Birth Defects in Connecticut 2001-2004

A Surveillance Report on Birth Defects Prevalence

Connecticut Department of Public Health
Public Health Initiatives Branch
Family Health Section
Connecticut Birth Defects Registry

CONNECTICUT DEPARTMENT OF PUBLIC HEALTH

Keeping Connecticut Healthy

Birth Defects in Connecticut, 2001-2004

A surveillance report on birth defects prevalence

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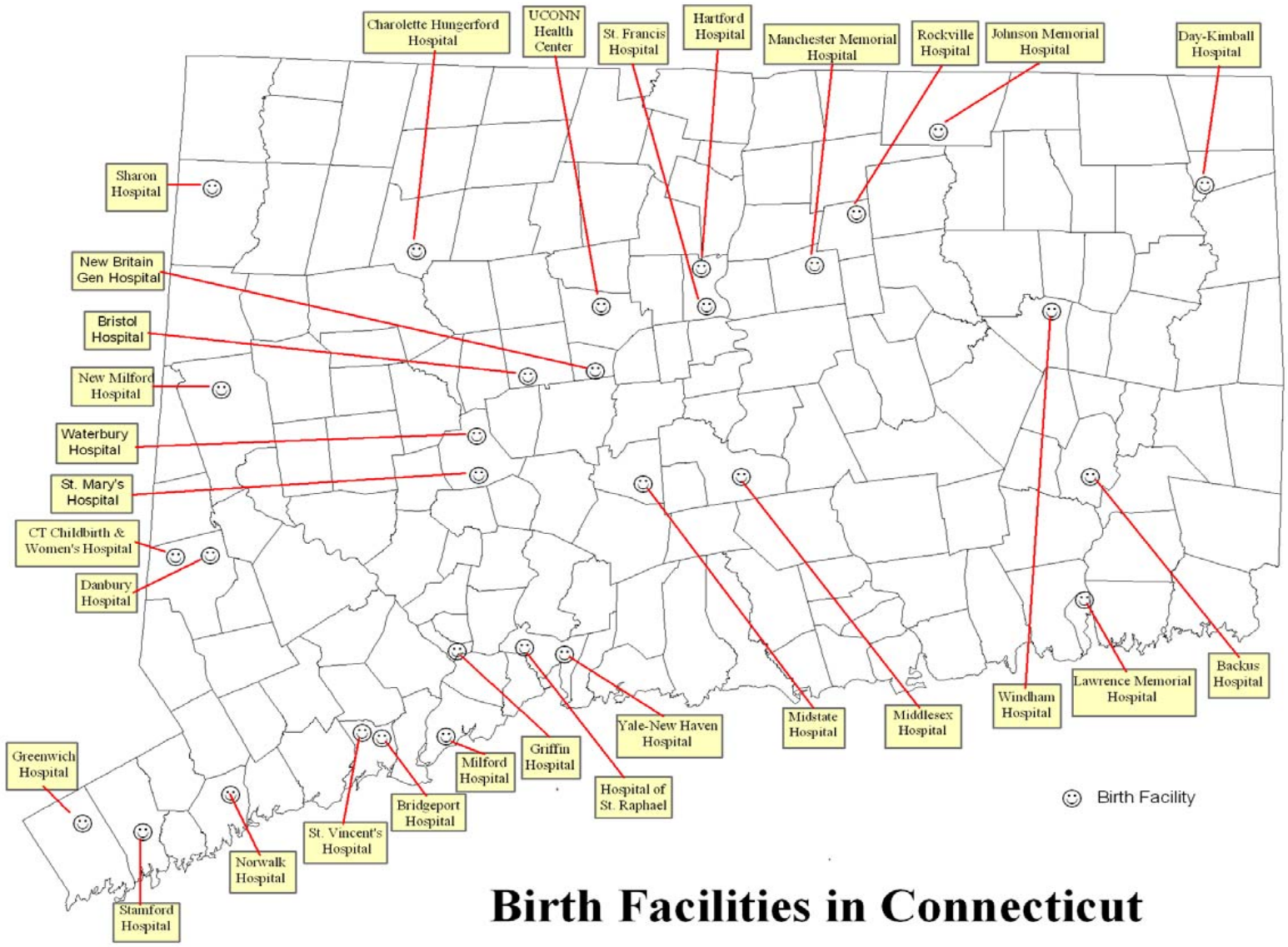
Understanding the burden of birth defects in Connecticut would not be possible without the enormous contribution of staff in the neonatal intensive care units and well baby nurseries of birth facilities across the state to identify and report such information to the Connecticut Birth Defects Registry. The Department of Public Health would like to acknowledge their commitment to completeness, accuracy, and integrity of birth defects data. Without the dedication of the following facilities, the pursuit of birth defects control and prevention would be greatly compromised.

Birth Facilities

Bridgeport Hospital
Bristol Hospital
CT Childbirth and Women's Hospital
Charlotte Hungerford Hospital
Danbury Hospital
Day-Kimball Hospital
Greenwich Hospital
Griffin Hospital
Hartford Hospital
John Dempsey Hospital
Johnson Memorial Hospital
Lawrence & Memorial Hospital
Manchester Memorial Hospital
Mid State Medical Center
Middlesex Hospital
Milford Hospital
New Britain General Hospital
New Milford Hospital
Norwalk Hospital
Rockville Gen. Hospital
St. Vincent Hospital
Sharon Hospital
St. Francis Hospital
St. Mary's Hospital
Hospital of St. Raphael
Stamford Hospital
Waterbury Hospital
William Backus Hospital
Windham Hospital
Yale-New Haven Hospital


Charolette Hungerford Hospital
UCONN Health Center
St. Francis Hospital
Hartford Hospital
Manchester Memorial Hospital
Rockville Hospital
Johnson Memorial Hospital
Day-Kimball Hospital
Sharon Hospital
New Britain Gen Hospital
Bristol Hospital
New Milford Hospital
Waterbury Hospital
St. Mary's Hospital
CT Childbirth & Women's Hospital
Danbury Hospital
Greenwich Hospital
Stamford Hospital
Norwalk Hospital
St. Vincent's Hospital
Bridgeport Hospital
Milford Hospital
Griffin Hospital
Yale-New Haven Hospital
Hospital of St. Raphael
Midstate Hospital
Middlesex Hospital
Windham Hospital
Lawrence Memorial Hospital
Backus Hospital
Birth Facility
Birth Facilities in Connecticut

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INTRODUCTION

Birth defects, as defined by the March of Dimes, is an abnormality of structure, function, or body metabolism that is present at birth and results in physical or mental disability. Most birth defects are included within the range of diagnosis codes 740.0 to 759.9, as defined within the International Classification of Disease (ICD), 9th Revision. Conditions within this range include: (1) malformations, which involve poor tissue formation; (2) deformations, which involve unusual forces on normal tissue; and (3) disruptions, which involve the break-down of normal tissue. Examples of birth defects in body systems include: (1) central nervous system defects; (2) congenital heart defects; (3) gastrointestinal defects; (4) oral cleft defects; (5) musculoskeletal defects; (6) genital and urinary defects; (7) chromosomal defects; and (8) other defects.

In the United States, about 3% of babies are born with birth defects. Some women are at increased risk of having a child with a birth defect. Certain risks include the use of certain medications during pregnancy; alcohol and/or tobacco use during pregnancy; and family history. Also, women over the age of 35 years have a higher chance of having a child with Down Syndrome than women who are younger.

Birth defects are one of the leading causes of infant mortality in Connecticut as they are in the United States [1]. Birth defects account for about 20% of all infant deaths in this country [2], and birth defects were the underlying causes for 15% of the infant deaths for Connecticut residents in 2003 [3]. National data indicate that a substantial percentage of deaths of older children are related to birth deaths (15.5% among 1-5 year olds and 8% among 5-9 year olds) [4]. Although 90% of infants born with birth defects survive their first year of life, some of these children will have life-long disabilities and significant medical and rehabilitation needs. Surgical intervention can prevent future sequelae in certain cases. Regardless, all of the families of children with major defects experience both financial and emotional consequences. Lifetime costs have been estimated at $6 billion for those infants born in a single year with 1 or more of 17 major birth defects [5].

A recent study examining hospitalization stays and charges for selected birth defects indicates the average length of hospital stay for newborns was longest for infants with surgically repaired gastroschisis for 41.0 days and omphalocele for 32.5 days. The most expensive average neonatal hospital charges were for congenital heart defects of hypoplastic left heart at $199,597 and common truncus arteriosus at $192,781. Six birth defects had total cumulative charges of approximately $200 million or greater in 2003: obstructive genitourinary defect, pulmonary value stenosis, coarctation of the aorta, transposition of the great arteries, and gastroschisis [6]. Based on information from the 1998 Connecticut birth cohort, the estimates lifetime costs associated with selected birth defects range from $1.4 million for reduction defects of the lower limb to over $28 million for Down Syndrome [7].

Although birth defects are a significant cause of morbidity and mortality, there is relatively limited information about their causes; approximately 70% of birth defects are of unknown cause. Known causes of birth defects include single gene mutations, chromosomal abnormalities, maternal illness such as diabetes or infections, and the use of certain medications [8]. Continued

surveillance, research, and prevention efforts are essential to reducing the morbidity and mortality of infants born with birth defects.

This report is organized into three major sections: (1) the summary of all birth defects by year, gender, race/ethnicity, and maternal age; (2) system-specific birth defects statistics, and (3) the appendices. Here are features of this report:

- A section on birth defects prevention is included to illustrate the importance of prevention at different levels to reduce birth defects burdens.
- Comparisons of prevalence rates by gender, race/ethnicity, and maternal age are included to illustrate the disparities of risks associated with birth defects.
- Trends of birth defects prevalence over time are examined, by system, to illustrate the effectiveness of birth defects prevention and intervention programs.
- Comparisons of prevalence rates between counties and the state are available to identify potential risks of birth defects associated with different life styles and possible environmental/occupational exposures.
- Geographic Information System (GIS) mapping is used to illustrate the differences in rates between counties and the state for cardiovascular system defects and genitourinary system defects.
- Comparison of prevalence rates for selected birth defects in Connecticut and states participating in the National Birth Defects Prevention Study.
- Definitions and ICD-9 CM codes associated with each birth defect are provided.
- Information from the 2003 Connecticut Pregnancy Risk Assessment Tracking System (PRATS) survey is included to provide information related to both risky and health-prompting behaviors- that are associated with certain types of birth defects.
- Information from the Healthy People 2010 is included to provide information on long-term objectives for achieving these goals.
- A section on “Frequently Asked Questions” is included to provide answers to widely received inquires from the public.
- Technical notes are included in the appendices to provide information on methodological issues.
- Sources of additional information are included in the appendices.

Family Health Section

The Family Health Section is located under the Public Health Initiatives Branch in the State of Connecticut's Department of Public Health. The Family Health Section is responsible for administering the State Title V Maternal Child Health Services Block Grant in addition to other state and federally funded programs. The section's mission is to continually improve the health of Connecticut's residents across the lifespan through culturally appropriate surveillance, public education, family-centered interventions, and community-based capacity building. Charged with the responsibility of optimizing the health of Connecticut families, the Family Health Section strives to assure that all individuals and families achieve optimal health through appropriate and comprehensive health services.

The section organizational structure is comprised of the following units – School and Adolescent Health, Primary Care and Prevention, Epidemiology, Newborn Screening, and Children and Youth with Special Health Care Needs.

The Connecticut Birth Defects Registry (CTBDR)

The Connecticut Birth Defects Registry is a passive surveillance system developed to collect information about birth defects that occur among state residents.

Mission

The mission of the Connecticut Birth Defects Registry is to: (1) maintain statewide surveillance through collecting information on birth defects in Connecticut; (2) monitor trends and patterns in birth defects; (3) conduct analyses to identify risk factors for birth defects; and (4) promote education activities for the prevention of birth defects.

Data Sources

The Connecticut Birth Defects Registry uses various sources to collect information on birth defects, including reporting from the birth facilities, electronic birth certificates, and in-patient hospital discharge data (Connecticut Health Information Management & Exchange - CHIME data). The reporting of birth defects information from birth facilities is part of the electronic Newborn Screening System (NSS) that includes Early Hearing Detection and Intervention (EHDI), newborn genetic screening, and the CTBDR. Newborns with birth defects diagnosed before discharge from hospitals are reported to the CTBDR through the NSS. This information is linked to the electronic birth certificates in the Vital Records Section of the Connecticut Department of Public Health for quality assurance purposes. The congenital anomalies recorded on the birth certificates are compared with those reported by birth hospitals, and such information are used to supplement the information collected in the CTBDR.

In addition, CTBDR uses in-patient hospital discharge data as a data source. The Connecticut Department of Public Health contracts with the Connecticut Hospital Association to obtain the hospital discharge data. The three data sources are merged to create a consolidated dataset for CTBDR. This consolidated dataset is used to produce statistical information for surveillance purposes.

Compliance of reporting on birth defects information from birth facilities is estimated to be 86.2% for 2005, and this information is shared with birth facilities on an ongoing basis to reinforce the continued reporting. Overall data quality in the CTBDR is assessed on a routine basis, in terms of completeness, timeliness, and accuracy [9-10].

Surveillance

The surveillance activities in CTBDR provide useful statistical information to health care professionals, researchers, and policy makers. Data from CTBDR have also been used for specific research projects [11]. Reporting of birth defects to the Registry is mandatory under the Connecticut State Statutes Sec. 19a-53, 19a-54, and 19a-56a.

Confidentiality

All data collected by the Connecticut Birth Defects Registry complies with the state and federal privacy and confidentiality regulations.

Referral

There are a multitude of specialty treatment centers, support services, and family advocacy groups available to assist families of a child with birth defects. The Child Development Infoline is the resource of referral services for children with birth defects and other special health care needs.

BIRTH DEFECTS PREVENTION

Several types of birth defects may be prevented, and considerable progress continued to be made to improve the quality of life and survival for children with birth defects. Prevention strategies are based on the natural history of the birth defects development, and are categorized into three levels of intervention.

Primary prevention – is to limit the occurrence of birth defects by controlling exposure to risk factors or increasing an individual's resistance to them (e.g., by taking folic acids). Clearly, the first step is to identify the relevant exposures and to assess their impact on the risk of developing disease in the population. For example, taking folic acid during pregnancy may prevent neural tube defects. Maternal smoking during pregnancy may increase risks for cleft lip, cleft palate, clubfoot, limb defects, heart defects, gastroschisis, and imperforate anus.

This report includes information from the second round of the Pregnancy Risk Assessment Tracking System (PRATS) survey conducted in 2003 whenever possible and appropriate.

This icon indicates data from the Pregnancy Risk Assessment Tracking System (PRATS) survey in 2003, a survey of CT women who recently gave birth that assessed risk behaviors and attitudes pertinent to pregnancy outcomes.

* The results of this survey are limited by the low response rate (44.2%). Although the demographic profile of respondents correlated well with the sample's characteristics, as well as the birth cohort in 2003, the results may be considered representative of the respondents only.

Secondary prevention – refers to detection of diseases at an early stage, when intervention is more effective than at the time of usual diagnosis and treatment. Early detection and intervention can reduce or eliminate the complications related to the condition, including death. Screening represents an important component of secondary prevention. Prenatal visits provide good opportunities to identify birth defects early and employ appropriate interventions to reduce the consequences of birth defects.

Tertiary Prevention – aims at improving the prognosis and quality of life of affected individuals by offering them the best available treatment and rehabilitation programs.

The ultimate goal of birth defects prevention is to reduce the associated morbidity and mortality. It is important to set up long-term objectives for achieving these goals through various birth defects prevention activities. Objectives from the Healthy People 2010 are included in this report whenever possible and appropriate.

This icon indicates goals of Healthy People 2010 from the CDC National Center for Health Statistics.

SUMMARY OF ALL BIRTH DEFECTS

Table 1. Frequency and Prevalence Rates of Birth Defects by Year, CT, 2001-04

	2001	2002	2003	2004	2001-04	
	N	N	N	N	N	Rate*
Central Nervous System						
Anencephalus	2	0	0	1	3	0.18
Spina bifida without anencephalus	7	17	21	7	52	3.07
Hydrocephalus without Spina bifida	0	0	0	0	0	0.00
Encephalocele	3	1	6	2	12	0.71
Microcephalus	13	26	16	13	68	4.01
Eye and Ear						
Anophthalmia/microphthalmia	2	6	4	1	13	0.77
Congenital cataract	11	10	13	7	41	2.42
Aniridia	0	0	0	0	0	0.00
Anotia/microtia	5	2	4	1	12	0.71
Cardiovascular						
Common truncus	4	1	2	6	13	0.77
Transposition of great arteries	10	21	20	18	69	4.07
Tetralogy of Fallot	23	19	20	20	82	4.84
Ventricular septal defect	221	206	220	139	786	46.38
Atrial septal defect	185	212	215	178	790	46.61
Endocardial cushion defect	19	16	20	11	66	3.89
Pulmonary valve atresia and stenosis	26	17	31	23	97	5.72
Tricuspid valve atresia and stenosis	1	1	4	5	11	0.65
Ebstein's anomaly	3	3	1	0	7	0.41
Aortic valve stenosis	5	7	5	6	23	1.36
Hypoplastic left heart syndrome	9	5	13	8	35	2.07
Patent ductus arteriosus (BW>=2500 grams)	233	130	116	102	581	34.28
Coarctation of aorta	19	18	23	6	66	3.89
Orofacial						
Cleft palate without cleft lip	23	33	29	22	107	6.31
Cleft lip with and without cleft palate	26	30	38	20	114	6.73
Choanal atresia	10	11	7	2	30	1.77
Gastrointestinal						
Esophageal atresia/tracheoesophageal fistula	19	13	13	12	57	3.36
Rectal and large intestinal atresia/stenosis	16	18	11	15	60	3.54
Pyloric stenosis	107	129	74	73	383	22.60
Hirschsprung's disease (congenital megacolon)	11	11	13	15	50	2.95
Biliary atresia	4	4	3	2	13	0.77
Genitourinary						
Renal agenesis/hypoplasia	15	13	16	15	59	3.48
Bladder exstrophy	0	3	1	2	6	0.35
Obstructive genitourinary defect	115	123	112	100	450	26.55
Hypospadias and Epispadias	190	190	201	144	725	83.45
Musculoskeletal						
Reduction deformity, upper limb	12	5	8	5	30	1.77
Reduction deformity, lower limb	3	2	4	1	10	0.59
Gastroschisis/Omphalocele	25	18	18	9	70	4.13
Congenital hip dislocation	46	41	37	33	157	9.26
Diaphragmatic hernia	7	10	12	9	38	2.24
Chromosomal						
Trisomy 13	0	6	2	2	10	0.59
Down syndrome	61	61	62	46	230	13.57
Trisomy 18	7	1	3	2	13	0.77
Other						
Fetal alcohol syndrome	5	4	4	5	18	1.06

* Rates are per 10,000 live births

Table 2. Frequency and Prevalence Rates of Birth Defects by Gender, CT, 2001-04

	Male		Female		Total	
	N	Rate*	N	Rate	N	Rate
Central Nervous System						
Anencephalus	0	0.00	3	0.36	3	0.18
Spina bifida without anencephalus	24	2.76	28	3.39	52	3.07
Hydrocephalus without Spina bifida	0	0.00	0	0.00	0	0.00
Encephalocele	10	1.15	2	0.24	12	0.71
Microcephalus	31	3.57	37	4.48	68	4.01
Eye and Ear						
Anophthalmia/microphthalmia	8	0.92	5	0.61	13	0.77
Congenital cataract	24	2.76	17	2.06	41	2.42
Aniridia	0	0.00	0	0.00	0	0.00
Anotia/microtia	9	1.04	3	0.36	12	0.71
Cardiovascular						
Common truncus	6	0.69	7	0.85	13	0.77
Transposition of great arteries	42	4.83	27	3.27	69	4.07
Tetralogy of Fallot	49	5.64	33	4.00	82	4.84
Ventricular septal defect	362	41.67	424	51.33	786	46.38
Atrial septal defect	419	48.23	371	44.92	790	46.61
Endocardial cushion defect	30	3.45	36	4.36	66	3.89
Pulmonary valve atresia and stenosis	42	4.83	55	6.66	97	5.72
Tricuspid valve atresia and stenosis	3	0.35	8	0.97	11	0.65
Ebstein's anomaly	3	0.35	4	0.48	7	0.41
Aortic valve stenosis	14	1.61	9	1.09	23	1.36
Hypoplastic left heart syndrome	19	2.19	16	1.94	35	2.07
Patent ductus arteriosus (BW>=2500 grams)	298	34.30	283	34.26	581	34.28
Coarctation of aorta	41	4.72	25	3.03	66	3.89
Orofacial						
Cleft palate without cleft lip	48	5.52	59	7.14	107	6.31
Cleft lip with and without cleft palate	71	8.17	43	5.21	114	6.73
Choanal atresia	16	1.84	14	1.69	30	1.77
Gastrointestinal						
Esophageal atresia/tracheoesophageal fistula	41	4.72	16	1.94	57	3.36
Rectal and large intestinal atresia/stenosis	30	3.45	30	3.63	60	3.54
Pyloric stenosis	297	34.19	86	10.41	383	22.60
Hirschsprung's disease (congenital megacolon)	31	3.57	19	2.30	50	2.95
Biliary atresia	8	0.92	5	0.61	13	0.77
Genitourinary						
Renal agenesis/hypoplasia	42	4.83	17	2.06	59	3.48
Bladder exstrophy	4	0.46	2	0.24	6	0.35
Obstructive genitourinary defect	330	37.98	120	14.53	450	26.55
Hypospadias and Epispadias	725	83.45	0	0.00	725	42.78
Musculoskeletal						
Reduction deformity, upper limb	21	2.42	9	1.09	30	1.77
Reduction deformity, lower limb	6	0.69	4	0.48	10	0.59
Gastroschisis/Omphalocele	35	4.03	35	4.24	70	4.13
Congenital hip dislocation	34	3.91	123	14.89	157	9.26
Diaphragmatic hernia	26	2.99	12	1.45	38	2.24
Chromosomal						
Trisomy 13	6	0.69	4	0.48	10	0.59
Down syndrome	132	15.19	98	11.86	230	13.57
Trisomy 18	5	0.58	8	0.97	13	0.77
Other						
Fetal alcohol syndrome	5	0.58	13	1.57	18	1.06

* Rates are per 10,000 live births

Table 3. Frequency and Prevalence Rates of Birth Defects by Race/Ethnicity, CT, 2001-04

	White		Black		Hispanic	
	N	Rate*	N	Rate	N	Rate
Central Nervous System						
Anencephalus	2	0.19	1	0.60	1	0.44
Spina bifida without anencephalus	39	3.69	8	4.82	11	4.81
Hydrocephalus without Spina bifida	0	0.00	0	0.00	0	0.00
Encephalocele	9	0.85	2	1.21	3	1.31
Microcephalus	48	4.54	15	9.04	16	7.00
Eye and Ear						
Anophthalmia/microphthalmia	11	1.04	1	0.60	1	0.44
Congenital cataract	28	2.65	10	6.03	6	2.63
Aniridia	0	0.00	0	0.00	0	0.00
Anotia/microtia	11	1.04	1	0.60	2	0.88
Cardiovascular						
Common truncus	11	1.04	1	0.60	6	2.63
Transposition of great arteries	57	5.40	8	4.82	7	3.06
Tetralogy of Fallot	71	6.72	6	3.62	13	5.69
Ventricular septal defect	643	60.86	104	62.70	128	56.02
Atrial septal defect	635	60.10	109	65.71	115	50.33
Endocardial cushion defect	50	4.73	13	7.84	12	5.25
Pulmonary valve atresia and stenosis	70	6.63	22	13.26	9	3.94
Tricuspid valve atresia and stenosis	10	0.95	1	0.60	3	1.31
Ebstein's anomaly	7	0.66	0	0.00	3	1.31
Aortic valve stenosis	20	1.89	3	1.81	1	0.44
Hypoplastic left heart syndrome	29	2.74	5	3.01	6	2.63
Patent ductus arteriosus (BW>=2500 grams)	457	43.25	88	53.05	84	36.76
Coarctation of aorta	54	5.11	6	3.62	8	3.50
Orofacial						
Cleft palate without cleft lip	92	8.71	7	4.22	13	5.69
Cleft lip with and without cleft palate	98	9.28	12	7.23	12	5.25
Choanal atresia	27	2.56	1	0.60	5	2.19
Gastrointestinal						
Esophageal atresia/tracheoesophageal fistula	47	4.45	6	3.62	5	2.19
Rectal and large intestinal atresia/stenosis	43	4.07	10	6.03	6	2.63
Pyloric stenosis	327	30.95	29	17.48	74	32.39
Hirschsprung's disease (congenital megacolon)	43	4.07	4	2.41	8	3.50
Biliary atresia	9	0.85	3	1.81	3	1.31
Genitourinary						
Renal agenesis/hypoplasia	53	5.02	5	3.01	8	3.50
Bladder exstrophy	6	0.57	0	0.00	0	0.00
Obstructive genitourinary defect	370	35.02	43	25.92	52	22.76
Hypospadias and Epispadias	634	116.77	64	75.26	67	57.62
Musculoskeletal						
Reduction deformity, upper limb	27	2.56	2	1.21	4	1.75
Reduction deformity, lower limb	4	0.38	3	1.81	0	0.00
Gastroschisis/Omphalocele	56	5.30	10	6.03	11	4.81
Congenital hip dislocation	141	13.35	10	6.03	16	7.00
Diaphragmatic hernia	33	3.12	3	1.81	2	0.88
Chromosomal						
Trisomy 13	9	0.85	1	0.60	0	0.00
Down syndrome	192	18.17	27	16.28	27	11.82
Trisomy 18	10	0.95	2	1.21	3	1.31
Other						
Fetal alcohol syndrome	8	0.76	8	4.82	4	1.75

* Rates are per 10,000 live births

Table 4. Frequency and Prevalence Rates of Birth Defects by Maternal Age, CT, 2001-04

	<20		20-34		35+	
	N	Rate*	N	Rate	N	Rate
Central Nervous System						
Anencephalus	1	0.87	1	0.08	1	0.27
Spina bifida without anencephalus	9	7.79	31	2.59	12	3.23
Hydrocephalus without Spina bifida	0	0.00	0	0.00	0	0.00
Encephalocele	0	0.00	8	0.67	4	1.08
Microcephalus	6	5.19	49	4.09	13	3.50
Eye and Ear						
Anophthalmia/microphthalmia	1	0.87	11	0.92	1	0.27
Congenital cataract	5	4.33	27	2.25	9	2.42
Aniridia	0	0.00	0	0.00	0	0.00
Anotia/microtia	0	0.00	12	1.00	0	0.00
Cardiovascular						
Common truncus	2	1.73	8	0.67	3	0.81
Transposition of great arteries	5	4.33	52	4.34	12	3.23
Tetralogy of Fallot	7	6.06	51	4.26	24	6.47
Ventricular septal defect	47	40.68	521	43.48	218	58.73
Atrial septal defect	49	42.41	552	46.07	189	50.92
Endocardial cushion defect	4	3.46	41	3.42	21	5.66
Pulmonary valve atresia and stenosis	8	6.92	65	5.42	24	6.47
Tricuspid valve atresia and stenosis	2	1.73	7	0.58	2	0.54
Ebstein's anomaly	0	0.00	7	0.58	0	0.00
Aortic valve stenosis	1	0.87	17	1.42	5	1.35
Hypoplastic left heart syndrome	3	2.60	28	2.34	4	1.08
Patent ductus arteriosus (BW>=2500 grams)	31	26.83	405	33.80	145	39.07
Coarctation of aorta	3	2.60	45	3.76	18	4.85
Orofacial						
Cleft palate without cleft lip	9	7.79	76	6.34	22	5.93
Cleft lip with and without cleft palate	12	10.39	76	6.34	26	7.01
Choanal atresia	3	2.60	18	1.50	9	2.42
Gastrointestinal						
Esophageal atresia/tracheoesophageal fistula	2	1.73	34	2.84	21	5.66
Rectal and large intestinal atresia/stenosis	3	2.60	38	3.17	19	5.12
Pyloric stenosis	34	29.42	281	23.45	68	18.32
Hirschsprung's disease (congenital megacolon)	4	3.46	38	3.17	8	2.16
Biliary atresia	2	1.73	10	0.83	1	0.27
Genitourinary						
Renal agenesis/hypoplasia	7	6.06	35	2.92	17	4.58
Bladder exstrophy	0	0.00	6	0.50	0	0.00
Obstructive genitourinary defect	23	19.90	327	27.29	100	26.94
Hypospadias and Epispadias	49	77.42	471	73.57	205	101.25
Musculoskeletal						
Reduction deformity, upper limb	3	2.60	19	1.59	8	2.16
Reduction deformity, lower limb	1	0.87	7	0.58	2	0.54
Gastroschisis/Omphalocele	13	11.25	47	3.92	10	2.69
Congenital hip dislocation	14	12.12	100	8.35	43	11.59
Diaphragmatic hernia	8	6.92	20	1.67	10	2.69
Chromosomal						
Trisomy 13	0	0.00	7	0.58	3	0.81
Down syndrome	8	6.92	98	8.18	124	33.41
Trisomy 18	1	0.87	6	0.50	6	1.62
Other						
Fetal alcohol syndrome	3	2.60	11	0.92	4	1.08

* Rates are per 10,000 live births

Figure 1. Gender-specific Prevalence Rates by System, Connecticut, 2001-04

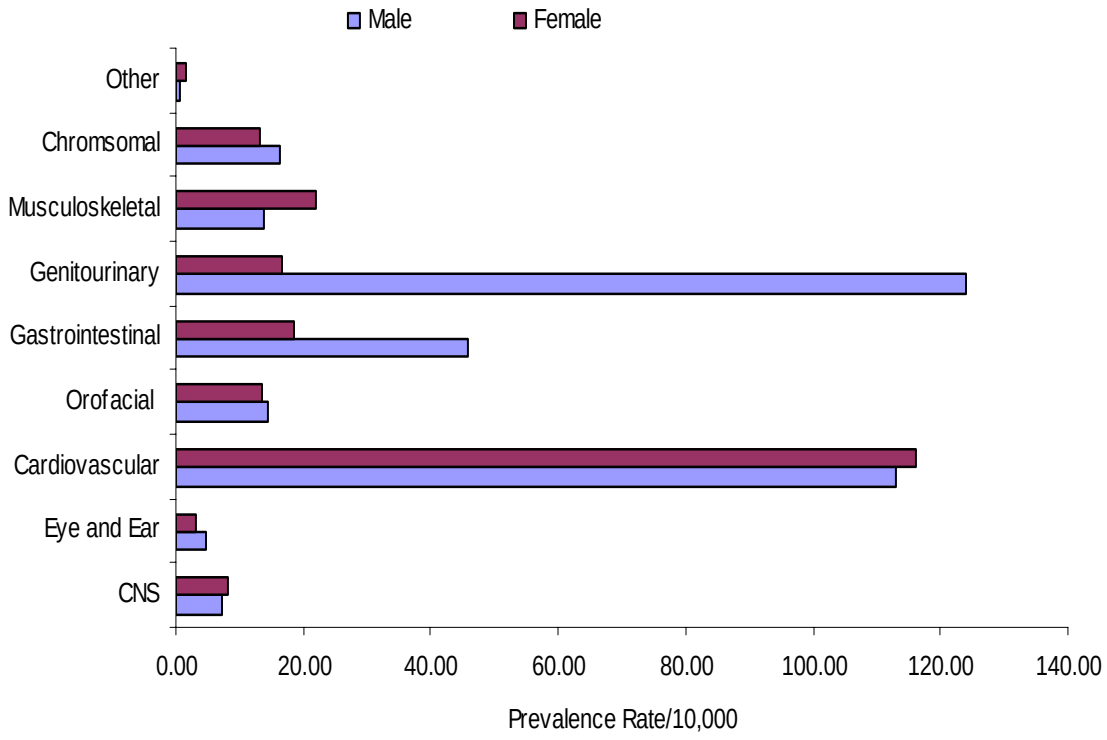


Figure 2. Race/Ethnicity-specific Prevalence Rates by System, Connecticut, 2001-04

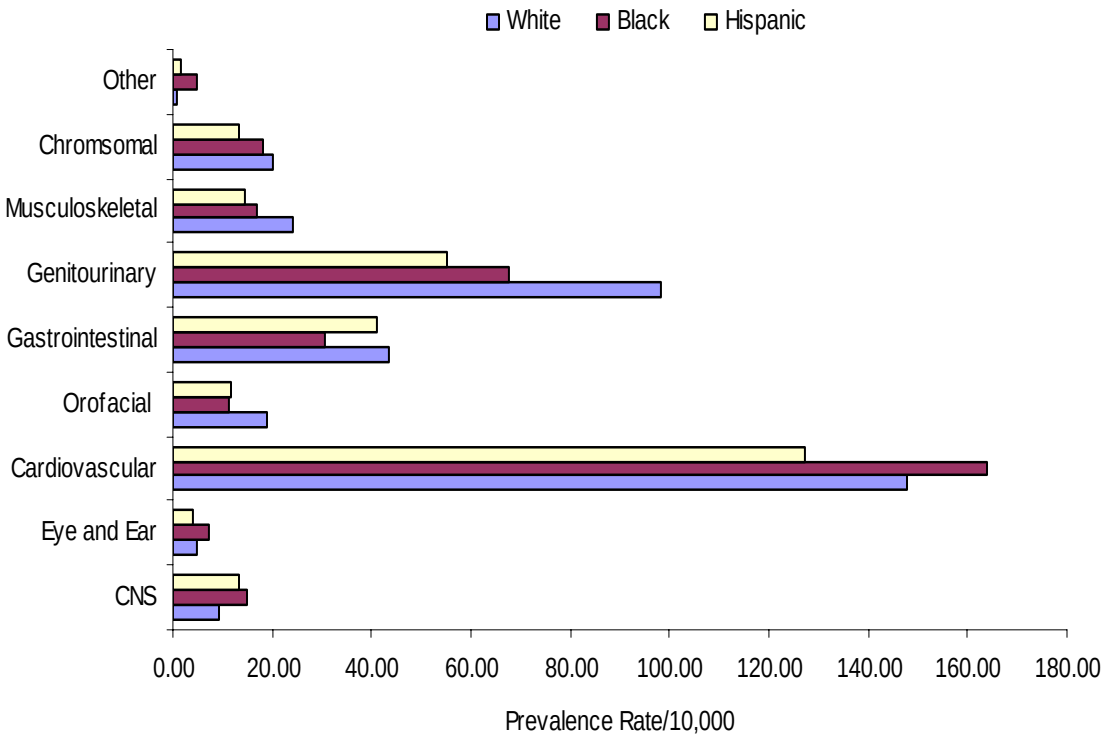


Figure 3. Maternal Age-specific Prevalence Rates by System, Connecticut, 2001-04

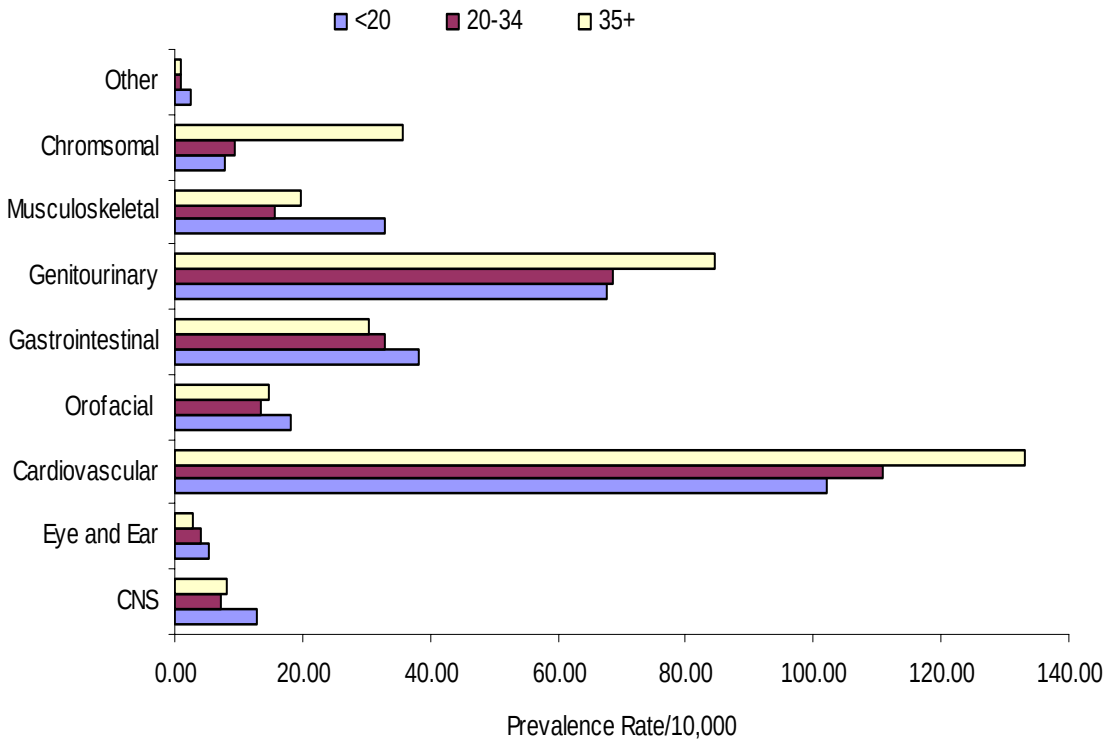
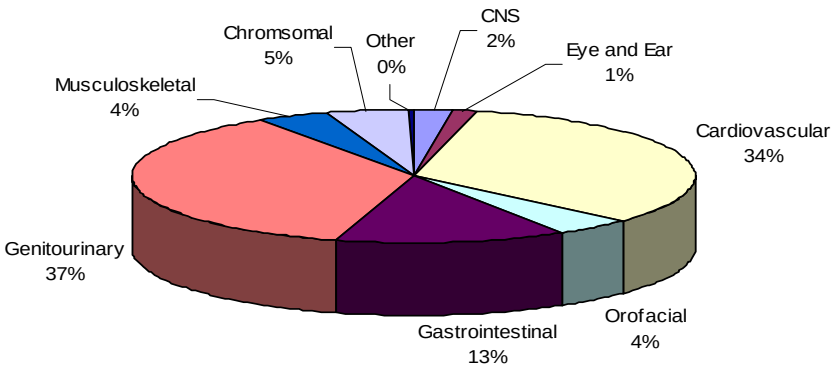


Figure 4. Distribution of Birth Defects Prevalence by System and Gender, Connecticut,
2001-04

Male
(n=2,954)



Female
(n=1,758)

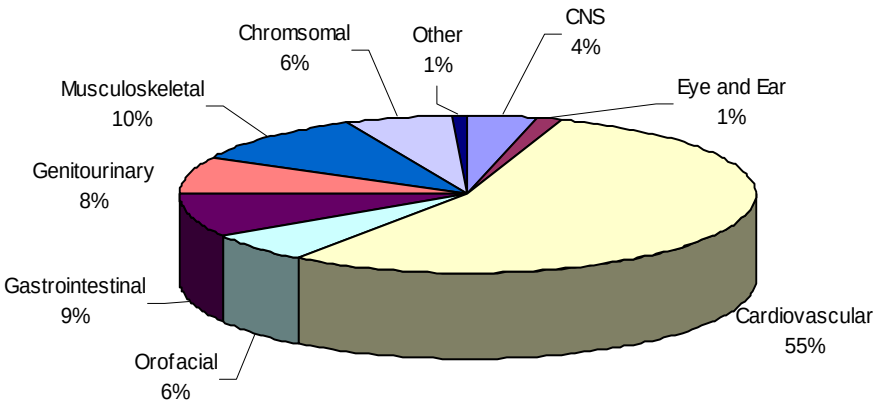


Table 5. Comparison of Prevalence Rates for Selected Birth Defects between CT and National Estimates[12]

	Connecticut 2001-04			National Estimates 1999-2001		
	Rate*	95% LCL	95% UCL	Rate	95% LCL	95% UCL
Central Nervous System						
Anencephalus	0.18**	0.00	0.38	1.10	0.97	1.25
Spina bifida without anencephalus	3.07	2.23	3.90	3.29	3.06	3.54
Encephalocele	0.71	0.31	1.11	0.65	0.55	0.76
Eye and Ear						
Anophthalmia/microphthalmia	0.77	0.35	1.18	0.99	0.87	1.14
Cardiovascular						
Common truncus	0.77	0.35	1.18	0.73	0.62	0.85
Transposition of great arteries	4.07	3.11	5.03	3.57	3.33	3.83
Tetralogy of Fallot	4.84	3.79	5.89	4.20	3.94	4.48
Endocardial cushion defect	3.89	2.95	4.83	3.12	2.90	3.37
Hypoplastic left heart syndrome	2.07	1.38	2.75	2.51	2.31	2.73
Orofacial						
Cleft palate without cleft lip	6.31	5.12	7.51	4.49	4.22	4.78
Cleft lip with and without cleft palate	6.73	5.49	7.96	8.05	7.69	8.44
Gastrointestinal						
Esophageal atresia/tracheoesophageal fistula	3.36	2.49	4.24	2.35	2.16	2.56
Rectal and large intestinal atresia/stenosis	3.54	2.64	4.44	3.73	3.48	4.00
Musculoskeletal						
Reduction deformity, upper limb	1.77	1.14	2.40	1.74	1.57	1.92
Reduction deformity, lower limb	0.59	0.22	0.96	1.03	0.91	1.18
Diaphragmatic hernia	2.24	1.53	2.96	2.90	2.68	3.13
Chromosomal						
Trisomy 13	0.59	0.22	0.96	0.90	0.79	1.04
Down syndrome	13.57	11.82	15.32	11.82	11.37	12.29
Trisomy 18	0.77	0.35	1.18	1.64	1.48	1.82

* Rates are per 10,000 live births

** Shaded cells indicate statistically significantly different from the National Estimates

Central Nervous System

Central nervous system (CNS) defects involve the brain, spinal cord, and associated tissues. They include neural tube defects (anencephaly, spina bifida and encephalocele), microcephalus and hydrocephalus. The table below gives the four-year prevalence rates for each defect, followed by a description of the condition. A comparison of CNS defect prevalence rates by county is also available. Taking folic acids during pregnancy can prevent some birth defects, especially neural tube defects. CDC and the U.S. Public Health Service urge every woman who could become pregnant to get 400 micrograms (400 mcg) of synthetic folic acid every day.

41.6% (95% CI: 38.78-44.42) of survey respondents took multivitamins everyday during the month before pregnancy. 73.6% (95% CI: 69.83-77.27) of these women took multivitamins containing 400 micrograms of folic acids.

Table 6. Frequency and Prevalence Rates of Central Nervous System Defects, CT, 2001-04

	ICD-9-CM Codes	Frequency	Rate/10,000
Anencephalus	740.0-740.1	3	0.18
Spina bifida without anencephalus	741.00-741.93	52	3.07
Hydrocephalus without Spina bifida	742.3	0	0.00
Encephalocele	742.0	12	0.71
Microcephalus	742.1	68	4.01

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.

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).

Hydrocephalus The abnormal accumulation of fluid within the spaces of the brain.

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

Microcephalus The congenital smallness of the head, with corresponding smallness of the brain.

Figure 5. Central Nervous System Defects by Year, CT, 2001-04

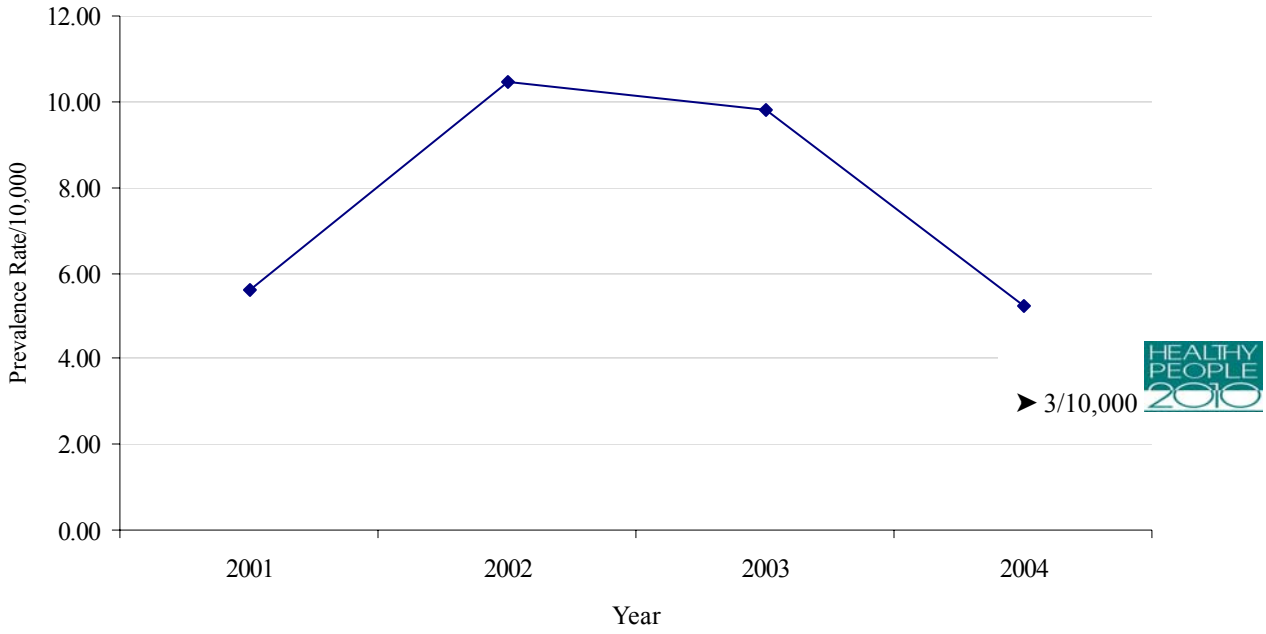
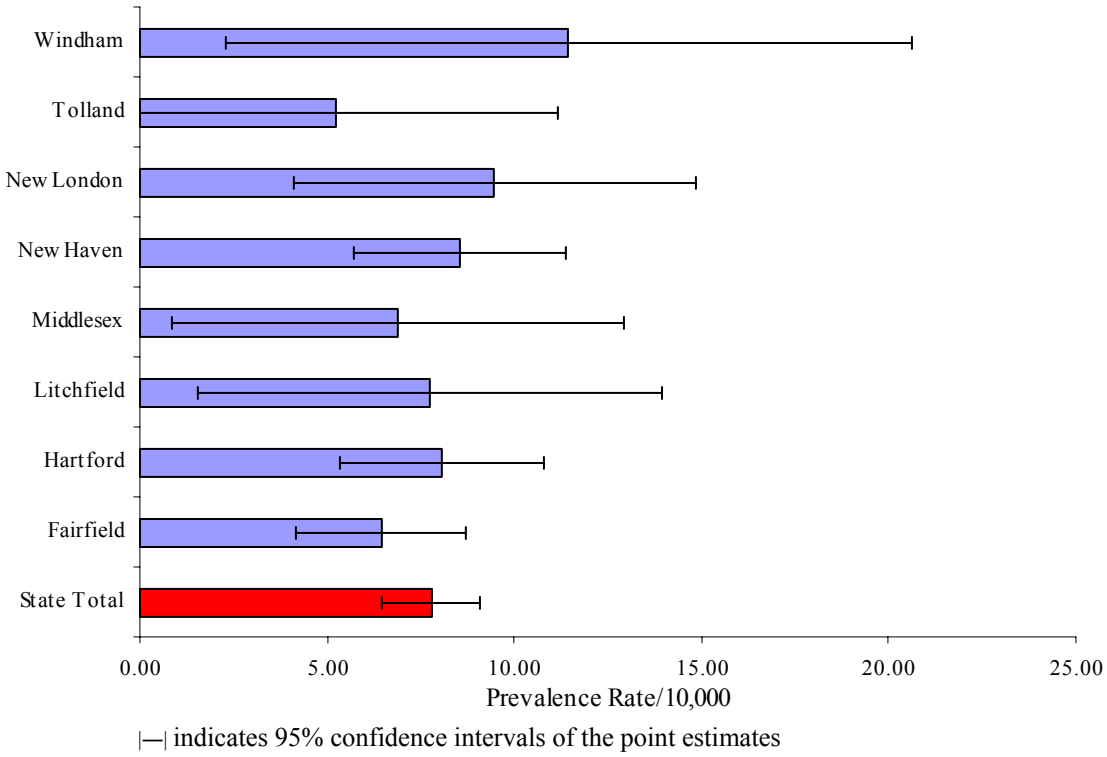


Table 7. Frequency and Prevalence of Central Nervous System Defects by County, CT, 2001-04

	Frequency	Rate/10,000	95% LCL	95% UCL
Fairfield	31	6.44	4.17	8.71
Hartford	34	8.09	5.37	10.81
Litchfield	6	7.76	1.55	13.97
Middlesex	5	6.90	0.85	12.95
New Haven	35	8.55	5.72	11.38
New London	12	9.48	4.12	14.84
Tolland	3	5.23	0.00	11.15
Windham	6	11.45	2.29	20.60
State Total	132	7.78	9.10	1.33

Figure 6. Central Nervous System Defects by County, CT, 2001-04



Eye and Ear Defects

Eye and ear defects include anophthalmia/microphthalmia, congenital cataract, aniridia, and anotia/micortia. The table below gives the four-year prevalence rates for each defect, followed by a description of each condition. A comparison of eye and ear defect prevalence rates by county is also available.

Table 8. Frequency and Prevalence Rates of Eye and Ear Defects, CT, 2001-04

	ICD-9-CM Codes	Frequency	Rate/10,000
Anophthalmia/ microphthalmia	743.0, 743.1	13	0.77
Congenital cataract	743.30 – 743.34	41	2.42
Aniridia	743.45	0	0.00
Anotia/microtia	744.01, 744.23	12	0.71

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

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

Congenital cataract An opacity (clouding) of the lens existing at or dating from birth.

Aniridia The complete absence of the iris of the eye or a defect of the iris. Can be congenital or traumatically induced.

Anotia A congenital absence of one or both ears.

Microtia A small or maldeveloped external ear and atretic or stenotic external auditory canal.

Figure 7. Eye and Ear Defects by Year, CT, 2001-04

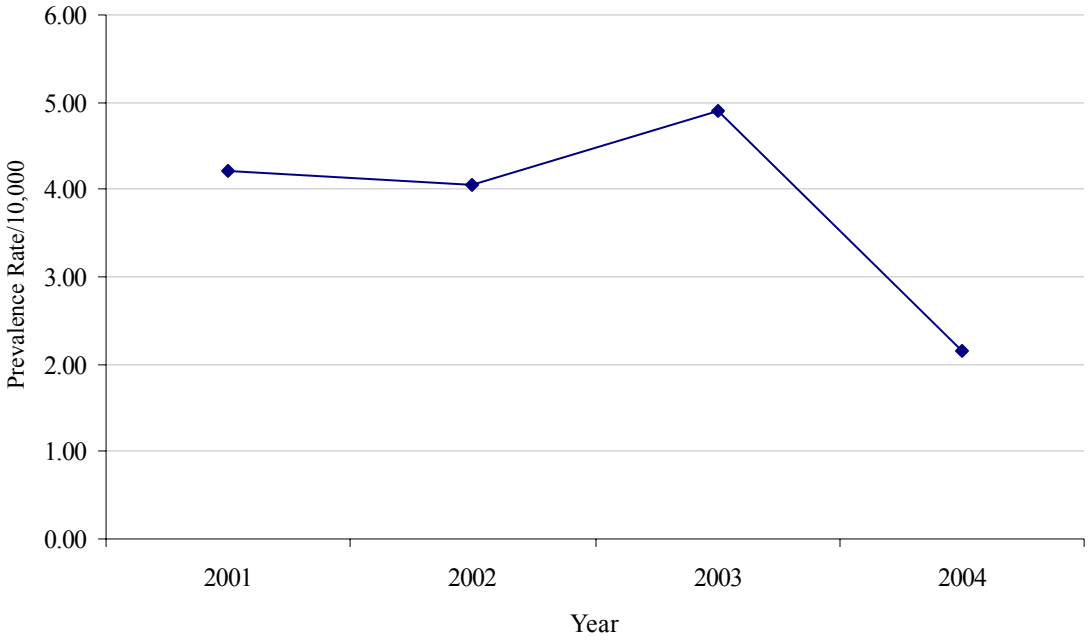
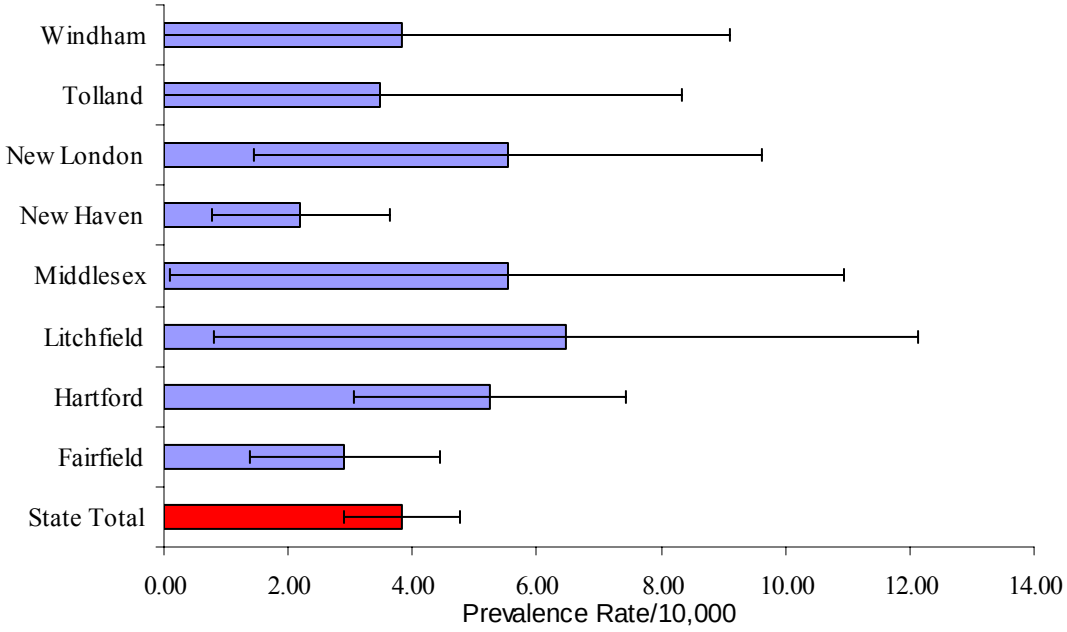


Table 9. Frequency and Prevalence of Eye and Ear Defects by County, CT, 2001-04

	Frequency	Rate/10,000	95% LCL	95% UCL
Fairfield	14	2.91	1.39	4.43
Hartford	22	5.23	3.05	7.42
Litchfield	5	6.47	0.80	12.13
Middlesex	4	5.52	0.11	10.93
New Haven	9	2.20	0.76	3.63
New London	7	5.53	1.43	9.63
Tolland	2	3.49	0.00	8.32
Windham	2	3.82	0.00	9.10
State Total	65	3.83	2.90	4.76

Figure 8. Eye and Ear Defects by County, CT, 2001-04



|—| indicates 95% confidence intervals of the point estimates

Cardiovascular System

Cardiovascular system defects involve the heart and circulatory systems. They are the most common group of birth defects. The table below gives the four-year prevalence rates for each defect, followed by a description of each condition. A comparison of cardiovascular system defect prevalence rates by county is also available.

Table 10. Frequency and Prevalence Rates of Cardiovascular System Defects, CT, 2001-04

	ICD-9-CM Codes	Frequency	Rate/10,000
Common truncus	745.0	13	0.77
Transposition of great arteries	745.1	69	4.07
Tetralogy of Fallot	745.2	82	4.84
Ventricular septal defect	745.4	786	46.38
Atrial septal defect	745.5	790	46.61
Endocardial cushion defect	745.6	66	3.89
Pulmonary valve atresia and stenosis	746.01, 746.02	97	5.72
Tricuspid valve atresia and stenosis	746.0	11	0.65
Ebstein's anomaly	746.2	7	0.41
Aortic valve stenosis	746.3	23	1.36
Hypoplastic left heart syndrome	746.7	35	2.07
Patent ductus arteriosus (BW>=2500 grams)	747.0	581	34.28
Coarctation of aorta	747.10	66	3.89

Common truncus	A congenital heart defect in which the common arterial trunk fails to divide into pulmonary artery and aorta. This is corrected surgically.
Transposition of great arteries	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. Immediate surgical correction is needed. When this is not associated with other cardiac defects, and not corrected, it is fatal.
Tetralogy of Fallot	A congenital cardiac anomaly consisting of four defects, ventricular septal defect, pulmonary valve stenosis, displacement of the aorta to the right, and hypertrophy of right ventricle. The condition is corrected surgically.
Ventricular septal defect	A congenital cardiac malformation in which there are one or several 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 unoxygenated blood. The openings vary in size and may resolve without treatment or require surgical intervention.
Atrial septal defects	A congenital cardiac malformation in which there are one or several openings in the atrial system (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 intervention. Also referred to as ostium secundum defect.
Endocardial cushion defect	A variety of septal defects (malformations of the walls separating the two atria and two ventricles of the heart) resulting from imperfect fusion of the endocardial cushions in the embryonic heart.
Pulmonary valve atresia and stenosis	A congenital heart condition characterized by the 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.
Tricuspid valve atresia and 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.

Ebstein's anomaly	A congenital heart defect in which the tricuspid valve is displaced downward into the right ventricle causing abnormal patterns of cardiac circulation.
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.
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 can be surgically repaired in a series of three procedures over a period of one year. Transplantation is also a treatment. This condition is usually fatal in the first month of life if not treated.
Patent ductus arteriosus	A blood vessel between the pulmonary artery and the aorta. This is normal in fetal life, but can cause problems after births, particularly in premature infants. This condition causes abnormal cardiac circulation and pressure in the heart during contractions. The vast majority close spontaneously and cause no problems. Medical or surgical correction may be done. This is only an abnormality if it causes significant medical problems.
Coarctation of 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. Surgical correction is recommended even for mild defects.

Figure 9. Cardiovascular System Defects by Year, CT, 2001-04

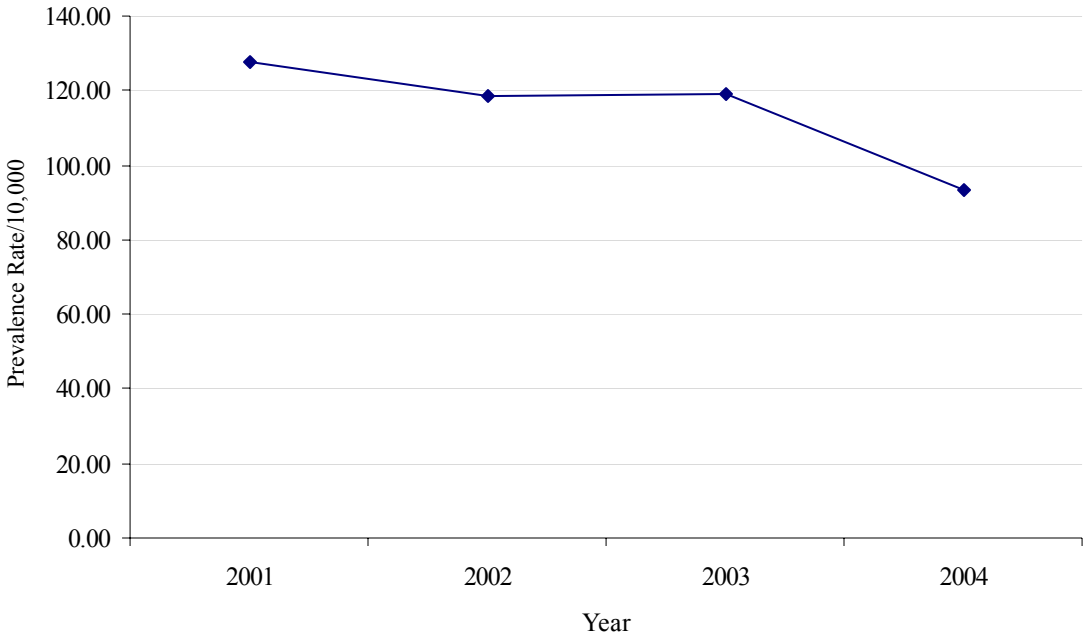


Table 11. Frequency and Prevalence of Cardiovascular System Defects by County, CT, 2001-04

	Frequency	Rate/10,000	95% LCL	95% UCL
Fairfield	527	109.50	100.15	118.85
Hartford	495	117.73	107.36	128.11
Litchfield	68	87.93	67.03	108.84
Middlesex	75	103.53	80.10	126.97
New Haven	479	117.01	106.53	127.49
New London	189	149.29	128.01	170.57
Tolland	57	99.39	73.59	125.19
Windham	51	97.29	70.59	123.99
State Total	1941	114.36	109.28	119.45

Figure 10. Cardiovascular System Defects by County, CT, 2001-04

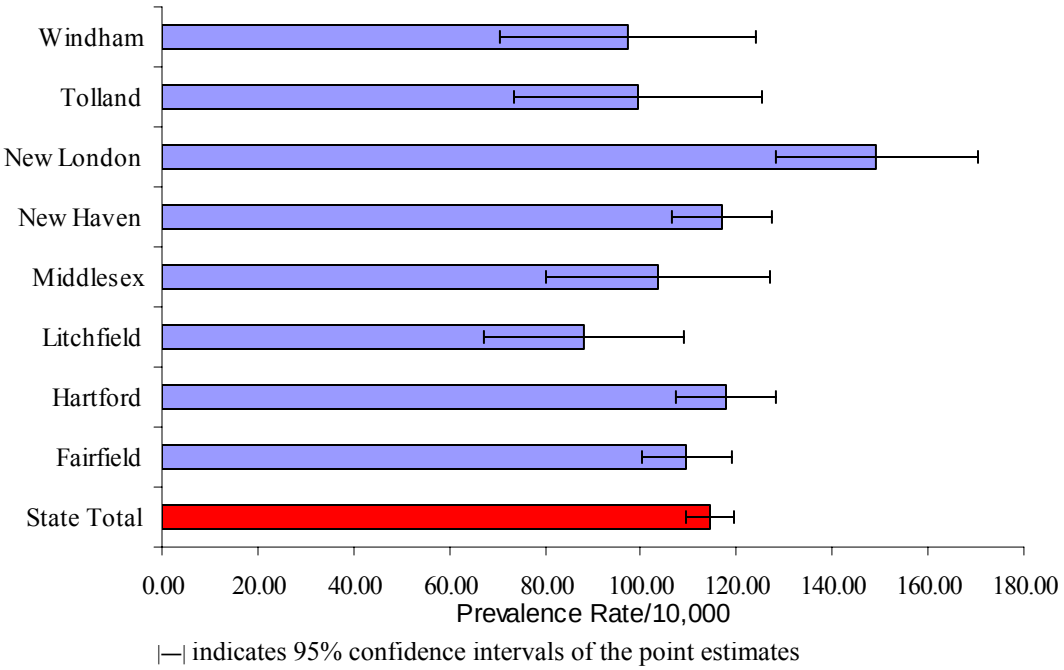
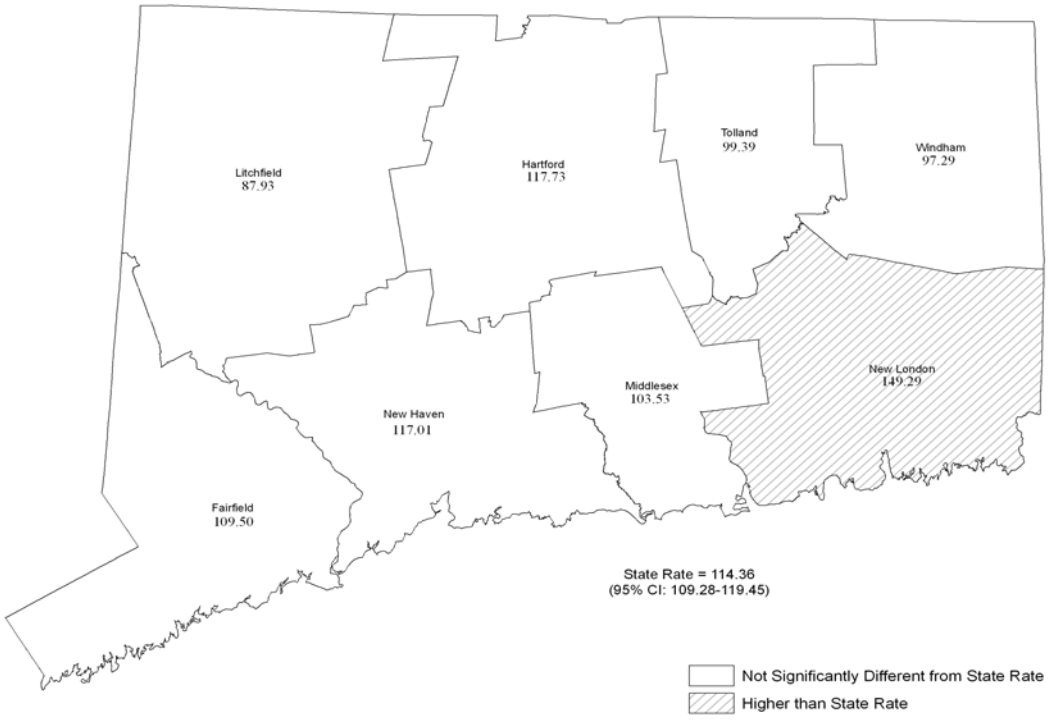


Figure 11. Cardiovascular System Defects by County, CT, 2001-04



Alimentary Tract Defects

Alimentary tract defects are made up of orofacial defects (cleft palate and lip, choanal atresia) and gastrointestinal defects (esophageal atresia, rectal and intestinal atresia and stenosis, and pyloric stenosis). Most of these defects can be repaired surgically. The table below gives the four-year prevalence rates for each defect, followed by a description of each condition. A comparison of alimentary tract defect prevalence rates by county is also available.

Table 12. Frequency and Prevalence Rates of Alimentary Tract Defects, CT, 2001-04

	ICD-9-CM Codes	Frequency	Rate/10,000
Cleft palate without cleft lip	749.0	107	6.31
Cleft lip	749.10-749.25	114	6.73
Choanal atresia	748.0	30	1.77
Esophageal atresia/tracheoesophageal fistula	750.3	57	3.36
Rectal and large intestinal atresia/stenosis	751.2	60	3.54
Pyloric stenosis	750.5	383	22.60
Hirschsprung's disease (congenital megacolon)	751.3	50	2.95
Biliary atresia	751.61	13	0.77

Cleft palate without cleft lip

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. Infants with this condition have difficulty feeding, and may use assistive devices for feeding. Surgical correction is begun as soon as possible. Children with cleft palates are at risk of hearing problems due to ear infections.

Cleft lip	The congenital failure of the fetal components of the lip to fuse or join, forming a groove or fissure in the lip. Infants with this condition can have difficulty feeding, and may use assistive devices for feeding. This condition is corrected when the infant can tolerate surgery.
Choanal atresia	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. Bilateral choanal atresia is a surgical emergency.
Esophageal atresia/tracheoesophageal fistula	A narrowing or incomplete formation of the esophagus. Usually a surgical emergency. Frequently associated with Tracheoesophageal Fistula.
Rectal and large intestinal atresia/stenosis	The absence, abnormal localization or blockage of the rectum, anus or large intestine. It may be corrected surgically or bypassed.
Pyloric stenosis	A narrowing of the pyloric sphincter at the outlet of the stomach. This causes a blockage of food from the stomach into the small intestine. Usually treated surgically.
Hirschsprung's disease (congenital megacolon)	The congenital absence of autonomic ganglia (nerves controlling involuntary and reflexive movement) in the muscles of the colon. This results in immobility of the intestines and may cause obstruction or stretching of the intestines. This condition is repaired surgically in early childhood by the removal of the affected portion of the intestine.
Biliary atresia	A congenital absence or underdevelopment of one or more of the ducts in the biliary tract. Correctable surgically.

Figure 12. Alimentary Tract Defects by Year, CT, 2001-04

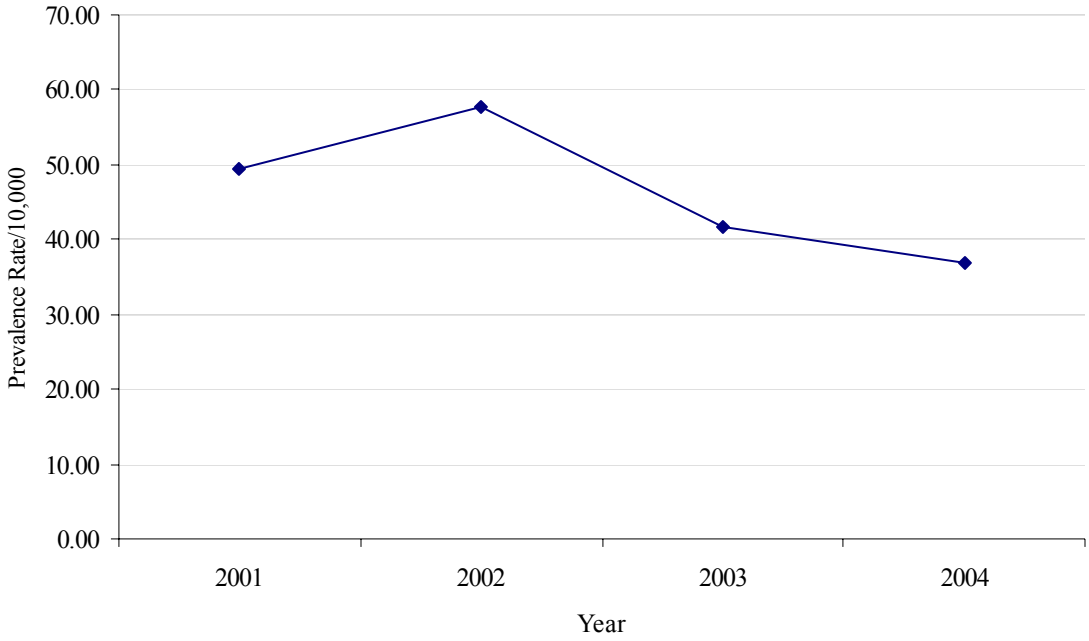
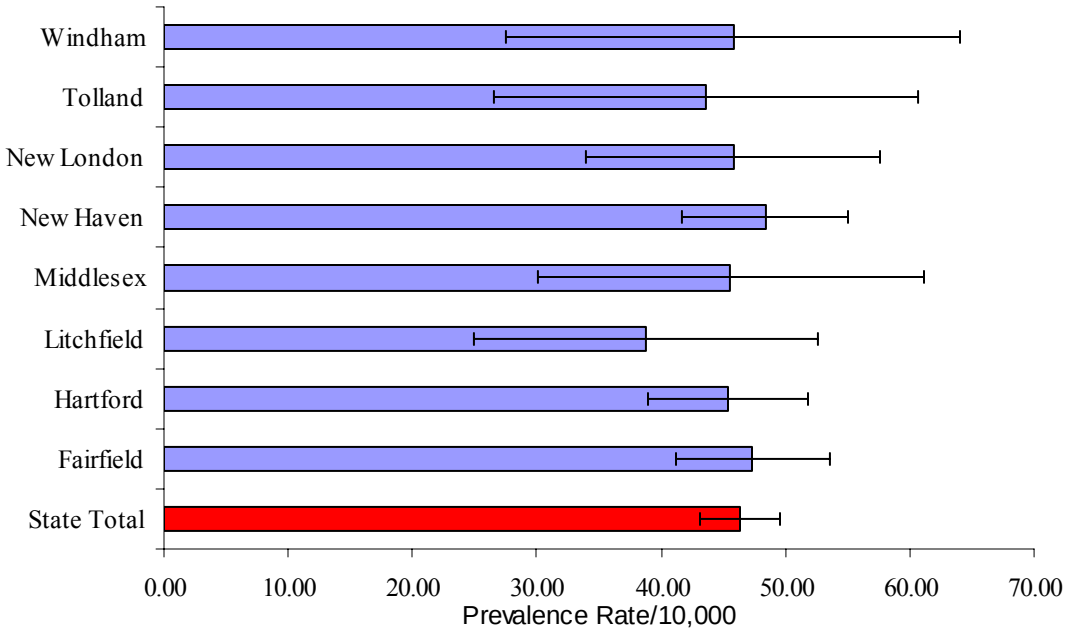


Table 13. Frequency and Prevalence of Alimentary Tract Defects by County, CT, 2001-04

	Frequency	Rate/10,000	95% LCL	95% UCL
Fairfield	228	47.37	41.22	53.52
Hartford	191	45.43	38.99	51.87
Litchfield	30	38.79	24.91	52.68
Middlesex	33	45.55	30.01	61.10
New Haven	198	48.37	41.63	55.10
New London	58	45.81	34.02	57.60
Tolland	25	43.59	26.50	60.68
Windham	24	45.78	27.47	64.10
State Total	787	46.37	43.13	49.61

Figure 13. Alimentary Tract Defects by County, CT, 2001-04



|—| indicates 95% confidence intervals of the point estimates

Genitourinary System

The defects in genitourinary system affect the male and female reproductive organs and urinary tracts. Some are relatively minor, fairly common defects that may be readily repaired by surgery. Others are more serious and potentially life-threatening malformations. The table below gives the four-year prevalence rates for each defect, followed by a description of each condition. A comparison of genitourinary system defect prevalence rates by county is also available.

Table 14. Frequency and Prevalence Rates of Genitourinary System Defects, CT, 2001-04

	ICD-9-CM Codes	Frequency	Rate/10,000
Renal agenesis/hypoplasia	753.0	59	3.48
Bladder exstrophy	753.5	6	0.35
Obstructive genitourinary defect	753.2, 753.6	450	26.55
Hypospadias and Epispadias	752.61, 752.62	725	83.45

Renal agenesis or dysgenesis The failure, or deviation, of embryonic development of the kidney.

Bladder exstrophy Incomplete closure of the anterior wall of the bladder and the abdominal cavity. The upper urinary tract is generally normal. Often associated with anorectal and genital malformations, and epispadias. Affected persons are at a markedly increased risk of bladder carcinoma (squamous cell). This condition is usually corrected surgically after birth.

Obstructive genitourinary defect Stenosis or atresia of the urinary tract at any level. Severity of the defect depends largely upon the level of the obstruction. Urine accumulates behind the obstruction and damages the organs.

Hypospadias A congenital defect 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 urinary sphincters are not defective so incontinence does not occur.

Epispadias A congenital defect in which the urinary meatus (urinary outlet) opens above (dorsal to) the normal position. The urinary sphincters are defective, so incontinence does occur. Surgical correction is aimed at correcting incontinence and permitting sexual functioning. The corresponding defect in females is rare.

Figure 14. Genitourinary System Defects by Year, CT, 2001-04

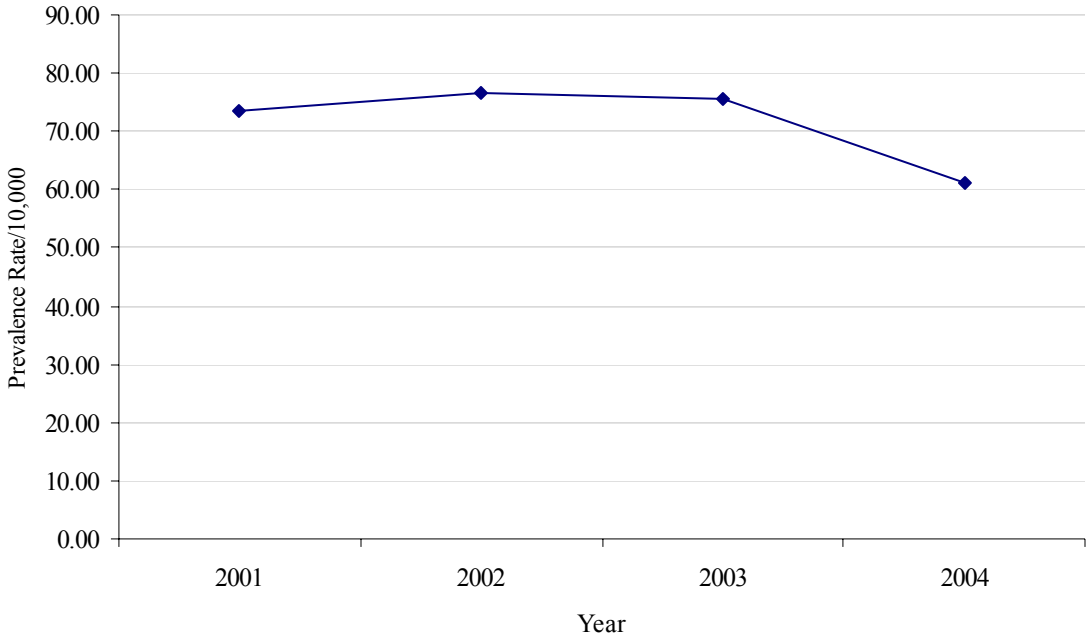


Table 15. Frequency and Prevalence of Genitourinary System Defects by County, CT, 2001-04

	Frequency	Rate/10,000	95% LCL	95% UCL
Fairfield	329	68.36	60.97	75.75
Hartford	363	86.34	77.46	95.22
Litchfield	58	75.00	55.70	94.31
Middlesex	55	75.92	55.86	95.99
New Haven	250	61.07	53.50	68.64
New London	79	62.40	48.64	76.16
Tolland	47	81.95	58.52	105.38
Windham	34	64.86	43.06	86.66
State Total	1215	71.59	67.56	75.61

Figure 15. Genitourinary System Defects by County, CT, 2001-04

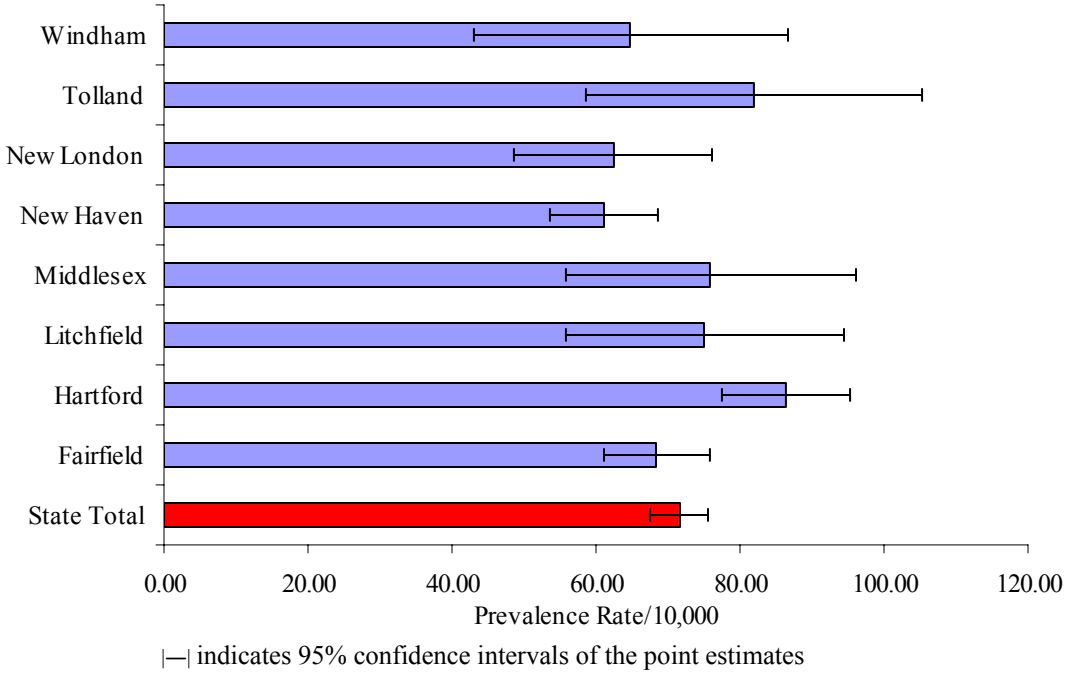
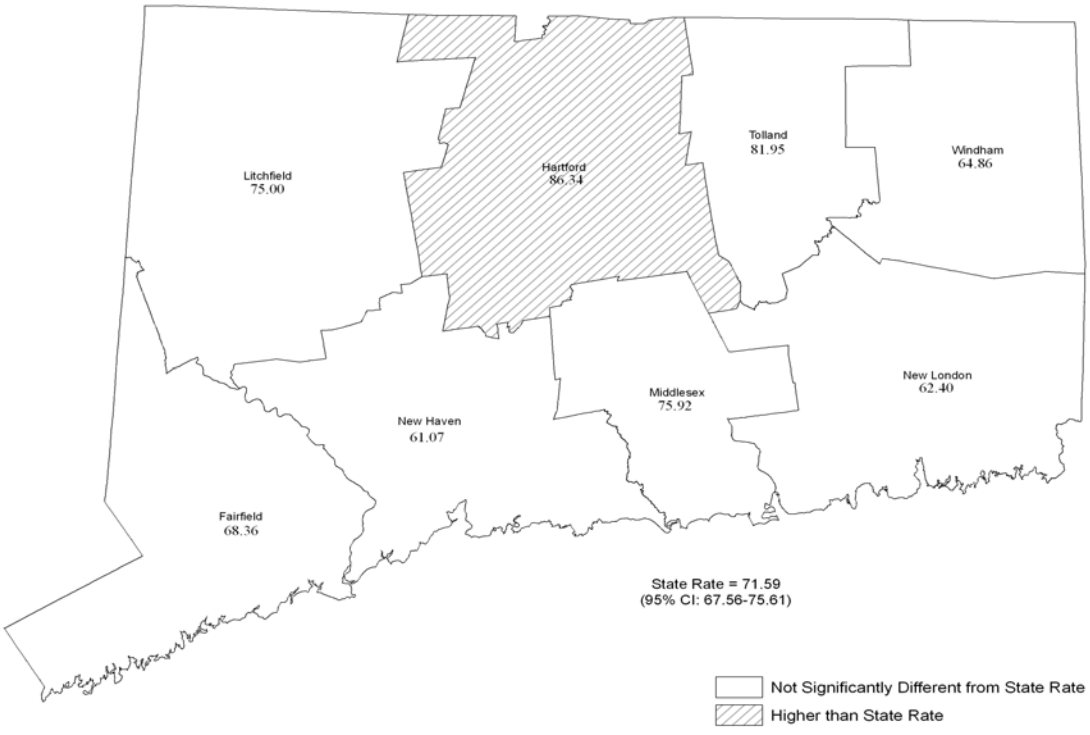


Figure 16. Genitourinary System Defects by County, CT, 2001-04



Musculoskeletal System

Musculoskeletal malformations make up a diverse group of defects that include congenital dislocation of the hip, a relatively common disorder, and several more rare and serious conditions. The table below gives the four-year prevalence rates for each defect, followed by a description of each condition. A comparison of musculoskeletal system defect prevalence rates by county is also available.

Table 16. Frequency and Prevalence Rates of Musculoskeletal System Defects, CT, 2001-04

	ICD-9-CM Codes	Frequency	Rate/10,000
Reduction deformity, upper limb	755.2	30	1.77
Reduction deformity, lower limb	755.3	10	0.59
Gastroschisis/Omphalocele	756.79	70	4.13
Congenital hip dislocation	754.30, 754.31, 754.35	157	9.26

Reduction deformity, upper limb — The congenital absence of a portion of the upper limb. There are two types of defects, transverse and longitudinal. Transverse defects appear like amputations, or like missing segments of the limb. Longitudinal defects are missing rays of the limb (e.g., a missing radius and thumb).

Reduction deformity, lower limb — The congenital absence of a portion of the lower limb. There are two types of defects, transverse and longitudinal. Transverse defects appear like amputations, or like missing segments of the limb. Longitudinal defects are missing rays of the limb (e.g., a missing tibia and great toe).

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

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

Congenital hip dislocation — A congenital defect in which the head of the femur does not articulate with the acetabulum of the pelvis because of an abnormal shallowness of the acetabulum. Treatment in early infancy consists of bracing of the joint to cause a deepening of the acetabulum.

Figure 17. Musculosketal System Defects by Year, CT, 2001-04

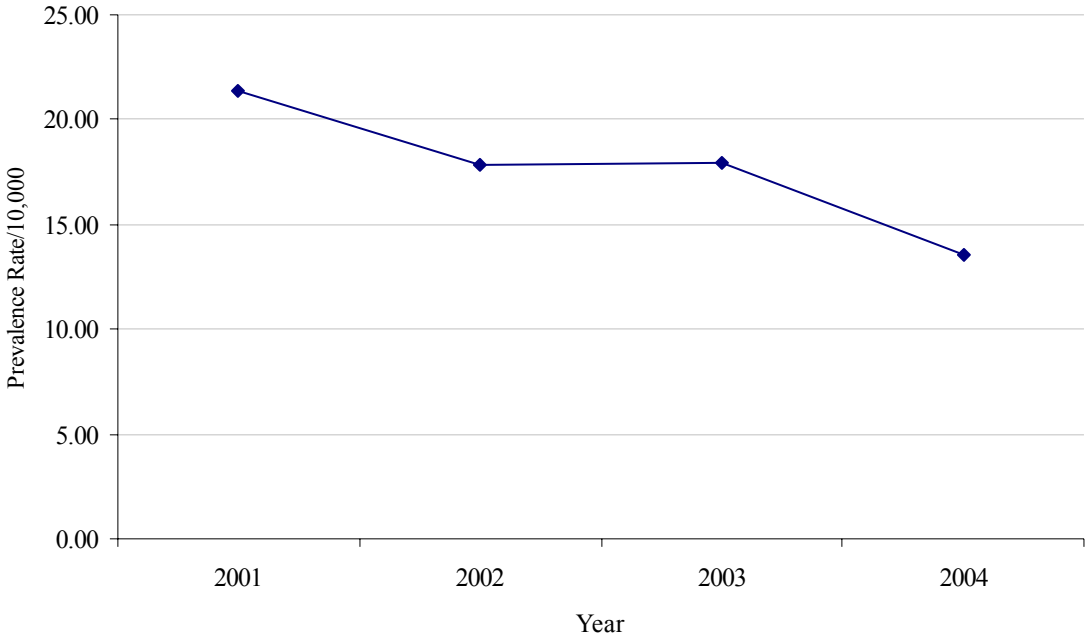
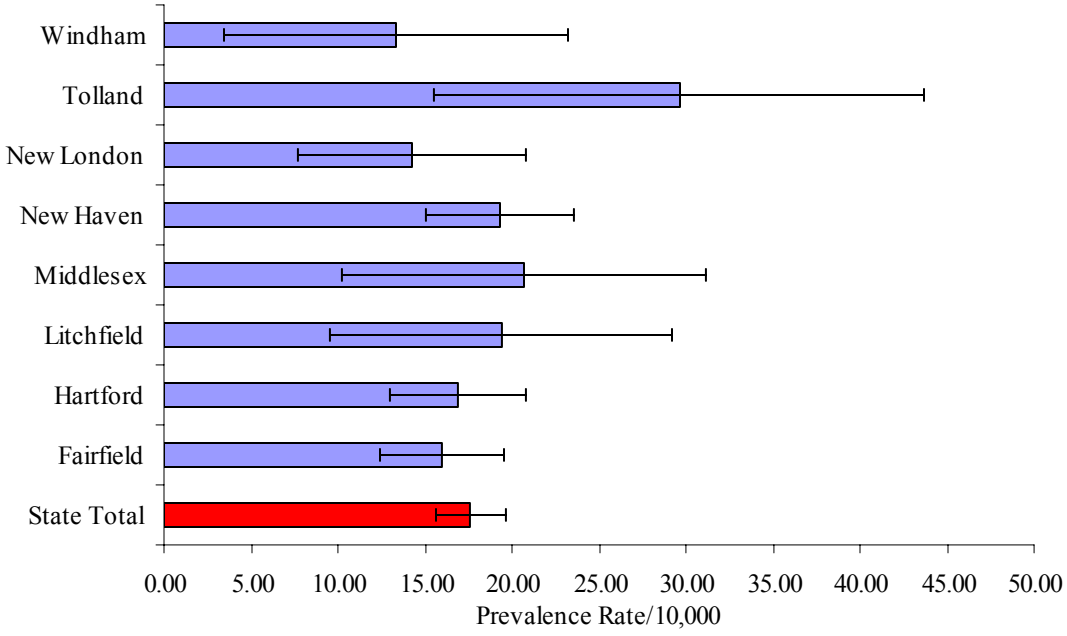


Table 17. Frequency and Prevalence of Musculoskeletal System Defects by County, CT, 2001-04

	Frequency	Rate/10,000	95% LCL	95% UCL
Fairfield	77	16.00	12.43	19.57
Hartford	71	16.89	12.96	20.82
Litchfield	15	19.40	9.58	29.21
Middlesex	15	20.71	10.23	31.19
New Haven	79	19.30	15.04	23.55
New London	18	14.22	7.65	20.79
Tolland	17	29.64	15.55	43.73
Windham	7	13.35	3.46	23.25
State Total	299	17.62	15.62	19.61

Figure 18. Musculoskeletal System Defects by County, CT, 2001-04



|—| indicates 95% confidence intervals of the point estimates

Chromosomal Defects

Chromosomal anomalies are disorders that usually arise from abnormal numbers of chromosomes or from breaks or deletions in specific fragments of the chromosomes. Each is associated with a characteristic pattern of defects that arises as a consequence of the underlying chromosomal abnormality. Congenital heart defects (especially septal defects) are very common among these infants and are a major cause of death. The table below gives the four-year prevalence rates for each defect, followed by a description of each condition. A comparison of chromosomal defect prevalence rates by county is also available.

Table 18. Frequency and Prevalence Rates of Chromosomal Defects, CT, 2001-04

	ICD-9-CM Codes	Frequency	Rate/10,000
Trisomy 13	758.1	10	0.59
Down syndrome	758.0	230	13.57
Trisomy 18	758.2	13	0.77

Trisomy 13 (Patau syndrome)

The chromosomal abnormality caused by an extra chromosome 13. The extra copy can be free-lying, or can be attached to some other chromosome. Patau syndrome can occur in *mosaic* so that there is a population of normal cells and a population of trisomy 13 cells. Patau syndrome is characterized by impaired midline facial development, cleft lip and palate, polydactyly and mental retardation. Most infants do not survive beyond six months of life.

Trisomy 21 (Down syndrome)

The chromosomal abnormality characterized by an extra copy of chromosome 21. In rare cases this syndrome is caused by *translocation*. The extra copy can be free-lying, or can be attached to some other chromosome, most frequently number 14. Down syndrome can occur in *mosaic* so that there is a population of normal cells and a population of trisomy 21 cells. Down syndrome is characterized by moderate to severe mental retardation, sloping forehead, small ear canals, flat bridged nose and short fingers and toes. One third of infants have congenital heart disease, and one third have duodenal atresia. (Both can be present in the same infant.) Affected people can survive to middle or old age. There is an increased incidence of Alzheimer disease in adults with Down syndrome.

Trisomy 18 (Edwards syndrome)

The chromosomal abnormality characterized by an extra copy of chromosome 18. The extra chromosome can be free lying or attached to another chromosome. Trisomy 18 can occur in *mosaic*. Edwards 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.

Figure 19. Chromosomal Defects by Year, CT, 2001-04

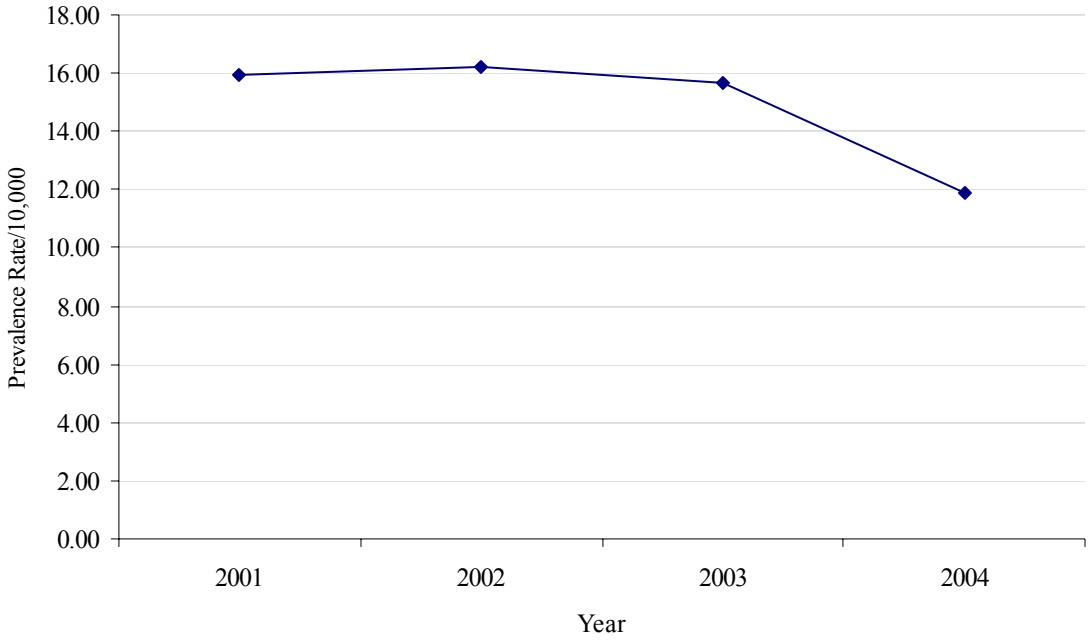
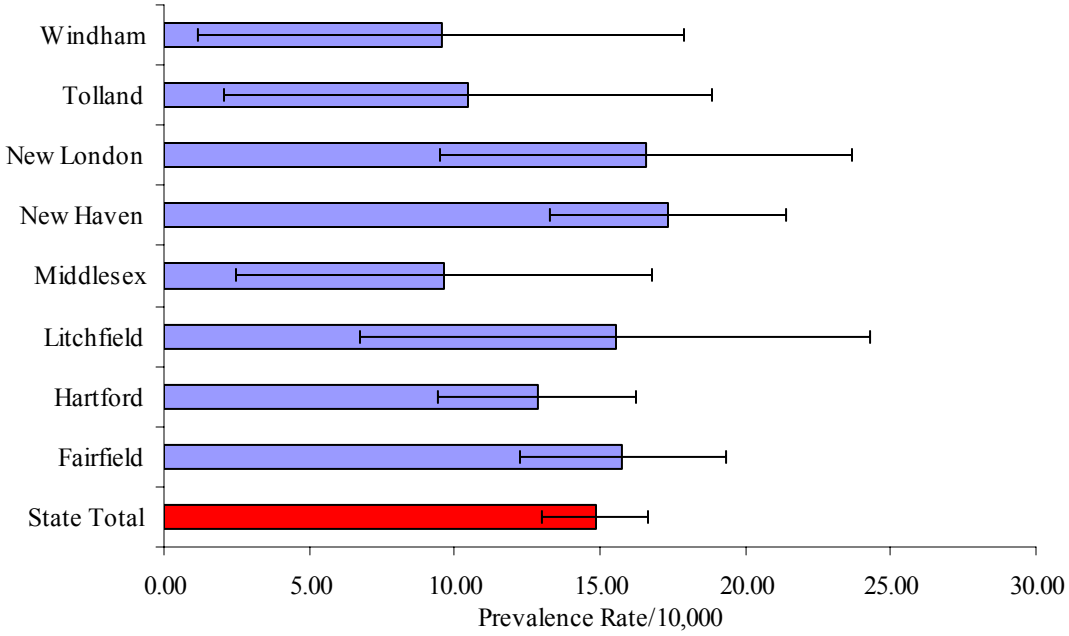


Table 19. Frequency and Prevalence of Chromosomal Defects by County, CT, 2001-04

	Frequency	Rate/10,000	95% LCL	95% UCL
Fairfield	76	15.79	12.24	19.34
Hartford	54	12.84	9.42	16.27
Litchfield	12	15.52	6.74	24.30
Middlesex	7	9.66	2.50	16.82
New Haven	71	17.34	13.31	21.38
New London	21	16.59	9.49	23.68
Tolland	6	10.46	2.09	18.83
Windham	5	9.54	1.18	17.90
State Total	252	14.85	13.01	16.68

Figure 20. Chromosomal Defects by County, CT, 2001-04



|—| indicates 95% confidence intervals of the point estimates

CONCLUSIONS

Overall, the birth defects prevalence in Connecticut for 2001-04 is comparable to that of the national estimates. The five most common birth defects for 2001-04 are hypospadias/epispadias, septal defect, patent ductus arteriosus, obstructive genitourinary defect, and pyloric stenosis. The prevalence of birth defects varies by sex, race/ethnicity, maternal age, county, and system over this four-year period. While results for birth defects differ by risk factors described in this report, some of the reported birth defects prevalence rates are not stable for interpretation due to small numbers. Each of the risk factors tabulated in this report are described below.

Sex

While the majority of birth defects do not substantially differ by sex of the infants, some conditions are more highly associated with sex. In general, birth defects of genitourinary and gastrointestinal systems are higher in males than in females. Common birth defects seen in both sexes include septal defect, patent ductus arteriosus, Down Syndrome, cleft palate, cleft lip, and pulmonary valve atresis and stenosis. The most common birth defects seen in males are hypospadias/epispadias, septal defect, and obstructive genitourinary defect; and the most common birth defects seen in females are septal defect, patent ductus arteriosus, and congenital hip dislocation.

Mother's Race/Ethnicity

The prevalence of birth defects varies by mother's race and Hispanic ethnicity. In general, birth defects of cardiovascular system are higher in Blacks than in Whites and Hispanics. In contrast, birth defects of genitourinary system are higher in Whites than in Blacks and Hispanics. The most common birth defects in Whites include hypospadias/epispadias, septal defects, and patent ductus arteriosus. In Blacks, the most common birth defects include hypospadias/epispadias, septal defects, and patent ductus arteriosus. The most common birth defects in Hispanics include hypospadias/epispadias, septal defect, and patent ductus arteriosus.

Maternal Age

The prevalence of birth defects varies by maternal age group. In general, prevalence of birth defects is higher in the maternal age group of 35 or older for birth defects of cardiovascular system, genitourinary system, and chromosomes. As expected, there is a strong association of Down Syndrome with advanced maternal age. Women 35 years or older had a Down Syndrome prevalence rate of 33.41 per 10,000 live births. This rate is five times that of any other maternal age group.

County

The prevalence of birth defects varies by county. The prevalence for cardiovascular system defects is statistically significantly higher in New London County (149.29/10,000) than the state average (114.36/10,000), and the prevalence for genitourinary system defects is statistically significantly higher in Hartford County (86.34/10,000) than the state average (71.59/10,000). This information is useful for identifying potential risks of birth defects associated with different life styles and possible environmental/occupational exposures.

The benefits of birth defects surveillance data to public health programs include: identifying population groups or geographic areas with a higher prevalence of birth defects; identifying children in need of services to ensure that they and their families are referred appropriately; evaluating service utilization by children with birth defects and their families; and planning the location of services for particular conditions in areas of highest frequency. An important use of surveillance data is monitoring birth defects trends following the initiation of prevention programs in order to evaluate their effectiveness.

The data collection in CTBDR has improved over the years through the incorporation of multiple sources of data. Continued efforts will be made to further enhance data collection, in terms of completeness, timeliness, and accuracy. CTBDR will further benefit from the collaboration with other programs that provide services to these children and their families, for example, Newborn Screening program and Children and Youth with Special Health Care Needs program (CYSHCN). The collaboration among these programs will facilitate the record linkage for more comprehensive data collection.

Information collected in the CTBDR has not been widely used to refer specific children and their families to appropriate services. Therefore, a need exists to establish referral networks as a resource for children and their families to learn about and access the available medical services, community programs, and social support. This can be achieved through the collaboration with other programs within DPH and other stakeholders to connect affected children and their families with appropriate services in a timely fashion. Further program evaluation and planning can be conducted through analyzing data collected in the CTBDR.

There are a certain number of steps a woman can take to reduce the risk of having a baby with a birth defect. An important step is to have a preconception visit with her health care provider. During this visit, the provider can identify, and often treat, health conditions that can pose a risk in pregnancy, such as high blood pressure or diabetes. The provider can provide advice on lifestyle factors, such as quitting smoking and avoiding alcohol, and occupational exposures that can pose pregnancy risks. The provider also can make sure that any medications a woman takes are safe during pregnancy. All of these steps help prevent birth defects.

Recommendations to improve preconception health and health care are available from CDC for consumers, public health and clinical providers, researchers, and policy makers [13]. In Connecticut, these recommendations are being implemented through a partnership DPH has with the Hartford Health Department as part of their CDC/CityMatch technical assistance grant to address systems of care as it relates to preconception and interconceptual care.

Consumer/provider education is a critical component in birth defects prevention. This can be accomplished through the dissemination of information on birth defects statistics and prevention at professional meetings and conferences. Pamphlets and brochures with information on birth defects prevention can be distributed at the providers' office to reach consumers. Such information can also be made available in the media like newspaper and public television.

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FREQUENTLY ASKED QUESTIONS

1. *I am interested in looking at birth defects rates for my county by year, but when I read the report, I find only 4-year rates by system. Why?*

   Calculating a rate based on less than 20 events in the numerator creates an unstable estimate that is not statistically reliable and varies greatly from year to year by chance alone. Therefore, generating rates based on small numbers can lead to misinterpretation. For this reason, 4 years of data are aggregated to create a more stable rate that can be used to compare birth defects by type or another variable of interest.

2. *I would like to see the different types of birth defects in my town, but cannot find this information in the report. Why doesn't this report show town-level data and where can I find this information?*

   Connecticut is a small state with 169 cities and towns. In a given year, the number of newly diagnosed birth defects is too small to generate meaningful results on a town level. Data are summarized by county to provide information that is more detailed than state level data. For information on a town level, please contact the Connecticut Birth Defects Registry at 860-509-8057.

3. *I have noticed many birth defects cases on my street and in my town. Who can I contact at the state if I want this investigated further?*

   One in every 33 babies is born with a birth defect in this country. Although birth defects are rare events, it is still possible to see the occurrence of cases in a neighborhood. Even in a neighborhood where several birth defects cases are observed, they might not be the same type of birth defects or share the same morphology. However, there are rare circumstances where birth defects clusters are observed. Please contact the Connecticut Birth Defects Registry (860-509-8057) for concerns on non-environmental birth defects clusters. For information or investigation of a suspected, environmentally related birth defects cluster, please contact the Environmental Health Program at 860-509-7740.

4. *Where can I get more detailed information on birth defects treatment, prevention, and research?*

   The Connecticut Birth Defects Registry (CTBDR) can be reached at 860-509-8057 for information regarding this report. The National Center on Birth Defects and Developmental Disability (NCBDDD) at CDC (http://www.cdc.gov/ncbddd) and the National Birth Defects Prevention Network (NBDPN) (http://www.nbdpn.org) are good resources for information on all types of birth defects.

5. *How is birth defects defined in this report?*

   Birth defects, as defined by the March of Dimes, is an abnormality of structure, function, or body metabolism that is present at birth and results in physical or mental disability. Most birth defects are included within the range of diagnosis codes 740.0 to 759.9, as defined by the International Classification of Disease (ICD), 9th Revision. The specific conditions included in this report are those recommended by the CDC's National Birth Defects Prevention Network (NBDPN) for surveillance and reporting.

6. *I am interested in some types of birth defects, but I do not see mention of them in this report. Why? How do I get this information?*

   The birth defects included in this report are those recommended by the CDC's National Birth Defects Prevention Network (NBDPN) for surveillance and reporting. Many rare types of birth defects are not included in this report because the numbers are too small for meaningful tabulations. Please refer to NCBDDD or NBDPN website for more information.

7. *This report summarizes data from 2001-04. It is now 2007. Why is there such a long time between data acquisition and publication?*

   There is a considerable lag time in the data acquisition process. Data from birth hospitals, Vital Records, and the CT Hospital Association (hospital discharge data) are used to provide information to the CTBDR. Each data source undergoes a quality assurance process before it is released to third parties. After data are obtained, information from these three sources are reconciled and consolidated into records stored in the CTBDR, a process that is time-intensive. All of these factors contribute to the length of time between the events and reporting.

8. *Why is the prevalence rather than incidence used in this report? How are they different?*

   The occurrence of birth defects is commonly reported as prevalence. Prevalence is calculated as the number of birth defect cases born at a point in time per 10,000 live births. Prevalence tables include the number of cases found, the estimated prevalence rate per 10,000 live births, and the 95% confidence interval for that rate. The incidence (new cases) of birth defects (based upon the number of embryos conceived within a year) is not easily measured because both the number of conceptions that occur and the number of these conceptions resulting in a defect are not known.

APPENDIX A: TECHNICAL NOTES

1. **Data Sources**
   The Connecticut Birth Defects Registry uses 3 different data sources to consolidate records in the database, including reporting from birth hospitals, vital records, and in-patient hospital discharge data. Vital records data were used as the source of information for mother's date of birth, race/ethnicity, and town of residence for the analysis.

2. **Prevalence**
   Prevalence is defined as the number of individuals with a disease or condition at a specific time divided by the number of individuals at risk. The numerator is the number of cases of birth defects. Since the preferred denominator is all pregnancies and since the number of pregnancies cannot be determined, the number of total births is normally used as an approximation.

3. **Rate**
   The rates provided in this report are estimations of the proportion of infants born with birth defects. This rate is usually expressed as birth defects births per 10,000 births and is calculated by the formula:

$$\text{Rate} = \frac{\text{Number of births with selected birth defects}}{\text{Number of live births}} * 10{,}000$$

4. **Confidence Interval (CI)**
   The confidence interval is a method of assessing the magnitude and stability of a rate or ratio. The 95% CI represents a range of values that has a 95% probability of including the true rate or ratio. Observed rates are subject to statistical variation. Thus, even if the underlying risk of an infant being born with a birth defect is identical in two subpopulations, the observed rates for the subpopulations may differ because of random variation. The confidence interval describes the precision of the observed rate as an estimate of the underlying risk of being born with a birth defect, with a wider interval indicating less certainty about this estimate. The width of the interval reflects the size of the subpopulation and the number of cases of birth defects. Smaller subpopulations with fewer defects lead to wider confidence intervals. The 95% confidence intervals used in the report are based on the Poisson distribution.

5. **Disparities on Race and Ethnicity**
   The Registry follows the recommendation of the National Center for Health Statistics of classifying births according to the self-reported race/ethnicity of the mother. The Connecticut Vital Records record mother's race and ethnicity, including Hispanic ethnicity, and was used to more accurately calculate Hispanic specific rates of birth

defect prevalence. Race/ethnicity is a self-reported item and is subject to the usual limitations of this type of information.

APPENDIX B: SOURCES OF ADDITIONAL INFORMATION

For more information on birth defects, risk factors or prevention strategies please refer to the following resources:

- International Clearinghouse for Birth Defects Surveillance and Research
  http://www.icbdsr.org

- The National Center on Birth Defects and Developmental Disability at CDC
  http://www.cdc.gov/ncbddd

- National Birth Defects Prevention Network
  http://www.nbdpn.org

- The Teratology Society
  http://www.teratology.org

- The March of Dimes
  http://www.marchofdimes.com

- Child Development Infoline
  http://www.infoline.org
  1-800-505-7000 or 211

- The Connecticut Department of Public Health
  http://www.dph.state.ct.us