Birth Defects in Connecticut 2018-2020: A Surveillance Report on Birth Defects

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Connecticut Birth Defects Registry

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About Birth Defects

Major structural or genetic birth defects affect 1 in every 33 babies born in the United States each year[1]. These major birth defects include conditions that result from a malformation, deformation, or disruption in one or more parts of the body, a chromosomal abnormality, or a known clinical syndrome; are present at birth; and have serious, adverse effect on health, development, or functional ability[1]. Birth defects can be caused by genetic or environmental factors or by a combination of both and may result in mental and physical disabilities, economic, emotional, and social distress for individuals[2].

Connecticut Department of Public Health

The Connecticut Department of Public Health's (CT DPH) mission statement is to protect and improve the health and safety of the people of Connecticut (CT) by assuring the conditions in which people can be healthy; preventing disease, injury, and disability; and promoting the equal enjoyment of the highest attainable standard of health, which is a human right and a priority of the State.

Within CT DPH is the Community, Family Health and Prevention Section (CFHPS). This section works to improve the health of the overall population across the lifespan, especially mothers, infants, children, adolescents and other vulnerable groups, by establishing opportunities that support healthy living habits through education, injury and disease prevention, early detection, and access to care. CT Birth Defects Program is within CFHPS.

Connecticut's Birth Defects Registry

The CT Birth Defects Program has a Birth Defects Registry, which is a passive surveillance system developed in 1998 to collect information about birth defects that occur among state residents. The mission of the CT Birth Defects Program is to: 1) maintain statewide surveillance through collecting information on birth defect incidence in CT; 2) monitor trends and patterns in birth defect statistics; 3) conduct research studies to identify risk factors for birth defects; and 4) promote education activities for the prevention of birth defects.

The CT Birth Defects Registry collects information on birth defects directly from birth hospitals across the state through the Newborn Screening System. Reporting of birth defects to the Registry is mandatory under the CT State Statutes (Sec. 19a-53, 19a-54, and 19a-56a).

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[1] Centers for Disease Control and Prevention (CDC). Update on overall prevalence of major birth defects--Atlanta, Georgia, 1978-2005. MMWR Morb Mortal Wkly Rep. 2008 Jan 11;57(1):1-5. PMID: 18185492.

[2] Boyle CA, Cordero JF. Birth defects and disabilities: a public health issue for the 21st century. Am J Public Health. 2005 Nov;95(11):1884-6. doi: 10.2105/AJPH.2005.067181. Epub 2005 Sep 29. PMID: 16195507; PMCID: PMC1449452.

Along with the Birth Defects Registry data, hospital discharge records (CT DPH Hospital Discharge Records, 2018-2020) indicating a major birth defect [3] from birth to 1 year after birth for 2018 to 2020 births are included in this report to determine the prevalence of birth defects in CT. This report presents findings from the linked dataset using hospital discharge records and the Birth Defects Registry. The linked dataset represents a more complete analysis of major birth defects in CT compared to the Birth Defects Registry alone.

Confidentiality

All data collected by the CT DPH complies with state and federal privacy and confidentiality regulations.

Limitations

There are limitations to the findings in this report. A birth defects diagnosis code on a hospital discharge record does not necessarily mean there is a confirmation of diagnosis. However, utilizing findings from the Birth Defects Registry alone may be an underrepresentation of the true burden of birth defects in CT. This report presents findings from the linked dataset using hospital discharge records and the Birth Defects Registry. The linked dataset may contain cases that are not confirmed cases. A medical records review will help identify true diagnosed birth defects in CT.

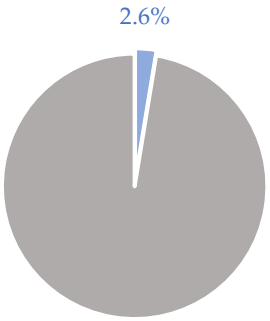
Additionally, this analysis does not include fetal or infant deaths or terminations with a birth defect diagnosis. It is possible that this analysis does not capture all diagnosed infants due to limiting data from diagnosed infants in the outpatient setting.

[3] National Birth Defects Prevention Network. Table 1: Birth defects for the National Birth Defects Prevention Network (NBDPN) annual report by disease classification codes. https://www.nbdpn.org/birth_defects_data_tables_and.php

Prevalence of Birth Defects in Connecticut

From 2018 through 2020, we identified 2,594 infants with one or more major birth defects. Given a denominator used is 99,525 live births to CT residents during this same period, there is an overall prevalence of 2.6% of major birth defects among live births in CT for 2018-2020 (Figure 1).

Figure 1: Infants With Major Birth Defects Among CT Infants, Combined 2018-2020



Most of the infants with major birth defects have one major birth defects (79.2% of 2,594 infants), 13.6% had two major birth defects, 4.5% had three major birth defects, and 2.7% had four or more major birth defects (Figure 2).

Figure 2: Distribution of Number of Major Birth Defects Among CT Infants with a Major Birth Defect, Combined 2018-2020

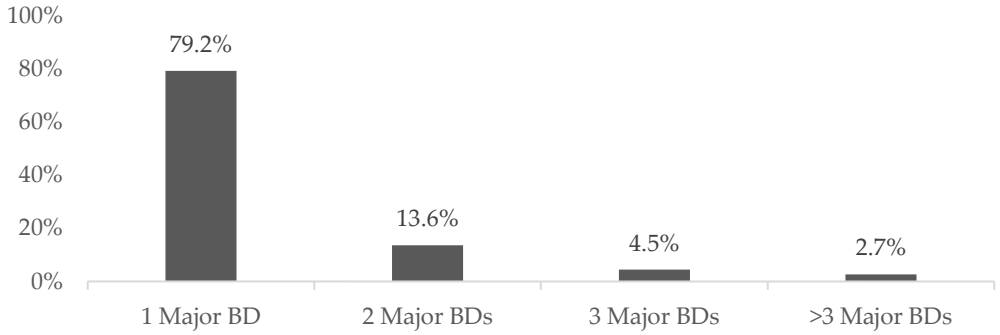
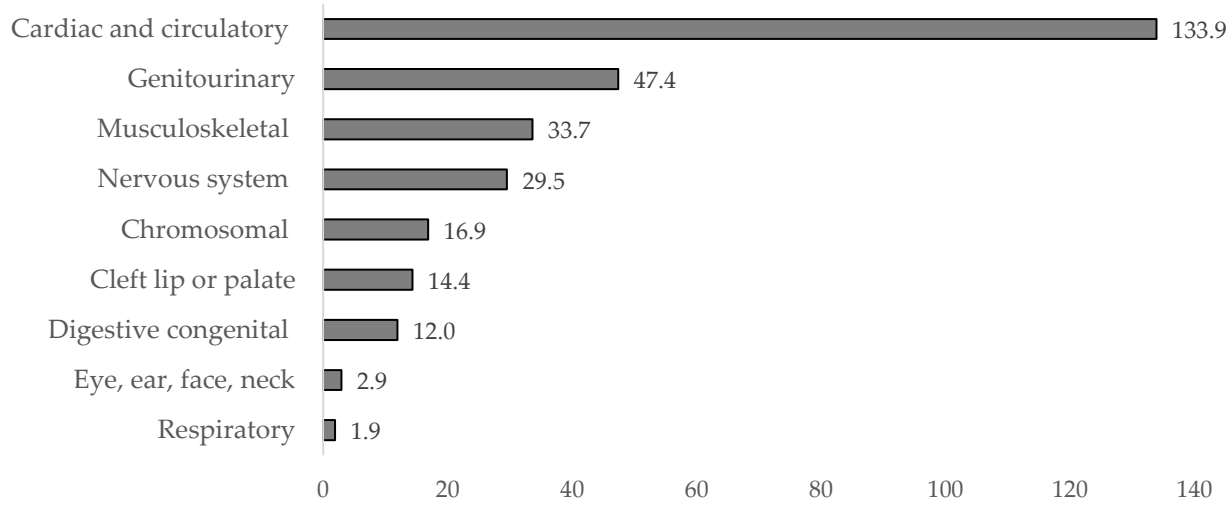


Table 1 shows the case counts for specific types of major birth defects. In addition, the prevalence of each type of defect (case counts divided by number of live births) is presented with 95% confidence intervals.

The most common type of major birth defect in CT is cardiac and circulatory congenital anomalies. Over 1 in 100 babies (about 1.3% or 133.4 per 10,000 babies) in CT are born with cardiac and circulatory congenital anomalies (Figure 3). The two most common major birth defects in CT are atrial and ventricular septal defects which are cardiac and circulatory congenital anomalies (Table 1). Please note that the most common major birth defects may be overrepresented in the hospital discharge data which contains no confirmation of diagnosis.

Figure 3: Rate of Infants with Major Birth Defects by Body System per 10,000 Infants



For more information on birth defects, please call the CT Department of Public Health at (860) 509-8000 or visit the following websites:

Connecticut Department of Public Birth Defects Registry: https://portal.ct.gov/dph/Family-Health/Birth-Defects-Registry/Connecticut-Birth-Defects-Registry
Connecticut Department of Public Health: www.ct.gov/dph
The National Center on Birth Defects and Developmental Disability at CDC: www.cdc.gov/ncbddd
March of Dimes: www.marchofdimes.org
National Birth Defects Prevention Network. https://www.nbdpn.org/

Suggested Citation

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Contact Information

For more information contact: CT Department of Public Health, Community, Family Health, and Prevention Section at (860) 509-8251

Table 1: Connecticut Birth Defects, 2018-2020

Body System	Defect	Live Birth Count	Prevalence per 10,000 Births (95% Confidence Interval)	
Cardiac and circulatory congenital anomalies	Atrial septal defect	902	90.6	(84.7-96.5)
	Ventricular septal defect	520	52.2	(47.8-56.7)
	Interrupted aortic arch	95	9.5	(7.6-11.5)
	Pulmonary valve atresia and stenosis	63	6.3	(4.8-7.9)
	Coarctation of aorta	55	5.5	(4.1-7.0)
	Atrioventricular septal defect	32	3.2	(2.1-4.3)
	Tetralogy of Fallot	30	3.0	(1.9-4.1)
	Transposition of great arteries	25	2.5	(1.5-3.5)
	Hypoplastic left heart syndrome	19	*1.9*	*(1.1-2.8)*[a]
	Double outlet right ventricle	18	*1.8*	*(1.0-2.6)*[a]
	Aortic valve stenosis	11	*s*[b]	
	Single ventricle	9	*s*	
	Epstein's anomaly	*s*	*s*	
	Total anomalous pulmonary venous return	*s*	*s*	
	Tricuspid valve atresia and stenosis	*s*	*s*	
	Common truncus	*s*	*s*	
Chromosomal abnormalities	Down syndrome	128	12.9	(10.6-15.1)
	Turners syndrome[c]	20	*4.1*	*(2.3-5.9)*[a]
	Trisomy 18	13	*1.3*	*(0.6-2.0)*[a]
	Trisomy 13	6	*s*	
	Deletion 22 q11.2	*s*	*s*	
Cleft lip or palate	Cleft palate without cleft lip	82	8.2	(6.5-10.0)
	Cleft palate with cleft lip	42	4.2	(2.9-5.5)
	Cleft lip without cleft palate	32	3.2	(2.1-4.3)
Congenital malformations of eye, ear, face, neck	Anotia/microtia	17	*1.7*	*(0.9-2.5)*[a]
	Congenital cataract	9	*s*	
	Anophthalmia/microphthalmia	*s*	*s*	
Digestive congenital anomalies	Small intestinal atresia/stenosis	48	4.8	(3.5-6.2)
	Rectal and large intestinal atresia/stenosis	37	3.7	(2.5-4.9)
	Esophageal atresia/tracheoesophageal fistula	30	3.0	(1.9-4.1)
	Biliary atresia	16	1.6	(0.8-2.4)
Genitourinary congenital anomalies	Hypospadias and Epispadias[d]	388	39.0	(35.1-42.9)
	Renal agenesis/hypoplasia	70	7.0	(5.4-8.7)
	Congenital posterior urethral valves[d]	11	*s*	
	Bladder exstrophy	6	*s*	
	Cloacal exstrophy	*s*	*s*	

Musculoskeletal congenital conditions	Clubfoot	199	20.0	(17.2-22.8)
	Craniosynostosis	34	3.4	(2.3-4.6)
	Omphalocele	33	3.3	(2.2-4.4)
	Diaphragmatic hernia	28	2.8	(1.8-3.9)
	Gastroschisis	28	2.8	(1.8-3.9)
	Deformities of upper limbs	13	*1.3*	*(0.6-2.0)* [a]
	Deformities of lower limbs	7	*s*	
	Microcephalus	260	26.1	(23.0-29.3)
	Spina bifida without anencephalous	24	*2.4*	*(1.4-3.4)* [a]
	Encephalocele	8	*s*	
	Holoprosencephaly	8	*s*	
	Anencephaly	*s*	*s*	
Respiratory congenital malformations	Choanal atresia	19	*1.9*	*(1.1-2.8)* [a]

Abbreviation: s, Suppressed

[a] Rates may be unstable due to low sample size. Take precaution when interpreting rates.

[b] Suppressed due to number and rates are suppressed due to low sample size and high error rates.

[c] Defect reported among females only.

[d] Defect reported among males only.

Appendix 1. Glossary of Terms Used in this Report

Agenesis The complete absence of part(s) of the body.

Agenesis, aplasia, or hypoplasia The absence or incomplete development of an organ or body part.

Anencephaly Congenital absence of the skull, with cerebral hemispheres completely missing or reduced to small masses attached to the base of the skull. Anencephaly is not compatible with life.

Anophthalmia A developmental defect characterized by complete absence of the eyes, or by the presence of vestigial eyes.

Anotia A congenital absence of one or both ears.

Aortic valve stenosis A cardiac anomaly characterized by a narrowing or stricture of the aortic valve. This condition causes abnormal cardiac circulation and pressure in the heart during contractions. This condition can be repaired surgically in some cases.

Atresia Absence or closure of a normal opening.

Atrial septal defect (ASD) A congenital cardiac malformation in which there are one or more openings in the atrial septum (muscular and fibrous wall between the right and left atria) allowing a mixing of oxygenated and unoxygenated blood. The openings vary in size and may resolve without treatment or may require surgical treatment.

Biliary atresia A congenital absence or underdevelopment of one or more of the ducts in the biliary tract. Correctable surgically.

Birth prevalence or rate (# of cases with birth defect A in an area and time period ÷ #of live births in that area and period) X 10,000. See also Prevalence.

Bladder exstrophy Incomplete closure of the anterior wall of the bladder and the abdominal cavity. The abdominal wall and underlying organs do not fuse properly so that the bladder is exposed on the outside of the body.

Cardiovascular See Heart Defects.

Cataract An opacity (clouding) of the lens of the eye.

Central Nervous System Related to the brain or spinal cord.

Choanal atresia or stenosis A congenital anomaly in which a bony or membranous formation blocks the passageway between the nose and the pharynx. This defect is usually repaired surgically after birth.

Chromosomal Relating to chromosomes. Chromosomal defects involve abnormal structure or number of chromosomes, including partial or total absence of chromosomes or presence of extra chromosomes or parts of chromosomes. Examples include trisomy 13, Turner syndrome, and Down syndrome (trisomy 21).

Cleft lip The congenital failure of the fetal components of the lip to fuse or join, forming a groove or fissure in the lip.

Cleft palate The congenital failure of the palate to fuse properly, forming a grooved depression or fissure in the roof of the mouth. This defect varies in degree of severity. The fissure can extend into the hard and soft palate and into the nasal cavities.

Clubfoot A deformity in which an infant's foot is turned inward, often so severely that the bottom of the foot faces sideways or even upward.

Coarctation of the aorta Localized narrowing of the aorta. This condition causes abnormal cardiac circulation and pressure in the heart during contractions. This condition can vary from mild to severe.

Common truncus See Truncus arteriosus.

Confidence interval (CI) (95%) A range that is calculated based on the standard error of a measurement and conveys how precise a measurement is. 95% confidence interval means that that the likelihood of the true mean falling within the interval is 95%.

Congenital Existing at or dating from birth.

Craniosynostosis A premature closing of the cranial sutures before or soon after birth. This condition is occasionally associated with other skeletal defects. If no surgical correction is made, the growth of the skull is inhibited, and the head is deformed.

Diaphragmatic hernia A failure of the diaphragm to form completely, leaving a hole. Abdominal organs may protrude through the hole into the chest cavity and interfere with development of the heart and lungs.

Down syndrome (Trisomy 21) The chromosomal abnormality characterized by an extra copy of chromosome 21. In rare cases this syndrome is caused by translocation. Down syndrome can occur in mosaic, where there are some normal cells and some trisomy 21 cells. Many infants with Down syndrome also have congenital heart disease.

Double outlet right ventricle (DORV) A congenital heart defect where the pulmonary artery and the aorta both connect to the right ventricle.

Ebstein anomaly A congenital heart defect in which the tricuspid valve is displaced downward into the right ventricle causing abnormal patterns of cardiac circulation.

Encephalocele The protrusion of the brain substance through a defect in the skull.

Epispadias A urogenital anomaly characterized by the failure of the urethral tube to tubularize on the dorsal aspect.

Esophageal stenosis or atresia A narrowing or incomplete formation of the esophagus. Usually a surgical emergency. Frequently associated with a tracheoesophageal fistula.

Fistula An abnormal passage from an internal organ to the body surface or between two internal organs or structures.

Gastrointestinal Related to the digestive system. Includes defects of the stomach, esophagus, and liver.

Gastroschisis A congenital opening of the abdominal wall with protrusion of the intestines. This condition is surgically treated.

Genital or Urinary (Genitourinary) Related to the genital or urinary organs.

Heart defects (Cardiovascular defects) Congenital heart defects affect the structure and function of a baby's heart and circulation system.

Hernia A protrusion of an organ or part through connective tissue or through a wall of the cavity in which it is normally enclosed.

Holoprosencephaly Failure of the brain to develop into two equal halves, so there is structural abnormality of the brain. There may be associated midline facial defects including cyclopia (fusion of the eye orbits into a single cavity containing one eye) in severe cases. Frequently occurs with Trisomy 13.

Hypoplasia A condition of arrested development in which an organ or body part remains below the normal size or in an immature state.

Hypoplastic left heart syndrome Atresia, or marked hypoplasia, of the aortic opening or valve, with hypoplasia of the ascending aorta and defective development of the left ventricle (with mitral valve atresia). This condition is usually fatal if not treated.

Hypospadias A congenital defect in males in which the urinary meatus (urinary outlet) is on the underside of the penis or on the perineum (area between the genitals and the anus). The condition may be surgically corrected if needed for cosmetic, urologic, or reproductive reasons.

Limb defects See Reduction deformities.

Live birth Any infant who breathes or shows any other evidence of life at birth.

Major structural or genetic birth defects includes conditions that result from a malformation, deformation, or disruption in one or more parts of the body, a chromosomal abnormality, or a known clinical syndrome; are present at birth; and have serious, adverse effect on health, development, or functional ability.

Microcephaly Congenital small size of the head relative to the height, with corresponding small brain size.

Microphthalmia The congenital abnormal smallness of one or both eyes. Can occur in the presence of other ocular defects.

Microtia A small or maldeveloped external ear and absent or closed off external auditory canal.

Musculoskeletal Related to the limbs or skeletal systems. Musculoskeletal defects may involve absence, abnormality or hypoplasia of limbs, like arms or legs, or structural abnormalities of limbs, muscles (such as diaphragmatic hernia), or abdominal wall (such as gastroschisis or omphalocele).

Neural tube defect A type of defect that occurs when the neural tube doesn't close properly in early pregnancy. Includes defects of the brain and spinal cord, like anencephaly, spina bifida and encephalocele.

Omphalocele The protrusion of an organ into the umbilicus. The defect is usually closed surgically soon after birth. Contrast with Gastroschisis.

Posterior urethral valves Posterior urethral valves (PUV) are tissue folds in the posterior urethra. Congenital PUV is an abnormal membrane in males in the posterior urethra and is the most common cause of bladder outlet obstruction in male children.

Prevalence The number of birth defects observed during a period of time divided by the number of live births during the same time period. (# of cases with birth defect A in an area and time period ÷ #of live births in that area and period) X 10,000. See also Birth prevalence.

Pulmonary valve atresia or stenosis A congenital heart condition characterized by absence or constriction of the pulmonary valve. This condition causes abnormal cardiac circulation and pressure in the heart during contractions. This condition can vary from mild to severe. Mild

forms are relatively well tolerated and require no intervention. More severe forms are surgically corrected.

Reduction defects of the lower limbs The congenital absence of a portion of the lower limb (examples: a missing or shortened leg, missing toes.)

Reduction defects of the upper limbs The congenital absence of a portion of the upper limb (examples: a missing or shortened arm, missing fingers.)

Renal agenesis The failure of embryonic development of the kidney.

Spina bifida A neural tube defect resulting from failure of the spinal neural tube to close. The spinal cord and/or meninges may or may not protrude. This usually results in damage to the spinal cord with paralysis of the involved limbs. Includes myelomeningocele (involving both spinal cord and meninges) and meningocele (involving just the meninges).

Stenosis A narrowing or constriction of the diameter of a bodily passage or orifice.

Stenosis or atresia of large intestine, rectum and anus The absence, closure or constriction of the large intestine, rectum or anus. Can be surgically corrected or bypassed.

Stenosis or atresia of the small intestine A narrowing or incomplete formation of the small intestine obstructing movement of food through the digestive tract.

Tetralogy of Fallot A congenital cardiac anomaly consisting of four defects: ventricular septal defect, pulmonary valve stenosis or atresia, displacement of the aorta to the right, and hypertrophy of right ventricle. The condition is corrected surgically.

Total anomalous pulmonary venous connection A congenital heart defect which all four pulmonary veins do not connect normally to the left atrium.

Tracheoesophageal fistula An abnormal passage between the esophagus and trachea. Corrected surgically. It is frequently associated with esophageal atresia.

Transposition of the great vessels (Transposition of the great arteries/TGA) A congenital malformation in which the aorta arises from the right ventricle and the pulmonary artery from the left ventricle (opposite of normal), so that the venous return from the peripheral circulation is recirculated without being oxygenated in the lungs. Can occur in Levo (L-) or Dextro (d-) form. Dextro usually requires immediate surgical correction.

Tricuspid valve atresia or stenosis A congenital cardiac condition characterized by the absence or constriction of the tricuspid valve. The opening between the right atrium and right ventricle is absent or restricted, and normal circulation is not possible. This condition is often associated with other cardiac defects. This condition is surgically corrected depending on the severity.

Trisomy 13 The chromosomal abnormality caused by an extra chromosome 13. The syndrome can occur in mosaic so that there is a population of normal cells and a population of trisomy 13 cells. The syndrome is characterized by impaired midline facial development, cleft lip and palate, polydactyly and mental retardation. Most infants do not survive beyond 6 months of life. Also known as Patau syndrome.

Trisomy 18 The chromosomal abnormality characterized by an extra copy of chromosome 18. Trisomy 18 can occur in mosaic. The syndrome is characterized by mental retardation, neonatal hepatitis, low-set ears, skull malformation and short digits. Cardiac and renal anomalies are also common. Survival for more than a few months is rare. Also known as Edwards syndrome.

Trisomy 21 See Down syndrome.

Truncus arteriosus (Common truncus) A congenital heart defect in which the common arterial trunk fails to divide into pulmonary artery and aorta. This is corrected surgically.

Turners syndrome A condition that affects only females, results when one of the X chromosomes (sex chromosomes) is missing or partially missing.

Ventricle One of the two lower chambers of the heart (plural ventricles). The right ventricle sends blood to the lungs, and the left ventricle passes oxygen-rich blood to the rest of the body.

Ventricular septal defect (VSD) A congenital cardiac malformation in which there are one or more openings in the ventricular septum (muscular and fibrous wall between the right and left ventricle or right and left lower chambers of the heart) allowing a mixing of oxygenated and deoxygenated blood. The openings vary in size and may resolve without treatment or require surgical treatment.

Definitions adapted from: Texas Department of State Health Services Glossary of Birth Defects Terms, last updated February 24, 2021: https://www.dshs.texas.gov/birthdefects/glossary.shtm