

## Occurrence of Birth Defects at the Philippine General Hospital: 2001-2010

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### ABSTRACT

**Introduction.** Birth defects are a global problem with impact particularly severe in middle- to low- income countries. In the Philippines, there is limited data on birth defects despite the fact that congenital anomalies have been in the top 10 causes of infant mortality. The objectives of the study were: 1) to determine the occurrence of birth defects among patients admitted to the Philippine General Hospital (PGH); 2) to present the distribution of patients by geographic location and age group distribution; 3) to categorize birth defects by organ systems; and 4) to categorize birth defects as either isolated, part of a recognizable syndrome, chromosomal syndrome or multi-malformed case.

**Methods.** Patients admitted to the PGH from 2001-2010 and reported to have major structural defects were included in this study. Case ascertainment was done through a review of medical records of all admitted patients age 0 to more than 65 years old. Patients with birth defects were assigned codes of the International Classification of Diseases (ICD)-10 classification.

**Results.** Of the 438,944 admissions to the PGH from 2001 to 2010, there were 8,686 (2.0%) patients with a diagnosis of at least one (1) birth defect. The most common birth defects are as follows: digestive system (3,605/8,686 or 41.5%), cardiovascular system (2,839/8,686 or 32.7%), nervous system (1,070/8,686 or 12.3%), and genital organ anomalies (755/8,686 or 8.7%). The most common digestive system anomalies were cleft lip and/or palate (1,548/8,686 or 17.8%), imperforate anus (698/8,686 or 8%) and Hirschsprung disease (582/8,686 or 6.7%). Most of the cardiovascular system anomalies were congenital malformations of the cardiac septa (1,160/8,686 or 13.4%) and the great arteries (769/8,686 or 8.9%), while most of the nervous system anomalies were due to congenital hydrocephalus (347/8,686

or 4%), encephalocele (303/8,686 or 3.5%) and spina bifida (193/8,686 or 2.22%). The most common genital organ anomalies were hypospadias (340/8,686 or 3.9%) and undescended testicle (233/8,686 or 2.7%). Majority (4,042/8,686 or 46.5%) of birth defect cases came from the National Capital Region (NCR) while 32.5% (or 2,827/8,686) of the cases came from Region IV-A or Cavite, Laguna, Batangas, Rizal and Quezon (CALABARZON) Region.

**Conclusion.** The results of this study show that the most common birth defects are digestive, cardiovascular, nervous system, and genital organ anomalies. This trend is similar to those reported internationally. The findings of the study can be the basis of policies toward the development and implementation of practical strategies for primary and secondary prevention of birth defects among Filipinos.

*Key Words:* birth defects, major birth defects, surveillance

### Introduction

Every year, an estimated 7.9 million children or about 6% of total births worldwide are born with a serious birth defect. At least 3.3 million children less than 5 years of age die annually because of serious birth defects and the majority of those who survive may be mentally and physically disabled for life. In the United States, structural or genetic birth defects affect approximately 3% of births and are a major contributor to infant mortality and result in billions of dollars in costs for care.<sup>1</sup>

The impact of birth defects is particularly severe in low- and middle-income countries (LMICs) where more than 94% of the births with serious birth defects and 95% of the deaths of these children occur. At present, most of these countries do not have established population-based birth defects surveillance programs.<sup>2</sup> The Centers for Disease Control and Prevention (CDC), Atlanta, Georgia, USA has taken the lead in surveillance programs through the development of the Metropolitan Atlanta Congenital Defects Program (MACDP) in 1967. Strengthening birth defects surveillance in the United States helped identify disparities in neural tube defect (NTD) prevalence and evaluation of the effectiveness of interventions, such as fortification of enriched cereal grain products with folic acid at the level of 140 µg/100 grams of grain.<sup>3</sup>

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Successful folic acid campaigns for the prevention of NTDs have also been carried out in China, Canada and Mexico.<sup>4,5,6</sup> In China, periconceptional intake of 400 µg of folic acid daily reduced the risk of neural tube defects in areas with high rates of these defects.<sup>4</sup> In Canada and Nova Scotia, a significant reduction in neural tube defects was observed after the implementation of food fortification.<sup>5</sup> In Mexico, free distribution of 5 mg tablets of folic acid weekly reduced the incidence of anencephaly and spina bifida cases into half.<sup>6</sup>

Cost-effective approaches to markedly reduce the toll of birth defects in lower-income countries are available. Evidence suggests strongly that with widespread implementation of these interventions, up to 70% of death and disability from birth defects could be prevented, ameliorated, or cured. The majority of recommended interventions such as controlling maternal infections and other health conditions that are known to increase risk of birth defects and improving access to perinatal health care services, including preconception services, are integral components of rational public health programs. Thus, such approaches will build on public health efforts by broadening the current scope of woman, maternal, newborn and child health services in primary health care and developing secondary and tertiary medical genetic services. Investment in the infrastructure of birth defects surveillance in LMICs is essential for the ultimate development, implementation, and evaluation of birth defects prevention activities in at-risk populations around the world.<sup>3,7</sup>

#### *Philippine Data on Birth Defects*

Congenital anomalies have been in the top 10 causes of infant mortality for the past 50 years.<sup>8</sup> However, there is no formal birth defects registry in the country. There have been no details on the congenital anomalies that have been identified by the national health statistics. A pilot study was conducted by the Department of Health-Essential National Health Research (DOH-ENHR) with the Institute of Human Genetics-National Institutes of Health in 1999-2000 involving 79 hospitals which showed that neural tube defects ranked 6<sup>th</sup>. A review of records of admitted patients from 1996-2000 at the Philippine General Hospital (PGH) showed that NTDs ranked 9<sup>th</sup>.<sup>9</sup>

The main objective of this study was to determine the occurrence of birth defects among admitted patients at PGH from 2001 to 2010. The specific objectives of the study were: 1) to categorize birth defects by organ systems; 2) to present the distribution of patients by origin of geographic location and age group distribution; and 3) to categorize birth defects as either isolated, part of a recognizable syndrome, chromosomal syndrome or multi-malformed case.

## Methods

### Description of the Study Setting

The PGH is the national university hospital and the largest tertiary referral hospital in the Philippines, with a 1,500-bed capacity and an average of 600,000 patients seen annually. It is state-owned, and administered and operated by the University of the Philippines Manila. There are 14 clinical departments, namely: Anesthesiology, Internal Medicine, Surgery, Neurosciences, Pediatrics, Otorhinolaryngology-Head & Neck Surgery, Ophthalmology, Orthopedics, Rehabilitation Medicine, Psychiatry, Radiology, Pathology, Emergency Medicine and Obstetrics & Gynecology. PGH also has the biggest number of subspecialty services. Being a tertiary referral center, it receives patient referrals needing specialized medical care and surgery from other institutions all over the country.

### Subjects

Patients admitted to PGH from 2001 to 2010 and reported to have major birth defects were included in this study.

### Data Collection

Medical records of all patients admitted from 2001-2010 were retrieved and reviewed by doctors and nurses trained for this project. Patients with major structural defects were given International Classification of Diseases (ICD)-10 codes in Chapters Q, P 2.8, and P 35.0. Metabolic disorders and functional problems without obvious structural deformity were excluded.

ICD-10 classifies birth defects cases as "congenital malformations, deformations and chromosomal abnormalities" or Q00-Q99. Cases classified under Amnion Rupture Sequence (P 02.8) and Congenital Rubella Syndrome (P 35.0) were also included in the study.

A secure data system was used to protect the patients. The following information were obtained: demographic information (identification of case patient, case mother, case father, nationality, sex, date of birth), specific written diagnoses, ICD-10 codes, prenatal data and family history.

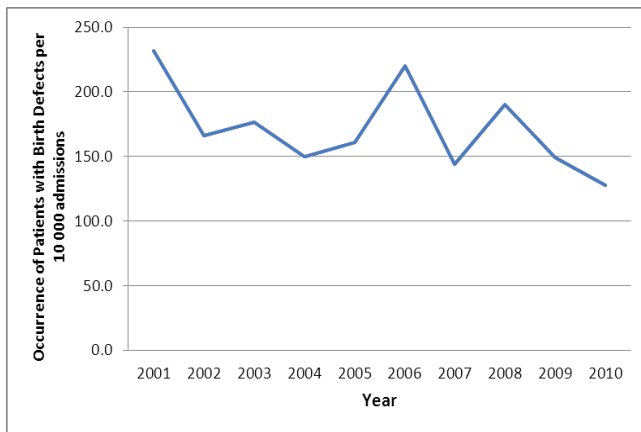
## Results

### General Findings

Of the 6,092,162 patients seen at PGH from 2001-2010, a total of 438,944 patients were admitted; and 8,686 (2.0%) had a diagnosis of at least one birth defect. Table 1 shows the total number of patients, admissions and occurrence of birth defects in PGH. Figure 1 shows the trend in the number of birth defect cases from 2001 to 2010.

**Table 1.** Total Number of Patients, Admissions, Birth Defects Cases and Prevalence at the Philippine General Hospital from 2001-2010

Year	Total Number of Patients	Total Number of Admissions	Total Number of Patients with Birth Defects	Occurrence of Patients with Birth Defects per 10,000 admissions
2001	629,328	59,609	1,168	195.94
2002	665,206	47,600	860	180.67
2003	582,186	35,705	902	252.62
2004	620,831	43,096	800	185.63
2005	631,725	47,499	877	184.63
2006	613,616	45,294	1,148	253.46
2007	576,419	43,917	715	162.81
2008	597,996	41,828	939	224.49
2009	596,849	34,157	687	201.13
2010	578,006	40,239	590	146.62
<b>Total</b>	<b>6,092,162</b>	<b>438,944</b>	<b>8,686</b>	



**Figure 1.** Occurrence of Patients with Birth Defects per 10 000 admissions at the Philippine General Hospital from 2001-2010

Table 2 shows the number of birth defects by ICD-10 classification. From 2001-2010, there were 8,686 patients with birth defects admitted at the PGH. Some of these patients have two or more birth defects, giving a total number of 11,062 defects. There were 6,917 patients (80%) with isolated defects; 1,206 (14%) with associated major defect in multi-malformed cases (MMC); 263 (3%) with defects that are part of recognizable syndromes; and 300 (3.4%) with defects as part of chromosomal syndromes. There were 3,605 (41.5%) digestive system anomalies, 2,839 (32.7%) cardiovascular system anomalies, 1,070 (12.3%) nervous system anomalies, and 755 (8.7%) genital organ anomalies.

For digestive system anomalies, combined cleft lip/palate (CLP) (975/3,605) topped the list, followed by imperforate anus (698/3,605), Hirschsprung disease (582/3,605), and isolated cleft lip (CL) (316/3,605). There was also a significant number of anomalies in gallbladder, bile

ducts and liver (437/3,605), the most common of which are biliary atresia (286/3,605) and choledochal cyst (131/3,605). Digestive system anomalies generally occurred as isolated defects (3,091/3,605). Of the 743 cases, 59 or 7.9% of cases of congenital absence, atresia, and stenosis of the large intestine, or imperforate anus occurred as part of chromosomal syndrome (Table 2).

For cardiovascular system anomalies, most of the cases (1,160/2,839) had congenital malformations of the cardiac septa particularly ventricular septal defect (576/2,839), atrial septal defect (297/2,839) and tetralogy of Fallot (281/2,839). This was followed by congenital malformations of the great arteries in the form of patent ductus arteriosus (672/2,839). Among these anomalies, 1,486 were isolated; 1,140 were part of multi-malformed case; 37 were part of a recognizable syndrome and 176 were part of a chromosomal syndrome (Table 2).

For nervous system anomalies, most of the cases had congenital hydrocephalus, encephalocele and spina bifida comprising 32.4% (347/1,070), 28.3% (303/1,070) and 18% (193/1,070) of the cases, respectively (Table 2).

For genital organ anomalies, most of the cases had hypospadias and undescended testicle comprising 45% (340/755) and 31% (233/755) of the cases, respectively (Table 2).

There were 263 cases of recognizable syndromes comprising 3% (263/8,686) of all patients. The most common recognizable syndromes identified were Neurofibromatosis (50/263), Congenital Rubella Syndrome (39/263) and Osteogenesis Imperfecta (20/263) (Table 2).

There were 300 cases of chromosomal anomalies comprising 3.4% (300/8,686) of all patients, 87.3% (262/300) of whom had Down Syndrome (Table 2).

Table 3 shows the top 10 birth defects in the PGH from 2001-2010. Four in the top 10 are digestive system anomalies (combined cleft palate and cleft lip, imperforate anus, Hirschsprung disease, and isolated cleft lip); three are cardiovascular system anomalies (patent ductus arteriosus, ventricular septal defect and atrial septal defect); two are nervous system anomalies (congenital hydrocephalus and encephalocele); and one is a genital organ anomaly (hypospadias). The high number of surgical cases is due to the status of PGH as a tertiary referral center.

Table 4 shows the breakdown of number of patients with birth defect cases classified according to region of residence. Majority (or 46.5%) of birth defect cases came from the National Capital Region (NCR) with 4,042 cases while 32.5% of the cases came from Region IV-A or Cavite, Laguna, Batangas, Rizal and Quezon (CALABARZON) Region with 2,827 cases. The numbers can be explained by the proximity of NCR and CALABARZON regions to PGH (Figure 2). Despite the distance, 2.2% of the cases (191 cases) and 0.9% (83 cases) came from Visayas and Mindanao, respectively.

**Table 2.** Frequency of Isolated Defects, Associated defects in Multi-malformed Cases (MMC), Defects as Part of Recognizable Syndromes, and Defects as Part of Recognizable Chromosomal Syndromes by ICD-10 Classification among Patients admitted at the Philippine General Hospital from 2001- 2010

ICD Code	Isolated defect / Sequence	Associated defect in MMC	Part of recognizable syndromes	Part of chromosomal syndromes	Total	Occurrence per 10 000 admissions
<b>Nervous System</b>	<b>753</b>	<b>311</b>	<b>5</b>	<b>1</b>	<b>1070</b>	<b>24.38</b>
Q 00 Anencephaly	3	0	0	0	3	0.07
Q 01 Encephalocele	247	55	1	0	303	6.90
Q 02 Microcephaly	2	29	1	1	33	0.75
Q 03 Congenital hydrocephalus	262	85	0	0	347	7.91
Q 04 Other congenital malformations of brain	57	40	0	0	97	2.21
Q 05 Spina bifida	127	63	3	0	193	4.40
Q 06 Other congenital malformations of spinal cord	2	6	0	0	8	0.18
Q 07 Other congenital malformations of nervous system	53	33	0	0	86	1.96
<b>Eye Anomalies</b>	<b>243</b>	<b>63</b>	<b>18</b>	<b>8</b>	<b>332</b>	<b>7.56</b>
Q 10 Congenital ptosis	35	16	0	1	52	1.18
Q 11 Anophthalmos, microphthalmos and macropthalmos	19	19	0	1	39	0.89
Q 12 Congenital lens malformations	141	13	16	6	176	4.01
Q 13 Congenital malformations of anterior segment of eye	7	6	0	0	13	0.30
Q 14 Congenital malformations of posterior segment of eye	3	1	1	0	5	0.11
Q 15 Other congenital malformations of eye	38	8	1	0	47	1.07
<b>Ear Anomalies</b>	<b>46</b>	<b>72</b>	<b>7</b>	<b>3</b>	<b>128</b>	<b>2.92</b>
Q 16 Congenital malformations of ear causing impairment of hearing	11	6	4	0	21	0.48
Q 17 Other congenital malformations of ear	1	45	2	3	51	1.16
Q 18 Other congenital malformations of face and neck	34	21	1	0	56	1.28
<b>Cardiovascular System Anomalies</b>	<b>1486</b>	<b>1140</b>	<b>37</b>	<b>176</b>	<b>2839</b>	<b>64.68</b>
Q 20 Congenital malformations of cardiac chambers and connections	50	150	3	2	205	4.67
Q 21 Congenital malformations of cardiac septa	612	437	9	102	1160	26.43
Q 22 Congenital malformations of pulmonary and tricuspid valves	48	166	7	4	225	5.13
Q 23 Congenital malformations of aortic and mitral valves	13	29	0	0	42	0.96
Q 24 Other congenital malformations of heart	61	91	2	8	162	3.69
Q 25 Congenital malformations of great arteries	436	257	16	60	769	17.52
Q 26 Congenital malformations of great veins	15	8	0	0	23	0.52
Q 27 Other congenital malformations of peripheral vascular system	106	2	0	0	108	2.46
Q 28 Other congenital malformations of circulatory system	145	0	0	0	145	3.30
<b>Respiratory System Anomalies</b>	<b>59</b>	<b>28</b>	<b>0</b>	<b>1</b>	<b>88</b>	<b>2.00</b>
Q 30 Congenital malformations of nose	2	13	0	1	16	0.36
Q 31 Congenital malformations of larynx	11	6	0	0	17	0.39
Q 32 Congenital malformations of trachea and bronchus	1	3	0	0	4	0.09
Q 33 Congenital malformations of lung	44	6	0	0	50	1.14
Q 34 Other congenital malformations of respiratory system	1	0	0	0	1	0.02
<b>Digestive System Anomalies</b>	<b>3091</b>	<b>417</b>	<b>18</b>	<b>79</b>	<b>3605</b>	<b>82.13</b>
Q 35 Cleft palate	210	38	5	4	257	5.85
Q 36 Cleft lip	293	22	0	1	316	7.20
Q 37 Cleft palate with cleft lip	889	79	4	3	975	22.21
Q 38 Other congenital malformations of tongue, mouth and pharynx	3	36	6	0	45	1.03
Q 39 Congenital malformations of oesophagus	44	26	1	2	73	1.66
Q 40 Other congenital malformations of upper alimentary tract	1	0	0	0	1	0.02
Q 41 Congenital absence, atresia and stenosis of small intestine	34	6	0	5	45	1.03
Q 42 Congenital absence, atresia and stenosis of large intestine	526	157	1	59	743	16.93
Q 43 Other congenital malformations of intestine	669	32	1	4	706	16.08
Q 44 Congenital malformations of gallbladder, bile ducts and liver	417	19	0	1	437	9.96
Q 45 Other congenital malformations of digestive system	5	2	0	0	7	0.16

<b>Genital Organ Anomalies</b>	<b>520</b>	<b>226</b>	<b>2</b>	<b>7</b>	<b>755</b>	<b>17.20</b>
Q 50 Congenital malformations of ovaries, fallopian tubes and broad ligaments	7	1	0	0	8	0.18
Q 51 Congenital malformations of uterus and cervix	51	15	1	0	67	1.53
Q 52 Other congenital malformations of female genitalia	46	13	0	1	60	1.37
Q 53 Undescended testicle	170	57	1	5	233	5.31
Q 54 Hypospadias	241	99	0	0	340	7.75
Q 55 Other congenital malformations of male genital organs	2	22	0	0	24	0.55
Q 56 Indeterminate sex and pseudohermaphroditism	3	19	0	1	23	0.52
<b>Urinary System Anomalies</b>	<b>159</b>	<b>60</b>	<b>3</b>	<b>1</b>	<b>223</b>	<b>5.08</b>
Q 60 Renal agenesis and other reduction defects of kidney	17	14	0	0	31	0.71
Q 61 Cystic kidney disease	51	11	0	0	62	1.41
Q 62 Congenital obstructive defects of renal pelvis and congenital malformations of ureter	48	16	2	0	66	1.50
Q 63 Other congenital malformations of kidney	10	4	1	0	15	0.34
Q 64 Other congenital malformations of urinary system	33	15	0	1	49	1.12
<b>Musculoskeletal System Anomalies</b>	<b>367</b>	<b>280</b>	<b>79</b>	<b>9</b>	<b>735</b>	<b>16.74</b>
Q 65 Congenital deformities of hip	27	7	0	0	34	0.77
Q 66 Congenital deformities of feet	69	70	7	3	149	3.39
Q 67 Congenital musculoskeletal deformities of head, face, spine and chest	3	9	4	0	16	0.36
Q 68 Other congenital musculoskeletal deformities	4	10	0	1	15	0.34
Q 69 Polydactyly	14	16	0	2	32	0.73
Q 70 Syndactyly	22	22	2	0	46	1.05
Q 71 Reduction defects of upper limb	0	14	3	0	17	0.39
Q 72 Reduction defects of lower limb	4	11	2	0	17	0.39
Q 73 Reduction defects of unspecified limb	4	3	0	0	7	0.16
Q 74 Other congenital malformations of limb(s)	24	18	0	0	42	0.96
Q 75 Other congenital malformations of skull and face bones	7	28	19	0	54	1.23
Q 76 Congenital malformations of spine and bony thorax	11	21	3	0	35	0.80
Q 77 Osteochondrodysplasia with defects of growth of tubular bones and spine	0	1	7	0	8	0.18
Q 78 Other osteochondrodysplasias	0	3	28	0	31	0.71
Q 79 Congenital malformations of the musculoskeletal system, not elsewhere classified	178	47	4	3	232	5.29
<b>Others</b>	<b>193</b>	<b>581</b>	<b>213</b>	<b>0</b>	<b>987</b>	<b>22.49</b>
Q 80 Congenital ichthyosis	0	1	0	0	1	0.02
Q 82 Other congenital malformations of skin	0	13	4	0	17	0.39
Q 83 Congenital Malformations of Breast	1	1	0	0	2	0.05
Q 84 Other congenital malformations of integument	1	1	0	0	2	0.05
Q 85 Phakomatoses, not elsewhere classified	1	0	70	0	71	1.62
Q 87 Other specified congenital malformation syndromes affecting multiple systems	0	11	99	0	110	2.51
Q 89 Other congenital malformations, not elsewhere classified	181	547	1	0	729	16.61
P 2.8 Newborn affected by other and unspecified abnormalities of membranes	9	7	0	0	16	0.36
P 35.0 Congenital rubella syndrome	0	0	39	0	39	0.89
<b>Chromosomal abnormalities</b>	<b>0</b>	<b>0</b>	<b>0</b>	<b>300</b>	<b>300</b>	<b>6.83</b>
Q 90 Down Syndrome	0	0	0	262	262	5.97
Q 91 Edwards and Patau Syndrome	0	0	0	28	28	0.64
Q 92 Other trisomies and partial trisomies of the autosomes, not elsewhere classified	0	0	0	1	1	0.02
Q 93 Monosomies and deletions from the autosomes, not elsewhere classified	0	0	0	1	1	0.02
Q 96 Turner Syndrome	0	0	0	4	4	0.09
Q 97 Other sex chromosome abnormalities, female phenotype, not elsewhere classified	0	0	0	2	2	0.05
Q 99 Other chromosomal abnormalities, not elsewhere classified	0	0	0	2	2	0.05
<b>TOTAL DEFECTS</b>	<b>6917</b>	<b>3178</b>	<b>382</b>	<b>585</b>	<b>11062</b>	
<b>TOTAL CASES</b>	<b>6917</b>	<b>1206</b>	<b>263</b>	<b>300</b>	<b>8686</b>	

**Table 3.** Top 10 Birth Defects at the Philippine General Hospital from 2001-2010

ICD Code	Diagnosis	Isolated defect/ Sequence	Associated defect in MMC	Part of recognizable syndromes	Part of chromosomal syndromes	Total	Occurrence per 10,000 admissions
Q 37	Cleft palate with cleft lip	889	79	4	3	975	22.21
Q 42.2 Q 42.3	Imperforate anus	520	127	1	50	698	15.90
Q 25.0	Patent ductus arteriosus	414	185	16	57	672	15.31
Q 43.1	Hirschprung disease	568	10	0	4	582	13.26
Q 21.0	Ventricular septal defect	267	255	6	48	576	13.12
Q 03	Congenital hydrocephalus	262	85	0	0	347	7.91
Q 54	Hypospadias	241	99	0	0	340	7.75
Q 36	Cleft lip	293	22	0	1	316	7.20
Q 01	Encephalocoele	247	55	1	0	303	6.90
Q 21.1	Atrial septal defect	135	129	3	30	297	6.77

**Table 4.** Patients with Birth Defects Admitted at Philippine General Hospital from 2001-2010 Classified according to Region of Residence

Region	Number of Admissions per Region	Number of Patients with Birth Defects	Percent of patients with birth defects among patients from the region
<b>LUZON</b>			
CAR - Cordillera Administrative Region	571	17	3.0
NCR - National Capital Region	293,700	4,042	1.4
Region I - Ilocos Region	3141	116	3.7
Region II - Cagayan Valley	2484	86	3.5
Region III - Central Luzon	22059	845	3.8
Region IVA - CALABARZON	107,791 <sup>a</sup>	2,827	2.6
Region IVB - MIMAROPA	See (a)	111	0.1
Region V - Bicol Region	4,007	161	4.0
<b>VISAYAS</b>			
Region VI - Western Visayas	1,575	131	8.3
Region VII - Central Visayas	419	21	5.0
Region VIII - Eastern Visayas	1,454	39	2.7
<b>MINDANAO</b>			
Region IX - Zamboanga Peninsula	423	15	3.5
Region X - Northern Mindanao	635	19	3.0
Region XI - Davao Region	281	12	4.3
Region XII - SOCCSKSARGEN	283	24	8.5
Region XIII - Caraga Region	83 <sup>b</sup>	8	1.5 <sup>d</sup>
ARMM - Autonomous Region of Muslim Mindanao	38 <sup>c</sup>	5	1.2 <sup>e</sup>
Unspecified		207	0.05 <sup>f</sup>

<sup>a</sup> No separate data on the number of admissions from Region IV-A and Region IV-B

<sup>b</sup> Data from 2005-2010. Provinces of the Caraga Region were previously part of Region X (Northern Mindanao)

<sup>c</sup> Data from 2005-2010. Provinces of ARMM were previously part of Region IX (Davao Peninsula) and Region XII (SOCCSKSARGEN)

<sup>d</sup> Denominator used was the sum of total number of admissions for Region X from 2001 to 2004 and total admissions from the region from 2005-2010.

<sup>e</sup> Denominator used was the sum of the total number of admissions for Regions IX and X from 2001 to 2004 and total admissions from the region from 2005 to 2010.

<sup>f</sup> Percent = Number of Defects unspecified according to the number of birth defects/Total number of birth defects

NCR and CALABARZON have 1.4% and 2.6% cases of birth defects, respectively, based on the number of admissions per region. Regions VI and XII had higher percentages of birth defects, 8.3% and 8.5%, respectively based on the number of admissions per region. This could be partly explained by the presence of another 10 government hospitals within NCR that can handle the specialized cases of neurosurgery, pediatric surgery and thoracovascular surgery. Thus, the numbers reflected in this paper could just be a share of PGH in the number of patients in NCR.

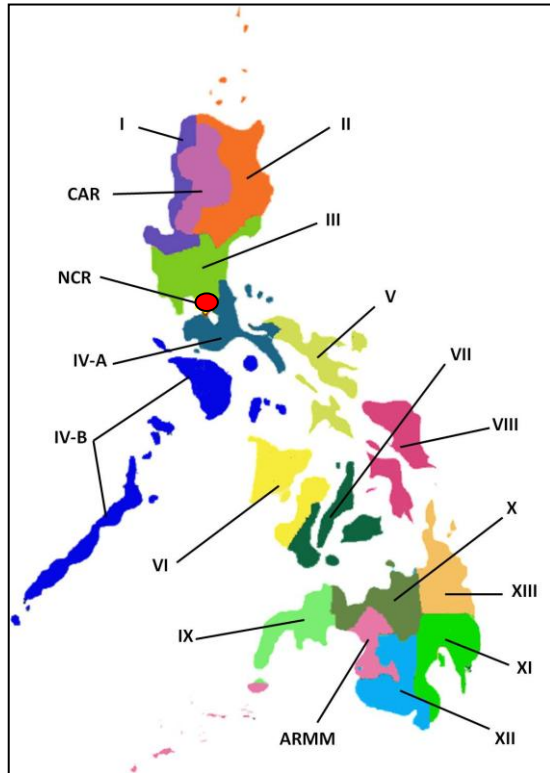
Table 5 shows the distribution of birth defect cases according to age group and organ system. The highest percentage of birth defects are from the <1 age group (38.5%), followed by the 1 to 4 age group (27.5%), and the 5 to 9 year old age group (11.2%). As previously shown, the most common birth defects are digestive system, cardiovascular system and nervous system anomalies. In the first two age groups, namely, <1 and 1 to 4 age groups, digestive system anomalies are the most common, followed by cardiovascular and nervous system anomalies. In the 5 to 9 age group, digestive system anomalies are the most common, followed by cardiovascular system, and genital

**Table 5. Distribution of Birth Defect Cases Admitted at Philippine General Hospital from 2001-2010 Categorized by Age Group**

ICD Category	Age Group												Total
	<1	1 to 4	5 to 9	10 to 14	15 to 19	20 to 24	25 to 34	35 to 44	>44	Un-specified	Cases	Percent (%)*	
	Cases	Cases	Cases	Cases	Cases	Cases	Cases	Cases	Cases	Cases	Cases	Percent (%)*	
<b>Digestive System</b>	1314	1091	341	137	100	52	43	18	18	13	3127	36.0	
	39.3	45.6	35.1	23.1	28.7	19.4	12.3	9.7	8.7	48.1			
<b>Cardiovascular System</b>	595	387	231	176	105	111	148	74	65	5	1897	21.8	
	17.8	16.2	23.8	29.6	30.1	41.4	42.3	40.0	31.4	18.5			
<b>Nervous system</b>	474	222	57	25	22	9	9	8	9	1	836	9.6	
	14.2	9.3	5.9	4.2	6.3	3.4	2.6	4.3	4.3	3.7			
<b>Genital Organs</b>	21	168	105	137	44	29	52	20	24	0	600	6.9	
	0.6	7.0	10.8	23.1	12.6	10.8	14.9	10.8	11.6	0.0			
<b>Musculoskeletal System</b>	214	102	44	22	22	14	11	5	8	1	443	5.1	
	6.4	4.3	4.5	3.7	6.3	5.2	3.1	2.7	3.9	3.7			
<b>Eye, ear, face and neck</b>	76	90	66	26	10	14	22	6	0	1	311	3.6	
	2.3	3.8	6.8	4.4	2.9	5.2	6.3	3.2	0.0	3.7			
<b>Chromosomal abnormalities</b>	170	91	14	9	4	2	4	0	2	1	297	3.4	
	5.1	3.8	1.4	1.5	1.1	0.7	1.1	0.0	1.0	3.7			
<b>Urinary System</b>	33	23	20	6	2	3	20	19	40	0	166	1.9	
	1.0	1.0	2.1	1.0	0.6	1.1	5.7	10.3	19.3	0.0			
<b>Respiratory System</b>	19	23	8	3	1	3	2	1	0	0	60	0.7	
	0.6	1.0	0.8	0.5	0.3	1.1	0.6	0.5	0.0	0.0			
<b>Multiple Congenital Anomalies</b>	307	97	40	20	8	5	7	1	2	0	487	5.6	
	9.2	4.1	4.1	3.4	2.3	1.9	2.0	0.5	1.0	0.0			
<b>Others</b>	121	96	46	33	31	26	32	33	39	5	462	5.3	
	3.6	4.0	4.7	5.6	8.9	9.7	9.1	17.8	18.8	18.5			
<b>TOTAL</b>	<b>3344</b>	<b>2390</b>	<b>972</b>	<b>594</b>	<b>349</b>	<b>268</b>	<b>350</b>	<b>185</b>	<b>207</b>	<b>27</b>	<b>8686</b>	<b>100</b>	
	100	100	100	100	100	100	100	100	100	100			

\* Percent=Number of cases/Total number of birth defects cases per age-group

organ anomalies. In the 10 to 14, 20 to 24, and 25 to 34 age groups, the most common anomalies are cardiovascular, followed by digestive or genital organ anomalies. In the 35 to 44 and >44 age groups, the most common anomalies are cardiovascular, followed by genital or urinary system anomalies.



**Figure 2.** Map of the Philippines showing the different Centers for Health Development or DOH Regional Centers – CHD I, CAR, CHD II, CHD III, NCR, CHD IVA, CHD IVB, CHD V, CHD VI, CHD VII, CHD VIII, CHD IX, CHD X, CHD XI, CHD XII, CHD XIII, and ARMM. The location of Philippine General Hospital is highlighted by the red dot.

### Discussion

Birth defects are a global problem but their impact is particularly severe in LMICs where the proportion of birth defects is much higher. This is due to sharp differences in maternal health, poverty, a high percentage of older mothers, a greater frequency of consanguineous marriages, and a lack of comprehensive health services needed to prevent birth defects.<sup>2</sup> Although consanguineous marriages are rare, the other factors are relevant to the Philippine setting. Therefore, just like other LMICs, primary health care, preconception care programs and strategies must be developed to improve the health of women, mothers, newborn and children for effective prevention and care of those with birth defects.

The March of Dimes, USA has involved the Philippines in the Global Network for Maternal and Infant Health

(GNMIH), whose goal is to reduce mortality and disability from birth defects and preterm births in lower-income countries. Together with China and Lebanon, the Philippines embarked on birth defects surveillance projects to identify and help the affected patients.<sup>10</sup> In the Philippines, activities under GNMIH include a prospective birth defects surveillance, prevention of birth defects campaigns, engaging the youth in GNMIH activities and preconception activities for the prevention of birth defects.<sup>9</sup> A retrospective study on birth defects at the Philippine General Hospital was conducted as an initiative to determine the occurrence of birth defects in a tertiary hospital. Simultaneously, a prospective Birth Defects Surveillance (BDS) is being undertaken in partnership with the DOH.<sup>8</sup>

Majority (79% or 6,917/8,686) of the patients in this study have isolated birth defects, while 13.9% (1206/8,686) are multi-malformed cases. Patients with defects that are part of recognizable syndromes comprise 3% (263/8,686) of the total cases while 3.4% (300/8,686) are patients with defects as part of chromosomal syndromes. The most common birth defect cases admitted at the PGH are classified under digestive system, followed by cardiovascular system, nervous system anomalies, and genital organ anomalies. These results show trends that are similar to some of the program reports of the International Clearinghouse for Birth Defects and Surveillance and Research (ICBDSR), i.e. the California Birth Defects Monitoring Program. It is however difficult to make straight comparisons because the ICBDSR programs report livebirths, stillbirths and termination of pregnancies whereas this study includes patients admitted in a tertiary hospital and covers all ages.<sup>11,12</sup>

The most common birth defect cases per 10,000 admissions are digestive system anomalies, which include cleft lip and palate (22.21), imperforate anus (15.90), and Hirschsprung disease (13.26); and cardiovascular system anomalies namely, patent ductus arteriosus (15.31) and ventricular septal defect (13.12). Japan and India are the two Asian countries included in the ICBDSR. In the 2010 Annual Report of the ICBDSR, the top five congenital anomalies per 10,000 population from 2004 to 2008 for both countries included cleft lip with or without cleft palate with rates of 8.22 and 21.09 for India and Japan, respectively. In a recent study from the birth defects surveillance in China, the top five congenital birth defects for 2009 also revealed cleft lip with or without cleft palate (13.17).<sup>13</sup>

Oral clefts are common birth defects of complex genetic and environmental etiology. Depending on geographic ancestry, it affects about 1 in 500 (Asian or Amerindian ancestry) to 2,500 births. Low socioeconomic status has been reported to increase the risk of oral clefts. There is some suggestive evidence for a possible role of folic acid in prevention of oro-facial clefts but studies have provided mixed results as regards to whether folic acid can prevent its



primary occurrence.<sup>14</sup> In the Philippine BDS, cleft lip and/or palate rank as the most common birth defect in the country with an occurrence of 1 in 1,164 live births, with a *cleft lip to cleft lip and palate to cleft palate* (CL:CLP:CP) ratio of 2.0:4.8:1.0.<sup>15</sup> A review of cases ascertained through Philippine surgical cleft missions revealed an incidence of 1 in 2,367 livebirths with a CL:CLP:CP ratio of 1.4:3.2:1.0.<sup>16</sup> Another study by Murray et al. reported an incidence of 1:500 using records from an 8-year period at a government hospital in Negros, the Corazon Locsin Montelibano Memorial Regional Hospital, revealing a 1:6.3 ratio for isolated cleft palate vs. cleft lip and palate.<sup>17</sup> The combined cleft lip and palate occur 3 to 4 times more than the isolated cleft lip or cleft palate. These findings are in contrast to the report of Fogh-Anderson with a ratio of 1:2:1, regarded as the normal ratio for different types of cleft.<sup>18</sup> Future studies determining the reasons for these differences in ratio for combined versus isolated oral clefts may help in elucidating possible etiologies.

Literature reveals that the international incidence of Hirschsprung disease and anorectal malformations (including imperforate anus) is 1/5000 live births.<sup>19,20</sup> The study by Torps revealed that the incidence varies according to ethnicity with 1.5, 2.1, and 2.8 per 10,000 live births in Caucasians, African Americans, and Asians.<sup>19</sup> This could explain the high number of cases in PGH.

The most common birth defect cases under nervous system anomalies are congenital hydrocephalus, encephalocele and spina bifida. The study results show more cases of encephalocele (303 cases) as compared to spina bifida (193 cases). In contrast, international trends show a significantly higher number of cases of spina bifida in relation to encephalocele.<sup>21</sup> Also, there were only three cases of anencephaly. It is presumed that most of the cases are stillbirths and are not admitted to the nursery. Except for the low number of anencephaly cases, the other nervous system anomalies are consistent with the findings of ICBDSR.

Another interesting result is the presence of 39 cases (0.89 per 10,000 admissions) of congenital rubella syndrome (CRS). CRS has been an important cause of severe birth defects since 1941 when Rubella virus was considered a teratogen by the World Health Organization.<sup>22,23</sup> CRS is a multi-systemic disease characterized by a combination of different birth defects in the context of a maternal rubella infection during early pregnancy.<sup>24</sup> These birth defects may include: sensorineural deafness which can progress after birth; ocular abnormalities such as cataract, retinopathy or glaucoma; cardiovascular defects such as patent ductus arteriosus, pulmonary artery stenosis or septal defects; and brain damage causing mild to severe mental retardation and spastic diplegia.<sup>25</sup> It is one of the preventable birth defect syndromes, through simple rubella vaccination of women in the childbearing age.<sup>23</sup> Despite the DOH's introduction

of the rubella vaccine in 1983, CRS has not been eradicated yet.<sup>26</sup>

A large number of cases (547) under Q89 Other congenital anomalies, not elsewhere classified (Table 2) may be due to the way the cases were ascertained and recorded in the chart. Many of these cases will probably fall under other categories such as in multiple congenital malformation (Q89.7).

The most common anomalies in the less than 5 years old age group are digestive followed by cardiovascular and nervous system anomalies. This may be due to the presence of symptoms evident in the newborn period, warranting hospital admission for immediate medical or surgical intervention. In the 5 to 9 age group, digestive system anomalies are the most common, followed by cardiovascular system, and genital organ anomalies. In the 10 to 14, 20 to 24, and 25 to 34 age groups, the most common anomalies are cardiovascular, followed by digestive or genital organ anomalies. In the 35 to 44 and >44 age groups, the most common anomalies are cardiovascular, followed by genital or urinary system anomalies. An increase in the numbers of cardiovascular, genital organ, and urinary system anomalies is noted with increasing age. This is probably because a significant percentage of these anomalies may not be readily apparent and symptomatic in the younger years and are detected later in life.

The PGH, being the largest government tertiary referral center in the country, is a rich source of information that can be used in exploring the occurrence patterns of birth defects. Although data generated cannot be compared with other birth defects surveillance programs that specifically monitor live births, the information from the patients of PGH will still be useful for planning. As shown in Table 4, all regions send patients to PGH for management of cases that cannot be handled by their hospitals. PGH has the biggest number of specialists and sub-specialists in the country.

The geographic proximity of NCR and CALABARZON (CHD IVA) regions to PGH makes the hospital accessible to patients coming from these areas, explaining why majority of the admitted cases come from these regions. In this study, there were 191/8,686 (2.2%) birth defect cases from the Visayas and 83/8,686 (0.9%) from Mindanao suggesting that the patients needed the specialized care of PGH.

This retrospective study has demonstrated an observable general decline in the number of admitted patients with birth defects from 2001-2010, most noticeable from 2008 to 2010 as shown in Table 1 and Figure 1. There are several possible explanations. Some patients are not able to come to seek consultation for financial reasons. Some patients could be seeking consultation in other regional hospitals. The implementation of the prospective BDS in 2008 in 82 hospitals and 18 community sites in different parts of the country could partly be contributory. The prospective BDS allows prompt diagnosis and referral to

nearby tertiary hospitals using the strategy of a sentinel site. A sentinel site is a group of hospitals and communities that are geographically linked. Every sentinel site has at least one tertiary government hospital, at least one tertiary private hospital, local government hospital/s (provincial, district and/or municipal/city hospital), and community-based health facilities (rural and urban). A tertiary hospital (private or government) provides clinical care and management on specialized and sub-specialized forms of treatment, surgical procedure and intensive care.<sup>8</sup>

Registries are important for planning. Considering the volume of patients at PGH, indeed, it is a rich source of information that can be used for program planning with outcome recommendations that can eventually be applied on a national scale.

### Conclusion

The results of this study showed that the most common birth defects are digestive, cardiovascular, nervous system, and genital organ anomalies. This trend is similar to those reported internationally.

Further studies must be done to look deeper into birth defects prevalence and causation in the Philippines. Considering the volume of patients at PGH, the findings of the study can be the basis of policies toward the development and implementation of practical strategies for primary and secondary prevention of birth defects among Filipinos.

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