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Prevalence of People with Down Syndrome in Brazil

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Down Syndrome (DS) is one of the most prevalent chromosomal changes, and therefore, deserves the study of chromosomal abnormalities. In Brazil there is no exact number for the quantity of people who have SD, only projections. Considering that better life and more efficient public policies aimed at those with SD directly depend on reliable data about its actual population, it is clear that there is a major deficiency in how they are surveyed and analyzed the information on the Brazilian population with SD.

Keywords: Down syndrome; population;data

Síndrome de Down (DS) é uma das mudanças mais prevalentes cromossómicas e, portanto, merece o estudo de anormalidades cromossómicas. No Brasil não há um número exato para a quantidade de pessoas que têm SD, apenas projeções. Considerando que a vida melhor e mais eficientes políticas públicas que visam as pessoas com SD dependem diretamente de dados fiáveis sobre a sua população real, é claro que há uma deficiência importante na forma como eles são pesquisadas e analisadas as informações sobre a população brasileira com SD.

Palavras-chave: Síndrome de Down; população; dados

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1. INTRODUCTION

Down Syndrome is one of the most prevalent chronic chromosomal abnormalities, which affects equally individuals of any geographic region, race and gender [1].

It is a chromosomal alteration characterized by the birth of a person with 47 chromosomes and not 46, as expected, in other words, the child has an extra chromosome among its millions of cells. [2].

People with Down Syndrome exhibit very particular physical characteristics (phenotype), which generally allows its identification on the moment of birth (which does not mean that further confirmation is not important). The main physical characteristics are short stature, microcephaly, flat profile, eyes with oblique palpebral fissures, small low set ears, clinodactyly (curvature) of the fifth fingers, increasing the distance between the first and second toe and fold only on the palms, protruding tongue (projected out of the mouth) and grooved, among others [3].

The person with Down Syndrome has three unique characteristics: phenotype, hypotonia and intellectual commitment, but the hypotonia, present in 99% of cases, determines the delay in psychomotor development. The baby takes longer to control the head, rolling over, sitting, crouching, crawling, walking and running and, because of this, the exploitation of the environment can be compromised [3].

However, not all patients with DS show these phenotypes, but the mental disability would be the only feature present in all cases. The identification of people affected at birth, provides early intervention, extended to all families involved [4].

With increasing population DS in Brazil, it is necessary to the country be better prepared to welcome these people. There must be a correct and accurate survey of the quantity of people with Down Syndrome in Brazil and its states. Thus it would be possible to base actions and policies in order to give appropriate care to people with Down Syndrome and therefore promote their citizenship.

The main feature of the genetic disease is extremely important, not only for the index patient, but also for future members of the family [5].

In this study, it was investigated how is made the survey of data on the Brazilian population with Down Syndrome.

2. MATERIALS AND METHODS

The study was conducted through analysis of other studies conducted by different authors, records of public and private institutions that serve people with Down Syndrome and field survey in Aracaju.

In these institutions it was raised the addresses of the DS patients registered in their records, since the initial operation of the entity. Registrants have been visited and their caregivers were asked to indicate other known people with DS and how to contact them. This procedure was repeated in order to do the survey, as more comprehensive as possible, with people with Down Syndrome living in Aracaju. To avoid duplication of location, the result of new information was filtered. The procedure was performed until all entities and persons registered in all listed by caregivers, since living in Aracaju, were sought.

3. RESULTS E DISCUSSION

About 5% of all pregnancies in Brazil result in the birth of children with congenital anomalies that hinder its development. Considering DS one of the most prevalent anomalies, this number appears to be quite significant. In developed countries, disorders of wholly or partly genetic causes account for 36% to 53% of admissions to pediatric hospitals. In developing nations, according to the still high prevalence of infectious and nutritional diseases, congenital anomalies / genetic disorders are between 15% to 25% of the causes of prenatal mortality and child, but these numbers are increasing. [5].

Analyzing initially the prevalence of DS in the Brazilian population, the pediatrician and geneticist Zan Mustacchi says that in Brazil, about five thousand children are born with the syndrome each year and in South America a baby with Down Syndrome is born among 600 live births. This prevalence of 1 / 600 live births would have been observed in Brazil over the past decade, responsible for the frequency of 1.7 per thousand in the whole population with some type of disability [6].

The estimated frequency of DS in a study from 2003 was also about 1 in 600 live births. The study also mentions that in populations where women have children until the end of their reproductive life, the incidence of births of children with chromosome disorders is up to 6/1.000 and that the proportion of live births with Down Syndrome is about 3/1.000, in other words, it is perceived that the universe of chromosomal defects, DS appears with a much higher incidence. However, there are conflicting results in this study, as it is shown as overall estimate the proportion of one person with Down Syndrome in every 600 live births, but by analyzing the relationship with maternal age it is shown an incidence of 3 people with the DS per 1000 live births, which are quite different results, in addition to not being shown how this calculation was made and neither the age of the mothers studied [7].

Maternal age would be one of the major factors related to the appearance of birth defects. According to a 2008 study, the ideal reproductive age of the mother is between 20 and 34 years old, and both ends of the distribution would be at risk for birth defects. In 20 years old mothers in the risk of emergence of any chromosomal abnormality would be 1:526 births, at 30 would be 1:384, and at 45 the risk would rise to 1 in 21 births. In the case of DS, the risk for a 20 years old mother would be 1:1.667 births, increasing to 1:952 at age 30, and 1:30 at age 45 [5].

For a 2009 study, DS is indicated as a genetic condition that would affect one in every 800 newborns in the world and considered the main genetic factor in the development of intellectual disability. In Brazil, would be born about 8000 people per year with DS, which is the most commonly diagnosed genetic alteration in the pediatric clinic. Due to improvements in family care, medical and rehabilitation in childhood, the life expectancy of patients with this syndrome has increased in recent years. It is considered that about 60% of the population live more than 50 years, 44% will reach 60 years and 14% at 68 years [8].

According to a 2006 study using data collected by the IBGE Census from 2000, there were approximately 300,000 people with Down Syndrome in Brazil, with a life expectancy of 50 years old. This statistic is similar to world data. [9].

The DATASUS data from 2007 revealed the birth of 3,035,096 people in Brazil in 2005, therefore, it is estimated that only in that year 5,058 people were born with DS and that about 300,000 Brazilians have the syndrome. The Information Technology Department of the Health Care System (DATASUS) is an agency of the Executive Secretariat of the Ministry of Health, which has, among other skills, research and incorporate technologies that enable computer systems implementation and dissemination of information necessary to the health actions, in agreement with the guidelines of the National Health Policy. [10].

Here we can see an inconsistency in the data, because the projection from IBGE and DATASUS is the same (300,000 people with Down Syndrome in Brazil), but one of the surveys was conducted in 2000, while the other was held in 2007.

The pediatrician and geneticist Zan Mustacchi, head of the Genetics Department in the State Child Hospital Darcy Vargas and responsible for handling 20% of the population with the syndrome in Brazil, also said that there are 300 000 people with SD in Brazil, and this survey is even more recent than the achievement from DATASUS in 2007, but has the same final projection [6].

According to the World Health Organization (WHO), the prevalence of DS in Brazil would be between 1 and 2% of its population. Here it is possible to see perhaps the greatest discrepancy in the data. According to the last IBGE census, conducted in 2010, there are 190.732. 694 inhabitants in Brazil today. In other words, according to the WHO, the population of people with Down Syndrome in Brazil would be between 1,900,000 and 3,800,000 people [11].

In the field survey conducted in Aracaju it was not possible to confirm the proportion of patients with DS established by WHO. Considering that in 2010 the population in Aracaju was 552,365 inhabitants [12], we would have 93,902 people with Down Syndrome (0.17%), considering the proportion of 1 / 600 [6,7]. If it were considered the proportion of 3 / 1000 [7], we would have 165,709 people with Down Syndrome (0.3%) and, if it were considered the proportion of 1 / 800 [8], we would have 69,045 people with Down Syndrome in Aracaju (0.125%).

However, even seeking information on specialized care services for patients with DS, like the Exceptional Parents and Friends Association (APAE), and even private clinics in serving people with special needs, it was possible to interview only 44 families of people with SD in Aracaju.

A previous study shows that the life expectancy of DS patients has been increasing in recent years due to improvements in family care, medical and rehabilitation in childhood [7]. In Aracaju it was not possible to reach a conclusion about the increase in life expectancy of patients with DS, since we did not find a significant portion of 40 years old or older patients with DS =, taking into account that the 2009 study says that about 60% of SD population live more than 50 years [7]. But it was possible to verify that there are indeed a greater family care, and a greater demand for medical treatments and procedures related to the early stimulation of patients with DS, particularly in families with higher family incomes and more education.

4. CONCLUSION

This study suggests that care for DS patients is not effective and it is necessary more efficient and effective public policies for this population. There is a great deficiency of scientific studies on the DS Brazilian population, that is not adequately known in its volume and main characteristics, which need to be revised to provide the necessary knowledge to the foundation of policies and actions for the effective inclusion of these people, which constitute a significant portion of the population.

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