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Methodological issues in setting up a surveillance system for birth defects in India

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ABSTRACT

India is undergoing an epidemiological transition—communicable diseases are on the decline due to better living conditions and healthcare delivery. On the other hand, the relative increase in the prevalence of non-communicable, chronic and genetic diseases threatens to be a public health problem in India. One such group of disorders is congenital malformations. Though several studies have been done on congenital malformations in India since the early 1960s, coinciding with the thalidomide tragedy in the West, no uniform methods are available for the surveillance of birth defects. Each study has come out with varying results, not only because of the geographic variation in birth defects but also due to the varying standards adopted by each study in data collection, case definition and other methodological issues.

Setting up a mechanism to understand the extent and nature of birth defects would involve the creation of a birth defects registry. The goals and objectives of such a registry should be formulated before it is set up. There are three types of registries—descriptive, analytical and preventive. These can also be classified as population- or hospital-based. Whether a registry is population- or hospital-based depends largely on the movement of mothers for delivery, registration of vital events in an area defined by the programme, as well as the resources available to the registry. Data can be collected in a passive or active manner, which also depends on the resources available to the registry. Every registry should have its own working definition of eligible cases to be reported, depending on the diagnostic services available in that area, and multiple sources of information should be used to improve the ascertainment rate. All the diagnostic terms should be coded and the information collected should be stored in a well-constructed database, preferably a relational type. Registries must evaluate their methods of data collection periodically to estimate the number of false-positive and false-negative reports. Ethical issues, cost and funding for the employment of various specialized professionals should be considered before setting up a registry.

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INTRODUCTION

The true magnitude of birth defects in India is not known, though research on congenital malformations started in India as early as 1963.[1] Congenital malformations remain one of the least focused areas of disease surveillance in India[2] compared with communicable and some chronic diseases. Unlike the situation in developed countries, where congenital malformations are the leading cause of infant mortality,[3–6] in India, low birth weight, prematurity, sepsis and infections are still the leading causes of neonatal and infant mortality.[7,8] Perhaps for this reason not much attention has been paid to the problem of congenital malformations in India. However, hospital-based studies published in the recent past have shown that birth defects are emerging as important causes of perinatal and neonatal mortality,[9,10] suggesting that India may be undergoing an epidemiological transition.[8,11] This could be because other causes of perinatal mortality have been controlled by improvement in obstetric and neonatal care.[9] All the hospital-based studies done so far in India have reported a high prevalence of congenital malformations at birth.[12–17] Although these figures cannot be extrapolated to the entire nation, the consistency of the existing reports suggests that the magnitude of this problem could be high.

Based on our experience of birth defects research and setting up surveillance systems for birth defects, we discuss the various methods available for setting up a registry for birth defects in India.

BIRTH DEFECTS REGISTRY: THE NEED OF THE HOUR

Statistics from developed countries suggest that 2%–3% of births are associated with major congenital anomalies.[6] In India, 25 million births occur every year.[18] If we assume that the birth prevalence of congenital malformations in India is 2% (it is likely to be more), then in absolute numbers it could mean that every year 500 000 babies are born with some form of birth defect; this is equal to the number of new cases of leprosy diagnosed in India in a year.[19] Fortunately, many of the major anomalies such as neural tube defects, conotruncal anomalies of the heart, anterior abdominal wall defects and oral clefts are preventable by periconceptional supplementation of folic acid alone.[20] Thus, data on the magnitude of birth defects are essential to plan preventive strategies and organize methods of supportive care for affected individuals and families.[21] This can be achieved only through an organized system such as a registry for birth defects.[21]

DEFINITION OF SURVEILLANCE

The epidemiological process of registering diseases or health conditions is called surveillance. The two most commonly used

definitions of surveillance are:

1. The ongoing systematic collection, analysis and interpretation of health data, essential to the planning, implementation and evaluation of public health practice, closely integrated with the timely dissemination of these data to those who need to know.[22]
2. Continuous analysis, interpretation and feedback of systematically collected data, generally using methods distinguished by their practicality, uniformity and rapidity rather than by accuracy or completeness.[22]

OBJECTIVES OF SURVEILLANCE FOR BIRTH DEFECTS

Before setting up a surveillance system for birth defects, the mission and objectives of the surveillance have to be defined. Defining the objectives will determine the type of surveillance to be set up. Usually, registries for birth defects are set up to ascertain the magnitude of birth defects in a defined population, monitor for secular trends in the prevalence of birth defects, and conduct aetiological research to determine risk factors for birth defects.

TYPES OF BIRTH DEFECT REGISTRIES

These are broadly classified into (i) descriptive, (ii) analytical and (iii) preventive, based on the purpose they serve.[23] A descriptive registry is primarily involved in describing the magnitude of congenital malformations in a given population in terms of prevalence. It also monitors for secular trends and occurrence of clusters of cases.[23] An analytical registry engages in aetiological research;[23] e.g. whether antiepileptic drugs increase the risk of congenital malformations. A preventive registry involves interventions[23] such as periconceptional folic acid supplementation for the reduction of neural tube defects. However, a descriptive programme is the basic aspect of research on birth defects.

Birth defect registries are also classified as population- and hospital-based.[24] In a population-based registry, cases are recorded on the basis of the residence of the mother. On the other hand, a hospital-based registry collects data from the place of birth (i.e. hospital).[24] In both types, the geographical area should be defined, though this is more appropriate in a population-based than a hospital-based registry. Selection bias is bound to occur in a hospital-based registry as a mother may be referred for delivery to a hospital outside the surveillance area due to certain risk factors, or a mother from outside the area may be referred for delivery to a hospital inside the surveillance area defined by the registry.[24] Hence, in a hospital-based registry, the prevalence is under- or overestimated depending on the location at which a mother delivers. However, no selection bias will exist in a hospital-based registry if it covers almost all the hospitals in a defined geographical area, provided the immigration and emigration of mothers is minimal.[24] A population-based registry must ensure that births in all the households in the registry area are traced, even if they take place outside the registry area. Therefore, data collection is difficult in a population-based registry compared with a hospital-based registry. Moreover, as only 50% of births are registered in India,[18] a hospital-based registry is ideal. However, one has to consider home or hospital delivery rates in an area before establishing a hospital-based birth defect surveillance system in that area; e.g. in Chennai, it has been estimated that 99.2% of deliveries take place in hospitals.[25]

METHOD OF DATA ASCERTAINMENT

Once the programme is defined, the next step is to decide the method of data collection. Surveillance data can be obtained by using either an active or passive method.[26] In an active surveillance method, which is more effective, the registry sends trained data abstractors to retrieve data from various sources such as hospitals and prenatal diagnosis centres where access to medical records is possible.[27] For example, the National Cancer Registry Programme in India sends data abstractors to hospitals and pathology laboratories to actively collect data on cancer cases.[28] In passive surveillance, instead of the registry staff actively collecting data, hospitals and other related sources send data to the registry. Passive surveillance is further classified into mandatory and voluntary. In the USA, it is mandatory to report the occurrence of birth defects to the respective state registry as per the Birth Defects Prevention Act.[23] However, the method of ascertainment depends on many factors—the types of medical services available in each area and their utilization, availability of diagnostic information to the registry, and manpower and financial resources available to the registry.[29]

SOURCES OF DATA ASCERTAINMENT

To accurately assess the magnitude of birth defects, a registry should use multiple sources of ascertainment,[29] such as birth and death certificates, maternity and hospital records including prenatal diagnosis, pathology services, maternal and child health services, etc. A surveillance programme which uses a single source of ascertainment, such as maternity records, would certainly underestimate the prevalence, because some congenital anomalies are not diagnosed at birth, particularly certain cardiac anomalies.[29]

DATA TO BE COLLECTED

The type of registry will determine the data to be collected. A descriptive registry's primary concern is the prevalence; hence, it collects the total number of births (denominator), and the number of malformed babies and type of malformation (numerator) in a given geographical area or a hospital. An analytical programme, on the other hand, has to collect obstetric, medical, family and conceptional details of both the numerator and the denominator to determine the risk factors.

VALIDITY OF CASE ASCERTAINMENT

Every birth defect surveillance programme must have an accurate prevalence estimate.[30] All surveillance programmes are vulnerable to false-positive as well as false-negative case reports,[30] especially registries with passive case ascertainment. Hence, it is mandatory for every surveillance programme to periodically evaluate the methodology of data collection, because no surveillance programme can claim to have achieved a 100% ascertainment rate.[29] The sensitivity and specificity of case ascertainment in registries with passive case ascertainment is evaluated by matching the reported cases with actively ascertained cases from a sample of randomly selected case sources.

CASE DEFINITION

A crucial step in surveillance for birth defects, is the definition of the case. All birth abnormalities should not be categorized as birth defects but, at the same time, the definition should not be very narrow. Because 'birth defect' is a broad term, encompassing both functional and structural deformities,[8] every registry should form its own working definition according to the resources available to it. Usually, the working definition used by registries is any structural or chromosomal malformation found in a live-born baby before its first birthday or in a stillborn baby or foetus that has been

medically terminated following the detection of an anomaly in the antenatal period.[27,31] Approximately 95% of birth defects are diagnosed before a child's first birthday.[27] Eligible conditions generally fall within the International classification of diseases (ICD–10) codes Q0–Q99.[32]

Exclusion criteria

Poor pregnancy outcomes such as prematurity, growth abnormalities (e.g. intrauterine growth retardation), placental and liquor abnormalities are usually not registered as birth defects.[27] Likewise, functional problems such as mental retardation or cardiac murmurs without obvious structural abnormalities are also not notified as birth defects.[27] Spontaneous/missed abortions, medical termination carried out for reasons other than malformations,[29] and molar and ectopic pregnancies are also excluded.

DEFINITION OF BIRTH CATEGORIES

Registries worldwide use several definitions for categories of birth. Some define spontaneous abortion as any foetal death before 20 weeks of gestation while others use the criterion of birth weight <500 g.[29] This information is crucial in comparing data from other registries. A definition based on gestational age rather than birth weight is better as many malformed babies would have lower birth weights than expected for that gestational age.[29] However, it may be difficult to assess the gestational age in rural pregnant women. This problem may be circumvented in hospital-based registries as women who go for an antenatal check-up might undergo an ultrasound assessment of the gestational age.

A live-born baby is defined as the delivery of a live infant who demonstrated signs of life such as respiration and heart beat after delivery.[33] Both intrauterine foetal death (IUFD) and stillbirth are grouped under one category—any foetal death on or after 24 weeks of gestation. Medical termination of pregnancy (MTP) (anomaly category) is defined as the induction of delivery with the intent of producing a non-viable infant because a prenatal screening test showed an abnormality in the foetus,[33] irrespective of the length of gestation. For example, if anencephaly was diagnosed in a 12-week foetus and the pregnancy was subsequently terminated, it has to be reported. Spontaneous or missed abortion is defined as any foetal death before 24 weeks of gestation.

The MTP for anomaly category is registered as many defects are now diagnosed prenatally and electively terminated.[34] Moreover, if they were not electively terminated they would either result in IUFD or stillbirth.[29] Hence, exclusion of this information would underestimate the prevalence.[34]

On the other hand, spontaneous or missed abortion is excluded from reporting as it may not be recognized by the woman and healthcare provider,[29] especially if it occurs very early in foetal life.[33] Even if it is recognized, not all abortions take place in a hospital setting.[33] Moreover, identifying a structural defect in foetuses at very early gestational ages, especially if maceration has occurred, is very difficult.[33] Because of the poor ascertainment of the denominator and numerator in this category, it has generally been excluded from reporting.

MAJOR AND MINOR ANOMALIES

Registries classify all the eligible anomalies reported to them as either major or minor. Major anomalies are those which are of medical, surgical or cosmetic significance.[27,34] For example, omphalocoele and talipes are grouped under major anomaly and polydactyly under minor anomaly. Some registries do not include minor anomalies as they are variably diagnosed and hence the completeness and accuracy of data are not assured.[35] However, some registries include all minor anomalies because they are often a sign of an underlying syndrome or chromosomal anomaly;[34] for example, the presence of Down syndrome is often manifested as minor anomalies.

CODING OF ANOMALIES

Often, various terminologies are used to describe an anomaly,[34] e.g. anencephaly may be refered to as acephaly. Hence, registries need a coding system that aggregates similar cases. Moreover, as a registry processes a large number of cases, coding of defects is necessary for data storage and retrieval.[34] Several coding systems are presently available, notable among them are the British Paediatric Association's classification of diseases[35] and the (ICD–10) published by WHO, Geneva.[32] ICD–10 uses a four-character alphanumeric code for congenital anomalies with a prefix 'Q', e.g. the code for anencephaly is Q0.00.

STORING DATA

Registries should have an exclusive database to store the data. To protect an individual's identity, data should be stored in a password-protected, secure database.[27] People other than the registry personnel should not be allowed access to data pertaining to the registry. Though various types of databases are available, a relational database should be preferred for ease of data entry and analysis. Data reported from multiple sources have to be matched for an overlap. Redundancy causes overestimation of prevalence. Therefore, registries that use multiple sources of information should have matching fields to avoid redundancy. Each case should be assigned a unique identification number. Cases reported from sources other than obstetricians must be matched to update the diagnosis. If no match is found, that case should be treated as a new one.

CALCULATION OF PREVALENCE

A baby/foetus may have several malformations, but while calculating the prevalence of malformed babies/foetuses, the number of babies/foetuses should be taken into account and not the number of malformations.[36] However, for calculating the anomaly-specific prevalence, each anomaly is counted separately.[36] For example, if a baby/foetus has talipes, omphalocoele and ventricular septal defect, each defect will be counted separately for the calculation of prevalence of such anomalies. However, when system-specific prevalence is calculated, the count would be two for the same child—omphalocoele and talipes are classified under the musculoskeletal system and ventricular septal defect under the circulatory system. While calculating the prevalence, isolated anomalies are not distinguished from anomalies that form part of syndromes or multiple malformations.

FEEDBACK

In a hospital-based registry, active cooperation of all registry members is essential for its effective long term functioning.[24,37] Hence, it is necessary to keep motivating members through meetings and newsletters.[24] During the meetings, registry investigators should present the results of surveillance to the registry members and discuss how the system is contributing to the generation of useful information. As government policy is based on evidence, it is mandatory to publish the findings of the registry in both scientific and lay magazines. Publication of data might also help the public to know the prevailing birth defects in their region and the likely aetiological factors.

ETHICAL ISSUES

It is important to consider ethical issues before setting up a registry. An institutional review board comprising of various sections of the population is mandatory. Informed consent should be taken from every woman who is interviewed by registry personnel in a population-based registry.

INFRASTRUCTURE, STAFF, COST AND FUNDING

Cost and funding are other important considerations in setting up a registry. In India, birth defects are not considered a public health problem, so an organization interested in setting up a registry may initially have to spend its own funds. Over the course of time it might get funds from government or non-governmental agencies. Since a registry of birth defects is a surveillance activity, epidemiologists and biostatisticians are essential. Obstetricians and neonatologists/paediatricians may be used for checking the diagnosis. The number and type of other staff needed to run a registry depends on the kind of registry; for example, in a hospital-based registry with active case ascertainment, trained medical data extractors are necessary and, for a population-based registry, trained social workers are essential to collect data from households. In addition to data entry operators, administrative and secretarial staff are also required for a registry.

CONCLUSION

Birth defect registries are essentially surveillance mechanisms to understand the epidemiology of birth defects. In India, such registries might play a vital role. The data collected by such programmes could be useful for policy-makers to tackle the problem of birth defects. Although three types of registries have been defined, setting up a descriptive type should be the primary objective. Finally, cooperation of all the participatory members is essential for effective functioning of the registry in the long run.

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