case is not available	12
20	Telangana*	Sindhi (n=3), Not mentioned	HbQ India trait→ 4 HbQ India β- thalassaemia[a] →1	α1 CD 64 (G→C) heterozygous α1 CD 64 (G→C)+ β-thalassemia[a]	13
21	Uttar Pradesh*	Not Mentioned	HbQ India -β-thalassaemia→1	α 1 CD 64 (G→C)+ β IVS 1-1 (G→T)	14
22	West Bengal*	Not Mentioned	HbD Punjab trait + α-thalassaemia→1	β CD 121 (G→C) heterozygous + $-\alpha^{3.7}/\alpha\alpha$	15

* States/UTs of Origin; # States/UTs where diagnosis has been done

a: β-thalassaemia mutations not available

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4. Novel and Rare Hb Variants

This sub-section includes 77 Case Reports on a large number of novel and rare α, β, δ or δβ hybrid Hb variants, some of them co-inherited with β thalassaemia or Hb S. Few variants like Hb D Agri with 2 amino acid substitutions in the same β chain were novel while many others were reported for the first time in the Indian population. Also included are case reports with co-inheritance of 2 Hb variants like Hb SE disease, Hb DE disease or Hb D Iran-Hb D Punjab disease which are rare.In many of these case reports the communities have not been mentioned. The states and union territories from where these rare variants originated or have been reported are shown in Fig 4.



Fig 4 : Map of India showing the states and union territories where novel or rare Hb variants originated or were reported

There are few rare Hb variants which were reported before molecular analysis was available. These have also been shown below Table 5.

Table 5: Novel and Rare Hb Variants

Sr. No.	States/UTs	Communities	Novel and Rare Hb Variants	Mutations	Ref.
1	Andhra Pradesh*	Tribals	Hb Godavari trait→1 HbS-Hb Godavari→1	α2 or α1 CD 95 (C→A) heterozygous β CD 6 (A→T)+ α2 or α1 CD 95 (C→A)	1
2	Andhra Pradesh*	Not Mentioned	HbM Boston trait→1	α2 or α1 CD 58 (C→T) heterozygous	2
3	Andhra Pradesh*	Not Mentioned	HbM Hyde Park trait→1	β CD 92 (C→T) heterozygous	2
4	Andhra Pradesh*	Not Mentioned	Hb Yaizu trait→1	β CD 79 (G→A) heterozygous	3
5	Assam*	Brahmin	HbE+ HbJ Meerut →1	β CD 26 (G→A)+ α2 or α1 CD120 (C→A)	4
6	Bihar*	Ansari, Shaikh	HbSE disease→3	β CD 6 (A→T)+ β CD 26 (G→A)	5
7	Chandigarh#	Khatri	HbQ India- HbD Punjab→ 1	α1 CD 64 (G→C) + β CD 121 (G→C)	6
8	Chandigarh#	Punjabi	Hb M Iwate Trait→1	α1 or α2 CD 87 (C→T) heterozygous	7
9	Chandigarh#	Not Mentioned	Hb M Saskatoon trait→3	β CD 63 (C→T) heterozygous	8
10	Chandigarh#	Sikh	HbD Punjab-Hb Brugg→1	β CD 121 (G→C)+ α1 CD 20 (C→A)	9
11	Chandigarh#	Not Mentioned	HbD Punjab β-thal-HbQ India→ 1	β CD 121 (G→C)+ CD 5 (-CT)+ α1 CD 64 (G→C)	10
12	Chhattisgarh*	Sindhi	HbS-HbQ India →1	β CD 6 (A→T) + α1 CD64 (G→C)	11
13	Delhi#	Not Mentioned	Hb Agenogi- β-thalassaemia→ 1	β CD 90 (G→A) + β IVS 1-5 (G→C)	12
14	Delhi#	Not Mentioned	HbD Iran-HbD Punjab →1	β CD 22 (G→C) + β CD 121 (G→C)	13
15	Gujarat*	Muslim Khoja	HbJ Paris-I - β-thalassaemia→1	α2 or α1 CD12 (C→A) + β 619 bp del	4
16	Gujarat*	Muslim Nizama	Hb Sun Prairie trait→1	α2 CD130 (G→C) heterozygous	4
17	Gujarat*	Koli	HbA2 Saurashtra trait→1	δ CD 100 (C→T) heterozygous	14
18	Gujarat*	Not Mentioned	Hb Agenogi trait→ 1	β CD 90 (G→A) heterozygous	15
19	Gujarat*	Not Mentioned	Hb Olympia trait→ 1	β CD 20 (G→A) heterozygous	16
20	Jharkhand*	Not Mentioned	HbSE disease→3	β CD 6 (A→T) + β CD 26 (G→A)	17
21	Jammu &Kashmir*	Not Mentioned	Hb Le Lamentin trait→1	α CD 20 (C→A) heterozygous	18
22	Madhya Pradesh*	Not Mentioned	Hb Hofu - β thalassaemia→1	β CD 126 (T→A) +β CD 8/9 (+G)	19
23	Madhya Pradesh*	Not Mentioned	Hb Lepore Hollandia trait→1	δβ hybrid (δ through 22; β from 50) heterozygous	20
24	Madhya Pradesh*	Gawali	HbS+ Hb Fontainebleau→1	β CD 6 (A→T)+ α2 or α1 21(G→C)	21
25	Madhya Pradesh*	Not Mentioned	HbS+ HbO Indonesia →1 HbO Indonesia trait→1	β CD 6 (A→T)+ α1 CD116 (G→A) α1 CD116 (G→A) heterozygous	22

(contd.)

Sr. No.	States/UTs	Communities	Novel and Rare Hb Variants	Mutations	Ref.
26	Madhya Pradesh*	Baiga	Hb Lepore Hollandia trait →1	δβ hybrid (δ through 22; β from 50) heterozygous	23
27	Madhya Pradesh*	Gond	HbS Disease-HbG -Waimanalo-->1	β CD 6 (A→T) + α2 CD64 (G→A)	24
28	Maharashtra*	Agri	Hb D Agri → 1	β CD 9 (C→A) and β CD 121 (G→C) heterozygous	25
29	Maharashtra*	Not Mentioned	HbM Ratnagiri trait→1	β CD 63 (C→T) heterozygous	26
30	Maharashtra*	Agri	Hb Showa Yakushiji trait→3 Hb Showa Yakushiji homozygous→1 HbE- Showa Yakushiji→1 Hb Showa Yakushiji- β-thalassaemia→1	β CD 110 (T→C) heterozygous β CD 110 (T→C) homozygous β CD 26 (G→A)+ β CD 110 (T→C) β CD 110 (T→C) + β CD 15 (G→A)	27
31	Maharashtra*	Sunni-Muslim	HbO Indonesia trait →1	α1 CD116 (G→A) heterozygous	4
32	Maharashtra*	Brahmin	Hb Jackson trait→1	α2 or α1 CD127 (G→C) heterozygous	4
33	Maharashtra#	North Indian	Hb Lepore Hollandia trait→1	δβ hybrid (δ through 87; β from 116) heterozygous	28
34	Maharashtra*	Agri	Hb Showa Yakushiji trait→7	β Cd 110 (T→C) heterozygous	29
35	Maharashtra*	Gond	HbS-Hb Koya Dora →1	β CD 6 (A→T) + α2 CD 142 (A→C)	21
36	Maharashtra*	Mahar	HbS-Hb O Indonesia→1	β CD 6 (A→T) + α1 CD 116 (G→A)	21
37	Maharashtra*	Agri	Hb Pelendri trait→1	δ CD 141 (T→C) heterozygous	30
38	Maharashtra*	Not Mentioned	Hb Tianhe- β-thalassaemia→1	δ CD 107 (G→A)+ β IVS I-5 (G→C)	31
39	Maharashtra#	Not Mentioned	HbD Punjab-HbQ India→1	β CD 121 (G→C)+ α CD 64 (G→C)	32
40	Maharashtra#	Sindhi	HbD Punjab-HbQ India→1	α1 CD 64 (G→C)+ β CD 121 (G→C)	33
41	Maharashtra*	Not Mentioned	Hb Koln trait→ 1	β CD 98 (G→A) heterozygous	34
42	Maharashtra*	Teli	HbSE disease→1	β CD 6 (A→T) + β CD 26 (G→A)	5
43	Maharashtra#	Muslim	HbD Iran trait→1	β CD 22(G→C) heterozygous	35
44	Maharashtra*	Nhavi	Hb St. Louis trait→1	β CD 28 (T→A) heterozygous	15
45	Maharashtra*	Baudha	Hb G Coushata trait→1	β CD 22 (A→C) heterozygous	15
46	Maharashtra*	Pathare Prabhus	Hb Saurashtra trait→ 1	δ CD 100 (C→T) heterozygous	36
47	Maharashtra*	Not Mentioned	Hb Alperton-β-thalassaemia→1	β CD 135 (C→T) + IVS I-5 (G→C)	37
48	Maharashtra#	Not Mentioned	Hb Beth Israel trait→3 Hb Hofu trait→2 HbJ Cambridge trait →2 HbJ Mizunami+HbS→1 Hb Sherwood Forest trait→1	β CD 102 (A→G) heterozygous β CD 126 (T→A) heterozygous β CD 69 (G→A) heterozygous β CD 83 (G→A)+ β CD 6 (A→T) β CD 104 (G→C) heterozygous	38

(contd.)

Sr. No.	States/UTs	Communities	Novel and Rare Hb Variants	Mutations	Ref.
49	Maharashtra*	Not Mentioned	HbJ Norfolk Trait→ 1	α2 or α1 CD 57 (G→A) heterozygous	39
50	New Delhi#	Not Mentioned	HbJ Meerut trait→7	α2 or α1 CD120 (C→A) heterozygous	40
51	New Delhi#	Not Mentioned	HbC-β-thalassaemia→1	β CD 6 (G→A)+ β CD 8/9 (+G)	41
52	New Delhi#	Not Mentioned	HbD Iran-HbD Punjab Disease→ 1	β CD 22 (G→C)+ β CD 121 (G→C)	42
53	New Delhi#	Not Mentioned	Hb Hope→1	β CD 136 (G→A) heterozygous	43
54	New Delhi#	Not Mentioned	HbD Iran trait → 25 HbD Iran homozygous →2 HbD Iran -β-thalassemia[a]→1 HbD Iran - HbD Punjab→1	β CD 22 (G→C) heterozygous β CD 22 (G→C) homozygous β CD 22 (G→C) + β-thalassemia[a] β CD 22 (G→C)+ β CD 121 (G→C)	44
55	Odisha*	Not Mentioned	HbSC Disease→1	β CD 6 (A→T) + β CD 6 (G→A)	45
56	Odisha*	Not Mentioned	Hb Hope → 1	β CD 136 (G→A) heterozygous	46
57	Odisha*	Chasa	HbS-Hb Hofu →3 Hb Hofu trait→1	β CD 6 (A→T)+ β CD 126 (T→A) β CD 126 (T→A) heterozygous	47
58	Odisha*	Not Mentioned	Hb A2 St. George-β-thalassaemia→1 HbA2 Saurashtra trait→ 1	δ CD 81 (C→T) + β IVS 1-5 (G→C) δ CD 100 (C→T) heterozygous	48
59	Odisha*	Not Mentioned	HbS-Hb Tianshui- →1	β CD 6 (A→T) + β CD39 (A→G)	49
60	Odisha*	Not Mentioned	Hb Limassole trait →1	β CD8 (G→C) heterozygous	50
61	Odisha*	Sikh	HbD Iran - β-thalassaemia→1	β CD 22 (G→C) + β CD 41/42 (-CTTT)	51
62	Puducherry*	Achari	Hb Andrew-Minneapolis homozygous→1	β CD 144 (G→T) homozygous	52
63	Punjab*	Not Mentioned	HbJ Paris-I trait→ 1	α2 or α1 CD12 (C→A) heterozygous	53
64	Punjab*	Punjabi	Hb Chandigarh trait→1	β CD 94 (A→G) heterozygous	54
65	Punjab*	Khatri	Hb Lepore Hollandia trait→1	δβ hybrid (δ through 22; β from 50) heterozygous	20
66	Punjab*	North Indian	HbD Iran - β-thalassaemia→1	β CD 22 (G→C)+ β IVS I-5 (G→C)	55
67	Punjab*	Not Mentioned	HbQ India-HbD Punjab→2	α1 CD 64 (G→C) + β CD 121 (G→C)	56
68	Punjab*	Not Mentioned	Hb Fontainebleau trait→1	α2 or α1 CD 21 (G→C) heterozygous	57
69	Punjab*	Not Mentioned	Hb Fontainebleau trait →1	α2 or α1 CD 21 (G→C) heterozygous	58
70	Punjab*	Punjabi	Hb British Columbia trait→ 1	β CD 101 (G→A) heterozygous	14
71	Punjab*	Brahmin	Hb Pyrgos trait →1	β CD 8 (G→A) heterozygous	14
72	Punjab*	Not Mentioned	Hb Fontainebleau trait→ 1	α2 or α1 CD 21 (G→C) heterozygous	59
73	Punjab*	Not Mentioned	Hb Rush trait →1	β CD 101 (G→C) heterozygous	60

(contd.)

Sr. No.	States/UTs	Communities	Novel and Rare Hb Variants	Mutations	Ref.
74	Tamil Nadu*	Not Mentioned	Hb Rajappan trait→1	α1 CD 90 (A→C) heterozygous	61
75	Tamil Nadu*	Koya Dora	Hb Koya Dora Trait → 7 Hb Rampa Trait →4 , Hb Koya Dora Homozygous →3 Hb Koya Dora + Hb Rampa → 2	α2 CD 142 (A→C) heterozygous α2 CD 95 (C→T) heterozygous α2 CD 142 (A→C) homozygous α2 CD 142 (A→C)+ α2 CD 95 (C→T)	62
76	Tamil Nadu#	Not Mentioned	Hb Lepore Hollandia - β-thalassaemia → 1 Hb Lepore Washington-Boston-β-thalassaemia→ 1 Hb Lepore Hollandia homozygous→2	δβ hybrid (δ through 22; β from 50)+ β IVS 1-5 (G→C) δβ hybrid (δ through 87; β from 116)+ β CD 30 (G→C) δβ hybrid (δ through 22; β from 50) homozygous	63
77	Tamil Nadu*	Not Mentioned	HbSE disease→1	β CD 6 (A→T)+ β CD 26 (G→A)	17
78	Tamil Nadu*	Tamil	Hb Titusville trait→1	α 2 or α 1CD 94 (G→A) heterozygous	64
79	Uttar Pradesh*	Hindu	Hb Lucknow-β-thalassaemia→ 1	β CD 8 (A→G)+ β IVS I-5 (G→C)	65
80	Uttar Pradesh*	Punjabi	HbD Iran - β-thalassaemia→1	β CD 22 (G→C)+ β 619 bp deletion	66
81	Uttar Pradesh*	Not Mentioned	Hb I Philadelphia trait→1	α2 CD 16 (A→G) heterozygous	67
82	Uttar Pradesh*	Varanasi	Hb Lepore Hollandia trait→1	δβ hybrid (δ through 22; β from 50) heterozygous	20
83	Uttar Pradesh*	Not Mentioned	Hb Lepore Hollandia-β-thalassaemia→1	δβ hybrid (δ through 22; β from 50) + β IVS I-5 (G>C)	68
84	West Bengal*	Gurkha	HbG Norfolk trait→1	α CD 57 (G→A) heterozygous	69
85	West Bengal*	Not Mentioned	Hb Sun Prairie homozygous→1	α2 CD130 (G→C) homozygous	70
86	West Bengal*	Not Mentioned	HbSE disease→ 4 HbED disease→3 HbE- Hb Lepore →1 [b]	β CD 6 (A→T)+ β CD 26 (G→A) CD 26 (G→A)+ β CD 121 (G→C) CD 26 (G→A)+ Hb Lepore [b]	17
87	West Bengal#	Not Mentioned	Hb Lepore Washington-Boston+ β-thalassaemia→1 HbE-Hb Lepore Washington-Boston→2	δβ hybrid (δ through 87; β from 116) + β IVS I-5 (G→C) β CD 26 (G→A)+ δβ hybrid (δ through 87; β from 116)	28
88	West Bengal*	Not Mentioned	Hb Vellore-β-thalassaemia→1	β CD 7 (G→C)+ β IVS I-5 (G→C)	71
89	West Bengal*	Burdwan	Hb Acharnes trait→1	β CD 53 (G→A) heterozygous	72
90	West Bengal*	Not Mentioned	Hb Hofu trait→1 Hb-Grange-Blanche-β-thalassemia[a]→2	β CD 126 (T→A) heterozygous β CD 27 (C→T)+ β-thalassemia[a]	73
91	West Bengal*	Kayastha, Jodi	HbED disease→3	β CD 26 (G→A) + β CD 121 (G→C)	5
92	West Bengal*	Brahmin	Hb Fannin- Lubbock-I trait→1	β CD 119 (G→A) heterozygous	74
93	West Bengal*	Bengali	Hb Midnapore trait →1	β CD 53 (C→T) heterozygous	75
94	West Bengal*	Not Mentioned	Hb Ty Guard Trait→1	β Codon 124 (C→A) heterozygous	76
95	West Bengal*	Not Mentioned	β-thalassaemia trait [c]→1	3'-UTR +1506 (A→C) heterozygous[c]	77

a: β-thalassaemia mutations not available; b: Hb Lepore mutations not reported; c: Novel mutation reported from West Bengal

* States/UTs of origin; # States/UTs where diagnosis has been done

There are few other rare alpha chain variants reported where molecular analysis data were not available. These include HbK Madras in an East Indian family (78), HbJ India in an Gujarati from Bombay (79), HbL in an Gujarati from Bombay (80), HbK Calcutta in an Bengali family from Calcutta (81) and HbL-Bombay in an Lohana from Bombay (82).

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HGVS Nomenclature for β-thalassaemia mutations

Common Name	HGVS Nomenclature
-28 (A→G)	HBB:c.-78A→G
-29 (A→G)	HBB:c.-79A→G
−86 (C→G)	HBB:c.-136C→G
-87(C→T)	HBB:c.-137C→T
-88(C→T)	HBB:c.-138C→T
-88 (C→A)	HBB:c.-138C→A
-90 (C→T)	HBB:c.-140C→T
5'UTR + 20 (-C)	HBB:c.-31del C
Capsite +1 (A→C)	HBB:c.-50A→C
Initiation CD (T→C)	HBB:c.2T→C
CD5 (-CT)	HBB:c.17_18delCT
CD 7/8 (+G)	HBB:c.24_25insG
CD8 (-AA)	HBB:c.25_26delAA
CD 8/9 (+G)	HBB:c.27_28insG
CD10 (C→A)	HBB:c.33C→A
CD13 (C→A)	HBB:c.41→A
CD14/15 (+G)	HBB:c.45_46insG
CD 15 (G→A)	HBB:c.48G→A
CD15 (-T)	HBB:c.46delT
CD 16 (-C)	HBB:c.51delC
CD 16 (C→T)	HBB:c.51C→T
CD 17 (A→T)	HBB:c.52A→T
CD19 (A→G) (Hb Malay)	HBB:c.59A→G
CD 22/23/24 (7 bp deletion)	HBB:c.68_74delAAGTTGG
CD26 (G→T)	HBB:c.79G→T
CD 27/28 (+C)	HBB:c.84_85insC
CD30 (G→C)	HBB:c.92G→C
CD30 (G→A)	HBB:c.92G→Aa

Common Name	HGVS Nomenclature
IVS-I-1 (G→T)	HBB:c.92+1G→T
IVS 1-1 (G→A)	HBB:c.92+1G→A
IVS 1-5 (G→T)	HBB:c.92+5G→T
IVS-I-5 (G→C)	HBB:c.92+5G→C
IVS I-5 (G→A)	HBB:c.92+5G→A
IVS 1-25 bp deletion	HBB:c.93-22_95del
IVS I-110 (G→A)	HBB:c.93-21G→A
IVS 1-128 (T→G)	HBB:c.93-3T→G
IVS 1-129 (A→C)	HBB:c.93-2A→C
IVS 1-130 (G→A)	HBB:c.93-1G→A
IVS I-130 (G→C)	HBB:c.93-1G→C
Codons 36/37 (-T)	HBB:c.112delT
CD 37 (-T)	HBB:c.112delT
CD 39 (C→T)	HBB:c.118C→T
CD 39 (-A)	HBB:c.119delA
CD 41 (-C)	HBB:c.126delC
CD 41/42 (-TCTT)	HBB:c.126_129delCTTT
CD 43 (G→T)	HBB:c.130G→T
CD44 (-C)	HBB:c.135delC
CD 47/48 (+ATCT)	HBB:c.146_147insATCT
CD 71/72 (+A)	HBB:c.216_217insA
619 bp deletion	NG_000007.3:g.71609_72227del619
IVS II-I (G→A)	HBB:c.315+1G→A
IVS II-654 (C→T)	HBB:c.316-197C→T
IVS II-745(C→G)	HBB:c.316-106C→G
IVS II-848 (C→A)	HBB:c.316-3C→A
IVS II-837 (T→G)	HBB:c.316-14T→G
CD 110 (T→C)	HBB:c.332T→C
CD 121 (G→T)	HBB:c.364G→T
CD126-131 (-17 bp deletion)	HBB:c.380_396delTGCAGGCTGCCTATCAG
Poly A (T→C)	HBB: c.*110T→C
Poly A (-AA)	HBB: c.*111_*112delAA

HGVS Nomenclature for α-thalassaemia mutations

Common Name	HGVS Nomenclature
Hb Constant Spring (Hb CS)	HBA2:c.427T→C
Hb Koya Dora	HBA2:c.428A→C
Hb Pakse (PS)	HBA2:c.429A→T
Hb Rampa	HBA2:c.286C→T (or HBA1)
Hb Sallanches	HBA2:c.314G→A
IVS 1-1 (G→A)	HBA2:c.95+1G→A
--MED	NG_000006.1:g.24664_41064del16401
Poly A(-AA)	HBB:c.*111_*112delAA
--SA	NG_000006.1:g.19464_43064del23601
--SEA	NG_000006.1:g.26264_45564del19301
α Seal Rock	HBA2:c.427T→G
α2 Codon 19 (-G)	HBA2:c.60delG
$-\alpha^{3.7}$	NG_000006.1:g.34247_38050del
$-\alpha^{4.2}$	NC_000016.10:g.169818_174075del
α Zürich-Albisrieden	HBA2:c.178G→C

HGVS Nomenclature for common and rare haemoglobin variants

Common Name	HGVS Nomenclature
Hb Variant (Common name)	HGVS Nomenclature
Hb A2 Saurashtra	HBD:c.301C→T
Hb A2 St. George	HBD:c.244C→T
Hb Acharnes	HBB:c.160G→A
Hb Agenogi	HBB:c.271G→A
Hb Alperton	HBB:c.407C→T
Hb Andrew-Minneapolis	HBB:c.435G→C
Hb Beth Israel	HBB:c.308A→G
Hb British Columbia	HBB:c.304G→A
Hb Brugg	HBA1:c.63C→A
Hb Chandigarh	HBB:c.284A→G
Hb D Agri	HBB:c.[29C→A;364G→C]
Hb D Punjab	HBB:c.364G→C
Hb E	HBB:c.79G→A
Hb Fannin- Lubbock I	HBB:c.359G→A
Hb Fontainebleau	HBA2:c.64G→C (or HBA1)
Hb G Coushata	HBB:c.68A→C
Hb Godavari	HBA2:c.286C→A (or HBA1)
Hb Hofu	HBB:c.380T→A
Hb Hope	HBB:c.410G→A
Hb I Philadelphia	HBA2:c.49A→G
Hb Jackson	HBA2:c.[384G→T (or HBA1) or 384G>C (or HBA1)]
Hb Koln	HBB:c.295G→A
Hb Koya Dora	HBA2:c.428A→C
Hb Le Lamentin	HBA2:c.63C→A
Hb Lepore Washington-Boston	NG_000007.3:g.63632_71046del
Hb Lepore-Hollandia	NG_000007.3:g.63290_70702del
Hb Limassole	HBB:c.27G→C
Hb Lucknow	HBB:c.26A→G
Hb M Iwate	HBA2:c.262C→T (or HBA1)
Hb M Saskatoon	HBB:c.190C→T

Common Name	HGVS Nomenclature
Hb Midnapore	HBB:c.161C→T
Hb Olympia	HBB:c.61G→A
Hb Pelendri	HBD:c.425T→C
Hb Pyrgos	HBB:c.251G→A
Hb Rajappen	HBA1:c.272A→C
Hb Rush	HBB:c.304G→C
Hb Sherwood Forest	HBB:c.314G→C
Hb St. Louis	HBB:c.86T→A
Hb Sun Prairie	HBA2:c.391G→C
Hb Tianhe	HBD: c.323G→A
Hb Titusville	HBA2:c.283G→A (or HBA1)
Hb Ty Guard	HBB:c.374C→A
Hb Vellore	HBB:c.22G→C
CD 6 (G→A) [HbC]	HBB:c.19G→A
HbDIran	HBB:c.67G→C
CD 121 (G→C) [HbD Punjab]	HBB:c.364G→C
CD26 (G→A) [HbE]	HBB:c.79G→A
HbG Norfolk	HBA2:c.256G→A (or HBA1)
HbG -Waimanalo	HBA2:c.193G→A
Hb-Grange-Blanche	HBB:c.83C→T
HbJ Cambridge	HBB:c.209G→A
HbJ Meerut	HBA2:c.362C→A (or HBA1)
HbJ Mizunami	HBB:c.251G→A
HbJ Norfolk	HBA2:c.173G→A (or HBA1)
HbJ Paris-I	HBA2:c.38C→A (or HBA1)
HbM Boston	HBA2:c.175C→T (or HBA1)
HbM Hyde Park	HBB:c.277C→T
HbM Ratnagiri	HBB:c.63C→T
HbO Indonesia	HBA1:c.349G→A
HbQ India	HBA1:c.193G→C
CD 6 (A→T) (HbS)	HBB:c.20A→T
HbYaizu	HBB:c.238G→A



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13th Floor, New Multistoreyed Building, K.E.M. Hospital Campus,
Parel, Mumbai - 400012 (INDIA). Web: www.niih.org.in