# Prevalence of Congenital Anomalies in a Tertiary Hospital in Southern Brazil (1998-2015)

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**Abstract.** *Objective:* To evaluate the prevalence of congenital anomalies in a tertiary hospital in southern Brazil.

*Methods:* Retrospective, cross-sectional study involving 22,317 births from 1998 to 2015 in the Gynecology and Obstetrics Department of the Caxias do Sul General Hospital /Caxias do Sul University. A descriptive univariate analysis was performed, followed by multivariate analysis, of the association between congenital anomalies and maternal, gestational, delivery, and newborn characteristics.

*Results:* The prevalence of congenital malformations was 1.5% (95% CI 1.4-1.7), distributed as follows: genitourinary tract (18.2%), central nervous system (16.5%), musculoskeletal system (16.3%), cardiovascular system (9.7%), gastrointestinal tract (10.8%), and multiple malformations (28.5%). In the multivariate analysis, the following neonatal variables remained associated with congenital malformation: 5-minute Apgar score 0 to 3 [aHR=2.09 (1.21-2.12); p<0.010], 5-minute Apgar score 4 to 7 [aHR=2.83 (1.53-3.72); p<0.001], infant mortality [aHR=10.34 (7.79-13.72); p<0.001], stillbirth [aHR=25.56 (24.61-64.44); p<0.0001], and cesarean delivery [aHR=2.51 (1.99-3.16); p<0.001].

**Keywords**: congenital anomalies, newborn, prevalence, risk factors, mortality

## Introduction

Congenital malformations (CMs) are important causes of death in newborns (NBs) and children under 5 years of age. It is estimated that worldwide, 1 in every 33 NBs has some type of CM. According to data from the World Health Organization/Pan American Health Organization, approximately 3% of NBs are affected by some type of CM every year in the United States (Brasil, Ministério da Saúde, 2018; Cremonese et al., 2014). In Brazil, approximately 2.9 million births were recorded in 2016, and live births with anomalies were 0.8% of all births. The main anomalies were musculoskeletal anomalies (40.7%), central nervous system anomalies (9.8%), and circulatory system anomalies (8.6%) (Brasil, Ministério da Saúde, 2018). Approximately 5% of NBs worldwide are affected by birth defects that contribute significantly to mortality rates in different ethnic groups (Cremonese et al., 2014;

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Sułkowski, 2012). CMs, in addition to being responsible for high perinatal mortality, are identified in a large percentage of miscarriages, suggesting that this is the natural progression of most pregnancies whose fetuses have embryonic malformations (Ren et al., 2018).

Little is known about the etiology of CMs. Mendelian inheritance seems to be the most plausible among the genetic causes (Fong et al., 2014). The main risk factors associated with CMs are environmental effects, occupational exposure, medication, smoking, use of illicit drugs or alcohol, pregestational diabetes mellitus, thyroid dysfunction, congenital infections, chronic hypertension (Ren et al., 2018; Fong et al., 2014) and asthma (Blais et al., 2015). However, approximately 70% of CMs have unknown etiologies (Fong et al., 2014). A study published in 2018 showed a rising number of cases of congenital anomalies when diabetes and obesity were present as risk factors. Most anomalies are rare, and large populations are needed to obtain relevant information and identify possible risk factors (Morris et al., 2018). Based on current indicators, we chose to conduct a study on the prevalence of CMs in the Gynecology and Obstetrics Department (GOD) of Caxias do Sul General Hospital (GH)/ Caxias do Sul University.

## Methods

This was a cross-sectional, retrospective study conducted in a tertiary teaching hospital in Rio Grande do Sul state, Brazil. GH is a reference center for 49 municipalities in the northeastern region of the state and serves only users of the National Health System (*Sistema Único de Saúde* - SUS). The average annual birth rate is 1,800. The target population included pregnant women seen from 1998 to 2015. All were systematically recorded in a hospital database. The following inclusion criteria were chosen: gestational age $\geq$ 22 weeks and delivery in GOD/GH. The fetal deformities were identified by ultrasound during gestation or at birth by the neonatologist team. Birth defects were systematized according to the International Classification of Diseases 10th revision.

Patients diagnosed with gestational diabetes mellitus or type 1 and type 2 diabetes mellitus were considered diabetic. Diabetes was diagnosed using the protocol recommended by the American Diabetes Association. Data on the Apgar score were obtained from evaluations performed by the neonatologist or resident doctor on call during care in the delivery room. The gestational age was calculated based on the date of the last menstrual period, early ultrasound, or the Capurro method, as applicable. A descriptive analysis was performed to correct any inconsistencies, and when necessary, the data were rechecked in the patient's medical records. Statistical analysis was performed using the *Statistical Package for the Social Sciences* (SPSS Inc., Chicago, IL, USA) version 23.0. For continuous data, normality was assessed using the Kolmogorov-Smirnov test with Lilliefors correction. Student's t-test for independent samples was used to evaluate possible differences between the mean values of the study groups. The crude hazard ratio as well as the respective 95% confidence intervals (CIs) and absolute and relative frequencies were estimated for categorical data using the Pearson chi-squared test or Fisher's exact test. The odds ratio, obtained by logistic regression, overestimates the data, distorting the magnitude of the effect to be estimated. Therefore, a Poisson regression was used because this model best fit the data, especially in terms of the effect size and the confidence interval. Values with $p<0.20$ were included in the multivariate analysis. In the final multivariate model, performed using Poisson regression, values with $p<0.05$ were considered statistically significant. The study was approved by the Research Ethics Committee of the University of Caxias do Sul, Brazil, under No. 1,453,509.

## Results

In the period mentioned, there were 22,317 births. The prevalence of CMs was 1.5% (95% CI 1.4-1.7) (n=351). Birth defects most often affected multiple systems (28.5%, n=100),

followed by the genitourinary tract (18.2%, n=64), central nervous system (16.5%, n=58), musculoskeletal system (16.3%, n=57), gastrointestinal tract (10.8%, n=38), and cardiovascular system (9.7%, n=34) (Table 1). The mean weight of NBs allocated to the group of malformed NBs and to the group of healthy NBs was 2,529 g ±987 and 3,035 g ±785 (p<0.001), respectively.

**Table 1. Distribution of data according to the type of congenital malformation (n=351)**

| Malformations | n | % |
|---|---|---|
| Multiple malformations | 100 | 28.5 |
| Genitourinary system | 64 | 18.2 |
| Central nervous system | 58 | 16.5 |
| Musculoskeletal system | 57 | 16.3 |
| Gastrointestinal tract | 38 | 10.8 |
| Cardiovascular system | 34 | 9.7 |

The mean age of the pregnant women whose NBs had some type of CM was 25.8±7.6 years. There was no significant difference between the groups with and without CMs. CMs were more prevalent in pregnant women who self-reported as white (n=263; 1.6%), though this difference was not significant. In the bivariate analysis, the maternal variables that showed statistical significance were multiparity (≥ 4 children) [cHR=1.29 (1.01-1.65); p<0.04], and cesarean delivery [cHR=3.10 (2.49-3.85); p<0.001]. However, after controlling for epidemiological confounding factors in multivariate analysis, the final model showed primiparity as a protective factor against CM [cHR=0.59 (0.46-0.77)]; p<0.001] and cesarean delivery as the only risk factor (p<0.001) (Table 2).

**Table 2. Distribution of maternal variables according to the occurrence or absence of congenital malformations (n=351)**

| Variable | Congenital malformation | | | | Crude hazard ratio (95% CI) | *P*-value | Adjusted hazard ratio (95% CI) | *P* value |
|---|---|---|---|---|---|---|---|---|
| | **Yes** | | **No** | | | | | |
| | n | % | n | % | | | | |
| Color/Race | | | | | | | | |
| White | 263 | 1.6 | 15.697 | 98.4 | 1.12 (0.87- 1.44) | 0.373 | | |
| Non-White | 76 | 1.5 | 5.098 | 98.5 | 1.00 ref. | | | |
| Maternal age | | | | | | | | |
| ≤20 | 110 | 2.0 | 6.205 | 98.0 | 1.09 (0.87 – 1.37) | 0.449 | | |
| ≥40 | 15 | 2.0 | 684 | 98.0 | 1.35 (0.81 – 2.27) | 0.251 | | |
| 21-39 | 226 | 1.6 | 14.029 | 98.4 | 1.00 ref. | | | |
| Previous miscarriages | | | | | | | | |
| Yes | 59 | 1.8 | 3.218 | 98.2 | 1.11 (0.84 – 1.46) | 0.499 | | |
| No | 292 | 1.6 | 1.7744 | 98.4 | 1.00 ref. | | | |
| Use of illicit drugs | | | | | | | | |
| Yes | 11 | 1.0 | 1.099 | 99.0 | 0.88 (0.46 – 1.67) | 0.811 | | |
| No | 56 | 1.1 | 4.903 | 98.9 | 1.00 ref. | | | |
| Diabetic syndrome | | | | | | | | |
| Yes | 5 | 3.2 | 152 | 96.8 | 0.67 (0.27 – 1.67) | 0.538 * | | |
| No | 54 | 4.7 | 1.096 | 95.3 | 1.00 ref. | | | |
| Type of delivery | | | | | | | | |
| Cesarean | 172 | 3.3 | 5.059 | 96.7 | 3.10 (2.49 – 3.85) | <0.001 | 2.51 (1.99–3.16) | <0.001 *** |
| Vaginal | 147 | 1.1 | 1.3725 | 98.9 | 1.00 ref. | | 1.00 ref. | |

| | | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| Gestational age | | | | | | | | |
| 22 to 34 | 89 | 5.4 | 1.572 | 94.6 | 4.08 (3.22– 5.17) | <0.001 | 0.80 (0.58– 1.11) | 0.180 |
| 34.1 to 41 | 252 | 1.3 | 1.8942 | 98.7 | 1.00 ref. | | 1.00 ref. | |
| No. of pregnancies | | | | | | | | |
| 1 | 81 | 1.1 | 7.494 | 98.9 | 0.59 (0.46 – 0.77) | <0.001 | 0.56 (0.46-0.74) | <0.001 *** |
| 2 to 3 | 166 | 1.8 | 9.122 | 98.2 | 1.00 ref. | | 1.00 ref. | |
| ≥ 4 | 103 | 2.3 | 4.351 | 97.7 | 1.29 (1.01 – 1.65) | 0.040 | 1.27 (0.98-1.65) | 0.08 |

Note: Diabetes: Diabetic syndrome (composed of gestational diabetes mellitus/type 1 diabetes mellitus/type 2 diabetes mellitus). * Fisher's exact test: 1.00 ref: reference category. *** Statistically significant results after fitting the Poisson regression. Gestational age in weeks. Preg: number of pregnancies. Maternal age in full years.

The presence of CM doubled the odds of the 5-minute Apgar score being between 0 and 3 [HR=2.09 (1.21-2.12); p<0.01] and almost tripled the odds of the Apgar score being between 4 and 7 [HR=4.28 (3.12-5.87); p<0.001]. The odds of infant mortality were ten times higher in the group of NBs with CM [HR=10.34 (8.1 - 13.0)]; p<0.001], and the odds of stillbirth were 25 times higher [HR=31.0 (22.0-42.0)]; p<0.001]. There was no predominance of either sex among the NBs of either group (Table 3).

**Table 3. Distribution of neonatal variables according to the occurrence or absence of congenital malformations (n=351)**

| Variable | Congenital Malformation | | | | Crude hazard ratio (95% CI) | *P*-value | Adjusted hazard ratio (95% CI) | *P* value |
|---|---|---|---|---|---|---|---|---|
| | Yes | | No | | | | | |
| | n | % | n | % | | | | |
| Sex of NB | | | | | | | | |
| Male | 36 | 1.1 | 3.161 | 98.9 | 1.20 (0.73 – 2.00) | 0.453 | | |
| Female | 27 | 1.0 | 2.872 | 99.0 | 1.00 ref. | | | |
| 1-minute Apgar score | | | | | | | | |
| 0-3 | 78 | 6.9 | 1.050 | 93.1 | 7.47 (5.71 – 9.76) | <0.001 | 0.79 (0.47 – 1.35) | 0.390 |
| 4-7 | 72 | 1.9 | 3.709 | 98.1 | 2.05 (1.55 – 2.72) | <0.001 | 0.79 (0.58 – 1.07) | 0.130 |
| 8-10 | 146 | 1.0 | 15.625 | 99.0 | 1.00 ref. | | | |
| 5-minute Apgar score | | | | | | | | |
| 0-3 | 43 | 14.6 | 251 | 85.4 | 13.71 (10.08 – 18.65) | <0.001 | 2.09 (1.21 – 2.12) | <0.010*** |
| 4-7 | 45 | 4.6 | 940 | 95.4 | 4.28 (3.12 – 5.87) | <0.001 | 2.83 (1.53 – 3.72) | <0.001*** |
| 8-10 | 208 | 1.1 | 19.300 | 98.9 | 1.00 ref. | | | |
| Fetal evolution | | | | | | | | |
| Infant mortality | 191 | 5.8 | 3.108 | 94.2 | 10.3 (8.1-13.0) | <0.001 | 10.34 (7.79-13.72) | <0.001*** |
| Stillbirth | 59 | 16 | 279 | 84 | 31.0 (22.0-42.0) | <0.001 | 25.56 (24.61 – 64.44) | <0.001*** |
| Healthy | 99 | 0.6 | 17.450 | 99.4 | 1.00 ref. | | | |
| Weight (SD) | 2.529 (987) | | 3.035 (785) | | | <0.001 *** | | < 0.480 |

Note: 1.00 ref.: reference category. *** Statistically significant results after fitting the Poisson regression. Weight: birthweight in grams. SD: standard deviation.

Table 4 shows the distribution of maternal variables associated with CM stratified by organ system. There was no association between the occurrence of system-stratified CM and maternal age, diabetic syndrome, or miscarriages before the pregnancy studied. The primiparity was still associated with CM after the multivariate analysis.

**Table 4. Distribution of maternal variables associated with the groups of congenital malformations (n=351)**

| Variable | Gastrointestinal (n) % | Genitourinary (n) % | CNS (n) % | MSS (n) % | Cardiovascular (n) % | Multiple (n) % | *p* * |
|---|---|---|---|---|---|---|---|
| Age | | | | | | | |
| < 20 years | (16) 42.1 | (16) 24.6 | (13) 22.4 | (21) 37.5 | (5) 14.7 | (11) 39.2 | NS |
| > 40 years | (1) 2.6 | (3) 4.6 | (2) 3.4 | (2) 3.6 | (1) 2.9 | (16) 57.1 | NS |
| 21-39 years | (21) 55.3 | (46) 70.8 | (43) 74.2 | (33) 58.9 | (28) 82.4 | (1) 3.7 | |
| Diabetic Syndrome | | | | | | | |
| Yes | (1) 50 | (0) 0 | (1) 7.7 | (2) 15.4 | (0) 0 | (1) 10 | NS |
| No | (1) 50 | (8) 100 | (12) 92.3 | (11) 84.6 | (5) 100 | (6) 90 | |
| Previous miscarriages | | | | | | | |
| Yes | (5)13.6 | (9)13.8 | (13) 22.4 | (10) 17.9 | (4) 11.8 | (4) 8 | NS |
| No | (33)86.8 | (56)86.2 | (945) 77.6 | (46) 82.1 | (30) 88.2 | (24) 92 | |
| Number of pregnancies | | | | | | | |
| 1 | (13) 34.2 | (13) 20.3 | (81) 23.1 | (11) 19.6 | (10) 29.4 | (39) 43* | <0.05 |
| > 4 | (13) 34.2 | (33) 28.1 | (103) 29.4 | (12) 21.4 | (7) 20.6 | (13) 10 | NS |
| 2 to 3 | (12) 31.6 | (18) 51.6 | (166) 47.4 | (33) 59.0 | (17) 50 | (42) 47 | |

Note: CNS: central nervous system; MSS: musculoskeletal system; Multiple: multiple malformations. * Student's t-test for independent samples. p<0.05: statistically significant results

Table 5 shows an association (p<0.001) between stillbirth and malformations of the musculoskeletal system. Infant mortality was associated with all types of malformations.

**Table 5. Distribution of the association between stillbirth and infant mortality and the systems affected by congenital malformations (n=351)**

| | Gastrointestinal n (%) | Genitourinary n (%) | CNS n (%) | MSS n (%) | Cardiovascular n (%) | Multiple n (%) | *p** |
|---|---|---|---|---|---|---|---|
| Stillbirth | | | | | | | |
| Yes | (0) | (2) 4.5 | (14) 70* | (6) 22* | (1) 12.5 | (7) 87.5 | <0.001* |
| No | (9) 100 | (42) 95.5 | (6) 30 | (28) 78 | (7) 87.5 | (1) 12.5 | |
| Infant mortality | | | | | | | |
| Yes | (29) 76* | (21) 33* | (36) 86* | (20) 42* | (26) 79* | (20) 95* | <0.001* |
| No | (9) 24 | (42) 67 | (6) 14 | (28) 58 | (7) 21 | (1) 5 | |

Note: CNS: central nervous system; MSS: musculoskeletal system; Multiple: multiple malformations. * p<0.05: statistically significant results

**Discussion**

The GH of Caxias do Sul accounts for approximately 50% of the births covered by the SUS in 49 municipalities in the region, so it can be an important and representative population sample. In our study, the prevalence of CMs was 1.5%, similar to data from other studies. The EUROCAT study, a 32-year cohort in European countries, found a CM prevalence that ranged from 1.4% to 4.1%. In some regions of Croatia, Ireland, and England, the prevalence of CM was 1.5% (Karbasi et al., 2009). A study conducted in Asia with 4,800 births showed a prevalence of 2.8% (Karbasi et al., 2009). In Brazil, among 487,953 live births in the municipality of Rio de Janeiro between 2000 and 2004, the CM prevalence was 0.9% (Reis, da Silva Santos, & Mendes, 2011). Almeida et al. (2016) showed a prevalence of 2.4% among 275 births. Another study, conducted between 2002 and 2011, identified a prevalence of 0.5% in the population analyzed (dos Santos Rodrigues et al., 2014). A recent study conducted in the state of Rio Grande do Sul (Luz, Karam, & Dumith, 2019) of 1,386,803 births from 2005 to 2014 found 12,818 (0.92%) with CM, corresponding to an overall mean rate of 9.2 per 1,000

births (95% CI 8.4-10.3). According to the authors, CMs were associated with an Apgar score lower than 7, birthweight ≤ 1.500 g, and gestational age ≤ 31 weeks.

Calone et al. (2009) found a prevalence of CM of 1.7%, and out of 172 CM births, cesarean delivery was the most common type of delivery. Other authors have shown results that confirm the association between different CM types and cesarean delivery, especially in a population with high percentages of CNS and gastrointestinal tract malformations (Fontoura, & Cardoso, 2014; Souza, Amorim, & Porto, 2010). However, in the aforementioned study, all patients whose NBs exhibited this type of birth defect underwent elective cesarean section (Calone et al., 2009). In a study conducted in Rio de Janeiro that evaluated the prevalence of CM between 2000 and 2006, cesarean delivery was the type of delivery in 10.3% of cases (Reis, da Silva Santos, & Mendes, 2011). In a study conducted at GOD/GH from 1998 to 2008 by Pante et al. (2011) that evaluated central nervous system malformations, cesarean delivery was the most frequent type of delivery, which was related to the attempt to avoid dystocia, especially in cases exhibiting volumetric increase of the fetal head. In CMs with anencephaly, the type of delivery chosen was the one that best preserved maternal health, considering the reserved prognosis of the NB. When the CM was accompanied by spina bifida, encephalocele, meningocele, or myelomeningocele, cesarean delivery was indicated mainly in cases that had a better prognosis to preserve the integrity of the herniated material (Pante et al., 2011).

Although there is no unanimity regarding the type of delivery, the rate of elective cesarean section can be increased to obtain successful perinatal management through delivery planning with a multidisciplinary team, as shown by Almeida et al. (2016) Other studies reported cesarean section rates of 3% and 34% (Almeida et al., 2016; Souza, Amorim, & Porto, 2010; Pante et al., 2011). Therefore, fetal prognosis must be assessed by analyzing the extent of the defect, the presence and progression rate of damage to the CNS, and other associated anomalies, such as macrocrania, with these factors determining the choice of delivery method and the best time to perform it (Pante et al., 2011).

As seen in Table 3, birthweight was significantly different between cases with and without CM in the bivariate analysis [2,529±987 g *vs.* 3,035±785 g ($p<0.001$)], but not in the multivariate analysis ($p=0.480$). In an Iranian study, the prevalence of NBs with low birthweight and CM was 22% (Daliri et al., 2019). Alijahan et al. (2013) saw a statistically significant association between the risks of low birthweight and CM. In the study by Khatami and Mamuri (2005), a significant relationship was observed between low birthweight and CM prevalence. Multiple births seem to induce a simultaneous increase in the low-birthweight rate (Daliri et al., 2019; Bazyar et al., 2015) and the risk of CM.

Regarding sex, we saw no association between CM and infant sex. A study conducted in China between 2004 and 2014, which evaluated urinary and renal CMs, recognized male sex as a risk factor (OR 1.83; 95% CI 1.53-2.19) for CM (Tain et al., 2016). In Iran, 136 NBs with CM were evaluated, and 51.9% of them were male (Karbasi et al., 2009). Also in Iran (Daliri et al., 2019), and based on a meta-analysis of the relationship between the sex of the NB and the CM prevalence, the risk observed in male NBs was 1.25 times that among female NBs. In the same study, there was no significant association between the risk of CM and the history of miscarriage or the type of delivery. In our study, which corroborates the findings by Lei (1992), the sex of the NBs was equally distributed in each group.

In the current study, multiparity was associated with 1.27 times the odds of CM than primiparity in the bivariate analysis, but this was not confirmed in the multivariate analysis. One study has reported that multiparous women have a higher risk of having children with CM than primiparous women (Almeida et al., 2016).

A study conducted in 2016 to identify risk factors and neonatal outcomes, including CM, showed that one risk factor was a 5-minute Apgar score lower than 7, which seems to be associated with complications at the time of delivery, difficult fetal extraction, dystocia,

skeletal dysplasia, fetal hydrops, pulmonary hypoplasia, and low birthweight (Almeida et al., 2016). These results are corroborated by the present study in GOD/GH, which also found that a 5-minute Apgar score between 0 and 3 [aHR 2.09 (1.21-2.12); p<0.010] or between 4 and 7 [aHR 2.83 (1.53-3.72); p<0.001] was associated with severe neonatal malformation in the population with CMs, as was perinatal mortality, encompassed by infant mortality [aHR 10.34 (7.79 to 13.72); p<0.001] and stillbirth [aHR 25.56 (24.61-64.44); p<0.0001]. Gaiva, Fujimori, and Sato (2014) reported that neonatal mortality was associated with <7 prenatal visits (OR=3.80; CI: 1.66-8.70), gestational age <37 weeks (OR=4.77; CI: 1.48-15.38), 1-minute Apgar score <7 (OR=4.25; CI: 1.84-9.81), 5-minute Apgar score <7 (OR=5.72; CI: 2.24-14.60), and the occurrence of congenital anomalies (OR=14.39; CI: 2.72-76.09) (Pinto, & Nascimento, 2007; Gaiva, Fujimori, & Sato, 2014). Additionally, according to Gaiva, Fujimori, and Sato (2014), a significant association was observed between congenital anomalies and shorter gestational length at delivery, higher number of dead children, type of delivery, low birthweight, and lower Apgar score.

The different CM types are presented in different ways in the scientific literature. In our study population, multiple malformations (28.5%) were the most prevalent, followed by genitourinary (18.2%), central nervous system (16.5%), skeletal muscle (16.2%), and cardiovascular anomalies (9.7%). A Chinese study conducted between 2012 and 2014 showed a prevalence of congenital heart disease of 2.3% (Sun et al., 2017), which corroborates data obtained from a Swedish cohort (2001 to 2014) involving 43,550 infants with CM (Persson et al., 2017), that found prevalence of 1.6%, 0.5%, 0.4%, 0.2%, 0.1%, and 0.1% for CMs of the cardiac system, genitourinary tract, upper and lower limbs, digestive tract, CNS, and orofacial region, respectively. In a systematic review conducted between 1986 and 2015 in Saudi Arabia and neighboring countries, the overall prevalence of orofacial clefts ranged from 0.3 to 2.4 per 1,000 live births (Sabbagh, Mossey, & Innes, 2012). In a Colombian study conducted between 1999 and 2008, which analyzed the percentage of fetal and neonatal death in infants with CMs, 32% of CMs were congenital heart diseases, 15.8% were CNS anomalies, and 8% were chromosomal anomalies (Roncancio et al., 2018). The same study reported that the fetal mortality rate due to CM was 9.9/10,000 live births, and the neonatal mortality rate was 20.8/10,000 live births (Roncancio et al., 2018). A European study conducted between 1998 and 2011 analyzed 73,337 births and found a perinatal mortality rate of 1.27:1,000 live births with congenital anomalies. The percentages of stillbirth and early and late neonatal mortality were 2.7%, 2.8%, and 0.9%, respectively (Groen et al., 2017).

The present study was performed with data from a hospital database, which limits some conclusions and imposes restrictions due to the retrospective design. In addition, for many of our cases, we did not obtain permission from the relatives to perform necropsy, which hindered a better characterization of the birth defect. Genetic evaluation was not routinely available in GOD/GH, and when performed in other departments, it was not recorded in the GH database. Finally, the difficulty in establishing a universal nomenclature that could distinguish the many types of congenital anomalies became evident, which made it difficult to compare results among similar studies. We would benefit from measures that raised the awareness of physicians about the need to better and more accurately record CMs in the hospital information system and in the neonatal medical records.

## Conclusion

The prevalence of CM in this population was 1.5%. or 1 per 64 births. Multiple anomalies were the most prevalent type, followed by CMs of the genitourinary tract, central nervous system, musculoskeletal system, gastrointestinal tract, and cardiovascular system. The presence of CM was associated with higher rates of cesarean delivery, low 5-minute Apgar

score—which characterizes a significant state of asphyxia and acidosis—and higher infant mortality and stillbirth rates.

Despite the cited limitations, the study described a qualitative and quantitative assessment of the profile of pregnant women and newborns over a period of 17 years. The results presented here were from multidisciplinary care provided by a maternal-fetal health care service in the northeast region of Rio Grande do Sul. Data on populations of NBs with CM identified in specialized services are essential for the management of care and of hospital expenses. Congenital anomalies, when present, contribute significantly to the increase in care costs due to longer hospitalization, more surgical interventions, and the need for an integrated team of specialists. It should be emphasized that this population requires continuous, prolonged, special care related to parallel comorbidities, psychosocial problems triggered in relatives, difficulties in social adjustment, and, consequently, greater demand for public health care services.

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