Comprehensive healthcare for individuals with sickle cell disease – a constant challenge

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Sickle cell disease (SCD) is one of the most common genetic disorders in Brazil and in the world. It is a chronic inflammatory disease permeated with manageable acute clinical events.[1,2]

Advances in the knowledge of molecular, cellular and clinical aspects of the disease associated with early diagnosis and treatment have reduced morbidity, increased survival and improved the quality of life of SCD individuals.[3,4]

Neonatal diagnosis, prompt treatment (vaccines, prophylactic penicillin) and guidance in respect to early recognition of splenic sequestration by mothers and caregivers have contributed to a reduction in the mortality rate of children in the first 5 years of life. Two other important factors were the identification of children at high risk for strokes with the adoption of a regime of chronic red blood cell transfusions aiming at preventing this serious complication, and the diagnosis and treatment of acute chest syndrome. These two conditions are currently the main causes of death in adolescents and young adults.[4-8]

The introduction of hydroxyurea (HU) has also had an impact on the quality of life of SCD patients by reducing the number of vaso-occlusive crises, the number of hospitalizations, length of hospitalization and the occurrence of acute chest syndrome as well as noticeably reducing the mortality rate of SCD patients treated with HU compared to those that do not take the drug.[9]

Blood transfusions have been increasingly used as a therapeutic resource, in part because of improvements in the safety of this procedure, but above all, because transfusions prevent serious complications. Recently, allogeneic peripheral stem cell transplantation, which is the only therapeutic option that provides a cure, has been indicated for this group of patients, especially for those with more severe disease in the first years of life and who have compatible donors.[8]

In Brazil, the first step toward the construction of comprehensive healthcare for SCD patients was with the establishment of neonatal screening in the National Health Service by the Ministry of Health on January 15, 1992, with testing for phenylketonuria and congenital hypothyroidism (Phase 1).

In 2001, with Decree 822/01 of the Ministry of Health, the National Neonatal Screening Program was extended to include screening for hemoglobinopathies (Phase 2).[10]

The addition of hemoglobin electrophoresis in neonatal screening represented a major step forward in the recognition of the importance of the hemoglobinopathies as a public health problem in Brazil and also the beginning of the change in the natural history of the disease in our country.[1]

Associated with the genetic aspect, the social conditions of SCD patients have direct implications that greatly affect the biological condition and decisively contribute by aggravating the disease. As a rule, SCD patients have low socioeconomic conditions, bad schooling and difficulties to access healthcare services; the great majority is attended by state health services. This explains the unquestionable difficulties they face in their daily lives, which gives them a condition of greater social vulnerability.[8,11]

Neonatal diagnosis is fundamental, but not enough. We know that the diagnosis needs to be linked to a wide-ranging support program for SCD individuals which includes: easy access to quality medical services, an appropriate multidisciplinary approach to treatment with prevention of acute and chronic complications, continual education programs about the disease for different medical specialists and professionals in healthcare services, adequate genetic counseling, availability of educational activities for patients and families about the disease and specific programs to improve the patient's socioeconomic conditions.[3,4,8]

In general, the clinical approach to SCD patients does not require complex or costly procedures. Up to the fifth year of life, the period of the highest rates of death and serious complications, prophylactic care is basically the essence of treatment. The follow-up and adequate treatment of these patients in state health clinics determine a better or worse prognosis following the occurrence of acute events.[8,10,11]

In spite of all the endeavors of the Ministry of Health, through the Coordenação Geral da Política Nacional de Sangue e Hemoderivados to regulate and implement the measures established by Decree 1391, which provides, within the National Health Service, guidelines on the national policy for comprehensive healthcare for people with SCD and other hemoglobinopathies, we are far from reaching our goals. These, in particular, include the organization of a care network for SCD patients in all Brazilian states and the development of concrete measures, especially with regards to the enrollment of these patients in ongoing governmental programs and the education and training of the healthcare team needed to prevent disease by identifying clinical complications at an early stage, which will help to reduce morbidity and mortality rates and improve the quality of life of SCD individuals in Brazil.[10]

The study by Santoro et al.,[12] published in this issue, points out some of the weaknesses in the methodology applied, which are inherent to a retrospective study, but it certainly has the merit of portraying how SCD patients are being cared for in the State of Rio de Janeiro. This study confirms the precariousness and vulnerability of care for these patients in state health clinics and the overuse of specialized services for clinical situations of low complexity. It also emphasizes the need for concrete measures with the objective of decentralizing healthcare, safeguarding the specialized centers of medium and high complexity as referral units for urgency and emergency care.

The issue of comprehensive healthcare for SCD individuals continues to be a political, social and economic challenge which requires the support of all: government bodies, referral centers, healthcare programs of the child, the woman and the family, healthcare professionals, patients and their representatives. We have come far, but we need to improve even more.

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