CHAPTER
15

Comprehensive health care for children and adolescents with congenital disorders: The impact of the COVID-19 pandemic on care

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ABSTRACT
Congenital anomalies are structural or functional changes that occur during intrauterine life. Approximately one in five deaths in the neonatal period are due to birth defects in Latin America and the Caribbean. There are several factors for their development, from infections, maternal nutritional status, use of medication, and environmental or genetic factors. Most anomalies are present in low-income countries where we have an inadequate prenatal context, a greater predisposition to infections due to precarious health, and the use of prohibitive substances (medicines and drugs) during pregnancy. From this, we have that several social determinants of health such as social, economic, cultural, ethnic, psychological, and behavioral factors influence the course of a disease in a certain population. When there is a system that integrates health care for children and adolescents with congenital disorders, including genetic syndromes, especially Down syndrome, care for children promotes better growth and development. A comprehensive health care service for children and adolescents inserted in a university hospital must be able to coordinate care actions. During the years 2020 to 2022, health services faced the covid-19 pandemic that the entire population experienced. There was an impact with a reduction in the number of daily visits to these patients and a reduction in the inclusion of new ones, leading to fragmentation or discontinuity of care.

Keywords: Congenital Abnormalities, Genetic Diseases, Comprehensive Health Care, Children, Adolescents, Covid-19.

1 INTRODUCTION

1.1 CONGENITAL MALFORMATIONS, DOWN SYNDROME AND HEALTHCARE

Birth defects or congenital malformations have significant public health impacts, affecting 3% to 4% of all live births and causing elective terminations of pregnancy or miscarriages. In the United States, approximately 150,000 babies are born each year with birth defects, these and genetic diseases being the main causes of infant deaths and important morbidities during childhood. Although they represent 15% to 30% of all pediatric hospitalizations, these children generate proportionately higher costs than other hospitalizations [1].

The overall incidence of birth defects in Latin America does not differ significantly from that found in other regions of the world [2]. In general, it can be considered that no less than 5.0% of live

births have some developmental anomaly, determined, totally or partially, by genetic factors, which will compromise their development and quality of life. Adding the disorders that manifest later, such as certain chronic degenerative diseases, the considerable effect that genetic factors have on health is even more evident [3,4].

In Brazil, an ecological study carried out by Siedersberger Neto et al. [5] analyzed deaths in children under one year old due to congenital anomalies between 1996 and 2008 and found that the proportion of infant deaths attributable to congenital malformations rose from 9.74% in 1996 to 18.22% in 2008, with an average annual increase of 0.71%, statistically significant values. In the year 2000, congenital anomalies were the third cause of infant mortality in Brazil, accounting for 13% of deaths [4]. In developing countries, diseases of predominantly genetic etiology account for 15 to 25% of the causes of infant mortality [6].

According to NUSSBAUM et al. [7] genetic factors are implicated in virtually all diseases, with a greater or lesser relative role depending on their interaction with the environment. Among the diseases in which the genetic component is preponderant, we can mention the monogenic, individually rare, but affecting as a group 2.0% of the general population; chromosomal, present in 0.7% of live births (and in half of the spontaneous abortions); and the multifactorial, responsible for most of the congenital malformations and also for many common problems of adult life.

Down Syndrome (DS) is a genetic alteration present in the human species since its origin. It is a chromosomal disease characterized by the presence and expression of three copies of chromosome 21. Its first description as a clinical condition with its own identity occurred in 1866, by the English pediatrician John Langdon Down. In 1958, the Frenchman Jérome Lejeune and the Englishwoman Pat Jacobs independently discovered the chromosomal origin, starting to be considered a genetic syndrome, naming it Down Syndrome or Trisomy 21 (T21), in homage to John Down who initiated the theme in the scientific environment [8,9].

DS is the most frequent chromosomal abnormality in humans [10,11]) and the most prevalent cause of intellectual disability [12]. People with DS have the potential to develop due to the broad neuroplasticity that can be stimulated from the first months of life by the family, health professionals, and education [13].

The National Down Syndrome Society (NDSS) reports that approximately one in every 700 babies in the United States is born with Down syndrome, about 6,000 per year [14]. No different, in Brazil, it is estimated that for every 700 births, there is 1 case of trisomy 21, totaling around 300,000 people with the syndrome, regardless of ethnicity, gender, or social class [15].

It is, therefore, a significant part of the Brazilian population, consisting of an important portion that may require timely interventions from the earliest childhood, as this chromosomal disorder is

related to organic changes and specific physical constitution and, a greater risk for developmental delay.

However, despite the impossibility of predicting the degree of autonomy that a child with DS will have in adult life, it is known that, when properly cared for from birth, and receiving adequate guidance and stimulation, they have the potential to lead a healthy and fulfilling life. social and educational inclusion [16]. This statement is also valid for all children born with congenital conditions.

The Ministry of Health's Guideline for Attention to People with DS, updated in 2020 by the Brazilian Society of Pediatrics, recommends that care for this population is guided by public policies and that the theoretical assumptions of the expanded clinic, integrality, and care be used. shared. It brings as a proposal for care conducts such as the expanded understanding of the health-disease process; construction shared by the multidisciplinary team of the situational diagnosis; and the individual care plan; definition of therapeutic goals and commitment of professionals, family, and individual with these goals [16, 17].

According to Roseni Pinheiro, "health care" is not just a level of care provided by the health system or a simplified technical procedure, but an integral action that has meanings and meanings aimed at understanding health as the 'right to be'. Through interdisciplinary work and the articulation of professionals, managers, and users, with active participation, care can be expanded and the social support network strengthened. Comprehensive health care is opposed to an economicist view and is concerned with the well-being of individuals, making them capable of judging what their health needs are, placing them as other subjects and not objects [18].

The Unified Health System (SUS) emerged in Brazil from a history of struggles by the Brazilian health movement to facilitate health care [19]. Comprehensiveness in the Unified Health System (SUS) should be widely applied to everyone, and I think especially to children and adolescents with congenital conditions, to promote a timely stimulus for their full growth and development. But also to prevent complications and carry out periodic investigations to detect comorbidities typical of malformations and congenital alterations or genetic syndromes [6].

Thus, congenital anomalies and genetically determined diseases represent an important public health problem in Brazil, and the understanding of their etiopathogenic mechanisms, their diagnostic elucidation, and comprehensive assistance to those who present these alterations and diseases should deserve special attention. The SUS, with a focus on this concern, instituted the National Policy for Comprehensive Care in Clinical Genetics in 2009, through Ordinance MS/GM nº 81 (BRASIL, 2009), but the Health Care Secretariat (SAS) did not disclose the necessary measures for the full implementation of this Policy. Thus, there is still a very large gap in the care of those with congenital and genetic conditions in Brazil [20].

The number of specific programs aimed at detecting these diseases and acting in health promotion and disease prevention is still very scarce in Brazil. What can be seen is that, as there is no broad structured public assistance for these patients, comprehensive care and the Health Care Network (RAS) have not been effective. It is frequent that non-governmental and philanthropic organizations, or even the private sector, end up taking on this care, which should be primarily the responsibility of the SUS [21].

Due to the capillarity of the UBS network, these actions must take place from the care provided in Primary Care, the main gateway to the SUS, with support from the RAS. However, for this to occur with quality, it is essential to train general professionals, from graduation and specialization, both in technical-scientific knowledge and in relational technologies, as proposed by Merhy [22]. The existence of medium and high-complexity Units and their integration into a regionalized network should guarantee support to the UBS and adequate care for the different needs of each patient.

Reinforcing this Comprehensive Care Policy, in 2014 the Ministry of Health instituted, through Ordinance 199/2014, the National Comprehensive Care Policy for people with rare diseases, which includes an axis for genetically determined ones. However, little progress has been made toward the recognition and adequate treatment of these people in the SUS [23].

1.2 THE COVID-19 PANDEMIC AND CHILD VULNERABILITY

In January 2020, the World Health Organization declared an international public health emergency with the worldwide spread of covid-19, starting the coronavirus pandemic that lasted until the beginning of May 2023, when after more than three years, the World Health Organization (WHO) declared in early May 2023 that Covid-19 no longer constitutes a public health emergency of international importance. According to the entity, the virus is now classified as an “established and ongoing health problem” [24,25]. In this scenario, the political response directed at the pediatric age group in the face of the pandemic is a constant concern, as these are individuals in a more vulnerable situation [26]. And, because the initial concern was with the disease itself, and with the multisystem inflammatory syndrome, much greater than with the care, follow-up, and impact on the growth and development of these children, demonstrated by the protocols at the time, which focused on this condition.

Also with the pandemic, there was a rupture in vaccination follow-up, both due to the overload of the health system due to covid-19 and the drop in the demand of families to vaccinate their children, whether due to social distancing or due to the very reluctance of communities in the face of the current scenario. This leads to increased susceptibility to vaccine-preventable diseases that have epidemic

potential. Children constitute an important group of people vulnerable to vaccine-preventable diseases and may suffer increased morbidity and mortality, further burdening health services [27].

The pandemic impact with social isolation measures, devastating economic damage, and closure of schools and so-called "non-essential" services also impacts access to health and how the relationship between health professionals and patients takes place, directly affecting basic health and prevention and health promotion measures, which with the pandemic gave way to care for patients infected with COVID 19 and its complications. Support networks, schools, physical activity, and social interactions were suddenly removed from children's daily lives. At the same time, routine consultations in pediatrics gave way to priority and urgent health care or those infected with SARS-COVID, and the health promotion measures so hard won and built in our country were distancing themselves from the reality of care in health for this age group [28].

2 FINAL CONSIDERATIONS

Childhood is a period of great vulnerability and adaptation to the environment in which the subject is inserted, therefore requiring continuous health monitoring that allows the promotion of good conditions for growth and prevention and treatment of diseases in this age group.

The covid-19 pandemic has emerged as an additional obstacle, important for the delay in the diagnosis and follow-up of patients with congenital anomalies. Many patients depend on public health services which changed mainly in the years 2020 and 2021 due to the pandemic and the need to face it. Schools and closed medical outpatient clinics, Basic Health Units, and health professionals (doctors and physiotherapists, among others) were redirected to the high demand of respiratory cases and led to the loss of follow-up of certain patients who need long-term follow-up to better growth and development.

Thus, this reflection on access to health and child care in health services is urgent and necessary, with the many new challenges brought by the pandemic and with the establishment of a new reality for everyone – patients and the health team.

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