APORS BIRTH DEFECT CASE FINDING STUDY – 2018 BIRTHS

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Adverse Pregnancy Outcomes Reporting System

PURPOSE

The Illinois Department of Public Health's (IDPH's) Adverse Pregnancy Outcomes Reporting System (APORS) collects information about Illinois infants born with birth defects or other abnormal conditions. APORS was established to identify children who require special services to correct or prevent developmental problems and to refer the children for case-management and other services. In addition, APORS conducts surveillance on birth defects and other adverse pregnancy outcomes to monitor the health of Illinois children and to provide information to guide public health policy.

Children who have an APORS condition (Appendix A) reported by a hospital are offered case management services through the Illinois Department of Human Services' High Risk Infant Follow-up Program (HRIF). The goal of the program is to assure children reach their full potential by identifying, as early as possible, conditions requiring further evaluation, diagnosis, and treatment and by facilitating environments that promote optimal growth and development.

Hospitals affiliated with the Illinois Perinatal Network are required to report any child born with a defect or other selected adverse pregnancy outcome to the APORS program at discharge from the newborn hospital stay. In addition, APORS receives bi-annual hospital discharge data from the IDPH Division of Patient Safety and Quality. This includes birth defects coded on any discharge records from Illinois hospitals for children less than 2 years old. APORS also has begun receiving reports of genetic and prenatal diagnoses from maternal fetal medicine

programs. Children identified from hospital discharge data are not referred for services since the reports are usually not available until children are at least 1 year old, and the discharge diagnoses are coded for billing purposes and include conditions that may have been subsequently ruled out.

Four abstractors reviewed the charts of children born in 2018 and identified from any data source with selected birth defects that are relatively common and significant in their impact on the child, chosen in collaboration with the National Birth Defect Prevention Network (NBDPN). A list of the conditions included in 2018 is given in Appendix B.

This study is completed to assess whether the APORS program is missing cases and the extent of work needed for APORS staff to identify any such cases. APORS staff also uses the study information to identify hospitals that need additional training in APORS reporting.

METHODS

Seventeen hospitals distributed around the state and providing various levels of care were selected (Table 1). They were among hospitals that reported the lowest percentage of their birth cases (as determined in the APORS QC report *Rates of Hospital Reporting of Adverse Pregnancy Outcomes in 2017)*.

A list of children with in-patient or out-patient visits at one of the hospitals in Table 1 and were under 2 years of age at the time of the visit were identified from the hospital discharge data file. From this list, children were selected for chart review if they had at least one of the following ICD-10-CM codes that had not already been reported to APORS: Q24.8 (Other specified heart defect), Q24.9 (Congenital malformations of the heart), Q74.8 (Other specified congenital malformations of limb), Q74.9 (Unspecified congenital malformations of limb), Q17.3 (Other misshapen ear), Q17.8 (Other specified congenital malformations of ear), Q17.9 (Congenital malformation of ear, unspecified), Q66.89 (Other specified deformities of feet), Q66.90 (Congenital deformity of feet, unspecified, unspecified foot), and within the range of Q87.0 (Malformation syndromes) and Q99.9 (Chromosomal abnormalities). Abstractors reviewed the charts for any unreported diagnoses. Any children identified as APORS cases were entered into the APORS case abstraction system.

Table 1: Hospitals Selected for Inclusion

Hospital	Annual Births	Reported Cases 2017 N (%) of hospital's births	Level	Abstractor
1	4,021	276 (6.9)	3	A
2	1,709	79 (4.6)	3	A
3	2,531	90 (3.6)	3	B
4	3,142	136 (4.3)	3	A
5	1,204	38 (3.2)	3	B
6	2,966	179 (6.0)	3	B
7	3,265	201 (6.2)	3	B
8	1,729	42 (2.4)	2+	C
9	687	11 (1.6)	2+	D
10	1,189	21 (1.8)	2+	A
11	1,441	35 (2.4)	2+	B
12	1,023	21 (2.1)	2+	B
13	1,095	12 (1.1)	2	B
14	123	2 (1.6)	2	C
15	1,513	26 (1.7)	2	D
16	1,349	15 (1.1)	2	B
17	1,431	16 (1.1)	2	B

RESULTS

The APORS abstractors identified missed conditions at all but two of the selected hospitals (Table 2). There were 20 children identified with NBDPN diagnoses who were unreported by hospital nurseries or reported only as a non-specific anomaly. The most common condition was club foot (15.8%). Thirty-two additional children were discovered with non-NBDPN conditions. There were a variety of conditions found, but the most frequently identified were gene or

chromosomal anomalies and syndromes (nine) and other foot deformities (six). The families of these 32 children should have been offered case management services through HRIF.

Table 2. Unreported APORS Cases, Born in 2018 and Identified at Selected Hospitals

Hospital	Level	2018 Reported Cases	Missed Referral Diagnoses			% Unreported
			NBDPN	non-NBDPN	Total	
1	3	252	2	8	10	3.9
2	3	91	3	2	5	5.5
3	3	78	3	5	8	10.3
4	3	149	0	3	3	2.0
5	3	35	0	1	1	2.9
6	3	172	1	1	2	1.2
7	3	207	3	1	4	1.9
8	2+	29	2	1	3	10.3
9	2+	16	0	0	0	0.0
10	2+	22	0	1	1	4.5
11	2+	33	1	3	4	12.1
12	2+	16	0	0	0	0.0
13	2	14	1	0	1	7.1
14	2	17	1	1	2	11.8
15	2	48	2	0	2	4.2
16	2	5	0	4	4	8.0
17	2	10	1	1	2	2.0
Total		**1,194**	**20**	**32**	**52**	**4.4**

DISCUSSION

QC 23:03 APORS

The APORS program did not originally ascertain 52 children who should have been reported to the program. This has an impact both on the support that the families of these children are offered and on the accuracy of the surveillance data released by APORS. These hospitals are not representative of all reporting hospitals since they were selected because of the possibility that they were underreporting; therefore, the results cannot be scaled to all hospitals and may represent the worst-case scenario for hospital reporting.

Impact on Follow-up

In total, the families of 52 children with known conditions were not offered the services available to them. Among them, 58% have diagnoses that might have required specialist follow-up or surgeries after the babies were discharged home. This would be a stressful time for families and some would have appreciated the support offered through HRIF.

Impact on Surveillance

APORS does not undertake intensive surveillance of every condition reported to the program since there are thousands that babies might experience and many of them are extremely rare. Program staff put efforts into surveillance of selected birth defects in collaboration with the NBDPN. During the reviewed year, there were a total of 20 such cases not identified either at discharge from the newborn stay or through hospital discharge data. The missed cases include clubfoot (15), ray defect with bilateral missing feet (1), neonatal abstinence syndrome (3), and absent ear canal (1). These missed conditions means that the APORS program underestimated the prevalence of these conditions in 2018.

Solutions

Referrals need to be made promptly so that the baby and his or her family can receive services as early as possible. The APORS program does not have the resources to review every delivery (approximately 145,000 annually) to assure that all referrals are made. The program does use two approaches to help improve the referral rate. The APORS program is using a data system

that is linked to the birth certificate registry. Babies born prematurely or with a birth defect or certain other conditions marked on the birth certificate have a case generated automatically. This is a reminder to the hospitals to report these cases to APORS. The program also has multiple staff available to train hospitals virtually. The information from this report will be used both in educating the hospitals that were evaluated as part of this report and in other hospitals with low reporting rates. Emphasis will be placed on conditions that were frequently missed.

Birth defect surveillance data is collected later than referral data. This assures that the chart is complete when it is reviewed by APORS abstractors and allows the program to identify diagnosis that may have been made after the newborn hospital visit. The hospital discharge data is a useful data source, since it identifies multiple birth defect diagnoses missed after the nursery reporting. APORS staff could review a wider range of diagnosis codes to identify diagnoses that are often missed, such as club foot.

It would be useful in the future to implement a strategy allowing abstractors to review hospital logs on a regular basis for any unreported cases.

Illinois Department of Public Health
Conditions for APORS Hospital Nursery Reporting **APPENDIX A**

Gestational age less than 31 completed weeks (based on physician's assessment)

Multiple birth, triplets, or higher order

Infant death (before discharge from the newborn stay) ***Expiration*** after showing signs of life, including breathing, heartbeat, pulsation of the umbilical cord, or definite movement of voluntary muscles. May have a zero APGAR score. A birth certificate should be issued.

Prenatal drug exposure

Diagnosis of a positive toxicology for any drug (except marijuana or drugs administered during labor and delivery).

Signs of drug toxicity or withdrawal (in the infant).
Children of mothers who admit to illicit drug use during pregnancy (except marijuana).

Birth defect or congenital anomaly *(except as listed below)*

Congenital pigment anomalies (stork bites, Mongolian spots etc.)	*Peripheral pulmonic stenosis (PPS)*	*Skin tag*
Dacrostenosis	*Persistent fetal circulation*	*Syndactyly*
Incomplete or redundant penile foreskin	*Polydactyly*	*Tongue tie*
Isolated choroid plexus cyst	*Preauricular sinus*	*Two-vessel cord*
Isolated simian crease	*Prenatal diagnosis of hydronephrosis, caliectasis or pelviectasis*	*Umbilical hernia*
Patent ductus arteriosus (PDA)	*Sacral dimple with visualized base or post-natal imaging ruling out problem*	*Undescended testes*
Patent foraman ovale (PFO)		*Vascular harmatomas (small or insignificant birth marks, port wine stains, strawberry nevi etc.)*

Serious congenital infections *(Excludes: Hepatitis C or HIV exposure, neonatal candidiasis (thrush), conjunctivitis, dacrocystitis, infective mastitis and omphalitis, and HIV)*

Chlamydia	Hepatitis B (disease or prenatal exposure)	Rubella
Confirmed septicemia (sepsis)	Herpes	Syphilis (disease or exposure to active disease)
Cytomegalovirus	Listeriosis	Tetanus neonatorum
Gonococcal conjunctivitis neonatorium	Meningitis	
Group B streptococcus	Necrotizing enterocolitis leading to surgery	

Endocrine, metabolic, or immune disorders

Combined immunity deficiency	Hypothyroidism

Blood disorder

Coagulation defects	Constitutional aplastic anemia	Hereditary hemolytic anemia	Leukemia

Other conditions

Bronchopulmonary dysplasia	Endocardial fibroelastosis	IVH grade III or IV
Cerebral lipidoses	Erb's palsy	Neurofibromatosis
Chorioretinitis	Fetal alcohol syndrome	Occlusion of cerebral arteries
Conditions leading to ECMO	HIE leading to cooling treatment	Retinopathy of prematurity
Conditions leading to > 72 hours on a ventilator	Intrauterine growth restriction leading to SGA	Strabismus
		Seizures

Centers for Disease Control and Prevention (CDC) Birth Defects and ICD-10-CM Codes

Appendix B

Chart Review

CDC Birth Defects	ICD-9-CM Codes
Anencephalus	Q00.0 – Q00.1
Encephalocele	Q01.0 – Q01.9
Microcephaly	Q02
Holoprosencephaly	Q04.2
Spina bifida without anencephalus	Q05.0 – Q05.9, Q07.01, Q07.03
Anophthalmia/microphthalmia	Q11.0 – Q11.2
Congenital cataract	Q12.0
Anotia/microtia	Q16.0, Q17.2
Common truncus/truncus arteriosus	Q20.0
Double outlet right ventricle	Q20.1
Transposition of great arteries	Q20.3, Q20.5
Single ventricle	Q20.4
Ventricular septal defect	Q21.0
Atrial septal defect	Q21.1
Atrioventricular septal defect	Q21.2
Tetralogy of Fallot/Pentalogy of Fallot	Q21.3
Pulmonary valve atresia and stenosis	Q22.0, Q22.1
Tricuspid valve atresia and stenosis	Q22.4
Ebstein's anomaly	Q22.5
Aortic valve stenosis	Q23.0
Hypoplastic left heart syndrome	Q23.4
Coarctation of aorta	Q25.1
Interrupted aortic arch	Q25.21
Total anomalous pulmonary venous connection (TAPVC/TAPVR)	Q26.2

CDC Birth Defects	ICD-9-CM Codes
Choanal atresia	Q30.0
Cleft palate without cleft lip	Q35.1 – Q35.9
Cleft lip without cleft palate	Q36.0 – Q36.9
Cleft lip with cleft palate	Q37.0 – Q37.9
Esophageal atresia/tracheoesophageal fistula	Q39.0 – Q39.4
Small intestinal atresia/stenosis	Q41.0 – Q41.9
Rectal and large intestinal atresia/stenosis	Q42.0 – Q42.9
Biliary atresia	Q44.2 – Q44.3
Hypospadias	Q54.0 – Q54.3, Q54.8 – Q54.9
Renal agenesis/hypoplasia	Q60.0 – Q60.6
Bladder exstrophy	Q64.10, Q64.19
Cloacal exstrophy	Q64.12
Congenital posterior urethral valves	Q64.2
Clubfoot	Q66.0
Limb reduction deformity	Q71.0 – Q73.8
Craniosynostosis	Q75.0
Diaphragmatic hernia	Q79.0
Gastroschisis	Q79.3
Omphalocele	Q79.2
Down syndrome	Q90.0 – Q90.9
Trisomy 18	Q91.0 – Q91.3
Trisomy 13	Q91.4 – Q91.7
Turner Syndrome	Q96.0 – Q96.9
Deletion 22q11.2	Q93.81

Special Instructions for CDC Diagnoses

Spina bifida	Do not code in the presence of anencephaly.
Atrial Septal Defect	Excludes PFO; follow flowchart to determine when to code PFO vs ASD.
Renal agenesis/hypoplasia	Do not collect if condition is only prenatally diagnosed, except in the case of immediate demise.
Transposition of great arteries	Code the VSD as well as the complete transition. If the word 'complete' or 'D' is not used in the chart, do not use this code with an unspecified VSD.