



COLORADO RESPONDS TO CHILDREN WITH SPECIAL NEEDS
Public Health Program for Monitoring and Preventing Birth Defects

Major Congenital Anomalies in Colorado from 1989-1996

This brief describes the occurrence of major congenital anomalies in Colorado residents from 1989 to 1996. Since 1989, Colorado Responds to Children with Special Needs (CRCSN) has been collecting data on the occurrence of congenital anomalies. CRCSN is the Colorado Department of Public Health and Environment's statewide, public health program for monitoring and preventing birth defects and developmental disabilities. The purposes of CRCSN are to

- ❖ develop and maintain a statewide database of young children with birth defects, developmental disabilities, and/or risk factors for developmental delay
- ❖ perform epidemiologic surveillance and investigations
- ❖ prevent birth defects and disabilities associated with birth defects and help connect children and families with early intervention services
- ❖ provide accurate, aggregate statistics and an unduplicated count of children with special needs to other programs and agencies
- ❖ provide statistics to researchers

To be included in CRCSN a child must be a Colorado resident diagnosed prenatally to three years of age with an eligible condition. Eligible conditions include medical conditions, medical risk factors for delay, or one of two environmental (maternal) risk factors for delay. Children meeting these criteria are identified from computer linkage of information from hospitals, vital records (birth, death and fetal death certificates), the Newborn Genetics Screening Program, the Newborn Hearing Screening Program, laboratories, physicians, and genetic, developmental and other specialty clinics. About 7,000 children or 13 percent of births in Colorado each year are identified because they meet CRCSN's eligibility criteria. About 4.5 percent of all births each year to Colorado residents (~2,400) have major congenital anomalies.

The terms "birth defect" and "congenital anomaly" are commonly used interchangeably. CRCSN collects information on both major and minor birth defects/congenital anomalies. Generally an anomaly is classified as major if it has medical, surgical or cosmetic importance whereas minor defects do not.¹ The classification of congenital anomalies is not always consistent and varies among states and studies. Whether a birth defect is classified as major or minor is not as important as how this information can be used to prevent future birth defects.

1. Leppig KA, Werler MM, Cann CI, Cook CA, Holmes LB. Predictive value of minor anomalies
I. Association with major malformations. J Pediatr 1987;110:531-537.

WHAT IS KNOWN ABOUT THE CAUSES OF CONGENITAL ANOMALIES?

The causes of the majority of congenital anomalies are not presently understood. A combination of genetic, biologic or environmental factors is considered to produce many of these conditions. An estimate of the percent of congenital anomalies attributed to each of these factors is shown in Table 1,² At least 50 percent of major congenital anomalies have unknown causes.

TABLE 1. CAUSES OF MAJOR CONGENITAL ANOMALIES

Causes	Percent (%)
Chromosome abnormalities*	6-7
Mutant genes**	7-8
Environmental factors (for example:drugs)	7-10
Multifactorial inheritance (a combination of genetic and environmental factors)	20-25
Unknown causes	50-60

* Chromosome Abnormalities- Chromosomes are structures in the center of our cells, which contain genes. Normally, each cell contains 46 chromosomes. A chromosome abnormality refers to an incorrect number of chromosomes or pieces of chromosomes that are missing or extra.

** Mutant genes refer to a change in genetic(hereditary) material. Genes are instructions that determine how we grow, develop and function. If a gene has a mutation, it may cause serious birth defects.

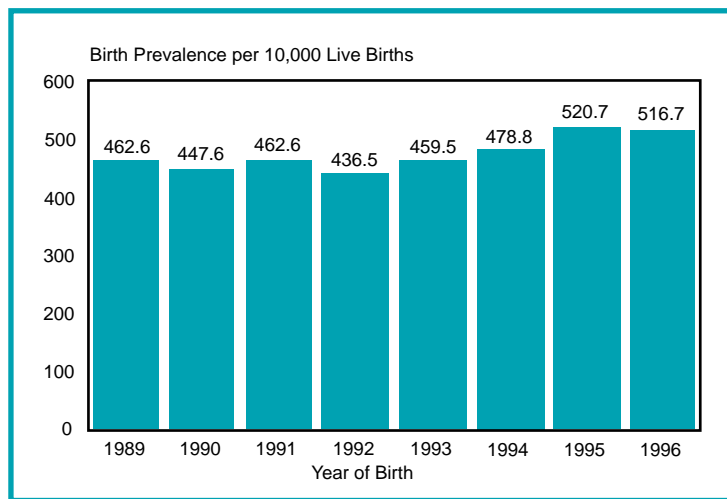
The intent of this brief is to provide an introductory description of the occurrence of major congenital anomalies in Colorado residents from 1989-1996. The live birth prevalence rates presented in the following figures were calculated by counting the number of births with at least one major anomaly, dividing by the total number of births which occurred in the group of interest and multiplying by 10,000. For example: In 1989, there were 52,697 live births to Colorado residents. Major congenital anomalies have been diagnosed in 2,438 of these births. The major congenital anomaly rate therefore was calculated as $(2,438/52,697) * 10,000$ which results in a live birth prevalence rate of 462.6 per 10,000 live births.

2. Moore KL, Persaud TVD(1993): Human Birth Defects. In: "The Developing Human" Philadelphia: W.B.Saunders Company. p.143.

MAJOR CONGENITAL ANOMALIES IN COLORADO OVER TIME

The rates of congenital anomalies can change over time. The live birth prevalence rate of major congenital anomalies in Colorado from 1989-1996 are presented below. Respectively, the most common anomalies by body system were: cardiovascular anomalies, musculoskeletal anomalies and genitourinary anomalies. Statistically, these data show that the live birth prevalence rate of major congenital anomalies increased over this time period.

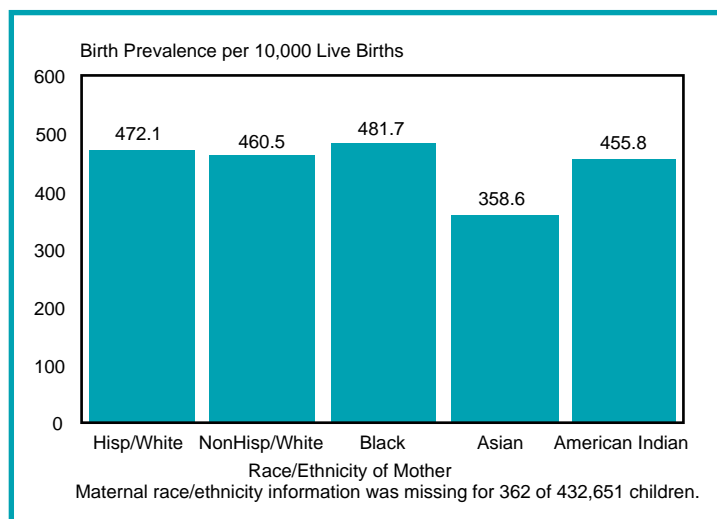
MAJOR CONGENITAL ANOMALY RATES BY YEAR OF BIRTH COLORADO RESIDENTS: 1989-1996



RACE/ETHNICITY

The overall live birth prevalence rates by race/ethnicity of the mother are presented below. Race/ethnicity categories were determined from the child's birth certificate. As can be seen in the graph below, Asian mothers have a notably lower live birth prevalence of major congenital anomalies.

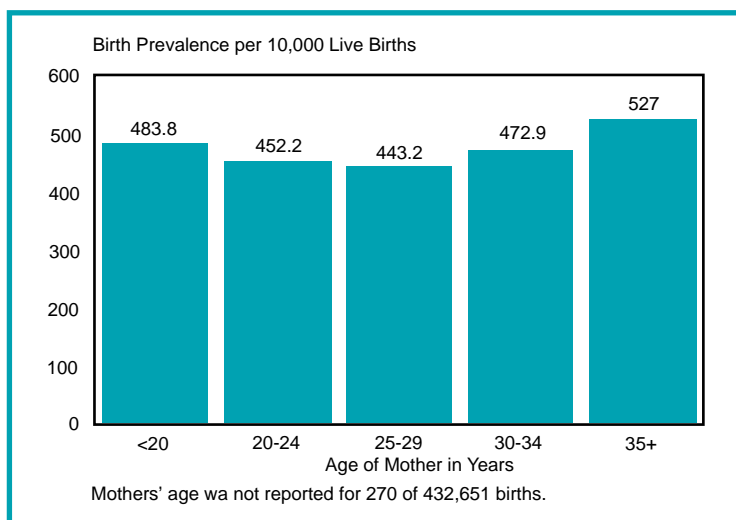
MAJOR CONGENITAL ANOMALY RATES BY RACE/ETHNICITY COLORADO RESIDENTS: 1989-1996



MATERNAL AGE

Rates of congenital anomalies change depending upon the age of the mother. Rates for major congenital anomalies were highest among mothers 35 years of age and older. Rates generally decreased across age groups from less than 20 years, to 20-24 years, and 25-29 years.

MAJOR CONGENITAL ANOMALY RATES BY AGE OF MOTHER COLORADO RESIDENTS: 1989-1996



HOW DO SPECIFIC MAJOR ANOMALY RATES IN COLORADO COMPARE WITH RATES IN OTHER STATES?

Accurate prevalence rates are essential to birth defects monitoring and surveillance programs. Among the many ways in which CRCSN monitors rates is by comparing Colorado rates to rates from similar programs in other states. Such comparisons are valuable because they are a useful step in evaluating the accuracy of Colorado's birth defect surveillance system and suggest areas for further investigation. The comparison rates presented in Table 2 were provided by the individual states reporting to the National Birth Defects Prevention Network.

The data in Table 2 are from states that based their rates on live births and fetal deaths. However, comparing data from different states must be done carefully because of methodological and statistical issues.

TABLE 2. RATES FOR SELECTED MAJOR ANOMALIES: COLORADO AND SELECTED STATES

STATE YEARS Rates based on live births and fetal deaths	Colorado 1989-96 435,615			IL	MA	NE	VA	WI	HI	CA
	Lower C.L.		Upper C.L.							
Anencephalus	1.27	1.63	2.06	1.83	0.73	3.21	0.12	2.02	3.59	2.80
Spina bifida without anencephalus	3.36	3.93	4.56	3.27	2.47	5.74	2.70	4.83	5.32	3.68
Hydrocephalus without Spina Bifida	8.13	9.00	9.93	5.97	4.46	8.11	3.26	5.61	11.53	7.01
Encephalocele	0.97	1.29	1.67	0.63	0.85	1.63	0.36	0.73	2.63	1.11
Microcephalus	6.58	7.37	8.22	2.92	2.84	4.63	2.29	3.32	10.40	--
Anophthalmia/micro- phthalmia	1.17	1.52	1.93	0.98	0.89	0.84	0.40	0.80	4.06	3.07
Congenital cataract	1.87	2.30	2.79	0.54	1.18	2.16	0.66	0.91	1.08	1.83
Aniridia	0.09	0.21	0.39	0.03	0.12	0.11	0.03	0.12	0.00	1.52
Anotia/microtia	1.68	2.09	2.56	0.26	0.61	1.16	0.23	12.74	3.94	2.51
Common truncus	1.25	1.61	2.03	0.40	0.45	0.63	0.45	0.41	0.66	0.83
Transposition of great arteries	3.21	3.76	4.39	2.01	1.54	4.69	2.42	1.38	4.18	4.44
Tetralogy of Fallot	3.25	3.81	4.44	1.86	2.51	3.79	2.44	1.88	4.24	3.35
Ventricular septal defect	34.80	36.57	38.41	16.79	22.80	38.08	17.83	17.82	46.13	16.60
Atrial septal defect	32.74	34.46	36.24	10.31	8.56	35.71	11.93	11.66	55.45	—
Endocardial cushion defect	3.21	3.76	4.39	1.23	1.22	4.00	1.71	1.07	2.27	3.69
Pulmonary valve atresia and stenosis	10.54	11.52	12.58	1.52	5.80	5.53	7.57	3.60	10.40	—
Tricuspid valve atresia and stenosis	1.23	1.58	2.00	0.39	0.16	10.06	0.58	0.28	27.55	—
Ebstein's anomaly	0.58	0.83	1.14	0.26	0.20	0.42	0.31	0.21	0.30	—
Aortic valve stenosis	1.97	2.41	2.92	0.50	0.24	2.53	0.72	1.15	1.25	2.00
Hypoplastic left heart syndrome	2.70	3.21	3.79	1.27	1.30	2.90	0.88	0.82	1.61	1.78
Patent ductus arteriosus	82.71	85.42	88.20	37.41	35.29	46.09	33.53	28.54	61.84	—

STATE YEARS Rates based on live births and fetal deaths		Colorado 1989-96 435,615		IL	MA	NE	VA	WI	HI	CA
Coarctation of aorta	7.69	8.54	9.45	1.53	1.34	5.21	2.50	1.37	2.81	3.89
Pulmonary artery anomalies	9.95	10.90	11.93	9.20	4.75	8.37	4.87	2.33	16.79	—
Lung agenesis/hypoplasia	8.87	9.78	10.75	2.93	2.88	5.11	0.97	4.10	5.44	—
Cleft palate without cleft lip	7.65	8.49	9.40	3.52	5.92	8.16	4.52	14.75	7.05	6.56
Cleft lip with and without cleft palate	10.74	11.73	12.79	6.09	7.75	11.59	8.63	15.62	12.25	10.94
Choanal atresia	2.03	2.48	2.99	0.86	1.42	1.05	0.66	1.24	1.61	1.47
Esophageal atresia/ tracheoesophageal fistula	4.13	4.75	5.45	2.40	2.27	3.90	1.18	2.72	2.03	2.53
Rectal and large intestinal atresia/ stenosis	5.55	6.27	7.06	2.51	2.88	4.37	2.02	4.30	4.90	3.73
Pyloric stenosis	16.67	17.91	19.21	1.16	0.49	17.22	7.57	0.25	8.96	15.40
Hirschsprung's disease (congenital megacolon)	2.49	2.98	3.54	1.10	0.61	1.90	1.04	0.69	2.09	1.44
Biliary atresia	0.85	1.15	1.51	0.19	0.12	0.68	0.42	0.46	1.20	0.87
Renal agenesis/hypoplasia	3.81	4.41	5.08	2.03	3.69	5.79	1.12	3.05	4.78	2.69
Bladder exstrophy	0.13	0.25	0.45	0.25	0.24	0.53	0.21	0.25	0.24	0.26
Obstructive genitourinary defect	19.44	20.78	22.17	6.39	28.68	17.85	5.27	6.50	14.82	14.66
Hypospadias and Epispadias	38.88	40.75	42.68	14.28	40.52	30.81	16.67	4.63	26.05	14.59
Reduction deformity, upper limbs	4.08	4.71	5.40	1.43	2.15	3.58	1.32	2.72	2.99	3.58
Reduction deformity, lower limbs	2.01	2.46	2.97	0.76	1.05	2.00	0.73	1.51	1.08	1.62
Congenital hip dislocation	21.89	23.30	24.78	1.86	18.54	2.21	7.67	25.90	12.73	—
Diaphragmatic hernia	3.76	4.36	5.03	2.46	2.19	4.00	1.21	3.20	2.45	2.83
Trisomy 13	0.93	1.24	1.62	0.99	0.32	1.53	0.35	1.03	2.39	0.97
Down syndrome	11.53	12.56	13.65	9.18	9.45	12.69	5.43	11.73	15.12	12.33
Trisomy 18	1.66	2.07	2.54	1.52	0.89	2.74	0.34	2.08	5.74	1.90

NOTES FOR TABLE 2

RATES ARE PER 10,000 LIVE BIRTHS AND FETAL DEATHS

Rates for this table were provided by the National Birth Defects Prevention Network (NBDPN). Time periods may vary by state and within state for specific conditions. An "-" indicates information was not available. For specific information concerning how rates were compiled the reader is referred to the: National Birth Defects Prevention Network. Congenital Malformations Surveillance Report. Teratology. 2000;61(1/2).

CRCSN would like to thank the NBDPN and the following states: **IL:** Adverse Pregnancy Outcomes Reporting System, Illinois Department of Public Health, **MA:** Massachusetts Congenital Anomaly Surveillance System, Massachusetts Department of Public Health, **NE:** Nebraska Birth Defects Registry, Nebraska Department of Health, **VA:** Virginia Congenital Anomalies Reporting and Education System, Commonwealth of Virginia Department of Public Health. **WI:** Birth and Developmental Outcome Monitoring Program, Wisconsin Department of Health and Family Services, **HI:** Hawaii Birth Defects Program, **CA:** California Birth Defects Monitoring Program, California Department of Health Services.

Confidence limits presented here are 95% exact binomial confidence limits. This means that the true rate of a defect should be contained within these limits 95 % of the time that the limits are calculated this way.

For more information contact:

Colorado Responds to Children with Special Needs

Colorado Department of Public Health and Environment

4300 Cherry Creek Drive South

Denver, CO 80246-1530

(303) 692-2700

e-mail: crcsn@state.co.us

website: www.cdphe.state.co.us/dc/dccrcsn.html



Colorado Department
of Public Health
and Environment

For more information on healthy pregnancies
or birth defects contact the

March of Dimes

888-663-4637

e-mail: resourcecenter@modimes.org

website: www.modimes.org