It is well known that with rapid advancement of medical technology various types of diseases of non-genetic origins (that is, environmental) have to a great extent been controlled and are successfully being cured. But the disorders which are of genetic origin can neither be controlled nor cured till today with advancement of medical sciences. It is true that modern medical technology has considerably brought about improvement in modes of treatment for such genetic disorders and consequently the average longevity of persons, suffering from various genetic disorders, has considerably increased. As a result the chance of survival of those individuals with various types of genetic disorders must have some serious social and biological implications which are certainly relevant and critical to man's future biological and social evolutions. For example, the inherited retinoblastoma the frequency of which at present is 1 in 40,000 is likely to increase to 1 in 15,000 in future after successful medical therapy (Reddy 1992). However, till today such problems have not occurred with the populations of simple culture who hardly get opportunity to take advantage of modern medical facilities.
In those societies deleterious genes are speedily removed through the process of natural selection. But the situation is not like that in modern societies which enjoy modern medical facilities. In modern society individuals with genetic disorder not only survive and enter into reproductive age group but also are likely to contribute those deleterious genes to the next generations and thereby there is every likelihood that both genetic and social load in such societies will further increase in future.

With this idea in view in the present study we have dealt with the case of homozygous β-thalassaemia patients in order to find out various physiological symptoms of disease, assessing the growth pattern of the thalassaemic children and various social and economic implications of such disease on the parents of the patients and the patients themselves. In this connection one can recall what Roberts (1975) has said, while working on genetic disorders, particularly hemophilia. He is of the opinion that in modern human societies the deleterious genes have assumed a greater importance beyond that which they confer on the life of the individuals. He has further suggested that it is not the problem of treatment, which are often very expensive and long continued, but of day to day care and maintenance, and finally he has suggested that with advancement of medical technology there are adverse effects on the society as such. As suggested earlier, in the
present study we have taken into consideration the homozygous 
ß-thalassaemia patients and have tried to examine the various 
social problems cropping up not only with the expensive mode 
of treatment but also various social and economic hardships 
on the parents of such patients.

The present study has been divided into two parts, namely biological aspects and social aspects. In biological aspects we have dealt with the symptoms of thalassaemia, to what extent the thalassaemic patients have to, visit hospitals, health condition of the thalassaemic patients before and after transfusion, requirement of operation, i.e., splenectomy, general mode of treatment of the thalassaemic patients and finally the physical growth of the thalassaemic patients.

The social aspects cover demographic composition of the families with thalassaemic patients, economic condition, psychological stresses and economic hardships of their parents. It also includes academic and extra-curricular activities of the thalassaemic patients.

In the present study we have collected data on 380 ß-thalassaemic patients out of which, 14 per cent never had any visible symptoms, though on doctors’ advice their parents brought them to hospitals and thalassaemia in them was detected. However, of all symptoms the most prominent ones are fever with vomiting tendency, loss of appetite, and
anaemic condition. To the best of our knowledge, nobody has ever suggested such symptoms by which a layman can suspect the presence of thalassaemia. It is well known that in homozygous β-thalassaemia there are severe anaemia and elevation of HbF and HbA₂, higher serum-iron concentration and some other haematological characteristics (Basu 1994). The point to be noted is that thalassaemia is generally detected before 1 year of age, though in many cases, perhaps due to negligence and ignorance, it is detected as late as within 5 years. This is by and large universally true.

In the present study, we have examined the frequency of hospital visits by the thalassaemic patients. It is seen very clearly that as age advances, the frequency of hospital visits by the thalassaemic patients also increases. The reason for such increased frequency of visits is that with advancement of age the thalassaemic patients start suffering more and more and they are compelled to visit hospitals in greater frequency for medical treatments and temporary comfort. However, to the best of our knowledge, no one has ever tried to examine this aspect.

In the present study we have examined the feelings of the patients before and after taking transfusions. It is well understood that transfusion from time to time is absolutely needed for any thalassaemic patient. In this connection, it may be recalled what Hardisty and Weatherall (1982) have
suggested "It has now been established quite unequivocally that homozygous thalassaemic children grow better and develop far fewer complications if their haemoglobin level is maintained as close to normal as possible... Immediately after diagnosis a full blood group genotype should be obtained and once steady-state, haemoglobin level has been reached which requires transfusion, the child should be started on a regular transfusion regina". The importance and method of transfusion for management of this genetic disorder have already been reviewed by Weatherall and Clegg (1981), Medell and Berdoukas (1981). So, we have not made any attempt to discuss the importance of blood transfusion in case of the thalassaemic patients. In the present study we have seen that nearly 64.21 per cent of all thalassaemic patients had never faced any serious problem before taking transfusion, whereas 35.79 per cent of them certainly had serious problems before taking transfusions. However, it is found that nearly 62.63 per cent of all thalassaemic patients have certainly felt an improvement in their health condition after having transfusions, whereas the rest have never felt so. Perhaps it may be due to the fact that those, who are older people and have had transfusions several times, can hardly make any difference in their health condition before and after transfusion. In this connection it may be mentioned that so far no body has made any attempt to find out the feelings of
the thalassaemic patients, about their health condition before and after transfusions. What we suggest now is that one has to look into the feelings of the thalassaemic patients regarding transfusions, though it is an established fact that transfusion is a 'must' for any homozygous thalassaemic patient.

In the present study we have made an attempt to see how menarche is delayed in the girls, suffering from \( \beta \)-thalassaemia major. It is seen that the mean age at menarche in case of homozygous \( \beta \)-thalassaemic girls is 13.82±0.49. Bhattacharjee et al. (1982) have reported that the mean age of the Bengali speaking women with \( \beta \)-thalassaemia trait is 13.63±0.12 years. But Bhattacharjee et al. (1977) have reported that the mean age at menarche among the normal Bengali girls is 13.21±0.04 years. It shows that in comparison to the normal girls and the girls with thalassaemia trait the mean menarcheal age in the girls with \( \beta \)-thalassaemia major is further delayed. Chatterjea (1965) has also made a similar observation. He has said, "the secondary sex characters appear late and menstruation is delayed and scanty" in case of girls with homozygous thalassaemia. The present study supports the findings of Chatterjea (1965).

Swarup Mitra et al. (1969), Ajmani et al. (1977) and many others have observed that usually the total haemoglobin
level is lower in those, who have abnormal haemoglobin genes and/or thalassaemia genes than in those with normal haemoglobin genotype (HbA/A). In the present study we have examined the total haemoglobin level among the thalassaemic patients by age and sex. It has been found that the total haemoglobin level in case of the male thalassaemic patients varies between 3.4 to 11.3 g/dl. and that in the female thalassaemic patients between 5.0 to 10.0 g/dl, depending on age of the patients. The WHO (1968a) has recommended that the total haemoglobin level in case of adult normal males should not be lower than 13.0 g/dl. and that for non-pregnant normal adult females 12.0 g/dl. So, in the present study, it is seen that no thalassaemic patient, male or female, can maintain that level of total haemoglobin level as recommended by WHO (1968a) Sukumaran (1975) has suggested that the mean total haemoglobin level in case of males and thalassaemia major is 4.7±0.30 g/dl. and that in females with thalassaemia major is 4.4±0.10 g/dl. In the present study it is found that the thalassaemic patients, either male or female, have slightly better mean value of the total Hb level than what Sukumaran (1975) has suggested. Das et al (1982) have suggested that among the minor male thalassaemic patients (0-14 years) the total haemoglobin level is 5.73±0.30 and that among the adult (15 years+) male thalassaemic patients is 9.98±0.30. They have further observed that the total haemoglobin level among
the minor girls with thalassaemia major is 5.98±0.62 g/dl. From that point of view in the present data the initial total haemoglobin content in the thalassaemic patients, male and female, is slightly higher. However, the present findings in no way contradict the findings of Sukumaran (1975) and Das et al. (1982) since the point to be noted is that the total haemoglobin content in the thalassaemic patients is always lower than that in the normal persons, minor or adult.

When the total Hb content in the person with β-thalassaemia major is examined after they have had some rounds of transfusion, it is found that in all cases the total haemoglobin content in thalassaemic patients, both male and female, has risen to certain level, though not to the level recommended by the WHO (1968). In this connection, it may be mentioned that Choudhury et al. (1997) have observed that there is no consistent pattern in rise of fall in the total haemoglobin content after transfusions and they have further observed that if there is rise in the Hb level after transfusion, the rise in the Hb level is not statistically significant. It may be mentioned that the UKTS News (September, 1993), quoted by the National Thalassaemia Bulletin (1993), has suggested that "Ideally the post transfusion level should be no more than 15 g/dl and pre-transfusion should be no less than 10 g/dl." However in the present study neither of the two has been observed.
The result of splenectomy have already been reviewed by Weatherall and Glegg (1981). Medell and Berdoukas (1981) and Hardisty and Weatherall (1982) have suggested that there is good evidence that young children are more prone to serious infections after splenectomy and the operation should be avoided in the first five years of life. In the present study it is seen that more than 6 per cent of the infants below 5 years of age have undergone splenectomy. It is further observed that as age advances with the thalassaemic patients splenectomy is performed more and more on them. The indications for splenectomy are on increase in transfusion requirements and it is mostly done, when physical discomfort for a large spleen increases. In the present study it is found that (combining both sexes together) nearly 22.22 per cent of the thalassaemic patients have undergone splenectomy. In the present sample majority of the patients belong to the lower income group and most of them just cannot afford the cost of operation. Consequently, in the present sample around 22 per cent of them, mostly belonging to the middle and high income groups, have gone for splenectomy. Since no such data are available we have not been able to compare the present findings with what is happening in other parts of the country.

In the present study we have not been able to collect information on differential blood count, osmotic fragility,
packed cell volume, alkali resistant haemoglobin, etc. since none of the five hospitals in Calcutta, where the present study has been done, keep those records properly. It is unfortunate that in spite of our best attempt we have not been able to study all haematological aspects of the thalassaemic patients.

In the present study we have examined the growth patterns of the thalassaemic individuals and have made a comparison with the normal individuals. Garin (1952) has suggested that height and weight are the two body dimensions which should be taken as good indicators of physical growth. Mills (1937), Wolff (1940), Grenlich (1957) and others have suggested that human physical growth is influenced by many factors including nutrition, genetic constitution, etc. Sarkar et al. (1984) have observed that haemoglobin genotypes and also β-thalassaemic genotypes do not have much influence on height and weight in adult population. Sanctis and Pinamonti (1995) have observed that pathogenesis of growth retardation in thalassaemic patients is complex and incompletely understood. They have suggested some factors like chronic anemia, endocrine disease, defeseral toxicity, etc. as the main factors affecting growth. However, in the present study we have taken into consideration 8 anthropometric and 5 skinfold measurements to understand the growth pattern of the β-thalassaemic individuals. When we
compared each of the measurements, taken on the thalassaemic individuals, male and female separately, with the normal male and female individuals by age-groups it is seen that the growth of the thalassaemic individuals in almost each of these measurements, is significantly lower in comparison to those in the normal individuals. It is further observed that the rate of increment in physical growth of the β-thalassaemic individuals is much slower than that of the normal individuals. It is true that there are a few exceptions in one or the other measurements. But it can be said that the physical growth of the β-thalassaemic persons is certainly retarded at least upto 15 years of age. It is further observed that after 15 years of age the rate of increment in physical measurements of the thalassaemic persons certainly improves, though it never catches up the rate of growth of the normal persons. The present findings do not corroborate with the observations, made by Sarkar et al. (1984). In this connection it may be noted that Johnston and Crogman (1964) have studied the pattern of growth in children with thalassaemic major in the USA. They have inferred that there is a retardation in normal growth expectation and a retardation in the rate of growth. They have further observed that a growth curve drawn on the measurements of the thalassaemic children looks similar to that drawn on the normal but not quite reaching normal values. The present
study fully corroborates the findings of Johnston and Crogman (1964).

As mentioned earlier in the present study we have tried to examine some social implications of thalassaemia disease.

In the present study we have seen that generally the fertility level of the mothers with thalassaemic children is very low. The reason is perhaps that most of the thalassaemic children were born between first and third birth orders. Consequently, the parents generally do not look for larger family size. But the reproductive wastage among the mothers with thalassaemic children is fairly high. However, it is seen that there is an inverse relationship between occurrence of thalassaemia and number of birth order, i.e. as the number of birth order increases, the probability of birth of a baby with thalassaemia decreases.

Since thalassaemia is a genetic disorder there is no correlation between occurrence of thalassaemia and educational standard of the parents. However, when parents are better educated they try to restrict their families after birth of a thalassaemic baby.

When economic condition of the families in which there are thalassaemia babies, is classified into 3 groups - low income group, middle income group and high income group, an interesting point emerges. It is found that the frequency of thalassaemic children is more in middle income and high
income families than that in the lower income group. The reason for this is probably that in the low income group many of the thalassaemic babies die early since the parents belonging to the low income group just cannot afford to give their diseased children proper medical treatment, which is very expensive and long continued. On the other hand the parents in the high income group are certainly in a better position to provide better medical treatment to their thalassaemic children. The parents, belonging to the middle income group, may not be that financially sound like the people of the high income group, but even then they do certainly try their best to provide proper medical treatment as far as possible to their thalassaemic children, in spite of their severe economic hardship.

In the present study it has been seen that the average expenditure on the thalassaemic children incurred by the parents of high income group is much higher than that incurred by the parents, belonging to the middle income group. The average expenditure per month on a thalassaemic child is found to be lowest in the low income group families. It is certainly understandable that the parents, belonging to the low income group, just cannot afford so much expensive treatment for their thalassaemic children, and consequently many of their children must have died in early age. In Table-4.9, it is presented the data on economic hardship, faced by
the parents of the thalassaemic children. It is found that nearly 95 per cent of all the parents, belonging to the low income group, are facing tremendous economic hardship to maintain their thalassaemic children. Even among the parents, belonging to the middle income group nearly 67 per cent of them have admitted such economic hardship, whereas nearly 35 per cent of the parents, belonging to high income group, have had such economic burden for the maintenance of their thalassaemic children. It clearly shows that lower the economic condition of the parents, higher is the economic burden on them for the maintenance and treatment of their thalassaemic children. In this connection one may recall what Tyler et al. (1982) have observed in England while studying the socio-economic burden of Huntington Chorea in South Wales. They have also observed that as the age of the person, suffering from Huntington Chorea, advances, cost of treatment also increases. It creates a serious economic burden on the family. However, in a place like England also where there are a lot of social security for the ailing persons, the families are facing tremendous economic burden for the treatment of their family members, suffering from Huntington’s Chorea.

In a place like Calcutta, where there is hardly any social security for the diseased people, the entire burden of their treatment is borne by the family members. Consequently, one can well assume that the parents, belonging to the low
income group, have to deprive their normal children with a view to meeting all expenses for the treatment of their thalassaemic children. The same is by and large true for the parents, belonging to the middle income group.

So far the thalassaemia patients are concerned, it is found that nearly 29 per cent of them do not go for schooling, perhaps for their bad health condition. It is further observed that as age of the thalassaemia patients advances, the frequency of attending schools drops sharply. Generally, they do not go beyond the secondary level. It is found in the present study that nearly 50 per cent of the thalassaemic patients do not perform satisfactorily in school. Perhaps due to their poor health condition they have to take time off often from their studies. It may be assumed that for their poor educational performances it may, in future, create problems for them to find out suitable jobs, and that in turn may create further economic burden on them.

It is further observed that so far physical activities of the thalassaemic children are concerned, nearly 20 per cent of them have become inactive by 15 years of age and nearly 40 per cent of them by that age cannot perform heavy physical activity. It shows that altogether nearly 60 per cent of the thalassaemic patients become dependent for their day to day work on others. It indirectly creates problems not only on the thalassaemic patients themselves but also on
other normal members of their family. It may be noted that in England most of the children with haemophilia are engaged in little or no physical exercise (Roberts, 1975). The same is by and large true for the thalassaemic children here.

In the present survey data on parents' idea of getting their thalassaemia children married have been collected. It is found that nearly 25 per cent of the parents having thalassaemic children firmly believe that their thalassaemic children should marry, whereas 38.36 per cent of them have expressed their opinion against marriage of such thalassaemic children. Nearly one-third of the total parents till the time of the present survey are found to be undecided. It shows that a good number of parents are ignorant about the serious genetic nature of this disease and many of them are found to be conservative in their outlook since they believe in all traditional system.

In the present survey it is found that nearly 93 per cent of all parents remain always extremely anxious about health condition of their thalassaemic children, whereas 7 per cent of such parents at times become anxious of their diseased children. However, it shows that nearly 100 per cent of all parents spend their time in anxiety for their thalassaemic children. Consequently, nearly 100 per cent of the parents never allow their thalassaemic children to move
freely, though 56 per cent of the parents rarely allow their thalassaemia children some freedom for moving alone.

In the present study, it is found that nearly 11 per cent of the parents with thalassaemic children never go out for any pleasure trip, nearly 9 per cent of them never take their thalassaemic children to visit their relatives or friends and 2 per cent of them never attend any social gathering along with their thalassaemic children. It shows to what extent the parents are depriving themselves and their other normal children from pleasures of life and many of them avoid visiting friends/relatives/or any other social gathering with thalassaemic children since they feel embarrassed to explain to others every now and then of health condition of their diseased children. Roberts (1975) has reported that nearly 50 per cent of the families with hemophiliac children never go out for holidays, considering the health condition of their diseased children and a fairly good number of mothers are unable to take part in any social activities outside their house. The situation is by and large same for the families with thalassaemic children.

In the present study, we have dealt with some biological aspects of $\beta$-thalassaemia and tried to find out some possible social implications of such serious genetic disorder. However, there are a lot of limitations in the present study, for example:
1) We have not tried to find out the present frequency of thalassaemia gene among the Bengalees. It has been reported by Dr. S. Majumder (Statesman, Calcutta dated 14.2.94) that the frequency of the thalassaemic gene is about 10 per cent among the Bengalees at present. It shows that the frequency of thalassaemia gene among the Bengalees has steadily been increasing over the last 40 years, since the time, Chatterjea et al. (1954) reported the frequency of thalassaemic gene was around 3 per cent. It needs an urgent study to determine the exact frequency.

2) The present study is confined only among the thalassaemic patients aged up to 20 years. As a matter of fact the average longevity of the thalassaemic patients have at present increased to 30/35 years (Ghosh, 1996). It shows that many of the thalassaemic patients, with the help of modern medical technology, now enter not only into the reproductive age group but also marry and reproduce. In this connection, it may be noted that Das et al. (1983) have reported that the reproductive fitness of the β-thalassaemic patients is now around 0.03, which means that it has increased from 0 to 3 per cent. It shows that the rate of segregation of the thalassaemic gene has increased to a great extent in view of the fact that previously the segregation of such genes used to take place only through heterozygotes and now both homozygotes and heterozygotes are passing such thalassaemic
Fig. 33: Social fitness of human genotypes.
genes to the next generations. It warrants an immediate study to find out the exact rate of segregation of the thalassaemic gene.

3) In the present study we have not included the married thalassaemic persons. Consequently, it has not been possible for us to find out the social implications of this genetic disorder on conjugal life.

4) In the present study, we have not been able to collect data on all haematological parameters before any treatment given to a thalassaemic patient. It is also needed that such data on haematological parameters must be looked into after giving treatment to the thalassaemic patients. It will help us to understand to what extent modern treatment can remove discomfiture of the thalassaemic patients.

As mentioned above there are some serious lacunae in the present study, which we shall try to make up in our future study.

In this connection, one can mention, the observations, made by Wright (1960), on social fitness of human genotypes. It is admitted that evaluation of social fitness of human genotype is very difficult. However, Wright has treated the problem in terms of the balance between the contributions to society by individuals of different genotypes and what they cost to the society. Figure-33, adopted from Wright (1960) shows the following categories of individuals:
1) Those - who contribute to the society approximately what they cost at relatively modest levels. It includes most of the people - ordinary persons, law abiding citizens, etc.

2) Those - in whom a balance between contribution and cost at relatively high levels. It includes professional persons, technicians, experts, specialists on whose education, society invests heavily, and who receive standard of living above the average by dint of their labour.

3) & 4) Those - make extra-ordinary contribution at either low or high costs to the society. For example, it includes high class artists or experts, persons of genius and others.

5) Those persons - whose capacities are like those in the first and second category. But they repay to the society much less than the cost of their maintenance.

6) It includes criminal and anti-social persons who are otherwise of normal mental capacity.

7) Those - who have sub-normal physical constitution and health. It seems that most of the thalassaemic patients should be included in this category since society pays a lot for them and their return to the society is minimal.

8) Those persons - who are of low intelligence but sufficient to take care of themselves under existing social conditions.
9) & 10) Those persons - who are normal in childhood, but experience early mental or physical breakdown. In this category many of the more severe hereditary diseases like Huntington's chorea come.

11) Those - who are incapacitated physically or mentally throughout life.

12) Those - who die before maturity. It means that they have no opportunity to make any appreciable contribution to society.

13) Those - who die at or before birth.

It is seen that each of these categories includes a great number of genotypes. The categories 1 to 5 are the normal persons, who keep society going and the rest i.e. 6 to 13 in which social cost is much more than returns and they become burden on society. Most of the genetic disorders have become heavy burden on society.

In the present study we have seen that with the development of medical technology the mode of treatment for thalassaemic patients has tremendously improved, and consequently, it may be assumed that many of them are surviving for quite some time and entering into reproductive age group. But there is very little opportunity for them to make any positive contribution to the society. On the other hand, the society is paying heavily on welfare and physical well-being of those thalassaemic patients. It is not
unreasonable to assume that with advancement of medical technology, the rate of segregation of the thalassaemic gene, as suggested by Das et al. (1983), is increasing in the population which means genetic load is accumulating at faster rate in the society. It is well known that if genetic load increases in society, social load is bound to increase concomitantly in view of the fact that society will have to maintain many more thalassaemic patients than it used to do a few decades earlier.

With all these ideas we may suggest some measures by which we can prevent the spread of this genetic disorder and by the process we can reduce to a great extent the social load. The suggestions are as follows:

1) The government must come forward to take up the responsibility of all medical treatment for all thalassaemic patients in this country as it is normally expected that any welfare state would do.

2) Alternatively, if the government itself cannot take up this responsibility the government should reimburse the parents the whole amount spent by them for the treatment of their thalassaemic children.

3) The government must introduce immediately a national register for all thalassaemic patients in this country.
4) The government should make all efforts to encourage and motivate people to go for marriage counselling.

5) The government should make a legislation to prevent marriage between two heterozygotes, i.e., those who are the carriers of this deleterious gene and also the legislation should prevent the thalassaemic patients from marrying.

With this idea in future we hope to make a further study on the β-thalassaemic patients and that may help us understand the biological and social implications of this dreaded disease.