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The Burden of Thalassemia in India: A Strategy for Disease Prevention and Control

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ABSTRACT:-

The most prevalent monogenic diseases worldwide are thalassemias and structural haemoglobin variations. A b thalassemia syndrome is thought to affect 100,000 people in India, and there are approximately 150,000 people who have sickle cell disease. However, only a small percentage of these patients receive optimal care, and the majority of families cannot afford allogeneic stem cell transplants. Promoting education and awareness campaigns, stepping up screening in all states with micromapping to determine the true burden, and creating enough facilities for genetic counselling and prenatal diagnosis in public sector institutions are all viable options for management. Government and non-governmental organisations have been working toward this aim for the past three to four decades, but community control in a large and diverse nation is difficult, and a national programme reaching all rural regions, where nearly 70% of the population dwells, has not yet started. 1) Education of health professionals, school and college students, expectant mothers, and the general public are necessary thalassemia control measures. 2) Opening prenatal diagnosis centres in various parts of the nation 3) Establishing more day care facilities for handling current thalassemia patients. 4) Building nationwide stem cell transplant facilities that are affordable. This study investigates methods for collaborating with the Central and State Governments, NGOs, Parents-Patients Societies, and Corporate Houses to successfully lessen the burden of hemoglobinopathies in India. With the assistance of numerous national specialists, the Nationwide Health Mission, Ministry of Health and Family Welfare has developed guidelines for carrying out such a national programme.

Keywords: Burden, Community control, Challenges, β thalassemia.

INTRODUCTION: -

Thalassemia and structural variations of haemoglobin are two examples of hereditary haemoglobin diseases. It is predicted that between 300,000 and 400,000 kids are born each year with a severe haemoglobin disease, making these the most prevalent single gene disorders in the world with an autosomal recessive inheritance [1]. A major thalassemia condition would be present in 56,000 conceptions worldwide, and of those, 30,000 would have major b thalassemia, with the majority of infants being born in middle- and low-income countries [2]. In India, sickle cell disease and b thalassemia are serious health problems. In our 1.21 billion person population, which is made up of a variety of ethnic, cultural, and

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linguistic groups and comprises around 8% of tribal groups, the average prevalence of b thalassemia carriers is 3e4%, or 35 to 45 million carriers. The frequency is significantly greater (4e17%) in some ethnic groups [3, 4].

Small geographic areas within Maharashtra (1-6%) and Gujarat (0-9.5%) have differing distributions of b thalassemia carriers, according to limited micromapping. For each district in these two states, it was also computed how many kids with b thalassemia major should be born each year. In Gujarat and Maharashtra, the annual homozygosity rate per 1000 live births was 0.39 and 0.28, respectively [5]. HbE is more common in the north-eastern and eastern regions, where HbE carrier frequencies range from 3 to over 50% [6-8], whereas HbS is more common among scheduled castes, scheduled tribes, and other backward castes, where carrier frequencies range from 5 to 35%. These haemoglobin variations frequently co-occur with b thalassemia, especially in areas where both conditions are common. Patients with b thalassemia major continue to have just one curative option: allogeneic stem cell transplant. In individuals with low risk characteristics, the likelihood of a successful transplant is >90%, while for patients with high risk characteristics, the prognosis is still uncertain [9]. The majority of families with children who have thalassemia major cannot afford the hefty expense. Thus, it is realistic and practical to try to prevent the birth of a child who is impacted.

Numerous institutions in India as well as charitable groups like Rotary Clubs, Lions Clubs, and numerous NGOs and Thalassemia Parents-Patients Societies have been putting on education and awareness campaigns for the past 30 to 40 years. But in the multicenter Jai Vigyan initiative, awareness of b thalassemia among pregnant women in 6 states ranged from 0.2% to 4.8% in Bangalore, Vadodara, Mumbai, Dibrugarh, and Ludhiana, and from 20.7% in Kolkata [10]. Only 37.93% of the population studied in the Knowledge, Attitude and Practice Study (KAPS), which was conducted among people with medium to low socioeconomic and educational backgrounds in the South 24 Parganas district of West Bengal, an area with a high risk of developing b thalassemia, knew the correct definition of the condition. When asked if a normal person could carry b thalassemia, just 10.07% of those in this group responded affirmatively [11]. Another recent survey-based KAP study on b thalassemia in a high-risk population, the Aroras in north India from rural Rohtak district in Haryana and an urban area in New Delhi, revealed that many people from the rural setting had not heard of thalassemia and the aetiology of the disease was believed to depend on orthodox feelings like the sins committed by the parents. Additionally, pre-marital b thalassemia screening was declined by more than 50% of urban residents [12]. It has been argued time and time again that the mass populace would be significantly affected by a brief segment on b thalassemia by a well-known film star during prime time television programmes over a period of time [13].

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DISCUSSION :-

Identification of carriers is crucial, but screening should be informed consent-based, and after participants have undergone screening, counselling should be the following step. Unfortunately, this is not always the case. In order to establish screening programmes and determine the prevalence of b thalassemia and other hemoglobinopathies in six states—Maharashtra, Gujarat, West Bengal, Karnataka, Punjab, and Assam—the multicenter Jai Vigyan programme of the Indian Council of Medical Research on Community Control of Thalassemia assisted centres in various states, particularly in medical colleges. Carriers of b thalassemia were found in 59 distinct ethnic groups, with rates ranging from 0 to 9.3%. HbE carriers were extremely prevalent in six Assamese ethnic groups (41e66%) [6].

Although it was a difficult effort, prenatal diagnosis facilities were developed in some of these states during the program's second phase. A number of states have now started thalassemia control programmes, and screening is being done in various target groups as a result of the tremendous impact these programmes made. Individual states are responsible for maintaining public health, however under the National Health Mission, the Indian government helps the states financially.

The West Bengal State Thalassemia Control Program consists of 2 nodal centres and the 21 centres that fall under them in various districts. Numerous studies on the prevalence of b thalassemia and HbE in urban and rural settings have been published, and awareness, screening, and counselling efforts are ongoing. B thalassemia prevalence has varied significantly by area, ranging from 4 to 10%. The massive screening and patient data from various places are assimilated into the state database for reporting and analysis using a programme called Thalaman, which has been developed. Additionally, prenatal diagnosis is increasingly frequently performed.

The Gujarat State Thalassemia Control Program, which has been in place for a while, has also provided as an example. State officials issued an order to all universities requiring thalassemia screening in college. The Gujarat Red Cross Society is very involved in this programme, and screenings are frequently conducted in institutions and schools. Programs for prenatal diagnosis and antenatal screening are also carried out in Ahmedabad, Vadodara, Rajkot, and Bhavnagar.

Initially conducted in Thane, Nashik, Satara, and Amravati districts in Maharashtra, a pilot programme for the screening, prevention, and treatment of current patients with thalassemia and sickle cell disease was then expanded to additional areas.

The Central and State Governments, various NGOs, Parents-Patients Societies, and Corporate Houses will collaborate for the care of thalassemia patients in Punjab as part of "Project Rainbow," another effort in the province. (Personal \scommunication).

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Individual centres in other states have started screening and counselling programmes after realising the severity of the issue. HbE is particularly prevalent in the tribally dominant north-eastern states. The majority of the screening programmes that have recently been done here have been hospital-based. In a large series of 9000 patients and other people who were referred to a diagnostic facility in Upper Assam for HPLC analysis, 2294 people had HbE trait and 1892 people had HbE disease. 114 patients with HbE-b thalassemia, 32 with b thalassemia major, and 313 b thalassemia carriers were also discovered, contrary to earlier beliefs that HbE-b thalassemia was either uncommon or under-reported in the north east [7].

As a result, every screening programme implemented in various areas has revealed that many ethnic groups, including Sindhis, Kutchi Bhanushalis, Lohanas, Punjabi Khatris and Aroras, Bengalees, some Muslim groups, and some tribal populations from Orissa and Gujarat, have prevalence rates that are significantly higher than the average, ranging from 4 to 17% [3-6, 8]. Due to the possibility that one of their friends or family members has a child with b thalassemia major, some of these populations are more informed, open, and willing to undertake screening. Therefore, focused screening in these high-risk groups is appropriate and need to be done. Our personal experience has also demonstrated that regardless of whether screening is carried out generally or for specific target populations, young people, such as high school students, frequently do not recall their carrier status, and one time counselling is insufficient and requires follow-up.

Accurate diagnosis of heterozygotes

The majority of centres in India now use CBC and HPLC haemoglobin tests for reporting the carrier frequencies of b thalassemia and other hemoglobinopathies. Capillary electrophoresis has only seldom been employed in contemporary papers. When diagnosing b thalassemia carriers, the cutoff HbA2 value is typically taken to be 3.5%, combined with decreased MCV (80fl) and MCH (27pg) levels, a reasonably high RBC count, and a normal RDW. The country has a large iron deficiency problem, and a primary care doctor should be able to tell the difference between microcytosis and hypochromia caused by iron deficiency when, in addition to low MCV and MCH levels, the RBC count is also low relative to haemoglobin value, RDW is increased, and HbA2 level is normal or low.

Genetic counselling and prenatal diagnosis

In particular, prenatal diagnosis programmes need pre- and post-test counselling to help people overcome unreasonable anxieties, particularly those related to stigmatisation. Additionally, it aids at-risk individuals and families in accepting their status and the effects of the disorder. This will assist at-risk couples in making informed decisions about their future pregnancies in order to prevent the birth of more sick children. The main goal of genetic counselling is to inform the family about the genetic problem, its clinical manifestation and severity, as well as the risk of disease recurrence and mortality. Simple language should be

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used to discuss the family history, psychological problems, moral dilemmas, and obstacles posed by different cultures and religions [14].

To include information on molecular genetics, population genetics, genotype-phenotype correlations, and the burden of disease in India, a country-specific web-based informatics resource called ThalInd has been created [15]. It's also crucial to keep in mind that certain conditions, such as homozygous HbD or HbE disease, hereditary persistence of foetal haemoglobin (HPFH), HbD-b-thalassemia, HbD-Q-b thalassemia, and HbD-b-thalassemia, which increase a couple's risk of having a baby, don't necessarily need to be diagnosed before conception [16]. Instead, they typically have Because foetal collection procedures are intrusive, there is a minor risk of foetal loss, and prenatal diagnoses using DNA have an error rate of about 1%, which must be disclosed during pre- and post-test counselling.

The plan for the journey ahead

The complex and diverse Indian population in both urban and rural areas of every state will continue to require a concerted effort in the direction of education and awareness generation. Although many states have extensive data on the prevalence of b thalassemia carriers, much of information is hospital-based and selective, making it difficult to simply exploit to determine the exact illness load. Many states still lack enough data, and many ethnic groups have not been examined. The requirement for micromapping in every state is demonstrated by the widely varying prevalence, even within small geographic boundaries, and the exceptionally high carrier frequencies in some localities. With a national thalassemia control programme set to launch soon, enough screening facilities of the right calibre It would be necessary to establish control, and both more trained ancillary staff members and genetic counsellors would be needed. Every state has at least one government-run facility. Prenatal diagnosis would be offered, with larger states requiring 2 to 3 centres.

CONCLUSION :-

Direct action Obstetricians and sonologists in smaller towns and rural areas need to regularly organise training sessions in foetal sampling techniques. For the care of the current b thalassemia patients, many of whom are ageing, more day care centres are required. For low risk patients and their families who can afford the cost, stem cell transplantation should be encouraged. It would be more cheap if government hospitals developed transplant programmes. The extensive public health system should be involved and this existing infrastructure should be exploited to reach every corner of the country for a successful programme for the care and control of b thalassemia in India, where 68.8% of the population lives in rural regions. The successful execution of a national control programme would involve a cooperative effort from all the State Governments supported by the Center, assistance from NGOs, thalassemia societies, and corporate houses as part of their social duties, and strong political backing. The National Health Mission, Ministry of Health and

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Family Welfare, along with a number of local specialists, recently developed guidelines for this [17] (National Health Mission Hemoglobinopathies Guidelines in India, 2016).

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