BIRTH DEFECTS ASCERTAINMENT IN THE PHILIPPINES

Carmencita D Padilla[1,2], Eva Maria Cutiongco[1,2], Joseph Martin Sia[1]

[1]Institute of Human Genetics, National Institutes of Health, University of the Philippines, Manila,
[2]Department of Pediatrics, College of Medicine, University of the Philippines, Manila

Abstract. In the Philippines, congenital anomalies rank among the top 20 causes of death across the life span and are already the third leading cause of death in the infancy period (Philippine Department of Health, 1996). Despite the magnitude of the problem, no formal systematic registration of birth defects has been done in the country up until 1999. Various attempts have been made by different study groups to gather data but there was never a formal effort to consolidate the information and establish a centralized registry. Data from various modes of ascertainment are hereby presented: 1) Philippine Birth Defects Registry Project, 2) Hospital In-Patient and Out-Patient Registries, 3) Prenatal Inventory and Neonatal Outcome Study Group, 4) Hospital Pathology Reports, and 5) Community Outreach Programs. Birth Defects Registry Project had the largest reporting of the different methods presented here. The most common birth defects were multiple congenital anomalies, congenital malformations of the tongue, mouth, and pharynx (ankyloglossia), cleft palate with cleft lip, Down Syndrome, congenital deformities of the feet (talipes equinovarus), anencephaly, other congenital malformations of the face and neck, congenital malformations of the musculoskeletal system (diaphragmatic hernia, gastroschisis), hypospadias, congenital hydrocephalus, polydactyly, syndactyly, and cleft lip.

INTRODUCTION

In the Philippines, congenital anomalies rank among the top 20 causes of death across the life span and are already the third leading cause of death in the infancy period (Philippine Department of Health, 1996). Despite the magnitude of the problem, no formal systematic registration of birth defects has been done in the Philippines up until 1999. Various attempts have been made by different study groups in the Philippines to gather data but there was never a formal effort to consolidate the information, hence no single reliable source for data on birth defects in the country presently exists. There is therefore a need to establish a centralized registry that will obtain baseline data for use in monitoring trends in the incidence and prevalence of specific birth defects; identify priority areas for research, such as those regarding risk factors and methods of prevention; recognize areas which may need policy intervention; and determine outcomes of treatment and/or preventive programs. Available data may also be used in recommending services for and giving counseling to patients and their families.

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Philippine Birth Defects Registry Project

The Philippine Birth Defects Registry Project is a joint project of the Department of Health and the Institute of Human Genetics-National Institutes of Health, University of the Philippines Manila. It commenced in February 1999 in 79 hospitals nationwide. For the years 1999-2000, the Project included reports from 191,576 deliveries. This represented only 6.3% of annual births in the country. A total of 1,240 cases of birth defects were tallied. The inclusion criteria limited reporting to those cases that could be diagnosed by routine physical examination (ie structural defects such as oral clefts, polydactyly, etc., and chromosomal abnormalities such as Down Syndrome). Inherited and metabolic diseases (eg congenital hypothyroidism), functional problems without obvious structural deformity (eg mental retardation), and poor pregnancy outcomes (eg low birth weight) were not included. The top 10 birth defects are presented in Table 1.

Table 1. Top ten birth defects: Philippine Birth Defects Registry Project, 1999-2000.

Rank	Birth defect	No. of cases	Rate per 10,000
1	Multiple Congenital Anomalies	991	9.9
2	Congenital malformations of tongue, mouth, and pharynx (eg ankyloglossia)	172	8.9
3	Cleft lip with cleft palate	110	5.7
4	Down Syndrome	82	4.3
5	Congenital deformities of the feet (eg talipes equinovarus)	73	3.8
6	Anencephaly and similar malformations	42	2.2
6	Other congenital malformations of the face and neck (eg preauricular skin tags)	42	2.2
7	Congenital malformations of the musculoskeletal system not elsewhere classified (eg diaphragmatic hernia, gastroschisis)	40	2.1
8	Hypospadias	28	1.5
9	Congenital hydrocephalus	27	1.4
9	Polydactyly	27	1.4
9	Syndactyly	27	1.4
10	Cleft lip only	25	1.3

The data collection form of the Philippine Birth Defects Registry Project ascertained information such as parents' data (age, occupation, race, province of origin), baby's data (including birthday, sex, weight, anthropometric measurements, and family history), maternal history (such as infections and exposures to smoke and other pollutants during pregnancy), and pertinent laboratory examination results. An illustration was also provided for the notifier to illustrate the birth defect/s.

Hospital in-patient and out-patient records

The Philippine General Hospital (PGH) is the largest tertiary government hospital in the Philippines. The hospital offers more than 1,400 beds distributed throughout 12 departments. In 2000, it serviced 639,760 persons either as in-patients, out-patients, or emergency patients (PGH, 2001). A review of in-patient records from 1996-2000 at the PGH revealed a total of approximately 6,662 cases with diagnoses that were considered congenital malformations under the International Statistical Classification of Diseases and Related Health Problems system, 10th revision (ICD-10). The top 20 cases of birth defects are listed in Table 2.

Prenatal Inventory and Neonatal Outcome Study Group

The Prenatal Inventory and Neonatal Outcome (PINO) Study Group was formed to determine the accuracy of detection and effectivity of perinatal and neonatal interventions on congenital anomalies. Members of this study group included representatives from various specialties based at the Philippine General Hospital. For the period 2000-2001, there were 73 mothers enrolled with a finding of a congenital anomaly on the fetus on routine obstetric ultrasound. Post-natal verification of the anomalies was assessed and 65.7% had confirmed abnormalities. The top congenital anomalies were: 1) multiple congenital anomalies; 2) congenital hydrocephalus; 3) neural tube defects; 4) cleft lip and /or palate; 5) hydrops fetalis; and 6) congenital heart disease and omphalocoele.

Hospital pathology reports

Autopsy reports from 1995 to 1999 at the Department of Pathology of the College of Medicine, University of the Philippines Manila were reviewed. A total of 68 cases were reported to have birth defects.

Table 2. Top 20 birth defects at the Philippine General Hospital, 1996 - 2000.

Rank	Number of cases	Diagnosis
1	816	Congenital malformation of the heart, unspecified
2	471	Hirschsprung's Disease
3	427	Congenital absence, atresia, and stenosis of anus without fistula
4	426	Unspecified cleft palate with cleft lip, bilateral
5	356	Congenital hydrocephalus, unspecified
6	226	Cleft palate with cleft lip
7	195	Cleft lip
7	195	Multiple congenital malformations, not elsewhere classified
8	189	Patent ductus arteriosus
9	184	Spina bifida, unspecified
10	183	Congenital cataract
11	165	Hypospadias, unspecified
12	164	Cleft palate, unspecified, unilateral
13	158	Cleft palate
14	143	Atresia of bile ducts
15	142	Down Syndrome, unspecified
16	136	Cleft lip, unilateral
17	90	Undescended testicle, unspecified
18	83	Talipes equinovarus
19	81	Encephalocele, unspecified
20	71	Peripheral arteriovenous malformation

The most common malformations were: 1) Congenital heart disease (mostly patent ductus arteriosus); 2) Multiple congenital anomalies; 3) Hydrops fetalis; 4) Down Syndrome with or without other congenital anomalies; 5) Neural tube defects; and 6) Neurofibromatosis.

Community outreach programs

To augment health services in the country, voluntary medical and surgical missions are conducted year-round by different sponsoring agencies. Operation Smile is one of the organizations that has been conducting free surgical missions in various provinces of the Philippines since 1982 with the main purpose of repairing oral clefts. As of 2000, Operation Smile had served at least 1,633 Filipino children aged 10 years old and below in 10 different provinces (Nepomuceno, 2001). Data from Operation Smile indicate that the Philippines has one of the highest rates of oral clefts in the world, with an incidence of 1:500. Under the auspices of Operation Smile, studies are being conducted to determine the genetics of oral clefting in the Philippines, with a focus on understanding the causes of cleft lip and palate and the development of methods to prevent occurrence and recurrence, and improve treatment. More than 6,000 blood samples have been collected from over 75 Filipino families with two or more individuals affected with clefts, and over 20 families with affected sibling pairs.

DISCUSSION

In a developing country such as the Philippines, creating and maintaining a nationwide birth defects monitoring system remains a challenge. Although various methods are in place in the country with regard

to birth defects ascertainment, none of these have produced data that are truly reflective of the situation as each source was limited in its coverage. The Philippine Birth Defects Registry Project had the largest reporting of the different methods presented here. It was implemented in different hospitals nationwide, yet actually covered only a small proportion of total births. This may be because a relatively small number of hospitals were involved in the project. Future expansion to include other hospitals is being considered. It is also important to realize that only 30% of births in the Philippines occur in hospitals, while the rest take place at home. There is a need to reach out to the community setting as an additional source of information. This can be done by involving the midwives, community health workers, and traditional birth attendants in case ascertainment.

The World Health Organization (WHO), in a report of a joint meeting with the World Alliance of Organizations for the Prevention of Birth Defects (WAOPD), has stated that the burden of genetic disorders in the developing world is dynamic, with an ongoing epidemiological transition from infections and malnutrition, to genetic disorders and birth defects as the principal causes of disease and disability. Research is an important component of medical genetics in developing countries, especially as insufficient data are available on the epidemiology of genetic disorders and birth defects (WHO, 1999).

Indeed, a registry of all Filipino newborns with birth defects will allow for the accumulation of data on incidence rates and other related statistics. Data generated from such a registry will be important not only in policy and program planning in health, but also in terms of program development in social and educational services for affected children. A national registry will be instrumental in addressing urgent public health concerns on prenatal care, genetic counseling, and risk factors. Most importantly, it may pave the way for more research that will elucidate the causes of birth defects and ultimately allow for the implementation of effective strategies for primary and secondary prevention. However, before the realization of this national birth defects registry, important issues remain to be addressed adequately.

It has been suggested that data should be collected by the register staff since it results in far more complete and accurate data collection than depending on voluntary registration from busy doctors, nurses, hospital or clinic personnel (Stanley, 1984). However, in a developing country where financial means are scarce, employing a dedicated staff may not be feasible. Instead, the registry must rely on the initiative of health care providers to voluntarily ascertain and report cases.

With either an employed staff or a volunteer health care provider, there is often a lack in the necessary know-how, thus leading to delay in diagnosis and reporting. There is also the unwillingness to fill out the registry forms completely and accurately. There is presently no standard for coding system and data analysis. This list of problems shows the need to conduct further education and training not only for registry team members but also for primary physicians, nurses and midwives. These health personnel must have the commitment and the desire to improve statistics, which in turn may help in the understanding and prevention of birth defects. An awareness campaign must include all persons whether or not they are part of the registry. A technical working group may be created with the purpose of planning for activities and policies, and drafting guidelines for improvement and expansion.

As it often takes several years of preliminary data production before valid trends in incidence are produced, funding may be difficult as research funding groups and governments do not wish to commit funds over long periods of time (Stanley, 1984). Funding must therefore include more than one source to ensure sustainability. The expenses incurred in a registry must be limited to essential administrative and logistic concerns, and innovative approaches to data collection may be employed. For example, the Philippine Birth Defects Registry Project is linked with the Philippine Newborn Screening Program, and both are based in the same institute. Completed forms for the Birth Defects Registry Project are sent to the Secretariat together with the samples for newborn screening, thus eliminating the expense of sending forms via mail or courier service. Linkages with other registry systems (local and international) must be established. Collaboration with the Local Government Unit (LGU) may be initiated for the purpose of manpower and funding. Institutionalizing of the birth defects registry may first start with LGU centers, working towards the involvement of all DOH hospitals, and finally of all hospitals in the country.

The success of a registry is dependent on many factors. All those who might potentially benefit from the data must be willing to actively participate in the project. Sustaining a registry is the bigger challenge. The stakeholders in a birth defects surveillance system include children and parents, health care providers, researchers, policy-makers, public health professionals, and voluntary advocacy agencies.

CONCLUSION

There is a need to consolidate the various methods of birth defects ascertainment in the Philippines in order to produce data that is reliable and accurate. The responsibility for this attempt rests on the potential beneficiaries of a successful registry: researchers, clinicians, policy makers, and even parents and relatives of affected children. It is a huge task that can be overcome with the commitment and willingness of all concerned.

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