CHAPTER 12

CONTROL AND PREVENTION PROGRAMME IN SAUDI ARABIA FOR BLOOD GENETIC DISORDERS

12.1 Introduction

Blood genetic disorders i.e. haemoglobinopathies, thalassaemias and enzymopathies occur at a high frequency in several regions of Saudi Arabia as revealed during our studies. Since there is currently no definitive cure, the genetic disorders constitute a major chronic health problem in several regions of Saudi Arabia particularly those with a past or present history of malaria endemicity. It is of interest to note that significant improvement in the morbidity and a decrease in the associated complications have been demonstrated as a result of application of proper management and care protocols. Increase with a higher standard of living, good hygeinic conditions and proper dietary habits the overall suffering of the patients appear to decrease considerably [Powars, 1975; Kohotey-Ahulu, 1973]. Many patients exercise a more or less normal life as a result of good compliance with the appropriate treatment regimens and avoidance of the factors known to precipitate complications [W.H.O. Technical Report, 1972]. In several countries major steps have been adopted to achieve better management, prevention and control. The successful implementation of these programmes have been obvious by the significant reduction in the homozygous states of the haemoglobin disorders [W.H.O. Bulletin, 1982]. Among the most successful programmes are those adopted in Cyprus, Sardinia and Greece for the control of β-thalassaemia major (Angastionitis, 1990; Coa et al, 1989).

These studies show that dissipation of knowledge and sensitization of the patients, parents, families and the community at large is essential for application of a control and prevention programme.

Since a better understanding of the pathophysiology, diagnosis, management

and prevention of these disorders is essential in order to reduce the burden on health care services and to minimize psychosocial problems faced by these parents and their families. We adopted several steps in an attempt to initiate effective prevention and control programmes.

12.1.1 Conception of sickle cell anaemia and allied syndrome study group

During the early 1980, the ideas of formulating a study group for the study of blood genetic disorders was conceived at the College of Medicine King Saud University. The Sickle Cell Anaemia and Allied Syndrome (SAS) Study Group for the sickle cell disease and thalassaemia patients and for adoption of comprehensive multidisciplinary heath care programmes, was formulated. The members were drawn from different medical specialities including clinical pathology, cardiology, pulmonary medicine, haematology, biochemistry, nephrology, radiology and obstetrics and gynaecology. The members adopted a multidisciplinary approach towards patients care, control and prevention. The major responsibilities of the SAS group are presented in Table 12.1.

Over the last five years the SAS group members actively applied comprehensive and individualised health care programmes both locally in Riyadh and in other regions of the country through coordinators. Weekly meetings were held amongst the members to discuss patients, their complications, detailed analysis to be conducted and management strategies. Meetings were held with the parents, patients, family members to improve awareness and better understanding of the disease state and its complications and to adopt steps for better home management. The SAS group also prepared a monographi entitled "Sickle Cell and Thalassaemia Disorders - Guidelines

Table 12.1: Outline of the Responsibilities of the SAS and National Working Group

Responsibilities of the SAS and National Work Group

A. Multidisciplinary Approach to Patient Care
   - Clinical monitoring of the patient.
   - Identification and treatment of complications.
   - Multidisciplinary approach to individual patients problems.
   - Steady state and crises period monitoring:
     - Clinical and Laboratory
   - Regular follow-up
   - Identification of psychosocial problems and the methods to solve these problems.

B. Creating awareness and educating the public and health care personnel:
   - Patient and family education.
   - Education programmes for under- and post-graduate students (medical and paramedical).
   - Publication of newsletters.
   - Seminars, workshops and symposia.
   - Public lectures, T.V. programmes, Newspaper articles.

C. Multidisciplinary approach to control of haemoglobin disorders
   - Adoption of preventive measures (vaccination, prophylaxis).
   - Pharmacological manipulation.
   - Screening and genetic counseling.

D. Research
   - Molecular basis of haemoglobin disorders.
   - New therapeutic measures.
   - Advances in bone marrow transplantation.
   - Gene therapy.

for Management" covering the various management strategies for patients suffering from haemoglobin disorders and thalassaemia. In addition, various management protocols i.e. the hydroxyurea, erythropoietin and piracetam protocols, were applied by the SAS members on their patients.

12.1.2 Conception of the National Working Group

To conduct comprehensive health care and control programmes at the national level, the `National Working Group' was conceived in collaboration with the Ministry of Health, Riyadh. The members were drawn generally from the Ministry of Health hospitals and health centres and other hospitals in the different regions of the country. The major responsibilities of the National Working Group were the same as those for the SAS group presented in Table 12.1 but were adopted at the national level. The major activities addressed by this group included (a) appropriate diagnosis using the facilities at the national referral centre in Riyadh (The Department of Medical Biochemistry at the College of Medicine was designated as the National Referral Centre as the complicated cases from other regions were referred to the centre).

12.1.3 Application of comprehensive management, therapeutic and care programmes for proper management of patients with haemoglobin disorders

A. Preparation of regional and hence a national registry of patients with blood genetic disorders.

B. Prevention and control through carrier detection and genetic counselling.

Regulary contacts consultations are actively maintained between the members of the National Working Group through yearly meetings held under the auspices of the

Ministry of Health, where personal experiences are presented, difficulties faced are outlined and suggestions for management are made.

12.1.4 The World Health Organization Collaborating Centre for Haemoglobinopathies, Thalassaemias and Enzymopathies

In recognition of the longstanding services offered by the Department of Medical Biochemistry in the field of haemoglobinopathies, thalassaemias, enzymopathies, the World Health Organization (W.H.O.) designated the department as the `W.H.O. Collaborating Centre' for Haemoglobinopathies, Thalassaemias and Enzymopathies in January, 1991. The major responsibilities as the W.H.O. Collaborating Centre are listed in Table 12.2. Steps were adopted to fulfill the responsibilities. Training programmes were held as workshops for technical staff and clinicians. Awareness programmes were carried out to investigate community knowledge about blood genetic disorders, and to improve health care delivery services to community. Studies were conducted to determine the psychosocial burden on the patients and family. These aspects are discussed in the following section.

12.2 Psychosocial effects of the haemoglobin disorders on the patient/family unit

All chronic illnesses, due to the continuity and persistence of symptoms, are frequently associated with psychosocial affects on the patients and the family. The patient, once he has grown out of infanthood, starts to feel this lack of his ability, during playing and other strenuous situations. Frequently, the steady state is interrupted with more acute complications and the patient has to be absent from the school. This influences the school performance and creates a psychological stress. Parents and other family members are more seriously affected. They are faced with the regular burden of

Table 12.2: Outline of the responsibilities of the Medical Biochemistry Department, (College of Medicine, King Saud University)
W.H.O. Collaborating Centre for Haemoglobinopathies and Enzymopathies

Responsibilities as W.H. O Collaborating Centre

- Improvement of delivery of thalassaemia control services to community.
- Investigation of community knowledge and attitude in control of hereditary diseases.
- Development of training aids for hereditary control programme.
- Assist in training for population screening and fetal diagnosis of the haemoglobin disorders.
- Develop and improve diagnostic methods for blood haemoglobin disorders and enzymopathies.
- Investigate structural, function and genetic aspects of haemoglobin disorders and enzymopathies.

leaving work, leaving other household chores or other children to take the child to the hospital for follow-up. They are faced with the financial burden due to extra dietary and medical requirements of the patients, they feel depression and anxiety due to the uncertainity of the future of the child and they pity the child due to the lack of normal development and growth. The treatment regimes, as in patients with ك-thalassaemia major, often require blood transfusions and chelation therapy. Both of these take considerable time of the family members and in addition to the complications produced in the child, they also create both financial burden and psychosocial trauma on the family. These frequently influence the relationship of the parents towards each other and towards other siblings who feel neglected and often feel envious or jealous of the sick child.

As in the case of other chronic illnesses, presence of haemoglobinopathies has an influence on the entire family and the challenge is expected to affect the cognitive, emotional and behavioural aspects of the family, often resulting in altered family dynamics. Several psychosocial problems affect the patient/family unit as a result of increased financial burden, uncertainty and anxiety of the future and emotional strain (resentment, disappointment, guilt, anger, anxiety or embarrassement). A few studies that have been conducted on families with sickle cell disease (SCD) have suggested that the chronically ill individual may experience depression, lower self-esteem, social disability, chronic anxiety and poor school achievements. More recently, Burlew et al (1989) have examined the effect of child with chronic illness on the family system, on the one hand, and the impact of family characteristics on the manifestations of illness, on the other. They have shown that the families of patients with SCD experience additional emotional strain and the relationship between the parents and between the

patients and parents and other siblings is affected adversely. It is suggested that patients with more knowledge of the disease cope more favourably with the illness and the fewer the psychosocial stresses in the family the better the patients response to illness.

Haemoglobin disorders occur frequently in the Saudi population, but to date no study has been conducted to identify the psychosocial problems associated with these disorders and how they influence the patient/family unit. In our study we evaluated patients and their families in order to identify the major psychosocial affects of haemoglobin disorders and to determine ways by which the psychosocial burden can be alleviated.

The study group comprised 164 patients, suffering from SCD, and their family members. All patients were regularly attending the out-patient clinics of the Ministry of Health hospitals in the different regions. Specific questionnaires were prepared and the patients and their family members were invited to answer the questions asked by their clinicians. The questionnaires were designed to assess the attitude and practice of SCD patients and their families towards SCD, the social effect of the disease and the social adjustments made by the patient and the family.

The key questions are summarized below:

1. What is the attitude of the family members to the child with haemoglobin disorders?
2. What is the parents' reaction to haemoglobin disorder?
3. What are the reactions of the other siblings towards the patient?
4. What is the effect of haemoglobin disorder on family income?
5. What are the effects on social relations of the family?

6. Does the family get extra financial support; if yes, from whom?
7. What is the knowledge of the family about the nature, cause, treatment, prognosis and complications of the haemoglobin disorders?
8. What is the degree of disease understanding of the family?
9. What is the time lapsed since the disease was diagnosed?
10. What is the frequency of follow-up?
11. What are the number of hospital admissions following diagnosis?
12. What is the family reaction to hospital admissions?
13. How is the performance of the child at school?

The following is a summary of the answers for each question:

12.2.1 The Families/Patients

Among the families questioned 41.7% had 1 child suffering from a severe haemoglobin disorder, 23.3% had 2, 17.5% had 3 and 17..5% had more than 3 members suffering from these disorders. In the majority of the cases (35%) over 5 years had passed since diagnosis, 17.5% had been diagnosed in the last 3-5 years, 24.2% in the last 1-3 years and in 11.7% the diagnosis had been made during the last year. One third of the patients had 1-3 hospital admission every year, and one third had over 6 admissions, 8.6% had between 4-6 admissions while 18.3% had none. The majority of the cases (75.8%) were taken to the hospital at regular interval for follow-up, while 8.3% never went for follow-up and in the rest the follow-up was irregular.

12.2.2 The knowledge of haemoglobin disorders

The knowledge of the nature of the haemoglobin disorders was poor. 53.4% of the individuals tested knew that the disorders resulted from either an abnormality of the

haemoglobin molecule (19.2%), low level of haemoglobin (17.5%) or abnormal red blood cells (16.7%), while 46.7% did not know the cause of these disorders. Out of the total, 68% had the knowledge that blood was affected, while 21.7% were of the opinion that it was a joint disorder. The rest (10.37%) believed that the major organs affected were heart, brain or lungs. On the other hand, 51.8% knew that the disorders were inherited while 8.6% believed that infections cause these disorders and 39.6% did not know the cause. The lack of awareness was also reflected in the openion of the individuals about the disease prognosis. Over 29% believed there is a cure for these disorders, 14.2% believed that there is no harmful effect, 23.3% thought that these disorders were fatal and 27.5% did not know . Only 22.5% believed that the disease is present at birth, while 49.2% stated that it started below the age of 3 years. 13.3% thought that the symptoms appeared between 3-5 years of age, while the rest (15%) thought that they appeared after 6 years of age.

12.2.3 Attitude of the family

One half (55.5%) of the family members tested felt normally towards the child with a haemoglobin disorder. 37.8% said they felt pity for him and 6.0% said that they avoided and neglected the child. The parents' reaction towards the sick child was normal in over half the parents (55% mothers and 52.5% of fathers); Almost 29.2% of the mothers felt depressed at the fact that their child had SCD, compared to 20.08% of the fathers. The fathers had more anger (13.7%) compared to the mothers (7.5%). Only 0.8% of the mothers were over-attentive while 1.7% of the fathers neglected the child. The majority of the siblings, 84.11%, did not feel differently towards the sick child, but 15.9% felt a great pity towards him.

12.2.4 Social influence

Among the families tested, 87.5% of the families were of the opinion that having a child with haemoglobin disorder had no effect on their social relationship with others. The others said they had lost their friends (11.7%) and had decreased social interaction (0.8%).

12.2.5 Effect on income

Almost 68% of the family did not feel any extra financial burden, but 30.8% felt the extra financial load. Only 2.5% had extra help from the government while 97.5% of the families were not getting any extra support from the government authorities or other family members.

12.2.6 Follow-up

The follow-up was regularly conducted by 79.1% of the families, 7.6% were irregular and 13.3% never took the child for follow-up. 73.3% thought that the follow-up was necessary for improving the patients' health, while the rest (26.7%) thought that follow-up was to avoid complications, or their doctors advised them and so it must be done or because of previous experience. Regarding the hospital admission of the child, 85.8% of the families willingly agreed to the hospital admission, only 2.5% did not agree and the rest, (11.7%), did not know whether there was any benefit in hospital admission or not.

12.2.7 Performance of the child at school

Over 50% of the children attended the school regularly and took active part in school activities. Almost 39.2% children were either very good or good in their studies, 3.37% were average, while 4.2% were poor in their performance at school. 17.5% of

the children attended school but were irregular in their attendance while 35.8% did not go to school. The majority of these were below the school going age (i.e. < 6 years).

Our study on the Saudi patients shows several differences compared to the results in the literature. In general, the knowledge of the parents and family members about the genetic disorders was very low. It did not seem to influence their routine family and social life. They, due to the lack of knowledge, did not anticipate what may be the complications of these disorders, and how may they be solved. They generally had a strong faith in Allah Almighty and often answered by saying that "Allah knows" why or what was in line for the future. Thus the majority of the patients were treated normally by the family members and friends. Similarly, due to the strong social bonds, and a sense of providing help, the majority of the families or children did not feel socially left out or neglected. In fact, the social system is such that a family with a sick child is given extra care and consideration by other relatives and friends. The visits of the neighbours and other family members at each occasion or complication, provides extra support to the family of the sick child.

In conclusion, the result of our study shows a significantly lower prevalence of psychosocial problems and nominal effect of haemoglobin disorder on the patient/family unit. It may be that as the knowledge of the patients and their families about the disease and its complications will increase the reactions will change. However, due to the strong faith in Allah, which is one of the major common factors in all families in Saudi Arabia, the psychosocial burden may be confidently overcome by the belief that "this is from Allah and must be accepted with strong faith".

12.3 An awareness programme of blood genetic disorders

The clinical manifestations of sickle cell anaemia (Hb SS), Hb S/β°-thalassaemia, Hb S/ α-thalassaemia ,α -and β-thalassaemias and glucose-6-phosphate dehydrogenase (G-6-PD) deficiency vary from mild haemolytic anaemia to a severe incapacitating haemolytic state requiring frequent hospitalization and blood transfusion due to associated complications affecting the major tissues of the body. Abnormal growth and development, organ dysfunction, transfusion reactions and complications due to iron overload in regularly transfused patients are frequently encountered and are precipitated by several factors including dehydration, infections, cold, fever and hypoxia.

Since, genetic disorders are chronic and are frequently associated with lifelong suffering, knowledge of the nature of the disease, mode of prevention, management and treatment has two major advantages. Firstly, the morbidity associated with these disorders can be reduced by avoidance of precipitating factors, proper care, management and improved compliance to the therapeutic regimes and secondly, by applying the general rules for the control of the disease i.e. performing premarital screening, prenatal screening, the homozygous disorders may be prevented and hence control achieved.

The chronic nature of these illnesses is believed to challenge the entire family and frequently alters interactions within the family. A recent study has shown that the level of education and knowledge of the parents plays an important role in coping favourably with these illnesses. The awareness of the complications and early proper home and hospital management can result in significant reduction of morbidity in these

patients. A knowledge of the aetiology, signs and symptoms, prognosis, complications and treatment of the disease, is necessary to understand the disease and to face its consequences with the least possible psychosocial trauma.

During our study, we initiated programmes to increase doctor/family/patient interactions and to determine ways to increase awareness and decrease psychological trauma caused by these disorders.

Initially, a study was initiated to determine the level of awareness of the families of patients suffering from blood genetic disorders, to enhance awareness and to suggest plans for improving the provision of the health care. In this study a health care team composed of at least three clinicians and two technical staff made local trips for a period of 2-7 days to regions where the gene frequency of the blood genetic disorders was high. Lectures were delivered to and discussions were held with the local treating doctors to highlight recent trends in management. The families (164) included in this study was randomly interviewed in different regions of Saudi Arabia. All families included in this study had at least one or more members suffering from a severe form of a blood genetic disorders (i.e. either Hb SS or Hb S/β°-thalassaemia). At least one parent was available for interview all cases, while in some both parent were available to answer the questions. An appointment was given to the families to attend the outpatient clinic. The questionnaires were prepared in English and Arabic and the family was asked by a clinician to answer questions, where the answers were filled on special forms. To enhance awareness, a lecture was given about the basic information on the disease and was followed by discussions with the families. The questions were repeated to the family and the questionnaires were refilled. The questions on sickle cell disease

i. What are genetic disorders?

ii. What is the mode of transmission?

iii. How are the genetic disorders transmitted?

iv. What can be done to prevent genetic disorders?

v. What is Sickle Cell Disease (SCD)?

vi. What are the major symptoms of SCD?

vii. What is home management of SCD?

viii. What is a crisis?

ix. How can you deal with a crisis?

x. What factors precipitate crises?

xi. How can you prevent a crisis?

The data before and after the teaching session was separately analysed and evaluated and the answers were judged as either `correct', `wrong' or `do not know'. The answers to the questions prior to and after the teaching sessions were analysed separately.

12.3.1 Prior to the teaching session

The percentage of families who respond to questions (gave a correct or a wrong answer, or did not know) prior to the teaching were calculated and the results were as follows:

12.3.1.1. Sickle cell disease (SCD)

The questions were more specifically directed to unveil the degree of the awareness about SCD. A total of 41% of the families knew correctly what is SCD, 20% had a

wrong conception and 39% were non committal (did not know). About half of the number of the families (51.8%) knew that it was genetic, 8.6% thought it was infectious disease (Fig. 12.1). The majority of the families (72%) had a correct knowledge of the major symptoms of the disease, only 2.4% had a wrong idea and 25.6% said they did not know (Fig. 12.2).

12.3.1.2 Genetic disorders and their transmission

The general questions asked were aimed to determine the general knowledge of the families about genetic disorders, how are they transmitted, and how can they be prevented. Only 7.6% of the families knew correctly as to what are genetic disorders. On the other hand, 11.8% gave a wrong answer, while 80.6% did not know (Fig. 12.3). 9.3% of the families knew the mode of transmission, 37.8% gave a wrong answer and 52.9% stated that they were not aware of the answer (Fig. 12.4). When asked how genetic disorders can be prevented, 49.5% gave a correct answer, 7.6% gave a wrong answer, while 42.9% give `did not know' answer (Fig. 12.5).

12.3.1.3 Crises and their management

All families identified the crises as sudden painful episodes affecting the joints, abdomen or bones or causing severe anaemia. However, only 29.4% knew what factors precipitated crises, 10.9% give wrong answer while 59.7% said they had no idea what caused these sudden severe episodes (Fig. 12.6). About dealing with crises 71.4% did not know, 24.4% give a correct answer and 4.2% did not know (Fig. 12.7). On the management, about half of the families (55%) gave an acceptable answer, 2.4% had wrong information, while 42.7% did not know (Fig. 12.8).

12.3.2 Following the Teaching Session

After the lecture of about 30-45 minutes and discussion for 10 min. with 4-5 families at each session, the same questions were repeated and the questionnaires were refilled and data analysed. Figures 12.1 to 12.8 present the prevalence of each answer after the teaching session. The results indicated that there was improvement in the knowledge after the teaching session; however, the results were not statistically significant in most cases.

12.3.3 Distribution of articles in Arabic

Following the distribution of written articles, and several teaching and discussion sessions, the awareness of the family members improved significantly. This was evident particularly in their dealing with crises and home management of the patients with sickle cell disease.

Our results showed that in Saudi Arabia, the frequency of the blood genetic disorders, particularly Hb SS and thalassaemia, is high in several regions, and the level of awareness of the families about these diseases is limited. This may be due to multiple factors including the fact that only a limited information is conveyed to the patient and their families in a busy clinic. The understanding of the terminologies is limited due to a non-scientific background of the population tested. The last hypothesis is evident from the results of this study since the information gained after single teaching session was not significantly higher than the information of the preteaching session. The limited knowledge gained following the single teaching session would be due to either an inadequate explanation during the teaching session, or due to inadequacy of the population to understand fully the lecture delivered. Repetition of the

Figure 12.1: Patient/Family Awareness –
Mode of Transmission of SCD

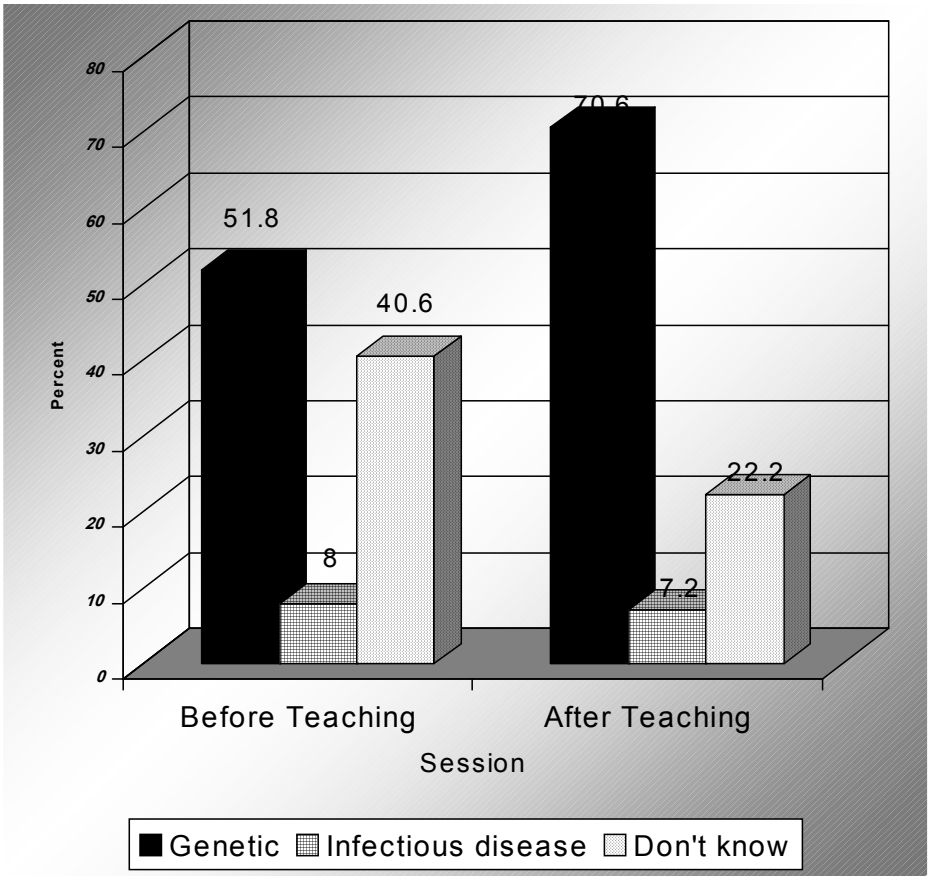


Figure 12.2: Patient/Family Awareness:
Major Symptoms of SCD

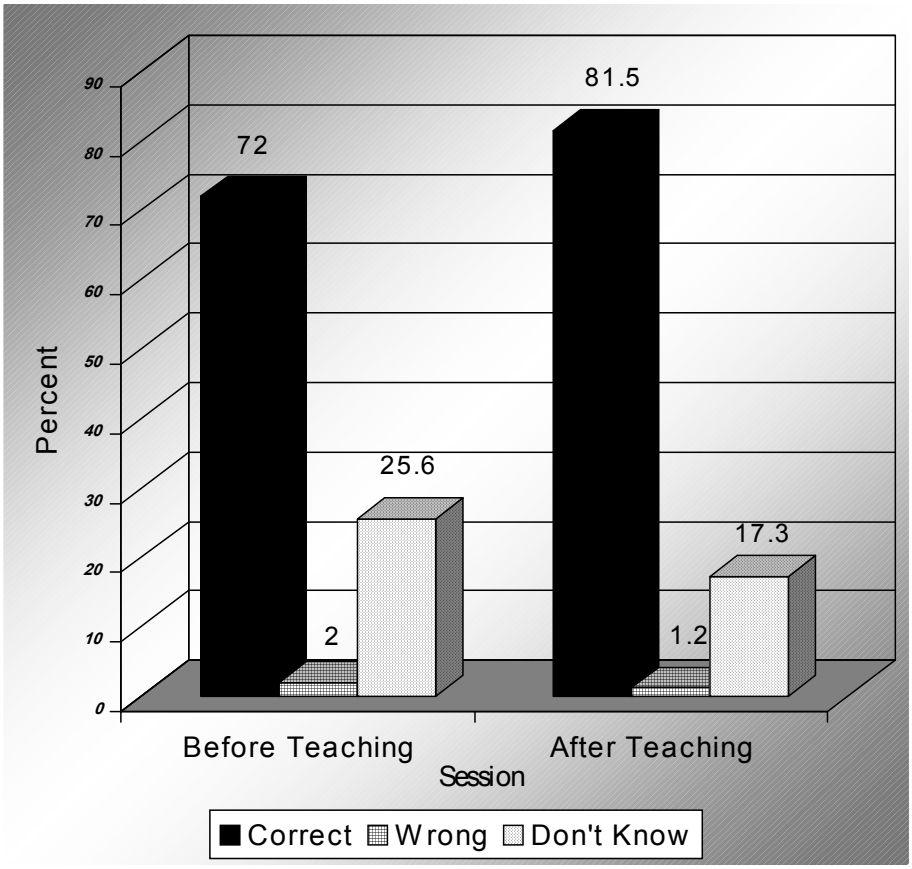


Figure 12.3: Patient/Family Awareness:
Understanding of genetic disorders

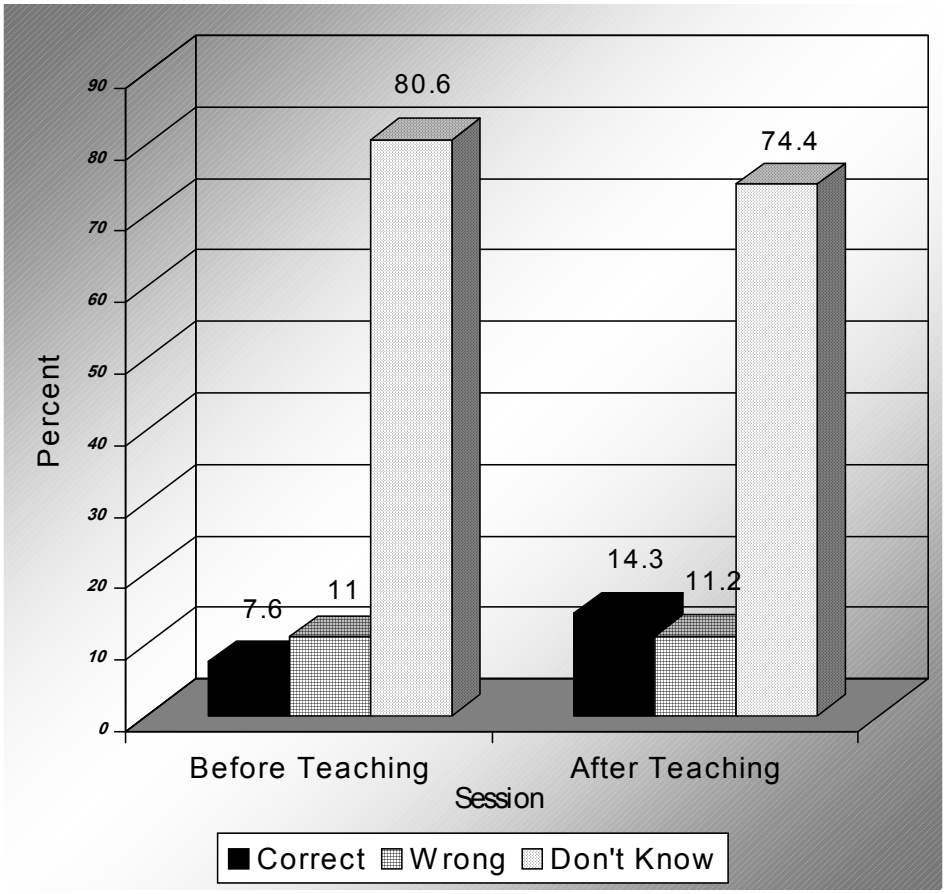


Figure 12.4: Patient/Family Awareness:
Conception of mode of transmission

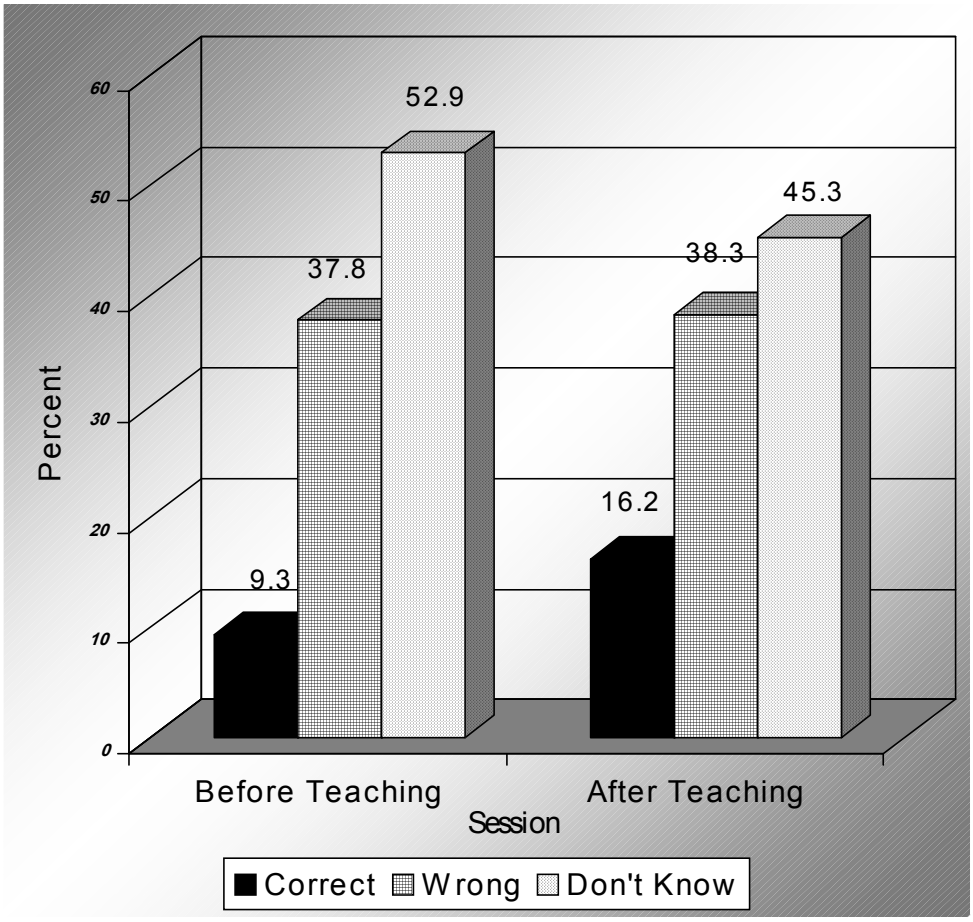


Figure 12.5: Patient/Family Awareness:
Prevention of genetic disorders

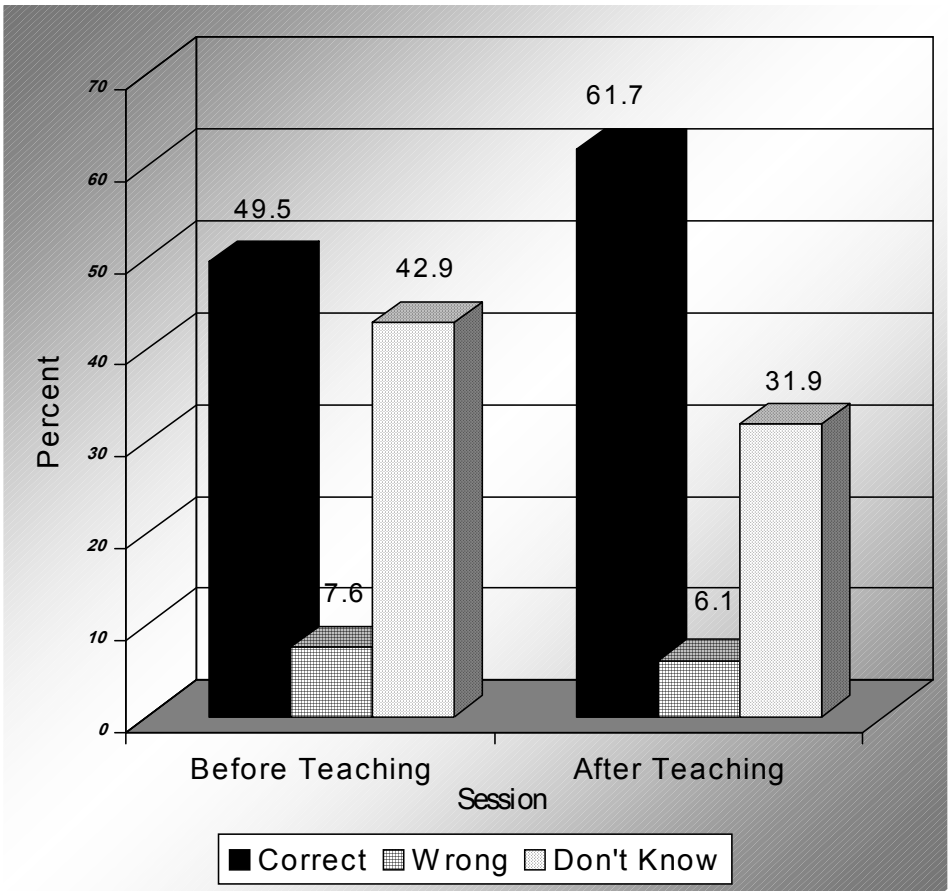


Figure 12.6: Patient/Family Awareness:
Factors precipitating crises

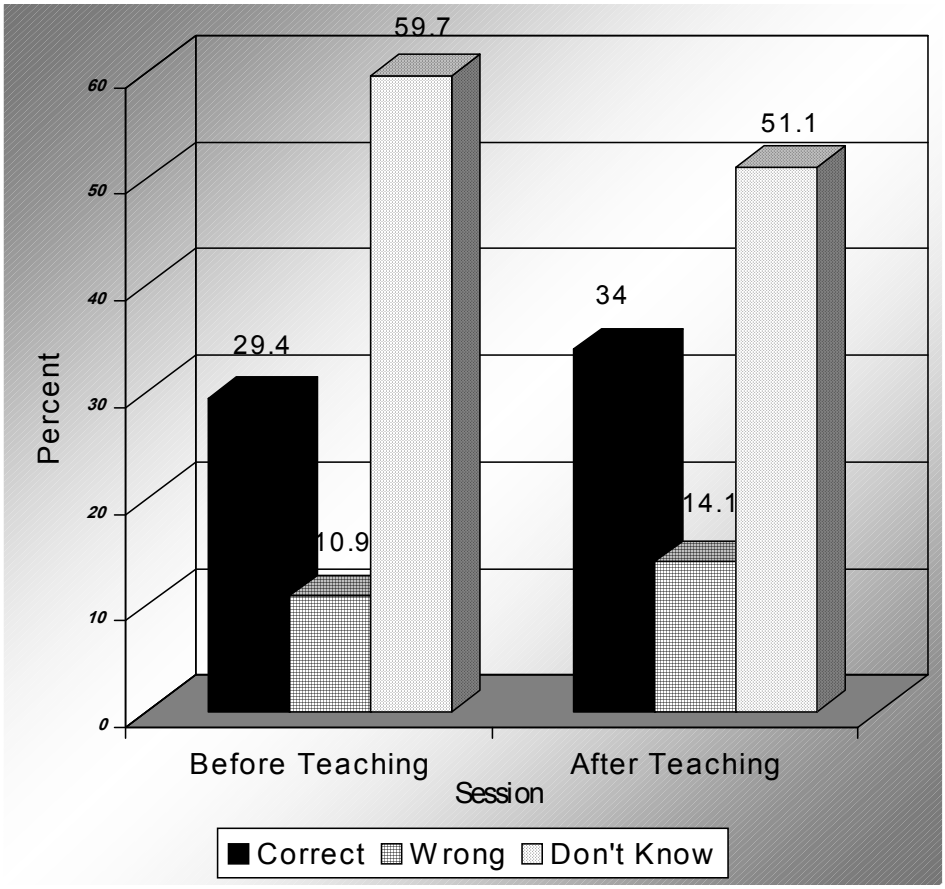


Figure 12.7: Patient/Family Awareness:
Dealing with crises

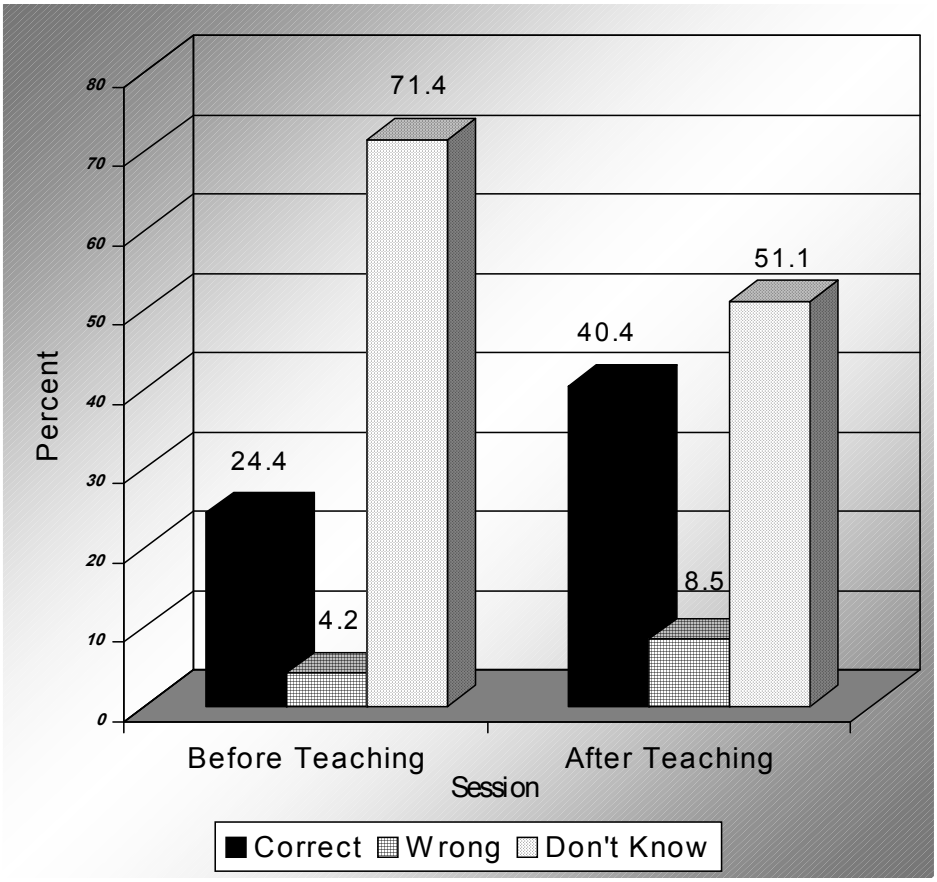
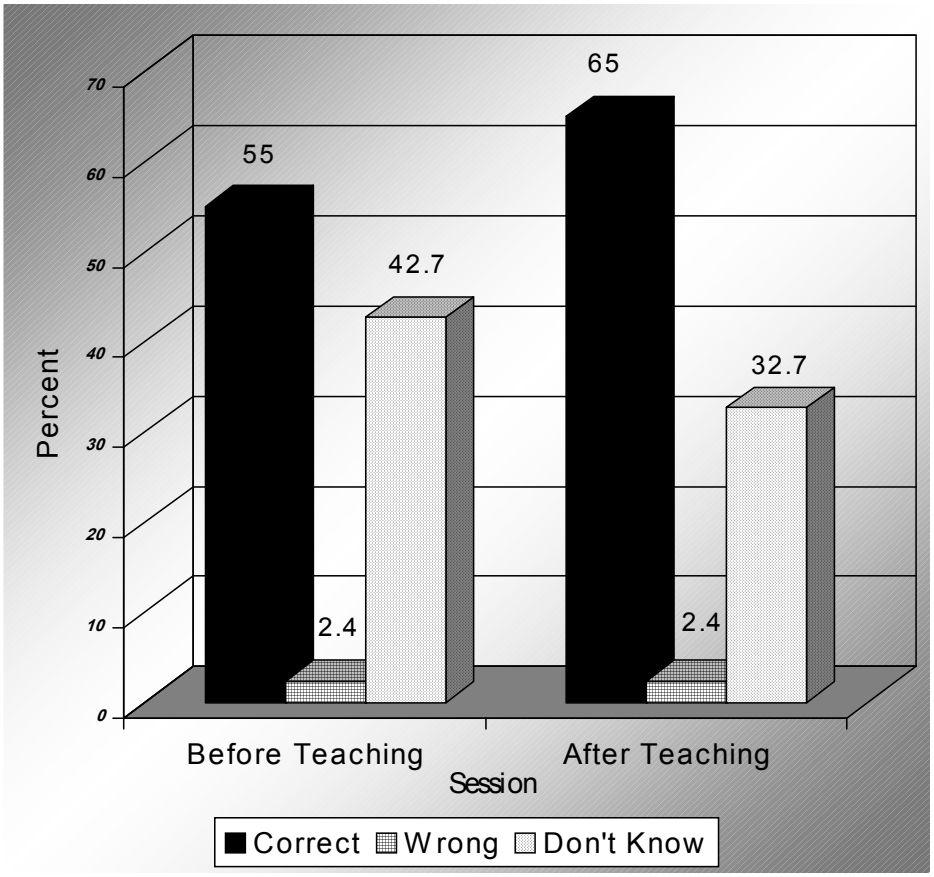


Figure 12.8: Patient/Family Awareness:
Home Management



same material, and informal discussions may improve the understanding. Written articles, in simple words, and repeated exposure to such literature has shown significant improvement. It is expected that further exposure through the newspapers, TV or general public lectures will prove to better enhance the knowledge regarding blood genetic disorders.

Parents tend to learn a lot from other parents, friends and families of patients, thus parent-parent meetings, may be of greater benefit in spreading the knowledge and enhancing the awareness of the public. Doctor-parents meetings in relaxed, friendly atmosphere may also convey more information. Inclusion of relevant information in the school curricula is another efficient way of enhancing the awareness. Parents generally follow the children's courses, help to prepare the child for examinations and discuss the studies with their children. Through this practice, patients, carriers and families can acquire sufficient information on the nature of genetic disorders.

It is of particular relevance to emphasize, that for better care of patients with blood genetic disorder, health care management must start early. After birth, proper vaccination regimes prevents infections and decreases morbidity. Awareness of the family helps in avoiding the factors that precipitate crises and improves compliance to the various therapeutic modalities - a necessity in case of chronic diseases. Early intervention and management by a conversant management team and through appropriate management protocols, minimizes complications. The Cyprus experience, where affected individuals, on appropriate management protocol, lead almost normal lives supports this assumption. Since these genetic diseases have currently no cure, at all cost, everything must be done to decrease the suffering and morbidity of the patient,

to encourage him/her to live a life as normal as that lived by others.

Steps to improve awareness of the patients, their families and the community at large were taken during our study. We prepared pamphlets in Arabic and circulated these to the different parts of the country and to other Arab countries. Articles were written for the newspapers and films were prepared to be shown on the television which covered basic information about blood genetic disorders.

12.4 Care and Control Programmes at National and Regional Level

Plans to link the national aprogrammes to the regional and international levels have been considered of particular relevance. The health care at the `National Level', is linked to the `Regional Level' which in turn is linked to the International level (Figure 12.9). The contributions at each level with particular stress to improve awareness, health care delivery and to work towards achieving control and prevention, are coordinated and it is anticipated that such cooperative and coordinative efforts at national, regional and international levels will provide suitable means aimed at decreasing morbidity and mortality from blood genetic disorders and will play a more active role in control and prevention.

Figure 12.9: The National, Referral and International Links