Birth Defects at the Outpatient Department of the Philippine General Hospital from 2000-2010

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ABSTRACT

Introduction. Birth defects or congenital anomalies are a major global concern. An estimated 7.9 million children are born worldwide each year. Birth defects are among the top ten leading causes of infant deaths in the Philippines for more than six decades. The objectives of this study were to: 1) determine the frequency of birth defects among patients seen at the Outpatient Department (OPD) of the Philippine General Hospital (PGH) from 2000 to 2010; 2) describe the birth defects by organ systems and presentation (isolated, part of a recognizable syndrome, chromosomal syndrome or multimalformed case); 3) present the distribution of patients by geographic origin; 4) describe the birth defects according to age group and organ system; and 5) compare the data from this study to the previously published report among admitted patients at PGH in the same time period.

Methods. Medical records of new patients seen at the PGH OPD from 2000 to 2010 were reviewed. Medical records that included written diagnosis of any of the following International Classification of Diseases (ICD) -10 codes (Q 00 – Q 99, P 35.0, P 83.5, K40, H49.0, H50.0, H50.1, H53.0, H54.42, H54.7, and H55.01) were considered birth defect cases.

Results. Out of the 804,410 new patients at the PGH OPD from 2000 to 2010, 12,827 patients (1.59%) had a diagnosis of at least one major structural birth defect. The most common birth defects were cardiovascular, digestive, genital organ and nervous system anomalies. The top 5 anomalies in this report were: congenital malformations of cardiac septa, other congenital malformations not elsewhere classified, cleft palate with cleft lip, congenital hydrocoele, and congenital hydrocephalus. The highest percentage of birth defects were from the < 1 age group (40.3%), followed by the 1 to 4 age group (29%) and the 5 to 9 age group (14.6%). NCR, Region IV-A and Region III had the highest percentages of patients with birth defects, 51.4%, 26.03% and 10.97%, respectively.

Conclusion. This study revealed a prevalence of birth defects among PGH OPD patients of 1.59%. The most common birth defects were possibly surgically correctable reflecting the nature of PGH as a referral center. Majority of patients affected were in the under-5 population. The study reflects the importance of a birth defects surveillance to develop policies on strategies that will reduce the burden of morbidity and mortality secondary to preventable birth defects like congenital rubella syndrome that can be aborted by a successful immunization program. The birth defects surveillance will generate data that will support strengthening the regional hospitals with a better complement of specialists and capability for both medical and surgical management of the patients.

Key Words: birth defects, congenital anomalies, congenital abnormalities, Philippine General Hospital

Introduction

Birth defects or congenital anomalies continue to be a major global concern. Serious birth defects of genetic or partially genetic origin are born to an estimated 7.9 million children or six percent of total births every year. Serious birth defects can be lethal and for those who survive, these disorders can cause lifelong disability. There are also hundreds of thousands more who are born with serious birth defects of post-conception origin, including maternal exposure to environmental agents (teratogens) such as alcohol, rubella, syphilis and iodine deficiency that can harm a developing fetus. The March of Dimes Global Report on Birth Defects revealed that at least 3.3 million children under five years of age die from birth defects each year and an estimated 3.2 million of those who survive may be disabled for life.[1]

In the Philippines, congenital anomalies have been consistently in the top ten causes of infant mortality for the past six decades.[2] To influence policy development for surveillance of birth defects, there have been several efforts to gather local data.[3,4] The retrospective study involving patients admitted at the PGH from 2001-2010 revealed a birth defect prevalence of 2% across all age groups.[4]

A birth defect is defined as any abnormality affecting body structure or function that is present from birth. It may be clinically obvious at birth or may be diagnosed only later in life.[1] Synonymous terms that are often used are 'congenital anomalies,' 'congenital abnormalities' and 'congenital malformations'.[5] There are two main categories of birth defects: structural birth defects and functional, or

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developmental birth defects. Structural birth defects are related to a problem with body parts and structure. Functional or developmental birth defects are related to a problem with how a body part or body system works. These problems often lead to intellectual and developmental disability and can include nervous system or brain problems, sensory problems, metabolic disorders, and degenerative disorders.[6]

Birth defects can be single (isolated or sequence) or multiple (associated defect in multimalformed case, part of recognizable syndrome, and chromosomal syndrome). Sequence refers to anomalies due to a single problem in morphogenesis that leads to a cascade of subsequent effects (e.g. amnion rupture sequence).[7] Multiple structural defects that occur due to a single inciting event are called malformation syndromes.[7] Those that are due to a chromosomal problem are called chromosomal syndromes. Those that have been described in literature have been classified as 'part of a recognizable syndrome.' Those that are labelled 'multiple congenital anomalies' with no specific syndrome have been classified 'associated defect in multimalformed case.'

There are major and minor anomalies. Major structural anomalies are defined as structural changes that have significant medical, social or cosmetic consequences, and typically require medical intervention. Examples include cleft lip and spina bifida. In contrast, minor congenital anomalies are structural changes that pose no significant health problem in the neonatal period and tend to have limited social or cosmetic consequences for the affected individual.[5] Examples include ear tags, high arched palate and clinodactyly.

The objectives of the study were to 1) determine the percentage of patients with birth defects at the PGH OPD; 2) describe the birth defects by organ systems and presentation (isolated, part of a recognizable syndrome, chromosomal syndrome or multi-malformed case); 3) present the distribution of patients by geographic origin; 4) describe the birth defects according to age group and organ system; and 5) compare the data from this study to the previously published report among admitted patients at PGH in the same time period.

Methods

Study Setting

The PGH has a 1,500-bed capacity with an average of 600,000 patients seen annually for both inpatients and outpatients. It is state-owned, administered and operated by the University of the Philippines Manila. The OPD houses clinics of the following 12 specialty services: Family Medicine, Internal Medicine, Neurosciences, Dermatology, Allergy and Immunology, Obstetrics and Gynecology, Ophthalmology, Orthopedics, Otorhinolaryngology-Head and Neck Surgery, Pediatrics, Psychiatry, Rehabilitation Medicine and Surgery. Other services/clinics at the OPD are Traditional Medicine, Nutrition Clinic, Anti-Rabies Unit, and Dentistry. Being a tertiary referral center, it receives patient referrals needing specialized medical care and intervention from other healthcare institutions all over the country.

Inclusion Criteria and Exclusion Criteria

Only new patients seen at the PGH OPD from 2000 to 2010 were included in the study. Medical records of outpatients were kept at the Main Medical Records Section and at the Cancer Institute. Only the medical records of outpatients at the Main Medical Records Section were included in this study. Medical records that included written diagnosis of any of the following ICD-10 codes (Q00 – Q99, P35.0, P83.5, K40, H49.0, H50.0, H50.1, H53.0, H54.42, H54.7, and H55.01) were considered birth defect cases. These codes correspond to congenital anomalies and syndrome diagnoses: specifically, Q00 – Q99 represent congenital malformations, deformations and chromosomal abnormalities, P35.0 congenital rubella syndrome, P83.5 congenital hydrocele, K40 inguinal hernia, H49.0 third oculomotor nerve palsy, H50.0 esotropia, H50.1 exotropia, H53.0 amblyopia ex anopsia, H54.42 blindness, left eye, normal vision right eye, H54.7 unspecified visual loss, and H55.01 congenital nystagmus.

In this study, birth defects referred to structural anomalies and not functional ones.

The following were excluded: 1) patients who were reported to have minor birth defects (Appendix A); 2) patients with metabolic disorders and functional problems; and 3) patients who have been initially identified as having birth defects but did not have final or definitive diagnoses such as patients with records containing the following words in the diagnosis: to consider, versus, probably and without supporting laboratory or ancillary procedures.

Birth defects were counted separately such as a patient may have more than one birth defect. Accordingly, the total number of birth defects were more than the total number of patients.

Data Collection

Medical records of all new patients seen at the PGH OPD meeting the inclusion criteria from 2000 to 2010 were retrieved and reviewed. This was done after securing approval from the institution's Ethics Review Board. The following data from the medical records were then encoded into a secure password-protected database: demographic information [name, case number, gender, date of birth, nationality, province of origin, plurality (singleton or twin etc.), parents' identifying data (name, age and occupation)], prenatal data (maternal illness and medications taken during pregnancy), family history, specific diagnoses, and

laboratory work-up. The written diagnoses and ICD-10 codes of the cases were then verified by the genetics fellows and consultants, and recorded.

Data Analysis

Of the total number of new patients seen at the OPD between the years 2000-2010, the frequency of birth defects was reported per 10,000 patients. The number of birth defects was also reported per year. The frequency of birth defects according to type was also reported and the top 10 birth defects were identified.

Results

From 2000 to 2010, there were 822,494 outpatients at the PGH. Of the 822,494 outpatients, there were 804,410 charts available at the Main Medical Records Section for review. Medical records at the Cancer Institute were not reviewed.

Out of the 804,410 new patients who consulted at the PGH OPD from 2000 to 2010, 12,827 patients (1.59%) had a diagnosis of at least one major structural birth defect (Table 1).

Table 1. Annual Number of Patients Seen, Birth Defects Cases and Prevalence at the Outpatient Department of the Philippine General Hospital from 2000 to 2010

Year	Total Number of New OPD Patients	Total Number of OPD Patients with Birth Defects	Patients with Birth Defects Per 10,000 OPD Patients
2000	91,889	1,300	141.5
2001	83,442	1,845	221.1
2002	79,706	1,335	167.5
2003	74,232	1,545	208.1
2004	73,202	1,513	206.7
2005	74,340	1,452	195.3
2006	74,939	914	121.9
2007	66,980	949	141.7
2008	66,033	470	71.2
2009	62,643	758	121.0
2010	57,004	746	130.9
TOTAL	**804,410**	**12,827**	**159.5**

* Cancer and Breast Clinic were excluded

* Included clinics were: Allergy, Dental, Dermatology, Family Medicine, General Surgery, Internal Medicine, Nutrition Clinic, Neurology, OB-Gyne, Orthopedics, Otorhinolaryngology, Pediatrics, Psychiatry, Rehabilitation Medicine, Traditional Medicine, Anti-Rabies Unit

Table 2 shows the top 10 birth defects at the PGH OPD. Three out of the top 10 birth defects were digestive system anomalies - combined cleft palate and cleft lip, isolated cleft palate and congenital absence, atresia and stenosis of the large intestine.

Among the 12,827 patients with birth defects, majority were isolated defects seen in 83% of the cases, 9.1% were associated defects in multimalformed cases, 1.8% with defects that are part of a recognizable syndrome, and 6.1% with defects that are part of chromosomal syndromes (Table 3).

Table 3 also shows that out of the 16, 316 birth defects (some patients had multiple defects), 20.4% were cardiovascular system anomalies, 20.3% were digestive system anomalies, 16.6% were genital organ anomalies and 11.5% were nervous system anomalies (see Appendix B for the complete list of birth defects and their respective ICD-10 codes).

Cardiovascular system anomalies (41.36 per 10,000 patients) and digestive system anomalies (41.14 per 10,000 patients) were the most prevalent birth defects in this study. This was followed by genital organ anomalies (33.61 per 10,000 patients) and nervous system anomalies (23.31 per 10,000 patients).

Among the 3,327 cardiovascular system anomalies, the majority were isolated at 60.4% (2,010 out of 3,327). Most were malformations of the cardiac septa (1,975 out of 3,327, 59.4%) which included ventricular septal defect and atrial septal defect, followed by malformations of the great arteries (814 out of 3,327, 24.5%) in the form of patent ductus arteriosus. For the digestive system anomalies, combined cleft lip and palate (1,153 out of 3,309, 34.8%) topped the list, followed by isolated cleft palate (605 out of 3,309, 18.3%), congenital absence, atresia and stenosis of small intestine (496 out of 3,309, 15%), and isolated cleft lip (433 out of 3,309, 13.1%). For genital organ anomalies, most of the defects identified were hydrocele (1,082 out of 2,704, 40%), undescended testicle (775 out of 2,704, 28.7%) and hypospadias (381 out of 2,704, 14.1%). For nervous system anomalies, the most common reported defect was congenital hydrocephalus (816 out of 1,875, 43.5%). There were no reported anencephaly cases in this study.

Table 2. Top 10 Birth Defects at the Outpatient Department of the Philippine General Hospital from 2000–2010

Rank in Frequency	ICD Code	Diagnosis	Isolated and Sequence	Associated defect in MMC	Part of recognizable syndrome	Part of chromosomal syndromes	Total	Occurrence per 10,000 OPD patients
1	Q21	Congenital malformations of cardiac septa	1,324	447	28	176	**1,975**	**24.55**
2	Q89	Other congenital malformations, not elsewhere classified	157	1181	2	0	**1,340**	**16.66**
3	Q37	Cleft palate with cleft lip	1,036	111	1	5	**1,153**	**14.33**
4	P83.5	Congenital hydrocoele	1,074	8	0	0	**1,082**	**13.45**
5	Q03	Congenital hydrocephalus	741	70	3	2	**816**	**10.14**
6	Q25	Congenital malformations of great arteries	443	263	40	68	**814**	**10.12**
7	Q53	Undescended testicle	630	126	3	16	**775**	**9.63**
8	Q90	Down syndrome	0	0	0	758	**758**	**9.42**
9	Q35	Cleft palate	507	84	7	7	**605**	**7.52**
10	Q42	Congenital absence, atresia and stenosis of large intestine	385	61	4	46	**496**	**6.17**

Table 3. Frequency of Isolated Defects, Associated Defects in Multimalformed Cases (MMC), Defects as Part of Recognizable Syndromes, and Defects as Part of Chromosomal Syndromes by ICD-10 Classification among Patients seen at the Outpatient Department of the Philippine General Hospital from 2000–2010

Organ system	Isolated And Sequence	Associated defect in MMC	Part of Recognizable syndrome	Part of Chromosomal syndromes	Total (% of total defects)	Occurrence per 10,000 patients
Nervous system	1,564	283	19	9	1,875 (11.5%)	23.31
Eye Anomalies	597	207	73	20	897 (5.5%)	11.15
Ear Anomalies, Face and Neck	357	87	34	7	485 (3%)	6.03
Cardiovascular System Anomalies	2,010	985	82	250	3,327 (20.4%)	41.36
Respiratory System Anomalies	40	13	1	1	55 (0.3%)	0.68
Digestive System Anomalies	2,887	336	19	67	3,309 (20.3%)	41.14
Genital Organ Anomalies	2,377	290	8	29	2,704 (16.6%)	33.61
Urinary System Anomalies	32	18	4	2	56 (0.3%)	0.70
Musculoskeletal System Anomalies	595	553	60	35	1,243 (7.6%)	15.45
Chromosomal abnormalities	0	0	0	783	783 (4.8%)	9.73
Others	182	1,196	201	3	1,582 (9.7%)	19.67
Total Number of Birth Defects	**10,641 (65.2%)**	**3,968 (24.3%)**	**501 (3.1%)**	**1,206 (7.4%)**	**16 316**	
Total Number of Patients	**10,641 (83%)**	**1,172 (9.1%)**	**231 (1.8%)**	**783 (6.1%)**	**12 827**	

There were 231 patients with recognizable syndromes comprising 1.8% of all patients included in the study. The most common was congenital rubella syndrome (110 cases, 1.37 per 10,000 patients). Table 3 shows that several organ systems were affected among the recognizable syndromes and the top 3 were cardiovascular system, eye anomalies and musculoskeletal system.

There were 783 cases of chromosomal syndromes comprising 6.1% of all patients included in the study. Down syndrome was the most common chromosomal abnormality (96.8%). Cardiovascular system anomalies and digestive system anomalies were often seen in patients with chromosomal syndromes.

Among the 10,641 recorded isolated defects, digestive and genital organ anomalies made up approximately half of the cases. On the other hand, cardiovascular system anomalies and musculoskeletal system anomalies were the common defects that were associated as part of multimalformed cases.

As shown in Figures 1 and 2, out of the 12,827 patients with birth defects, 94.7% were from Luzon with majority from NCR (51.41%) and IV-A CALABARZON (26.03 %). The hospital also received referrals from the Visayas (2.5%) and Mindanao (1%).

Table 4 shows the breakdown of patients with birth defects according to place of residence, and the percentage of patients of birth defects among the total number of patients with birth defects.

Table 5 shows the distribution of birth defect cases categorized by age group and organ system. The age groups were divided into 5-year categories. The highest percentage of birth defects were from the < 1 age group (40.3%), followed by the 1 to 4 age group (29%) and the 5 to 9 age group (14.6%). In the < 1 age group, the most common isolated defects were digestive system anomalies (28.47%), nervous system anomalies (18.82%) and cardiovascular system anomalies (13.53%). Chromosomal abnormalities (6.6%) and multiple congenital anomalies (11.83%) occurred mostly in the < 1 age group. In younger age groups up to the 25 to 34 age group, genital organ, cardiovascular system anomalies, and digestive system anomalies were in the top three, with slight differences in order/rank. In the 35 to 44 and > 44 age groups, the top three birth defects included genital organ anomalies (most common for both), cardiovascular system anomalies and other anomalies.

It was observed that there has been a decline in the number of patients consulting at the PGH OPD from 2000 to 2010. A similar observation was noted in the retrospective study for PGH inpatients from 2001-2010[4] (Figure 3).

Table 4. Number and percentage of patients with birth defects according to place of regional residence of outpatients at PGH

Regional Residence of patients	Number of Patients with Birth Defects (a)	Percent of Patients with Birth Defects per Region among Total Number of Patients with Birth Defects (a/n*100 where n = 12,827)
Cordillera Administrative Region (CAR)	54	0.42%
National Capital Region (NCR)	6,594	51.41%
Ilocos Region (I)	194	1.51%
Cagayan Valley (II)	97	0.76%
Central Luzon (III)	1,407	10.97%
CALABARZON (IV-A)	3,339	26.03%
MIMAROPA (IV-B)	210	1.64%
Bicol (V)	250	1.95%
Western Visayas (VI)	185	1.44%
Central Visayas (VII)	38	0.30%
Eastern Visayas (VIII)	99	0.77%
Zamboanga Peninsula (IX)	19	0.15%
Northern Mindanao (X)	42	0.33%
Davao (XI)	6	0.05%
SOCCSKSARGEN (XII)	27	0.21%
Caraga (XIII)	24	0.19%
Autonomous Region of Muslim Mindanao (ARMM)	6	0.05%
Unspecified Region (address incomplete)	236	1.84%
Total number of patients with birth defects	12,827	

Table 5. Distribution of Birth Defect Cases seen at PGH OPD from 2000 to 2010 Categorized by Age Group and Organ System

ICD Category	Age Group										
	<1 n=5,168	1 to 4 n=3,723	5 to 9 n=1,869	10 to 14 n=885	15 to 19 n=441	20 to 24 n=235	25 to 34 n=234	35 to 44 n=97	>44 n=47	Unspecified n=128	Total n=12,827
	Percent (%)*	Percent (%)*	Percent (%)*	Percent (%)*	Percent (%)*	Percent (%)*	Percent (%)*	Percent (%)*	Percent (%)*	Percent (%)*	Percent (%)*
Digestive System	28.47	20.08	17.17	13.30	26.40	18.18	12.34	9.28	4.44	23.47	22.51
Genital Organs	9.17	24.77	22.42	29.09	16.78	33.47	38.30	31.96	51.11	2.04	18.53
Cardiovascular System	13.53	15.17	18.78	20.86	21.48	16.12	17.45	18.56	13.33	8.16	15.67
Nervous System	18.82	10.45	6.28	5.30	3.13	2.07	2.55	3.09	0.00	7.14	12.19
Eye, ear, face and neck	4.30	7.75	11.48	8.46	13.65	12.40	10.64	9.28	8.89	24.49	7.44
Musculoskeletal System	5.08	4.64	4.02	4.96	3.80	3.72	3.83	2.06	4.44	1.02	4.64
Respiratory System	0.15	0.29	0.70	0.45	0.67	0.00	0.00	0.00	2.22	0.00	0.31
Urinary System	0.12	0.21	0.32	0.11	0.45	0.41	0.85	4.12	4.44	0.00	0.25
Other Anomalies	0.27	1.29	2.47	2.25	1.57	4.96	3.40	11.34	11.11	11.22	1.42
Multiple Congenital Anomalies	11.83	8.26	7.03	6.43	5.59	2.48	3.83	5.15	2.22	17.35	9.14
Chromosomal Abnormalities	6.60	5.57	8.15	5.75	3.13	0.83	2.55	2.06	0.00	6.12	6.10
Recognizable Syndromes	1.41	1.31	1.45	2.82	2.01	2.48	3.83	3.09	2.22	29.59	1.80

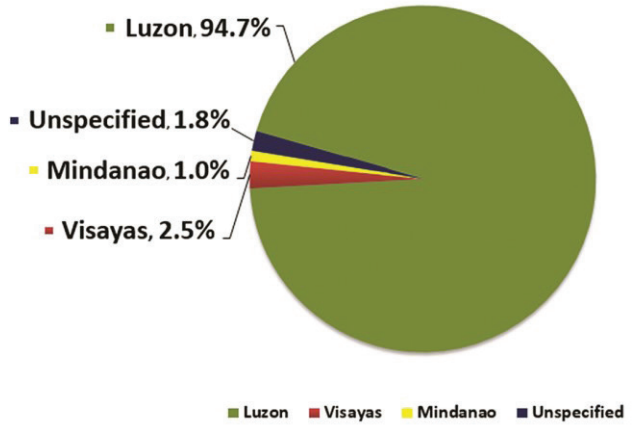

Figure 1. Percentage distribution of patients with birth defects at the Outpatient Department of the Philippine General Hospital from 2000-2010 according to place of residence by island groups.

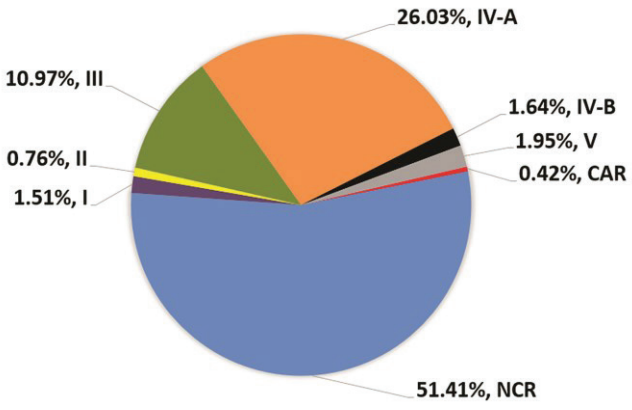

Figure 2. Percentage distribution of patients with birth defects at the Outpatient Department of the Philippine General Hospital from 2000-2010 according to regional resident in Luzon.

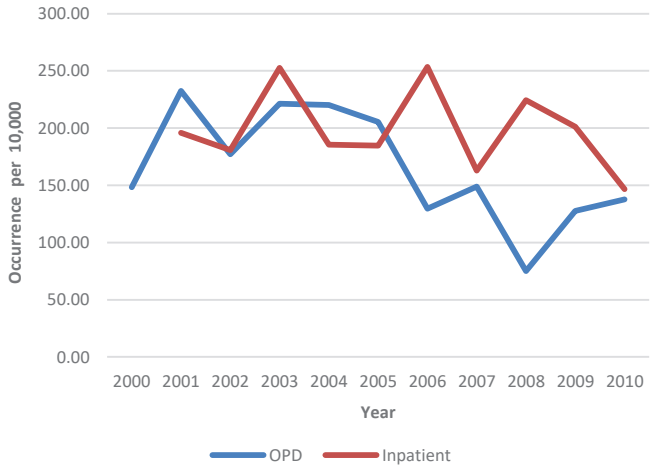

Figure 3. Patients with Birth Defects per 10, 000 OPD patient vs inpatients[4] at PGH from 2000 to 2010.

Discussion

Congenital malformations occur in 2-3% of all pregnancies caused by several contributing factors such as single-gene mutations (20%), chromosomal abnormalities (25%), environmental teratogen (5%), multifactorial (40%), and copy number variants (10%).[8,9] In countries where prenatal diagnosis (ultrasound, chorionic villus sampling or amniocentesis, *etc*) is routinely offered to pregnant women, detection of birth defects or congenital anomalies is made prior to the birth of the baby. This allows psychological preparation of the parents as well as referral to a tertiary hospital for anticipatory care of the baby with serious birth defects. In the Philippines, the great majority of the babies are diagnosed only at birth. In the 2006 March of Dimes Global Report on Birth Defects, the Philippines had an estimated prevalence of 52.9 birth defects per 1,000 live births or 1 birth defect per 19 deliveries.[1] With this estimate, we expect 105,263 birth defects (all types) every year with 2 million babies a year.

The reported prevalence of birth defects in this study was 1.59%, which was lower than most reported prevalence data in other birth defects registry reports.[10,11] This can be attributed to the bigger denominator with the inclusion of all age groups in this study. Most prevalence studies include only the perinatal and neonatal period. The inclusion of all age groups was intentional to capture the burden of birth defects at all age groups.

The prevalence in this study is lower than the prevalence of birth defects among PGH inpatients of 2% during the same time period.[4] The comparison with the PGH inpatients data is an attempt to compare the cases more commonly seen among outpatients vs cases more commonly seen only among inpatients. In both outpatients and inpatients, majority of the patients had isolated birth defects. Birth defects associated in multi-malformed cases ranked second in both studies.

Among outpatients, the less serious cases of birth defects will not need admission and may need only outpatient care. Diagnoses which were more commonly seen among outpatients rather than inpatients were third (oculomotor) nerve palsy, esotropia, exotropia, amblyopia ex anopsia, blindness, unspecified visual loss, congenital nystagmus, congenital hydrocele, and inguinal hernia. In contrast to the PGH inpatient study,[4] there was no reported case of anencephaly among the reported neutral tube defects in this study since patients with anencephaly usually succumb to death within the first few hours to days of life. Among inpatients, the fewer occurrences of chromosomal abnormalities and multiple congenital anomalies in the older age groups reflect the shorter life span of patients with such conditions. Only recognizable syndromes compatible with life reach the older age groups.

There is the possibility on the overlap of cases between the outpatients and the inpatients since patients with surgically correctible birth defects will eventually be admitted. There is also the possibility that the surgery was performed in other hospitals and thus will not be reflected in the PGH inpatient data. There was no attempt to analyze the overlap of cases.

Most birth defects were observed in the under-5 age group with a combined percentage of 69.3% from both the < 1 age group and the 1 to 4 age group. This is a major finding since the under-5 mortality rate in the Philippines remains high at 30 per 1,000 live births.[12-14] Birth defects remain to be a leading cause of infant mortality in the Philippines in the past six decades.[2] The absence of a national birth defect surveillance could be the reason for the absence of focused programs that specifically address its reduction. There is no formal detailed study on the real burden of birth defects or congenital anomalies in the national data on infant mortality where congenital anomalies have consistently been in the top 5.

Just like the March of Dimes Global Report on Birth Defects where congenital heart defects were the most common form of birth defects occurring in 4-8 per 1,000 live births,[1] this study revealed that cardiovascular system anomalies were the most common.

Digestive system anomalies and nervous system anomalies were included in the top birth defects in Iran and Japan as reported in the 2011 Annual Report of the International Clearinghouse for Birth Defects and Surveillance and Research (ICBDSR).[15] For the years 2007–2011, cleft lip with or without cleft palate occurred in 26.01 and 21.67 per 10,000 in Iran and Japan, respectively. Hydrocephalus on the other hand is more common in Iran with a prevalence of 11.38 per 10,000 compared to Japan's 7.82 per 10,000.[15] Although trends seen in this study were similar, it was difficult to make straight comparisons since the patients in this study included all age groups from various outpatient clinics at PGH whereas those of the ICBDSR only included live births, stillbirths and termination of pregnancies.

In both inpatients and outpatients, congenital rubella syndrome (CRS) was prominently observed. There were 110 patients with congenital rubella syndrome (CRS) occurring at 1.37 per 10,000 outpatients in this study whereas 39 patients with CRS were reported in the PGH inpatient study at 0.89 per 10,000 admissions.[4] This study presented an even higher number as compared to the study by Agnas et al which reported the occurrence of 58 cases of CRS for a period of 10 years (1993 – 2002) at PGH.[16] The significance of this data is the fact that it is preventable. Rubella, one of the vaccine-preventable viral conditions, can potentially cause birth defects to the unborn fetus if the mother becomes infected during the first 16 weeks of pregnancy.[1] About 25% of mothers who had rubella during the first trimester of pregnancy will have babies affected with CRS. Birth defects in CRS include blindness, hearing impairment, heart defects, and intellectual disability.[1] With the wide availability of MMR vaccine in the market, national efforts to improve MMR vaccination among Filipinos need to be undertaken and strengthened.

Similarly, there is an opportunity to reduce the burden of neural tube defects (NTD) with anencephaly at the end of the spectrum. Studies have shown that adequate intake of folic acid results in a range of 37% to 92% reduction of NTD occurrence.[17-20] Thus, it is recommended that women of childbearing age should supplement with 0.4mg (400ug) of folic acid daily, two months prior to conception and until the 12th week of gestation.[21-24] In the Philippines, 16% of pregnancies are unplanned and 20% are mistimed.[25]

The hospital received referrals from other regions in as far as the Mindanao areas. The reason for the referrals could be the need for specialized services which are not available in the hospitals in the said areas. One of these specialized services is a formal genetic assessment for proper diagnosis

and management. Currently, there are only ten geneticists who can serve the 100 million Filipinos[12,13] and the majority are practicing in the NCR. While training of additional geneticists is a challenge, efforts have been made to make genetics services more available for Filipinos despite geographical adversities. One major effort is the Telegenetics Referral system, which has been piloted by the Institute of Human Genetics-National Institutes of Health, University of the Philippines Manila in collaboration with the DOH. The aim of this program is to make the services of a genetics team available even in remote areas through email and/or teleconference video calls. Another program to address the lack of genetic services is the offering of the Master of Science in Genetic Counseling by the Department of Pediatrics, College of Medicine, University of the Philippines Manila.[26] It aims to train genetic counselors who can be fielded to the different regions in the country.

The PGH is probably the biggest government hospital with the widest complement of specialized services that can help patients with birth defects. Its residency and fellowship programs have graduated specialists and subspecialists that now man the different hospitals in the country. It is the only hospital with a genetics clinic among its OPD clinics. The PGH, together with National Institutes of Health, provides comprehensive genetic services to its patients through the geneticists, genetic counselors, genetic nurses of the Section of Genetics, PGH Department of Pediatrics and the Institute of Human Genetics, National Institutes of Health.

The data generated by this study may be limited being collected only in one hospital but the number of patients coming from all regions in the country makes it an excellent surrogate of the Philippine scenario. Both studies, the inpatients and the outpatients, can be the basis of programs that can reduce the burden of birth defects through: 1) better preparation of pregnancies (immunization programs, folic acid supplementation, *etc*); 2) early diagnosis of birth defects for anticipatory care of the baby in a well-equipped facility (improved capability of physicians through training of specialists and subspecialists in designated regional and provincial hospitals, upgrading of designated regional and provincial hospitals, *etc*); 3) development of a comprehensive program (both diagnosis, short term and long term management) for patients with birth defects; and 4) the integration of the affected children into society so that that can be productive adults.

The forged partnership between government (Department of Health and the Department of the Interior and Local Government) and academe (National Institutes of Health, University of the Philippines Manila) can develop programs that will redound to the reduction of morbidity and mortality among newborns, children and adults with birth defects or congenital anomalies.

Conclusions

Birth defects occurred in 1.59% of new patients at the OPD of PGH from 2000 to 2010. The most common defects seen were possibly surgically correctable, reflecting the nature of PGH being a tertiary referral center. Majority of patients affected were in the under-5 population, which suggests that birth defects are possibly a contributing factor to morbidity and/or mortality in the country's under-5 population.

The results of this study complement the previously published inpatient data. Both studies similarly show that the most common birth defects were cardiovascular, digestive, genital organ and nervous system anomalies. The top 5 anomalies in this report were: congenital malformations of cardiac septa, other congenital malformations not elsewhere classified, cleft palate with cleft lip, congenital hydrocoele, and congenital hydrocephalus.

The study reinforces the importance of a birth defects surveillance to develop policies on strategies that will reduce the burden of morbidity and mortality secondary to preventable birth defects. Excellent examples are congenital rubella syndrome that can be aborted by a successful immunization program and the folic acid supplementation and fortification that can dramatically reduce neural tube defects. The birth defects surveillance will also generate the data that will support strengthening the regional hospitals with a better complement of specialists and capability for both medical and surgical management of the patients.

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Statement of Authorship

All authors have approved the final version submitted.

Author Disclosure

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Appendix A: List of minor defects excluded from this study

- frontal bossing
- epicanthal folds
- proptosis
- hypertelorism
- hypotelorism
- low set ears
- deformation of ears
- folded superior helix
- prominent ears/ antihelix
- flat nasal bridge
- upturned nose
- shallow nasolabial fold
- long philtrum
- maxillary hypoplasia
- widely spaced teeth
- macroglossia
- ankyloglossia
- high arched palate
- narrow palate
- congenital prognathia
- micrognathia
- webbed neck
- sandal gap deformity
- midphalanx hypoplasia
- clinodactyly
- simian crease
- phimosis
- sacral dimpling
- skin tag
- hemangioma
- lymphangioma
- melanocytic nevi

Appendix B: Complete list of birth defects and their respective ICD codes

ICD Code		Isolated and Sequence	Associated defect in MMC	Part of recognizable syndrome	Part of chromosomal syndromes	Total	Occurrence per 10,000 patients
Nervous system		**1, 564**	**283**	**19**	**9**	**1, 875**	**23.31**
Q00	Anencephaly and similar malformations	0	0	0	0	**0**	**0.00**
Q01	Encephalocele	315	42	0	0	**357**	**4.44**
Q02	Microcephaly	198	118	15	7	**338**	**4.20**
Q03	Congenital hydrocephalus	741	70	3	2	**816**	**10.14**
Q04	Other congenital malformations of the brain	47	25	0	0	**72**	**0.90**
Q05	Spina bifida	226	25	1	0	**252**	**3.13**
Q06	Other congenital malformations of the spinal cord	4	1	0	0	**5**	**0.06**
Q07	Other congenital malformations of the nervous system	33	2	0	0	**35**	**0.44**
Eye Anomalies		**597**	**207**	**73**	**20**	**897**	**11.15**
Q10	Congenital malformations of eyelid, lacrimal apparatus and orbit	101	25	4	8	**138**	**1.72**
Q11	Anophthalmos, microphthalmos and macrophthalmos	37	57	2	0	**96**	**1.19**
Q12	Congenital lens malformations	288	56	64	6	**414**	**5.15**
Q13	Congenital malformations of anterior segment of eye	20	18	2	0	**40**	**0.50**
Q14	Congenital malformations of posterior segment of eye	2	2	0	0	**4**	**0.05**
Q15	Other congenital malformations of eye	41	15	1	1	**58**	**0.72**
H49.0	Third (oculomotor) nerve palsy	1	0	0	0	**1**	**0.01**
H50.0	Esotropia	96	30	0	5	**131**	**1.63**
H50.1	Exotropia	7	0	0	0	**7**	**0.09**
H53.0	Amblyopia ex anopsia	0	1	0	0	**1**	**0.01**
H54.42	Blindness, left eye, normal vision right eye	1	0	0	0	**1**	**0.01**
H54.7	Unspecified Visual Loss	0	2	0	0	**2**	**0.02**
H55.01	Congenital nystagmus	3	1	0	0	**4**	**0.05**
Ear Anomalies, Face and Neck		**357**	**87**	**34**	**7**	**485**	**6.03**
Q16	Congenital malformations of ear causing impairment of hearing	263	27	29	4	**323**	**4.02**
Q17	Other congenital malformations of ear	61	35	2	1	**99**	**1.23**
Q18	Other congenital malformations of face and neck	33	25	3	2	**63**	**0.78**
Cardiovascular System Anomalies		**2, 010**	**985**	**82**	**250**	**3, 327**	**41.36**
Q20	Congenital malformations of cardiac chambers and connections	34	90	0	1	**125**	**1.55**
Q21	Congenital malformations of cardiac septa	1, 324	447	28	176	**1, 975**	**24.55**
Q22	Congenital malformations of pulmonary and tricuspid valves	87	134	8	3	**232**	**2.88**
Q23	Congenital malformations of aortic and mitral valves	11	7	4	0	**22**	**0.27**
Q24	Other congenital malformations of heart	61	40	2	2	**105**	**1.31**
Q25	Congenital malformations of great arteries	443	263	40	68	**814**	**10.12**
Q26	Congenital malformations of great veins	12	3	0	0	**15**	**0.19**
Q27	Other congenital malformations of peripheral vascular system	1	1	0	0	**2**	**0.02**
Q28	Other congenital malformations of circulatory system	37	0	0	0	**37**	**0.46**
Respiratory System Anomalies		**40**	**13**	**1**	**1**	**55**	**0.68**
Q30	Congenital malformations of nose	6	12	1	0	**19**	**0.24**
Q31	Congenital malformations of larynx	26	0	0	1	**27**	**0.34**
Q32	Congenital malformations of trachea and bronchus	0	0	0	0	**0**	**0.00**
Q33	Congenital malformations of lung	2	1	0	0	**3**	**0.04**
Q34	Other congenital malformations of respiratory system	6	0	0	0	**6**	**0.07**
Digestive System Anomalies		**2, 887**	**336**	**19**	**67**	**3, 309**	**41.14**
Q35	Cleft palate	507	84	7	7	**605**	**7.52**
Q36	Cleft lip	405	26	1	1	**433**	**5.38**
Q37	Cleft palate with cleft lip	1, 036	111	1	5	**1, 153**	**14.33**
Q38	Other congenital malformations of tongue, mouth and pharynx	3	16	3	0	**22**	**0.27**
Q39	Congenital malformations of esophagus	3	10	3	0	**16**	**0.20**
Q40	Other congenital malformations of upper alimentary tract	0	0	0	0	**0**	**0.00**
Q41	Congenital absence, atresia and stenosis of small intestine	2	1	0	1	**4**	**0.05**
Q42	Congenital absence, atresia and stenosis of large intestine	385	61	4	46	**496**	**6.17**
Q43	Other congenital malformations of intestine	245	12	0	4	**261**	**3.24**
Q44	Congenital malformations of gallbladder, bile ducts and liver	301	15	0	3	**319**	**3.97**
Q45	Other congenital malformations of digestive system	0	0	0	0	**0**	**0.00**
Genital Organ Anomalies		**2, 377**	**290**	**8**	**29**	**2, 704**	**33.61**
Q50	Congenital malformations of ovaries, fallopian tubes and broad ligaments	0	1	0	0	**1**	**0.01**
Q51	Congenital malformations of uterus and cervix	7	2	0	0	**9**	**0.11**
Q52	Other congenital malformations of female genitalia	63	13	2	1	**79**	**0.98**
Q53	Undescended testicle	630	126	3	16	**775**	**9.63**
Q54	Hypospadias	295	77	1	8	**381**	**4.74**
Q55	Other congenital malformations of male genital organs	0	19	0	0	**19**	**0.24**
Q56	Indeterminate sex and pseudohermaphroditism	11	30	1	3	**45**	**0.56**
P83.5	Congenital hydrocoele	1, 074	8	0	0	**1, 082**	**13.45**
K40	Inguinal hernia	297	14	1	1	**313**	**3.89**

Urinary System Anomalies		**32**	**18**	**4**	**2**	**56**	**0.70**
Q60	Renal agenesis and other reduction defects of kidney	2	3	1	0	**6**	**0.07**
Q61	Cystic kidney disease	14	2	0	0	**16**	**0.20**
Q62	Congenital obstructive defects of renal pelvis and congenital malformations of ureter	6	3	1	1	**11**	**0.14**
Q63	Other congenital malformations of kidney	3	3	2	1	**9**	**0.11**
Q64	Other congenital malformations of urinary system	7	7	0	0	**14**	**0.17**
Musculoskeletal System Anomalies		**595**	**553**	**60**	**35**	**1, 243**	**15.45**
Q65	Congenital deformities of hip	6	9	2	0	**17**	**0.21**
Q66	Congenital deformities of feet	301	128	4	14	**447**	**5.56**
Q67	Congenital musculoskeletal deformities of head, face, spine and chest	14	56	5	2	**77**	**0.96**
Q68	Other congenital musculoskeletal deformities	3	13	0	0	**16**	**0.20**
Q69	Polydactyly	37	39	3	3	**82**	**1.02**
Q70	Syndactyly	36	73	7	3	**119**	**1.48**
Q71	Reduction defects of upper limb	5	16	0	4	**25**	**0.31**
Q72	Reduction defects of lower limb	0	21	1	0	**22**	**0.27**
Q73	Reduction defects of unspecified limb	0	5	0	0	**5**	**0.06**
Q74	Other congenital malformations of limb(s)	26	95	14	7	**142**	**1.77**
Q75	Other congenital malformations of skull and face bones	78	42	1	1	**122**	**1.52**
Q76	Congenital malformations of spine and bony thorax	40	27	5	0	**72**	**0.90**
Q77	Osteochondrodysplasia with growth of tubular bones and spine	0	2	9	0	**11**	**0.14**
Q78	Other osteochondrodysplasias	0	0	6	0	**6**	**0.07**
Q79	Congenital malformations of the musculoskeletal system, not elsewhere classified	49	27	3	1	**80**	**0.99**
Others		**182**	**1, 196**	**201**	**3**	**1, 582**	**19.67**
Q80	Congenital ichthyosis	0	0	11	0	**11**	**0.14**
Q82	Other congenital malformations of skin	6	1	1	0	**8**	**0.10**
Q83	Congenital malformations of breast	0	4	0	0	**4**	**0.05**
Q84	Other congenital malformations of integument	3	6	0	0	**9**	**0.11**
Q85	Phakomatoses, not elsewhere classified	1	0	31	0	**32**	**0.40**
Q86	Congenital malformation syndromes due to known exogenous causes, not elsewhere classified	0	0	1	0	**1**	**0.01**
Q87	Other specified congenital malformation syndromes affecting multiple systems	15	4	45	3	**67**	**0.83**
Q89	Other congenital malformations, not elsewhere classified	157	1, 181	2	0	**1, 340**	**16.66**
P35	Congenital rubella syndrome	0	0	110	0	**110**	**1.37**
Chromosomal abnormalities		**0**	**0**	**0**	**783**	**783**	**9.73**
Q90	Down syndrome	0	0	0	758	**758**	**9.42**
Q91	Edwards and Patau syndrome	0	0	0	5	**5**	**0.06**
Q92	Other trisomies and partial trisomies of the autosomes, not elsewhere classified	0	0	0	0	**0**	**0.00**
Q93	Monosomies and deletions from the autosomes, not elsewhere classified	0	0	0	4	**4**	**0.05**
Q96	Turner syndrome	0	0	0	12	**12**	**0.15**
Q97	Other sex chromosome abnormalities, female phenotype, not elsewhere classified	0	0	0	0	**0**	**0.00**
Q98	Other sex chromosome abnormalities, male phenotype, not elsewhere classified	0	0	0	1	**1**	**0.01**
Q99	Other chromosomal abnormalities, not elsewhere classified	0	0	0	3	**3**	**0.04**
TOTAL DEFECTS		10, 641	3, 968	501	1, 206	**16, 316**	
TOTAL CASES		10, 641	1, 172	231	783	**12, 827**	