Thalassemia in Messina: a sociological approach to chronic disease

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Abstract

Changing the care along with improved treatment, have altered the life of thalassemia patients, one of the world's most common genetic diseases (Thalassemia International Federation; http://www.thalassemia.org.cy). The new demography of the disease, with its widely variable phenotypes, has implications for its diagnosis, counseling, and management. Improved of the new treatment of this ancient disease is essential for optimizing survival. From June 2010 to January 2011, we interviewed 36 people with thalassemia from a primary care in Messina (Center of Genetics and Immunology). The aim of this study is to show the results of a survey conducted in this Center in Messina. This study shows the importance of influence of a multidisciplinary approach, medical, psychological and social, that addresses the changing treatment and epidemiology of thalassemia in order to ensure a better quality of life and survival. Understanding the influence of all three types of resources, medical, psychological and social, is critical for constructing ways to enhance health capability, chronic disease self-management, and health.

Introduction

Thalassemias are a heterogeneous group of inherited disorders of hemoglobin synthesis resulting in life-threatening anemia and requiring regular blood transfusion for survival.[1] It's particularly associated with people of Indian subcontinent, Middle East and Mediterranean origin. Since the last survey results that in the Mediterranean basin is the most common β-thalassemia (Cooley's disease), also called anemia for this *Mediterranean*; are approximately 7000 patients in Italy.[2] Sicily and Sardinia where there is a rate of thalassemia equal to 12 per cent. In the latter two Regions carriers are more than 700 thousand out of a total population of just under 7 million.[3] In Sicily there are 1752 patients with thalassemia (1141 whit thalassemia major; 621 whit thalassemia intermedia).[4] β-thalassemia is an inherited blood disorder for which the body produces, in red blood cells, hemoglobin fewer than normal. Hemoglobin requires iron to transport oxygen in the body. In thalassemic this failure causes a reduction of the volume of red blood cells and the subsequent onset of anemia. Hemoglobin is normally composed of 4 prosthetic groups that contain iron, responsible for the red, and globin, a protein part consists of two α chains and two-β globin. Through blood tests, we are becoming aware of the failure of one of these two protein chains that become the cause of two different thalassemia syndromes: the α-thalassemia and β-thalassemia. The majority of *carriers* do not show any signs of relief. In fact, many are unaware of having these disorders. It is important for these individuals to become aware of their own fault because in the case of union with another immune carrier is the probability of generating: 25% a child with β-thalassemia major, that is to say a child of 4 will be having Alzheimer's Cooley; another 25% of all healthy children; finally the remaining 50% of children suffering from β-thalassemia even they, or immune carriers. The first symptoms of the disease are recognizable around the third/fifth month of life and occur usually within the first/second year. The child is not capable of producing adult hemoglobin and then falls ill with a severe anemia. It becomes very pale and suffered need for transfusion of red blood cells should be repeated approximately every 15-20 days for a lifetime. They can be paler than the others, and to practice before a therapy based on iron must ensure that the value of the iron in the blood is very low (serum iron). Can still live and do it all without any risk. Transfusions, however, inevitably involve an excessive intake of iron, which eventually accumulate in major organs: the heart, the endocrine glands and the liver. All this undermines the functions. The only way to avoid this detrimental accumulation of iron is to make a proper treatment with medication (iron chelator). The disease does not have a truly effective therapy, although today the thalassemic live a much more normal for improvements in treatments. The extension and improvement of the living conditions of people with thalassemia affect much on the attention that they have against the disease, and consequently in living life. The purpose of the research is to test this hypothesis that some studies show: the positive attitude of thalassemia is linked not only to the change in the prognosis and treatment strategies, but also psycho-social medical intervention that some facilities today are able to offer.[5]

Materials and Methods

In Sicily there are 20 centers for the treatment of thalassemia, one of which is UOC Center Genetics and Immunology Pediatric. The other is in the province of Messina, Sant'Agata di Militello PO. The total of approximately 130 patients in Messina province, and of these about 70 treated in UOC. The present research was a cross- sectional (descriptive, analytical) one. This study has been conducted on 36 patients with thalassemia syndromes (18 men and 18 women) by the UOC of Genetics and Immunology Pediatric Department of Messina with Prof. Salpietro and Dr. Piraino, in collaboration with the Pediatric Clinic University of Milan Bicocca with Dr. Masera, and the Department of Human and Social Science, University of Messina with Prof. Cammarota and Dr. Raffa. For these reasons the cross- section can be considered representative of the reference universe, despite the smallness. The participants of this study were the individuals on the basis of the frequency of transfusions, by a seventh to maximum of three weeks. The questionnaire consists of 68 questions (closed) and 96 variables. All respondents agreed voluntarily to participate in research, after being fully informed about. It was also guaranteed their anonymity and the rules of privacy law were respected. It was also ensured that the data were communicated or disseminated in anonymous and aggregate statistics. The detection was made with unanimous opinion of the Ethics Committee. A detailed chart review was undertaken to define the relationships between socio-demographic conditions and epidemiological conditions. This research examined the demography and natural history of all patients with

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thalassemia who are registered in this Center. To facilitate research, it was developed a cross-sectional registry to characterize the demographic and clinical features of Messina patients. Data were included from all living patients who gave informed consent and were diagnosed with β-thalassemia major, or β-thalassemia intermedia, obtained by using a self-compile questionnaire. Data were collected once for each subject from June 2010 to January 2011. The data were analyzed through the StatSoft statistical software (StatSoft, Inc., Tulsa, OK, USA). In addition, descriptive results were presented in graphs and charts in the form of numbers and percents. Moreover, in order to compare the quantitative variables, two-sample t-tests were run.

Results

Thirty-six enrolled patients completed collection of the registry data: 77% of the patients with β-thalassemia major, 23% of the patients with β-thalassemia intermedia. A total of 63% of the patients who come to the Center were born in Messina city, 11% were born in Milazzo and 8% in Taormina (the remaining 3% in Africo, 3% in Locri, 3% in Barcelona, 3% in Rivoli, 3% in Palermo, 3% in Brolo). The median age is 36 years. Thirty-six percent of patients were older than 40 years. Moreover, 44% of the patients who referring to the center have high school education and 17% have university education. Also 59% is nubile/bachelor, 32% married and 9% separated (Figure 1). Among married and separated on 74% have children. The correlation between diagnosis and marital status was statistically significant (P=0.04). The 29% of the patients believe that the disease conditions (conditional or not having) nothing for the possibility of finding a job, 11% a little, while the majority, or 40% thought so as well. Instead, the 9% and 11% of patients believe that the disease condition (or has affected), respectively, very and very much the opportunity to find work. The correlation between diagnosis and work was statistically insignificant (P=0.27). Forty percent of patients are unemployed, 20% are employed, 11% seeking employment, and 9% retired (Figure 2). Twenty-five of employees said that the disease has influenced the career choice, while the remaining 75% say the opposite. Start transfusion for the 90% is started between 0 and 5 years. Fifty-six of the patients stated that they do transfusions every 3 weeks; 8% every 15 days. According to many studies, transfused patients may develop complications related to iron overload. Later iron overload-related complications include involvement of the heart (dilated myocardiopathy or rarely arrythmias), liver (fibrosis and cirrhosis), and endocrine glands (diabetes mellitus, hypogonadism and insufficiency of the parathyroid, thyroid, pituitary, and, less commonly, adrenal glands).[6] Eleven percent of patients claimed to suffer from heart slightly. There is 69% of patients say they practice sports, all at an amateur level. The correlation between heart problems and sport was statistically significant (P=0.1789). Among other complications at 33% there are those who contracted hepatitis C virus, and another 33% problems with the liver/hepatitis C. Survival of individuals who have been regularly transfused and treated with appropriate chelation extends beyond age of 40 years. Fatal in the absence of modern medical care, thalassemia is associated with extended survival with adherence to transfusion and effective iron-chelation regimens.[7] Thalassemia has become a chronic adult illness with a median life span approaching 40 years. For this reasons it's important that these complex standard-care guidelines will require a close working relationship between thalassemia centers and the growing number of patients. Patients have also expressed an opinion with respect to the perception of the services provided by the Centre regarding the relationship with the staff that works there daily. More than half of the patients said that doctors and nurses of the center are very much available: to discuss their health problems; to give information about the disease and treatment; to be interested in a lot of their prob-

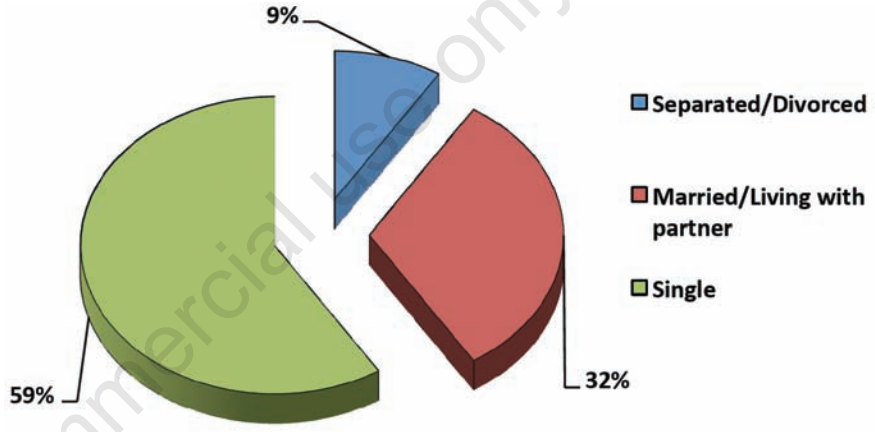

Figure 1. Marital status of the patients interviewed (%).

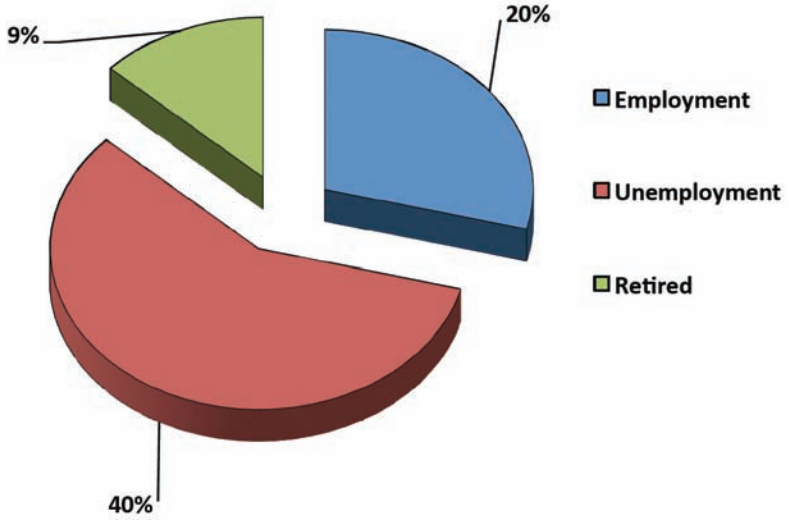

Figure 2. Employment and unemployment of the patients interviewed (%).

lems (Figure 3). Only 3% of patients reported a lack of willingness on the part of doctors and nurses for times of sampling and transfusions. Eleven-one of patients felt very much tiring to follow the treatment, 17% very tiring, 31% on average, just 22%, and the remaining 19% not at all tiring. The cross section is generally satisfied in the relationship with the doctors and nurses. While presenting a general tendency to satisfy an element of dissatisfaction seems to be the lack of professionals such as psychologists and social workers. Among 78% of patients claimed to have turned to a psychologist, 22% never went. But, 59% of patients believe that the figure of a psychologist at the Centre of Care is useful; 6% instead considers it unnecessary, while 35% do not says (Figure 4). Eighty-eight of patients states to become members of the family association, and of these, 6% actively participate very much, 16% much, 42% on average; while 32% are less active. For more than 60% of patients it is important to have friends and a wild economic stability, while for less than 50% of patients it is important to have a job and children (Figure 5).

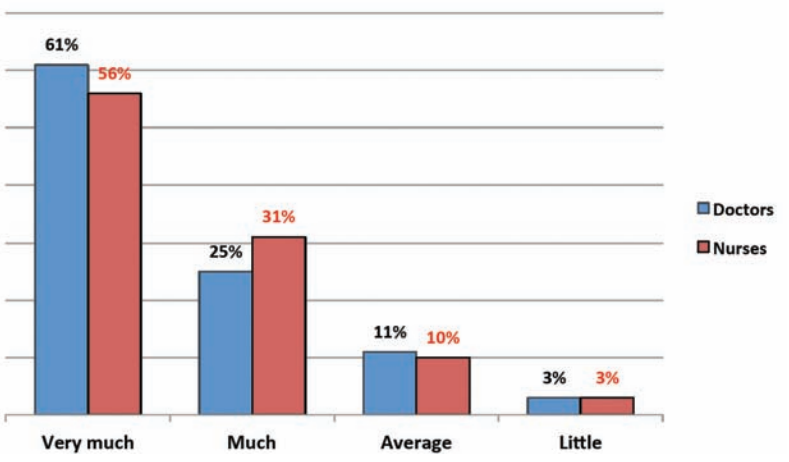

Figure 3. Availability of doctors and nurses (%).

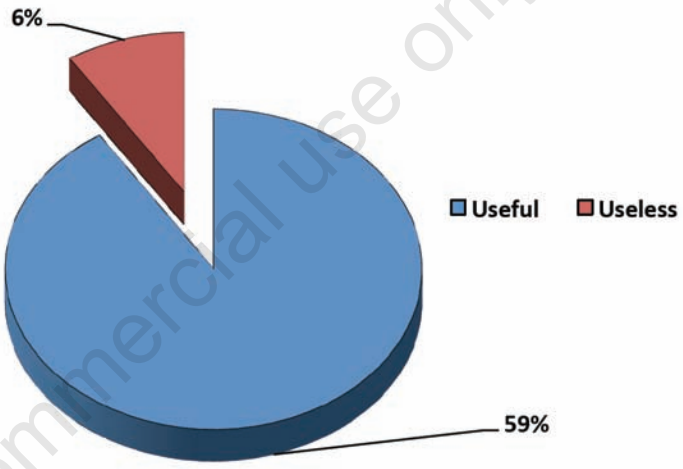

Figure 4. Usefulness of the figure of a psychologist at the Centre of Care (%).

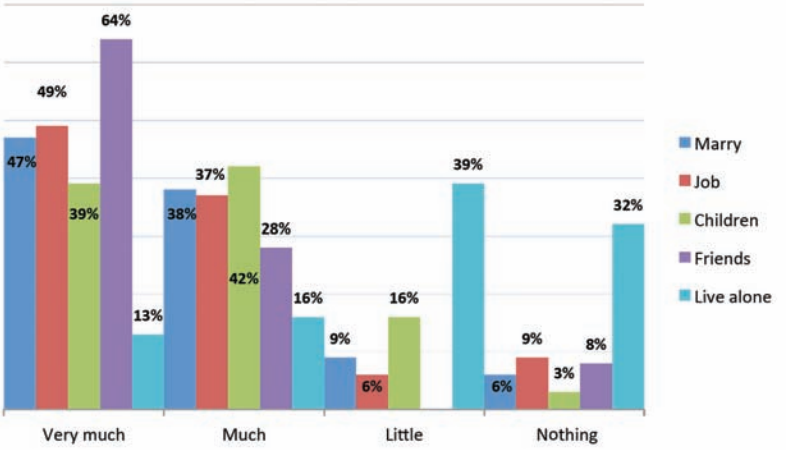

Figure 5. Importance of: being married; finding a job; having children; having friends; living alone (%).

Conclusions

The cure of thalassemia has evolved considerably over the past thirty years. In the opinion of 83% of patients own state of health is good. Thalassemia is less frightening. In Messina, the quality of adult life is satisfactory both in relation to the physical conditions both personal accomplishments in the field of work and emotional level. This gradual improvement should be considered an important result, as compared to situation of other chronic diseases. Today, living with this disease is possible, thanks to new therapeutic options and diagnostic supports that have significantly improved the quality of life for patients. Firstly, survival of individuals who have been regularly transfused and treated with appropriate chelation extends beyond age of 40 years. Fatal in the absence of modern medical care, thalassemia is associated with extended survival with adherence to transfusion and effective iron-chelation regimens. Secondly, a strategic element that emerges from the interviews is the need for a strengthening of nurses and other medical specialists, such as psychologists and social workers. The creation of a multidisciplinary team can be a boon for patients and their families, who are helped by considering their medical needs, but also psychological and social. This research shows clearly the importance of the relational dimension and collaborative that develops between staff and patients. Support and confidence are combining the system of care with the social system of the patient.

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