Study of Selected Congenital Anomalies in Pennsylvania

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A STUDY of selected types of congenital anomalies was undertaken by the Pennsylvania Department of Health. The five specific congenital defects selected for study—imperforate anus, omphalocele, tracheo-esophageal fistula, diaphragmatic hernia, and intestinal obstruction—were chosen because they are usually discernible at birth and require immediate surgery or care to prevent the death of the newborn. Because these anomalies are more readily observed than less obvious defects, such as congenital defects of the circulatory system, they should be reported more completely on birth certificates and hospital records. We would thus be able to obtain more accurate counts of the number of these defects, their characteristics, and their frequency of occurrence.

By collecting data on these five congenital anomalies using birth and death certificates and hospital records, we could compute the frequency and rate of occurrence by type for use in program planning, measure the degree of reporting and accuracy of the birth certificate, and compile the characteristics of the newborn and his mother.

Methodology

The initial step in the project was to collect the basic data by reviewing the birth certificate of each infant born in Pennsylvania in 1962 to ascertain whether one or more of the congenital anomalies selected for study was noted. Death certificates of babies born in 1962 also were reviewed for mention of these anomalies. The birth and death certificates were matched by name to provide an unduplicated count of the anomalies. Birth certificates were obtained for any unmatched infant death, and a review of the death certificate file was made for all unmatched births. These matched certificates gave the background for the study plus the frequency with which these congenital anomalies were reported on birth and death certificates.

A congenital anomalies questionnaire and instructions for its completion were mailed to all hospitals in Pennsylvania that reported births in 1962. Identifying information (name of child, mother's name and address, date of birth, sex, race, type of congenital anomaly) was requested for all babies born in that hospital in 1962 with one or more of the five congenital anomalies under study. The instruction sheet included a listing of the code numbers and inclusions according to the "Standard Nomenclature of Diseases and Operations" for the five anomalies. Letter followups and telephone calls were made to hospitals not returning the

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Table 1. Number and rate of selected congenital anomalies, Pennsylvania, 1962

Congenital anomaly	Number	Rate [1]
Total	235	10.9
Imperforate anus	66	3.1
Omphalocele	39	1.8
Tracheo-esophageal fistula	40	1.9
Diaphragmatic hernia	18	.8
Intestinal obstruction	72	3.3

[1] Per 10,000 live births occurring in reporting hospitals.

original questionnaire. Each returned questionnaire was matched by name with the previously matched birth-death file. Questionnaires for which we had no birth or death certificate were set against the complete 1962 birth file and infant death file to obtain those certificates on which there was no reported congenital anomaly of the type included in the study.

With the matching of the questionnaires and birth and death certificates, all the basic history of the newborn was obtained. Tabulation of this file of unduplicated counts of anomalies would give an accurate estimate of the frequency of each type of congenital defect plus characteristics of the newborn and his mother.

Results

The questionnaires were mailed to 249 hospitals in Pennsylvania, all of which reported live births during the 1962 calendar year. Questionnaires were returned by 236 hospitals (94.8 percent). The hospitals either completed the form or stated that no babies with these malformations had been born there during 1962. A total of 225,490 live births occurred in Pennsylvania in 1962 in the 249 hospitals originally queried, while 216,005 births occurred in the 236 hospitals responding to the questionnaire. Thus, 95.8 percent of all hospital births are included in this study. In Pennsylvania more than 99 percent of all births occur in hospitals.

An unduplicated count of the five selected congenital anomalies showed 235 occurrences, a rate of 10.9 per 10,000 live births. Table 1 gives the number and rate for each specific type of congenital defect. Based on these data, we can estimate that one congenital defect of these types will be found in every 1,000 live births. In 10,000 live births, we would find approximately three imperforate anus, three intestinal obstructions, two omphaloceles, two tracheo-esophageal fistulas, and about one diaphragmatic hernia.

Three documents were used to determine the total or unduplicated count of the congenital anomalies found in this study. Table 2 shows a breakdown of the number and percent of each specific type as recorded from the birth certificate, death certificate, or hospital questionnaire. Of the 235 birth defects found, 129 were reported on birth certificates and 109 on death certificates. If the questionnaire were used as the only reporting instrument, 206 (87.7 percent) of the total congenital anomalies found would have been noted. Those reported in the questionnaire from the hospitals when compared to the total enumerated congenital de-

Table 2. Number and percent of selected congenital anomalies as recorded on birth and death certificates and hospital questionnaire, Pennsylvania, 1962

Congenital anomaly	Unduplicated count	Birth certificates		Death certificates		Questionnaire	
		Number	Percent [1]	Number	Percent [1]	Number	Percent [1]
Total	235	129	54.9	109	46.4	206	87.7
Imperforate anus	66	44	66.7	18	27.3	59	89.4
Omphalocele	39	30	76.9	17	43.6	37	94.9
Tracheo-esophageal fistula	40	21	52.5	24	60.0	34	85.0
Diaphragmatic hernia	18	11	61.1	13	72.2	15	83.3
Intestinal obstruction	72	23	31.9	37	51.4	61	84.7

[1] Based on unduplicated count of congenital anomalies of each type.

fects showed little variance by type ranging from a high of 94.9 percent for imperforate anus to a low of 83.3 percent for diaphragmatic hernia. Those anomalies reported on birth certificates showed the highest percentage in the most discernible malformations, omphalocele and imperforate anus, and the lowest percent for intestinal obstructions. The greatest number of congenital defects reported on death certificates were those most likely to result in the death of the newborn—diaphragmatic hernia and tracheo-esophageal fistula.

Table 3 shows the frequency distribution of each specific type of congenital anomaly by the age of mother at time of birth of the infant, while table 4 shows the birth order of the combined congenital defects distributed by age of mother. The distribution of the congenital anomalies by age of mother (table 3) was not significantly different from what would be expected based on the distribution of total births by age of mother. The distribution by birth order (table 4) also did not differ significantly from the birth order distribution of all births. Although the difference was not statistically significant, there were more first- and second-born babies than would be expected. In first-born infants, imperforate anus and diaphragmatic hernia occurred more frequently, while the second borns had more intestinal obstructions. More babies were born with an omphalocele than expected when the birth order was greater than four.

The total number of congenital anomalies found in the study is distributed by sex and weight at birth in table 5. The ratio of males to females born with these congenital defects was 1.61 : 1. The ratio of males to females for all births in Pennsylvania in 1962 was 1.05 : 1. The distribution of birth weight of the babies born with these congenital anomalies differed significantly from the birth weight distribution based on total births ($P=0.01$). The difference was also significant for the weight at birth of males and females. Approximately 32 percent (76) of the babies born with the congenital

Table 3. Number of infants born with selected congenital anomalies by age of mother, Pennsylvania, 1962

Age group (years)	Total	Imperforate anus	Omphalocele	Tracheo-esophageal fistula	Diaphragmatic hernia	Intestinal obstruction
All ages	235	66	39	40	18	72
Under 20	32	12	6	4	1	9
20–24	66	14	18	8	7	19
25–29	68	19	7	16	5	21
30–34	38	12	3	6	1	16
35–39	21	6	3	3	3	6
Over 39	10	3	2	3	1	1

Table 4. Number of infants with selected congenital anomalies, birth order by age of mother, Pennsylvania, 1962

Age of mother (years)	Total	Number of previous births					
		0	1	2	3	4	More than 4
All ages	235	70	63	41	22	18	21
Under 20	32	23	6	3	0	0	0
20–24	66	28	21	7	7	1	2
25–29	68	14	23	14	6	4	7
30–34	38	5	7	10	7	7	2
35–39	21	0	3	7	0	4	7
Over 39	10	0	3	0	2	2	3

Table 5. Selected congenital anomalies by sex and birth weight, Pennsylvania, 1962

Birth weights (grams)	Total	Male	Female
All weights	235	145	90
Less than 1,500	7	5	2
1,501–2,500	69	45	24
2,501–3,500	120	64	56
3,501–4,500	39	31	8
More than 4,500	0	0	0

Table 6. Number of selected congenital anomalies by sex, race, and specific type, Pennsylvania, 1962

Congenital anomaly	Total cases	White	Nonwhite
Both sexes	235	216	19
Imperforate anus	66	61	5
Omphalocele	39	35	4
Tracheo-esophageal fistula	40	40	0
Diaphragmatic hernia	18	17	1
Intestinal obstruction	72	63	9
Male	145	137	8
Imperforate anus	46	45	1
Omphalocele	27	25	2
Tracheo-esophageal fistula	25	25	0
Diaphragmatic hernia	7	7	0
Intestinal obstruction	40	35	5
Female	90	79	11
Imperforate anus	20	16	4
Omphalocele	12	10	2
Tracheo-esophageal fistula	15	15	0
Diaphragmatic hernia	11	10	1
Intestinal obstruction	32	28	4

anomalies included in the study weighed 2,500 grams or less at birth. In comparison only about 8 percent of all infants born are in this weight group. Seven times more babies are born with an omphalocele who weigh under 2,501 grams than would be expected if these infants were distributed by the birth weight of the total birth cohort. This ratio of 7 to 1 for omphaloceles is followed by tracheo-esophageal fistulas (5:1), intestinal obstructions (3.5:1), and imperforate anus (3:1). There is no substantial difference for those born with diaphragmatic hernia.

Table 6 depicts the sex and race distribution for each type of congenital anomaly in the study. In Pennsylvania in 1962, 88.4 percent of the infants born were white, while in the survey 91.9 percent were white. The remaining 8.1 percent in the survey were classified as nonwhite as compared to 11.6 percent for all births. In the nonwhite group the number of females born with these congenital anomalies was greater than the number of males, and almost half of the nonwhite infants were born with intestinal obstruction.

The percentage of newborns who died before completing the first year of life is shown for each type of congenital defect by sex in table 7. The highest percentage or case fatality rate was for those babies born with diaphragmatic hernias, of whom almost three of every four died before reaching age 1. While the overall case fatality rate for males and females was the same, this did not hold true for each specific type of congenital malformation. The highest percentage of male deaths occurred in those born with tracheo-esophageal fistulas and for females, in those born with diaphragmatic hernias.

Summary

Two hundred and thirty-five cases of imperforate anus, omphaloceles, tracheo-esophageal fistulas, diaphragmatic hernias, and intestinal obstructions were found in a study using hospital questionnaires, birth certificates, and

Table 7. Number and percent of infants dying before age 1 by sex and specific congenital anomaly, Pennsylvania, 1962

Congenital anomaly	Total cases	Deaths	
		Number	Percent
Both sexes	235	122	51.9
Imperforate anus	66	20	30.3
Omphalocele	39	25	64.1
Tracheo-esophageal fistula	40	26	65.0
Diaphragmatic hernia	18	13	72.2
Intestinal obstruction	72	38	52.8
Male	145	75	51.7
Imperforate anus	46	15	32.6
Omphalocele	27	19	70.4
Tracheo-esophageal fistula	25	16	76.2
Diaphragmatic hernia	7	4	57.1
Intestinal obstruction	40	21	52.5
Female	90	47	52.2
Imperforate anus	20	5	25.0
Omphalocele	12	6	50.0
Tracheo-esophageal fistula	15	10	66.7
Diaphragmatic hernia	11	9	81.8
Intestinal obstruction	32	17	53.1

death certificates. These congenital anomalies occurred in 216,005 infants born in Pennsylvania during 1962. It was estimated that about one congenital defect of these types would be found in each 1,000 births.

Two hundred and six (87.7 percent) of these malformations were reported on the questionnaires, and 129 (55 percent) on birth certificates. The majority of the anomalies reported on the birth certificate were imperforate anus and omphaloceles. Of the 122 infant deaths in the study, 109 (89.3 percent) were reported on death certificates as caused by these congenital anomalies. All 13 newborns who died from diaphragmatic hernias were reported on death certificates.

The distribution of newborns with these congenital defects by weight at birth was significantly different from the birth weight distribution of all births. Approximately 32 percent (76) of the babies born with the congenital anomalies included in the study weighed 2,500 grams or less at birth as compared to about 8 percent of the total number of babies born weighing this little. The distribution by age of mother or birth order of babies born with these malformations was not significantly different. There was, however, a slight excess in the number of first-and second-born infants.

Of the 235 babies born with these congenital anomalies in 1962, more than half died before reaching age 1. The percentage of males and females dying was about the same. Of the 18 babies born with diaphragmatic hernias, 13 (72.2 percent) died.

Conclusions

While it is not possible to say that this survey discovered all the babies born in Pennsylvania in 1962 with imperforate anus, omphaloceles, tracheo-esophageal fistulas, diaphragmatic hernias, and intestinal obstructions, the methodology used enabled us to obtain a reasonably accurate count of the number born out of the 216,005 births included in the study. By using extrapolation we could estimate that a total 246 babies were born with these congenital anomalies.

This study verifies the fact that congenital malformations are not reported completely and accurately. When only 32 to 77 percent of such discernible and fatal congenital defects as tabulated in this study are reported on birth certificates, there is a need for more complete early observance of the defect, or a better method of getting known information onto the birth record, or both.

International Reference Center for Comparative Oncology

The World Health Organization has designated the Armed Forces Institute of Pathology as the International Reference Center for Comparative Oncology, to work in collaboration with the Registry of Comparative Pathology. Facilities will be available on a limited basis for advanced study, training, and research.

An important aim of the program is to establish a classification and nomenclature for animal tumors as a basis for epidemiologic studies and to bring out similarities and differences between species, including man. Work will focus initially on spontaneous tumors of the six most common domestic mammals (horses, oxen, sheep, pigs, dogs, and cats) and nonhuman primates, but other species may eventually be included. The center will also prepare a yearly bibliography of world literature on comparative oncology.

Further information about the center is available from the Director, Armed Forces Institute of Pathology, Washington, D.C. 20305.