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ORIGINAL ARTICLE

Congenital disorders prevalence in a neonatal ICU in the south of Rio Grande do Sul

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Abstract

Congenital disorders are structural or functional anomalies present at birth. This study aims to identify the most frequent birth defects in a neonatal intensive care unit (NICU) over a period of five years. A retrospective cross-sectional study was carried out by reviewing patients files admitted to the neonatal intensive care unit. Data collection was done through standardized charts and then analyzed using Excel program. Sixty children were included in the study, representing 8.6% of hospitalized children in that period. Congenital anomalies were present in male patients (57.33%), preterm (39.33%), and aged between 0 and 7 days (55.7%). Regarding etiology, multifactorial inheritance was responsible for more than 50% of disorders. Gastrointestinal (33.3%) and cardiac (28.3%) anomalies were the most prevalent among them. Down syndrome was the most frequent chromosomal syndrome (11.6%). Given the prevalence and importance of these disorders, this study suggests a clinical geneticist as a consultant or a member of neonatal ICU team is necessary to neonate investigation and to offer an appropriate genetic counseling to families.

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INTRODUCTION

Congenital defects (CD) are anomalies present at birth, and they can be morphological or functional, genetic and-or environmental[1]. They encompass all the functional or structural anomalies arising from factors associated to the time before birth, being that genetical, environmental or unknown. They may be already apparent in the newborn, or manifest later[2-4]. In Brazil, we estimate that about 3-5% of all newborns have some type of these defects[1]. The incidence of congenital anomalies in Rio Grande do Sul, varies between 10 and 15/1,000 live newborns[5]. Congenital defects are associated with high morbimortality, representing a very high cost for public healthcare[6,7]; thus, the importance of surveillance for prevention and diagnostic purposes. Since 2005, congenital malformations and chromosomic anomalies make up the second cause of pediatric mortality in all Brazilian regions, corresponding to about 20% of deaths[2,8,9]. In Rio Grande do Sul, CD represent the second cause of pediatric mortality[10,11].

Malformations correspond to 25.6% of pediatric causes of death in neonatal intensive care units (NICU)[4]. In the city where the study was carried out, the number of fetal and infant deaths associated with the chapter XVII of the International Code of Diseases (congenital malformations, deformities and chromosome anomalies), corresponded to 14% in the year of 2014 and, among these, 33% happened during the neonatal period only[12].

This study aimed to report on the frequency of congenital defects and their etiology in the neonatal ICU of a University Hospital.

METHODS

The city of Rio Grande boasts about 210,000 dwellers, and it is in the south coast of Rio Grande do Sul, at about 350 km of Porto Alegre - State Capital. The main sources of income are the chemical industry, fishing and port activities[13]. This city has a University Hospital, which has a Neonatal ICU that serves 22 nearby towns. We ran a cross-sectional study, reviewing the charts from patients admitted to this NICU from January of 2006 through March of 2011. As for inclusion criteria, we considered all the cases of suspected genetic disease - according to Chapter XVII of the International Code of Diseases (congenital malformations, deformities and chromosomal anomalies), included in the unit's admissions logbook. We excluded those cases in which the diagnosis of genetic disease or congenital defect was ruled out after a pediatric and specialized clinical investigation, including genetic evaluation, gestational assessment, sex, city of origin, reason for admission, dysmorphological diagnosis (isolated defect, syndrome, sequence or association), etiological diagnosis (Mendelian, chromosomal, multifactorial or inconclusive) and case outcome, identifying whether the patient remained under investigation in the service, if there was loss of follow-up or if genetic follow-up was discharged. After collection, we entered the data into the Excel software for processing.

The archived medical records were accessed through the Medical Archive and Statistics Service (SAME) of the institution after the hospitalization of the patients. Regarding genetic research during the period evaluated, common imaging and clinical analyzes were performed at the hospital itself and cytogenetic and/or molecular tests were carried out in partner laboratories.

The present study was approved by the Institution's Health Research Ethics Committee.

RESULTS

During the study period, 697 newborns were admitted to the Neonatal ICU of the HU-FURG, 97 with initial suspicion of congenital defect and/or genetic disease. Of these, 37 were excluded from the study; there were 23 deaths during hospitalization, for which there was no genetic evaluation in time and whose medical records could not be evaluated; 9 medical records with incomplete information and 5 for not presenting a disease of confirmed genetic etiology, totaling 60 cases in the present study.

Table 1 shows that there was a predominance of males and that most hospitalized children were full term. Table 2 shows the diagnosis by etiology. Most of those included in the study had problems of multifactorial origin. Among these, gastrointestinal malformations accounted for 33.3%; 95% of them (n=19) in isolation, and 5% (n=1) accompanied by other defects (data not shown). Cardiac malformations accounted for 28.3% and were subdivided into isolated (41.1%), multiple cardiac anomalies (41.1%) or cardiac anomalies accompanied by defects in other organs (17.6%), as shown in Graph 1.

DISCUSSION

The prevalence of CD among patients admitted to the NICU was 8.6%. Such prevalence is higher than that described in the general population, 3-5%[1]; however, like the one reported by other authors[14,15]. In addition, this is the inpatient unit that receives high-risky patients and is therefore expected to concentrate such problems. In a time, series analysis from Rio Grande does Sul, between 2005 and 2014, the highest prevalence of CD was associated with maternal age higher than or equal to 40 years of age, black or brown skin color and having less than 3 prenatal consultations[5], teenage pregnancy, self-medication, smoking, unplanned and unwanted pregnancies, as well as abortion attempts, consanguineous unions[4] and conditions of exposure to biochemicals from agriculture[16].

The sex most affected by malformations was male (57.33%), in agreement with previously reported data, 64%, 51.9% and 58.2%[17-19], respectively.

In the study, most of the patients in the NICU - who made up the sample - had gestational age at term (55%), despite the prevalence - estimated by DataSUS - of premature infants having increased between 2000 and 2009, and this is, one of the indicators of increased demand in the indication of intensive care for newborns in the country[20].

Table 1. Distribution of birth defects in the Neonatal ICU according to sex, gestational age, origin and age upon admission.

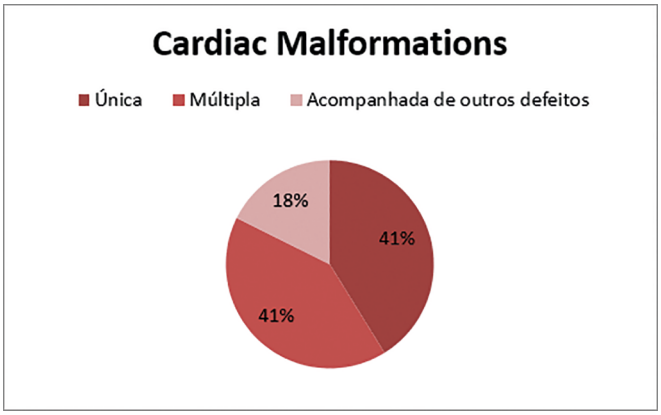
Sex	N	%
Males	35	57.3
Females	25	42.6
Gestational Age		
Preterm	23	39.3
Term	33	55.0
Ignored	4	6.6
Origin		
Rio Grande	41	68.3
Other cities	19	31.6
Age upon admission		
0-7 day	33	55.0
8-30 days	27	45.0
Total	60	

Table 2. Diagnosis of congenital defect. According to etiology.

	N (%)
Multifactorial	
Gastrointestinal malformation	20 (33.3)
Cardiac malformation	17 (28.3)
Genito-urinary malformation	4 (6.6)
Multiple malformation	3 (5.0)
CNS malformation	2 (3.3)
Limb defects	1 (1.7)
Chromosomic	
Down syndrome	7 (11.7)
Rubinstein-Taybi syndrome	2 (3.3)
DiGeorge syndrome	1 (1.7)
Beckwith-Wiedmann syndrome	1 (1.7)
Others	2 (3.4)
Total	60 (100)

Regarding measures of length of stay in perinatal care services, there is no information in the literature associated with sets of morbidities. However, the average number of 17 days is used as a national comparative assessment[20].

As for location, the main malformations found were gastrointestinal, followed by cardiac malformations, the opposite of what was found in the study by Arruda et al[9], in which cardiovascular malformations are the most prevalent and have high mortality in the first year of life. Cardiovascular diseases add a significant impact, both in morbidity and mortality, and in cost to the healthcare system, representing the main cause of death among CDs. The frequency of cardiovascular CD associated with extracardiac CD is consistent with the percentage between 5 and 50% present in the literature, leading to the discussion of the cost-effectiveness of screening, through



Graph 1. Distribution of Cardiac Malformations according to single, multiple or accompanied by other defects.

complementary tests, for extracardiac malformations for all children with cardiac CD[21].

As for the diagnoses of multiple malformations and/or classic genetic syndromes, it was only possible to establish a conclusive diagnosis (clinical and cytogenetic) for seven cases of trisomy 21 (7/11 cases). This reflects the reality of other studies in which Down Syndrome is one of the most prevalent genetic alterations in the population, having its incidence of 1 in every 600 to 800 live births, and being related to advanced maternal age[22,23].

The difficulty in establishing a definitive etiological diagnoses is due, in part, to three factors: non-use or under-use of the dysmorphological physical examination performed by a geneticist as a standard approach in conducting the investigation[21]; the restriction for cytomolecular and molecular laboratory investigation, nor always available by the Brazilian Public Healthcare System and the low number of authorizations for necropsy. Even though this procedure can help answer important questions, such as the diagnosis itself, transmissibility and risk of recurrence of the pathology that affected the newborn, parents often do not authorize, as well as there is reluctance to request the medical team, according to Adappa et al[24].

In patients diagnosed with diseases of genetic etiology, genetic counseling was carried out with family members, advising on the risk of disease recurrence. Children with Trisomy 21 and other chromosomal disorders continued to be monitored at the institution's own genetics clinic.

A limitation of this study is the high number of newborns for whom there is no record of gestational age, many of them from other cities. This is also a limitation of the design. In a prospective study, the loss of these data would likely be minimized.

CONCLUSION

Although rare when alone, congenital defects together represent a large portion of the pathologies of the neonatal

period, being present in 8.6% of the patients in this study. The considerable number of deaths for which there was no timely evaluation suggests that the prevalence of CD in the present study could be even higher.

Thus, the importance of having a Geneticist to care for patients hospitalized in the neonatal ICU is highlighted, since many of these defects have a genetic origin - a qualified professional to help conduct the investigation in the search for an early diagnosis, intervention clinic, when necessary, as well as for adequate genetic counseling for the family[25]. Furthermore, the investigation of the points discussed as possible causes of the higher prevalence of birth defects in the study emerges as an open field.

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