INTERNATIONAL SEVEN JOURNAL
OF HEALTH RESEARCH
ISSN: 2764-9415

INCIDENCE OF CONGENITAL HEART DISEASE IN THE MUNICIPALITIES OF NORTHWESTERN SÃO PAULO: AN ANALYSIS BASED ON DATASUS

https://doi.org/10.56238/isevjhv4n1-006

Receipt of originals: 01/20/2025 **Acceptance for publication:** 02/20/2025

Mariana Gatto Juliano[1], Ellie Daiane Teodoro[2], Larissa Vilela Perroni Silva Soler[3], Rafael Alves de Oliveira[4], Cindy Larielli Vasconcelos[5], Sthefany Cristine Avila Mendes[6], Ana Clara Antonino Pereira[7], Matheus Sousa Ferreira[8], Lucas Martins Kamiyama[9], Alba Regina de Abreu Lima[10] and Uderlei Doniseti Silveira Covizzi[11]

ABSTRACT

The chromosomopathies that generate congenital heart diseases represent one of the main challenges for neonatal public health in Brazil, affecting about 30 thousand newborns annually. Data from DATASUS and the Information System on Live Births (SINASC) indicate significant underreporting, with only 5.3% of the estimated cases officially registered. The global prevalence of 9 cases per 1,000 live births is used as a parameter to estimate the national figures, which point to a significant burden of the disease. About 40% of affected newborns require surgical intervention in the first year of life, highlighting the importance of early diagnosis and specialized follow-up. This article discusses the challenges related to data collection, the formulation of public policies, and strategies to improve the care of patients with chromosomal diseases in the Northwest of São Paulo.

Keywords: Chromosomopathies. Heart Diseases Congenital. Neonatal Health.

[1] Undergraduate student in Medicine at Universidade Brasil – Fernandópolis.
LATTES: http://lattes.cnpq.br/1993837818012776
[2] Undergraduate student in Medicine at Universidade Brasil – Fernandópolis.
LATTES: http://lattes.cnpq.br/1375746972249051
[3] Undergraduate student in Medicine at Universidade Brasil – Fernandópolis.
LATTES: http://lattes.cnpq.br/9964946949053610
[4] Graduating in Medicine at Universidade Brasil – Fernandópolis.
LATTES: https://lattes.cnpq.br/0952211920930078
[5] Undergraduate student in Medicine at Universidade Brasil – Fernandópolis.
LATTES: https://lattes.cnpq.br/0561573212272139
[6] Undergraduate student in Medicine at Universidade Brasil – Fernandópolis.
LATTES: https://lattes.cnpq.br/2056708369922294
[7] Undergraduate student in Medicine at Universidade Brasil – Fernandópolis.
LATTES: https://lattes.cnpq.br/5451362117873669
[8] Graduating in Medicine at Universidade Brasil – Fernandópolis.
LATTES: https://lattes.cnpq.br/8655238169822199
[9] Graduating in Medicine at Universidade Brasil – Fernandópolis.
LATTES: http://lattes.cnpq.br/0833186582433013
[10] Professor of Medicine at FAMERP – São José do Rio Preto.
LATTES: http://lattes.cnpq.br/0802737766517359
[11] Professor at UNORTE, UNIFEV and Universidade Brasil.
LATTES: http://lattes.cnpq.br/3432578311647024

INTRODUCTION

Chromosomal abnormalities during pregnancy, although not very frequent, represent a risk of perinatal mortality (Silva, Vasconcellos, 2021). It is observed worldwide that pregnancy at an advanced maternal age is an increasingly frequent reality in obstetric practice, with the occurrence of gestational diabetes, gestational hypertension, chromosomopathies, fetal death, cesarean section, and congenital malformation being common (Costa, 2023). Chromosomal diseases and congenital heart defects are conditions of great medical relevance due to their significant impact on child health and clinical management. Congenital heart diseases, which comprise a set of structural anomalies of the heart present at birth, represent one of the main causes of morbidity and mortality in newborns. Chromosomopathies, on the other hand, which involve numerical or structural alterations in chromosomes, such as Down syndrome (trisomy 21), are often associated with a wide spectrum of clinical manifestations, including heart diseases. Over the years, studies involving classical gene mapping and subsequent large-scale exome and genome sequencing have provided ample evidence that alterations of nuclear function can cause developmental defects and heart disease in adults (Bertero, Rosa-Garrido, 2020).

The relationship between these conditions is striking, since chromosomal diseases often predispose to congenital malformations, including heart defects. For example, more than 40% of patients with Down syndrome have some form of congenital heart disease. This intersection requires a multidisciplinary approach, from prenatal diagnosis, made possible by advances such as genetic testing and fetal echocardiography, to specialized clinical and surgical follow-up.

The incidence and epidemiological profile of these conditions, especially in specific regions, such as the northwest of São Paulo, is essential to improve the planning of public health policies and optimize care, contributing to the reduction of complications and increase the quality of life of patients.

DEFINITION AND RELEVANCE

Congenital heart defects represent one of the main malformations in newborns, with an estimated global prevalence of 8 to 10 cases per 1,000 live births. These conditions involve anatomical changes in the heart and great vessels, with clinical manifestations ranging from asymptomatic to severe conditions that require immediate

interventions. On the other hand, chromosomal diseases, such as Down syndrome (trisomy 21), result from changes in the number or structure of chromosomes, and are often associated with comorbidities, including heart defects.

INTERSECTION BETWEEN CONGENITAL HEART DEFECTS AND CHROMOSOMAL DISORDERS

The association between chromosomal diseases and congenital heart diseases is widely documented. About 40% to 50% of individuals with Down syndrome have heart diseases, such as atrioventricular septal defect. These conditions require specific diagnostic and therapeutic approaches, reinforcing the importance of early screening by tests such as echocardiograms and genetic testing.

EPIDEMIOLOGICAL DATA IN BRAZIL

In Brazil, DATASUS is a fundamental tool for understanding the magnitude and patterns of these conditions. The platform allows access to information on births, mortality and hospital admissions associated with congenital heart diseases and chromosomal diseases. An important instrument to analyze the significance of this issue in the country is the Information System on Live Births (SINASC) (Carvalho *et al*, 2021). According to SINASC data, the rate of congenital malformations reported at birth has increased, which may reflect greater diagnostic coverage.

In regions such as the northwest of São Paulo, it is possible to identify specific epidemiological patterns, helping in the planning of public health actions. Data from the Hospital Information System (SIH/SUS) indicate that hospitalizations due to congenital malformations and complications related to chromosomal diseases are among the main causes of neonatal hospitalization. A study by Moreno and Moreira (2023) demonstrated that high parental age is associated with congenital malformations and genetic syndromes, constituting an important factor for genetic counseling of couples. They also associated parental age with congenital malformations and genetic syndromes and syndromes.

IMPORTANCE OF USING DATASUS DATA

DataSUS plays a crucial role in monitoring the health conditions of the population. It allows the identification of temporal trends, comparison between

municipalities and states, in addition to subsidizing public policies. In the case of congenital heart diseases and chromosomal diseases, the data can guide:

- The strengthening of early diagnosis programs.
- The allocation of resources for corrective surgeries and supportive therapies.
- The training of health teams for specialized management.

MULTIDISCIPLINARY APPROACH

The integration of DATASUS data with clinical strategies and public policies is essential to improve the survival and quality of life of patients with these conditions. Regional programs aimed at early diagnosis, access to treatment, and social support are examples of how epidemiological data can be applied in a practical and efficient way.

Studies based on DataSUS data have the potential to expand knowledge about the epidemiology of congenital heart diseases and chromosomal diseases, especially in regional contexts such as the northwest of São Paulo. Understanding the distribution, associated factors, and clinical outcomes of these conditions is an essential step to improve neonatal, genetic, and pediatric health care in Brazil.

METHODOLOGY

This is a descriptive and retrospective study, based on vital statistics obtained from the Health Information System (TABNET), accessible by the Department of Informatics of the Unified Health System (DATASUS), referring to the last ten years.

The data were analyzed among live births with geographic coverage in Brazil by municipalities. The variables selected were municipality, mother's age, and births to the mother's residence in the period from 2013 to 2023.

The selected municipalities of residence were all those belonging to the Regional Department of Health XV – São José do Rio Preto of the state of São Paulo, namely: Adolfo, Álvares Florence, Américo de Campos, Aparecida D'oeste, Ariranha, Aspasia, Bady Bassit, Bálsamo, Cardoso, Catanduva, Catiguá, Cedral, Cosmorama, Dirce Reis, Dolcinópolis, Elisiário, Embaúba, Estrela D'oeste, Fernandópolis, Fernando Prestes, Floreal, Gastão Vidigal, General Salgado, Guapiaçu, Guarani D'oeste, Ibirá, Icém, Indiaporã, Ipiguá, Irapuã, Itajobi, Jaci, Jales, José Bonifácio, Macaubal, Macedonia, Magda, Marapoama, Marinópolis, Mendonça, Meridiano, Mesópolis, Mira Estrela,

Mirassol, Mirassolândia, Monções, Monte Aprazível, Neves Paulista, Nhandeara, Nipoã, Nova Aliança, Nova Canaã Paulista, Nova Granada, Novais, Novo Horizonte, Onda Verde, Orindiúva, Ouroeste, Palestina, Palmares Paulista, Palmeira D'oeste, Paraíso, Paranapuã, Parisi, Paulo de Faria, Pedranópolis, Pindorama, Pirangi, Planalto, Poloni, Pontalinda, Pontes Gestal, Populina, Potirendaba, Riolândia, Rubinéia, Sales, Santa Adélia, Santa Albertina, Santa Clara D'oeste, Santa Fé do Sul, Santa Rita D'oeste, Santa Salete, Santana da Ponte Pensa, São Francisco, São João das Duas Pontes, São João de Iracema, São José do Rio Preto, Sebastianópolis do Sul, Tabapuã, Tanabi, Três Fronteiras, Turmalina, Ubarana, Uchoa, União Paulista, Urânia, Urupês, Valentim Gentil, Vitória Brasil, Votuporanga, Zacarias.

In addition, another search was performed repeating all the selections from the first and including the filter: Type of congenital anomaly – NCOP chromosomal abnormalities (Not Elsewhere Classified).

The data were tabulated in Microsoft Excel, analyzed based on their absolute frequency and, from this, graphs were created to discuss the results. Because these were data in the public domain, the study did not require approval by the Ethics Committee.

RESULTS AND DISCUSSIONS

The descriptive analysis of the data collected in TABNET/DATASUS revealed significant information about the profile of congenital anomalies and chromosomal diseases in the municipalities belonging to the Regional Health Department XV (DRS XV) of São José do Rio Preto, from 2013 to 2023. The results are organized based on the variables studied:

1. Absolute Frequency of Live Births and Chromosomal Abnormalities

   Between 2013 and 2023, approximately 199,419 live births were registered in the region covered by DRS XV. Of the total number of live births, 59 cases were diagnosed with NCOP chromosomal abnormalities, representing about 0.03% of the total births.

2. Distribution by Municipalities

   São José do Rio Preto led in absolute number of live births, corresponding to 30% of the total in the region, followed by Catanduva (7.21%) and Fernandópolis (4.18%). Smaller municipalities, such as Zacarias and Dirce

Reis, recorded the lowest number of cases of chromosomal abnormalities, which can be attributed to the low population density.

3. Maternal Age and Prevalence of Chromosomal Abnormalities

The prevalence of chromosomal abnormalities was correlated with maternal age, being more frequent in mothers over 35 years of age (74.5% of cases). This data reinforces the importance of prenatal care in high-risk pregnancies.

4. Temporal Evolution

There was a slight increase in reported cases of chromosomal abnormalities over the 10 years analyzed. This may reflect improvements in diagnosis, increased awareness of healthcare professionals, or advances in data recording.

Studies by Nascimento (2021) and collaborators pointed to a frequency of 10% of patients who were referred to a general genetics outpatient clinic had a karyotype indicative of chromosomal abnormalities. These data indicated the importance of adequate cytogenetic investigation for diagnosis and family genetic counseling in situations of congenital defects, intellectual disability, pathological short stature in girls, primary amenorrhea, infertility, recurrent miscarriage and clinical suspicion of chromosomal syndromes.

The results point to epidemiological trends relevant to the understanding of chromosomal diseases in the northwestern region of São Paulo:

1. Distribution Standards

The higher concentration of births and chromosomal abnormalities in larger municipalities, such as São José do Rio Preto, can be explained by the higher population density and the availability of more specialized health services. On the other hand, smaller municipalities have potential underreporting due to limitations in access to diagnosis.

2. Impact of Maternal Age

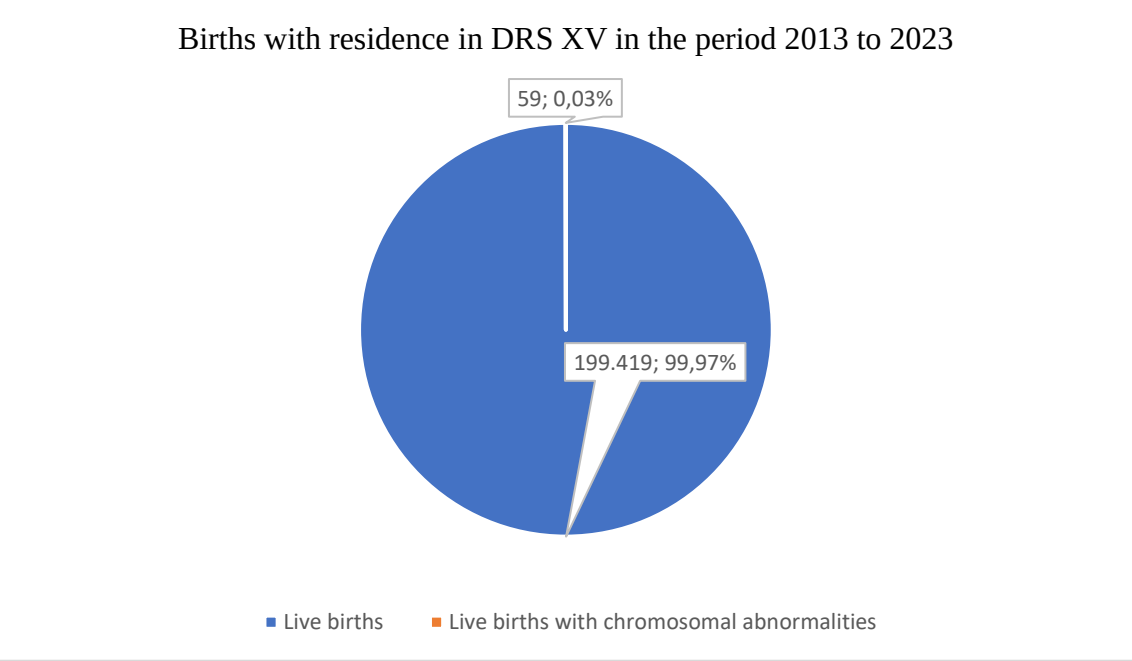
The association between advanced maternal age and a higher prevalence of chromosomal abnormalities is in accordance with the literature, which points to a higher risk for trisomies, such as Down syndrome, in mothers over 35 years of age. These data highlight the importance of public policies aimed at family planning and genetic screening in high-risk pregnancies.

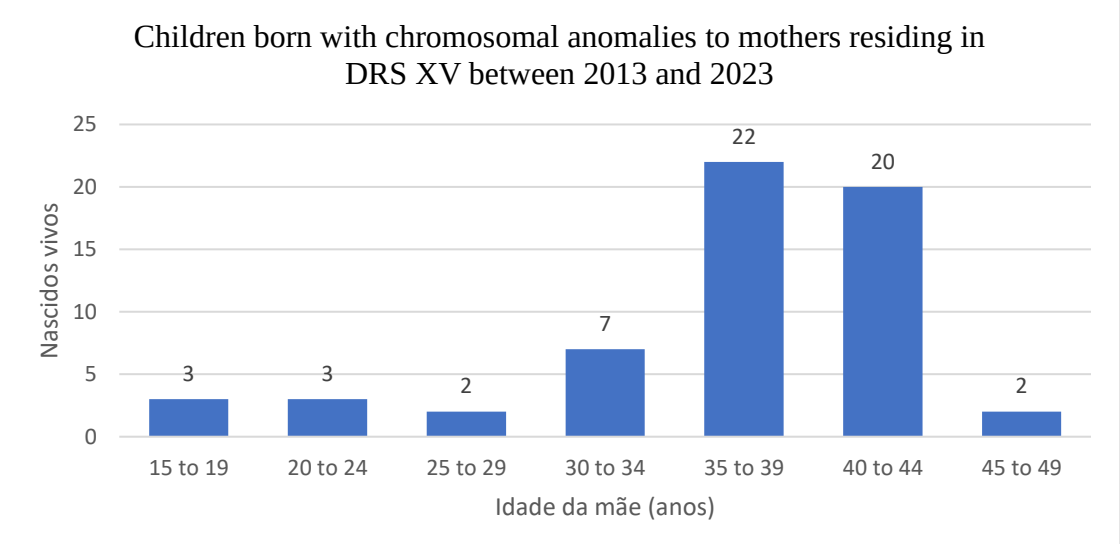
3. Temporal Evolution and Diagnosis

The increase in the number of diagnoses of chromosomal abnormalities over the decade may be related to improvements in registration systems and access to technology for prenatal diagnosis, such as karyotype examinations and advanced ultrasounds.

4. Regional Perspectives and Public Health Strategies

The regional analysis highlights the need for greater attention in smaller municipalities, where health infrastructure is limited. Investments in training health professionals and early diagnosis are essential to ensure equity in care.





CONCLUSION

Congenital heart diseases have always been defined as a multifactorial etiology that affects about 1% of live births. Recent advances in the identification of genetic factors have shown great importance in the clinical and surgical treatment of syndromic and non-syndromic heart defects (Saliba *et al*, 2020). Studies suggest that variations in specific genes, such as the transcription factors T-box, NKx, and GATA may increase the risk of congenital heart defects, but it is also recognized that environmental factors, such as exposure to certain substances during pregnancy, may play a significant role in congenital heart defects (Wolf, Craig, 2010, Barros, *et al*. 2023).

The data obtained from DataSUS highlight the relevance of regional analyses to understand the impact of chromosomal diseases on births and to plan effective public health actions. The implementation of screening programs and the expansion of access to specialized care can contribute to the improvement of neonatal outcomes in the DRS XV region and in other areas of Brazil.

REFERENCES

1. BARROS, Eliab Batista *et al*. Perfil epidemiológico de nascidos vivos com cardiopatia congênita nas regiões brasileiras. *Brazilian Journal of Implantology and Health Sciences*, v. 5, n. 5, p. 2316-2328, 2023.

2. BERTERO, A.A., ROSA-GARRIDO, M. Three-dimensional chromatin organization in cardiac development and disease, *Journal of Molecular and Cellular Cardiology,* Volume 151, 2021, Pag 89-105. https://doi.org/10.1016/j.yjmcc.2020.11.008.

3. BRASIL Ministério da Saúde. Política Nacional de Atenção Integral à Saúde da Criança: orientações para promoção, proteção e cuidado. Brasília: *Ministério da Saúde*, 2018.

4. CARVALHO, N. B. *et al.* Levantamento e análise dos dados sobre anomalias congênitas nos recém-nascidos em município de minas gerais. *Recisatec-revista científica saúde e tecnologia*, v. 1, n. 4, p. e1445-e1445, 2021.

5. COSTA, A. R. F. Impacto da idade materna avançada nos desfechos obstétricos e perinatais. 2023. Tese de Doutorado. https://repositorio.ulisboa.pt/handle/10451/61967 Acesso em 12/02/2025.

6. DATASUS – Departamento de Informática do Sistema Único de Saúde.

7. LIMA, M. C.; MONTEIRO, C. A.; SILVA, A. A. M. Prevalência de anomalias congênitas no Brasil: uma revisão de dados do SINASC. Revista Brasileira de Saúde Materno Infantil, v. 20, n. 4, p. 567-577, 2020.

8. Ministério da Saúde. DATASUS. Tabnet. Brasília, DF: Ministério da Saúde, 2024. Disponível em: <https://datasus.saude.gov.br/nascidos-vivos-desde-1994>. Acesso: 06 nov. 2024.

9. Ministério da Saúde. Manual de instruções para preenchimento da Declaração de Nascidos Vivos (DN). Brasília: *Ministério da Saúde*, 2011.

10. MORENO, V. C.S. R., MOREIRA, L. M. A. Distúrbios congênitos e idade parental em um programa de genética comunitária: estudo retrospectivo. Revista de Ciências Médicas e Biológicas, v. 22, n. 2, p. 274-282, 2023.

11. NASCIMENTO, A. C. P., de ABREU, T., LIBERATO, É. L. A., dos SANTOS, T. R., MARSIGLIO, H. P., MELO, D. G. Anormalidades cromossômicas identificadas em um serviço de genética médica do interior do Estado de São Paulo, Brasil. *Saúde e Desenvolvimento Humano*, *9*(3), 1-11. 2021.

12. Organização Mundial da Saúde (OMS). Relatório sobre malformações congênitas e sua relação com determinantes genéticos e ambientais. Geneva: OMS, 2021.

13. Plataforma TABNET. Disponível em: http://www.datasus.gov.br. Acesso em: dezembro de 2024.

14. SALIBA, A., FIGUEIREDO, A.C.V., BARONEZA, J. E., AFIUNE, J. Y., PIC-TAYLOR, A., OLIVEIRA, S.F.D., MAZZEU, J.F. Genética e genômica na cardiopatia congênita: uma revisão clínica. *Jornal de Pediatria*, v. 96, p. 279-288, 2020.

15. SILVA, A.H., VASCONCELLOS, M.J.A. Atualização no rastreio de Cromossomopatias. *Revista da faculdade de medicina de teresópolis* v. 5, n.1, (2021).

16. VIEIRA, A. C.; PEREIRA, K. R.; OLIVEIRA, A. J. Associação entre idade materna e prevalência de trissomias: estudo de base populacional no Brasil. *Journal of Medical Genetics,* v. 57, p. 293-300, 2022.

17. WOLF, M., CRAIG, T. B. The molecular genetics of congenital heart disease: a review of recent developments. *Cur Op Card.* 2010; 25(3): 192.