

HAEMOGLOBINOPATHIES IN THE INDIAN SUBCONTINENT

A Review of Literature

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SUMMARY

An up-to-date review of literature on the distribution of haemoglobinopathies in the Indian subcontinent has been presented. Haemoglobins S, D, E, J, K, L, M, Q, β -thalassaemia major, β -thalassaemia minor, α -thalassaemia minor, haemoglobin H, haemoglobin Lepore and persistence of foetal haemoglobin have been reported from India. β -thalassaemia in combination with haemoglobins S, E, D, J, K, Q has also been reported. Haemoglobin S is found to be prevalent among tribal populations in various regions of India, while haemoglobin D is prevalent among Sikhs, Pathans and Gujratis. Haemoglobin E is present in high frequency among populations of Assam, Bengal and Veddahs of Ceylon. β -thalassaemia major is widespread in various populations. There is paucity of data regarding distribution of β -thalassaemia minor and α -thalassaemias in this region.

The Indian subcontinent is a vast reservoir of abnormal haemoglobins and thalassaemias. Several of the abnormal haemoglobins were first detected in persons of Indian origin, e.g., Hbs D, J, K, and L, and prevalence of abnormal haemoglobins is widespread in many of the racial and tribal groups of India over wide areas. However, the only available reviews on the distribution of haemoglobinopathies in the Indian subcontinent is that of Chatterjea (1966) and of Livingstone (1967) as part of a study of the world distribution of abnormal haemoglobins. Since then, many reports have appeared in the literature and because of the medical and scientific significance of haemoglobinopathies it is desirable to present an up-to-date review of the subject.

The results of all the published surveys have been presented in tabular form according to geographical areas arranged in alphabetical order. The accompanying map shows the different States of the Indian subcontinent with capital cities (shown by solid square) and other relevant places (shown by solid circle). Isolated case reports and the most important points of the table have been described in the text under the headings for appropriate haemoglobins.

Haemoglobin S

The first case of sickle-cell anaemia in an Indian was reported from Capetown by Berk and Bull (1943). The patient was an anaemic married woman aged 22 years, born of Indian parents in Durban. The first cases in India itself were reported by Dunlop and Mozumder

(1952) who found 5 cases of sickle-cell trait and 3 presumptive cases of sickle-cell anaemia among tea garden labourers of upper Assam, originating from the tribal population of Orissa and Bihar.

In the same year the presence of sickle-cell trait among the aboriginal tribes (the Pre-Dravidians) of the Nilgiri Hills was reported by Lehmann and Cutbush (1952). After that there have been several surveys in the Nilgiris by Lehmann's group and others and in other areas. The details of these surveys have been presented in the table.

High frequencies of the sickle-cell gene have been found in tribal population of the Nilgiris, and in several tribes of Gujrat, Madhya Pradesh, and Orissa. There are wide variations of the frequencies in different tribes in the same locality. Foy et al. (1956) reported 55% sickle-cell positive among Parja Kondhs and 3% among Jijmor Kondhs in South India. The details have not been published.

In contrast to the tribal populations the sickle-cell gene is almost absent in the different caste groups of India over the wide areas screened: there are in general only sporadic instances of sickle-cell anaemia in the caste populations which may be due to occasional admixture of tribal blood. Sur et al. (1968) recorded 65 cases of sickle-cell haemoglobinopathy among children at Nagpur between 1962 and 1966: the highest incidence was among Neo-Buddhists. Several other sporadic instances of sickle-cell gene have been reported from all over the sub-continent (Shukla and Parande 1956, Nail et al. 1957, Shukla et al. 1958, Khandelwal and Paithankar 1961, Beohar et al. 1963, Salgia et al. 1965, Reddy and Baruah 1966, Reddy et al. 1966, Nanda et al. 1967, Subbarao et al. 1968, Praharaj et al. 1969, Rao et al. 1969, Guha and Bhattacharjee 1971). However, there are a few pockets of high frequencies of the sickle-cell gene reported among the caste groups: Sorathis (30%) in Gujrat, Danukhs (10.5%) in Uttar Pradesh, and Mahars (20%) in Madhya Pradesh, although the Mahars of Bombay have a much lower frequency of the gene (2%).

From the above it is clear the sickle-cell gene is widespread among many of the tribal populations of India, and is not unknown among the caste-groups of Indians. Further work is needed to have more systematic data for the different caste groups from all parts of the country.

Haemoglobin D

Bird et al. (1955) recorded the first case of Hb D in a 19-year-old Sikh soldier from the Hosiarpur district of East Punjab. Since then several surveys have been carried out, and Sikhs and Punjabi Hindus have been found to have Hb D with a frequency between 1 and 2%. In addition Hb D has been reported among Lohanas of Gujrat (1%), Gujratis (< 1%), Muslims of Bombay (0.2%), and among Indians in Goa and Due (very low). Pathans have been reported to have 1.4% of Hb D. Brahuís and Baluchis of N.W. Pakistan also have been reported to have D haemoglobin. Ghai et al. (1961) recorded D haemoglobin in a Punjabi family and Lele et al. (1962) recorded one case in a Kunbi family in Aurangabad. Sporadic cases have been reported among Brahmins and Vaidyas of Bengal, Tamil Muslims, and from unspecified population groups in Mysore and Uttar Pradesh.

Haemoglobin E

Hb E was first discovered simultaneously in Thais by Chernoff et al. and in a Guatemalan by Itano et al. in 1954. Hb E is widespread in S.E. Asian regions, with highest incidence in Thailand. High frequency of this haemoglobin is found in the eastern wing of India: Ahoms, Khasis, Assamese, and Totos, have frequencies ranging from 58 to 20%. Bhutanese have a frequency of 6.5%. Hb E is present among Bengalee Hindus (3%) and Bengalee Muslims (4%). It seems that there is a lower frequency of Hb E among Bengalee Brahmins compared to Kayasthas. Veddahs of Ceylon have also a high incidence of Hb E (17%). Sporadic instances of Hb E have been reported from Uttar Pradesh, and among Tamils and Malayalis of Singapore and Malaya. Cases of Hb E have also been reported among Sinhalese of Ceylon (Nagaratnam et al. 1958, De Silva et al. 1959) and in Bombay (Udani et al. 1963). Punt and Goel (1957) recorded two instances of Hb E in Indo-Europeans.

It appears from the above that Hb E is more prevalent among people of Eastern India, although it is not unknown in other areas. More systematic study is necessary of the distribution of Hb E in South India.

Haemoglobin J

Raper (1957) reported the first case of Hb J in a Gujrati woman in a survey of 500 Gujrati residents in Uganda. Sanghvi et al. (1958) recorded two cases of Hb J in 2 unrelated women belonging to the Gujrati-speaking Lohana community in Bombay, one of which was associated with thalassaemia trait. Vella (1962a) reported one instance of Hb J in a Sikh from a survey of 3341 Indians in Singapore. Subhedar et al. (1961) reported one case of Hb J in a Harijan family from Nagpur. Swarup et al. (1966b) reported Hb J in a Bengalee family in which there were two more instances of Hb J.

Haemoglobin K

Ager and Lehmann (1957a) reported the first case of Hb K in two unrelated East Indians in London. Vella (1962a) reported 5 cases of Hb K among 3341 Indians surveyed in Singapore. Labie et al. (1961) recorded 3 cases of Hb K among 114 Hindus of lower caste and another of unknown identity in Pondicherry. De Traverse et al. (1963) reported 3 instances of Hb K among 101 South Indians in Madras. Trincão et al. (1963) reported 2 instances of Hb K in a survey of 1843 Indians in Goa.

Haemoglobin L

Haemoglobin L was first reported by Ager and Lehmann (1957b) in a Punjabi Hindu of Kshatri caste resident in London. Vella (1962a) reported 2 instances of Hb L from Singapore, one in a Sindhi and the other in a Sikh. Sukumaran et al. (1959) reported 8 instances of Hb L in three Gujrati-speaking Lohana families in Bombay.

Haemoglobin M

Only one family with haemoglobin M has so far been detected, in a Punjabi family from Amritsar (Chatterjea 1966). Three members of the family were found to have Hb M levels of 7%, 33%, and 50%.

Haemoglobin Q

Trincão et al. (1963) reported 4 instances of Hb Q in a survey of 1843 Indians in Goa. Sukumaran et al. (1971) recorded a new Hb Q²⁶⁴ (aspartic acid→histidine), or Hb Q (India), in two Sindhi families in Bombay.

THALASSAEMIA SYNDROMES

The earliest case of thalassaemia in India was reported by M. Mukherji (1938) in a two-and-a-half-year-old Bengalee boy from Calcutta. Due to lack of diagnostic facilities it was not possible to know whether the boy was suffering from thalassaemia major or E-thalassaemia. Since then until the early sixties, the literature on thalassaemia in India contained only a few scattered case reports of thalassaemia from various parts (Coelho 1939, Napier et al. 1939, Patel and Bhende 1939, Dhayagude 1944, Malhotra and Chhuttani 1944, K.C. Chaudhuri 1947, Chanda and Chaudhuri 1950, Pirzada and Kapoor 1950, De Silva and Weeratungee 1951, Tiagi et al. 1954, Ganguli and Lahiri 1955, Coelho et al. 1958, Das Gupta et al. 1958, Narayanappa 1963, and Srinivasan et al. 1966).

 β -THALASSAEMIAS

β -thalassaemia major. This condition is caused by the homozygous state of a mutant gene resulting in suppression of β -chain synthesis and is present throughout the country. Chatterjea (1966) recorded 190 cases of thalassaemia major among Hindus, consisting of 175 Bengalees, 5 Biharis, 6 Punjabis, 2 each of Oriahs and Sindhis, and also in 3 Bengalee Muslims. Sanghvi et al. (quoted by Chatterjea 1966) recorded 128 cases of thalassaemia major at the Tata Cancer Institute, Bombay. From the J.J. Group of Hospitals in Bombay 157 cases of thalassaemia major have been reported also (Chatterjea 1966). Sharma et al. (1963) recorded a further 80 cases of thalassaemia major in Bombay and the regional distribution was as follows: Gujratis 35, Maharashtrians 21, Sindhis 12, Goanese 9, Bengalee 1, Uttar Pradesh 1. Udani et al. (1961) reported 14 cases of thalassaemia among Lohanas. Mathur et al. (1962) and Atal and Mittal (quoted by Chatterjea 1966) reported cases of thalassaemia major in the local populations and among Punjabis and Sindhis in Uttar Pradesh. Mariswamy and Pierce (1959) reported 5 cases of thalassaemia major from Mysore. Weatherall and Vella (1960) reported one case of thalassaemia in a Gurkha family in Singapore.

β -thalassaemia minor. This is caused by the heterozygous state of a mutant gene resulting in suppression of β -chain synthesis and has been reported among Bengalees (26%), Sherpas

(1%), Cutchhi Bhanushalis (44%), Sikhs (6%), higher and lower caste Hindus of Pondicherry (20% and 14%), Indians in Khartoum (32%). Flatz et al. (1972) recently reported the presence of this trait among Assamese (5%), Ahoms (1%), and Khasis (< 1%) in Assam.

α -THALASSAEMIAS

α -thalassaemia minor. This is caused by the heterozygous state of a mutant gene resulting in suppression of synthesis of α -chains. This is expressed as increased Hb Bart's in cord blood. Hb-Bart's has been reported to be present among Bengalees (4%), Maharashtrians (1%) (Chouhan et al. 1969), Indians in Singapore and Malaya (1-2%).

Haemoglobin H. This is probably produced by the interaction of two α -thalassaemia genes (WHO 1972). Brain and Vella (1958) reported one case of Hb H trait in a Nepalese woman in Singapore. Hb H was reported from India by Swarup et al. (1963) in a 19-year-old Bengalee Hindu. Saha and Banerjee (1971) reported two cases of Hb H trait among Malayalis, and one each among Tamils, Gujratis, and Sindhis in Singapore. Nagaratnam and Sukumaran (1967) reported one case of Hb H from Ceylon.

THALASSAEMIA WITH OTHER ABNORMAL HAEMOGLOBINS

S-thalassaemia. Chatterjea (1966) reported 15 cases of S-thalassaemia, 8 in Oriah Hindus, 1 each in Bengalee Hindu and Muslim, and 1 in a South Indian Hindu and 2 in Tamil Muslims. Sanghvi et al. and Parekh respectively recorded 16 and 6 cases of S-thalassaemia in Bombay (quoted by Chatterjea 1966). Mital et al. (1962) reported a high incidence of S-thalassaemia among Sorathis in Palghar (3.7%). Lele et al. (1962) recorded one family of S-thalassaemia in a survey of 100 students of scheduled caste in Aurangadabad.

E-thalassaemia. Chatterjea (1966) reported 526 cases of E-thalassaemia investigated in Calcutta among Indian Hindus and the regional distribution was as follows: Bengalees 508, Oriahs 10, Biharis 4, Assamese 2, Punjabi 1, South Indian 1, and 48 cases among Bengalee Muslims and 1 in a Bihari Muslim. Khaleque (1961) reported one family with E-thalassaemia in Bangladesh. Sarkar et al. (1959) reported 14 cases of E-thalassaemia from Calcutta. Sanghvi et al. recorded 5 instances of E-thalassaemia in Bombay (quoted by Chatterjea 1966). Occasional cases have been reported from Uttar Pradesh. Kochhar and Kathpalia (1963) and Praharaj et al. (1969) reported solitary instances of E-thalassaemia in a Canarese and an Oriah family.

D-thalassaemia. Chatterjea (1966) reported 9 cases of D-thalassaemia - 6 from Bengal, and 1 each from Bihar, Punjab, and South India. Sanghvi et al. recorded 7 cases of D-thalassaemia in Bombay (quoted by Chatterjea 1966). Occasional cases of D-thalassaemia have been reported in and around Delhi (Ghai et al. 1961). Lele et al. (1962) reported one case in a Kunbi family from Aurangadabad. Sukumaran et al. (1960) reported one case each in a Sindhi and Gujrati-Lohana family.

J-thalassaemia. Sanghvi et al. (1958) recorded one case of J-thalassaemia in a Gujrati-speaking Lohana. Swarup et al. (1966b) reported 4 cases of J-thalassaemia in Bengalee Hindus.

K-thalassaemia. Swarup et al. (1966a) reported an interaction of Hbs E and K with thalassaemia in a Bengalee family of Calcutta.

Q-thalassaemia. Sukumaran et al. (1972) recorded one case of Q-thalassaemia major and 2 cases of Q-thalassaemia minor in Sindhi families in Bombay.

Hereditary Persistence of Foetal Haemoglobin

Sukumaran et al. (1961) recorded 13 instances of this variant in heterozygous form, out of which in 3 they found double heterozygous state, i.e., Hb F and thalassaemia major. Similar interaction was observed in two Bengalee families by Chatterjea (1966) and by Barkhan and Adinolfi (1962) in a family of mixed Indian and Portuguese ancestry in London, by Parekh et al. (1963) in Indian families in Bombay, and by Bird et al. (1964) in an Indian boy at Poona.

Haemoglobin Lepore

Chouhan et al. (1971) reported the only case of Hb Lepore in an Indian family from Coondapur of Mysore state. Out of 23 members of the family, 10 had between 7.9% and 14.2% of this haemoglobin.

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FIGURE. *The Indian Subcontinent [showing different states with state capitals (■) and other relevant places (●)].*

TABLE
DISTRIBUTION OF SICKLING TRAIT AND ABNORMAL HAEMOGLOBINS IN THE INDIAN SUBCONTINENT

| Reference | Population | Locality | No. tested | No. with positive sickling or Hb-S | No. with other abnormal haemoglobins ^a |
|--|------------------------|-------------------------|------------|------------------------------------|---|
| ANDHRA PRADESH | | | | | |
| Lehmann & Cutbush 1952 | Telegus | Unspecified | 109 | 0 | — |
| Chaudhuri et al. 1964 <i>a</i> | Chenchus | Unspecified | 9 | 0 | 0 |
| ASSAM, NEPAL, TIBET | | | | | |
| Chaudhuri et al. 1964 <i>a</i> | Totos | Totopara, Assam | 116 | 0 | AE-23 |
| Pande et al. 1970 | Assamese | Poona | 8 | 0 | AE-1, EE-1 |
| Das B.M. et al. 1971 | Khasis | Assam | 80 | 0 | AE-31, EE-2 |
| | Ahoms | Assam | 82 | 0 | AE-37, EE-11 |
| Flatz et al. 1972 | Assamese | Assam | 182 | 0 | AE-29, EE-5, Thal. minor-10 |
| | Ahoms | Assam | 129 | 0 | AE-60, EE-15, Thal. minor-2 |
| | Khasis | Assam | 140 | 0 | AE-51, EE-6, Thal. minor-4 |
| | Kacharis | Assam | 5 | 0 | AE-4, EE-1 |
| Aksoy et al. 1955 | Nepalese | Poona | 199 | 0 | 0 |
| Bird et al. 1957 | Gurkhas | Nepal | 200 | 0 | 0 |
| Vella & Field 1958 | Gurkhas | Singapore | 560 | 0 | AE-3 |
| Chatterjea 1959 | Nepalese | Calcutta | 109 | 0 | AE-1 |
| Lehmann 1959 | Nepalese | Unspecified | 99 | 0 | 0 |
| Nijenhuis 1963 | Nepalese | Pokhara, Nepal | 14 | 0 | 0 |
| | Tibetans | Nepal | 47 | 0 | 0 |
| Swarup et al. 1966 <i>c</i> | Nepalese | Calcutta | 134 | 0 | High A ₂ -40 |
| Glasgow et al. 1968 | Bhutanese | Luana & Thimphu, Bhutan | 31 | 0 | AE-2 |
| Jackson et al. 1960 | Sherpas | Khembu, Tibet | 128 | 0 | AE-1, Thal. minor-1 |
| BENGAL (WEST BENGAL & BANGLADESH) | | | | | |
| Buchi 1955 <i>a</i> | Indians | Calcutta | 400 | 0 | — |
| Chatterjea et al. 1957 | Bengalees | Calcutta | 700 | 0 | AE-25, EE-2, Thal. minor-26 |
| Swarup et al. 1965 | Bengalees (cord blood) | Calcutta | 100 | 0 | Bart's-4 |
| Chaudhuri et al. 1969 | Brahmins | Calcutta | 235 | 0 | 0 |
| | Kayasthas | Calcutta | 229 | 0 | AE-6 |
| | Vaidyas | Calcutta | 129 | 0 | AE-1 |
| Bhattacharjee 1956 | Rahri Brahmins | Hooghly | 100 | 0 | — |
| | Muslims | Hooghly | 100 | 0 | — |

TABLE - continued

| Reference | Population | Locality | No. tested | No. with positive sickling or Hb-S | No. with other abnormal haemoglobins ^a |
|------------------------------------|------------------|------------------------------------|------------|------------------------------------|---|
| Kumar 1957 | Duley Bagdis | Hooghly | 69 | 0 | — |
| | Tentulia Bagdis | Hooghly | 89 | 0 | — |
| Chaudhuri et al. 1964 ^b | Santals | Midnapore | 119 | 2 (AS) | 0 |
| | | | families | | |
| Chaudhuri et al. 1967 ^a | Santals | Midnapore | 336 | 4 (AS) | 0 |
| Das et al. 1967 | Rajbanshis | Midnapore, Jalpaiguri, Cooch Bihar | 300 | 0 | — |
| | Padmaraj (Pods) | 24-Parganas | 100 | 0 | — |
| | Mahishyas | 24-Parganas | 60 | 0 | — |
| Chaudhuri et al. 1963 | Anglo-Indians | Kharagpur | 89 | 0 | — |
| Pande et al. 1970 | Bengalees | Poona | 65 | 0 | AE-1 |
| Swarup et al. 1958 ^b | Bengalee Muslims | Bangladesh | 424 | 1 | AE-25, E-thal.-50, S-thal.-1 |
| Lehmann et al. 1961 | Bengalee Muslims | Karachi | 23 | 0 | AE-2 |
| Sen 1960 | Brahmins | U.K. | 188 | 0 | AE-1, EE-1, AD-1, E-thal.-1 |
| | Kayasthas | U.K. | 269 | 0 | AE-7 |
| | Vaidyas | U.K. | 88 | 0 | AD-1 |
| | Miscellaneous | U.K. | 57 | 0 | AE-2 |
| Pande et al. 1972 | Bengalees | Unspecified | 41 | 0 | AE-1 |
| BIHAR | | | | | |
| Lehmann 1954 | Oraons | Bihar | 100 | 0 | — |
| | Kharias | Bihar | 23 | 0 | — |
| Chatterjea 1959 | Biharis | Calcutta | 54 | 0 | 0 |
| Kirk et al. 1962 ^a | Oraons | Chota Nagpur | 56 | 0 | 0 |
| Pande et al. 1970 | Biharis | Poona | 57 | 0 | 0 |
| Pande et al. 1972 | Biharis | Unspecified | 25 | 0 | 0 |
| GUJRAT | | | | | |
| Sukumaran et al. 1956 | Anavil Brahmins | Surat | 53 | 0 | — |
| | Leva Patidars | Kheda | 150 | 0 | — |
| Mital et al. 1962 | Sorathis | Palghar | 325 | 99 (AS-81, SS-6) | S-thal.-12 |
| Sanghvi 1962 | Brahmins | Gujrat | 203 | 0 | 0 |
| | Lohanas | Gujrat | 603 | 0 | AD-5, AJ-2, AL-3 |
| Vyas et al. 1962 | Bhils | Panchmahal | 206 | 32 (AS) | 0 |
| | Gamils | Surat | 207 | 65 (AS-26 ^c , SS-2) | 0 |

TABLE - *continued*

| Reference | Population | Locality | No. tested | No. with positive sickling or Hb-S | No. with other abnormal haemoglobins ^a |
|------------------------------------|----------------------|-----------------------|--------------------|------------------------------------|---|
| Vyas et al. 1962 (<i>contd.</i>) | Dublas | Surat | 211 | 20 (AS) | 0 |
| | Kolis | Surat | 182 | 8 (AS) | 0 |
| | Naikas | Surat | 174 | 28 (AS-26 ^c) | 0 |
| | Dhodias | Surat | 213 | 38 (AS) | 0 |
| | Dhankas | Broach | 215 | 44 (AS-41, SS-3) | 0 |
| Sayed & Amin 1966 | Bhils | Baroda | 220 | 38 | — |
| Jacob et al. 1956 | Gujratis | Uganda | 326 | 0 | AD-1 |
| Raper 1957 | Gujratis | Uganda | 500 | 0 | AD-4, AJ-1 |
| KERALA | | | | | |
| Lehmann & Cutbush 1952 | Malayalis | Unspecified | 110 | 0 | — |
| Bird et al. 1962 | Malayalis | Poona | 190 | 0 | 0 |
| Chaudhuri et al. 1964a | Indians | Kerala | 35 | 0 | — |
| Buchi 1955c | Malapandarams | Quilon | 116 | 0 | — |
| | Kurubans | Quilon | 36 | 0 | — |
| | Ullatans | Quilon | 142 | 0 | — |
| | Malavedans | Quilon | 69 | 0 | — |
| | Kadars | Trichur | 167 | 0 | — |
| | Saha & Banerjee 1971 | Malayalis | Kerala & Singapore | 314 | 0 |
| MADHYA PRADESH | | | | | |
| Negi 1962 | Dorlas | Konta (Bastar) | 200 | 26 | — |
| | Dhurwas | Jagdalpur (Bastar) | 218 | 7 | — |
| | Northern Dhurwas | Jagdalpur | 60 | 10 | — |
| Negi 1963 | Bade Bhattras | Jagdalpur | 153 | 25 | — |
| | Manjhela Bhattras | Jagdalpur | 64 | 7 | — |
| | San Bhattras | Jagdalpur | 88 | 17 | — |
| | Mahars | Jagdalpur | 123 | 47 | — |
| | Eastern Murias | Kondagaon (Bastar) | 143 | 15 | — |
| | Western Murias | Kondagaon | 169 | 27 | — |
| Negi 1964 | Marias | Bijapur | 185 | 29 | — |
| | (Bison-horned) | (Bastar) | | | |
| | Raj Gonds | Bastar | 68 | 8 | — |
| | Dorlas | Bastar | 27 | 3 | — |
| | Halbas | Bastar | 34 | 9 | — |
| | Mahars | Bhopalpatnam (Bastar) | 30 | 12 | — |
| | Telangas (Telegu) | Bastar | 19 | 0 | — |
| | Others | Bastar | 34 | 3 | — |

TABLE - *continued*

| Reference | Population | Locality | No. tested | No. with positive sickling or Hb-S | No. with other abnormal haemoglobins ^a |
|-----------------------|------------------|----------------|------------|------------------------------------|---|
| Kumar 1966 | Bhils | Indore | 46 | 11 | — |
| | Balais | Indore | 73 | 4 | — |
| | Chamars | Indore | 22 | 2 | — |
| | Others | Indore | 95 | 0 | — |
| Das et al. 1967 | Balais | Unspecified | 104 | 5 | — |
| | Bhilalas | Unspecified | 139 | 39 | — |
| | Bhils | Unspecified | 174 | 21 | — |
| Kumar & Ghosh 1967 | Mehtars | Ujjain | 72 | 1 | — |
| | Brahmins | Ujjain | 28 | 0 | — |
| | Rajputs | Ujjain | 42 | 0 | — |
| | Jains & Vaishyas | Ujjain | 15 | 0 | — |
| | Dhakars | Dewas | 25 | 0 | — |
| | Nayata Muslims | Ujjain & Dewas | 65 | 1 | — |
| | Chamars | Ujjain & Dewas | 27 | 0 | — |
| | Muslims | Ujjain & Dewas | 12 | 0 | — |
| | Balais | Ujjain & Dewas | 10 | 0 | — |
| | Others | Ujjain & Dewas | 17 | 0 | — |
| Sharma 1968 | Raj Gonds | Bastar | 54 | 15 | — |
| | Murias | Bastar | 35 | 10 | — |
| | Bhattras | Bastar | 25 | 7 | — |
| | Halbas | Bastar | 26 | 6 | — |
| Pande et al. 1970 | Indians | Poona | 67 | 1 (AS) ^d | 0 |
| Pande et al. 1972 | Indians | Unspecified | 34 | 0 | AD-1 |
| MAHARASHTRA | | | | | |
| Shukla & Solanki 1958 | Mahars | Nagpur | 450 | 100 (AS) ^e | — |
| | Kunbis | Nagpur | 116 | 11 (AS) ^e | — |
| | Telis | Nagpur | 89 | 9 (AS) ^e | — |
| | Koshtis | Nagpur | 46 | 0 | — |
| | Ghonds | Nagpur | 53 | 0 | — |
| | Muslims | Nagpur | 68 | 0 | — |
| | Brahmins | Nagpur | 26 | 0 | — |
| | Others | Nagpur | 171 | 0 | — |
| | Das et al. 1961 | Mahars | Nagpur | 482 | 87 |
| Lele et al. 1962 | Scheduled caste | Aurangabad | 700 | 39 (AS-36, SS-2) | S-thal.-1 |
| Sanghvi 1962 | Marathas | Maharashtra | 201 | 0 | 0 |
| | Gurjars | Maharashtra | 203 | 0 | 0 |
| | Pajnas | Maharashtra | 200 | 0 | 0 |
| | Chamars | Maharashtra | 208 | 0 | 0 |
| | Mahars | Maharashtra | 200 | 4 (AS) | 0 |
| | Mixed | Maharashtra | 222 | 0 | 0 |

TABLE - *continued*

| Reference | Population | Locality | No. tested | No. with positive sickling or Hb-S | No. with other abnormal haemoglobins ^a |
|--------------------------------|--------------------------|--------------|------------|------------------------------------|---|
| Sanghvi 1962 (<i>contd.</i>) | Thakurs | Maharashtra | 264 | 0 | 0 |
| | Koknas | Maharashtra | 190 | 8 (AS) | 0 |
| | Katkaris | Maharashtra | 262 | 21 (AS) | 0 |
| | Warlis | Maharashtra | 225 | 36 (AS) | 0 |
| Deshmukh 1968 | Mahars | Aurangadabad | 100 | 24 (AS) | 0 |
| Parikh et al. 1969 <i>b</i> | Mahars | Bombay | 200 | 0 | 0 |
| Parikh et al. 1969 <i>a</i> | Audich Brahmins | Bombay | 200 | 0 | 0 |
| | Lad Vanias | Bombay | 200 | 0 | 0 |
| Undevia 1969 | Visa Oswal Jains | Bombay | 200 | 0 | 0 |
| | Parsis | Bombay | 1290 | 5 (AS) | 0 |
| | Iranis | Bombay | 160 | 0 | 0 |
| Baxi et al. 1970 | Scavengers | Bombay | 140 | 0 | 0 |
| Pande et al. 1970 | Maharashtrians | Poona | 355 | 5 (AS) ^f | 0 |
| Hakim et al. 1972 | Muslims | Bombay | 854 | 0 | AD-2 |
| Mehta et al. 1972 | Cutchhi Bhanushali | Bombay | 599 | 9 | High A ₂ -44 |
| Pande et al. 1972 | Indians | Unspecified | 34 | 0 | 0 |
| MYSORE | | | | | |
| Lehmann & Cutbush 1952 | Canarese | Unspecified | 95 | 0 | — |
| Swarup et al. 1959 | Indians | Shimoga | 550 | 1 | 0 |
| | | Unspecified | 68 | 1 (AS) | AD-1 |
| ORISSA | | | | | |
| Batabyal & Wilson 1958 | Oriahs | Assam | 100 | 15 | — |
| | Griza Oriahs | Assam | 100 | 29 | — |
| Das et al. 1967 | Bado Gadabas | Koraput | 99 | 0 | — |
| | Pareng Gadabas | Koraput | 225 | 28 | — |
| | Ollaro Gadabas | Koraput | 225 | 6 | — |
| | Bareng Porojas | Koraput | 104 | 0 | — |
| | Konda Porojas | Koraput | 225 | 30 | — |
| | Roy & Roy Chaudhuri 1967 | Tribals | Koraput | 770 | 86 |
| | Tribals | Umerkote | 630 | 64 | — |
| | Tribals | Kondaga | 73 | 5 | — |
| PUNJAB, HARYANA & KASHMIR | | | | | |
| Aksoy et al. 1955 | Sikhs | Poona | 100 | 0 | 0 |
| Siddoo et al. 1956 | Sikhs | Vancouver | 80 | 0 | Thal.minor-5 |
| Bird et al. 1956 | Sikhs | N.W.India | 279 | 0 | AD-4, DD-1 |
| Bird & Lehmann 1956 | Hindus | N.W. India | 13 | 0 | AD-1 |
| Saha & Banerjee 1965 | Sikhs | Ludhiana | 100 | 0 | AD-1, Thal. major-1 |
| Chaudhuri et al. 1967 <i>b</i> | Sikhs | Calcutta | 427 | 0 | AD-5, AE-1 |

TABLE - continued

| Reference | Population | Locality | No. tested | No. with positive sickling or Hb-S | No. with other abnormal haemoglobins ^a |
|-----------------------------|-------------------------|--------------------|----------------|------------------------------------|---|
| Pande et al. 1970 | Sikhs | Poona | 117 | 0 | 0 |
| | Hindus | Poona | 50 | 0 | 0 |
| | Others | Poona | 70 | 0 | 0 |
| Saha & Banerjee 1971 | Jats | Poona | 41 | 0 | 0 |
| | Sikhs | Punjab, | 378 | 0 | AD-5, DD-1 |
| Mathur et al. 1962 | Hindus | Singapore | 123 | 0 | AD-1 |
| | Punjabis | Agra | 11 | 0 | Thal.major-2 |
| Pande et al. 1972 | Punjabis & Kashmiris | Unspecified | 66 | 0 | AD-1 |
| TAMILNADU | | | | | |
| Lehmann & Cutbush 1952 | Tamils | Unspecified | 128 | 0 | — |
| Lalie et al. 1961 | Higher caste | Pondicherry | 28 | 0 | High A ₂ -5 |
| | Lower caste | Pondicherry | 114 | 0 | High A ₂ -16, AK-3 |
| | Untouchables | Pondicherry | 11 | 0 | 0 |
| | Nairs | Pondicherry | 3 | 0 | 0 |
| | Kharwars | Pondicherry | 1 | 0 | 0 |
| | Muslims | Pondicherry | 1 | 0 | 0 |
| | Unknown | Pondicherry | 12 | 0 | High A ₂ -2, AK-1 |
| | De Traverse et al. 1963 | Indians | Madras city | 101 | 0 |
| Wickremasinghe et al. 1963b | Tamils | Ceylon | 660 | 0 | 0 |
| | Muslims | Ceylon | 252 | 0 | 0 |
| Saha & Banerjee 1971 | Tamils | Kerala & Singapore | 1310 | 0 | AE-6, EE-2, AH-2, AD-1 |
| Lehmann 1954 | Paniyans | Nilgiri | 61 | 21 | — |
| | Kurumbas | Nilgiri | 16 | 3 | — |
| | Kotas | Nilgiri | 86 | 0 | — |
| | Irulas | Nilgiri | 124 | 39 | — |
| | Badagas | Nilgiri | 191 | 26 | — |
| | Todas | Nilgiri | 84 | 3 | — |
| | Buchi 1955a | Paniyans | Wynad, Nilgiri | 74 | 22 |
| Buchi 1955b | Pallars | Tinnevelly | 112 | 1 | — |
| Lehmann & Sukumaran 1956 | Kurumbas | Nilgiri | 26 | 7 (AS) | 0 |
| | Kotas | Nilgiri | 22 | 0 | 0 |
| | Badagas | Nilgiri | 30 | 2 (AS) | 0 |
| | Todas | Nilgiri | 50 | 1 (AS) | 0 |
| | Irulas | Nilgiri | 18 | 4 (AS) | 0 |
| | Buchi 1959 | Kurumbas | Nilgiri | 112 | 20 |
| Kirk et al. 1962b | Kurumbas | Nilgiri | 43 | 10 (AS) | 0 |
| | Irulas | Nilgiri | 15 | 6 (AS) | 0 |

TABLE - *Continued*

| Reference | Population | Locality | No. tested | No. with positive sickling or Hb-S | No. with other abnormal haemoglobins ^a |
|---|---------------|-------------------------------|------------|------------------------------------|---|
| Kirk et al. 1962 <i>b</i> (<i>contd.</i>) | Todas | Nilgiri | 60 | 2 (AS) | 0 |
| Das et al. 1967 | Paniyans | Wynad, Nilgiri | 955 | 265 | — |
| Chaudhuri et al. 1964 <i>a</i> | Kotas | Nilgiri | 12 | 0 | — |
| | Todas | Nilgiri | 12 | 0 | — |
| UTTAR PRADESH | | | | | |
| Bhatia et al. 1955 | Dhanukh caste | Mainpur | 335 | 32 | — |
| Dube et al. 1959 | Indians | Lucknow | 235 | 0 | 0 |
| Misra 1961 | Indians | Agra | 250 | 0 | 0 |
| Mathur et al. 1962 | Indians | Agra | 410 | 0 | AE-1, Thal.major-6, Thal.minor-1 |
| | | | (anaemic) | | |
| | Indians | Western U.P. | 102 | 0 | 0 |
| Atal. & Mital 1966 ^b | Indians | U.P. | 97 | 0 | Thal.major-2, AE-1 |
| Mehrotra et al. 1968 | Indians | U.P. | 755 | 0 | AD-2, AE-1 |
| Pande et al. 1970 | Indians | Poona | 352 | 0 | 0 |
| Gupta et al. 1970 | Indians | Allahabad | 1270 | 0 | E-thal.-1 |
| Pande et al. 1972 | Indians | Unspecified | 193 | 0 | 0 |
| ANDAMANS | | | | | |
| Lehmann 1954 | Andamanese | Andaman | 16 | 0 | — |
| | Onges | Little Andaman | 52 | 0 | — |
| Agrawal 1968 | Nicobarese | Great Nicobar | 113 | 0 | — |
| GOA, DAMAN, DIU | | | | | |
| Lessa & Dessai 1955 | Indians | Goa | 684 | 0 | — |
| Trincão et al. 1963 | Indians | Goa | 1843 | 0 | AD-5, AK-2, AQ-4 |
| | Indians | Goa | 833 | 0 | High F-20 |
| | Indians | Diu | 379 | 1 (AS) | AD-2 |
| CEYLON | | | | | |
| De Silva 1957 | Ceylonese | Colombo | 800 | 0 | 0 |
| Wickremasinghe et al. 1963 <i>b</i> | Sinhalese | Ceylon | 1068 | 0 | 0 |
| | Burghers | Ceylon | 60 | 0 | 0 |
| Graff et al. 1954 | Veddahs | Ceylon | 9 | 0 | AE-2 |
| Aksoy et al. 1955 | Veddahs | Ceylon | 158 | 0 | AE-3 |
| Kirk et al. 1962 <i>b</i> | Veddahs | Ceylon | 30 | 0 | AE-14, EE-2 |
| Wickremasinghe et al. 1963 <i>a</i> | Veddahs | Pollebedda, Central Ceylon | 38 | 0 | AE-4, EE-1 |

TABLE - *Continued*

| Reference | Population | Locality | No. tested | No. with positive sickling or Hb-S | No. with other abnormal haemoglobins ^a |
|--|-------------------------|-------------------------------|------------|------------------------------------|---|
| Wickremasinghe et al. 1963a (<i>contd.</i>) | Veddahs | Dambana, Central Ceylon | 27 | 0 | 0 |
| | Veddahs | Ginidamana, Central Ceylon | 87 | 0 | AE-26, EE-2 |
| | Veddahs | Adampane, Northern Ceylon | 32 | 0 | AE-5 |
| PAKISTAN | | | | | |
| Lehmann et al. 1961 | West Pakistanis | Karachi | 76 | 1 (AS) | 0 |
| Mathur et al. 1962 | Sindhis | Agra | 43 | 0 | Thal.major-2 |
| Bolton et al. 1964 | Brahuis | N.W. Pakistan | 4 | 0 | AD-1 |
| | Sindhis | N.W. Pakistan | 6 | 0 | AJ-1 |
| | Baluchis | N.W. Pakistan | 9 | 0 | AD-2 |
| | Pathans | N.W. Pakistan | 18 | 0 | AJ-2, JD-1 |
| | Others | N.W. Pakistan | 18 | 0 | 0 |
| Stern et al. 1968 | Pathans | N.W. Pakistan | 129 | 0 | AD-1, Thal.minor-5 |
| UNSPECIFIED | | | | | |
| Lie-Injo & Ti 1961 | Indians (cord blood) | Kuala Lumpur | 278 | 0 | Bart's-3 |
| Vella 1962a | Indians | Singapore & Malaya | 3341 | 0 | AD-15, AE-15, AL-2, AJ-1, AK-5 |
| | Indians (cord blood) | Singapore & Malaya | 222 | 0 | Bart's-2 |
| Vella 1962b | Indians | Khartoum | 31 | 0 | High A ₂ -10 |
| Pande et al. 1970 | South Indians | Poona | 135 | 1 (AS) | 0 |
| Saha 1970 | Indians | Singapore | 143 | 0 | AD-2, AE-1 |
| Lopez & Lie-Injo 1971 | Indians (newborns) | Malaya | 226 | 0 | Bart's-4 |
| Saha & Banerjee 1971 | Gujratis & Sindhis | Singapore & Punjab | 150 | 0 | AD-1, AE-1, AH-1 |
| | | Unspecified | 116 | 0 | 0 |

^a The indication "—" stands for no electrophoresis performed.

^b Unpublished observation quoted by Chatterjea 1966.

^c Electrophoresis was not performed on the rest of the samples.

^d The subject with AS belonged to Mahar caste.

^e Electrophoresis was only performed on sickling-positive samples.

^f Four belonged to Mahar caste and the other to Maratha caste.

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ADDED IN PROOF. Sinha et al. (1973) investigated 2075 subjects in the Indian Armed Forces personnel, coming from different states of India, and reported the presence of Hbs AD in 13, D-thalassaemia in 1, AE in 3, and EE in 3 individuals. Schroeder et al. (1973) reported the presence of hereditary persistence of foetal haemoglobin in the heterozygous state γ glycine (136) and γ alanine (136) in 6 persons from four Indian families. In 3 of them it was associated with β -thalassaemia. Further, Sukumaran et al. (1972) reported homozygous state of hereditary persistence of foetal haemoglobin γ glycine (136) in two Indian families.

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RIASSUNTO

Viene presentata una aggiornata rassegna bibliografica sulla distribuzione delle emoglobinopatie nel subcontinente indiano. In India sono state finora descritte le emoglobine S, D, E, J, K, L, M, Q, la β -talassemia major, la β -talassemia minor, la α -talassemia minor, l'emoglobina H, l'emoglobina Lepore e la persistenza dell'emoglobina fetale. È anche stata riportata la β -talassemia in combinazione con le emoglobine S, E, D, J, K e Q. L'emoglobina S è risultata più frequente nelle popolazioni tribali di varie regioni dell'India, mentre l'emoglobina D prevale nei Sikh, Pathani e Gujrati. L'emoglobina E ha una frequenza elevata nelle popolazioni di Assam e Bengala e nei Veddah di Ceylon. La β -talassemia major è diffusa in diverse popolazioni. Sulla distribuzione della β -talassemia minor e delle α -talassemie, vi è scarsità di dati.

RÉSUMÉ

Une revue bibliographique est présentée concernant la distribution des hémoglobinopathies dans le subcontinent indien. Jusqu'à présent y ont été décrites les hémoglobines S, D, E, J, K, L, M, Q, la β -thalassémie major, la β -thalassémie minor, l' α -thalassémie minor, les hémoglobines H et Lepore, ainsi que la persistance de l'hémoglobine fétale. La β -thalassémie en combinaison avec les hémoglobines S, E, D, J, K et Q a aussi été rapportée. L'hémoglobine S est plus fréquente parmi les populations tribales de différentes régions de l'Inde, alors que l'hémoglobine D est plus fréquente parmi les Sikhs, Pathans et Gujratis. L'hémoglobine E est très fréquente parmi les populations de l'Assam, du Bengale et les Veddahs de Ceylon. La β -thalassémie major est répandue dans plusieurs populations, alors qu'il y a peu de données sur la distribution de la β -thalassémie minor et des α -thalassémies.

ZUSAMMENFASSUNG

Übersicht über die neuesten Arbeiten, die sich mit der Distribution der Hämoglobinopathien auf dem indischen Subkontinent befassen. Bisher wurden in Indien folgende Hämoglobine beschrieben: S, D, E, J, K, L, M, Q, β -Thalassämie major, β -Thalassämie minor, α -Thalassämie minor, Hb H, Hb Lepore und das persistente fötale Hb. Es wurde auch über β -Thalassämie in Verbindung mit den Hämoglobinen S, E, D, J, K, Q berichtet. Bei den noch in Volksstämmen lebenden Bevölkerungen der verschiedenen Regionen Indiens war Hb S häufiger, während Hb D bei den Sikh, Pathani und Gujrati vorwiegen. Hb E findet sich recht häufig bei den Populationen von Assam, von Bengala und bei den Veddah auf Ceylon. Die β -Thalassämia major ist bei verschiedenen Bevölkerungen verbreitet. Über die Diffusion der β -Thalassämia minor und der α -Thalassämien bestehen noch zu geringe Angaben.

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