Puerto Rico

Puerto Rico Birth Defects Surveillance and Prevention System (PR-BDSPS)

Purpose: Surveillance, Referral to Services, Referral to Prevention/Intervention Services
Partner: Local Health Departments, Hospitals, Advocacy Groups, Early Childhood Prevention Programs
Program status: Currently collecting data
Start year: 1995
Earliest year of available data: 1995
Organizational location: Department of Health (Services for Children with Special Medical Needs Division)
Population covered annually: 23,000
Statewide: Yes
Current legislation or rule: Law #351
Legislation year enacted: 2004

Case Definition
Outcomes covered: Selected birth defects covered: neural tube defects, microcephaly, holoprocencephaly, cleft lip and/or cleft palate, anotia, microtia, anophthalmia, microphthalmia, limb defects, talipes equinovarus, gastroschisis, omphalocele, craniosynostosis, diaphragmatic hernia, trisomy 13, 18 and 21, Tuner syndrome, 22q11.2 deletion syndrome, albinism, Jarcho-Levin syndrome, Prader Willi syndrome, major congenital heart defects, ambiguous genitalia, hypospadias , and bladder extrophy. Outcomes covered: live-births, stillbirths, terminations and spontaneous abortion.
Pregnancy outcome: Livebirths (All gestational ages and birth weights), Fetal deaths - stillbirths, spontaneous abortions, etc. (All gestational ages), Elective terminations (All gestational ages)
Age: Up to 6 years after delivery
Residence: In-state births to state residents

Surveillance Methods
Case ascertainment: Active Case Finding
Vital records: Birth certificates, Death certificates
Other state based registries: Programs for children with special needs, Newborn hearing screening program, Newborn metabolic screening program
Delivery hospitals: Disease index or discharge index, Discharge summaries, Obstetrics logs (i.e., labor & delivery), Regular nursery logs, ICU/NICU logs or charts, Pediatric logs, Postmortem/pathology logs, Surgery logs, Specialty outpatient clinics
Pediatric & tertiary care hospitals: Disease index or discharge index, Discharge summaries, ICU/NICU logs or charts, Pediatric logs, Postmortem/pathology logs, Surgery logs
Other specialty facilities: Prenatal diagnostic facilities (ultrasound, etc.), Cytogenetic laboratories
Other sources: Physician reports

Case Ascertainment
Conditions warranting chart review in newborn period: Any chart with an ICD-9-CM code 740-759/ICD-10-CM code Q00-Q99, Any birth certificate with a birth defect box checked, Any chart with selected defects or medical conditions (i.e. abnormal facies, congenital heart disease), All stillborn infants, All elective abortions, All neonatal deaths, All infants in NICU or special care nursery, All infants with low APGAR scores, All prenatally diagnosed or suspected cases
Conditions warranting chart review beyond the newborn period: Facial dysmorphism or abnormal facies, Cardiovascular condition, Ocular conditions, Auditory/hearing conditions, Any infant with a codable defect
Coding: ICD-9-CM/ICD-10-CM

Data Collected
Infant/fetus: Identification information (name, address, date-of-birth, etc.), Demographic information (race/ethnicity, sex, etc.), Birth measurements (weight, gestation, Apgars, etc.), Tests and procedures, Infant complications, Birth defect diagnostic information
Mother: Identification information (name, address, date-of-birth, etc.), Demographic information (race/ethnicity, sex, etc.), Gravidity/parity, Illnesses/conditions, Prenatal care, Prenatal diagnostic information, Pregnancy/delivery complications, Family history
Father: Identification information (name, address, date-of-birth, etc.), Demographic information (race/ethnicity, sex, etc.)

Data Collection Methods and Storage
Data collection: Printed abstract/report filled out by staff, Printed abstract/report submitted by other agencies (hospitals, etc.)
Database collection and storage: Access, REDCap

Data Analysis
Data analysis software: SPSS, Excel
Quality assurance: Validity checks, Re-abstraction of cases, Double-checking of assigned codes, Comparison/verification between multiple data sources, Clinical review, Timeliness
Data use and analysis: Routine statistical monitoring, Baseline rates, Rates by demographic and other variables, Monitoring outbreaks and cluster investigations, Time trends, Service delivery, Referral, Grant proposals, Education/public awareness, Prevention projects

Funding
Funding source: 67.2% MCH funds, 32.7% CDC grant

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DATA TABLES

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Birth Defects Counts and Prevalence 2014 - 2018 (Prevalence per 10,000 Live Births)

	Maternal Race/Ethnicity		
Defect	**Hispanic**	**Total***	**Notes**
Anencephalus	45 ***3.2***	45 ***3.2***	
Anophthalmia/microphthalmia	25 ***1.8***	25 ***1.8***	
Anotia/microtia	41 ***2.9***	41 ***2.9***	
Aortic valve stenosis	19 ***1.4***	19 ***1.4***	
Atrial septal defect	372 ***26.6***	372 ***26.6***	
Atrioventricular septal defect (Endocardial cushion defect)	67 ***4.8***	67 ***4.8***	1
Bladder exstrophy	3 ***0.2***	3 ***0.2***	
Cleft lip alone	34 ***2.4***	34 ***2.4***	
Cleft lip with cleft palate	82 ***5.9***	82 ***5.9***	
Cleft palate alone	89 ***6.4***	89 ***6.4***	
Clubfoot	276 ***19.7***	276 ***19.7***	
Coarctation of the aorta	49 ***3.5***	49 ***3.5***	
Common truncus (truncus arteriosus)	8 ***0.6***	8 ***0.6***	
Craniosynostosis	24 ***3.2***	24 ***3.2***	2
Deletion 22q11.2	4 ***0.3***	4 ***0.3***	
Diaphragmatic hernia	33 ***4.4***	33 ***4.4***	2
Double outlet right ventricle	31 ***2.2***	31 ***2.2***	
Ebstein anomaly	14 ***1.0***	14 ***1.0***	
Encephalocele	16 ***1.1***	16 ***1.1***	
Gastroschisis	72 ***5.1***	72 ***5.1***	
Holoprosencephaly	17 ***2.3***	17 ***2.3***	2
Hypoplastic left heart syndrome	28 ***2.0***	28 ***2.0***	
Hypospadias	417 ***57.9***	417 ***57.9***	3
Interrupted aortic arch	12 ***0.9***	12 ***0.9***	
Limb deficiencies (reduction defects)	82 ***5.9***	82 ***5.9***	
Omphalocele	54 ***3.9***	54 ***3.9***	
Pulmonary valve atresia and stenosis	140 ***10.0***	140 ***10.0***	
Pulmonary valve atresia	11 ***0.8***	11 ***0.8***	
Single ventricle	6 ***0.4***	6 ***0.4***	
Spina bifida without anencephalus	54 ***3.9***	54 ***3.9***	
Tetralogy of Fallot	56 ***4.0***	56 ***4.0***	
Total anomalous pulmonary venous connection	17 ***1.2***	17 ***1.2***	

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Birth Defects Counts and Prevalence 2014 - 2018 (Prevalence per 10,000 Live Births)

	Maternal Race/Ethnicity		
Defect	**Hispanic**	**Total***	**Notes**
Transposition of the great arteries (TGA)	44 ***3.1***	44 ***3.1***	
Dextro-transposition of great arteries (d-TGA)	19 ***1.4***	19 ***1.4***	
Tricuspid valve atresia and stenosis	16 ***1.1***	16 ***1.1***	
Tricuspid valve atresia	14 ***1.0***	14 ***1.0***	
Trisomy 13	9 ***0.6***	9 ***0.6***	
Trisomy 18	22 ***1.6***	22 ***1.6***	
Trisomy 21 (Down syndrome)	159 ***11.4***	159 ***11.4***	
Turner syndrome	7 ***1.0***	7 ***1.0***	4
Ventricular septal defect	439 ***31.4***	439 ***31.4***	5
Total live births	**140,029**	**140,029**	**6**
Male live births	**72,055**	**72,055**	
Female live births	**67,970**	**67,970**	

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Birth Defects Counts and Prevalence 2014 - 2018 (Prevalence per 10,000 Live Births)

	Maternal Age (Years)			
Defect	**Less than 35**	**35+**	**Total***	**Notes**
Gastroschisis	72	0	72	
	5.8	***0.0***	***5.1***	
Trisomy 13	5	4	9	
	0.4	***2.7***	***0.6***	
Trisomy 18	12	10	22	
	1.0	***6.7***	***1.6***	
Trisomy 21 (Down syndrome)	76	83	159	
	6.1	***55.3***	***11.4***	
Total live births	**125,024**	**15,005**	**140,029**	**6**

Notes

1. Data for this condition only include atrioventricular canal.
2. Data for this condition begin in 2016.
3. Data for this condition include male and unknown gender cases only. Prevalence is calculated per 10,000 male live births.
4. Data for this condition include female and unknown gender cases only. Prevalance is calculated per 10,000 female live births.
5. Data for this condition exclude probable diagnosis and exclude inlet/posterior type ventricular septal defect only in the presence of atrioventricular canal.
6. Data for total live births include unknown gender.

General comments

*Data for totals include unknown and/or other.