

Epidemiological analysis of priority congenital anomalies for the birth surveillance purposes in a health regional in Maranhão

Hidário Lima da Silva¹, Ismália Cassandra Costa Maia Dias¹, Floriacy Stabnow Santos¹, Leonardo Hunaldo dos Santos¹, Lívia Maia Pascoal¹, Marcelino Santos Neto¹, Janaína Miranda Bezerra¹

ABSTRACT

Introduction: Priority congenital anomalies for birth surveillance purposes constitute a group of anomalies defined by the Brazilian Ministry of Health that can be identified in a timely manner after birth, mainly because they have characteristics that facilitate postpartum identification, as well as the early management and the accomplishment of appropriate therapies. **Objective:** To determine the incidence trend and characterize priority congenital anomaly cases for birth surveillance purposes in a health regional in the southwest of Maranhão, Brazil. **Methods:** Ecological study, typified as time series, carried out with data on live births with anomalies in the Imperatriz Health Regional, Maranhão (2011-2020), based on data from the Information System on Live Births. The temporal trend analysis of incidence rates was performed using Prais-Winsten linear regressions, through the SPSS software, version 24.0, at 5% significance. **Results:** There were 222 priority anomaly cases. Limb defects had a higher incidence (0.88/1,000), with an annual variation rate of -5.6 (95% CI: -15.7; 5.7) and a stable p-value of 0.35. There was a predominance of single women (69.36%), up to 29 years of age (72.98%), full-term (77.03%) and single (95.94%) pregnancies. The majority of newborns were males (60.81%), with adequate weight (70.72%) and brown color (79.28%). **Conclusion:** The occurrence of priority congenital anomalies in the southwest of Maranhão was stable, with a predominance of limb defects. Prenatal care provides early diagnosis, although data shows that congenital anomalies are still prevalent.

Keywords: Congenital anomalies, Incidence, Epidemiology, Time series studies, Health information system.

INTRODUCTION

Congenital Anomalies (CAs) stem from changes in the structure and/or function of parts of the body or organs, which may begin during intrauterine life and be detected early during prenatal care, delivery or after birth. They occur due to different causal factors, although they are related to the triad of chemical, physical and/or biological nature¹.

According to the World Health Organization (WHO), around 6% of live births

(LBs) around the world have some type of CA, resulting in approximately 295 thousand child deaths within the first four weeks of life. In Brazil, CAs represent the second leading cause of death in children less than five years of age, with around 24 thousand of LB cases with CA notified annually².

In Brazilian territory, data on LB are obtained through the Brazilian Information System on Live Births (SINASC, as per its Portuguese acronym), since 1990, through the Statement of Live Birth (DNL, as per its Portuguese acronym), which has had a specific field for the filling out information

Federal University of Maranhão, Graduate Program in Health and Technology, Imperatriz, (MA), Brazil



on CA since 1999, in order to improve tracking and monitoring of the occurrence of this condition.¹ Data on LBs with CAs began to be notified to SINASC compulsorily since 2018, through Law nº 13,685, dated June 25, 2018³.

Concerning priority CAs, they are considered when they have possibilities for diagnosis, either at birth or at the earliest possible intervention. This special attention to a group of anomalies is an important tool for surveillance, diagnosis, monitoring and management purposes, given that Brazil has social, demographic and territorial diversities that need to be fully addressed⁴.

From this perspective, the Brazilian Ministry of Health defined the priority CAs for birth surveillance, with the purpose of minimizing the impact that LB with CA can have on the individual, family and socioeconomic levels, as well as resulting in better clinical outcomes, continuity of the offspring and organization of the health system to meet the demands arising from the disease²⁻⁴.

The proposed categorization of priority CAs by the Brazilian Ministry of Health has been introduced as an important strategy for health surveillance. The incipient scientific production on the topic stands out, as well as the scarcity of studies on this area of understanding at the level of the state of Maranhão, given that a study analyzed CAs in a local setting and pointed out the growth of cases in recent years⁵.

Due to the above, this study had the objective of determining the incidence trend and characterizing priority congenital anomaly cases for birth surveillance purposes in a health region in the southwest of Maranhão, Brazil.

METHODS

This is an ecological study, typified as time series⁶, carried out by collecting data on LBs with CAs in the Imperatriz Health Regional, Maranhão, from 2011 to 2020. Data collection took place in October 2022, through the Information System on Live Births (SINASC).

The study setting was one of the 19 health regions in the state of Maranhão, the Imperatriz Health Regional, located in the southwest of the state of Maranhão, in region XV, whose Imperatriz Health Regional Management Unit (UGRSI, as per its Portuguese acronym) comprises 16 municipalities municípios⁷⁻⁸.

CAs notified during the period were classified as priority for birth surveillance purposes, in accordance with the recommendations of the Brazilian Ministry of Health, using the classification established according to ICD-10⁴, divided into eight groups: neural tube defects, microcephaly, congenital heart diseases, oral clefts, genital organ anomalies, limb defects, abdominal wall defects and Down Syndrome.

The incidence calculation was performed considering the number of LBs with CAs by the total number of LBs, with the constant 10^3 , equivalent to 1,000 live births⁹.

The temporal trend analysis of incidence rates was performed using the linear regression method proposed by Prais-Winsten, considering autocorrelation in time series¹⁰, which aims to relate a series of values of a measurement in periods. For the Annual Increment Rates (AIR) and respective Confidence Interval (CI), the calculation proposed by Antunes and Cardoso¹¹ was used. Based on these parameters, it

could be classified as increasing, stable or decreasing. The test was performed via syntax in the IBM SPSS software, version 24.0, at 5% significance¹².

For the epidemiological characterization of priority LB cases with CA, the following maternal variables were considered: marital status, age group, years of education, gestational age, type of pregnancy, type of delivery and number of prenatal consultations. In turn, the following LB variables were used: sex, birth weight and color/race. LB weight less than 2,500 grams was considered underweight, LB weight between 2,500 and 3,999 grams was considered adequate, and LB weight more than 4,000 grams was considered overweight. Using descriptive statistics, absolute frequency (Fa) and relative frequency (Fr) values of the variables under investigation were expressed.

This study did not require approval from the Research Ethics Committee (REC) with human beings, as it was carried out with secondary data available in a public domain database.

RESULTS

In the period from 2011 to 2020, 98,086 LB births took place in the Imperatriz Health Regional, with 596 LB being registered with CAs.

The municipalities that make up the UGRSI showed CA cases in the period from 2011 to 2020, although, when classifying them as priority, of the 596 notified cases, 222 were allocated into five priority groups: limb defects (87); oral clefts (58); genital organ anomalies (43); congenital heart diseases (19); and neural tube defects (15). It is underlined that it was not possible to classify 374 cases, due to the lack of sufficient support/data when filling out DNV in SINASC, in line with the ICD-10. Therefore, for this reason, it was not possible to classify priority CAs into the groups of microcephaly, abdominal wall defects and Down Syndrome.

Table 1 shows the number of priority CA cases by groups, distributed by year of occurrence, during the analyzed period.

Table 1

Priority CA cases by year in the Imperatriz Health Regional, Maranhão (2011-2020).

Priority CA cases	2011	2012	2013	2014	2015	2016	2017	2018	2019	2020	Total Period
Congenital heart diseases	0	2	5	3	2	0	3	2	1	1	19
Oral clefts	5	5	7	6	7	6	9	5	4	4	58
Genital organ anomalies	4	2	3	5	5	5	7	1	3	8	43
Limb defects	10	11	4	12	10	9	9	6	5	11	87
Neural tube defects	3	0	3	1	0	3	1	0	4	0	15
Regional	22	20	22	27	24	23	29	14	17	24	222

Source: MS/SVS/SINASC (2022).

In the period from 2011 to 2020, the incidence rate of CA in the five identified priority groups corresponded to 2.17:1,000LB. As for the years, 2017 had the highest incidence of the period (2.96:1,000LB), followed by 2014 (2.74:1,000LB) and 2020 (2.59:1,000LB), as displayed in Table 2.

Table 2

Incidence of priority CAs by group, year and period for every 1,000 live births in the Imperatriz Health Regional, Maranhão (2011-2020).

Anomaly groups	2011	2012	2013	2014	2015	2016	2017	2018	2019	2020	Total Period
Neural tube defects	0.29	0.00	0.31	0.10	0.00	0.31	0.10	0.00	0.39	0.00	0.15
Congenital heart diseases	0.00	0.20	0.51	0.30	0.19	0.00	0.30	0.19	0.09	0.10	0.19
Oral clefts	0.49	0.52	0.72	0.61	0.68	0.63	0.91	0.48	0.39	0.43	0.59
Genital organ anomalies	0.39	0.20	0.31	0.50	0.49	0.52	0.71	0.09	0.29	0.86	0.43
Limb defects	0.99	1.15	0.41	1.22	0.98	0.94	0.91	0.58	0.49	1.19	0.88
Regional	2.19	2.09	2.28	2.74	2.35	2.41	2.96	1.37	1.68	2.59	2.17

Source: MS/SVS/SINASC (2022).

Regarding the priority CA groups per year, limb defects had the highest occurrence in 2014; oral clefts in 2017; genital organ anomalies in 2020; congenital heart diseases in 2013; and neural tube defects in 2019.

variation of 0.0 (95% CI: -0.4; 0.5). Conversely, the group of CAs related to limb defects was the one that had the highest occurrence in the analyzed period (0.88:1,000LB), ranging from -5.6 (95% CI: -15.7; 5.7).

It is highlighted that, between 2011 and 2020, CA related to neural tube defects had the lowest incidence (0.15:1,000LBV), with a

As for the incidence trend, it remained stable in the priority CA groups, as displayed in Table 3.

Table 3

Trends in the incidence rates of priority CAs in the population of live births in the Imperatriz Health Regional, Maranhão (2011-2020).

	Annual variation rate %	Low_ CI*	High_ CI**	P-value	Situation
Congenital heart diseases	-2.1	-10.5	7.2	0.65	Stable
Oral clefts	-2.5	-11.3	7.2	0.63	Stable
Genital organ anomalies	5.9	-5.4	18.6	0.35	Stable
Limb defects	-5.6	-15.7	5.7	0.35	Stable
Neural tube defects	0.0	-0.4	0.5	0.98	Stable
Regional	-5.2	-25.7	21.0	0.69	Stable

Prais-Winsten regression.

*Lower confidence interval

**Higher Confidence Interval

Source: Authors (2022).

Regarding maternal characterization, Table 4 provides an overview of the variables of mothers who had LBs with priority CAs. There was a predominance of single women (69.36%), aged up to 29 years (72.98%), with more than eight years of education (67.57%), with gestational age suitable for the birth of a full-term baby (77.03%), with a single pregnancy (95.94%), with progression to caesarean section (54.06%) and with seven or more prenatal consultations (41.00%).

Table 4

Characterization of the mothers who had LBs with priority CAs in the Imperatriz Health Regional, Maranhão (2011-2020).

Variables	n (222)	
	Fa*	Fr%**
Marital status		
Single	154	69.36
Married/Stable Union	67	30.19
Separated	1	0.45
Age group (years)		
Up to 29	162	72.98
≥ 30	60	27.02
Years of education (years)		
Up to 7	72	32.43
≥ 8	150	67.57
Gestational age (weeks)		
Up to 36	45	20.27
≥ 37	171	77.03
Ignored	6	2.70
Type of pregnancy		
Single	213	95.94
Double	7	3.16
Ignored	2	0.90
Type of delivery		
Vaginal	100	45.04
Cesarean	120	54.06
Ignored	2	0.90
Number of prenatal consultations		
0 to 3	44	19.82
4 to 6	86	38.73
≥ 7	91	41.00
Ignored	1	0.45

*Absolute frequency

**Relative frequency

Source: MS/SVS/SINASC (2022).

As for the characterization of LBs with priority CAs, between 2011 and 2020, there was a predominance of births of male children (60.81%), with adequate birth weight (70.72%) and brown color (79.28%). This information can be checked in Table 5.

Table 5

Characterization of live births with priority CAs in the Imperatriz Health Regional, Maranhão (2011-2020).

Variables	n (222)	
	Fa*	Fr%**
Sex		
Male	135	60.81
Female	81	36.49
Ignored	6	2.70
Birth weight		
underweight	53	23.88
Adequate weight	157	70.72
Overweight	11	4.95
Ignored	1	0.45
Color/race		
Brown	176	79.28
White	29	13.07
Black	4	1.80
Yellow	1	0.45
Indigenous	9	4.05
Ignored	3	1.35

*Absolute frequency

**Relative frequency

Source: MS/SVS/SINASC (2022).

DISCUSSION

Priority anomalies for birth surveillance purposes are classified considering the ease they have for early diagnosis during prenatal care. They have common characteristics that are occasionally related to body structures and can be recognized during physical examination^{1-2,12}, which may justify timely registration in SINASC.

In this study, limb defect anomalies had the highest incidence in the analyzed period (0.88:1,000LB), with an annual variation of -5.6 (95% CI: -15.7; 5.7), corroborating the data from the Epidemiological Bulletin of the Brazilian Ministry of Health, which analyzed the situation of CAs in the country between 2010 and 2021, showing limb defects as the most prevalent in all analyzed years of the series. The overall prevalence of CA in 2021 among the num-

ber of LB was 82.6%, with limb defects accounting for 25.4%. In 2021, the Northeast Region had the highest prevalence rate in the country among priority CAs, 29.8%; and, in Maranhão, the prevalence of limb defects reached 22.8:10,000LB¹³.

Limb defects are non-chromosomal congenital anomalies that are most common in newborns. They may manifest different phenotypic characteristics, from partial changes in fingers or toes and/or complete absence of limbs. The risk of these defects occurring is accentuated between the 24th and 36th day after fertilization, due to the formation of limb buds¹⁻².

The most common types of CA limb defects are congenital deformities of the feet, polydactyly, reduction defects of the upper and/or lower limbs, and arthrogryposis multiplex congenita^{1-2,14}. Forefoot CAs have two different forms: malformations, when they

occur during the embryonic period and result in anatomical defects; and deformations, when they occur in the fetal period, in a foot with an apparently normal configuration¹⁴.

Regarding Oral Clefts (OC), they are classified as craniofacial CAs that affect the oral and/or nasal cavities. The occurrence of this illness can affect the lip and/or palate, being classified according to the anatomical location¹⁵⁻¹⁶.

It is underlined that, in this study, OC were the second most common CA between 2011 and 2020 (0.59:1,000LB), whereas, in 2013, it was the priority CA group, with the highest incidence (0.72:1,000LB), with a variation of -2.5 (95% CI: -11.3;7.2).

Regarding the OC incidence, this study introduced data similar to national and international studies. A study held with SINASC data from all states of the federation, from 2005 to 2016, showed an incidence rate similar to this study, 0.51:1,000LB¹⁵. In Brazil, studies indicate a variation in prevalence between 0.19 and 1.54:1,000LB¹⁷. As for the international setting, studies indicate that OC constitute one of the most prevalent craniofacial malformations around the world (0.5 to 2.0:1,000LB)¹⁸, 1:700LB¹⁷.

With respect to sex, it is estimated that 60% to 80% of individuals with OC are males. A study conducted in Paraná revealed males as the most prevalent¹⁹, showing data similar to this study (60.81%).

With regard to CA cases related to genital organ anomalies, these had the third highest incidence in this study (0.43:1,000LB), with 2020 being the year with the highest occurrence of cases

(0.86:1,000LB), with a variation annual rate of 5.9 (95% CI: -5.4;18.6).

The Epidemiological Bulletin of the Brazilian Ministry of Health revealed genital anomalies occurring at 1:300NB, as well as undifferentiated genitalia at 1:4,500NB², which is similar to the data found in this study.

A literature review showed that hypospadias and cryptorchidism are the most common CAs in men²⁰. In addition to them, there is also indeterminate sex and pseudohermaphroditism, which are CAs of a morphological and physiological nature, where the newborn has atypical development of chromosomes, gonads and/or anatomical sex^{1,13}.

Concerning the congenital heart diseases, they are changes in the structure of the heart pump and/or blood vessels. Some of these changes can be identified by observing abnormalities in cardiac auscultation, cyanosis, hepatomegaly, edema of the lower limbs, respiratory distress, tachypnea, hyperdynamic precordium, irregular rhythm, third heart sound, difference in pulses and shock²¹⁻²².

In this study, congenital heart diseases had an incidence of 0.19:1,000 LB, with the highest occurrence in 2013 (0.51:1,000 LB) and annual variation of -2.1 (95% CI: -10.5;7.2).

The Epidemiological Bulletin of the Brazilian Ministry of Health identified congenital heart diseases as the second most prevalent group of anomalies (11:10,000 LB). In 2021, they had a prevalence of 35:10,000LB in the state of São Paulo, Brazil¹³. Nevertheless, in states like São Paulo, there are projects that aim to raise

awareness and train professionals for early identification of CAs, thus contributing to high notification rates in SINASC²³.

In the United States, approximately 44.5% of LBs with congenital heart diseases die within the first year of life. In Latin America, CAs of the cardiac system are the second leading cause of death in the first year of life²¹. Another study analyzed the panorama of congenital heart diseases around the world, showing an increase of 18.7% from 1990 to 2017 in prevalence rates, and a decline in mortality rates of around 34.5% for the same analyzed period. Nevertheless, deaths from congenital heart diseases prevailed in children less than one year of age, corresponding to 67% of deaths²⁴.

A study conducted in Saudi Arabia evaluated the association of heart diseases in patients with OC, showing a prevalence of 41% of associated CAs, where atrial septal defect is the most prevalent congenital heart disease, with 37%²⁵.

Congenital heart diseases, as they do not manifest physical characteristics visible on physical examination, are less registered in notification systems, such as SINASC, but mortality rates are much higher when infant mortality records are analyzed^{5,26}.

Regarding the CAs of neural tube defects, they stem from problems and/or failures in the embryogenesis process up to the 29th day of embryonic life. These CAs involve malformations of the brain and spinal cord, which can compromise the skin, muscles, bones and adjacent soft tissues^{1-2,13}.

Neural tube defects involve malformations due to incomplete closure of the

neural tube in the embryonic phase, the most prevalent being anencephaly and spina bifida²⁷.

Among the priority CAs introduced in this study, neural tube defects had the lowest incidence (0.15:1,000LB). The highest occurrence of this condition was in 2019 (0.39:1,000LB), showing an annual variation of 0.0 (95% CI: -0.4; 0.5). A systematic literature review showed variability in global prevalence rates of CAs related to neural tube, 0.3 to 199.4:10,000LB²⁸. This happens mainly due to the specificities of the public policies adopted in each country. In Brazil, there are public policies that aim to encourage folic acid supplementation early in pregnancy as a strategy to prevent the occurrence of neural tube defects²⁹.

It is noted that this study did not introduce data on microcephaly cases; however, between 2015 and 2016, there was an epidemic of cases related to Zika Virus infection⁴. A study emphasized that the number of microcephaly cases in Brazil increased nine times in relation to the annual average, when compared to the period from 2010 to 2014, especially in newborns of women living in the Brazilian Northeast³⁰. Another highlight was the lack of notified Down Syndrome cases.

In light of the foregoing, it is inferred that the scarcity of data on microcephaly and Down Syndrome may raise questions regarding the identification and timely notification of cases in SINASC, as well as regarding the reorganization of health services, in order to avoid undernotification.

Regarding the incidence trend, the rates remained stable in the five groups during the analyzed period (p-value 0.69), when considering the rates for the health

regional, with an annual variation of -5.2 (95% CI: - 25.7; 21.0). The stability of the trend in the incidence of priority CAs can be attributed to the average number of notifications, with a small discrepancy in the analyzed time frame, as well as to the incidence rates of the health regional, also due to the high number of live births in the health regional, considering that the number of cases showed a pattern of similar averages, when analyzed year by year within the time frame.

Concerning the socioepidemiological characterization of mothers, in this study, there was a difference with the findings of a study conducted in Salvador, Bahia, where the predominance was of women less than 18 years of age and more than 35 years of age, and with multiple pregnancies. Conversely, there was similarity in the type of delivery, with a predominance of surgical birth³¹. A study conducted in São Luís, Maranhão, also showed a higher prevalence of CAs in women who had a cesarean section and had a good level of education⁵.

As for the number of prenatal consultations, the women in this study had a number of seven or more, above that recommended by the Brazilian Ministry of Health, which considers a minimum of six consultations²⁹. The findings of this study were similar to a study conducted in São Luís, Maranhão, where CA cases were more prevalent in women with seven or more prenatal consultations⁵. It is assumed that these women would have more access to health services, providing early diagnosis. This finding allows reflection on the importance of prenatal care as a strategy capable of providing early identification of cases, guidance for mothers and appropriate professional intervention³².

Regarding the characterization of LBs, the data are similar to a study conducted in Salvador, Bahia, with a predominance of males³¹; however, the percentage of LBs with CAs and adequate birth weight was higher in this study.

It is highlighted that low birth weight, linked to genetic and environmental factors, is commonly associated to explain the occurrence of CAs, being considered the genesis of LBs with low birth weight³¹. A systematic review and meta-analysis study showed low birth weight as a predictive factor for reduced survival for any type of CA³³. Therefore, LBs with CAs tend to have low birth weight, although the majority of them were born with adequate weight in this study. It is noted that the presence of CA is more common among those born with low birth weight (< 2,500g) (370: 10,000 LB)¹³.

Similarly, the Epidemiological Bulletin of the Brazilian Ministry of Health indicated that the prevalence of congenital anomalies in premature infants (267: 10.000LB) was three times higher than in full-term infants (86:10,000LB)¹³.

It is pointed out that, despite the improvement in the quality of data made available at SINASC in recent years, there is still weakness in filling out DNV, in addition to the probable undernotification of CA cases, revealing data that may distort the reality of cases in Brazil³¹. Furthermore, SINASC does not show data regarding family income, leaving a gap for a possible association between low family income and the occurrence of CA.

It is emphasized that this study has limitations regarding the use of secondary data available at SINASC, given that los-

ses, inadequate completion and/or lack of data may occur when filling out DNV. Nonetheless, despite the limitations, the study highlighted the priority CAs and can serve as a basis for developing programmatic health actions that reiterate the importance of preventing these illnesses.

CONCLUSION

The occurrence of priority congenital anomalies in the southwest of Maranhão was stable in the analyzed time series, with a predominance of cases related to congenital anomalies of limb defects. Maternal and neonatal characteristics corroborate findings in the scientific literature, showing similarities in the occurrence of certain anomalies.

The need to preserve the effectiveness of prenatal consultations is evident, especially as an opportune moment to identify priority congenital anomalies that occasionally prove to be prevalent and neglected in prenatal care.

Knowledge on priority congenital anomalies at local levels, such as the state of Maranhão, can provide support for the development of regional and specific public policies aimed at preventing and/or minimizing these illnesses, as well as providing support for early diagnosis and monitoring of cases.

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HLS: responsible for designing, collecting data and writing of the manuscript.

ICCMD, FSS, LMP, MSN: responsible for analyzing and interpreting the results, writing and critically reviewing the content of the manuscript.

LHS: responsible for statistical analysis.

JMB: responsible for analyzing and interpreting the results, writing and critically reviewing the content.

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Corresponding Author:

Hidário Lima da Silva

hidariolimadasilva@gmail.com

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