Research on Current Status and Countermeasures for Comprehensive Prevention and Control of Birth Defects under the "321" Hierarchical Architecture: A Case Study of Nanning City

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Abstract

Through the application of the "321" hierarchical architecture comprehensive prevention and control model for birth defects, this study analyzes and investigates the current status and countermeasures of birth defect prevention and control in Nanning City. With Nanning City as the research subject, the "321" hierarchical architecture comprehensive prevention and control model for birth defects was implemented for the management and treatment of neonatal birth defects among newborns with Nanning household registration within the jurisdiction of Nanning City. Statistics were compiled on an annual basis, and birth defect indicators before and after the implementation of the model were compared to explore its practicality and rationality for broader implementation and promotion. Analysis demonstrates that following the implementation of the "321" hierarchical architecture comprehensive prevention and control model for birth defects, both the incidence rate of severe fatal and disabling birth defects and the birth rate of severe fatal and disabling birth defects among perinatal infants with Nanning household registration exhibited a significant declining trend, while the intervention rate for severe fatal and disabling birth defects was substantially improved. The "321" hierarchical architecture comprehensive prevention and control model for birth defects can effectively reduce the incidence of birth defects and enable timely and effective intervention for severe fatal and disabling birth defects.

Full Text

Research on the Current Status and Countermeasures of Comprehensive Birth Defect Prevention and Control Under the "321" Hierarchical Structure: A Case Study of Nanning City

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Abstract

This study analyzes and investigates the current status and countermeasures of birth defect prevention and control in Nanning City through the application of the "321" hierarchical architecture model for comprehensive birth defect prevention and control management. Taking Nanning City as the research subject, we implemented this model to manage and address birth defects among newborns with household registration in Nanning. Using annual statistical comparisons of birth defect indicators before and after implementation, we explored the model's practicality and rationality for broader promotion. Our analysis revealed that following implementation, both the incidence rate and birth rate of severe fatal and disabling birth defects among perinatal infants registered in Nanning showed significant downward trends, while the intervention rate for such defects markedly increased. The "321" hierarchical architecture model effectively reduces birth defect incidence and enables timely, effective intervention for severe fatal and disabling birth defects.

Keywords: "321" hierarchical structure; birth defects; prevention and control model; fatal and disabling birth rate; countermeasure research; Nanning City

Introduction

Birth defects refer to structural, functional, and metabolic abnormalities that occur during embryonic or fetal development due to internal and external factors. These defects arise from diverse causes, including multifactorial inheritance, single-gene disorders, chromosomal abnormalities, environmental teratogens, and micronutrient deficiencies. Birth defects often lead to adverse outcomes such as early miscarriage, fetal death, infant mortality, and congenital disabilities, imposing heavy burdens on families and society. Consequently, they represent a critical public health issue requiring heightened attention and

control. These defects severely impact population quality and serve as an important indicator for evaluating primary healthcare service quality in a country or region.

In recent years, China has achieved certain successes in reducing neonatal mortality rates and the incidence of major birth defects. However, challenges persist, including high numbers of birth defects, complex varieties, and extremely complicated etiologies. Previous research has primarily analyzed individual factors affecting birth defect occurrence, such as pregnancy methods, gravidity, fetal number, maternal nutritional status, medication use, and secondhand smoke exposure. Beyond individual factors, social factors—including socioeconomic status, meteorological conditions, environmental factors, healthcare service utilization, and preventive intervention policies—also correlate with birth defects, potentially leading to regional variations in incidence. Further research is needed to explore the role of these social factors in birth defect occurrence to develop better prevention and management strategies that reduce risks and safeguard population quality and health.

Currently, birth defect research data primarily derives from hospital-based surveillance systems. Due to variations in monitoring capabilities and screening strategies across regions, results differ. Hospital monitoring data enables region-specific risk factor identification studies, and combined with regional statistical yearbooks, facilitates macro-level understanding of regional epidemiological trends and etiological factors.

Based on this context, our study utilized surveillance data from Nanning's maternal and child health information system (2020–2022) and relevant Nanning statistical data to investigate birth defect occurrence and trends in Nanning. We explored potential social factors influencing birth defect rates and established a "Nanning Model" comprehensive birth defect prevention architecture, led by Nanning Maternal and Child Health Hospital and jointly implemented by multiple institutions across municipal, county (district), and township levels. This provides reference applications and preventive measures for reducing birth defect rates and disability rates while improving newborn survival and quality of life. The research findings are presented below.

The "321" Hierarchical Architecture Model for Comprehensive Birth Defect Prevention and Control

As the capital of Guangxi Zhuang Autonomous Region, Nanning represents a high-level, well-developed region in Guangxi for medical care, health prevention, and disease control. However, Guangxi remains a severely affected area for birth defects, with Nanning and other Guangxi cities consistently reporting high numbers of birth defect cases. There is an urgent need to establish a coordinated prevention, control, and timely treatment plan and model suitable for Guangxi and its majority of regions. Through investigation and analysis of regions, cities, and rural areas in other countries with high birth defect incidence rates world-

wide, combined with Nanning's current birth defect reporting situation and emergency response measures, and considering comprehensive geographic, environmental, and climatic factors, we propose the "Nanning 321 Hierarchical Architecture Model for Comprehensive Birth Defect Prevention and Control."

The "321" hierarchical structure establishes a three-tier comprehensive joint prevention and control model for birth defects based on municipal, county (district), and township levels, integrating factors such as grade, severity, and urgency. This architecture enables effective and timely feedback on birth defect data, facilitates prompt intervention, and helps accurately understand, monitor, and implement targeted prevention and control measures. This approach improves feedback reporting quality and reliability while enhancing patient satisfaction and core technical competitiveness. Therefore, during model development, we emphasized and adopted the "321" hierarchical architecture to effectively address shortcomings in comprehensive birth defect prevention and control systems.

This model centers on Nanning Maternal and Child Health Hospital, with its key established facilities serving as the core diagnostic and treatment network: the Municipal Birth Defect Prevention and Control Key Laboratory, Obstetrics Quality Control Center, Thalassemia Prenatal Diagnosis Sub-center, Neonatal Disease Screening Center, Neonatal Hearing Disorder Diagnosis and Treatment Center, and Congenital Heart Disease Screening Center. Seven municipal hospitals within Nanning's jurisdiction coordinate clinical treatment for birth defect-related diseases, fully leveraging integrated prenatal and postnatal birth defect comprehensive prevention and control systems and multidisciplinary collaborative treatment systems for critical, difficult, and severe cases. Eighteen county (district)-level healthcare institutions or maternal-child health medical consortiums under Nanning's jurisdiction serve as the initial screening network for birth defect-related diseases.

Since implementing and deeply promoting birth defect prevention and control programs—including premarital medical examinations, folic acid supplementation, prenatal screening, thalassemia prevention and control, and neonatal disease screening—Nanning has achieved obvious control over the birth rate of severe fatal and disabling birth defects. However, a small number of major lethal and disabling birth defect cases continue to be born, creating heavy psychological and economic burdens for families and society. Implementing a first-diagnosis responsibility system enables specialized case management for pregnancies identified as at risk for severe fatal and disabling birth defects. When necessary, multiple departments conduct follow-up tracking for cases not actively pursuing further medical intervention, providing face-to-face education on birth defect prevention and treatment knowledge for couples of childbearing age while offering green channels for further examination and treatment to save time and improve compliance rates among high-risk couples, thereby reducing births of infants with severe fatal and disabling birth defects and improving Nanning's birth population quality.

2.1 Data Sources

The study subjects comprised 272,218 perinatal infants born in all midwifery institutions in Nanning from January 1, 2020, to December 31, 2022, covering the period from 28 weeks of gestation to 7 days postpartum. Using Nanning's maternal and child health information system, we collected hospital monitoring information including perinatal infant numbers, basic conditions, defect status, and outcomes. Birth defect diagnoses and classifications strictly followed the *China Maternal and Child Health Surveillance Manual* and *Manual for Diagnosis of Major Congenital Malformations*. According to the *Guangxi Birth Defect Hospital Monitoring Program*, birth defect information reported through the hospital monitoring system was exported from the Guangxi Maternal and Child Information Management System for cases where the mother's household registration was in Nanning.

2.2 Survey Methods

Birth defect hospital monitoring targeted perinatal infants born in Nanning. In each monitoring hospital, clinical observation and physical examination, ultrasound diagnosis, chromosome detection technology, blood biochemistry, and immunological tests were combined with diagnosis, reporting, and quality control procedures from the *Guangxi Birth Defect Hospital Monitoring Program* for data submission. This study conducted relevant statistical analyses and dynamic comparative analyses of birth defect occurrence.

2.3 Quality Control

To ensure authentic and accurate monitoring data, participating institutions were required to conduct monthly self-inspections of birth defect monitoring information reported by all monitoring institutions within their jurisdiction. Other municipal medical institutions reviewed birth defect registration card completion monthly and performed quarterly checks for missed and erroneous reports. Nanning Maternal and Child Health Hospital conducted annual quality control of birth defect monitoring data to ensure accuracy.

2.4 Statistical Methods

Birth defect case lists for Nanning-registered households were exported from the Guangxi Maternal and Child Information Management System. An Excel database was established, and SPSS 26.0 software was used for relevant statistical analyses. Descriptive analysis was primarily employed, with chi-square tests and trend chi-square tests used for comparing count data. The significance level was set at α=0.05.

3.1 Birth Defect and Perinatal Birth Defect Occurrence

Table 1 Incidence Rate of Birth Defects Among Nanning-Registered Perinatal Infants (2020–2022) (per 10,000)

3.2 Distribution of Major Birth Defect Diseases in Nanning (2020–2022)

Statistical data indicate that neural tube defects (anencephaly, spina bifida, encephalocele), congenital hydrocephalus, cleft lip with cleft palate, cleft palate alone, cleft lip alone, microtia (including anotia), other external ear malformations (excluding microtia and anotia), rectal and anal atresia or stenosis, hypospadias, bladder exstrophy, limb reduction, congenital diaphragmatic hernia, omphalocele, gastroschisis, Down syndrome (trisomy 21), severe thalassemia, intermediate thalassemia, fetal hydrops syndrome, esophageal atresia or stenosis, talipes equinovarus, polydactyly, syndactyly, and hydrocele maintained fluctuating trends at certain levels. With changing years, congenital heart disease and chromosomal/gene abnormalities showed 逐年升高 [year-by-year increasing] trends. The top five birth defects were: congenital heart disease, polydactyly, hydrocele, other external ear malformations (excluding microtia and anotia), and talipes equinovarus. See Table 2 .

Table 2 Incidence Rate of Major Birth Defects Among Nanning Perinatal Infants (2020–2022) (per 10,000)

3.3 Occurrence of Severe Fatal and Disabling Birth Defects in Perinatal Infants

Trend chi-square tests revealed that the incidence rate of severe fatal and disabling birth defects showed an increasing trend with changing years (^{2}=7.124, P=0.008). Complex congenital heart disease, severe thalassemia, and Down syndrome (trisomy 21) constituted the main severe fatal and disabling birth defects. The birth rate of severe fatal and disabling birth defects showed a 逐年上升 [year-by-year increasing] trend (^{2}=8.668, P=0.003). From 2020–2022, the intervention rate for severe fatal and disabling birth defects before 28 weeks among Nanning-registered households exceeded 90%. See Table 3 and Table 4 .

Table 3 Incidence Rate of Severe Fatal and Disabling Birth Defects Among Nanning-Registered Perinatal Infants (2020–2022) (per 10,000)

Table 4 Intervention Rate for Severe Fatal and Disabling Birth Defects in Nanning (2020–2022) (%)

4 Discussion and Outlook

In recent years, Nanning has adopted the “321” hierarchical architecture model for comprehensive birth defect prevention and control, organizing efforts around government leadership, medical institutions, and community levels. This model

has yielded certain achievements in birth defect prevention. However, with continuous advancement in medical diagnostic technology and environmental deterioration, Nanning's birth defect incidence continues to increase, indicating that birth defect prevention remains an important and challenging public health task. Practical difficulties persist in policy implementation, resource allocation, target population participation, and multi-center collaborative governance.

Effective policy execution must be strengthened to ensure governments at all levels fully perform their functions and provide robust support for birth defect prevention. Rational resource allocation is crucial—Nanning should ensure adequate medical facilities and personnel to provide high-quality diagnosis and treatment services. Simultaneously, enhancing community-level participation to promote health awareness and behavioral change among residents warrants attention. Finally, Nanning should actively promote and improve multi-center collaborative governance mechanisms to enhance overall effectiveness in birth defect prevention.

Research results show that perinatal birth defect incidence is increasing annually. This trend can be attributed to three main reasons: First, continuous improvement in prenatal screening and diagnostic technologies enables more comprehensive detection of various birth defects, including rare conditions. Second, Nanning has actively implemented multiple phases of birth defect prevention measures and conducted training programs for birth defect monitoring personnel, deepening their understanding of reporting standards. Third, according to the 2022 edition of the *National Birth Defect Monitoring Guidelines*, more detailed requirements and explanations for reporting congenital heart disease and minor malformations have driven increases in reported perinatal birth defect numbers. Additionally, implementation of fertility policies has increased the proportion of advanced maternal age, raising risks for chromosomal abnormalities and consequently increasing reported defect cases among infants born to older mothers. In summary, these factors collectively explain the 逐年上升 [year-by-year increasing] trend in perinatal birth defect incidence.

Congenital heart disease is a common defect present at birth, with an increasing incidence trend attributable to Guangxi's implementation of free newborn congenital heart disease screening in 2020. Since then, screening rates have increased annually from 83.89% in 2020 to 99.19% in 2022, with corresponding increases in positive cardiac ultrasound examinations. These factors have collectively increased confirmed congenital heart disease cases. Meanwhile, widespread application of newborn disease screening technology has led some institutions to incorporate B-ultrasound examinations for congenital heart disease into routine screening, further increasing reported cases. Additionally, extensive application of non-invasive prenatal testing (NIPT) and CNV-Seq technologies has gradually increased reported numbers of chromosomal/gene abnormalities and rare birth defects, with perinatal chromosomal abnormality incidence rising annually. Consequently, incidence rates for congenital heart disease and chromosomal/gene abnormalities continue to increase, reflecting improved early

diagnosis rates, though resulting in increased reported numbers.

Recent statistics show the top five birth defects in the past three years include polydactyly, hydrocele, other external ear malformations (excluding microtia and anotia), and talipes equinovarus. Notably, severe thalassemia and Down syndrome rank beyond tenth place in birth defect prevalence. Fortunately, incidence rates for severe thalassemia and Down syndrome have decreased, primarily due to free thalassemia prevention programs and prenatal screening subsidies that enable earlier detection and intervention.

The intervention rate for severe fatal and disabling birth defects before 28 weeks exceeds 90%, yet birth rates continue to rise annually. Approximately 15% of birth defects citywide in recent years have been severe fatal and disabling defects. While intervention rates before 28 weeks surpass 90%—demonstrating certain achievements in preventing severe birth defects—the live birth rate for severe defects continues increasing. Three main factors explain this: First, complex congenital heart disease has the highest birth rate, and despite effective treatment options for some severe defects, some pregnant women choose to continue pregnancy (e.g., tetralogy of Fallot). Second, some pregnant women maintain a 侥幸心理 [wishful thinking mentality], refusing timely induced labor despite medical advice and repeated counseling, insisting on giving birth to affected infants. Third, an incomplete referral and tracking mechanism within the jurisdiction has resulted in inadequate management and tracking interventions for confirmed severe birth defect cases in some healthcare institutions, allowing these infants to be born. Strengthening relevant policy development and implementation is necessary to reduce severe birth defect incidence.

In summary, Nanning has achieved certain effectiveness in birth defect prevention and control but faces challenges from rising incidence rates and implementation difficulties. Only through strengthened policy implementation, rational resource allocation, enhanced target population participation, and improved multi-center collaborative governance can Nanning effectively address birth defect challenges and safeguard public health.

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Note: Figure translations are in progress. See original paper for figures.