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## Surveillance of birth defects: Brazil and the US

Vigilância epidemiológica em anomalias congênicas:  
Brasil e Estados Unidos

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**Abstract** *The impacts of birth defects in the society have substantially increased over the last decades in countries where the reduction of infant mortality by other causes has occurred. Birth defects surveillance represents an important source of information for planning X health policies and resource allocation. In this article, we discuss the potential utilizations, methodology options, limitations, and policy issues related to birth defects surveillance. Also, the birth defects surveillance programs from US and Brazil are described and compared as an illustration of the development of surveillance systems in two countries with clearly dissimilar health systems and resource allocation for birth defects programs. Finally, we propose measures for the improvement of the existing systems in both countries focusing at the utilization of preexisting resources*

**Key words** *Surveillance, Birth defects, Health information systems*

**Resumo** *O impacto das anomalias congênicas na sociedade vem aumentando substancialmente nas últimas décadas nos países onde ocorreu a diminuição da mortalidade infantil por outras causas. A vigilância epidemiológica em anomalias congênicas representa uma importante fonte de informação para o planejamento de políticas de saúde e alocação de recursos. Neste artigo, os potenciais usos, opções de metodologia, limitações e questões de políticas de saúde relacionadas à vigilância epidemiológica em anomalias congênicas são abordados. Além disso, os programas dos Estados Unidos e Brasil são descritos e comparados para ilustrar sistemas em dois países com sistemas de saúde e alocação de recursos para as anomalias congênicas claramente diferentes. Finalmente, apresentamos propostas de medidas para melhorar os sistemas existentes em ambos os países, focalizando na utilização de recursos pré-existent*

**Palavras-chave** *Anomalias congênicas, Vigilância epidemiológica, Sistemas de informação em saúde*

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## Introduction

Although individually rare, birth defects taken together account for a significant proportion of mortality among infants and children, particularly in areas where infant mortality due to more common causes has been reduced. Major birth defects are diagnosed in three to four percent of infants in their first year of life<sup>1</sup>.

The impact of birth defects on the future child, on the child's family and on the community is not restricted to mortality; it also involves the morbidity and disability experienced by those who survive. Birth defects are responsible for a high proportion on years of potential life lost, infant hospital admissions and medical costs<sup>2</sup>.

Birth defects surveillance and prevention programs is among the three programs most commonly identified as a contribution of genetics to public health besides newborn screening programs, and clinical genetics programs<sup>3</sup>.

The strategy proposed by the World Health Organization (WHO) for significantly reducing the public health impact of birth defects has three stages. The first involves interventions to prevent birth defects, what is possible in approximately 50% of them. The second stage involves improving the care locally available for affected infants. The third involves genetic screening and treatment of infants with treatable genetic diseases. At all three stages, the process of reducing birth defects impact involves surveillance of birth defects<sup>4</sup>.

Each surveillance system is created within a unique political, social, geographic, economic and historical context. It also reflects the particular interests, training and philosophy of the creators of each system. There is no single ideal model universally applicable, however the diversity of the structure of the programs results in a variability of the surveillance systems and, inevitably in the characteristics of the obtained data<sup>1</sup>.

The main issues in the development of birth defects surveillance policies regard usefulness of data and resource allocation. While virtually everyone agrees that there is societal demonstrated benefit, through its foundation for many public health activities and epidemiological studies, to what extent funding should be provided remains a central question for policy makers. The frail balance between data quality and expenses on financial and human resources has to be constantly evaluated to assure as much as possible the ultimate goal of surveillance that is to take the further step from "counting" to "action".

In this article, we discuss methodological aspects and applications of birth defects surveil-

lance. The US and Brazil programs are described to illustrate birth defects surveillance performed at two different health systems scenarios. Finally, we propose two measures that could improve those programs through rational utilization of already existing resources.

## Potential applications of birth defects surveillance data

In the late 1960s, after the thalidomide episode, the first registries for birth defects were created. Their primary reason was surveillance to avoid a repetition of a similar tragedy. One of the purposes for surveillance still is to detect changes in the prevalence of a specific defect or pattern of defects that might indicate the presence of a new causative factor, and to identify such hazard. However, the information has also been increasingly used for various other reasons such as epidemiologic studies concerned with the etiology of birth defects, studies of the societal impact of birth defects, census of disabled people for the planning on social welfare and medical services, assisting the development of clinical genetic services for the care and prevention of birth defects, and evaluation of effectiveness of preventive measures to tailor them for maximum effectiveness<sup>5</sup>.

The detection of the almost worldwide increasing prevalence of gastroschisis, a congenital condition characterized by an abdominal wall defect consisting in visceral herniation, is a good example of the utilization of birth defects surveillance to detect changes in prevalence. Epidemiologic findings such as the increased risk associated with very young maternal age are unique to gastroschisis and, although not yet understood, it is thought to indicate environmental exposures common among this age group.

Primary prevention of birth defects concerns the prevention of the origin of such disorders. By definition, this is only possible if the causative factor that induces the defect is known and the exposure prevented before conception and/or during gestation<sup>1</sup>. However, nearly 50% of birth defects have an unknown etiology. As birth defects are usually rare diseases etiological hypotheses are usually based on a small number of cases. Data from birth defects surveillance systems has also been used to test these hypotheses, the advantage, apart from the larger number of cases, is the possibility of collected data on exposure. Many different risk factors, varying from pregnancy-related factors to parental occupation and drugs, have been studied with the use of birth defects registries<sup>6</sup>.

The identification of disabled children in need of special education, social services, and other programs is another important contribution of birth defects surveillance. One of the most important goals of early intervention is to prevent secondary disabilities that result from an infant's primary condition, a goal that is more likely to be achieved the earlier an infant and family are identified. The use of birth defects registries data can be a very efficient and cost-effective means for identifying children in need of special care and providing them timely referral to specialized services<sup>7</sup>.

Besides early referral, the estimation of health resource needs and priorities for interventions based on birth defects prevalence is an additional significant direct social benefit of birth defects surveillance. Accurately predicting the demand for various interdisciplinary clinics, social, and educational services is critical for children born with birth defects. Estimating future service needs can also direct capacity building to ensure that necessary human resources will be accessible and that appropriate professionals will be available to provide the services.

Data from birth defects surveillance programs can also be used to determine whether reported cases of birth defects represent existing cases in other databases, such as records in interdisciplinary clinics and schools with programs to assist children with disabilities. The ability to link records on individuals in more than one database can streamline the treatment and referral processes and help maintain a certain level of fidelity and trust in prevalence data. Another utility of record linkage is the ability to supply crucial data required for various research efforts. The potential to link records and consolidate information from different databases contributes to the public health applications of birth defects surveillance data<sup>8</sup>.

The use of folates, through folic acid food fortification or supplementation, is an example of a potential use of birth defects surveillance data: evaluation of effectiveness of preventive measures. In 1980, the results of a nonrandomized trial revealed that taking multivitamins during the periconceptional period reduced the risk of having a fetus or infant with a neural-tube defect<sup>9</sup>. Since then, observational studies demonstrated a reduced risk among women who took multivitamin supplements containing folic acid and those who had higher dietary intakes of folate during early pregnancy<sup>10-12</sup>. Most studies to demonstrate the impact of health policies on folic acid, such as better source, dosage, target public, prevention of other birth defects and side effects were only

possible to be performed because of ongoing birth defects surveillance systems allowing for analysis of secular trends before and after folate interventions.

### **Birth defects surveillance**

Public health surveillance is defined by the Center for Disease Control and Prevention as "the ongoing systematic collection, analysis, and interpretation of outcome-specific data for use in the planning, implementation, and evaluation of public health practice"<sup>13</sup>. A surveillance system includes not only data collection and analysis, but also the application of these data to control and prevention activities by disseminating information to practitioners of public health and to others who need to know for planning, implementing and evaluation of interventions and programs<sup>14</sup>.

The term "birth defect" encompasses, in the broadest sense, a diversity of conditions including physical malformations, sensory deficits, chromosomal abnormalities, metabolic defects, neurodevelopmental disorders, and complications related to prematurity and low birth weight, among others<sup>5</sup>. However, traditionally, birth defects surveillance programs have monitored major structural and genetic defects that adversely affect health and development.

### **Methodology options for birth defects surveillance**

"Birth defects surveillance and prevention programs protect against environmental hazards by diagnosing and investigating health problems and health hazards in the community"<sup>3</sup>. Notwithstanding, the value of birth defects surveillance lies in how the data are collected and how they are used towards reducing the birth defects impact on society. All programs should establish goals and objectives making it clear that the ultimate rationale for conducting public health surveillance is to have data that can be used to improve the health of the public.

On planning a birth defects surveillance program, many distinct aspects have to be considered about the methodology to be employed based in the objectives and primary goals of the program and available resources (Panel 1). Firstly, there is the option of a population-based registry which tries to identify all births of malformed infants within the population of a given geographic area. Secondly, there is the hospital-

**Panel 1.** Methodological approaches for birth defects surveillance.

Elements	Options	
Registry	Population-based	Hospital-based
Surveillance	Active	Passive
Case Ascertainment	Multi-source	Single source
Case Definition	All BD	Selected BD
Description	Verbatim	Checkbox
Period (age)	Up to adulthood	Newborn
Pregnancy outcomes	All (LB, SB, Abortion)	Live birth
Coding system	Own	ICD, BPA
Coding process	Central	Local

BD: Birth Defects; ICD: International Classification for Diseases; BPA: British Pediatric Association; LB: Live birth; SB: Stillbirth

based registry where the registry receives notification from a certain number of hospitals. The ideal birth defects system is one in which a population-based information is reported in a timely manner. However, in many areas of the world a hospital-based system is all that is feasible because of the high costs usually involved in a population-based system.

Another methodology issue to be decided is whether to perform passive or active surveillance. The former involves the identification of cases from vital records or from reports submitted by health care facilities. Active surveillance involves the identification of cases by trained surveillance program staff who actively seek cases in the facilities periodically. Case ascertainment tends to be more complete than passive surveillance yet it is an expensive modality.

There are wide variety of data sources including vital records (birth and death certificate), hospital discharge summaries (newborn or other), hospital records, specific birth defects forms, and data from laboratories. Multiple source case ascertainment provides the best potential for case finding and precise and accurate diagnosis. However it usually involves higher costs, often limiting it to small populations, and increases the time needed to establish baseline data. In the era of electronic forms, linkage of health information systems is highly feasible and, at least in part, could lower the costs of multiple sources helping to populate the database.

Data collection using a predetermined list (for example, in a checkbox format) limits ascertain-

ment to some birth defects, leaving out unusual birth defects or new patterns, therefore potentially masquerading new epidemics. Therefore, the verbatim description of the birth defect is preferred, following a manual of procedures to ensure detailed descriptions and diagnosis confirmation in a central level.

Case definition in a birth defect surveillance system mainly involves the decision as whether to include only specific birth defects, all major and/or minor defects, and maximum age for inclusion on the database. Whether an individual program is able to ascertain defects beyond the newborn period will depend on the accessibility of information from sources other than the newborn nursery and the availability of staff and resources to add these additional sources. Information on other pregnancy outcomes (stillbirths, spontaneous abortions, and termination of pregnancy) should ideally be present, especially with the improvement of prenatal diagnosis leading to termination of pregnancy in countries where this is legal.

The coding process is an important component of the storage of surveillance information as it has to be easily retrieved to be useful. Some programs opt for the International Classification of Diseases and Related Health Problems (ICD), while others, for further specificity, opt for the ICD with the expanded fifth digit from the British Pediatric Association (BPA); and some choose to create their own code. When choosing for own code, this should have a compatible code with the ICD so that comparisons with other systems can be easily done. While local coding could speed the process, central coding is usually preferable as a guarantee of uniformity of data.

### Limitations of birth defects surveillance

Although one of the purposes of birth defects surveillance is the detection of new health hazards, it is apparent that environmental agents causing dramatic increases in specific types of adverse reproductive outcomes are the exception rather than the rule, and that no new teratogenic agent has been directly identified as a result of even the best surveillance systems created in the wake of the thalidomide episode in the early 1960s. There seems to be needed extreme, and unlikely conditions: either very high exposure rates (an agent that suddenly become very common) or very strong teratogenic properties to detect an increase in the rate of one of the common de-

fects<sup>15</sup>. Otherwise, the appearance of a previously very rare type of defect or a pattern of defects may be detected. For this reason some authors advocate that the main purpose should be register and analyze unusual events: rare defects or combinations of defects<sup>16</sup>.

Interpretation of long-term changes in birth defects rates is likewise rather difficult. The possibility of changed ascertainment by the introduction of new diagnostic tools, improvement of the old ones or new interventions, such as termination of pregnancy have to be first excluded, as well as change in population characteristics, such as maternal age distribution and changing trends with modifications in cultural, social, and environmental factors.

Effective coding and ascertainment are essential components of birth defects surveillance. Miscoding and underreporting can limit the usefulness of birth defects surveillance data by masking the degree to which birth defects are affecting a population, therefore leading to erroneous conclusions about the effectiveness of programs and influencing health policies as well as allocation of resources. Cuniff *et al.*<sup>17</sup> and Reefhuis *et al.*<sup>18</sup> present alarm situations associated with interpretation of birth defects surveillance data resulting from inappropriate coding.

Depending on the methods and sources of case ascertainment used, the various surveillance systems produce substantially varying birth defects rates making data comparison difficult. The use of systems that can collaborate will substantially increase the usefulness of the data nationally and internationally. Efforts towards collaboration revolve around the development of uniform methods and standards and the use of equivalent case definitions and coding systems.

### Policy issues

An integrated public health surveillance system is the result of policy-level agreements that exist between those who provide data and those who use the information. Public health officials, policy makers, researchers, and others who use the data should describe their needs for this information and should demonstrate that these needs justify the burden imposed on those who provide the data<sup>14</sup>.

The critical challenge in birth defects surveillance is the assurance of its usefulness. However, once usefulness is demonstrated, policies on allocation of resources are the main issues to as-

sure that surveillance is translated into public health action. Policies have to be developed according to the available resources for health care of a determined population.

Methodology strategies have to be weighted against available funding. Nowadays, with even more limited funds for health care, policy makers face difficult choices when deciding on funds allocation whether in developed or developing countries. Budget allocation involves government agencies, legislative bodies, special interest groups and professional organizations.

### Proposal of strategies for improvement of birth defects surveillance

The US is an example of a developed country where birth defects are recognized as conditions of high public impact and surveillance was established, in some states, more than 40 years ago. Brazil is a developing country where birth defects surveillance was implemented recently, during the epidemiologic transition, when birth defect morbidity and mortality pattern, previously obscured by the predominance of infectious disease, assumed great relevance (Panel 2). These two countries, with considerably different settings of health care systems and of resources availability, will be used as examples to illustrate two procedures that could improve birth defects surveillance quality data, and amplify the usefulness of data, provided that they were truly implemented.

### Birth defects surveillance in the United States

Birth defects currently represent the leading cause of infant mortality in the United States and have been identified as the primary contributor to infant mortality for at least 20 years<sup>19</sup>. Currently, 38 states report birth defects surveillance data to the National Birth Defects Prevention Network (NBDPN) and the CDC periodically reports national prevalence estimates for selected birth defects derived from the average prevalence of the birth defects in 15 states with active surveillance systems. Nonetheless, nationally representative estimates of birth defects prevalence are largely unavailable due to variations in state surveillance methodologies and the lack of a standardized national surveillance system<sup>20</sup>.

In the absence of a single national birth defects surveillance program in the United States,



**Panel 2.** Comparison between the US and Brazil birth defects population-based surveillance systems.

	US	Brazil
Population-based	52 programs	National Program (Birth Certificate)
National Coverage	38/50 states	Nation-wide
Active Surveillance	15 states	None*
Year of Implementation	1967 (1 <sup>st</sup> )	2000
Standardized National System	No	Yes
Data on Maternal Risk Factors	Yes	No
Birth Defects Research Centers	8 centers (CBDRP-CDC)	ECLAMC

\* Except the 32 hospitals (2% of Brazilian livebirths) participating in the Latin-American Collaborative Study of Congenital Malformations (ECLAMC).

pooled data from state-based programs across the country serve to estimate national rates, indicate regional variations, and describe the epidemiology of defects that occur rarely.

Currently, ECLAMC is collaborating with the Brazilian Ministry of Health in a project for the improvement of the use of the birth certificate for the birth defects surveillance<sup>23</sup>.

### Birth defects surveillance in Brazil

In Brazil, birth defects were the second leading cause of infant mortality (16.3%) in the first year of life in 2005, making evident a clear rise in the proportion of the mortality in the last 20 years: 7.1% in 1985-1987 and 11.2% in 1995-1997 triennial (www.datasus.gov.br). In the 2000 Birth Certificate Revision a field for the register of birth defects was introduced, in this way the basis for the permanent observation of the prevalence at birth of birth defects was established constituting a potential nationwide population based surveillance system of approximately 3 million live births per year<sup>21</sup>. Nonetheless, the data is not being currently used.

Other system for birth defects surveillance in Brazil, and from where data about birth defects has been extrapolated, is the Latin-American Collaborative Study of Congenital Malformations (ECLAMC), a program for the clinical and epidemiological investigation of risk factors in the etiology of birth defects using a hospital-based registry operating since 1967, in Latin America with a current net of 130 hospitals, 32 of these in Brazil. This system, even if not designed for this purpose, in the absence of other specific ones, fulfills the role of only source of information for the surveillance of this morbidity. Nevertheless, it is important to point out the reduced coverage of this study, only 2% of all Brazilian births, and also that the generated information is hospital-based and not population-based<sup>22</sup>.

### Strategy 1: the birth certificate

Birth certificates (BC) are an attractive source of information about birth defects because they are universal, standardized and inexpensive<sup>24</sup>. Birth certificates have been evaluated in the US as a potential source for the prevalence of birth defects, however have been found to substantially underestimate such prevalence varying from 55% to 88%<sup>24-29</sup>. In Brazil no extensive studies have been published yet. In a study in Rio de Janeiro city using the BC database, a birth defects prevalence of 83 per 10,000 live births was reported<sup>30</sup>. This result also suggests an underreporting when compared to birth defects prevalence at birth (170 per 10,000 live births) as reported in a previous cross-sectional study in the same city<sup>31</sup>. Besides, the overall reported prevalence suggests an underreporting of approximately 60%<sup>21</sup> if we consider mean estimates of birth defects prevalence worldwide.

The strengths of birth certificates include the complete coverage of the population, the availability of some medical and parental data, ready availability of data from previous years, low cost and the potential for follow-up of birth defect cases. The weakness include the lack of timeliness in reporting, the underreporting of birth defects (limited to data obtained during newborn period, thus less severe birth defects have a higher chance of being overlooked than more severe defects), and lack of specificity in the description of most birth defects. On one hand the underreport-

ing in prevalence can hide the degree to which the birth defects are affecting a population and lead to erroneous conclusions about the effectiveness of programs influencing health policies as well as allocation of resources. However, considering the resource limitations for surveillance activities, systems implemented that presents limitations should not be abandoned but improved. In a practical perspective, BC can provide, at least, low-end estimates of the prevalence of birth defects<sup>24</sup>.

The improvement of birth certificate as a data source could improve birth defects surveillance in both countries though in different perspectives for each one. In Brazil, the use of the birth certificate, which is integrated into a national pre-existing health program, would mean rational utilization of resources and surveillance at a high decision-making level with low operational costs and potential for high sensibility and specificity and wide and complete coverage. In the US, the improvement of the birth certificate could lead to the data collection standardization (at least for birth defects diagnosed at birth), reduction of the costs of overlapping systems for surveillance, and improvement on case ascertainment, especially for the passive systems.

The interventions needed to improve birth defects surveillance and its appropriate implementation through birth certificate involves multiple stakeholders. Advisory committees should have representation of health care services and government departments involved in maternal and child health, public health education, health legislators, disability and rehabilitation programs, experts in birth defects surveillance and patient/parent support organizations.

### **Strategy 2: routine health information systems database linkage**

The potential now exists for extensive computerized record linkage in birth defects surveillance programs allowing for the tracking of children with a health-related condition from the point of identification through access to services. Many computer-based systems already exist for documenting health care delivery, including diagnostic and procedure codes in the US and Brazil. Birth defects surveillance records can be linked to many other public health program databases. These include, for example, newborn screening to conduct epidemiologic studies, special education data to predict the need for services for children with mental retardation, and early inter-

vention program data to assess the overlap and utility of a birth defects surveillance program as a “child find” resource<sup>5</sup>.

However, many birth defects surveillance programs in the US – based both in health departments and in other institutions such as universities – have encountered increasing concerns and pressures as a result of Health Insurance Portability and Accountability Act (HIPAA) regulations and issues surrounding their interpretation and implementation. Program regulations frequently impede attempts to link records between case-finding databases and service-delivery databases. As a result, attempts to meet the very reasonable public health goal of ensuring access to services by those in need may be thwarted<sup>14</sup>.

In Brazil, in the other hand, privacy issues are not the ones who are preventing a better use of the information from the existing national health systems such as vital records, hospital discharge summary, morbidity, and health care delivery. In fact the unidentified information is available online for anyone who wishes to access it. However, linkage of data from the various existing systems is not routinely done at the Ministry level, where the data is compiled and analyzed. Some municipalities, though, are using the information from the birth certificate to indicate to their primary health care units, newborns that are at higher risks such as low birth weight, low maternal age or birth defects, assuring that they can receive special care and showing that this is a feasible action.

### **Concluding remarks**

A desired birth defect surveillance system would be an integrated population-based system with standardized data collection and qualified information. In addition, the information produced should have an efficient flow to appropriate users by avoiding duplication, minimizing burden, protecting confidentiality, and maximizing analytic study. Usually it is thought that this can be better achieved with an active and multi-source surveillance system. However, health care systems and services vary widely both among and within countries. Effective strategies to address birth defects must take into account the competing needs for resources and social, economic, and other factors that constraint health. Thus, to be effective, strategies and interventions need to be tailored to the specific population served.

In the US, efforts should be made towards standardization of data and use of the data gen-



erated specially trying to surpass privacy issues that are currently undermining the use of the existing data so that resources are used to its maximum effectiveness. This issue should be specially discussed in panels with patient/parent associations so that the balance between privacy and benefit can be reached and policies implemented.

In Brazil, surveillance may be strengthened with concentration of efforts on the improvement of birth certificate trying to allocate resources for capacity building in collection, coding and analysis of data. National guidelines as the one provided by the CDC in the US<sup>5</sup> are also of extreme importance in this matter.

In both countries, members of the public, including parents of children identified through these programs, are often not well informed about birth defects surveillance activities. Policy making process should be broadening to engage consumer and parent participation.

Surveillance information about the public health impact of birth defects must be effectively conveyed to legislators and makers of public health policy. In Brazil, there is almost no information available on the financial implications of birth defects in general or of individual birth defects it is often difficult to persuade international agencies and government of the importance of developing national programs for the care and prevention of birth defects. Close interaction with health economists towards developing better knowledge of the economics aspects of birth defects is therefore imperative.

Successful public health policy based on epidemiological data depends on the quality and strength of the evidence, interactions with multiple stakeholders, and the clarity with which this information is communicated to the public. These should be continuously sought by birth defects surveillance systems.

### Collaborators

DV Luquetti conducted the literature search and wrote the paper. RJ Koifman commented on the draft of the paper and edited sections of the paper.

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