Original Article

DISTRIBUTION OF SICKLE CELL GENE IN KORKU TRIBE OF CENTRAL INDIA

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ABSTRACT

Background: The intension of the study was not only to reveal current prevalence rate of sickle cell disease (SCD) but also to adduce most probable reason for its high prevalence in Korku population of Central India and its implications on their health.

Methods: During the study, a total of 865 subjects belonging to different age groups (0-60 years) were randomly sampled for estimating the incidence of sickle cell genotype in them. Blood samples were collected from all (n=865) individuals and positive samples were subjected to cellulose acetate Hb electrophoresis at pH 8.6 for confirmation of their patterns. Prepared questionnaire and previous medical reports were used as a tool to diagnose the clinical presentations of SCD.

Result: Prevalence of sickle cell trait among Korku people was found to be 5.3% (46/865), of which 4.4% with heterozygous gene AS (Carrier) whereas 0.9% with homozygous recessive gene SS (Disease). The frequency of SCD gene was reported more in males and in lower age groups especially in people belonging to the age group of 0-25 years. In homozygous individuals 87.8% of them had no history of pain crisis or blood transfusion. In heterozygous patients most frequently observed clinical features were body pain, joint pain, fever, abdominal pain, pallor and limited complaints about chest pain and intermittent jaundice. No complaint of gall stone was reported. The data also reflects high rate of mortality especially in children.

Conclusion: The high degree of consanguinity and lack of knowledge about sickle cell disorder are major reasons for high SCD gene frequency amongst Korku tribesmen. Premarital screening and genetic counseling needs to be considered as tools to reduce its rate of prevalence.

Key words: Sickle cell disease, Korku, central India, homozygous, heterozygous

INTRODUCTION

In India SCD is most common hemoglobinopathy after thallesaemia[1]. The sickle cell gene is highly distributed in central and southern parts of Indian subcontinent[2]. High frequency of sickle cell gene was also reported from north eastern states, especially from Orissa[3-5].

Central India is dominated by anthropologically important caste and tribal groups along with few migratory groups. It is one of the hubs of sickle cell gene. In Maharashtra three tribes namely Katkaria (Kathodi), Maria-Gond and Kolam are amongst the primitive groups of India and they were diagnosed as high prevalent groups for SCD[6,7].

Korku tribe that we have selected for current investigation is thought to be one of primitive tribe and is believed to be similar to the Santhal tribe of North India. They came to global map before couple of decades as thousands of tribesmen were died because of severe malnutrition. However, it is really unfortunate thing that, they are neglected people and very little information especially about sickle cell trait has been documented from them.

MATERIAL AND METHODS

Study design and Study area: The study was conducted as per cross-sectional survey in Amravati district of Maharashtra state between February 2009 and May 2010. Amravati district comprises of 14 tehsils although, only two tehsils, Chikhaldara and Dharni were selected as most of the Korku population living in them.

Study population: The Korku population is predominantly found in Melghat region of Amravati district of Maharashtra state. Majority of them are distributed in two tehsil, Dharni and Chickaldara.

Sample collection: The samples were collected from Dharni and Chikhaldara blocks of Amravati district of Maharashtra state by arranging camps in schools, residential schools or community halls preferentially on holidays or at the time of evening assisted by local leaders, social workers, or local social organizations.

The informed consent was obtained from study participants prior to the commencement of screening either from the individuals in case of mature participants or from their parents in case of children. The prepared questionnaires were provided only to SCD positive subjects divided into two parts, Part A and B. Part A contains personal information such as educational qualification, occupation and financial status. Part B contains information related with date or year of diagnosis, hematological data, death history in family, cause and age of death and clinical condition of patients. Clinical condition includes, the number of painful crises and number and types of infections per year.

Immediate after signed an informed consent from the participants, a volume of 20 µl blood samples were collected by finger prick from each individual in EDTA containing tubes and screened by solubility test. Screening was followed by taking 2 ml of blood sample from each positive subject by using BD vacutainer (USA), stored in cool until it brought to the Zoology Departmental Laboratory at RTM Nagpur University Nagpur for further investigation. Standard procedures were employed for hematological investigations[8]. All the samples along with known controls were subjected to hemoglobin electrophoresis on cellulose acetate membrane in TEB buffer at pH 8.6 for confirmation of their patterns. The majority of samples were investigated at aforementioned center and the rest were performed by other reputed laboratories.

Statistical analysis: The collected data was checked and entered on Microsoft Excel Worksheet. Statistical analysis was done by using SAS version 9.1.3 (SAS institute inc., Cary, NC, USA). Gene frequency was assessed by using Hardy-Weinberg law. A p-value of≤ 0.05 was considered statistically significant.

RESULTS

Age wise distribution of SCD gene has shown in Table-1. During this investigation, a total of 865 Korku subjects were randomly sampled, of which 46 had hemoglobin profile compatible with SCD. More positive cases were observed from the age group bracket between 0 to 25 years as compared to higher age peoples. No homozygous cases were reported from the age group 41-60 years. Males were observed to be more prevalent to sickle cell gene as compared to females.

Table 1: Age wise distribution of sickle cell disease in Korku tribe

Age	Population screened	Positive	Male	Female	Carriers (HbS)	Sufferers (HbSS)
0-5	102	05 (4.9)	03	02	05 (4.9)	-
6-15	213	12 (5.6)	07	05	08 (3.7)	04 (1.8)
16-25	240	17 (7.0)	09	08	14 (5.8)	03 (1.2)
26-40	178	08 (4.4)	05	03	07 (3.9)	01 (0.5)
41-60	111	04 (3.6)	03	01	04 (3.6)	Nil (-)
Total	865	46 (5.3)	27	19	38 (4.4)	08 (0.9)

Figures in parenthesis indicate percentage

Out of total SCD subjects, 89.13% had provided their information in questionnaire and few of them 28.26% have shown their previous medical reports. The extracted data from the sickle cell carriers (n=33) revealed that, 87.8% cases had no history of pain crisis or blood transfusion. The complaints of mild joint pain and abdominal pain once or twice a year made by 9.09% individuals whereas 3.03% carriers had vaso-occlusive crisis or some other morbidities.

Table-2depicts that, the most frequently observed clinical features among homozygous (SS) patients have body pain, joint pain, fever, abdominal pain, pallor and limited complaints about chest pain and intermittent jaundice. Severe joint pain was noted in 37.5% patients whereas 25% patients had moderate joint pain. More than 50% patients had joint pain with fever and 37.5% sufferers had shown joint pain, abdominal pain and fever. No complaint of gall stone was reported.

Table 2: Common Clinical symptoms observed in sickle cell patients

Clinical complaints	Percentage
Body pain	50
Joint pain	62.5
Fever	75
Abdominal pain	62.5
Pallor	62.5
Bony pain	37.5
Chest pain	12.5
Intermittent jaundice	12.5

(Data retrieved from questionnaires and previous medical reports)

With relation to mortality, out of total SCD positive families (n=35), 28.5% families had lost their one family member and 5.7% families had lost their two family members. 57.14% patients of them could not celebrate their 5th birthday. The data related with parental SCD status of positive children's was obtained only from those parents(n=6) who were participated in screening camps along withthem.66.6% of the parents were found to be heterozygous (AS) and 33.3% were found normal (AA).

According to information available about financial status, more than 95% patients were belonging to very lower income group families. With relation to literacy,

58.69% of them had not completed primary education and 23.91% were illiterate.

DISCUSSION

This is first ever study for assessing prevalence of SCD in central India exclusively dedicated to Korku population. This survey extracted 5.3% prevalence rate of the disease from this tribe. The overall distribution of sickle cell gene in tribal groups of Maharashtra ranges in between 0 and 35%. Highest frequency (35%) has been reported from Otkar tribe of and lowest (1%) from Thakkar tribe. The reported prevalence of SCD in Korku population is somewhat lower than the data published by previous authors[7]. A difference may be attributed probably to the differences in study period. The previous study was carried out before ten years thus, the migrations, improved health services and some sort of awareness might also have responsible for lower down the frequency of sickle cell gene among them.

We have documented more SCD patients from males in common with reports published by other authors[9-11]. There is no significant difference in sickle cell gene frequency was diagnosed among different age groups ($p < 0.01$). Despite that fact, SCD gene frequency was found to be decline with increasing age contrary to earlier findings[12]. The reason for that could be traced in early deaths of SS individuals. Secondly, we have sampled the population randomly and on the basis of convenient sampling because most of the study population living in remote areas covered by thick forest. Therefore it may be possible that rate of prevalence may be different if comprehensive screening program is available there.

The data documented in questionnaire revealed that, the prime complaints included moderate anemia and high mortality rate amongst children. The mortality could be the outcome of poverty, illiteracy, unawareness of food nutrition and good health[13]. They are also facing severe problem of malnutrition. Teenage mothers, large families, lack of medical care and superstitions are also led to early death of SCD children's. Majority of heterozygous individuals sustain normal life without any clinical complaint whereas homozygote faces severe clinical manifestations. [Table-II]

Although, the awareness about SCD is important for early diagnosis, although majority of Korku population is financially weaker and illiterate. The tribal areas also possess difficulty in diagnosis and management, as they are living in remote areas.

CONCLUSION

This study highlights the distribution of SCD gene in Korku Population. The high incidences of sickle cell disease are the result of high degree of consanguinity and lack of knowledge about this disease. Korku population is limited and living in small pocket of central India, the premarital screening and genetic counseling needs to be considered as important tools to control its gene frequency.

RECOMMENDATIONS

In order to restrict further spread and severity of SCD in Korku people steps should be taken towards educating and improving the social, economic and cultural conditions of the families. Capacitating primary health care centers (PHCs) should be made a priority, since urgent medical facility would be provided to improve public health. Educational campaigns aimed at health professionals and at SCD patients' families should be stepped up in order to reduce the mortality and morbidity caused by SCD.

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