Frequency of Congenital Anomalies in the State of Amazonas-Brazil for the Period 2008 to 2017

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Abstract: There are few studies on the frequency of congenital anomalies in Brazil, particularly in the state of Amazonas. Therefore, the aim of this study was to investigate the number of live births with congenital anomalies in Amazonas, in the period 2008-2017, based on the Live Birth Information System (SINASC), as well as to discuss the relevance of the professionals being trained for the correct completion of the notice of live birth. In the study period, of the 773,313 live births in Amazonas, 4,593 were diagnosed with some type of congenital anomaly, according to DATASUS. This corresponds to a prevalence of 5.9 cases per 1,000 live births. The most frequent cases were congenital deformities of the musculoskeletal system, with 23% of the reports, congenital deformity of the feet, with 16%, and other congenital malformations, with 15% of the registrations. The need for investments in the training and qualification of professionals was observed, so that they can act, in a qualified way, in the cycle of diagnosis, registration and support for families, since they have not been trained to deal with this new reality. The importance is emphasized of continuing the research in the field of genetic diseases in Amazonas, in order to assist the individuals and families affected, as well as supporting the decisions related to the planning and implementation of public policies directed toward the issue.

Key words: Congenital anomalies, information systems, notice of live birth.

1. Introduction

Congenital anomalies are developmental disorders of embryonic origin, and may be structural, functional or metabolic changes in the fetus present at birth or that manifest after several years [1-4]. Inherited or acquired, their causes may be associated with genetic, environmental, physical, chemical, biological factors, or multifactorial when they are due to more than one of these factors [1, 2, 5]. In the majority of cases, the etiology of the congenital anomalies remains unknown. Those of genetic origin have been investigated more and the chromosomes and malformations with Mendelian inheritance have been covered [1, 6]. Those dependent on environmental factors or gene-environment interactions are more difficult to recognize because cases caused by teratogens are still poorly studied, especially in developing countries such as Brazil [1, 2, 6]. Some studies in Brazil show that the estimated frequency of congenital anomalies is 2% to 3% of live births [2, 6-8]. In developing countries, such as Brazil, there is little research on the prevalence of these congenital malformations. The scarcity of official data in Brazil on the issue of these anomalies highlights the need to improve the existing information systems [6, 7].

In 1990, the Ministry of Health, through DATASUS, developed the Live Birth Information System (SINASC) with the aim of gathering epidemiological information regarding births reported throughout the country, based on the Notice of Live Birth (NLB). As of 1999, a field was included in the NLB, coded according to Chapter XVII of the 10th Revision of the International Statistical Classification of Diseases and Related Health Problems (ICD-10), which allowed the notification of congenital anomalies, making it possible, when duly completed, to evaluate their frequency and the nature of the events. The aim of this was to develop reliable health and surveillance indicators, in order to facilitate the planning of health...
policies, especially for children, which made SINASC an important tool for monitoring congenital anomalies in Brazil [1, 2, 6-9].

The characterization of these congenital anomalies is important information for the planning and implementation of programs directed toward individuals with these conditions and their families [1]. In order to improve data completion at SINASC, a partnership was established between the Ministry of Health and the Center for Medical Genetics of the Federal University of São Paulo in 2005 and a booklet was prepared in 2008 to reduce underreporting and stimulate the early diagnosis of congenital defects and guide the correct completion of this field. Each municipality can construct its own clinical and epidemiological reference to achieve a certain standard of excellence [1, 6].

In 2011, the Ministry of Health distributed the new form of the Notice of Live Birth, which had the field related to the registration of congenital anomalies—formerly field 34, now fields 6 and 41, implying changes in SINASC, with the production of a new instruction manual: the Manual for improvement in the diagnosis of congenital anomalies [10]. The Live Birth Information System proposes to collect and produce information on the occurrence of births in Brazil. The correct completion of the NLB is fundamental, since this instrument is the source of the information system [6]. In Brazil, the NLB is the data collection instrument and is characterized by being universal, standardized and of low cost. The vital data generated by it provide a wealth of information for epidemiological studies, for the collection of health indicators and for the basis of planning and prevention activities, as well as policies in the area of maternal and child health [7, 11].

Many authors have emphasized the importance of the quality of recorded data, especially with regard to coverage of events, reliability of the information and completeness of the data. However, SINASC still presents limitations as a source for determining the prevalence of congenital anomalies. Underreporting is still common and the quality of the recorded information needs to be improved [6]. Due to being an important instrument used for populational monitoring, efforts must be made to ensure better gathering of information [11]. This premise is due to the fact that, in all stages of the information production cycle, problems can occur in the quality of the information. If these issues are not taken into account, the knowledge generated from these data may not adequately represent the reality studied [12]. The under-registration of congenital anomalies in the NLB is a reality that hinders the epidemiological study of these malformations and impairs prevention actions. Probably, its poor completion is due to the lack of knowledge about its importance in public health and the impact that congenital defects have on society as a whole [6].

The registration of information in the hospital can be hampered by inadequate throughput, especially when different people collect and complete the NLB data. Therefore, it is recommended that the NLB be completed prior to the moment of discharge of the mother and by a single person [2]. However, there are studies that affirm that the NLB is completed by more than one member of staff. Failure to correctly enter the data may occur due to a lack of training of the staff responsible or failure to perceive the importance of the correct completion [8]. Therefore, it is necessary to invest in the qualification of the information, either through the personnel involved in this task or through recommendations regarding the completion of the NLB in cases of congenital defects, which would result in a greater commitment in the transcription of the eventual diagnosis performed by the physicians [7]. Therefore, if the professionals can be helped to correctly diagnose and notify congenital defects, this would be reflected in the NLB, with it becoming an excellent monitoring and surveillance system for congenital anomalies in Brazil [6].

It is necessary to reflect on the existence and quality
of the care provided by the healthcare providers, requiring the health and education system to invest more in infrastructure and human resources to institute rehabilitation and prevention of diseases [4]. To overcome the adversities, it is essential that the families receive support and information from professionals involved in the care, preferably from a multidisciplinary team. Clinical treatment alone is not sufficient, with it being necessary to identify ways that promote the insertion of this child into the family environment and society, as well as to prevent it from suffering discrimination [13].

A study that reports the family’s experience in caring for a child with a congenital anomaly found that when the family interacts with trained professionals that provide support and clarification they generally accept the child in a realistic way and feel empowered to cope with the situation. The literature, therefore, makes it clear that the child with congenital malformation needs specialized care and a multidisciplinary team, in which the primary focus of the recovery involves the integration of the patient into the family and social environment [5].

In order to identify the frequency of genetic diseases in the state, this study aimed to investigate the number of live births with congenital anomalies in the state of Amazonas, in the period 2008-2017, using the Live Birth Information System (SINASC). The study also sought to discuss the importance of having professionals trained for the proper diagnosis and correct completion of the NLB, thus facilitating the planning and implementation of public policies focused on the issue of genetic diseases in the state of Amazonas.

2. Methodology

2.1 Study

This study quantified data on live births that presented congenital anomalies, broken down by municipality of the state of Amazonas, recorded from 2008 to 2017 and based on information found in the Live Birth Information System (SINASC), with the search carried out in January 2019. At the same time, searches for articles published in scientific journals of the Scientific Electronic Library Online (SciELO) database were also conducted. Using certain keywords, the search result was: “congenital anomalies” (691 articles, 195 articles in Portuguese); “datasus” (335 articles, 270 articles in Portuguese); “diagnosis of anomalies” (179 articles in Portuguese); “frequency of anomalies” (71 articles in Portuguese). In order to select the articles most related to the theme of this study, the search was refined with the following words: “congenital anomalies”, “diagnosis”, “live births”, “information validation”. From this, 13 articles were selected for the base of the present article. It should be emphasized that due to the existence of few studies on the subject in question, it was decided not to limit the year of publication.

2.2 DATASUS Data Collection

In order to achieve the aims of this article, DATASUS was used through SINASC to tabulate the data. In the Vital Health Statistics Portal, the Live Births 1994 to 2016 and Preliminary Data of 2017 options were chosen, followed by the state. On the Amazonas page, the following filters were selected: Birth by residence/mother; Period from 2008 to 2016 and 2017; Municipalities; and Congenital anomalies. Three live births with congenital anomalies were excluded, due to the fact that the municipality had not been reported in the SINASC registry. After performing this investigation, another search was performed in DATASUS, with the following variables: Birth by residence/mother; Period from 2008 to 2016 and 2017; Municipalities; Congenital anomalies; and the Types of congenital anomalies, in order to investigate the types of anomaly that were reported more in the Notice of Live Births in the state.

The types of anomalies registered in SINASC are:
(1) Spina bifida;
(2) Other congenital malformations of the nervous...
system;
(3) Congenital malformations of the circulatory system;
(4) Cleft lip and cleft palate;
(5) Absence, atresia and stenosis of the small intestine;
(6) Other congenital malformations of the digestive tract;
(7) Undescended testis;
(8) Other malformations of the genitourinary system;
(9) Congenital deformity of the hip;
(10) Congenital deformity of the feet;
(11) Other malformations and congenital deformations of the musculoskeletal system;
(12) Other congenital malformations;
(13) Unspecified chromosomal abnormalities;
(14) Congenital syphilis;
(15) Hemangioma and lymphangioma;
(16) Dentofacial anomalies;
(17) Other conditions of integument specific to the fetus and newborn;
(18) Without congenital anomaly/not reported.

This second cataloging was carried out in order to provide a basis for the discussion regarding the importance of having trained professionals in the state of Amazonas to perform the proper diagnosis and correct completion of the NLB, according to the aims of this work.

3. Results and Discussion

From 2008 to 2017, 773,313 live births were reported in the state of Amazonas. Of these, 4,593 were diagnosed with some type of congenital anomalies according to DATASUS. This corresponds to the prevalence of 59 cases per 10,000 live births within the period studied (Table 1). According to the State Health Plan—Amazonas 2016-2019—SUSAM, the metropolitan region of Manaus, alone, represents 60% of the population of the state of Amazonas [14]. As expected, the capital of Amazonas, Manaus, was the city with the highest number of live births with congenital anomalies, with 2,816 registered cases.

It can be seen that there are municipalities in Amazonas that registered more than 70 cases of births with congenital anomalies, during that time period studied. In São Gabriel da Cachoeira, for example, for every 10,000 live births, there were 72 with congenital anomalies. Parintins is the second most populated municipality in Amazonas [15], as indicated by the IBGE, and presented 65 cases of births with congenital anomalies for every 10,000 live births. These two municipalities presented higher prevalences than that of the entire state, which was 59 cases for every 10,000 live births.

Table 2 shows the frequencies of the types of anomalies reported by DATASUS. The following types of congenital anomalies: Congenital syphilis; Dentofacial anomalies; and Other conditions of integument specific to the fetus and newborn; were not included in the table, because there were no records of these anomalies in the state of Amazonas for the period covered by this study. The most frequent cases were congenital deformities and malformations of the musculoskeletal system, accounting for 23% of the reports, congenital deformity of the feet, with 16%, and other congenital malformations, with 15% of the reports.

These results are similar to those of other studies performed in other locations in Brazil (São Paulo, MatoGrosso and Rio de Janeiro), as well as in other European countries, the United States and Venezuela, in which malformations of the musculoskeletal system, nervous system and the cleft lip and palate were the most frequent. These studies stated that this predominance in the outcomes may be related to the ease of diagnosis, because they are easily visible in physical examination at birth [1, 2, 8]. Considering the tables below, it is possible that this may be one of the realities of Amazonas. According to the Brazilian Society of Medical Genetics, there is only one physician-geneticist to attend 4,593 individuals with
Table 1  Records of live births with congenital Anomalias, Amazonas.

| MUNICIPALITY/YEAR | 2008 | 2009 | 2010 | 2011 | 2012 | 2013 | 2014 | 2015 | 2016 | 2017 | TOTAL |
|-------------------|------|------|------|------|------|------|------|------|------|------|-------|
| 1. Alvarães       | 1    | 1    | 0    | 1    | 2    | 4    | 4    | 1    | 2    | 5    | 21    |
| 2. Amaturá        | 0    | 1    | 3    | 3    | 4    | 1    | 1    | 2    | 2    | 2    | 19    |
| 3. Anamã          | 0    | 2    | 0    | 0    | 1    | 1    | 1    | 0    | 3    | 2    | 10    |
| 4. Anori          | 0    | 0    | 0    | 0    | 1    | 0    | 0    | 0    | 0    | 0    | 2     |
| 5. Apuí           | 0    | 0    | 0    | 1    | 2    | 1    | 1    | 3    | 0    | 3    | 11    |
| 6. Atalaia do Norte | 0  | 0    | 1    | 4    | 1    | 2    | 2    | 2    | 1    | 2    | 15    |
| 7. Autazes        | 4    | 6    | 3    | 12   | 11   | 0    | 5    | 7    | 2    | 2    | 52    |
| 8. Barcelos       | 0    | 1    | 1    | 2    | 2    | 2    | 1    | 1    | 1    | 1    | 12    |
| 9. Barreirinha    | 3    | 6    | 2    | 6    | 5    | 5    | 1    | 6    | 7    | 7    | 48    |
| 10. Benjamin Constant | 0 | 1    | 0    | 2    | 4    | 2    | 3    | 3    | 1    | 4    | 20    |
| 11. Beruri        | 0    | 0    | 0    | 1    | 2    | 2    | 3    | 0    | 2    | 0    | 10    |
| 12. Boa vista do Ramos | 3 | 0    | 0    | 1    | 0    | 0    | 1    | 1    | 0    | 2    | 8     |
| 13. Boca do Acre   | 2    | 2    | 1    | 2    | 2    | 2    | 1    | 4    | 8    | 2    | 26    |
| 14. Borba         | 5    | 3    | 7    | 6    | 0    | 3    | 1    | 6    | 3    | 9    | 43    |
| 15. Caapiranga    | 1    | 0    | 1    | 1    | 1    | 0    | 1    | 3    | 1    | 1    | 10    |
| 16. Canutama      | 1    | 0    | 0    | 2    | 2    | 3    | 1    | 0    | 1    | 0    | 10    |
| 17. Carauari      | 0    | 0    | 0    | 0    | 0    | 1    | 0    | 1    | 1    | 1    | 4     |
| 18. Careiro       | 1    | 1    | 2    | 3    | 1    | 1    | 2    | 4    | 3    | 2    | 20    |
| 19. Careiro da Várzea | 1  | 3    | 2    | 0    | 0    | 1    | 0    | 2    | 1    | 2    | 12    |
| 20. Coari         | 10   | 13   | 8    | 3    | 14   | 14   | 11   | 12   | 2    | 6    | 93    |
| 21. Codajás       | 0    | 1    | 4    | 2    | 2    | 4    | 2    | 1    | 0    | 1    | 17    |
| 22. Envira        | 6    | 1    | 1    | 2    | 0    | 8    | 6    | 10   | 6    | 5    | 45    |
| 23. Eirunepé      | 1    | 6    | 12   | 11   | 3    | 6    | 5    | 8    | 4    | 4    | 60    |
| 24. Fonte Boa     | 2    | 7    | 3    | 7    | 10   | 5    | 2    | 4    | 2    | 4    | 46    |
| 25. Guajará       | 1    | 3    | 1    | 2    | 0    | 1    | 0    | 1    | 0    | 1    | 10    |
| 26. Humaitá       | 0    | 3    | 2    | 5    | 5    | 1    | 3    | 1    | 9    | 5    | 34    |
| 27. Ipiúna        | 1    | 1    | 0    | 1    | 1    | 0    | 0    | 0    | 0    | 0    | 4     |
| 28. Iranduba      | 1    | 4    | 8    | 7    | 10   | 6    | 7    | 3    | 6    | 3    | 55    |
| 29. Itacoatiara   | 3    | 4    | 1    | 1    | 6    | 7    | 3    | 1    | 3    | 8    | 37    |
| 30. Itamarati     | 1    | 0    | 0    | 0    | 0    | 1    | 0    | 0    | 0    | 0    | 2     |
| 31. Itapiranga    | 0    | 1    | 0    | 1    | 0    | 0    | 1    | 0    | 1    | 1    | 5     |
| 32. Japurá        | 2    | 1    | 0    | 2    | 5    | 4    | 1    | 5    | 2    | 1    | 23    |
| 33. Jutaí         | 0    | 0    | 1    | 0    | 2    | 1    | 1    | 1    | 4    | 4    | 14    |
| 34. Jurua         | 0    | 0    | 0    | 2    | 0    | 3    | 4    | 2    | 2    | 3    | 16    |
| 35. Lábrea        | 0    | 0    | 1    | 0    | 8    | 2    | 0    | 0    | 0    | 0    | 11    |
| 36. Manacapuru    | 20   | 9    | 19   | 11   | 9    | 8    | 10   | 8    | 11   | 15   | 120   |
| 37. Manaquiri     | 0    | 0    | 2    | 3    | 4    | 4    | 2    | 2    | 1    | 4    | 22    |
| 38. Mauáns        | 300  | 320  | 338  | 361  | 321  | 325  | 273  | 211  | 199  | 168  | 2,816 |
| 39. Manicoré      | 5    | 1    | 4    | 1    | 1    | 4    | 6    | 5    | 1    | 7    | 35    |
| 40. Maués         | 3    | 2    | 4    | 9    | 12   | 15   | 11   | 4    | 6    | 9    | 75    |
| 41. Maraã         | 2    | 4    | 1    | 5    | 3    | 0    | 0    | 1    | 1    | 2    | 19    |
| 42. Nhamundá      | 1    | 3    | 0    | 2    | 0    | 3    | 6    | 6    | 1    | 6    | 28    |
| 43. Nova Olinda do Norte | 0 | 0    | 3    | 3    | 1    | 0    | 3    | 0    | 0    | 1    | 11    |
| 44. Novo Airão    | 0    | 1    | 1    | 5    | 1    | 2    | 2    | 3    | 0    | 2    | 17    |
| 45. Novo Aripuanã | 0    | 0    | 1    | 6    | 2    | 1    | 0    | 2    | 2    | 2    | 16    |
| 46. Parintins     | 15   | 12   | 15   | 10   | 15   | 12   | 7    | 15   | 11   | 11   | 123   |
| 47. Pauini        | 0    | 0    | 0    | 1    | 0    | 0    | 0    | 0    | 1    | 1    | 3     |
congenital anomalies in Amazonas, a state with 62 municipalities and difficult access logistics. For this reason, it is of great relevance that people who are involved in the delivery of these live-born infants with congenital anomalies, know how to properly complete the NLB. The support of a multidisciplinary-geneticist team to assist families who receive the diagnosis is essential.

The lack of this diagnosis prevents the preparation of the mourning phase in the family nucleus, as the birth of a malformed and previously undiagnosed child causes great suffering for the couple [3]. A project carried out in the city of São Paulo showed that if maternity hospitals were to establish partnerships with
centers specialized in the diagnosis and registration of congenital defects in the NLB, it would be possible to shorten the time until reliable records of congenital anomalies are available in Brazil. This would improve care in terms of early recognition of genetic syndromes and the consequent intervention in preventive terms, through genetic counseling for the families and care for the patient [6]. The conduct of healthcare providers in relation to the newborn with a congenital anomaly must be specific and of high quality. Thus, knowledge about malformations and the behaviors to be adopted by team members is of paramount importance in guiding parents and family members, enabling them to clarify their doubts about the disability and encouraging them to seek quality of life within the limits imposed on the child [5].

Considering that SINASC can be used as an important tool for the populational monitoring of congenital anomalies, efforts should be made to ensure better gathering of information [11]. The reliability of the information contained in the NLB is evaluated worldwide because it serves as the basis for planning health policies in this area and is widely disseminated in the bulletins of the majority of epidemiological surveillance centers. Therefore, greater commitment of the person in charge of completing the NLB in relation to obtaining and correctly transcribing the data is necessary, as is the adequate training of this professional to carry out this task, with this affecting the quality of the information provided to the interested parties.

4. Conclusions

This study is important because, by investigating the reality of the state of Amazonas, the need for investments in the area of medical genetics was confirmed, which has already been highlighted by several studies. These investments should be made with the aim of assisting all the staff in the hospitals involved before, during and after completion of the NLB. It is essential to have professionals trained to deal with the possibility of a child with a congenital anomaly, capable of properly diagnosing, accompanying and instructing the family for the best possible treatment.

This work explored a topic that has not been approached, presenting results that could help in the planning of Public Health actions, guiding and deepening discussions regarding the scope of actions in the public policies focused on the issue of Congenital Anomalies. However, this needs to be discussed further. Accordingly, there are still many possibilities for future studies which would be of great relevance, considering the lack of studies on congenital anomalies in Brazil, especially in the state of Amazonas.

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