Alaska Maternal and Child Health Data Book 2005:

Birth Defects Surveillance Edition

Alaska Maternal and Child Health Epidemiology Unit
Section of Women's, Children's and Family Health
Alaska Division of Public Health

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How to Learn More About the Alaska Birth Defects Registry (ABDR)

To find out more about the ABDR and other projects conducted by the Alaska MCH Epidemiology Unit, visit
http://www.epi.hss.state.ak.us/mchepi/

To find out about
the National Birth Defects Prevention Network
and birth defects surveillance projects in other states visit
http://www.nbdpn.org

Alaska Maternal and Child Health Data Book 2005: Birth Defects Surveillance Edition

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To the Alaskan families who provided photos for this book

Introduction

We are pleased to present the first comprehensive analysis of Alaska's birth defects surveillance data in the third edition of the *Alaska Maternal and Child Health (MCH) Data Book.*

The first edition of the MCH Data Book, published in 2003, provided comprehensive information on the health status of Alaskan mothers, infants, children and families. The *Alaska MCH Data Book 2003* synthesized information from statewide surveillance systems, surveys, vital records and program services to provide critical data on leading health status indicators and emerging issues in maternal and child health. We will continue to publish comprehensive summaries of maternal and child health status every three years.

In interim years, the MCH Data Book focuses on specific topics in Maternal and Child Health. The *Alaska MCH Data Book 2004: PRAMS Edition* presented detailed information from the Pregnancy Risk Assessment Monitoring System (PRAMS), providing the first regional and univariate analyses of PRAMS survey data.

The current publication, the *Alaska MCH Data Book 2005: Birth Defects Surveillance Edition*, features seven years of data from the Alaska Birth Defects Registry (ABDR). The birth prevalence of major congenital anomalies reported to the ABDR is presented by birth year, region of maternal residence, and demographic categories. Univariate analyses provide the user with a comparison of the relative distribution of major congenital anomalies within important maternal and infant subgroups.

The *Alaska MCH Data Book* is produced by the MCH Epidemiology Unit of the Section of Women's, Children's and Family Health. Our mission is to provide reliable and statistically accurate information for MCH program planning and evaluation. We trust the *Alaska MCH Data Book 2005: Birth Defects Surveillance Edition* will be a helpful reference for all Alaskans concerned with improving the health and well being of Alaskan families.

Janine Schoellhorn, MS, MPH
MCH Epidemiologist

HOW TO USE THIS BOOK

Birth defects registry data are useful for estimating the burden of congenital anomalies in the state and for identifying service delivery and intervention needs. In this book, we present temporal patterns in the occurrence of major anomalies and the relative frequency of birth defects among different populations.

We encourage readers to use the data presented in this book to improve the health of Alaska's children. However, **if you intend to use these data, please become familiar with the Data Limitations section of the book and the surveillance methods we used for collecting information on the occurrence of birth defects in Alaska, presented in Chapter 2.** Understanding the data limitations and surveillance methods will help you to correctly interpret the information presented in this book.

The Data Book is divided into chapters based on the anatomical site of the malformation, a common practice for birth defects reporting. Detailed epidemiological information is given for the group as a whole and for the specific birth defects within each group that have an Alaskan birth prevalence of at least 12.0 per 10,000 live births. The following information is presented:

- Trends and Geographic Distribution: Because the health care service delivery system in Alaska has agencies that specifically serve the Alaska Native population, we present trend lines for the overall population, Alaska Natives, and non-Natives. Regional prevalence estimates are presented in a bar chart with corresponding 95% confidence intervals. We analyzed data by the six labor market regions used by the Alaska Department of Labor and Workforce Development*. Sample size limitations prevent analysis by smaller geographical units.

- Epidemiological Characteristics: We evaluated prevalence by sex, birth weight, maternal race, maternal age, trimester of prenatal care, prenatal alcohol use and prenatal tobacco use. For

each characteristic, the tables provide relative prevalence estimates and 95% confidence intervals for the estimate. Relative prevalence estimates were derived from bivariate analysis and were not adjusted for the influence of other factors.

- Specific Anomalies: For each major anatomical grouping, we present the prevalence of specific anomalies that are designated as "major congenital anomalies" by the National Birth Defects Prevention Network, a coalition of state birth defects registries that works to establish standards for birth defects surveillance and reporting.

*See http://almis.labor.state.ak.us/

Please share with us how you have used the data published here.
We also welcome feedback on the usefulness of this format.
You may contact ABDR staff by e-mail at
AK_MCFHFacts@health.state.ak.us

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In an average year in Alaska…

Major congenital anomalies are identified in 556 infants (6% of live births)

16% of pre-term infants have an identified major congenital anomaly

Major congenital anomalies are identified in 10% of Alaska Native infants

38% of infants with an identified major congenital anomaly have a cardiovascular anomaly

25% of infants with an identified major congenital anomaly have a fetal alcohol spectrum disorder

Higher rates, compared to National data, are found for almost all major congenital anomalies

In Alaska, during 1996-2002 …

There was a statistically significant declining trend in the overall prevalence of microcephalus, pulmonary valve defects, congenital hip dislocation and Hirschsprung's disease

The prevalence of neural tube defects declined

Among all Alaskans, the only major anomalies that demonstrated significant overall increases in annual prevalence were atrial septal defects and patent ductus arteriosus

Among Alaska Natives, there was a statistically significant increase in the prevalence of Down syndrome

Older maternal age was associated with cardiovascular and chromosomal anomalies

Younger maternal age was associated with alimentary, central nervous system and musculoskeletal anomalies

Alaska Natives had higher rates than non-Natives for 10 of the 15 most commonly identified major congenital anomalies

Data Limitations

While birth defects surveillance data can provide accurate prevalence estimates, which in turn can assist with identifying service delivery and intervention needs, the reader should recognize the following limitations:

- ABDR is a passive surveillance system. Thus, prevalence estimates were based on cases reported under qualifying ICD-9 codes and were not verified through medical record reviews. Previous evaluations have demonstrated that the positive predictive value of reports to ABDR vary substantially by condition.

- We categorized birth defects by anatomical groupings used by most birth defects surveillance projects. Within anatomical groupings, specific birth defects may have diverse etiologies and epidemiological characteristics.

- Except where noted in the text, prevalence estimates included all reported individuals with an anomaly regardless of whether the anomaly occurred in isolation or in association with other anomalies, including as part of a syndrome.

- Birth defects are rare events and Alaska's population is relatively small. In order to provide reliable statistical estimates, we present detailed epidemiological information (trend analysis, regional analysis and sub-group analysis) only for the 15 most common major anomalies that occurred during the seven years under study (1996-2002). For less common major anomalies, the overall and race group specific prevalence estimates are presented. When less than 5 events occurred within a subgroup, prevalence estimates were not calculated.

- Although birth defects are reportable in Alaska up to one year of age (up to age six years for alcohol-related birth defects), many reporting sources reported birth defects diagnosed in older

children. The prevalence estimates presented here include all reports for children born during 1996-2002 that were received before January 1, 2005, regardless of the age at diagnosis or the age at which the child was first reported to the ABDR. Many states include only children who were diagnosed or reported before their first birthday.

- Data were collected from a variety of health care providers and medical records sources and thus were subject to diagnostic bias. For example, the availability of more sophisticated ultrasound machines and clinical specialists in some areas likely resulted in increased diagnosis of anomalies such as asymptomatic ventricular or atrial septal defects. Differences between reporting sources in record keeping and reporting methods may also have affected results.

- All risk factor information came from birth certificates through linking birth certificate and ABDR databases. Variables included on birth certificates may not accurately reflect the true prevalence of some risk factors such as prenatal care, alcohol use during pregnancy and tobacco use during pregnancy.

- Elevated prevalence estimates within particular risk groups do not imply that a causal relationship existed between the risk factor and the outcome. Associations instead may have occurred as a result of the presence of numerous unmeasured or unanalyzed confounding variables. Nevertheless, these associations may indicate appropriate groups for targeting of services or conducting more thorough evaluations of causal associations.

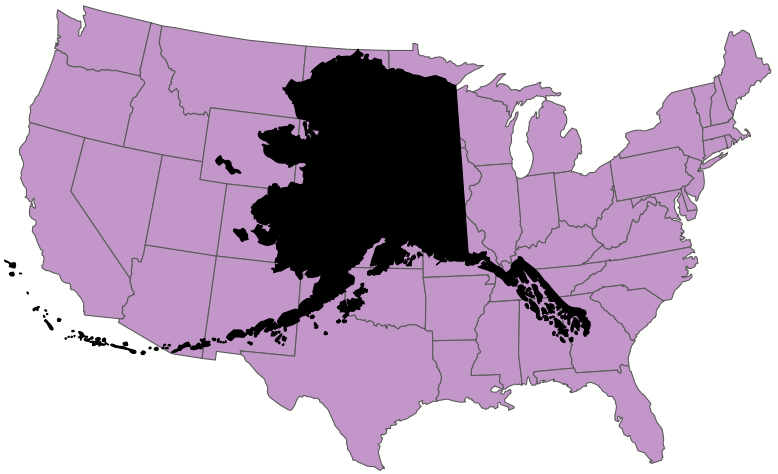
Population Characteristics

Population Distribution

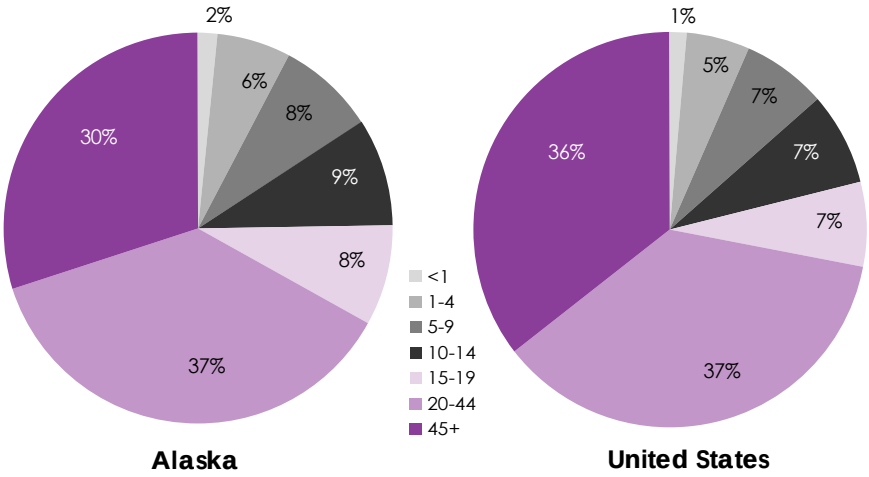
Geographically the nation's largest state, Alaska makes up approximately 16% of the United States land area, but only 0.2% of the population. The land area of Alaska is 570,373 square miles. Alaska's population was 648,280 in 2002, making it one of the least-populated states, ranking 48th.

- Alaska's population is young. Proportionately, there are 33% more 0-4 year olds in Alaska than in the overall U.S. population.
- The Anchorage/Mat-Su region is home to 51.4% of Alaska's population.
- Whites account for the 75% of the state's population, followed by Alaska Natives (16%), blacks (5%), and Asian or Pacific Islanders (4%). About 4% of Alaskans, regardless of race, indicate they are of Hispanic ethnicity.

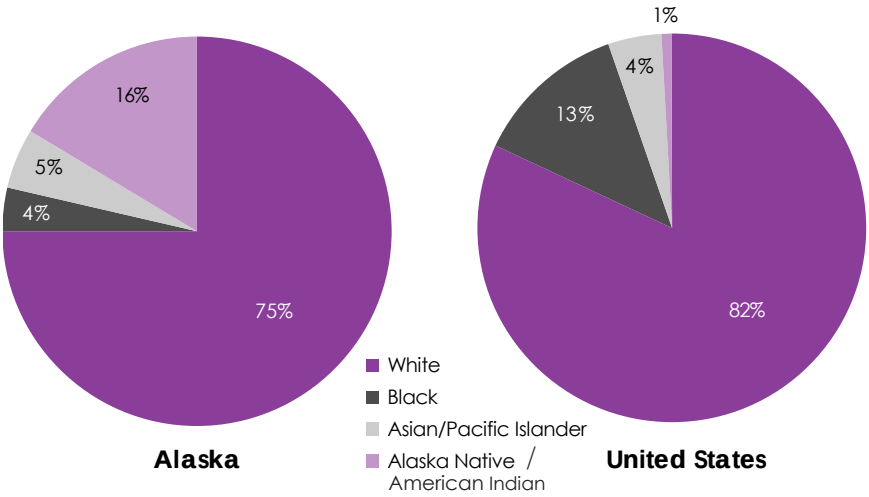
Size Comparison, Alaska and United States



Population Distribution by Age
Alaska and United States, 2002



Population Distribution by Race
Alaska and United States, 2002



Birth Rate

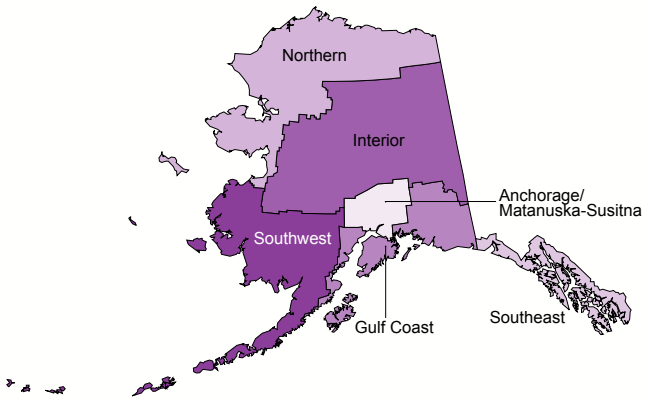
Historically, population growth in Alaska was characterized by periods of rapid growth due to in-migration during economic booms. Since 1993, the major stimulus of Alaska's growth has been natural population increase, despite significant declines in the birth rate. In recent years, population growth in Alaska averaged about 1.3% annually -- one of the highest rates of population growth in the nation (1).

- Compared to 1981, there has been a 33% decrease in the crude birth rate in Alaska. The national rate has also declined over the last two decades (11%).

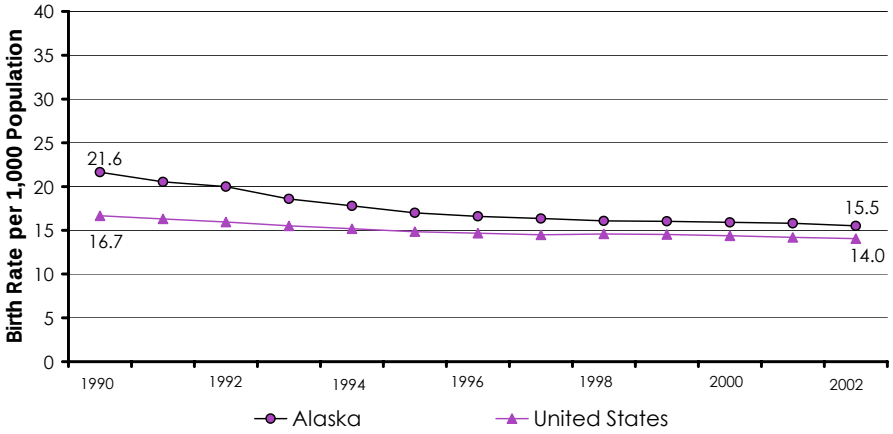
- Disparities in birth rates between Alaska and the U.S. are diminishing with time. In 1990, Alaska's crude birth rate was 29% higher than the U.S. rate. In 2002, Alaska's rate was 11% higher.

- Alaska Native women have the highest crude birth rate, 23 per 1,000 population – 1.6 times that of whites. Alaska Native women make up 18% of the female population and deliver approximately one-fourth of the total Alaskan births.

- During 1996–2002, birth rates were highest in the Northern and Southwestern regions of the state. The majority of people living in Alaska's Northern and Southwest regions are Alaska Native (76% and 70% respectively).

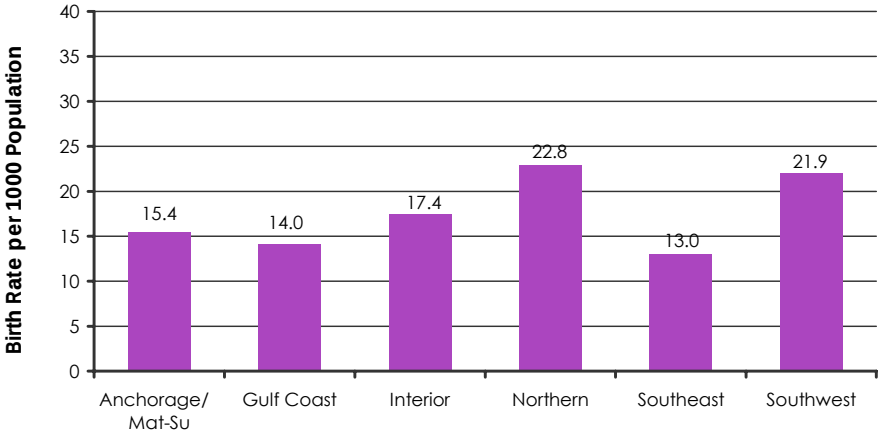


Crude Birth Rate by Year
Alaska and United States, 1990-2002



Data Sources: Alaska Department of Labor; U.S. Census Bureau, Population Estimates. Prepared by MCH Epidemiology Unit.

Average Annual Birth Rate by Region
Alaska, 1996 - 2002



Data Sources: Alaska Department of Labor; Alaska Bureau of Vital Statistics. Prepared by MCH Epidemiology Unit.

Characteristics of Live Births

Characteristics of live births are documented on an infant's birth certificate and registered as a vital record with the state of Alaska. These characteristics include details of the infant's birth, as well as demographic, medical and behavioral factors affecting the pregnancy.

In this data book, we present information on the number of infants reported with birth defects during 1996-2002 by child sex, birth weight, maternal race and ethnicity, maternal age, prenatal care category, reported prenatal alcohol use and reported prenatal tobacco use. The distribution of these characteristics within the total population of Alaska live births during 1996-2002 is presented on the facing page.

Distribution of Live Births by Selected Birth Characteristics, Alaska, 1996-2002

	n	Percent of Live Births
Child Sex		
Female	33962	**48.6**
Male	35790	**51.3**
Missing	78	**0.1**
Birth Weight		
Low and Very Low	3986	**5.7**
Normal	65844	**94.3**
Maternal Race		
White	45227	**64.8**
Alaska Native	16988	**24.3**
Black	3042	**4.4**
Asian or Pacific Islander	3896	**5.6**
Missing	677	**1.0**
Maternal Ethnicity		
Hispanic	4501	**6.4**
Non-Hispanic	61659	**88.3**
Missing	677	**1.0**
Maternal Age		
15-19 years	7708	**11.0**
20-29 years	37929	**54.3**
30-39 years	22125	**31.7**
40-45 years	1890	**2.7**
Missing and Other	178	**0.3**
Prenatal Care		
First Trimester	55019	**78.8**
Second Trimester	10354	**14.8**
Later or None	2666	**3.8**
Missing and Unknown	1791	**2.6**
Maternal Alcohol Use		
Reported	2295	**3.3**
Not Reported	66626	**95.4**
Missing	909	**1.3**
Maternal Tobacco Use		
Reported	12887	**18.5**
Not Reported	66626	**95.4**
Missing	535	**0.8**
Total Live Births	**69830**	**100.0**

Chapter References

Chapter 1: Population Characteristics

1. Alaska Department of Labor and Workforce Development, Research and Analysis Section. Annual Components of Population Change for Alaska, 1945-2005. Available at: http://www.labor.state.ak.us/research/pop/estimates/05t1.1.xls. Accessed February 16, 2006.

Birth Defects Surveillance

The ABDR

The Alaska Birth Defects Registry (ABDR) was established in 1996 under Alaska statute 7 AAC 27.012. Health care providers, hospitals and other health care facilities are required to report to the ABDR when they have cared for a child with a birth defect listed as a *Condition Reportable to Public Health.* A list of Alaska's reportable birth defects and their International Classification of Disease Version 9 (ICD-9) diagnosis codes is presented in the facing table.

Public health surveillance systems such as the ABDR provide information on the occurrence and distribution of reportable health conditions within populations.

ABDR data are used to:

- Estimate the prevalence of congenital anomalies within populations and identify temporal and geographic trends.
- Investigate unusual patterns of occurrence.
- Monitor the prevalence of birth defects in populations with identifiable or preventable exposures and determine whether known exposures have increased the risk of birth defects.
- Conduct analytic studies of high prevalence conditions to elucidate possible etiologies and prevention strategies.
- Observe and evaluate the effects of interventions and policy changes.

ICD-9 Code	Conditions Reportable to the ABDR
237.7	Neurofibromatosis
243	Congenital hypothyroidism
255.2	Adrenogenital disorders
277 – 277.9	Other and unspecified disorders of the Metabolism
279 – 279.9	Disorders involving the Immune Mechanism
282 – 282.9	Hereditary hemolytic anemias
284	Constitutional aplastic anemia
331 – 331.9	Other cerebral degenerations
334 – 334.9	Spinocerebellar disease
335 – 335.9	Anterior horn cell disease
343 – 343.9	Infantile cerebral palsy
359 – 359.9	Muscular dystrophies and other myopathies
362.74	Pigmentary retinal dystrophy
740 – 740.2	Anencephalus and similar anomalies
741 – 741.9	Spina bifida
742 – 742.9	Other congenital anomalies of the nervous system
743 – 743.9	Congenital anomalies of the eye
744 – 744.9	Congenital anomalies of the ear, face and neck
745 – 745.9	Bulbus cordis anomalies and anomalies of cardiac septal
746 – 746.9	Other congenital anomalies of the heart
747 – 747.9	Other congenital anomalies of the circulatory system
748 – 748.9	Congenital anomalies of the respiratory system
749 – 749.25	Cleft palate and cleft lip
750 – 750.9	Other congenital anomalies of the upper alimentary tract
751 – 751.9	Other congenital anomalies of the digestive system
752 – 752.9	Congenital anomalies of genital organs
753 – 753.9	Congenital anomalies of the urinary system
754– 754.89	Certain congenital musculoskeletal deformities
755 – 755.9	Other congenital anomalies of limbs
756 – 756.9	Other congenital musculoskeletal anomalies
757 – 757.9	Congenital anomalies of the integument
758 – 758.9	Chromosomal anomalies
759 – 759.9	Other and unspecified congenital anomalies
760 – 760.9	Fetus or newborn affected by maternal conditions which may be unrelated to the present pregnancy
760.71	Alcohol affecting fetus via placenta or breast milk; including Fetal Alcohol Syndrome

Surveillance Methods

The ABDR conducts passive surveillance with data collection relying on mandatory reporting by health care providers. Other state-based registries may rely on information reported on the birth certificate or on information gathered by actively searching medical records for cases of reportable birth defects. Differences between states in reported birth defects prevalences might reflect true differences in risk factor prevalence or may be due to these differences in surveillance methodology.

- The reporting facility screens patient records for reportable ICD-9 codes and submits quarterly reports to the ABDR.
- Reports to the ABDR include: the child's name, birth and diagnosis date, community of birth, race and ethnicity, sex, community of residence, and diagnosis information.
- Reportable birth defects identified in children from birth to 1 year of age must be reported to the ABDR; infants or children affected by maternal alcohol use (ICD-9 code 760.0 and 760.71) must be reported up to the child's 6th birthday.
- The ABDR is a multiple-reporting source registry that maintains information from all reporting sources for each infant or child reported.
- Data are cross-linked to ensure that each occurrence of a specific defect is tallied only once.
- A single child may be reported to the registry several times, for one or more congenital conditions.
- Data is maintained in a secure, confidential database. Individual data and personal identifiers are not released. Only summarized data are reported.

Alaska Birth Defects Registry Reporting Form

Completion date: __ __/__ __/__ __ __ __

Person completing form: _____

Medical facility name: _____

Patient name: _____ _____ _____
(last name) (first name) (middle)

Date of birth: __ __/__ __/__ __ __ __
(mm/dd/yyyy)

Patient community of birth:

Race: White __ Alaska Native __ Asian __ Black __ Other __ Unknown__
(check one)

Is patient of Hispanic? Yes __ No __ Unknown __
(check one)

Patient sex: Male __ Female __
(check one)

Patient community of residence:

ICD-9 Code	Specific Diagnosis Narrative	Date of Diagnosis
__ __ __.__ __	_____	__ __/__ __/__ __ __ __
__ __ __.__ __	_____	__ __/__ __/__ __ __ __
__ __ __.__ __	_____	__ __/__ __/__ __ __ __
__ __ __.__ __	_____	__ __/__ __/__ __ __ __

Alaska Birth Defects Registry
Mail:
3601 C Street, Suite 424
PO Box 240249
Anchorage, Alaska 99524-0249
Telephone: 1-907-269-8097

Case Ascertainment

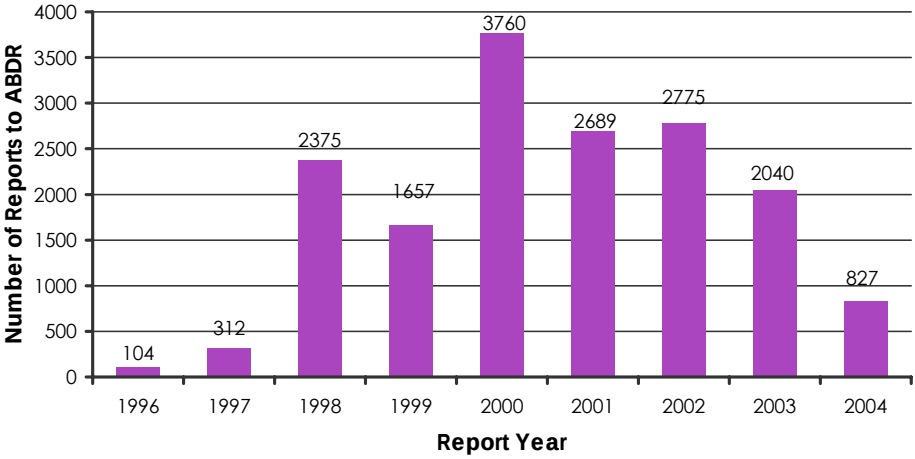
Surveillance issues such as incomplete case ascertainment, late or delayed case ascertainment, variation in diagnostic techniques, over- and under-reporting, coding errors and differences in methodology may influence the reliability of prevalence estimates derived from surveillance data. The ABDR periodically conducts surveillance evaluations to quantify, address and minimize biases associated with these effects. The completeness of case ascertainment is an important consideration in selecting which birth cohorts to include in an analysis of surveillance data. Evaluation of the ABDR indicates good case ascertainment for children born in 1996-2002.

- The number of children reported to the ABDR each year increased after the initial startup years of 1996-1997. Efforts to improve reporting and educate providers about ABDR reporting requirements are most apparent in 2000 when reports for 3760 children were submitted, including many reports for defects occurring during previous years.

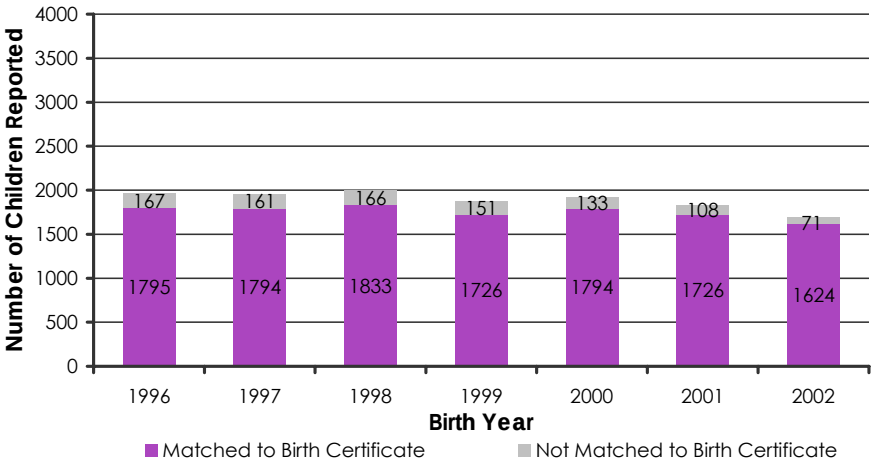
- Thirty five percent of children born during 1996-2002 who were identified with reportable birth defects (other than those classified under ICD-9 codes 760.0–760.71) were reported before their first birthday.

- During 1996-2002, the ABDR identified an average of 1893 children who were born each year with a reportable birth defect (standard deviation: 103, standard error: 39). This represents about 19% of an annual Alaskan birth cohort.

Number of Reports to the ABDR by Year of Report Alaska, 1996-2004



Number of Children Reported to the ABDR by Birth Year Alaska, 1996-2002



Prevalence

Prevalence is the preferred measure of frequency of birth defects (1). Prevalence is defined by the ABDR as the number of reported children born with a birth defect per 10,000 live births during the specified birth years. Before analysis, birth defects reports are matched to an Alaska birth certificate. This ensures an unduplicated count of children reported with birth defects and identifies children who were not born to Alaska resident mothers. In this publication, only reported children born in 1996-2002, who were matched to an Alaska birth certificate, are included in prevalence estimates. Prenatal diagnoses and fetal deaths are not included.

- There are three categories of reportable birth defects in Alaska: major anomalies (as defined by the National Birth Defects Prevention Network)(2), non-major anomalies, and congenital conditions of Alaska interest (Appendix A). This edition of the *MCH Data Book* focuses only on birth defects classified as major anomalies.

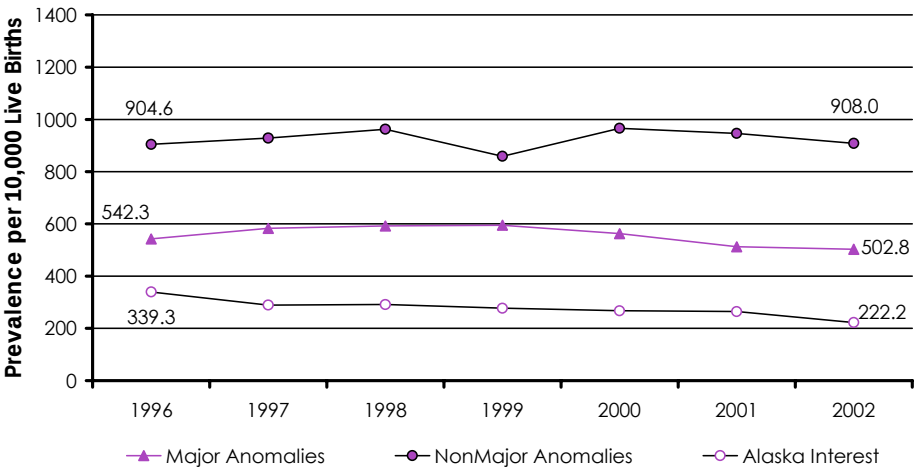
- During 1996-2002, there was no statistically significant change in the overall annual prevalence of reportable birth defects.

- Thirty two percent of children reported to the ABDR each year have at least one major congenital anomaly.

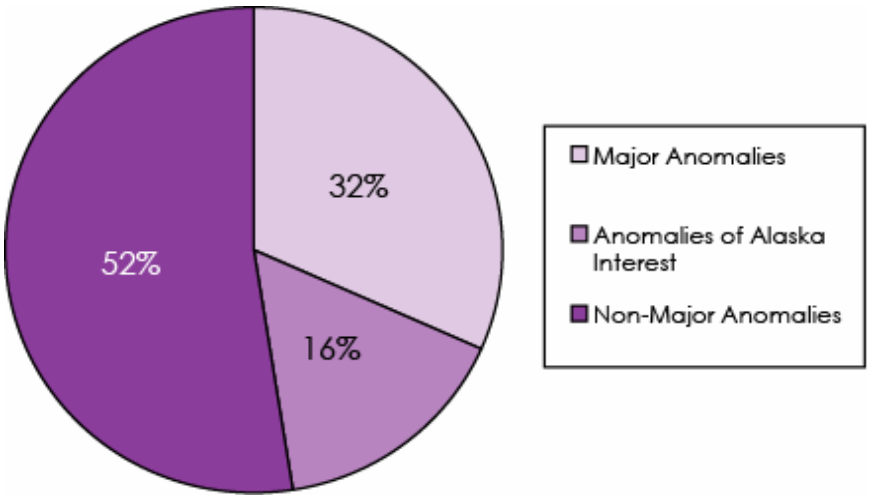
- Sixteen percent of children reported to the ABDR (an average of 276 births per year) had no major anomalies but at least one reportable congenital condition generally classified as a Congenital Condition of Alaska Interest.

- Fifty-two percent of the children reported to the ABDR had only birth defects that were considered to be non-major anomalies.

Prevalence of Reportable Birth Defects by Birth Year and Reporting Category, Alaska, 1996-2002



Distribution of Children Reported to the ABDR (and Matched to Alaska Birth Certificates) by Reporting Category Alaska, Birth Years 1996-2002



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Chapter 2: Birth Defects Surveillance

1. Mason CA, Kirby RS, Sever LE, Langlois PH. Prevalence is the preferred measure of frequency of birth defects. *Birth Defects Research (Part A): Clinical and Molecular Teratology.* 2005;73:400-408.
2. National Birth Defects Prevention Network. *Birth Defects Research (Part A): Clinical and Molecular Teratology.* 2004; 70:772.

Major Congenital Anomalies

Trends and Distribution

The National Birth Defects Prevention Network (NBDPN), an organization that works to promote birth defects research and integrate information collected by state birth defects registries, has defined 45 birth defects that are considered major congenital anomalies (Appendix A). This *MCH Data Book* presents epidemiological information on 44 of these 45 congenital anomalies, including alcohol-related birth defects and not including amniotic bands.

Alcohol-related birth defects are frequently not diagnosed until age 5 or older. Because the defects are recognized later in childhood, surveillance data for more recent birth years will demonstrate lower case counts. As a result, time trends for major congenital anomalies as a group are influenced by the inclusion of alcohol-related birth defects.

- An average of 556 Alaskan children (6% of live births) were born each year with at least one major congenital anomaly (555.9 per 10,000 live births) during 1996-2002.

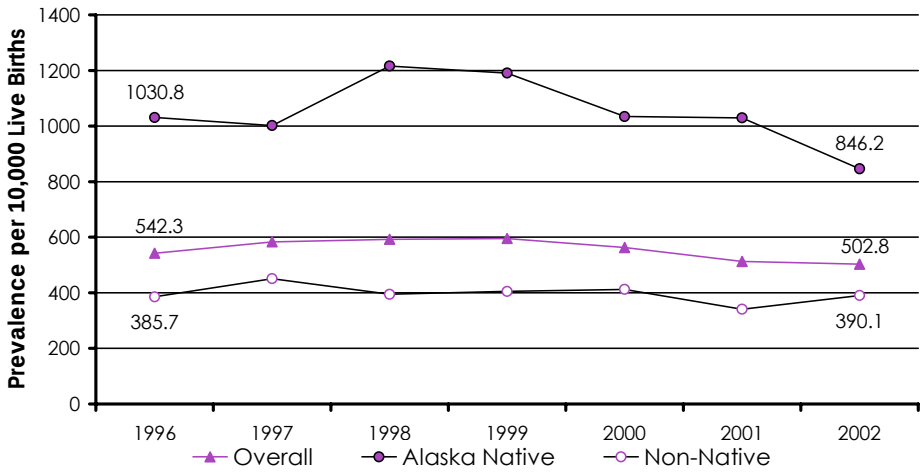
- There were no significant trends in the birth prevalence of major anomalies during 1996-2002 when alcohol-related conditions were removed from the analysis.

- The prevalence of major congenital anomalies was highest in the North and Southwest regions (11% and 9% of live births respectively).

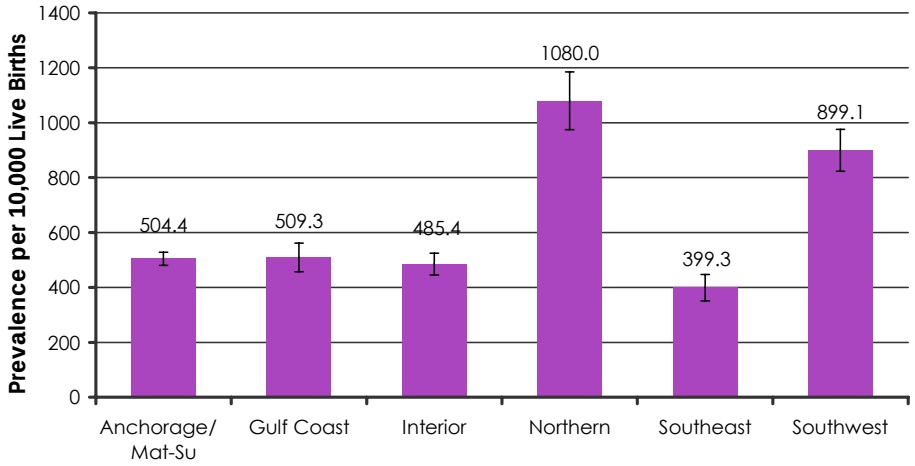
- In the remaining four evaluated regions, about 4% to 5% of live births were affected by a major congenital anomaly.

Major Congenital Anomalies

Prevalence of Major Congenital Anomalies by Birth Year and Race Group, Alaska, 1996-2002



Prevalence of Major Congenital Anomalies by Region Alaska, 1996-2002



Epidemiological Characteristics

Birth defects are one of the most common causes of death among infants and newborns. The cause of about 70% of birth defects has not yet been determined. Epidemiological information on the occurrence of birth defects may elucidate etiologies and assist in resource allocation for medical care and public health efforts.

- In Alaska, during 1996-2002, male infants were more likely to be reported with a major congenital anomaly than females.

- Alaskan infants with low and very low birth weights were 3.7 times more likely to be reported with a major anomaly than infants of normal birth weight.

- The prevalence of major congenital anomalies among Alaska Natives was about 2.5 times that of other race groups. When children reported with fetal alcohol spectrum disorder are excluded from the analysis, the probability of an Alaska Native infant having a major congenital anomaly was still over twice that of any other race group.

- Women aged 30-39 years were least likely to deliver an infant with a major congenital anomaly and teenage mothers were the most likely.

- The probability of delivering an infant with a major congenital anomaly was 1.6 times greater for women who received late or no prenatal care compared to women who began prenatal care in the first trimester.

- The probability of delivering an infant with a major birth defect was 4.3 and 2.2 times higher respectively for women who reported drinking or smoking during their pregnancies.

Note: Fetal alcohol syndrome and fetal alcohol spectrum disorder are considered major congenital anomalies even if children experience no associated major anatomical malformations. When these two conditions are excluded from the evaluation of birth certificate risk factors, prevalence ratios did not change substantially and thus are not presented.

Prevalence of Major Congenital Anomalies by Selected Birth Characteristics Alaska, 1996-2002

	n	Prevalence per 10,000 Live Births	Prevalence Ratio	95% CI
Child Sex				
Female	1631	**480.0**	ref	-
Male	2173	**607.0**	1.3	(1.2 - 1.4)
Birth Weight*				
Low and Very Low	638	**1600.0**	3.7	(3.4 - 4.0)
Normal	2826	**429.0**	ref	-
Maternal Race				
White	1089	**400.0**	ref	-
Alaska Native	1784	**1050.0**	2.6	(2.5 - 2.8)
Black	119	**391.0**	1.0	(0.8 - 1.2)
Asian or Pacific Islander	144	**370.0**	0.9	(0.8 - 1.1)
Maternal Ethnicity				
Hispanic	217	**482.0**	0.9	(0.8 - 1.0)
Non-Hispanic	3473	**563.0**	ref	-
Maternal Age				
15-19 years	520	**675.0**	1.4	(1.3 - 1.5)
20-29 years	2130	**562.0**	1.2	(1.1 - 1.2)
30-39 years	1081	**489.0**	ref	-
40-45 years	125	**661.0**	1.4	(1.1 - 1.6)
Prenatal Care				
First Trimester	2803	**509.0**	ref	-
Second Trimester	714	**690.0**	1.4	(1.3 - 1.5)
Later or None	213	**799.0**	1.6	(1.4 - 1.8)
Maternal Alcohol Use				
Reported	494	**2153.0**	4.3	(4.0 - 4.7)
Not Reported	3319	**498.0**	ref	-
Maternal Tobacco Use				
Reported	1284	**996.0**	2.2	(2.1 - 2.3)
Not Reported	2562	**454.0**	ref	-

*418 Infants with patent ductus arteriosus were excluded from birth wieght analysis because the surveillance case definition for patent ductus arteriosus specifies that only infants ≥2500g are counted.

Specific Anomalies

Major congenital anomalies are categorized into eight groupings – fetal alcohol spectrum disorder and seven anatomical groupings: Cardiovascular, Alimentary Tract, Genitourinary, Central Nervous System, Musculoskeletal, Chromosomal and Eye or Ear Anomalies.

- Cardiovascular birth defects are the most frequently reported major congenital anomalies in Alaska. Two percent of Alaskan infants are reported with a cardiovascular birth defect.

- The second most commonly reported major anomalies are fetal alcohol spectrum disorders. An average of 137 children (1% of live births) are reported annually as having been affected by maternal alcohol use.

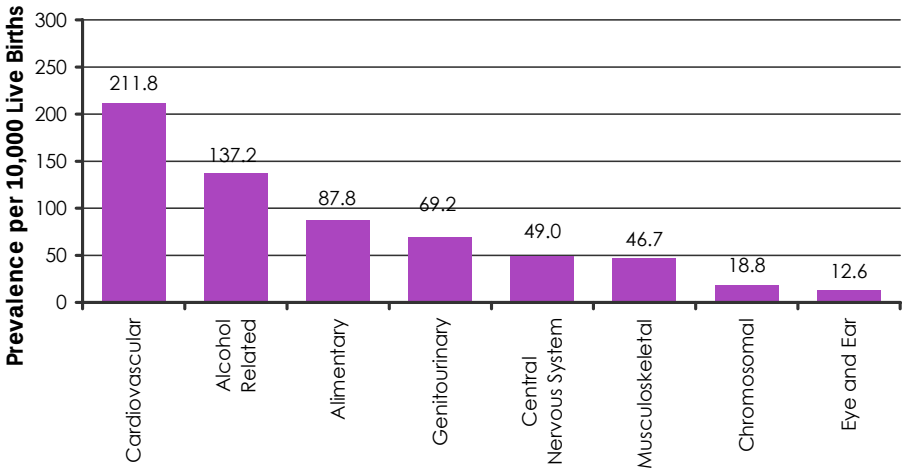
- An annual average of 88 infants are reported to the ABDR as having been born with alimentary tract anomalies, 69 with genitourinary anomalies, 49 with central nervous system anomalies, 47 with musculoskeletal anomalies, 19 with chromosomal anomalies, and 13 with eye or ear anomalies.

- During 1996-2002, there were 15 specific major anomalies with a sufficient number of cases to conduct detailed statistical analysis. Detailed epidemiological information on each anatomical grouping and on each of Alaska's 15 most common specific major anomalies is presented.

Note: Prevalence estimates for specific major anomalies with prevalence of less than 12 per 10,000 live births during 1996-2002 are provided in the detailed tables section of the book.

Major Congenital Anomalies

Prevalence of Major Congenital Anomalies by Anatomical Grouping, Alaska, 1996 –2002



Fifteen Most Frequently Reported Major Anomalies, Alaska, Birth Years 1996-2002

Congenital Anomaly	n	Prevalence
Atrial Septal Defect	641	91.8
Ventricular Septal Defect	591	84.6
Patent Ductus Arteriosus	418	59.9
Hypospadias and Epispadias	254	36.4
Microcephalus	213	30.5
Obstructive Genitourinary Defect	212	30.4
Pyloric Stenosis	210	30.1
Cleft Lip and Cleft Palate	205	29.4
Pulmonary Valve Atresia/Stenosis	195	27.9
Congenital Hip Dislocation	189	27.1
Fetal Alcohol Syndrome (1996-1999)	66	16.5
Down Syndrome (Trisomy 21)	107	15.3
Hydrocephalus	95	13.6
Hirschsprung's Disease	93	13.3
Neural Tube Defects	54	7.7

Cardiovascular Anomalies

Trends and Distribution

Cardiovascular birth defects affect the heart or blood vessels surrounding the heart. Cardiovascular defects are generally estimated to be present in about 1% of live births and are the most commonly diagnosed congenital anomalies. Cardiovascular anomalies usually result in either obstructed or abnormal blood flow to or from the heart. They range in seriousness from minor self-correcting anomalies to fatal conditions. Prevalence estimates for cardiovascular anomalies are highly influenced by the availability of modern diagnostic techniques that can identify less serious defects.

- Cardiovascular birth defects affected about 2% of Alaska live births annually during 1996-2002.

- The overall prevalence of cardiovascular birth defects increased significantly, from 212 to 251 per 10,000 live births, during 1996-2002 ($p=0.02$).

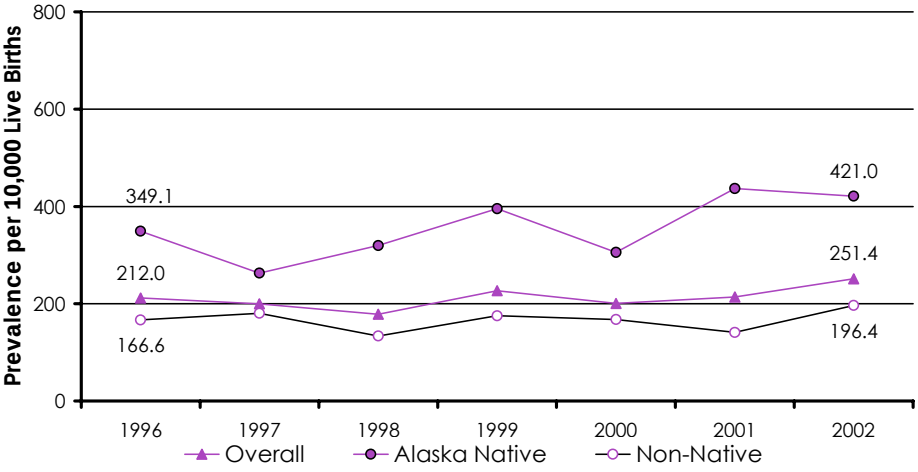
- When examined separately, increasing prevalence during 1996-2002 was significant for Alaska Natives ($p=0.01$), but not for non-Natives.

- Alaska Natives had consistently higher rates of cardiovascular anomalies than non-Natives during 1996-2002. The disparity between Native and non-Native rates remained at a fairly consistent two-fold difference during the time period.

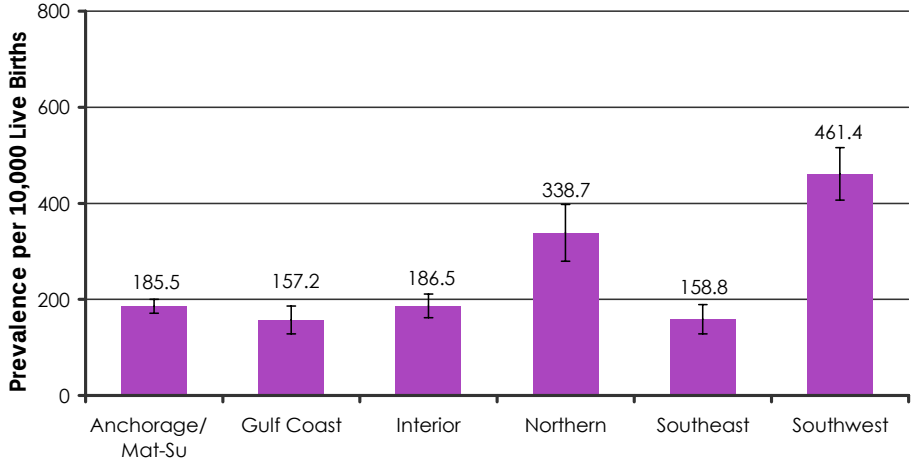
- The prevalence of cardiovascular anomalies in the North and Southwest regions was significantly higher than in other regions of the state.

- Almost 5% of newborns in the Southwest region and over 3% of newborns in Northern Alaska had cardiovascular birth defects during 1996-2002.

Prevalence of Cardiovascular Anomalies by Birth Year and Race Group, Alaska, 1996-2002



Prevalence of Cardiovascular Anomalies by Region Alaska, 1996-2002



Epidemiological Characteristics

The cause of most cardiovascular birth defects is unknown. Cardiovascular defects are assumed to have a multi-factorial etiology with both genetic and environmental components. Known causes include chromosomal abnormalities (5-6%), single gene defects (3-5%) and definable environmental exposures (2%) (1). In 85-90% of cases, there is no identifiable cause, but family history increases the risk of having a child with a cardiovascular anomaly.

- In Alaska, male and female babies had an equal risk of being born with a cardiovascular anomaly during 1996-2002.
- Low birth weight infants were 7 times more likely to have a cardiovascular anomaly than normal birth weight infants.
- Alaska Native mothers were twice as likely to deliver an infant with a cardiovascular birth defect as women of any other race. Maternal Hispanic ethnicity did not increase the risk of a cardiovascular anomaly.
- Older mothers (aged 40-45) were at greater risk for delivering a baby with a cardiovascular birth defect than younger women.
- Late prenatal care was not strongly associated with delivering a baby with a cardiovascular anomaly.
- Reported prenatal alcohol use and reported maternal prenatal cigarette smoking were both associated with an increased risk of having a baby with a cardiovascular birth defect during 1996-2002.

Prevalence of Cardiovascular Anomalies by Selected Birth Characteristics, Alaska, 1996-2002

	n	Prevalence per 10,000 Live Births	Prevalence Ratio	95% CI
Child Sex				
Female	722	**212.6**	ref	-
Male	748	**209.0**	1.0	(0.9 - 1.1)
Birth Weight*				
Low and Very Low	320	**802.0**	7.1	(6.3 - 8.1)
Normal	741	**113.0**	ref	-
Maternal Race				
White	751	**166.1**	ref	-
Alaska Native	605	**356.1**	2.1	(1.9 - 2.4)
Black	46	**151.2**	0.9	(0.7 - 1.2)
Asian or Pacific Islander	68	**174.5**	1.1	(0.8 - 1.3)
Maternal Ethnicity				
Hispanic	92	**204.4**	1.0	(0.8 - 1.2)
Non-Hispanic	1293	**209.7**	ref	-
Maternal Age				
15-19 years	170	**220.6**	1.2	(1.0 - 1.5)
20-29 years	833	**219.6**	1.2	(1.1 - 1.4)
30-39 years	400	**180.8**	ref	-
40-45 years	65	**343.9**	1.9	(1.5 - 2.5)
Prenatal Care				
First Trimester	1110	**201.7**	ref	-
Second Trimester	244	**235.7**	1.2	(1.0 - 1.3)
Later or None	65	**243.8**	1.2	(0.9 - 1.5)
Maternal Alcohol Use				
Reported	80	**348.6**	1.7	(1.4 - 2.1)
Not Reported	1372	**205.9**	ref	-
Maternal Tobacco Use				
Reported	382	**296.4**	1.5	(1.4 - 1.7)
Not Reported	1083	**192.0**	ref	-

*418 Infants with patent ductus arteriosus were excluded from birth wieght analysis because the surveillance case definition for patent ductus arteriosus specifies that only infants ≥2500g are counted.

Specific Anomalies

Individual cardiovascular birth defects may occur as an isolated anomaly or in combination with other birth defects. They are also associated with syndromic conditions caused by chromosomal abnormalities (including Down syndrome and Turner syndrome). Individual cardiovascular birth defects are classified as either cyanotic or acyanotic.

Cyanotic heart disease is a defect or group of defects in the structure or function of the heart or the great vessels consisting of abnormal blood flow from the right to the left part of the circulatory system (either at the level of the atria, the ventricles, or the great vessels). This abnormal communication (called right-to-left shunt) results in poor oxygenation of the body and therefore cyanosis (bluish coloration of the body).

Acyanotic heart disease results from a defect in the structure or function of the heart or great vessels but results in little or no mixing between the two sides of the circulatory system.

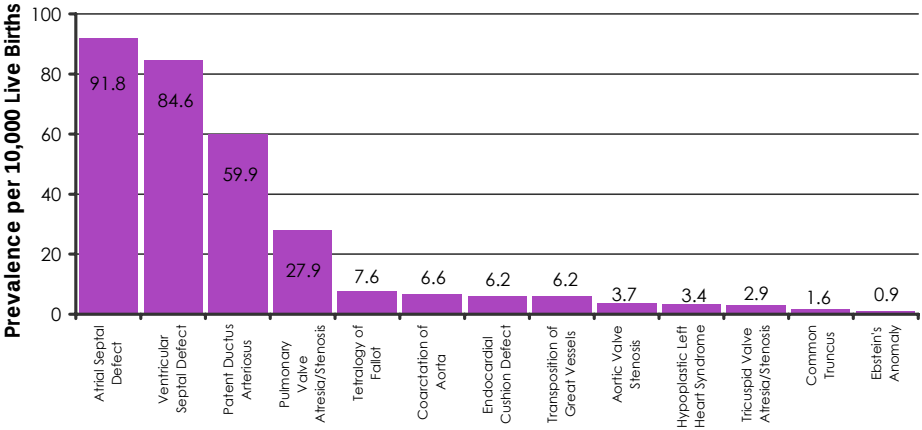
- Twenty five percent of children reported to the ABDR with major anomalies during 1996-2002 had a cardiovascular anomaly. Nineteen percent of these infants had birth defects in other anatomical groupings.

- The most common cardiovascular anomalies in Alaska during 1996-2002 were atrial septal defects (ASDs) (0.9% of live births) and ventricular septal defects (VSDs) (0.8% of live births).

- VSDs, ASDs, patent ductus arteriosus and pulmonary valve atresia/stenosis together comprised 87% of all cardiovascular birth defects reported in Alaska.

- Cyanotic anomalies occurred in 0.3% of live births (32.4 per 10,000 live births) and made up 11% of all cardiovascular anomalies reported during 1996-2002.

Prevalence of Specific Cardiovascular Anomalies Alaska, 1996-2002



Classification of Major Congenital Anomalies of the Cardiovascular System

Acyanotic	Cyanotic
Atrial Septal Defect Ventricular Septal Defect Patent Ductus Arteriosus Pulmonary Valve Atresia/ Stenosis Coarctation of the Aorta Aortic Stenosis	Tetralogy of Fallot Endocardial cushion defect Transposition of the Great Vessels Hypoplastic Left Heart Syndrome. Tricuspid Valve Atresia/ Stenosis Common Truncus

Trends and Distribution

Septal defects are the most common cardiovascular birth defects. During fetal development, an opening in the septum, or wall, between the upper chambers of the heart (atria) provides one of the mechanisms by which blood bypasses the lungs. The opening closes when the lungs begin to function at birth. An atrial septal defect (ASD) occurs when this opening does not close completely. Small ASDs do not considerably hinder heart function and may go undiagnosed and untreated with eventual closure on their own. Large, symptomatic ASDs are closed surgically. Advances in diagnostic techniques for detecting less serious defects may account for increasing trends in ASD prevalence (2). The majority of ASDs now diagnosed are minor -- as many as 87% of ASDs close spontaneously without medical intervention (3).

- The most common cardiovascular birth defect in Alaska, ASDs occurred in almost 1% of Alaska live births (92 per 10,000) during 1996-2002.

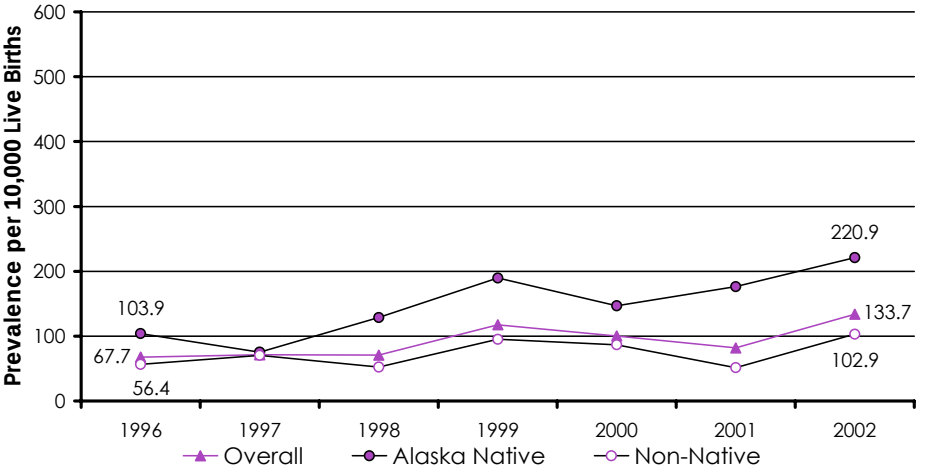
- The prevalence of ASDs almost doubled during 1996-2002, increasing from 67.7 to 133.7 per 10,000 live births.

- Statistically significant trends in the reported prevalence of ASDs were seen for both Natives ($p<0.0001$) and non-Natives ($p=0.009$), with the prevalence among Native Alaskans increasing 112%. In 2002, 2% of Alaska Native infants were born with an ASD.

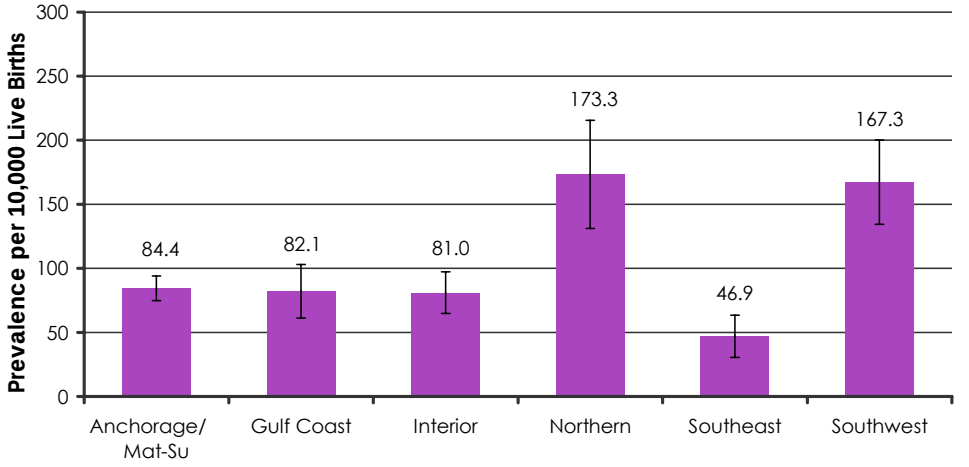
- The North and Southwest regions of Alaska had the highest prevalence of ASD – almost 2% of infants born in these regions during 1996-2002 had an ASD.

- The Southeast region had a significantly lower ASD prevalence than other regions of the state, with only about 0.5% of infants reported with an ASD.

Prevalence of Atrial Septal Defect by Birth Year and Race Group, Alaska, 1996-2002



Prevalence of Atrial Septal Defects by Region Alaska, 1996-2002



Epidemiological Characteristics

An ASD may occur as an isolated anomaly or in combination with other birth defects. A family history of cardiovascular anomalies is associated with ASDs (4) and an ASD susceptibility gene has been identified (5). In addition to genetic and chromosomal anomalies, maternal infections* are important risk factors for ASD. Epidemiological studies have linked ASDs with several environmental exposures, including maternal alcohol consumption (6), chemical exposure (6), and maternal prenatal residence at high altitude (7). Low birth weight has been postulated to have a causal role (8). While ASDs are commonly considered to occur more often in females than males, this appears to be true only for defects greater than 5mm in size (3). There is no documented intervention for the prevention of ASDs.

- In Alaska, female and male infants were equally likely to be reported with an ASD during 1996-2002.
- Low birth weight infants were 8.5 times more likely to have an ASD than normal birth weight infants.
- Alaska Native mothers were twice as likely as white mothers to deliver an infant with an ASD. Black, Asian and Hispanic mothers had a similar probability of delivering an infant with an ASD as whites.
- Women aged 40-45 years had the highest risk of delivering an infant with an ASD -- 2.4 times that of 30-39 year old mothers. Compared to women aged 30-39 years, teenagers were 50% more likely and 20-29 year olds 30% more likely to deliver an infant with an ASD.
- The probability of delivering an infant with an ASD was twice as high for women who smoked or used alcohol during pregnancy.

* Maternal infections associated with cardiovascular anomalies include toxoplasmosis, cytomegalovirus, herpes, and rubella

Prevalence of Atrial Septal Defect by Selected Birth Characteristics Alaska, 1996-2002

	n	Prevalence per 10,000 Live Births	Prevalence Ratio	95% CI
Child Sex				
Female	315	**92.8**	ref	-
Male	324	**90.5**	1.0	(0.8 - 1.1)
Birth Weight				
Low and Very Low	217	**544.4**	8.5	(7.2 - 9.9)
Normal	424	**64.4**	ref	-
Maternal Race				
White	325	**71.9**	ref	-
Alaska Native	253	**148.9**	2.1	(1.8 - 2.4)
Black	22	**72.3**	1.0	(0.7 - 1.5)
Asian or Pacific Islander	36	**92.4**	1.3	(0.9 - 1.8)
Maternal Age				
15-19 years	84	**109.0**	1.5	(1.2 - 2.0)
20-29 years	363	**95.7**	1.3	(1.1 - 1.6)
30-39 years	157	**71.0**	ref	-
40-45 years	32	**169.3**	2.4	(1.6 - 3.5)
Prenatal Care				
First Trimester	476	**86.5**	ref	-
Second Trimester	102	**98.5**	1.1	(0.9 - 1.4)
Later or None	26	**97.5**	1.1	(0.8 - 1.7)
Maternal Alcohol Use				
Reported	43	**187.4**	2.1	(1.6 - 2.9)
Not Reported	585	**87.8**	ref	-
Maternal Tobacco Use				
Reported	198	**153.6**	2.0	(1.7 - 2.4)
Not Reported	435	**77.1**	ref	-
OVERALL	641	**91.8**	-	-

Trends and Distribution

Although less commonly reported in Alaska than atrial septal defects (ASDs), ventricular septal defects (VSDs) are generally thought to be the most common cardiovascular anomaly, representing about 25% of congenital heart defects (9). Early in fetal development, a muscular wall forms to separate the right and left ventricles of the heart. If formation of the wall is incomplete, a hole remains and is referred to as a VSD. Most VSDs are minor and close spontaneously postnatally as the infant continues to grow. Large, symptomatic VSDs are closed surgically.

- Increases in the prevalence of VSDs have been observed since the 1970's (10). The ability to diagnose minor defects through more standard use of echocardiography is now thought to explain a large part of this increase, since the prevalence of more serious septal defects has not been shown to have increased significantly (11).

- For birth years 1996-2002, VSDs made up about 28% of cardiovascular anomalies reported to the ABDR.

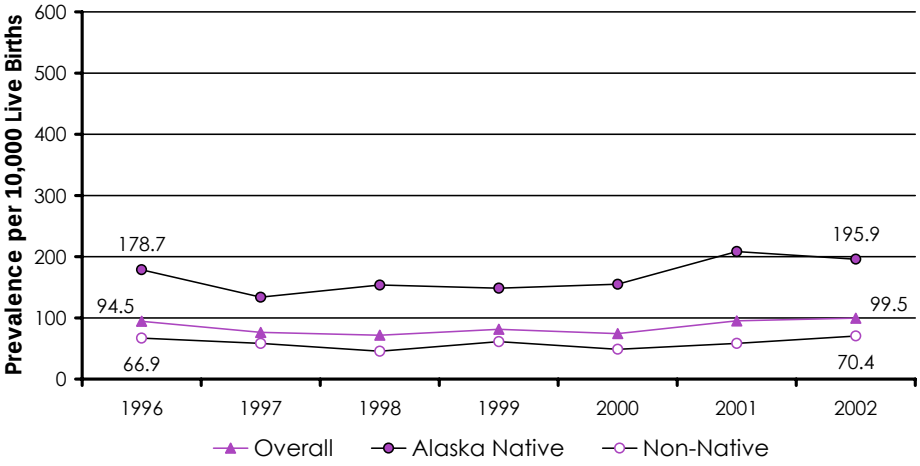
- During 1996-2002, the overall prevalence of VSDs in Alaska was 84.6 per 10,000 live births. There was no significant trend over the time period.

- As with ASDs, VSDs were more common among Alaska Natives than non-Natives. A two-fold racial disparity existed consistent during 1996-2002.

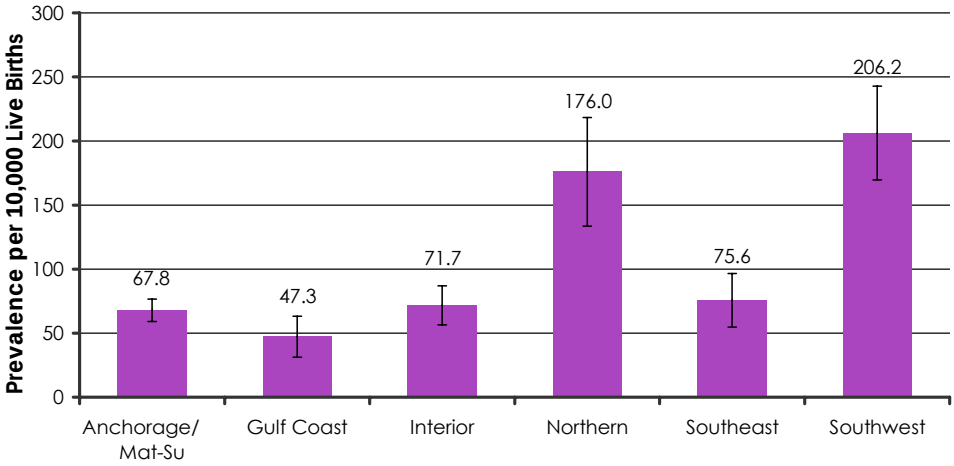
- The regional prevalence of VSDs was similar in the Anchorage/ Mat Su, Gulf Coast, Interior and Southeast regions, but significantly higher in the North and Southwest regions, where about 2% of infants were born with a VSD.

Ventricular Septal Defect

Prevalence of Ventricular Septal Defect by Birth Year and Race Group, Alaska, 1996-2002



Prevalence of Ventricular Septal Defects by Region Alaska, 1996-2002



Epidemiological Characteristics

VSDs frequently occur as isolated birth defects, but may also occur with other congenital anomalies and inheritable syndromes. Known risk factors, such chromosomal anomalies and maternal infection explain only a small proportion of VSDs. Research suggests several factors may be associated with VSDs, including family history of congenital heart disease, maternal alcohol use (9,12), cannabis use (12), exposures to solvents (13,14), pesticides (14), and air pollution (15). There are no known interventions for the prevention of VSDs. Because the concordance rate in twins is only about 10%, and because most VSDs do not appear to be associated with prenatal care, socioeconomic status, or maternal age or race, many VSDs may be the result of random errors in development, and therefore unpreventable based on current medical knowledge (16).

- In Alaska, males were slightly less likely to be reported with a VSD than females during 1996-2002.
- Infants with low birth weight were 3.2 times more likely to have a VSD than normal birth weight infants.
- In contrast to surveillance reports showing no association between maternal race and VSDs (13), there was a significant racial disparity for VSDs in Alaska. Alaska Natives were 2.8 times more likely to be reported with a VSD than other races. There was no significant difference in the prevalence of VSDs among whites, blacks, Asians or Hispanics.
- Women aged 40-45 had almost double the risk of delivering an infant with a VSD than younger mothers during 1996-2002.
- Women with reported alcohol use during pregnancy were almost twice as likely, and women with reported tobacco use 1.6 times as likely, to deliver a baby with a VSD.

Prevalence of Ventricular Septal Defect by Selected Birth Characteristics Alaska, 1996-2002

	n	Prevalence per 10,000 Live Births	Prevalence Ratio	95% CI
Child Sex				
Female	324	**95.4**	ref	-
Male	261	**72.9**	0.8	(0.6 - 0.9)
Birth Weight				
Low and Very Low	95	**238.3**	3.2	(2.5 - 3.9)
Normal	496	**75.3**	ref	-
Maternal Race				
White	274	**60.6**	ref	-
Alaska Native	285	**167.8**	2.8	(2.3 - 3.3)
Black	11	**36.2**	0.6	(0.3 - 1.1)
Asian or Pacific Islander	20	**51.3**	0.8	(0.5 - 1.3)
Maternal Age				
15-19 years	66	**85.6**	1.2	(0.9 - 1.6)
20-29 years	336	**88.6**	1.2	(1.0 - 1.5)
30-39 years	160	**72.3**	ref	-
40-45 years	26	**137.6**	1.9	(1.3 - 2.9)
Prenatal Care				
First Trimester	449	**81.6**	ref	-
Second Trimester	101	**97.5**	1.2	(1.0 - 1.5)
Later or None	25	**93.8**	1.1	(0.8 - 1.7)
Maternal Alcohol Use				
Reported	34	**148.1**	1.8	(1.3 - 2.5)
Not Reported	546	**81.9**	ref	-
Maternal Tobacco Use				
Reported	156	**121.1**	1.6	(1.3 - 1.9)
Not Reported	429	**76.1**	ref	-
OVERALL	591	**84.6**	-	-

Trends and Distribution

The ductus arteriosus is a vessel that provides one of the mechanisms by which blood bypasses pulmonary circulation before birth. No longer needed once pulmonary circulation increases and lung oxygenation occurs postnatally, the vessel normally closes 48-72 hours after birth. A patent ductus arteriosus (PDA) is a condition where the ductus arteriosus fails to close within 10 days after birth*. PDAs are one of the more common congenital heart defects, particularly in pre-term infants. They are estimated to comprise about 12% of cardiovascular anomalies.

- The overall birth prevalence of PDAs in Alaska during 1996-2002 was 59.9 per 10,000 live births. PDAs made up almost 20% of cardiovascular anomalies reported during the time period.

- There was a statistically significant increase in the reported prevalence of PDAs during 1996-2002 for both Alaska Natives and non-Natives. Overall, reported prevalence increased 93%, from 44.8 per 10,000 live births in 1996 to 86.5 in 2002.

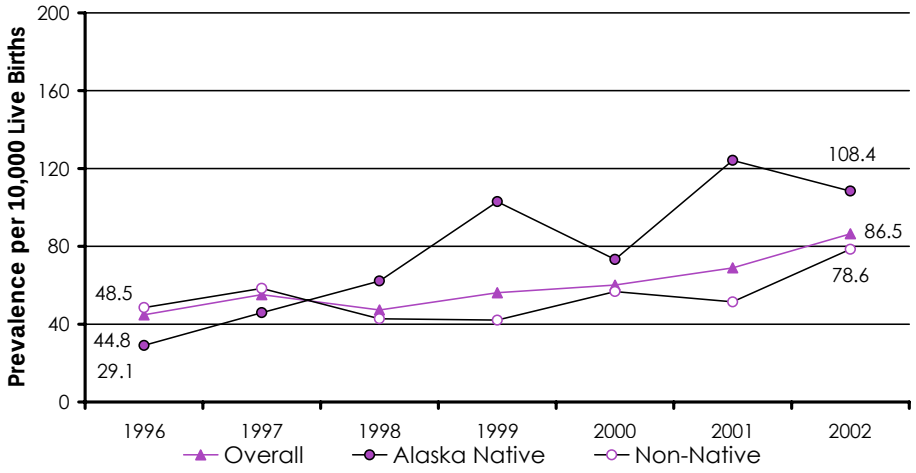
- Alaska Natives had a higher reported PDA prevalence than non-Natives at the end of the 1996-2002 time period. The three-year average prevalence more than doubled for Alaska Natives and the gap between Native and non-Native prevalence widened. At the end of the time period, the three-year average prevalence of PDA for Alaska Natives was 64% higher than the prevalence among non-Natives.

- PDA prevalence was higher (about 0.8% of live births) in the North and Southwest regions of Alaska, but regional differences in prevalence were not statistically significant.

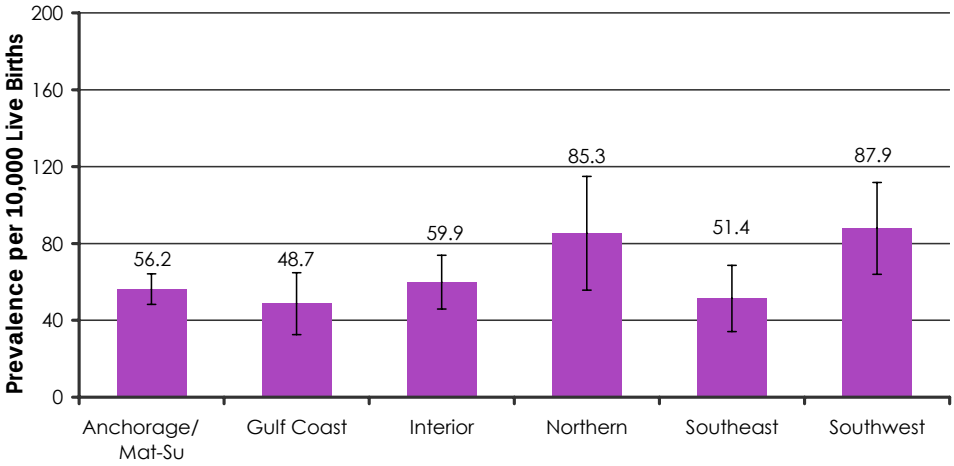
*Data from the ABDR includes PDAs reported at any age; however, low birth weight infants reported with PDAs are excluded from analysis.

Patent Ductus Arteriosus

Prevalence of Patent Ductus Arteriosus By Birth Year and Race Group, Alaska, 1996-2002



Prevalence of Patent Ductus Arteriosus by Region Alaska, 1996-2002



Epidemiological Characteristics

PDAs are common problems in premature infants (affecting 20–60%) and are less likely to be noted as gestational age increases to full term. Along with infants born prematurely, those with respiratory distress syndrome are at a higher risk for a PDA. Females develop PDAs 2-3 times more often than males (17). As with other cardiovascular anomalies, chromosomal anomalies and maternal infections, notably maternal rubella infection in the first trimester (18), are risk factors for PDA. PDA has also been associated with maternal residence at high altitude (7). Improved diagnostic testing and an increase in preterm deliveries may contribute to observed increases in prevalence of PDA. While no specific public health interventions for prevention of PDAs have been recommended, the March of Dimes nationwide campaign on prematurity (2003) may play a role in reducing the number of cases.

- In contrast to well-established epidemiological findings, analysis of ABDR surveillance data for birth years 1996-2002 showed no association between female sex and PDA.

- The infants of Alaska Native mothers were 1.5 times more likely to have a PDA reported than infants born of women of other races or Hispanic ethnicity.

- Older mothers (aged 40-45) were twice as likely to deliver an infant with a PDA as younger women during 1996-2002.

- Early prenatal care and reported prenatal alcohol use were not associated with PDAs, but PDAs were 30% more likely to occur among women who reportedly used tobacco during pregnancy.

- Note: Low birth weight infants with PDA were excluded from analysis of ABDR data.

Prevalence of Patent Ductus Arteriosus by Selected Birth Characteristics Alaska, 1996-2002

	n	Prevalence per 10,000 Live Births	Prevalence Ratio	95% CI
Child Sex				
Female	194	**57.1**	ref	-
Male	222	**62.0**	1.1	(0.9 - 1.3)
Birth Weight*				
Low and Very Low	-	**-**	-	-
Normal	418	**63.5**	-	-
Maternal Race				
White	243	**53.7**	ref	-
Alaska Native	133	**78.3**	1.5	(1.2 - 1.8)
Black	16	**52.6**	1.0	(0.6 - 1.6)
Asian or Pacific Islander	23	**59.0**	1.1	(0.7 - 1.7)
Maternal Age				
15-19 years	46	**59.7**	1.2	(0.9 - 1.7)
20-29 years	239	**63.0**	1.3	(1.0 - 1.6)
30-39 years	110	**49.7**	ref	-
40-45 years	18	**95.2**	1.9	(1.2 - 3.1)
Prenatal Care				
First Trimester	320	**58.2**	ref	-
Second Trimester	68	**65.7**	1.1	(0.9 - 1.5)
Later or None	19	**71.3**	1.0	(1.0 - 1.0)
Maternal Alcohol Use				
Reported	13	**56.6**	0.9	(0.5 - 1.6)
Not Reported	400	**60.0**	ref	-
Maternal Tobacco Use				
Reported	98	**76.0**	1.3	(1.1 - 1.7)
Not Reported	318	**56.4**	ref	-
OVERALL	418	**59.9**	-	-

*418 Infants with patent ductus arteriosus were excluded from birth wieght analysis because the surveillance case definition for patent ductus arteriosus specifies that only infants ≥2500g are counted.

Trends and Distribution

Pulmonary valve stenosis refers to the congenital narrowing of the pulmonary valve, resulting in obstruction of blood flow from the right ventricle. Narrowing to the point of complete obstruction is called pulmonary valve atresia, a condition where a solid sheet of tissue prevents blood flow from the right ventricle to the lungs.

- The overall birth prevalence of pulmonary valve atresia and stenosis in Alaska during 1996-2002 was 27 per 10,000 live births. These valve anomalies made up 9.2% of cardiovascular birth defects reported during the period.

- There was a statistically significant decrease in the prevalence of pulmonary valve stenosis and atresia during 1996-2002 ($p=0.0004$). Overall, the three-year average prevalence declined 42%.

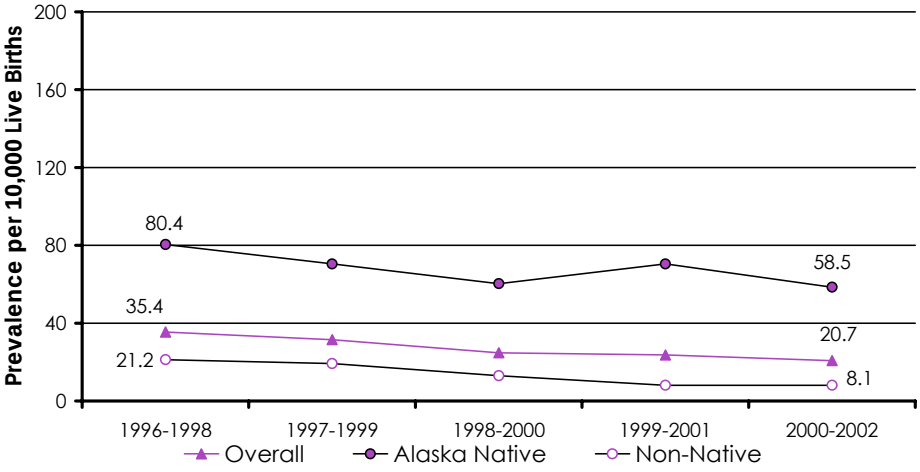
- Significant declines in pulmonary valve stenosis and atresia prevalence were observed for both Alaska Natives and non-Natives during 1996-2002.

- Alaska Natives had higher rates of pulmonary valve stenosis and atresia than non-Natives during 1996-2002 and less of a decline. Additionally, the three-year average prevalence for Alaska Natives declined 27% compared to a decline of 62% for non-Natives.

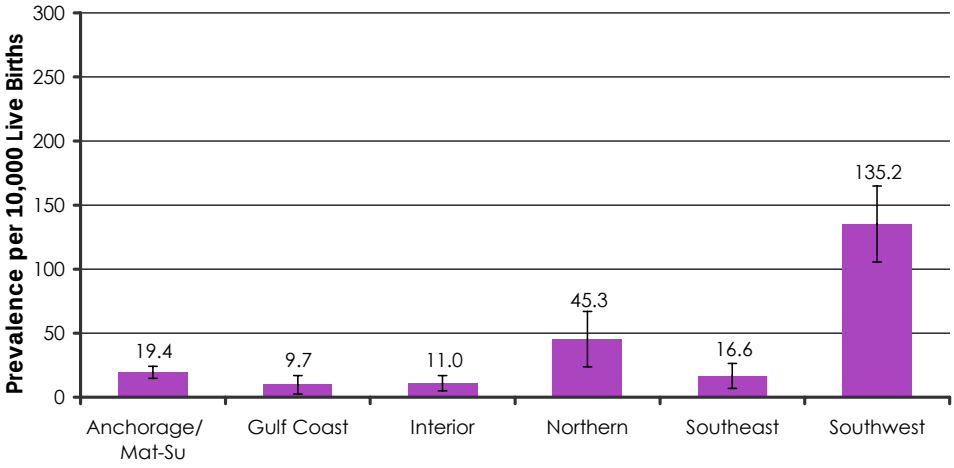
- The prevalence of pulmonary valve stenosis and atresia was significantly higher in the Southwest region of Alaska during 1996-2002, where one in every 100 infants was reported with the anomaly.

Pulmonary Valve Atresia/Stenosis

Prevalence of Pulmonary Valve Atresia and Stenosis By Birth Year and Race Group, Alaska, 1996-2002



Prevalence of Pulmonary Valve Atresia and Stenosis by Region, Alaska, 1996-2002



Epidemiological Characteristics

Pulmonary valve stenosis and atresia are often accompanied by other congenital heart defects. Although the causes of these pulmonary valve defects are unknown, they occur more frequently in families with a history of cardiovascular anomalies (19). Improved diagnostic testing and an increase in preterm deliveries may contribute to observed increases in prevalence of pulmonary valve stenosis and atresia.

- Pulmonary valve stenosis and atresia were equally as common among male and female infants in Alaska during 1996-2002.
- Low birth weight infants were six times as likely to have pulmonary valve stenosis or atresia as infants of normal birth weight.
- Alaska Native women were five times more likely to deliver an infant with pulmonary valve stenosis or atresia than women of other races.
- There were no associations between pulmonary valve stenosis and atresia and maternal age, the trimester prenatal care began, or prenatal tobacco use.
- Reported maternal alcohol use during pregnancy was associated with a 2.5-fold increase in the probability of pulmonary valve stenosis and atresia among Alaskan infants born in 1996-2002.

Prevalence of Pulmonary Valve Atresia and Stenosis by Selected Birth Characteristics Alaska, 1996-2002

	n	Prevalence per 10,000 Live Births	Prevalence Ratio	95% CI
Child Sex				
Female	95	**28.0**	ref	-
Male	100	**27.9**	1.0	(0.8 - 1.3)
Birth Weight				
Low and Very Low	53	**133.0**	6.2	(4.5 - 8.4)
Normal	142	**21.6**	ref	-
Maternal Race				
White	63	**13.9**	ref	-
Alaska Native	121	**71.2**	5.1	(3.8 - 6.9)
Black	n < 5	**-**	-	-
Asian or Pacific Islander	6	**15.4**	1.1	(0.5 - 2.6)
Maternal Age				
15-19 years	19	**24.6**	0.9	(0.5 - 1.5)
20-29 years	109	**28.7**	1.1	(0.8 - 1.5)
30-39 years	60	**27.1**	ref	-
40-45 years	6	**31.7**	1.2	(0.5 - 2.7)
Prenatal Care				
First Trimester	141	**25.6**	ref	-
Second Trimester	33	**31.9**	1.2	(0.9 - 1.8)
Later or None	11	**41.3**	1.6	(0.9 - 3.0)
Maternal Alcohol Use				
Reported	15	**65.4**	2.5	(1.5 - 4.2)
Not Reported	177	**26.6**	ref	-
Maternal Tobacco Use				
Reported	52	**40.4**	1.6	(1.2 - 2.2)
Not Reported	142	**25.2**	ref	-
OVERALL	195	**27.9**	-	-

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Fetal Alcohol Spectrum Disorders

Trends and Distribution

Various birth defects that have been associated with maternal alcohol use during pregnancy are referred to collectively as fetal alcohol spectrum disorders (FASD). Infants with FASD have physical, mental, behavioral or learning disabilities. Only fetal alcohol syndrome (FAS), the most severe subset of FASD, has a clinical definition developed specifically for surveillance purposes. Other birth defects associated with prenatal alcohol exposure are difficult to diagnose and are characterized by a variety of mild to severe disabilities. These include alcohol related neurodevelopmental disorders (ARND) and alcohol related birth defects (ARBD).

In the absence of a specific ICD-9 code for FAS and FASD, the ABDR evaluates ICD-9 code 760.71, *infant affected by prenatal alcohol exposure*, which is a reportable condition in Alaska. In ABDR analysis, FASD is defined as a report to the registry of ICD-9 code 760.71.

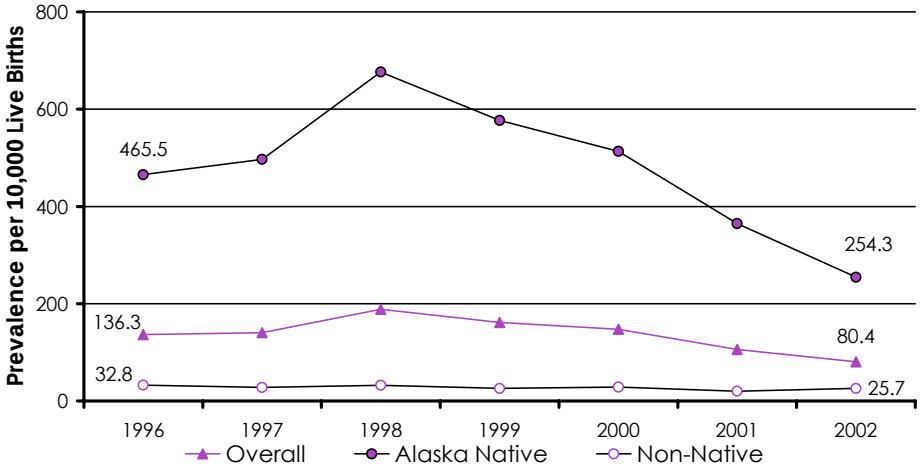
- After cardiovascular anomalies, FASD is the most common type of birth defect reported to the ABDR. As of December 31, 2004, 958 of Alaskan children born in 1996-2001 (1.4% of live births) were reported as having been affected by prenatal alcohol exposure.

- Although the prevalence of FASD appears to have declined during 1996–2002, this trend may not reflect the true birth prevalence of affected children. Because FASD is typically not diagnosed until after the child enters school, prevalence for more recent birth years may be underestimated (children born in 1999-2002 were under age six at the time of this analysis).

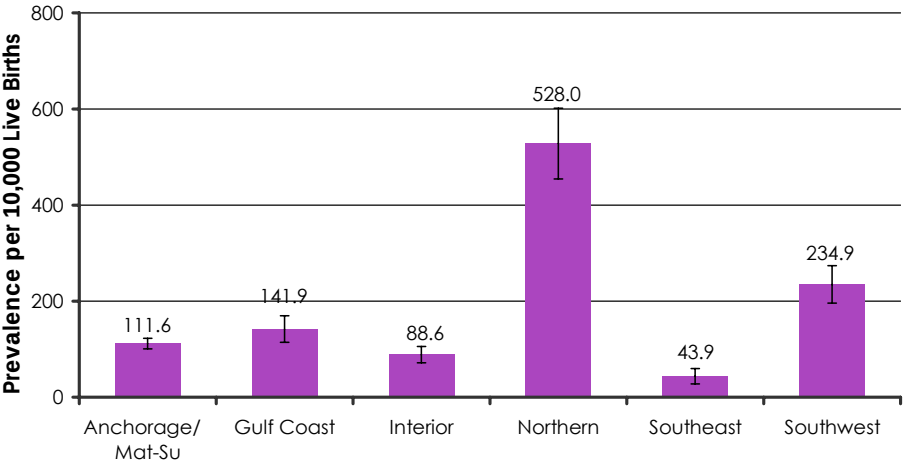
- For each annual birth cohort during 1996-2002, the prevalence of FASD was at least ten times higher for Alaska Natives than non-Natives.

Fetal Alcohol Spectrum Disorders

Prevalence of Fetal Alcohol Spectrum Disorders By Birth Year and Race Group, Alaska, 1996-2002



Prevalence of Fetal Alcohol Spectrum Disorders by Region Alaska, 1996-2002



Epidemiological Characteristics

The U.S. Substance Abuse and Mental Health Services Administration estimates the prevalence of FASD at about 100 per 10,000 live births (1). Brain damage can occur when alcohol crosses the placenta and damages developing tissues. The result may be mild to severe cognitive impairment, mental retardation, social and emotional problems, learning disabilities, visual impairment, neurobehavioral problems and other structural birth defects. Although other etiologies may lead to similar clinical presentations, prenatal alcohol exposure is by definition the only cause of FASD. Other cofactors may be important in modifying the outcome, including maternal alcohol dehydrogenase genotype (2,3).

- Four percent of low birth weight infants in Alaska were reported as having FASD during 1996-2002.

- Five percent of Alaska Native infants born during 1996-2002 were reported as having FASD. The prevalence among whites and blacks was about 16 times lower. Infants delivered of Asian women were significantly less likely than infants of white women to be affected by prenatal alcohol use.

- During 1996-2002, FASD was significantly related to the timing and receipt of prenatal care.

- The prevalence of FASD was strongly associated with a report on the birth certificate of prenatal tobacco use.

- The birth certificates of 58% of infants reported with FASD did not indicate maternal alcohol use during pregnancy.

Note: In analyses of ABDR data, FASD is defined as a report to the registry of ICD-9 code 760.71.

Prevalence of Fetal Alcohol Spectrum Disorder by Selected Birth Characteristics Alaska, 1996-2002

	n	Prevalence per 10,000 Live Births	Prevalence Ratio	95% CI
Child Sex				
Female	417	**122.8**	ref	-
Male	484	**135.2**	1.1	(1.0 - 1.3)
Birth Weight				
Low and Very Low	145	**363.8**	2.9	(2.5 - 3.5)
Normal	813	**123.5**	ref	-
Maternal Race				
White	132	**29.2**	ref	-
Alaska Native	812	**478.0**	16.4	(13.6 - 19.7)
Black	10	**32.9**	1.1	(0.6 - 2.1)
Asian or Pacific Islander	n < 5	**-**	-	-
Maternal Ethnicity				
Hispanic	24	**53.3**	0.4	(0.2 - 0.5)
Non-Hispanic	915	**148.4**	ref	-
Maternal Age				
15-19 years	138	**179.0**	1.2	(1.0 - 1.5)
20-29 years	475	**125.2**	0.9	(0.7 - 1.0)
30-39 years	321	**145.1**	ref	-
40-45 years	21	**111.1**	0.8	(0.5 - 1.2)
Prenatal Care				
First Trimester	557	**101.2**	ref	-
Second Trimester	266	**256.9**	2.5	(2.2 - 2.9)
Later or None	79	**296.3**	2.9	(2.3 - 3.7)
Maternal Alcohol Use				
Reported	-	**-**	-	-
Not Reported	-	**-**	-	-
Maternal Tobacco Use				
Reported	617	**478.8**	8.3	(7.2 - 9.4)
Not Reported	327	**58.0**	ref	-

*Maternal alcohol use is part of the case definition for FASD and is not analyzed as a risk factor.

Specific Anomalies

While diagnostic criteria for FAS have been established, scientific evidence for establishing diagnostic criteria for the two remaining categories of FASD, ARBD and ARND, are insufficient (4). The National Institute on Alcohol Abuse and Alcoholism sponsors research that might lead to evidence-based diagnostic criteria for persons with conditions other than FAS that are caused by prenatal alcohol use (4).

The ABDR reviews the medical records of children reported with FASD (ICD-9 code 769.71) to determine FAS case status using a standardized FAS case definition developed by the FAS Surveillance Network (5). The majority of children reported with FASD in Alaska do not meet the case definition for FAS. Frequently, children who have the neurodevelopment deficits required for FAS diagnoses do not have all of the facial features or growth deficits needed to meet FAS case criteria.

- Infants and children reported with FAS made up 11% of births affected by FASD during 1996-1999.

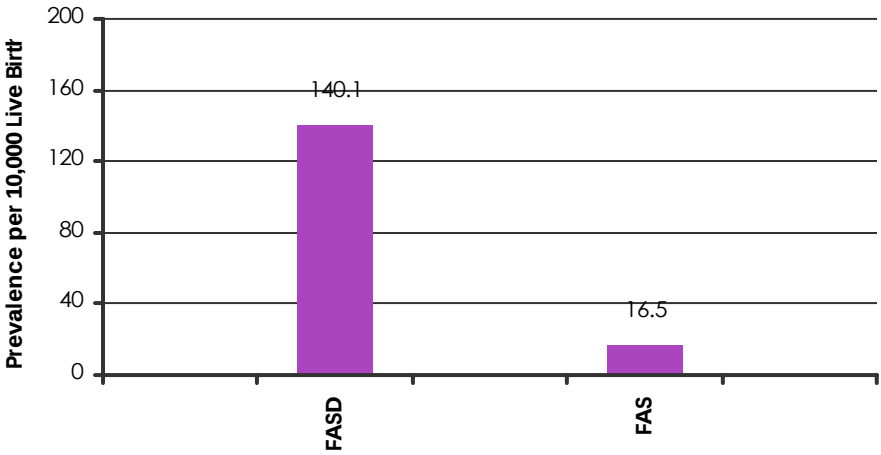
- FAS was diagnosed in an average of 0.2% of children from each annual birth cohort during 1996-1999 (range: 12-25 FAS cases per birth year). About 1.4% of children born each year in Alaska were reported to the ABDR with other conditions related to prenatal alcohol exposure.

- Surveillance data for FASD are presented for birth years 1996-1999 because case ascertainment for more recent birth years will not be complete until these children reach 6 years of age.

Note: In ABDR analysis, FASD is defined as a report to the registry of ICD-9 code 760.71.

Fetal Alcohol Spectrum Disorders

Prevalence of Specific Fetal Alcohol Spectrum Disorders Alaska, 1996-2002



FAS and other congenital conditions associated with prenatal alcohol exposure will be covered in more detail in a future edition of the Alaska MCH Data Book.

Chapter References

Chapter 5: Fetal Alcohol Spectrum Disorders

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Alimentary Tract Anomalies

Trends and Distribution

Alimentary tract anomalies involve the oral cavity, pharynx, esophagus, stomach, and intestine. These birth defects are often referred to as orofacial and gastrointestinal anomalies. Birth defects may occur at multiple sites along the alimentary system and can be severe. Most alimentary tract anomalies can be corrected surgically, but in many cases, even when corrected, respiratory and gastrointestinal complications may persist throughout life.

As a group, alimentary tract anomalies are some of the most common birth defects, often occurring in conjunction with other congenital anomalies. Orofacial anomalies, the most common type of alimentary tract defects, have significant importance internationally and occur in 1 of 700 infants in some parts of the world (1).

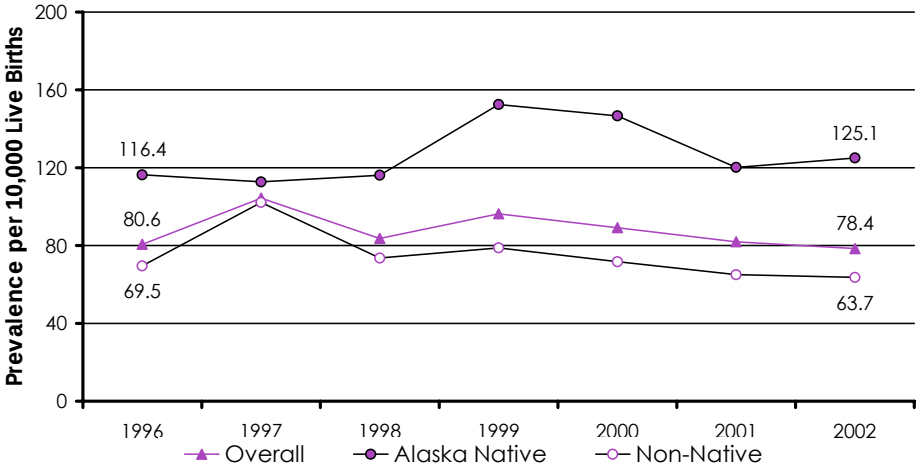
- Alimentary tract anomalies were the third most common group of birth defects in Alaska and affected an average of 88 infants annually during 1996-2002.

- There was no significant trend in annual prevalence of alimentary tract anomalies during 1996-2002. Alaska Natives had a higher prevalence with the annual disparity ranging from one (1997) to two (1999) times that of non-Natives.

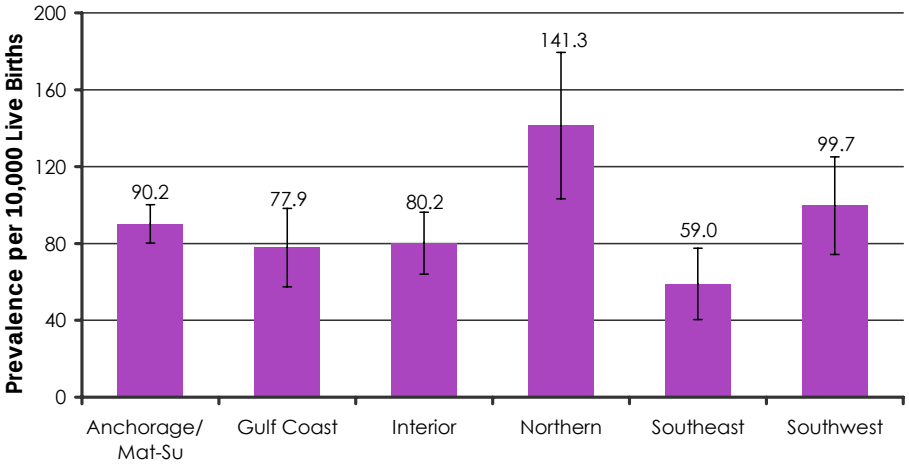
- The Northern region of Alaska had a significantly higher prevalence of alimentary tract anomalies than all other regions except the Southwest. Infants born in the Northern region were over twice as likely to have an alimentary anomaly as infants in the Southeast region, the area with the lowest prevalence.

- At least one in every hundred infants born in the Northern and Southwest regions of Alaska during 1996-2002 was reported with an alimentary tract anomaly.

Prevalence of Alimentary Tract Anomalies By Birth Year and Race Group, Alaska, 1996-2002



Prevalence of Alimentary Tract Anomalies by Region Alaska, Birth Years 1996-2002



Epidemiological Characteristics

While alimentary tract anomalies are related embryologically, epidemiological characteristics of specific conditions are very different and arise from a variety of etiologies. The causes of alimentary tract anomalies are largely unknown, but family history, prescription drugs, infection, gene-environment interactions, environmental toxins, and nutritional deficiencies have reported associations with specific alimentary anomalies.

- Alimentary tract anomalies as a group were significantly associated with male sex during 1996-2002, reflecting the well-established preponderance of male cases in the most commonly diagnosed conditions. A strong association was also observed for low birth weight.

- Infants of Alaska Native mothers were 1.6 times as likely as infants of any other maternal race to be reported with an alimentary tract anomaly during 1996-2002. There was no association between Hispanic ethnicity and the prevalence of alimentary tract anomalies.

- There was an observed linear association between alimentary tract anomalies and maternal age, with the youngest mothers at highest risk for delivering an infant with an alimentary tract anomaly.

- Women who reported alcohol or tobacco use during pregnancy were almost twice as likely (1.6 and 1.8 times respectively) to deliver an infant with an alimentary tract anomaly during 1996-2002.

Prevalence of Alimentary Tract Anomalies by Selected Birth Characteristics Alaska, 1996-2002

	n	Prevalence per 10,000 Live Births	Prevalence Ratio	95% CI
Child Sex				
Female	215	**63.3**	ref	-
Male	392	**109.5**	1.7	(1.5 - 2.0)
Birth Weight				
Low and Very Low	84	**210.7**	2.6	(2.1 - 3.3)
Normal	529	**80.3**	ref	-
Maternal Race				
White	351	**77.6**	ref	-
Alaska Native	216	**127.1**	1.6	(1.4 - 1.9)
Black	19	**62.5**	0.8	(0.5 - 1.3)
Asian or Pacific Islander	21	**53.9**	0.7	(0.4 - 1.1)
Maternal Ethnicity				
Hispanic	38	**84.4**	1.0	(0.7 - 1.3)
Non-Hispanic	539	**87.4**	ref	-
Maternal Age				
15-19 years	92	**119.4**	1.8	(1.4 - 2.3)
20-29 years	354	**93.3**	1.4	(1.1 - 1.7)
30-39 years	150	**67.8**	ref	-
40-45 years	12	**63.5**	0.9	(0.5 - 1.7)
Prenatal Care				
First Trimester	459	**83.4**	ref	-
Second Trimester	101	**97.5**	1.2	(0.9 - 1.4)
Later or None	32	**120.0**	1.4	(1.0 - 2.1)
Maternal Alcohol Use				
Reported	31	**135.1**	1.6	(1.1 - 2.2)
Not Reported	573	**86.0**	ref	-
Maternal Tobacco Use				
Reported	177	**137.3**	1.8	(1.5 - 2.1)
Not Reported	432	**76.6**	ref	-

Specific Anomalies

Eight major anomalies occur at different sites along the alimentary tract. Four of these; pyloric stenosis, cleft lip, Hirshsprung's disease and cleft palate, are included among Alaska's most common birth defects. Other major anomalies of the alimentary tract include gastrointestinal stenosis or atresia, choanal atresia, and biliary atresia.

- Sixteen percent of children reported to the ABDR during 1996-2002 had alimentary tract anomalies.

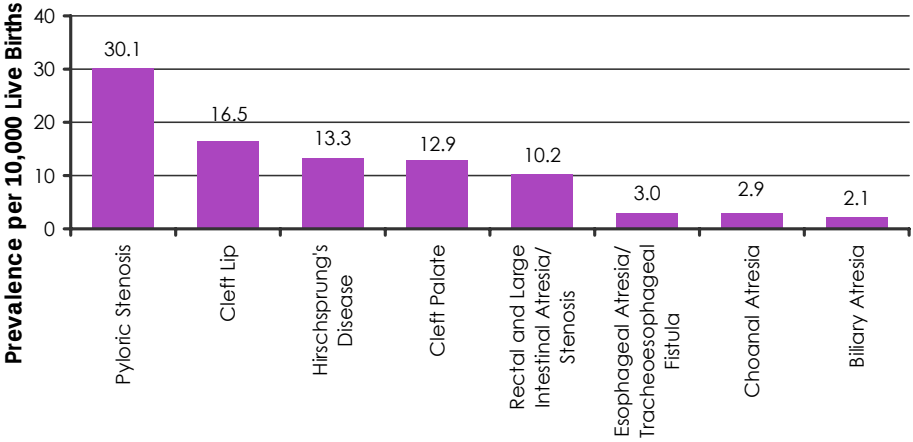
- Twenty three percent of infants born with an alimentary tract anomaly during 1996-2002 had other reported birth defects.

- The most common alimentary tract anomalies were pyloric stenosis, cleft lip, Hirshsprung's disease and cleft palate. Together, these defects made up 83% of alimentary tract anomalies reported to the ABDR and affected an average of 72 infants annually.

- Orofacial anomalies made up 33% and gastrointestinal anomalies 68% of alimentary tract anomalies reported during 1996-2002.

Alimentary Tract Anomalies

Prevalence of Specific Alimentary Tract Anomalies Alaska, 1996-2002



Classification of Major Congenital Anomalies of the Alimentary Tract

Orofacial	Gastrointestinal
Cleft Palate without Cleft Lip Cleft Lip with or without Cleft Palate	Esophageal Atresia Pyloric Stenosis Hirshsprung's Disease Rectal and Intestinal Atresia/Stenosis Tracheoesophageal Fistula Choanal Atresia Biliary Atresia

Trends and Distribution

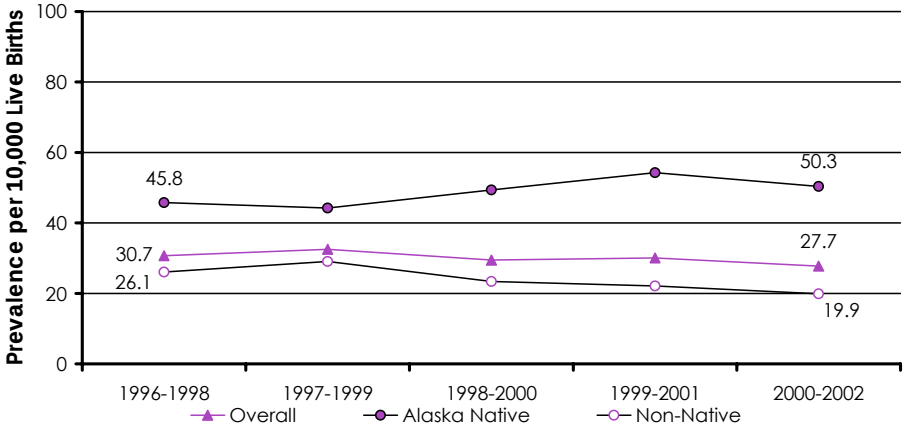
Pyloric stenosis is a condition where a thickening of muscles narrows the opening of the stomach into the small intestine. Hindering or preventing the stomach from emptying, pyloric stenosis causes external symptoms such as vomiting, diarrhea, dehydration and failure to gain weight. One of the most common causes of gastrointestinal obstruction in infants, it is usually not diagnosed at birth, but within the first 3-12 weeks of life (2). The condition must be repaired surgically. Increasing trends in the prevalence of pyloric stenosis have been observed and may be explained by the introduction of diagnostic radiography, which has increased the recognition of mild cases.

- An average of 30 Alaskan infants were born each year with pyloric stenosis during 1996-2002.

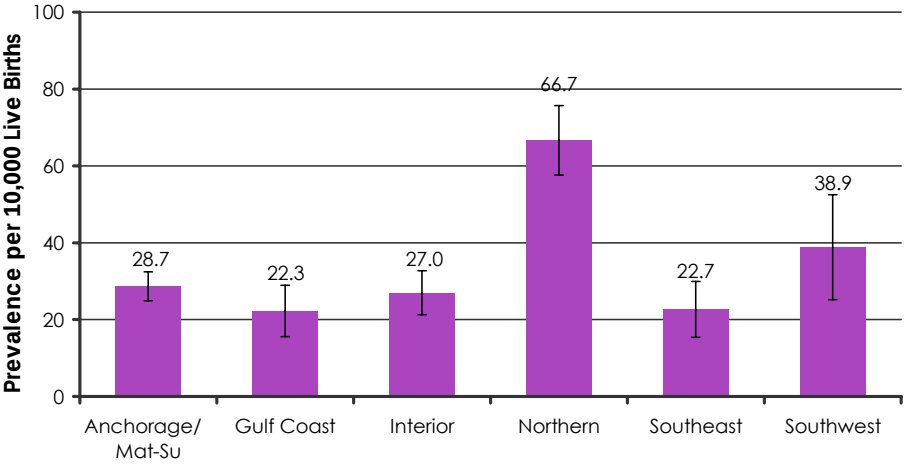
- There was no significant trend in the annual prevalence of pyloric stenosis during 1996-2002 for either Alaska Natives or non-Natives. Throughout the time period, however, the rates were higher for Alaska Natives and the racial disparity widened.

- The prevalence of pyloric stenosis in the Northern region was significantly higher than any other region of the state. This condition, however, affects only 3 to 4 births annually in the low-populated region.

Prevalence of Pyloric Stenosis by Birth Year and Race Group, Alaska, 1996-2002



Prevalence of Pyloric Stenosis by Region Alaska, 1996-2002



Epidemiological Characteristics

The most striking epidemiological feature of pyloric stenosis is its strong association with male sex. Reports from outside Alaska indicate that the condition is from 3 to 6.5 times more common among males and more likely to appear in whites than other races (3). Pyloric stenosis is strongly associated with a positive family history for the condition and is also associated with chromosomal syndromes. Research to identify the mode of inheritance, however, has been inconclusive (4).

- Consistent with well established epidemiological findings, male sex was associated with pyloric stenosis in Alaskan infants born during 1996-2002.

- Low birth weight displayed a weak association with pyloric stenosis during 1996-2002. Other studies have shown discordant findings on the association of pyloric stenosis with low birth weight.

- Alaska Native mothers were almost twice as likely as whites, and Asian and Pacific Islanders were significantly less likely, to deliver an infant with pyloric stenosis during 1996-2002.

- Pyloric stenosis was more common among infants of young Alaskan mothers than those of women thirty years of age and older. Other studies have demonstrated an association with young maternal age as well as low parity (3).

- Maternal tobacco use was significantly associated with pyloric stenosis in Alaska. One in every 167 births where prenatal smoking was documented on the birth certificate was affected by pyloric stenosis. Maternal alcohol use displayed no such association.

Prevalence of Pyloric Stenosis by Selected Birth Characteristics Alaska, 1996-2002

	n	Prevalence per 10,000 Live Births	Prevalence Ratio	95% CI
Child Sex				
Female	53	**15.6**	ref	-
Male	155	**43.3**	2.8	(2.0 - 3.8)
Birth Weight				
Low and Very Low	20	**50.2**	1.7	(1.1 - 2.8)
Normal	190	**28.9**	ref	-
Maternal Race				
White	116	**25.6**	ref	-
Alaska Native	84	**49.4**	1.9	(1.5 - 2.6)
Black	5	**16.4**	0.6	(0.3 - 1.6)
Asian or Pacific Islander	n < 5	**-**	-	-
Maternal Age				
15-19 years	30	**38.9**	2.2	(1.4 - 3.6)
20-29 years	137	**36.1**	2.0	(1.4 - 2.9)
30-39 years	39	**17.6**	ref	-
40-45 years	n < 5	**-**	-	-
Prenatal Care				
First Trimester	160	**29.1**	ref	-
Second Trimester	31	**29.9**	1.0	(0.7 - 1.5)
Later or None	11	**41.3**	1.4	(0.8 - 2.6)
Maternal Alcohol Use				
Reported	10	**43.6**	1.5	(0.8 - 2.8)
Not Reported	197	**29.6**	ref	-
Maternal Tobacco Use				
Reported	77	**59.8**	2.6	(1.9 - 3.4)
Not Reported	132	**23.4**	ref	-
OVERALL	210	**30.1**	-	-

Trends and Distribution

Incomplete fusion of the lip during the 6th through the 9th week of gestation is known as cleft lip. Severity can vary from a subtle notch not exteriorly visible to a complete fissure in the upper lip involving the floor of the nose. In addition to physical deformation, cleft lip can be the cause of feeding and speech problems. Cleft lip is usually closed surgically between 3-9 months of age. Extensive nasal involvement may warrant further surgery.

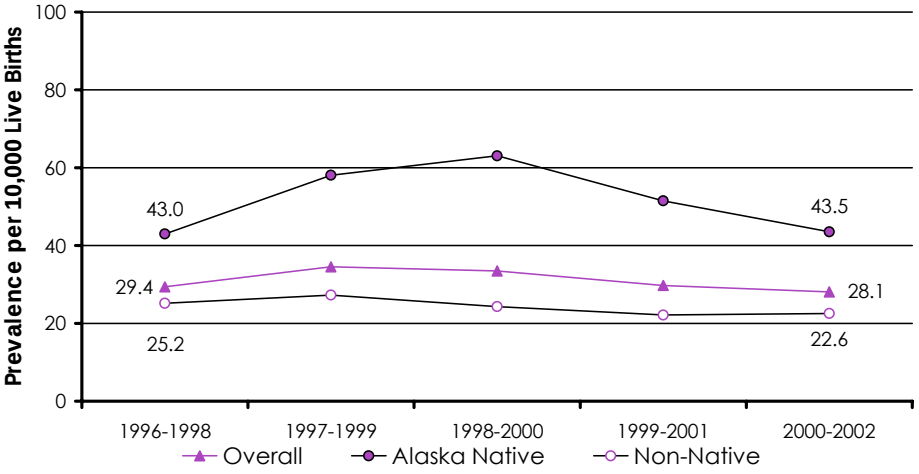
In normal fetal development, the palate (or roof of the mouth) closes by the tenth week. Incomplete closure of the palate is referred to as cleft palate. Depending on the severity, cleft palate can cause dental problems, difficulty with feeding, difficulty with speech development, and ear disease. Cleft palate can be surgically repaired. The initial surgery usually takes place between 9 and 18 months of age and may be followed by additional surgeries as the child grows.

- Oral clefts include cleft lip with or without cleft palate and cleft palate without cleft lip. Oral clefts comprised 5% of major anomalies reported to the ABDR and affected an average of 29 Alaskan infants annually during 1996-2002.

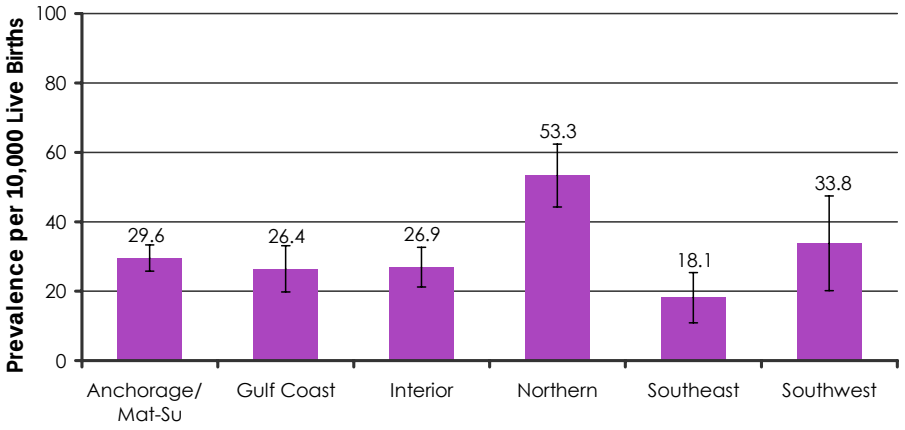
- There were no statistically significant trends in the annual prevalence of oral clefts during 1996-2002, regardless of whether oral clefts were grouped or examined separately.

- For both cleft lip with or without cleft palate and isolated cleft palate, the prevalence in the Northern region was highest. Cleft lip with or without cleft palate was more common than isolated cleft palate in all regions of Alaska except the Northern and Southeast regions.

Birth Prevalence of Oral Clefts by Birth Year and Race Group, Alaska, 1996-2002



Prevalence of Oral Clefts by Region Alaska, 1996-2002



Epidemiological Characteristics

Oral clefts occur as isolated conditions and in association with other birth defects. Over 150 syndromes include oral clefts (5). Genetic factors are generally considered to be more important than environmental exposures in the etiology of oral clefts, in part due to a strong association with race. The prevalence of oral clefts is highest among Asians, and lowest among blacks (6). Several studies have also found high rates among Native Americans (7). Maternal tobacco use, alcohol consumption, poor nutrition, and some prescription drugs have been reported to increase the risk of oral clefts, but study findings are inconsistent and probably influenced by gene-environment interactions (7,8). There is growing evidence that taking folic acid supplements during pregnancy and the pre-conception period could reduce the risk of oral clefts (7).

- Oral clefts were more common among male than female infants in Alaska during 1996-2002, a finding consistent with other epidemiological studies (6,7). Low birth weight was associated strongly with oral clefts, particularly isolated cleft palate.

- Teenage mothers were more likely than other age groups to deliver an infant with an oral cleft during 1996-2002. This association was strongest for cleft lip with or without cleft palate.

- Early prenatal care had a protective effect on the prevalence of oral clefts overall, but the association was only statistically significant for cleft lip with or without cleft palate. Women who began prenatal care in the second trimester were twice as likely to deliver an infant with cleft lip with or without cleft palate than women who began care in the first trimester.

- Oral clefts were 1.5 times more prevalent among women with reported prenatal tobacco use during 1996-2001. The association was strongest for cleft lip with or without cleft palate.

Prevalence of Oral Clefts by Selected Birth Characteristics Alaska, 1996-2002

	n	Prevalence per 10,000 Live Births	Prevalence Ratio	95% CI
Child Sex				
Female	86	**25.3**	ref	-
Male	117	**32.7**	1.3	(1.0 - 1.7)
Birth Weight				
Low and Very Low	25	**62.7**	2.3	(1.5 - 3.5)
Normal	180	**27.3**	ref	-
Maternal Race				
White	107	**23.7**	ref	-
Alaska Native	80	**47.1**	2.0	(1.5 - 2.7)
Black	6	**19.7**	0.8	(0.4 - 1.9)
Asian or Pacific Islander	10	**25.7**	1.1	(0.6 - 2.1)
Maternal Age				
15-19 years	34	**44.1**	1.8	(1.2 - 2.8)
20-29 years	108	**28.5**	1.2	(0.8 - 1.6)
30-39 years	54	**24.4**	ref	-
40-45 years	7	**37.0**	1.5	(0.7 - 3.3)
Prenatal Care				
First Trimester	143	**26.0**	ref	-
Second Trimester	43	**41.5**	1.6	(1.1 - 2.2)
Later or None	10	**37.5**	1.4	(0.8 - 2.7)
Maternal Alcohol Use				
Reported	11	**47.9**	1.7	(0.9 - 3.1)
Not Reported	190	**28.5**	ref	-
Maternal Tobacco Use				
Reported	51	**39.6**	1.5	(1.1 - 2.0)
Not Reported	151	**26.8**	ref	-
OVERALL	201	**28.8**	-	-

Trends and Distribution

Hirschsprung's disease, or congenital megacolon, occurs when a section of the colon lacks nerve cells, halting movement of material in the intestinal tract and causing an obstruction. Depending on the severity of the anomaly, the obstruction can result in chronic constipation and abdominal distension or in an inability to pass fecal matter. To treat the anomaly, the affected section of colon must be surgically removed, eliminating symptoms in 90% of patients (9).

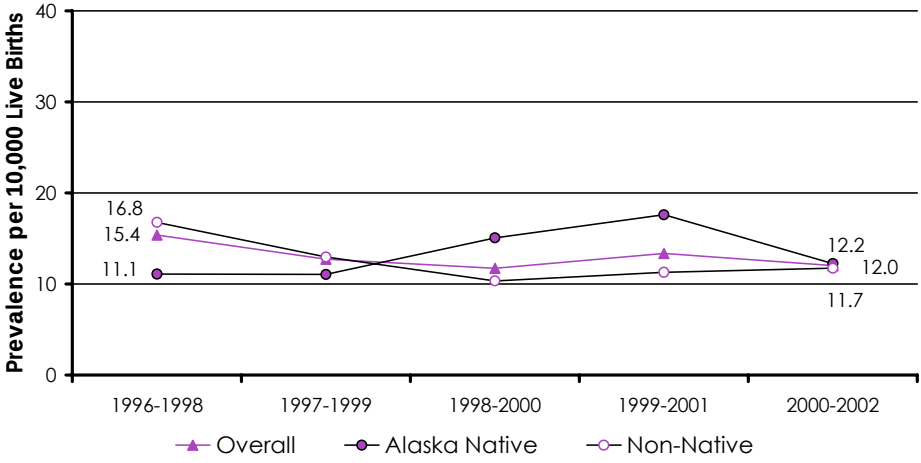
- Hirschsprung's disease is epidemiologically significant in Alaska because of its high overall prevalence. While it is generally thought to occur in one in every 5,000 infants (10), Hirschsprung's disease was reported in an average of six in every 5000 infants born in Alaska during 1996-2002.

- A small, but statistically significant decrease in the prevalence of Hirschsprung's disease among non-Natives during 1996-2002 ($p=0.05$) resulted in a marginally significant overall decline. There were no significant racial disparities.

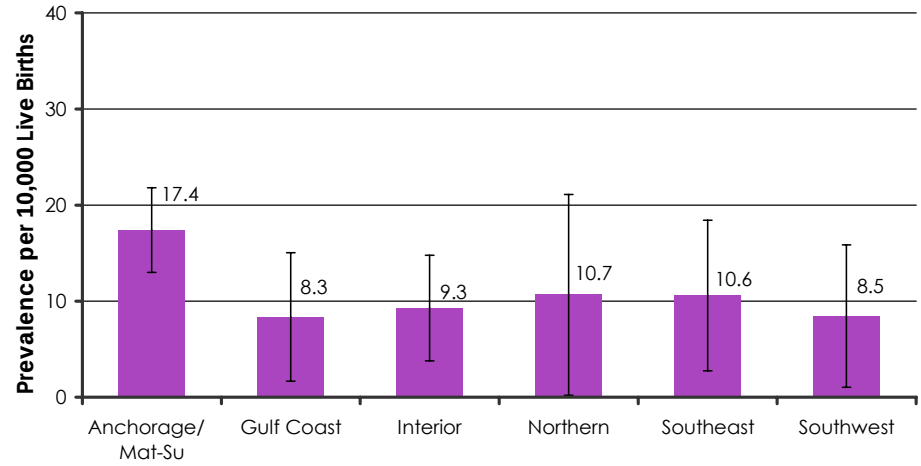
- During 1996-2002, the highest rate of Hirschsprung's disease was reported from the Anchorage/Mat-Su region; however, there were no statistically significant differences in Hirschsprung's disease prevalence by region.

- Temporal and regional analyses did not present any important clues to the high prevalence of Hirschsprung's disease in Alaska.

Prevalence of Hirschsprung's Disease by Birth Year and Race Group, Alaska 1996-2002



Birth Prevalence of Hirschsprung's Disease by Region Alaska, 1996-2002



Epidemiological Characteristics

Hirschsprung's disease presents more commonly in males than females and is nearly always diagnosed within the first two years of life. Caused by improper migration of neural cells early in fetal development, the disease was widely believed to be spontaneous until recent advances in genetic studies identified Hirschsprung's disease as primarily an inherited condition (10). Genetic studies are exploring specific inherited gene mutations believed to be responsible for familial cases (11). No environmental factors have been linked to Hirschsprung's disease.

- For Alaskan infants born in 1996-2002, the prevalence of Hirschsprung's disease was twice as high among males as females. Other studies have reported four to six fold differences (9,10).

- Low birth weight infants were three times more likely than normal birth weight infants to have Hirschsprung's disease.

- Maternal race and age were not associated with Hirschsprung's disease in Alaskan infants born in 1996-2002. Trimester of prenatal care and reported maternal alcohol use also showed no association with the condition in Alaska.

- Infants born to mothers who reported prenatal tobacco use were twice as likely to have Hirschsprung's disease as infants born to mothers without reported prenatal tobacco use during 1996-2002.

Prevalence of Hirschsprung's Disease by Selected Birth Characteristics Alaska, 1996-2002

	n	Prevalence per 10,000 Live Births	Prevalence Ratio	95% CI
Child Sex				
Female	28	**8.2**	ref	-
Male	65	**18.2**	2.2	(1.4 - 3.4)
Birth Weight				
Low and Very Low	15	**37.6**	3.2	(1.8 - 5.5)
Normal	78	**11.8**	ref	-
Maternal Race				
White	61	**13.5**	ref	-
Alaska Native	21	**12.4**	0.9	(0.6 - 1.5)
Black	6	**19.7**	1.5	(0.6 - 3.4)
Asian or Pacific Islander	n < 5	**-**	-	-
Maternal Age				
15-19 years	12	**15.6**	1.5	(0.7 - 3.0)
20-29 years	55	**14.5**	1.4	(0.9 - 2.3)
30-39 years	23	**10.4**	ref	-
40-45 years	n < 5	**-**	-	-
Prenatal Care				
First Trimester	73	**13.3**	ref	-
Second Trimester	12	**11.6**	0.9	(0.5 - 1.6)
Later or None	5	**18.8**	1.4	(0.6 - 3.5)
Maternal Alcohol Use				
Reported	n < 5	**-**	-	-
Not Reported	90	**13.5**	ref	-
Maternal Tobacco Use				
Reported	28	**21.7**	1.9	(1.2 - 2.9)
Not Reported	65	**11.5**	ref	-
OVERALL	93	**13.3**	-	-

Chapter References

Chapter 6: Alimentary Tract Anomalies

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Genitourinary Anomalies

Trends and Distribution

Genitourinary anomalies are congenital malformations of the urinary tract and reproductive system. As a group, these anomalies are relatively common and include both rare, life threatening anomalies and less severe, but more common anomalies that may be corrected surgically.

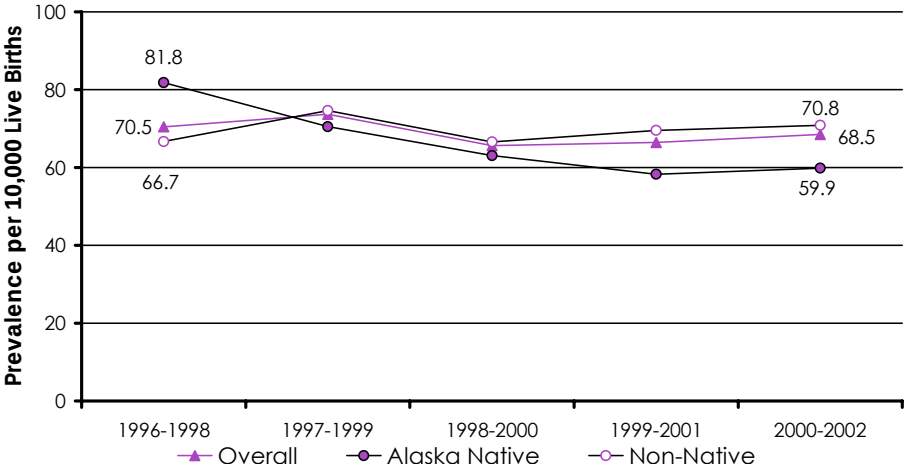
- During 1996-2002 there was no significant change in the prevalence of genitourinary anomalies in Alaska. In contrast, increasing trends in the prevalence of genitourinary anomalies have been widely reported for other populations (1,2).

- Although there was a 27% decline in the three-year average prevalence for Alaska Natives, this decline was not statistically significant.

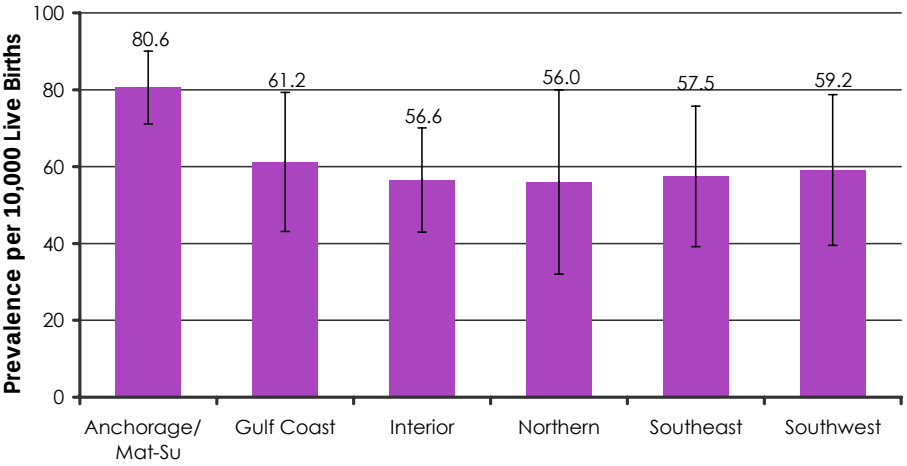
- Alaska Natives had lower rates of genitourinary anomalies than non-Natives during 1996-2002.

- The regional distribution of genitourinary anomalies was consistent during 1996-2002 (56-61 reported cases per 10,000 births) for all regions except Anchorage/Mat-Su, where genitourinary anomalies affected 81 infants per 10,000 live births. Although higher, the prevalence in the Anchorage Mat-Su region was not statistically different from the prevalence reported in other regions.

Prevalence of Genitourinary Anomalies by Birth Year and Race Group, Alaska, 1996-2002



Prevalence of Genitourinary Anomalies by Region Alaska, 1996-2002



Epidemiological Characteristics

The prevalence of genitourinary anomalies is increasing internationally. Some of the increase may be due to improved diagnoses of less severe and more common conditions. Risk factors for genitourinary malformations include chromosomal and other genetic anomalies as well as male gender.

- Genitourinary anomalies were reported five times more frequently in male Alaskan infants than females during 1996-2002.
- Low birth weight infants were almost three times more likely to have a genitourinary birth defect than normal birth weight infants.
- There was no association between race or Hispanic ethnicity and genitourinary anomalies during 1996-2002.
- Although a slightly increased prevalence of genitourinary anomalies was observed among infants of teenage mothers, there was no significant association with maternal age, or trimester of prenatal care.
- Neither prenatal maternal tobacco use nor alcohol use had any demonstrable association with genitourinary anomalies during 1996-2002.

Prevalence of Genitourinary Anomalies by Selected Birth Characteristics Alaska, 1996-2002

	n	Prevalence per 10,000 Live Births	Prevalence Ratio	95% CI
Child Sex				
Female	75	**22.1**	ref	-
Male	405	**113.2**	5.1	(4.0 - 6.6)
Birth Weight				
Low and Very Low	67	**168.1**	2.7	(2.1 - 3.4)
Normal	416	**63.2**	ref	-
Maternal Race				
White	314	**69.4**	ref	-
Alaska Native	112	**65.9**	0.9	(0.8 - 1.2)
Black	25	**82.2**	1.2	(0.8 - 1.8)
Asian or Pacific Islander	26	**66.7**	1.0	(0.6 - 1.4)
Maternal Ethnicity				
Hispanic	35	**77.8**	1.1	(0.8 - 1.6)
Non-Hispanic	422	**68.4**	ref	-
Maternal Age				
15-19 years	63	**81.7**	1.3	(1.0 - 1.8)
20-29 years	267	**70.4**	1.2	(0.9 - 1.4)
30-39 years	134	**60.6**	ref	-
40-45 years	15	**79.4**	1.3	(0.8 - 2.2)
Prenatal Care				
First Trimester	389	**70.7**	ref	-
Second Trimester	58	**56.0**	0.8	(0.6 - 1.0)
Later or None	23	**86.3**	1.2	(0.8 - 1.9)
Maternal Alcohol Use				
Reported	18	**78.4**	1.1	(0.7 - 1.8)
Not Reported	455	**68.3**	ref	-
Maternal Tobacco Use				
Reported	99	**76.8**	1.1	(0.9 - 1.4)
Not Reported	379	**67.2**	ref	-

Specific Anomalies

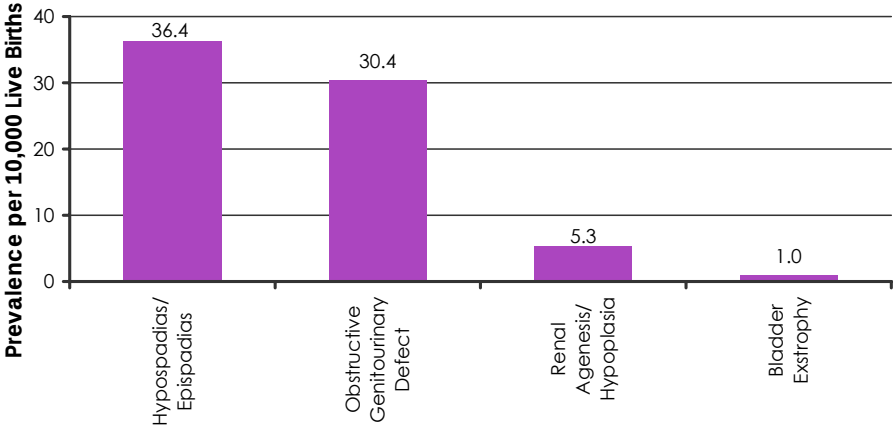
Four genitourinary anomalies are classified as major anomalies: hypospadias and epispadias, obstructive genitourinary defect, renal agenesis/hypoplasia and bladder exstrophy. The latter two are rare congenital anomalies, particularly bladder exstrophy, which is estimated to occur in 3 births per 100,000 nationally (3).

- Twelve percent of children with major anomalies reported to the ABDR during 1996-2002 had genitourinary anomalies.

- The two most common major anomalies of the genitourinary system, hypospadias/epispadias and obstructive genitourinary defect, are among the 15 most frequently reported specific anomalies in Alaska, ranking 4th and 6th respectively. These conditions make up 96% percent of genitourinary anomalies reported in 1996-2002 and affected an average of 67 live births annually.

- An average of five infants were born each year with renal agenesis/hypoplasia and only one each year with bladder exstrophy during 1996-2002.

Prevalence of Specific Genitourinary Anomalies Alaska, 1996-2002



Trends and Distribution

Hypospadias is a malformation of the male urinary tract where the opening of the urethra is located on the underside as opposed to the end (glans) of the penis. Far less common, epispadias is a malformation where the opening of the urethra is located on the upper side of the penis in boys and usually between the clitoris and labia in girls. Both hypospadias and epispadias are treatable by surgical procedures. In classification as major anomalies by the NBDPN, hypospadias and epispadias are grouped together.

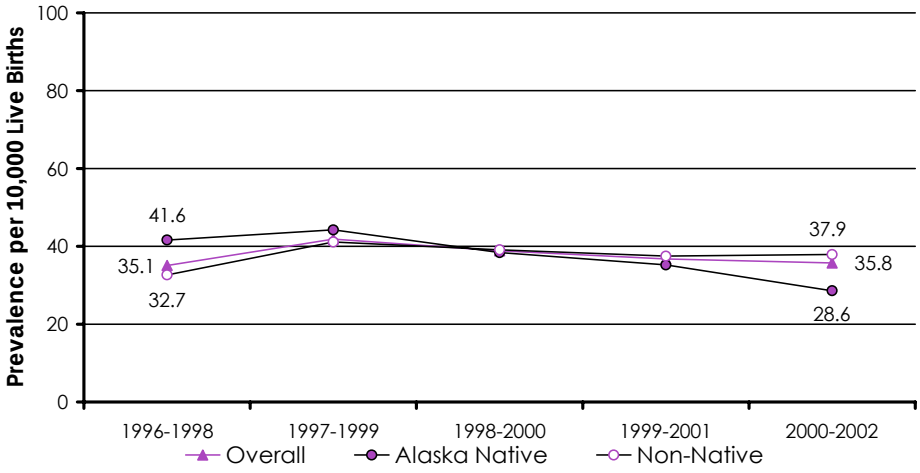
- Grouped together, hypospadias and epispadias were the fourth most commonly reported major anomaly in Alaska during 1996-2002, affecting an average of 36 children annually.

- There was no change in the overall prevalence of hypospadias/epispadias during 1996-2002 in Alaska. The three-year average prevalence of these conditions declined for Alaska Natives, but the trend was not statistically significant.

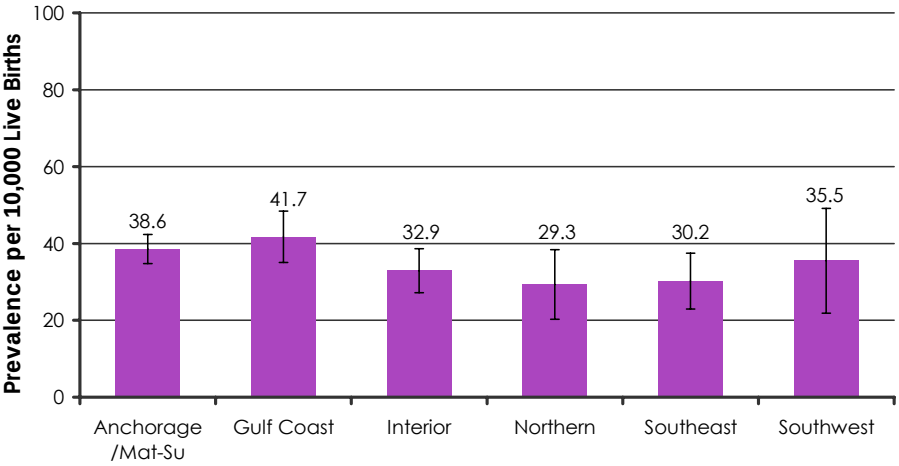
- The occurrence of hypospadias and epispadias was uniformly distributed across the state with no significant regional differences. Prevalence estimates for hypospadias/epispadias during 1996-2002 ranged from 29 (Northern) to 42 (Gulf Coast) cases per 10,000 live births.

Hypospadias and Epispadias

Prevalence of Hypospadias and Epispadias by Birth Year and Race Group, Alaska, 1996-2002



Prevalence of Hypospadias and Epispadias by Region Alaska, 1996-2002



Epidemiological Characteristics

Hypospadias rates in the U.S. and some European countries increased during the 1970s and 1980s, which was thought to be a result of more sensitive surveillance and diagnosis of mild cases (4). Studies suggest an increased risk of hypospadias with maternal intake of progestins, commonly prescribed during pregnancy (5) and in cases of in-vitro fertilization (6). Increasing maternal age has also been identified as a potential risk factor (7).

Less conclusive research exists that examines the possible risk factors associated with epispadias. Due to difficulty in diagnosis, incidence of epispadias in females is expected to be higher than current rates reflect (8).

- Only about 1% of hypospadias/epispadias cases reported during 1996-2002 in Alaska were females.
- Low and very low birth weight were associated with an increased prevalence of hypospadias/epispadias, with these conditions reported in almost 1% of low and very low birth weight infants.
- There were no significant racial differences in the prevalence of hypospadias/epispadias during 1996-2002.
- Hypospadias/epispadias was more common among infants born of teen mothers than those born of mothers aged 30-39, a finding that contrasts with that reported for other populations 7).
- There was no association between hypospadias/epispadias and timing of prenatal care, reported maternal alcohol use, or reported maternal tobacco use.

Prevalence of Hypospadias and Epispadias by Selected Birth Characteristics Alaska, 1996-2002

	n	Prevalence per 10,000 Live Births	Prevalence Ratio	95% CI
Child Sex				
Female	n < 5	-	-	-
Male	248	**69.3**	78.4	(25.1 - 244.9)
Birth Weight				
Low and Very Low	35	**87.8**	2.6	(1.9 - 3.8)
Normal	219	**33.3**	ref	-
Maternal Race				
White	163	**36.0**	ref	-
Alaska Native	60	**35.3**	1.0	(0.7 - 1.3)
Black	11	**36.2**	1.0	(0.5 - 1.8)
Asian or Pacific Islander	17	**43.6**	1.2	(0.7 - 2.0)
Maternal Age				
15-19 years	35	**45.4**	1.4	(1.0 - 2.2)
20-29 years	142	**37.4**	1.2	(0.9 - 1.6)
30-39 years	70	**31.6**	ref	-
40-45 years	5	**26.5**	0.8	(0.3 - 2.1)
Prenatal Care				
First Trimester	204	**37.1**	ref	-
Second Trimester	34	**32.8**	0.9	(0.6 - 1.3)
Later or None	12	**45.0**	1.2	(0.7 - 2.2)
Maternal Alcohol Use				
Reported	8	**34.9**	1.0	(0.5 - 2.0)
Not Reported	240	**36.0**	ref	-
Maternal Tobacco Use				
Reported	45	**34.9**	1.0	(0.7 - 1.3)
Not Reported	205	**36.3**	ref	-
OVERALL	254	**36.4**	-	-

Trends and Distribution

Obstructive genitourinary defects refer to congenital anomalies that cause a blockage at any point along the urinary tract, including the kidneys, ureters, bladder, urethra and genitals. Urinary tract anomalies are among the most common of birth defects, and the resulting symptoms vary widely depending upon the severity and the tissue affected. Obstructive genitourinary defects are among the major causes of chronic kidney disease in infants and children. Many aspects of the pathogenesis, etiology and treatment of the defects, however, have not been well studied or defined (9). Some defects can go untreated while others may require surgical correction.

- Obstructive genitourinary defects are the sixth most commonly reported major congenital anomalies in Alaska. During 1996-2002, an average of 30 infants each year were born in Alaska with an obstructive genitourinary defect.

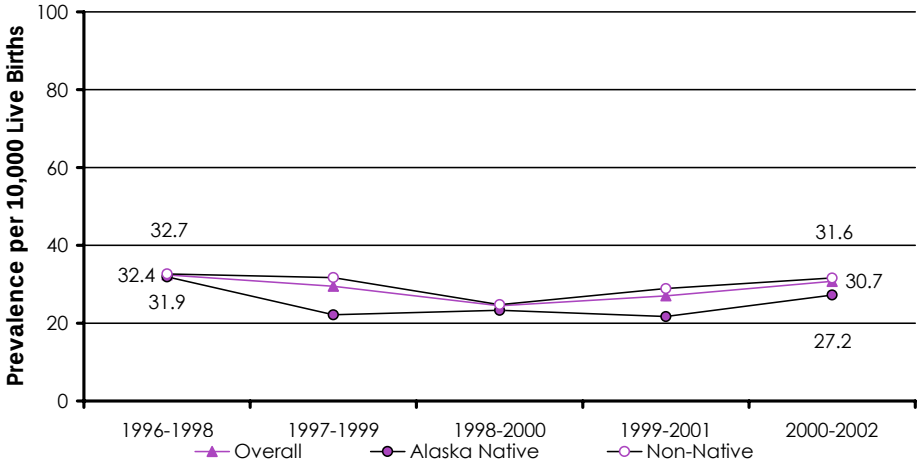
- The prevalence of obstructive genitourinary defects remained fairly constant during 1996-2002, with no significant trend in the prevalence for either Alaska Natives or non-Natives. In general, Alaska Natives had slightly lower rates than non-Natives during the time period.

- A significantly higher rate of obstructive genitourinary defects was reported from the Anchorage/Mat-Su region during 1996-2002. An average of 20 infants per year were born in this region with an obstructive genitourinary defect.

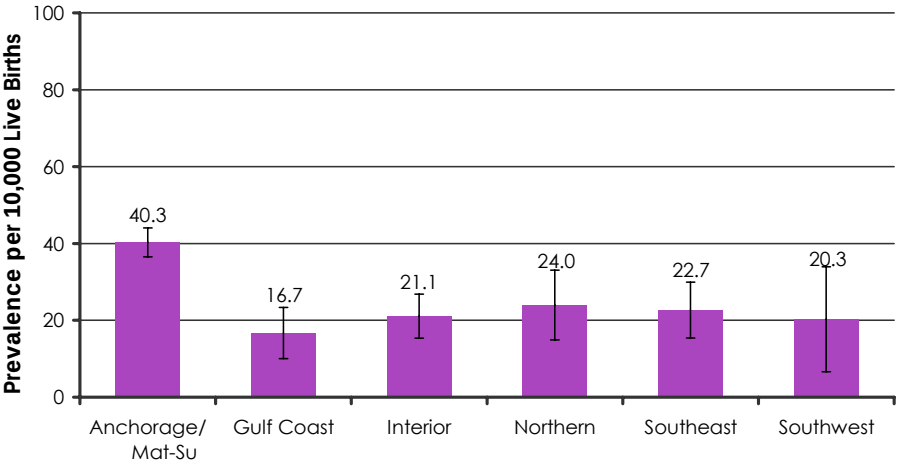
- Obstructive genitourinary defects were much less common in other regions of Alaska where prevalence ranged from 17-24 per 10,000 live births. Interregional differences in obstructive genitourinary defects prevalence for regions outside Anchorage/Mat-Su were not statistically different.

Obstructive Genitourinary Defects

Prevalence of Obstructive Genitourinary Defects by Birth Year and Race Group, Alaska, 1996-2002



Prevalence of Obstructive Genitourinary Defects by Region, Alaska, 1996-2002



Epidemiological Characteristics

The cause of obstructive genitourinary defects appears to be multifactorial, although inherited cases are known. Studies suggest an increased risk of congenital urinary tract anomalies associated with maternal cocaine use (10) and maternal smoking (11). The epidemiology of obstructive genitourinary defects has not been well documented. In general, however, congenital anomalies of the urinary tract occur more frequently in males than in females (12).

- Male infants in Alaska were twice as likely to have an obstructive genitourinary defect as females during 1996-2002.
- Obstructive genitourinary defects were three times more likely to be reported among low and very low birth weight infants.
- Obstructive genitourinary defect prevalence did not differ significantly by maternal race.
- Obstructive genitourinary defects were not significantly associated with maternal age, trimester of prenatal care during 1996-2002, or maternal prenatal tobacco or alcohol use.

Prevalence of Obstructive Genitourinary Defect by Selected Birth Characteristics Alaska, 1996-2002

	n	Prevalence per 10,000 Live Births	Prevalence Ratio	95% CI
Child Sex				
Female	61	**18.0**	ref	-
Male	151	**42.2**	2.3	(1.7 - 3.2)
Birth Weight				
Low and Very Low	32	**80.3**	2.9	(2.0 - 4.3)
Normal	180	**27.3**	ref	-
Maternal Race				
White	140	**31.0**	ref	-
Alaska Native	44	**25.9**	0.8	(0.6 - 1.2)
Black	14	**46.0**	1.5	(0.9 - 2.6)
Asian or Pacific Islander	11	**28.2**	0.9	(0.5 - 1.7)
Maternal Age				
15-19 years	25	**32.4**	1.3	(0.8 - 2.0)
20-29 years	119	**31.4**	1.2	(0.9 - 1.7)
30-39 years	57	**25.8**	ref	-
40-45 years	9	**47.6**	1.8	(0.9 - 3.7)
Prenatal Care				
First Trimester	170	**30.9**	ref	-
Second Trimester	25	**24.1**	0.8	(0.5 - 1.2)
Later or None	9	**33.8**	1.1	(0.6 - 2.1)
Maternal Alcohol Use				
Reported	11	**47.9**	1.6	(0.9 - 3.0)
Not Reported	198	**29.7**	ref	-
Maternal Tobacco Use				
Reported	48	**37.2**	1.3	(0.9 - 1.8)
Not Reported	163	**28.9**	ref	-
OVERALL	212	**30.4**	-	-

Chapter References

Chapter 7: Genitourinary Anomalies

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Central Nervous System Anomalies

Trends and Distribution

The brain and spinal cord make up the central nervous system. The basic structures of the central nervous system have formed by the 6th to 7th week of gestation, often before the woman is aware of her pregnancy. Structural anomalies of the central nervous system therefore arise very early in pregnancy and are typically severe. Because of the severity of the anomalies, many infants are not carried to term. Miscarriage is common, and central nervous system anomalies are associated with higher rates of elective abortion (1). For this reason, estimates of birth prevalence underestimate the frequency of central nervous system anomalies. Among children whose death was associated with a birth defect, 15% had a central nervous system anomaly (2).

- The overall prevalence of central nervous system anomalies in Alaska during 1996-2002 was 49 per 10,000 live births.

- During 1996-2002, there were no significant trends in the annual prevalence of central nervous system anomalies for either Natives or non-Natives.

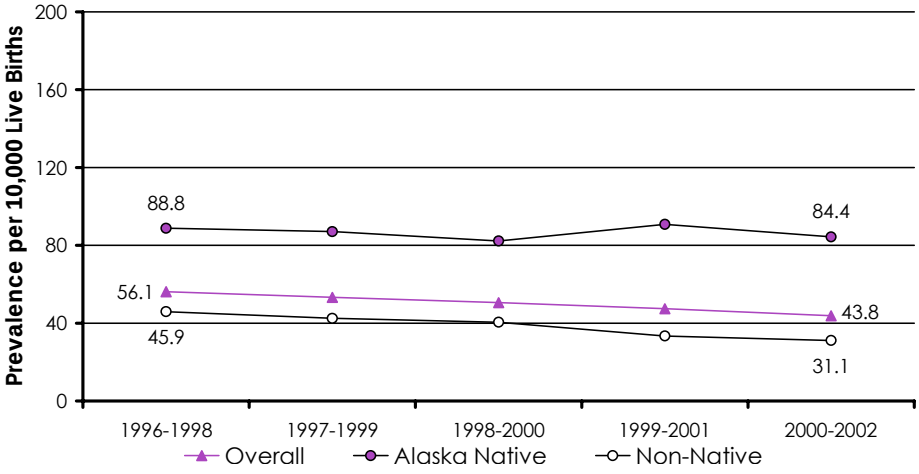
- Alaska Natives had higher rates of central nervous system anomalies than non-Natives during 1996-2002. The ratio of the Native to non-Native prevalence increased from 1.9 in 1996-1999 to 2.7 in 2000-2002.

- The prevalence of central nervous system anomalies was highest in the Southwest region during 1996-2002, where an average of eight infants per year were reported with central nervous system anomalies.

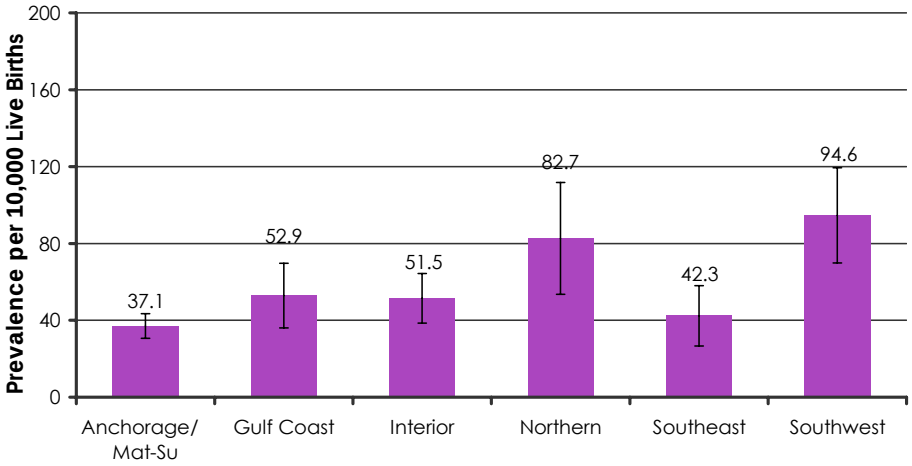
- The prevalence of central nervous system anomalies was lowest (37.1 per 10,000 live births) in the Anchorage/Mat-Su region where an average of 18 infants were reported annually during 1996-2002.

Central Nervous System Anomalies

Prevalence of Central Nervous System Anomalies by Birth Year and Race Group, Alaska, 1996-2002



Prevalence of Central Nervous System Anomalies by Region, Alaska, 1996-2002



Epidemiological Characteristics

Birth defects of the central nervous system are thought to be caused by interacting genetic and environmental factors. Causes of central nervous system anomalies include gene mutations, exposure of the fetus to alcohol or other toxic elements, and deficiencies of critical nutrients such as folic acid. Studies have reported increased risks of specific central nervous system anomalies among obese women and women with diabetes (3). Women who undergo surgery with general anesthesia early in pregnancy may be at increased risk of delivering an infant with a central nervous system anomaly (4).

- Central nervous system anomalies were almost seven times more common among low and very low birth weight infants than infants of normal birth weight during 1996-2002.

- Alaska Native mothers were at substantially increased risk of delivering an infant with a central nervous system anomaly during 1996-2002 compared to mothers of other races. The prevalence of central nervous system anomalies was over twice as high in Alaska Natives as in whites.

- Teenage mothers were almost twice as likely, and women who began prenatal care in the third trimester almost three times as likely, to deliver an infant with a central nervous system anomaly during 1996-2002.

- Reported maternal tobacco and alcohol use associated strongly with central nervous system anomalies. The prevalence of central nervous system anomalies among infants of mothers with these behaviors was two and four times higher respectively when compared to infants whose birth certificates did not indicate maternal tobacco or alcohol use.

Prevalence of Central Nervous System Defects by Selected Birth Characteristics Alaska, 1996-2002

	n	Prevalence per 10,000 Live Births	Prevalence Ratio	95% CI
Child Sex				
Female	154	**45.3**	ref	-
Male	186	**52.0**	1.1	(0.9 - 1.4)
Birth Weight				
Low and Very Low	99	**248.4**	6.7	(5.3 - 8.4)
Normal	243	**36.9**	ref	-
Maternal Race				
White	166	**36.7**	ref	-
Alaska Native	144	**84.8**	2.3	(1.8 - 2.9)
Black	15	**49.3**	1.3	(0.8 - 2.3)
Asian or Pacific Islander	16	**41.1**	1.1	(0.7 - 1.9)
Maternal Ethnicity				
Hispanic	23	**51.1**	1.1	(0.7 - 1.6)
Non-Hispanic	299	**48.5**	ref	-
Maternal Age				
15-19 years	56	**72.7**	1.7	(1.2 - 2.3)
20-29 years	170	**44.8**	1.0	(0.8 - 1.3)
30-39 years	96	**43.4**	ref	-
40-45 years	12	**63.5**	1.5	(0.8 - 2.7)
Prenatal Care				
First Trimester	230	**41.8**	ref	-
Second Trimester	60	**57.9**	1.4	(1.0 - 1.8)
Later or None	32	**120.0**	2.9	(2.0 - 4.1)
Maternal Alcohol Use				
Reported	43	**187.4**	4.2	(3.1 - 5.8)
Not Reported	295	**44.3**	ref	-
Maternal Tobacco Use				
Reported	105	**81.5**	2.0	(1.6 - 2.5)
Not Reported	235	**41.7**	ref	-

Specific Anomalies

Five central nervous system anomalies are classified as major anomalies: hydrocephalus, microcephalus, and the neural tube defects, including anencephalus, encephalocele and spina bifida. A single infant may have one or more of these anomalies. Central nervous system anomalies also occur in association with other birth defects – for example, about 10% of children with neural tube defects also have chromosomal anomalies. Hydrocephalus is often accompanied by spina bifida; however, children reported with both hydrocephaly and spina bifida are counted only under spina bifida.

- About 9% of children reported to the ABDR who were born in 1996-2002 had central nervous system anomalies, an average of 49 infants per year.

- Microcephalus, the most common central nervous system anomaly reported to the ABDR during 1996-2002, occurred twice as often as hydrocephalus and almost four times as often as a neural tube defect. Sixty two percent of children reported with central nervous system anomalies had microcephalus.

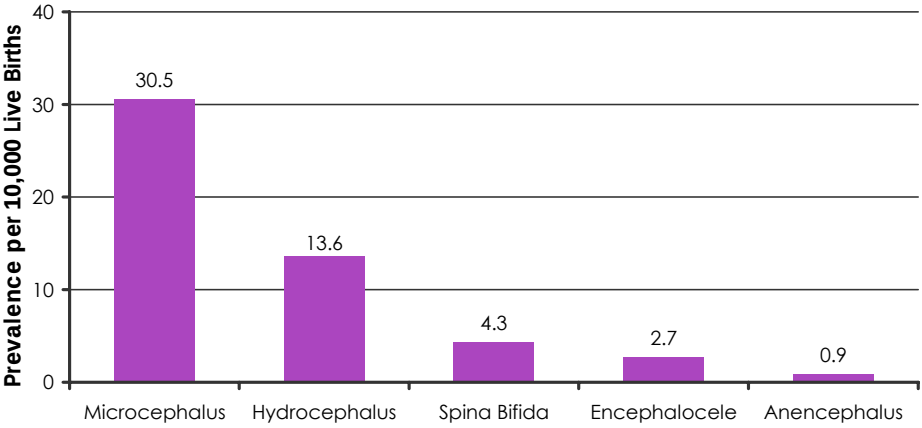
- Hydrocephalus without spina bifida was present in 28% of children reported to the ABDR with central nervous system anomalies during 1996-2002.

- NTDs made up 16% of Alaskan children reported with central nervous system anomalies during 1996-2002.

- Thirty six percent of infants born with central nervous system anomalies during 1996-2002 also had birth defects in other anatomical groups.

Central Nervous System Anomalies

Prevalence of Specific Central Nervous System Anomalies Alaska, 1996-2002



Trends and Distribution

Congenital microcephalus refers to an infant born with a head circumference that is less than the 10th percentile for gestational age and may be caused by improper or incomplete development of the brain. While there is no physical treatment for microcephalus, pediatric neurologists and early childhood intervention teams can treat the neurological and social disabilities that often accompany the anomaly. These disabilities can include mental retardation, delayed motor functions, facial distortions, dwarfism, hyperactivity and seizures (4).

- The prevalence of microcephalus among Alaskan children born in 1996-2002 was 30.5 per 10,000 live births. About 5% of children reported with at least one major anomaly had microcephalus.

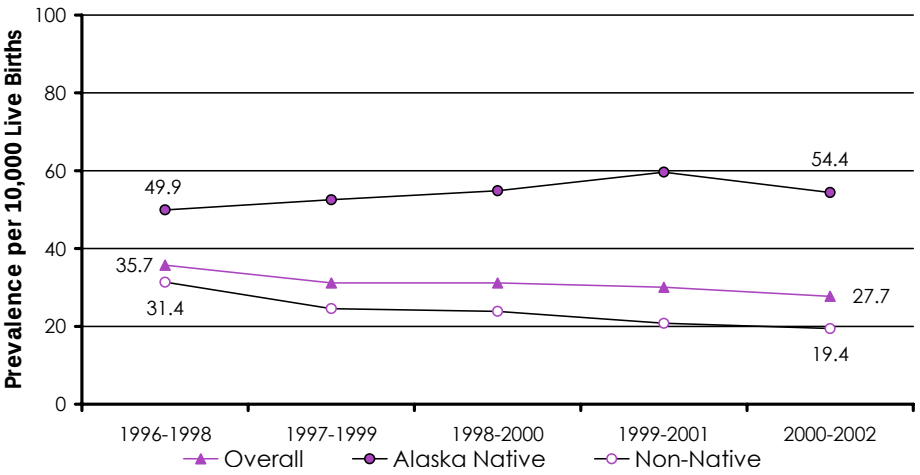
- There was a significant overall decrease in the annual prevalence of microcephalus during 1996-2002, explained primarily by the declining prevalence among non-Natives. Microcephalus prevalence among non-Natives decreased by 38% between 1996-1998 and 2000-2002.

- There was no significant change in the annual birth prevalence of microcephalus among Alaska Natives during 1996-2002. Alaska Natives had consistently higher annual microcephalus rates than non-Natives with the disparity increasing to almost three times that of non-Natives by 2000-2002.

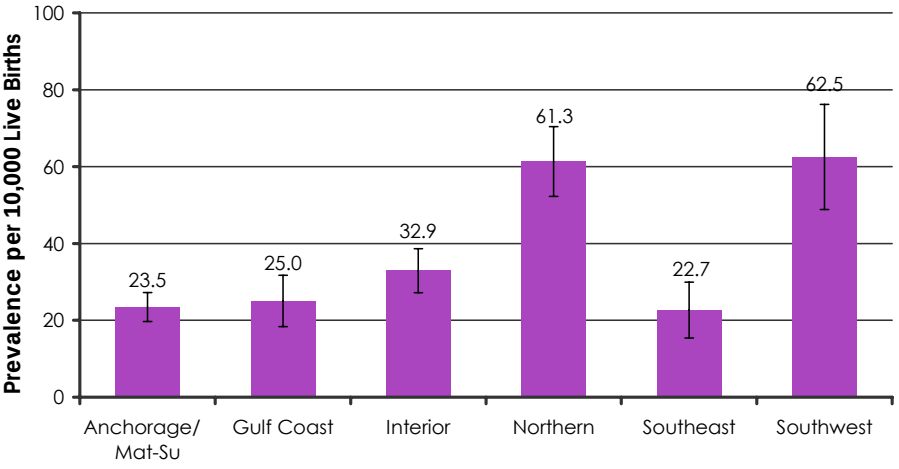
- Regional prevalence estimates for microcephalus reflected the excess risk reported for Alaska Natives. The regions with the highest proportional distributions of Alaska Natives (Southwest and Northern regions) had significantly higher microcephalus rates than other regions.

- Despite higher rates, cases from the Southwest and Northern regions made up less than a third of the total number of microcephalus cases reported statewide.

Prevalence of Microcephalus by Birth Year and Race Group, Alaska, 1996-2002



Prevalence of Microcephalus by Region Alaska, 1996-2002



Epidemiological Characteristics

Microcephalus due to abnormal brain development is most often caused by genetic abnormalities such as Down syndrome and trisomy 13. Prenatal environment, however, can also play a role. Pathological etiologies of decreased brain growth and subsequent microcephalus include infections, severe maternal malnutrition and environmental exposures. Microcephalus has been reported in children who were prenatally exposed to drugs, alcohol and environmental toxins. Prenatal genetic testing can help determine whether a fetus has one of numerous genetic disorders known to accompany microcephalus.

- Microcephalus was reported in 1.5% of low birth weight infants during 1996-2002.
- The prevalence of microcephalus among infants of Alaska Native mothers was over twice that of infants of white mothers in Alaska during 1996-2002.
- Teenage mothers were almost twice as likely, and women who began prenatal care in the third trimester about 3.5 times as likely, to deliver an infant with microcephalus during 1996-2002.
- Microcephalus was strongly associated with prenatal alcohol use. One in every one-hundred infants born to women who drank during pregnancy was reported to the ABDR with microcephalus during 1996-2002.
- During 1996-2002, women who smoked cigarettes prenatally were twice as likely to deliver an infant with microcephalus when compared to those who did not.

Prevalence of Microcephalus by Selected Birth Characteristics Alaska, 1996-2002

	n	Prevalence per 10,000 Live Births	Prevalence Ratio	95% CI
Child Sex				
Female	93	**27.4**	ref	-
Male	120	**33.5**	1.2	(0.9 - 1.6)
Birth Weight				
Low and Very Low	61	**153.0**	6.6	(4.9 - 8.9)
Normal	152	**23.1**	ref	-
Maternal Race				
White	106	**23.4**	ref	-
Alaska Native	87	**51.2**	2.2	(1.6 - 2.9)
Black	7	**23.0**	1.0	(0.5 - 2.1)
Asian or Pacific Islander	13	**33.4**	1.4	(0.8 - 2.5)
Maternal Age				
15-19 years	38	**49.3**	1.9	(1.3 - 2.9)
20-29 years	108	**28.5**	1.1	(0.8 - 1.6)
30-39 years	56	**25.3**	ref	-
40-45 years	5	**26.5**	1.0	(0.4 - 2.6)
Prenatal Care				
First Trimester	137	**24.9**	ref	-
Second Trimester	38	**36.7**	1.5	(1.0 - 2.1)
Later or None	24	**90.0**	3.6	(2.3 - 5.6)
Maternal Alcohol Use				
Reported	29	**126.4**	4.6	(3.1 - 6.8)
Not Reported	183	**27.5**	ref	-
Maternal Tobacco Use				
Reported	69	**53.5**	2.1	(1.6 - 2.8)
Not Reported	142	**25.2**	ref	-
OVERALL	213	**30.5**	-	-

Trends and Distribution

Hydrocephalus is an abnormal build up of cerebrospinal fluid (CSF) in the ventricles of the brain. The buildup of CSF can be caused by a blockage in its circulation or absorption, or when too much CSF is produced. The excess fluid causes pressure on the brain, which can result in brain damage. Hydrocephalus is treated by addressing the underlying cause or palliated through surgical insertion of a shunt to improve the flow of CSF. Advances in diagnostic imaging technology may contribute to an increasing trend in the number of diagnoses for hydrocephalus.

Hydrocephalus is often accompanied by spina bifida. Only reports of hydrocephalus without spina bifida are included here.

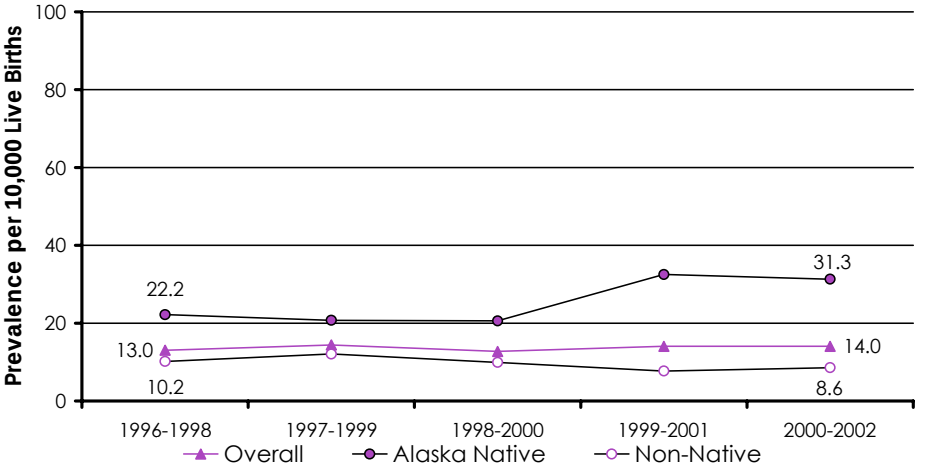
- The prevalence of hydrocephalus among Alaskan children born in 1996-2002 was 13.6 per 10,000 live births. There was no significant change in the overall prevalence during the time period.

- Alaska Natives had higher rates of hydrocephalus than non-Natives during 1996-2002. An apparently increasing annual trend in the prevalence of hydrocephalus among Alaska Natives was not statistically significant; nevertheless, the Native to non-Native disparity increased from 2.2 in 1996-1998 to 3.6 in 2000-2002.

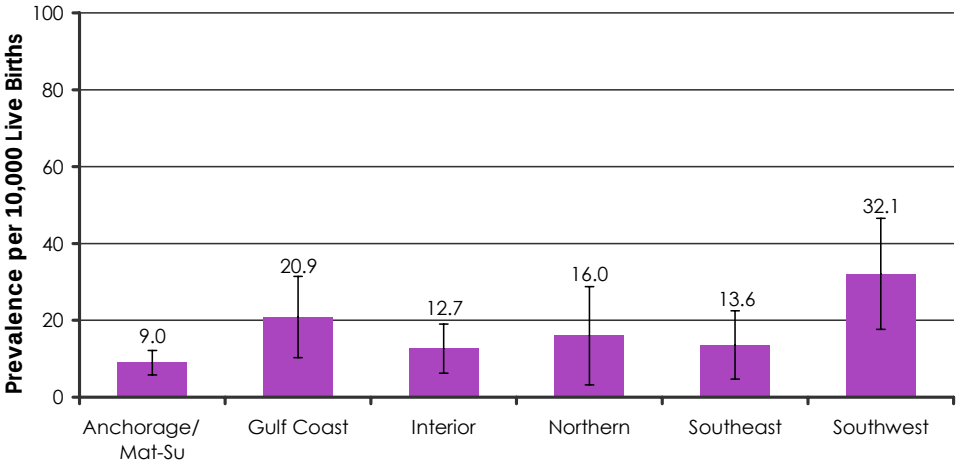
- The Anchorage/Mat-Su region had the lowest prevalence of hydrocephalus among children born in 1996-2002, and the Southwest region the highest.

- Statistically significant regional differences were only apparent when comparing Anchorage/Mat-Su with the Southwest and Gulf Coast regions. The Anchorage/Mat-Su region had statistically lower rates of hydrocephalus than either the Southwest or Gulf Coast regions.

Prevalence of Hydrocephalus by Birth Year and Race Group, Alaska, 1996-2002



Prevalence of Hydrocephalus by Region Alaska, 1996-2002



Epidemiological Characteristics

Congenital hydrocephalus can be caused by genetic anomalies or by prenatal environmental influences such as infections, injuries, toxins and some medications. Examples of environmental exposures that have been associated with hydrocephalus include prenatal exposure to aminopterin, a cancer treatment medication that inhibits folic acid metabolism (5), and prenatal exposure to general anesthesia (6). Studies show no obvious trends in sex, race or socioeconomic status.

- Hydrocephalus occurred eight times more frequently among low and very low birth weight infants than normal birth weight infants during 1996-2002.

- The prevalence of hydrocephalus among infants of Alaska Native mothers was three times that of infants of white mothers in Alaska during 1996-2002.

- Teenage mothers were twice as likely as women aged 30-39 years to deliver an infant with hydrocephalus during 1996-2002.

- Hydrocephalus was strongly associated with prenatal alcohol use. Women who drank alcohol during pregnancy were over three times as likely to deliver an infant with hydrocephalus as women who did not drink alcohol.

- During 1996-2002, women who smoked cigarettes prenatally were twice as likely to deliver an infant with hydrocephalus as those who did not.

Prevalence of Hydrocephalus by Selected Birth Characteristics Alaska, 1996-2002

	n	Prevalence per 10,000 Live Births	Prevalence Ratio	95% CI
Child Sex				
Female	41	**12.1**	ref	-
Male	53	**14.8**	1.2	(0.8 - 1.8)
Birth Weight				
Low and Very Low	32	**80.3**	8.4	(5.5 - 12.8)
Normal	63	**9.6**	ref	-
Maternal Race				
White	41	**9.1**	ref	-
Alaska Native	45	**26.5**	2.9	(1.9 - 4.5)
Black	n < 5	**-**	-	-
Asian or Pacific Islander	n < 5	**-**	-	-
Maternal Age				
15-19 years	16	**20.8**	2.0	(1.1 - 3.8)
20-29 years	49	**12.9**	1.2	(0.8 - 2.0)
30-39 years	23	**10.4**	ref	-
40-45 years	n < 5	**-**	-	-
Prenatal Care				
First Trimester	66	**12.0**	ref	-
Second Trimester	16	**15.5**	1.3	(0.7 - 2.2)
Later or None	6	**22.5**	1.9	(0.8 - 4.3)
Maternal Alcohol Use				
Reported	10	**43.6**	3.5	(1.8 - 6.6)
Not Reported	84	**12.6**	ref	-
Maternal Tobacco Use				
Reported	31	**24.1**	2.1	(1.4 - 3.3)
Not Reported	64	**11.3**	ref	-
OVERALL	95	**13.6**	-	-

Trends and Distribution

Early in conception, the neural groove of the developing fetus folds into a structure called the neural tube. The neural tube eventually develops into the spinal cord and brain. By day 28 of conception, the neural tube should be closed and fused. If it does not close, the result is a neural tube defect. Neural tube defects are serous birth defects and include spina bifida, anencephaly and encephalocele. In many cases, neural tube defects can be diagnosed during pregnancy with ultrasound and less often by other tests such as amniocentesis. Most prevalence estimates do not include prenatal diagnoses that were not carried to term; for this reason, the prevalence of neural tube defects in many populations is underestimated.

- A total of 54 Alaskan infants (7.0 per 10,000 live births) were reported to the ABDR as having been born with a neural tube defect in 1996-2002.

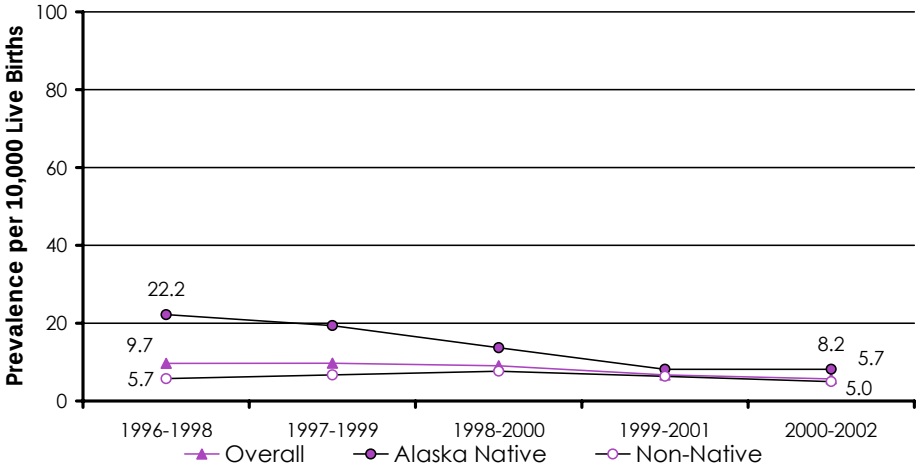
- During 1996-1998, an annual average of 10 neural tube defect-affected infants were born in Alaska. In 2000-2002, the annual number of neural tube defect cases declined to six.

- Alaska Natives reported higher rates of neural tube defects than non-Natives at the beginning of the study period. The average number of cases among Alaska Natives decreased by 60% in 2000-2002 compared to 1996-1998, minimizing the racial disparity. (Preliminary data for 2003-2004 indicate continued declines in neural tube defect prevalence for Alaska Natives.)

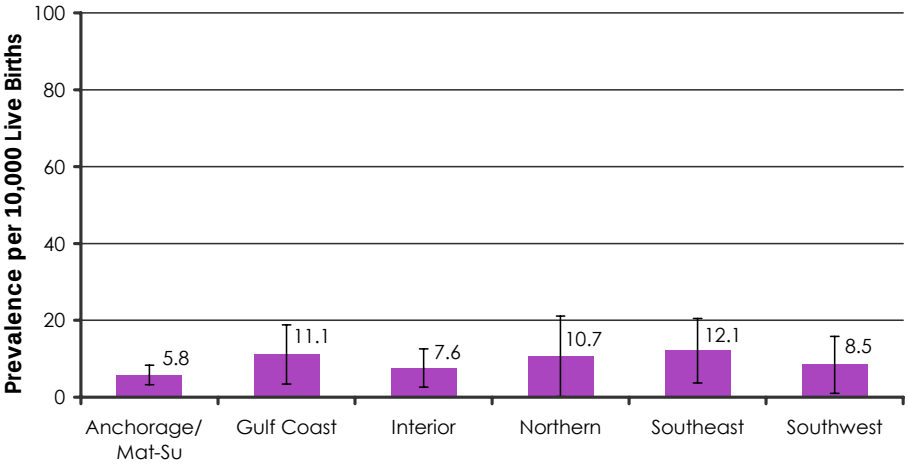
- For birth years 1996-2002, there were no significant regional differences in neural tube defect prevalence.

Note: The small number of neural tube defect cases during birth years 1996-2002 limits the statistical power of the trend analysis

Prevalence of Neural Tube Defects by Birth Year and Race Group, Alaska, 1996-2002



Prevalence of Neural Tube Defects by Region Alaska, 1996-2002



Epidemiological Characteristics

Genetic and environmental factors working in combination are thought to cause most neural tube defects. Women who have relatives with a neural tube defect or who have had a previous neural tube defect affected pregnancy are at increased risk. The prevalence of neural tube defects varies by race and ethnicity, with Hispanic populations having higher rates, and blacks and Asians lower rates. A large proportion of neural tube defects are related to folate metabolism and maternal folic acid deficiency -- 50-70% of neural tube defects can be prevented if women take 400 µg of folic acid before conception and during the first four weeks of pregnancy. A reduction in the prevalence of neural tube defects in the U.S. was demonstrated following mandated fortification of grain products in 1998 (7).

- Low and very low birth weight infants were four times as likely to be born with a neural tube defect as normal birth weight infants during 1996-2002.

- Despite declining neural tube defect rates for Alaska Natives during 1996-2002, the overall risk of a neural tube defect birth for Alaska Natives was 2.6 times greater than that of whites during the period.

- In Alaska, Hispanic ethnicity was not associated with neural tube defect risk. Less than five Hispanic infants were reported to the ABDR with a neural tube defect over the seven-year study period (1996-2002).

- Neural tube defect rates were not associated with maternal age or trimester of prenatal care in Alaska during 1996-2002.

- Neural tube defect affected births were four times as common among women who reportedly drank alcohol during pregnancy than those who did not. Prenatal tobacco use was not associated with increased risk of a neural tube defect.

Prevalence of Neural Tube Defects by Selected Birth Characteristics Alaska, 1996-2002

	n	Prevalence per 10,000 Live Births	Prevalence Ratio	95% CI
Child Sex				
Female	25	**7.4**	ref	-
Male	28	**7.8**	1.1	(0.6 - 1.8)
Birth Weight				
Low and Very Low	11	**27.6**	4.2	(2.2 - 8.2)
Normal	43	**6.5**	ref	-
Maternal Race				
White	25	**5.5**	ref	-
Alaska Native	24	**14.1**	2.6	(1.5 - 4.5)
Black	n < 5	**-**	-	-
Asian or Pacific Islander	n < 5	**-**	-	-
Maternal Ethnicity				
Hispanic	n < 5	**-**	-	-
Non-Hispanic	46	**7.5**	ref	-
Maternal Age				
15-19 years	6	**7.8**	0.9	(0.4 - 2.3)
20-29 years	24	**6.3**	0.7	(0.4 - 1.3)
30-39 years	19	**8.6**	ref	-
40-45 years	n < 5	**-**	-	-
Prenatal Care				
First Trimester	40	**7.3**	ref	-
Second Trimester	9	**8.7**	1.2	(0.6 - 2.5)
Later or None	n < 5	**-**	-	-
Maternal Alcohol Use				
Reported	6	**26.1**	3.8	(1.6 - 8.9)
Not Reported	46	**6.9**	ref	-
Maternal Tobacco Use				
Reported	9	**7.0**	0.9	(0.4 - 1.8)
Not Reported	45	**8.0**	ref	-

Chapter References

Chapter 8: Central Nervous System Anomalies

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Musculoskeletal Anomalies

Trends and Distribution

Musculoskeletal anomalies include diverse congenital anomalies of the limbs, abdominal wall and diaphragm. Major skeletal anomalies occur when one or more parts of a limb are missing or abbreviated (reduction deformities of the arms and legs) or when the hip joint capsule is so relaxed that it dislocates at birth (congenital hip dislocation). Abdominal wall anomalies are formed early in gestation when the wall fails to close properly, causing part of the gut to protrude outside the abdomen (gastroschisis or omphalocele). A diaphragmatic hernia occurs when there is an incomplete separation of the thorax (containing the heart and lungs) from the abdomen (containing the gastrointestinal organs). Most musculoskeletal defects are repaired surgically.

- Musculoskeletal anomalies affected an average of 47 Alaskan infants annually during 1996-2002.

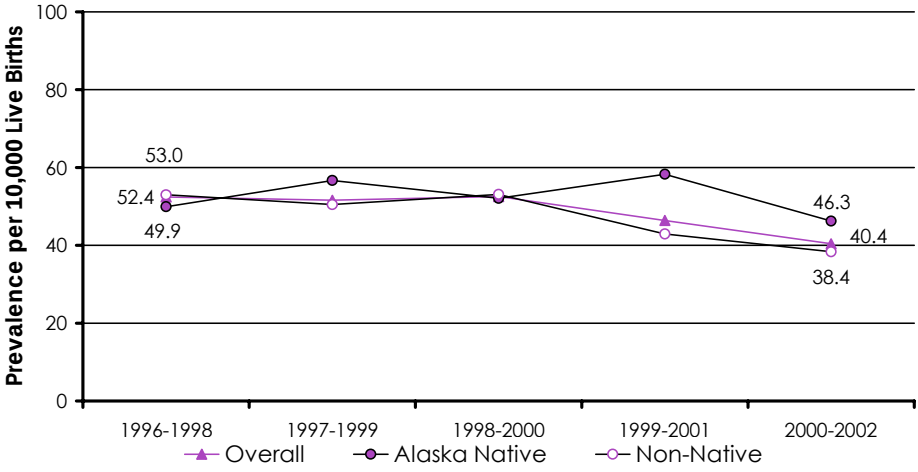
- There was a significant declining trend in the annual prevalence of musculoskeletal anomalies in Alaska during 1996-2002. Overall prevalence declined 40% between 1996-1998 and 2000-2002.

- The decline in prevalence of musculoskeletal anomalies was statistically significant for non-Natives born in 1996-2002, but not for Alaska Natives.

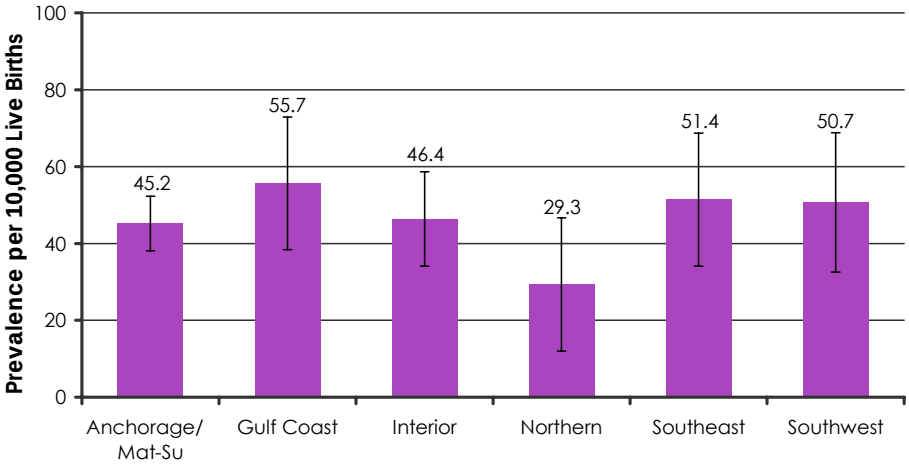
- There was no significant Native/non-Native disparity in the prevalence of musculoskeletal anomalies during 1996-2002.

- There were no significant regional differences in the distribution of musculoskeletal birth defects in Alaska during 1996-2001.

Prevalence of Musculoskeletal Anomalies by Birth Year and Race Group, Alaska, 1996-2002



Prevalence of Musculoskeletal Anomalies by Region Alaska, 1996-2002



Epidemiological Characteristics

Genetic and environmental causes have been identified for musculoskeletal anomalies. There is a broad diversity of etiologies among the congenital anomalies included in this group. For example, some limb anomalies are caused when the embryo becomes entangled in pieces of the membrane that encircles the amniotic sac. Mechanical force can be a factor in causing the hip to dislocate. A well known cause of limb anomalies was the drug thalidomide, given to women in the 1950's and 1960's to control morning sickness. Chromosomal abnormalities can cause musculoskeletal anomalies as well. About one-third of the children born with omphalocele have other birth defects, including chromosomal abnormalities.

- Female sex and low birth weight were associated with the prevalence of musculoskeletal anomalies in Alaska during 1996-2002. Low and very low birth weight infants were 2.4 times as likely to have a musculoskeletal birth defect.

- As a group, musculoskeletal anomalies were not associated with maternal race or Hispanic ethnicity.

- Younger women were more likely to deliver an infant with a musculoskeletal anomaly than older women. The prevalence was almost twice as high for teenage mothers compared to women aged 30-39.

- Prenatal alcohol and tobacco use were not associated with musculoskeletal birth defects in Alaskan infants during 1996-2002.

Prevalence of Musculoskeletal Anomalies by Selected Birth Characteristics Alaska, 1996-2002

	n	Prevalence per 10,000 Live Births	Prevalence Ratio	95% CI
Child Sex				
Female	181	**53.3**	ref	-
Male	142	**39.7**	0.7	(0.6 - 0.9)
Birth Weight				
Low and Very Low	42	**105.4**	2.4	(1.8 - 3.4)
Normal	284	**43.1**	ref	-
Maternal Race				
White	213	**47.1**	ref	-
Alaska Native	87	**51.2**	1.1	(0.8 - 1.4)
Black	9	**29.6**	0.6	(0.3 - 1.2)
Asian or Pacific Islander	14	**35.9**	0.8	(0.4 - 1.3)
Maternal Ethnicity				
Hispanic	27	**60.0**	1.3	(0.9 - 1.9)
Non-Hispanic	286	**46.4**	ref	-
Maternal Age				
15-19 years	51	**66.2**	1.8	(1.3 - 2.6)
20-29 years	189	**49.8**	1.4	(1.0 - 1.8)
30-39 years	81	**36.6**	ref	-
40-45 years	n < 5	**-**	-	-
Prenatal Care				
First Trimester	256	**46.5**	ref	-
Second Trimester	53	**51.2**	1.1	(0.8 - 1.5)
Later or None	8	**30.0**	0.6	(0.3 - 1.3)
Maternal Alcohol Use				
Reported	7	**30.5**	0.6	(0.3 - 1.4)
Not Reported	316	**47.4**	ref	-
Maternal Tobacco Use				
Reported	59	**45.8**	1.0	(0.7 - 1.3)
Not Reported	265	**47.0**	ref	-

Specific Anomalies

Six musculoskeletal anomalies are classified as major congenital anomalies. These birth defects affect anatomical structures that develop around the end of the fourth week of pregnancy, after closure of the neural tube. Reduction deformities of the lower or upper limbs, abdominal wall anomalies (gastroschisis and omphalocele) and diaphragmatic hernia occur four to five times less frequently than congenital hip dislocation, the most common musculoskeletal anomaly.

- Eight percent of children reported to the ABDR who were born during 1996-2002 had birth defects of the musculoskeletal system.

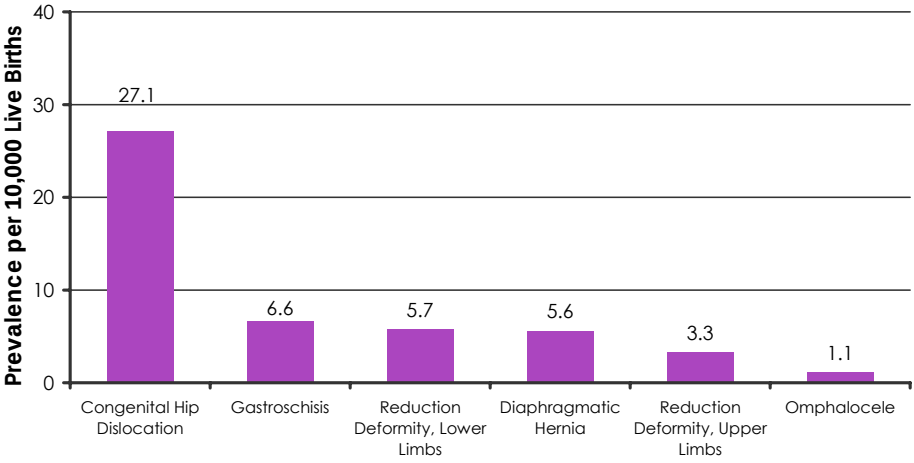
- Congenital dislocation of the hip was the most frequently reported musculoskeletal anomaly and is included among Alaska's 15 most common birth defects. Congenital hip dislocation made up about 55% of musculoskeletal anomalies reported to the ABDR during 1996-2002.

- Abdominal wall defects (omphalocele and gastroschisis) comprised 16% of musculoskeletal anomalies reported for children born in 1996-2002 and affected an average of eight infants each year during that time.

- Nine percent (an average of nine per year) of infants born with musculoskeletal anomalies during 1996-2002 had reduction deformities of the upper or lower limbs.

- Twenty four percent of infants born with musculoskeletal anomalies during 1996-2002 had birth defects in other anatomical groups.

Prevalence of Specific Musculoskeletal Anomalies Alaska, 1996-2002



Trends and Distribution

Congenital hip dislocation refers to a condition in which one or both of the hips are dislocated at birth. This often occurs due to improper formation of components of the joint or due to loose ligaments and muscles involved in hip movement. The actual dislocation usually occurs postpartum. If congenital hip dislocation is diagnosed in infancy, treatment with bracing is usually successful. As age at diagnosis increases, the condition becomes harder to treat with braces and may necessitate surgery and body casting.

- An average of 27 Alaskan infants were born annually during 1996-2002 with congenital hip dislocation. About 5% of children reported with at least one major anomaly had congenital dislocation of the hip.

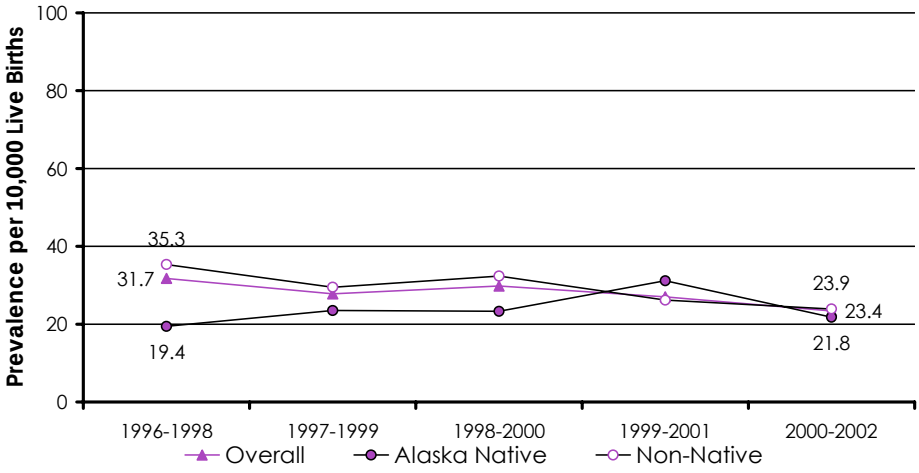
- The annual prevalence of congenital hip dislocation declined significantly during 1996-2002. The overall decline was explained by the significant 26% decline in prevalence among non-Natives between 1996-1998 and 2000-2002.

- There was no significant change in the annual birth prevalence of congenital hip dislocation among Alaska Natives during 1996-2002. In general, Alaska Natives born in 1996-2002 had slightly lower rates of congenital hip dislocation than non-Natives.

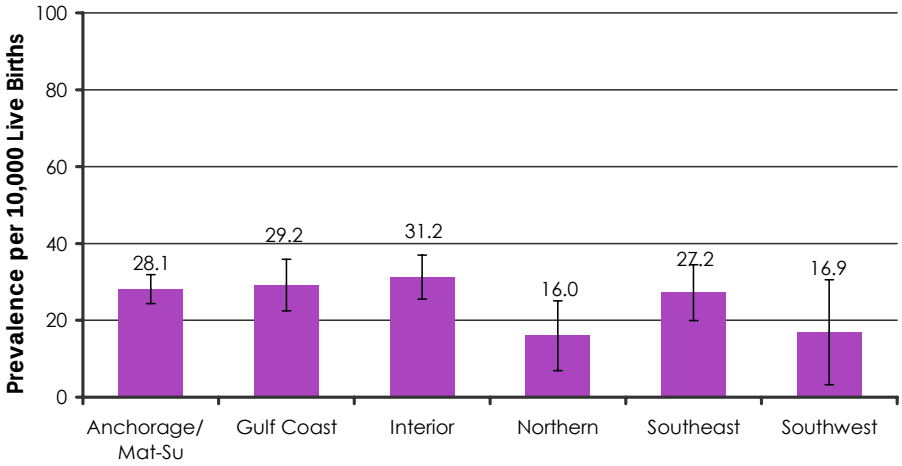
- There were no significant regional differences in congenital hip dislocation prevalence for Alaskan children born during 1996-2002.

Congenital Hip Dislocation

Prevalence of Congenital Hip Dislocation by Birth Year and Race Group, Alaska, 1996-2002



Prevalence of Congenital Hip Dislocation by Region Alaska, 1996-2002



Epidemiological Characteristics

Females are diagnosed with congenital hip dislocation more often than males (1). Furthermore, a higher incidence is reported in first-born children and in children born breech (2). The cause of congenital hip dislocation is multifactorial, and no environmental risk factors have been successfully explored. Congenital hip dislocation diagnoses in children born breech and in cultures that practice swaddling indicate that many cases are brought on by environment at the time of and after birth, not by developmental irregularity. Mild cases of congenital hip dislocation can go undetected well into adulthood when symptoms begin to present. Routine ultrasound screening, however, can detect cases earlier and lead to more effective treatment (3).

- Among Alaskan infants born during 1996-2002, females were significantly more likely than males to have congenital hip dislocation, a finding consistent with the established epidemiology of this condition.

- No significant associations were found between congenital hip dislocation and maternal race, ethnicity, age, trimester of prenatal care or prenatal tobacco or alcohol use for Alaskan infants born during 1996-2002.

Prevalence of Congenital Dislocation of Hip by Selected Birth Characteristics Alaska, 1996-2002

	n	Prevalence per 10,000 Live Births	Prevalence Ratio	95% CI
Child Sex				
Female	128	**37.7**	ref	-
Male	61	**17.0**	0.5	(0.3 - 0.6)
Birth Weight				
Low and Very Low	14	**35.1**	1.3	(0.8 - 2.3)
Normal	175	**26.6**	ref	-
Maternal Race				
White	134	**29.6**	ref	-
Alaska Native	39	**23.0**	0.8	(0.5 - 1.1)
Black	5	**16.4**	0.6	(0.2 - 1.4)
Asian or Pacific Islander	9	**23.1**	0.8	(0.4 - 1.5)
Maternal Age				
15-19 years	27	**35.0**	1.6	(1.0 - 2.6)
20-29 years	110	**29.0**	1.3	(1.0 - 1.9)
30-39 years	48	**21.7**	ref	-
40-45 years	n < 5	**-**	-	-
Prenatal Care				
First Trimester	146	**26.5**	ref	-
Second Trimester	33	**31.9**	1.2	(0.8 - 1.8)
Later or None	n < 5	**-**	-	-
Maternal Alcohol Use				
Reported	n < 5	**-**	-	-
Not Reported	186	**27.9**	ref	-
Maternal Tobacco Use				
Reported	29	**22.5**	0.8	(0.5 - 1.2)
Not Reported	160	**28.4**	ref	-
OVERALL	189	**27.1**	-	-

Chapter References

Chapter 9: Musculoskeletal Anomalies

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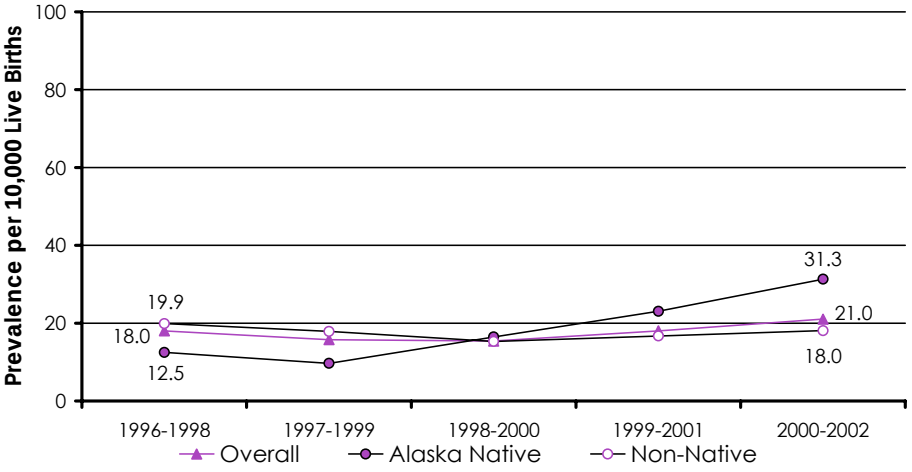
Chromosomal Anomalies

Trends and Distribution

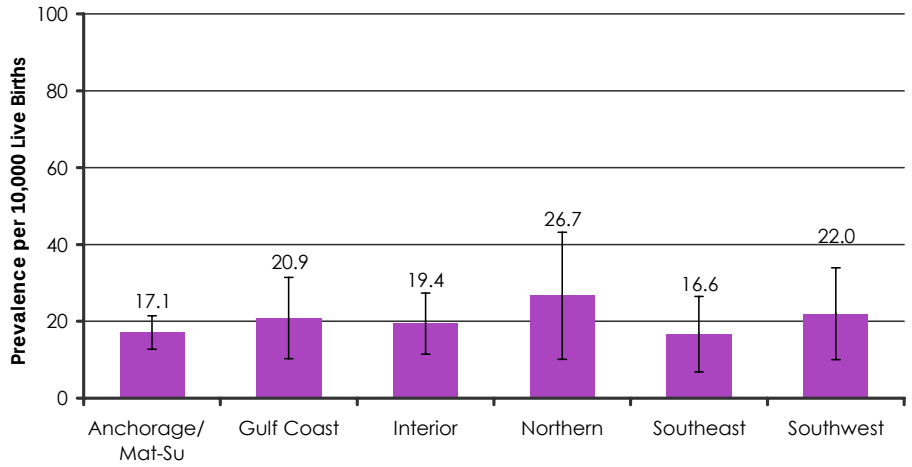
Birth defects categorized as chromosomal anomalies refer to those that are caused by abnormal numbers of chromosomes, or deletions or damage to the structure of the chromosome. Chromosomal anomalies usually occur when the sperm and egg are developing, before the egg is fertilized. A trisomy is a common type of chromosomal anomaly. Humans have 22 matched pairs of autosomal chromosomes plus the pair that determines sex. Trisomy occurs when an infant has an extra copy of a chromosome, forming a triad instead of a pair. A characteristic syndrome results, depending on which chromosome pair was affected.

- Chromosomal anomalies affected an average of 19 Alaskan infants annually during 1996-2002.
- While there was no change in the overall prevalence of chromosomal anomalies during 1996-2002, there was a significant increasing trend for Alaska Natives during the study period.
- The prevalence of chromosomal anomalies among Alaska Natives increased 150% from 12.5 in 1996-1998 to 31.3 in 2000-2002.
- There were no significant regional differences in the distribution of chromosomal anomalies in Alaska during 1996-2002.

Prevalence of Chromosomal Anomalies by Birth Year and Race Group, Alaska, 1996-2002



Prevalence of Chromosomal Anomalies by Region Alaska, 1996-2002



Epidemiological Characteristics

Most causes of chromosomal anomalies, including trisomy, are unknown. No studies have successfully identified behavioral or environmental risk factors, but the risk clearly increases with maternal age and some studies suggest an association with older paternal age (>50 years) (1,2). Women who have had more than four previous pregnancies may also be more likely to deliver an infant with a chromosomal anomaly (1).

- In Alaska, low birth weight infants were almost seven times more likely to have a major chromosomal anomaly than normal birth weight infants.

- Increasing maternal age was strongly associated with a higher prevalence of chromosomal anomalies in Alaskan infants born in 1996-2002. Women aged 40-45 years had 5.5 times the risk of delivering an infant with a chromosomal anomaly.

- ABDR surveillance data for infants born in 1996-2002 did not indicate a linear relationship between maternal age and chromosomal anomalies.

- Chromosomal anomalies were not associated with maternal race, Hispanic ethnicity, trimester of prenatal care, or prenatal tobacco or alcohol exposure.

Prevalence of Chromosomal Anomalies by Selected Birth Characteristics Alaska, 1996-2002

	n	Prevalence per 10,000 Live Births	Prevalence Ratio	95% CI
Child Sex				
Female	62	**18.3**	ref	-
Male	69	**19.3**	1.1	(0.7 - 1.5)
Birth Weight				
Low and Very Low	38	**95.3**	6.7	(4.6 - 9.8)
Normal	93	**14.1**	ref	-
Maternal Race				
White	88	**19.5**	ref	-
Alaska Native	34	**20.0**	1.0	(0.7 - 1.5)
Black	n < 5	**-**	-	(-)
Asian or Pacific Islander	7	**18.0**	0.9	(0.4 - 2.0)
Maternal Ethnicity				
Hispanic	12	**26.7**	1.5	(0.8 - 2.8)
Non-Hispanic	108	**17.5**	ref	-
Maternal Age				
15-19 years	12	**15.6**	0.7	(0.4 - 1.4)
20-29 years	47	**12.4**	0.6	(0.4 - 0.9)
30-39 years	47	**21.2**	ref	-
40-45 years	22	**116.4**	5.5	(3.3 - 9.1)
Prenatal Care				
First Trimester	94	**17.1**	ref	-
Second Trimester	25	**24.1**	1.4	(0.9 - 2.2)
Later or None	5	**18.8**	1.1	(0.4 - 2.7)
Maternal Alcohol Use				
Reported	5	**21.8**	1.2	(0.5 - 2.9)
Not Reported	121	**18.2**	ref	-
Maternal Tobacco Use				
Reported	20	**15.5**	0.8	(0.5 - 1.3)
Not Reported	109	**19.3**	ref	-

Specific Anomalies

Three trisomies are classified as major anomalies. These are trisomy 13 (Patau syndrome), trisomy 18 (Edward syndrome), and trisomy 21 (Down syndrome). Each of these syndromes has a characteristic set of findings, which include a variety of other structural birth defects. Cardiovascular anomalies, for example, are present in about 80% of individuals with trisomy 13 and 90% of those with trisomy 18 (3). Infants with trisomy 13 or 18 usually die within a year of birth. Although structural birth defects are also common in infants with Down syndrome, they have a better survival outlook than infants with trisomy 13 or 18.

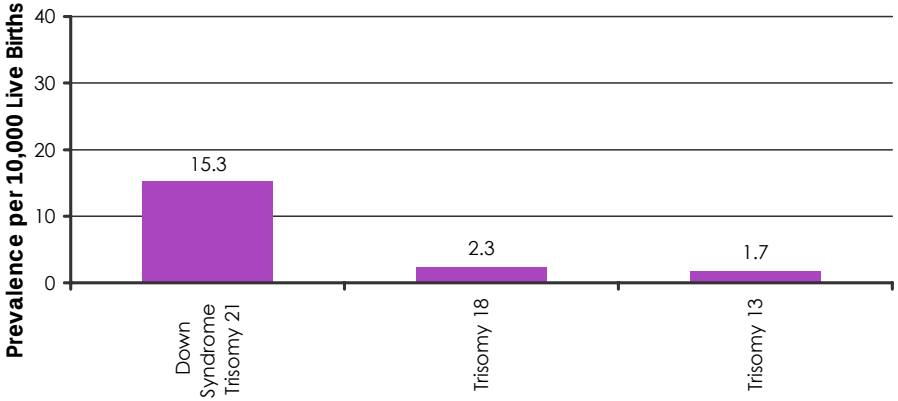
- Three percent of Alaskan infants who were born during 1996-2002 were reported with major chromosomal anomalies.

- Down syndrome was the most frequently reported major chromosomal anomaly, occurring seven and nine times more often than trisomy 18 and trisomy 13 respectively, and comprising 82% of infants born with reported chromosomal defects during the study period.

- Down syndrome ranked 11th among Alaska's 15 most common major anomalies during birth years 1996-2002.

- Sixty nine percent of infants born with chromosomal anomalies during 1996-2002 were reported with major anomalies in other anatomical groups.

Prevalence of Specific Chromosomal Anomalies Alaska, 1996-2002



Trends and Distribution

Individuals with Down syndrome, or trisomy 21, have extra chromosome 21 material. Ninety five percent of patients have non-disjunction Down syndrome, where the duplication of chromosome 21 is present in every cell. The remaining 5% of individuals have only a partial duplication of the chromosome (translocation Down syndrome) or have trisomy 21 in only some of their cells (mosaic Down syndrome). Incurable, Down syndrome is accompanied by characteristic facial anomalies and growth retardation. People with Down syndrome have some degree of mental retardation, are likely to be susceptible to infections, and may have vision or hearing problems. Cardiovascular anomalies are present in about 50% of Down syndrome cases and 10% have intestinal malformations that require surgery.

- An average of 15 Alaskan infants were born each year with Down syndrome during 1996-2002.

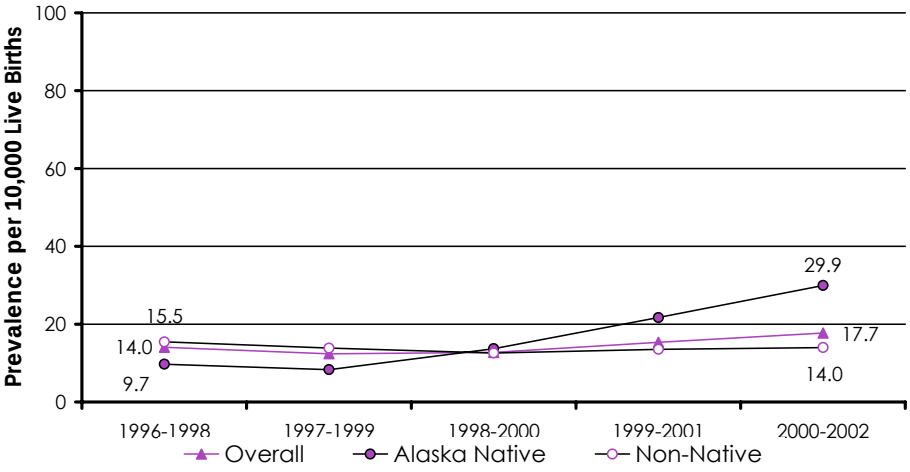
- There was no change in the annual overall prevalence of Down syndrome during 1996-2002; however there was a statistically significant tripling in the prevalence among Alaska Natives during the period.

- There was no Alaska Native/non-Native disparity in Down syndrome prevalence for Alaskan infants born during 1996-2002.

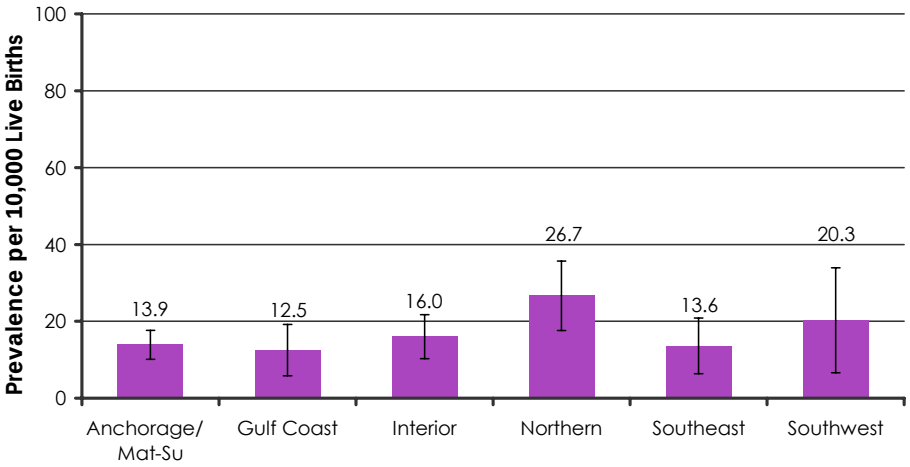
- There were no significant regional differences in the distribution of Down syndrome during birth years 1996-2002. Although the rate of Down syndrome was twice as high in the Northern region as the Gulf Coast region, these prevalence estimates are based on too few cases for meaningful statistical analysis.

Down Syndrome (Trisomy 21)

Prevalence of Down Syndrome by Birth Year and Race Group, Alaska, 1996-2002



Prevalence of Down Syndrome by Region Alaska, 1996-2002



Epidemiological Characteristics

Down syndrome occurs in one out of every 660 births (4). The risk of Down syndrome increases with maternal age. No behavioral or environmental risk factors have been identified. Reported rates of Down syndrome often do not reflect the actual incidence of the disorder because surveillance programs focus primarily on live births and do not include prenatal diagnoses that may result in pregnancy termination (5).

- Low birth weight infants were 4.5 times more likely to be born with Down syndrome than normal birth weight infants during 1996-2002. There was no association between Down syndrome and sex of the infant.

- While there were no differences in the prevalence of Down syndrome by maternal race, Alaskan mothers of Hispanic ethnicity had a marginally significant increased risk for delivering an infant with Down syndrome during 1996-2002.

- Women aged 40-45 years had five times the risk of having an infant with Down syndrome during 1996-2002 than women aged 30-39. While Down syndrome rates were lower in younger maternal age groups, there was no significant linear relationship between Down syndrome and maternal age.

- Down syndrome among Alaskan infants born in 1996-2002 was not associated with the trimester when prenatal care began, prenatal tobacco use or prenatal alcohol use.

Prevalence of Down Syndrome by Selected Birth Characteristics Alaska, 1996-2002

	n	Prevalence per 10,000 Live Births	Prevalence Ratio	95% CI
Child Sex				
Female	48	**14.1**	ref	-
Male	59	**16.5**	1.2	(0.8 - 1.7)
Birth Weight				
Low and Very Low	23	**57.7**	4.5	(2.9 - 7.2)
Normal	84	**12.8**	ref	-
Maternal Race				
White	67	**14.8**	ref	-
Alaska Native	31	**18.2**	1.2	(0.8 - 1.9)
Black	n < 5	**-**	-	-
Asian or Pacific Islander	7	**18.0**	1.2	(0.6 - 2.6)
Maternal Age				
15-19 years	8	**10.4**	0.6	(0.3 - 1.3)
20-29 years	41	**10.8**	0.6	(0.4 - 1.0)
30-39 years	39	**17.6**	ref	-
40-45 years	17	**89.9**	5.1	(2.9 - 9.0)
Prenatal Care				
First Trimester	77	**14.0**	ref	-
Second Trimester	20	**19.3**	1.4	(0.8 - 2.3)
Later or None	5	**18.8**	1.3	(0.5 - 3.3)
Maternal Alcohol Use				
Reported	5	**21.8**	1.5	(0.6 - 3.6)
Not Reported	98	**14.7**	ref	-
Maternal Tobacco Use				
Reported	19	**14.7**	1.0	(0.6 - 1.6)
Not Reported	87	**15.4**	ref	-
OVERALL	107	**15.3**	-	-

Chapter References

Chapter 10: Chromosomal Anomalies

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Eye and Ear Anomalies

Trends and Distribution

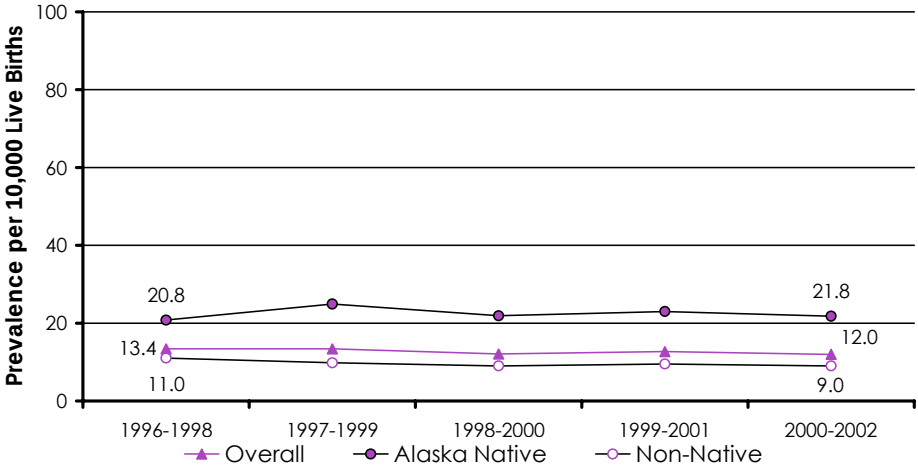
Eyes and ears start developing in the 4th gestational week. Major birth defects of the eye and ear generally are identifiable during the 2nd month of gestation and include: aniridia, absent or incomplete iris; anophthalmia, the absence of the eye (technically, the absence of the globe and ocular tissue from the orbit); microphthalmia, an abnormally small eye; congenital cataract, an opaque lens of the eye; anotia, the absence of an ear; and microtia, an abnormally small ear.

- During 1996-2002 there was no significant change in the prevalence of eye and ear anomalies in Alaska.

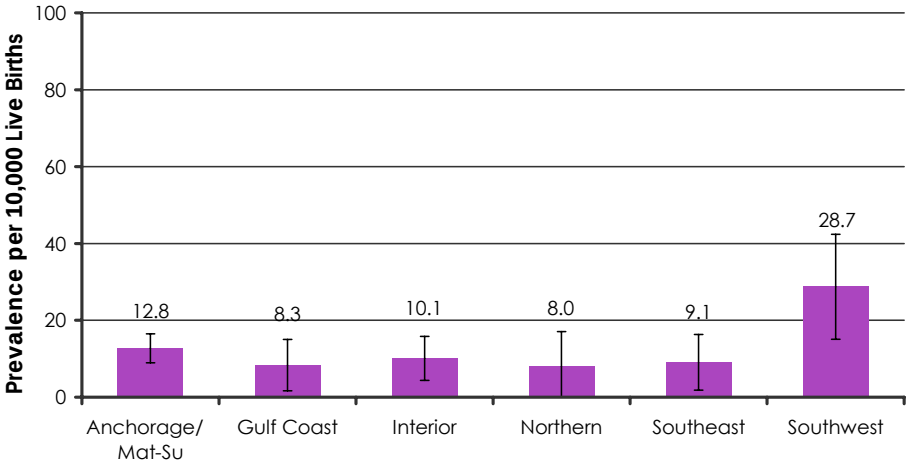
- Alaska Natives had higher rates of eye and ear anomalies than non-Natives during 1996-2002. A two-fold racial disparity was consistent throughout the study period.

- While regional differences in the prevalence of eye and ear anomalies during 1996-2002 did not reach statistical significance, prevalence in the Southwest region was two to three times higher than in other regions of the state.

Prevalence of Eye and Ear Anomalies by Birth Year and Race Group, Alaska, 1996-2002



Prevalence of Eye and Ear Anomalies by Region Alaska, 1996-2002



Epidemiological Characteristics

Most ear and eye anomalies have a genetic etiology. Aniridia, for example may occur as an autosomal disorder, an identifiable deletion of the short arm of chromosome 11, or as a sporadic case (1). External ear anomalies are one of the defining characteristics of many of the identified birth defect syndromes.

Fetal exposure to the rubella (German measles) virus can lead to congenital rubella syndrome. Along with cardiovascular anomalies and developmental delay, congenital rubella syndrome is associated with eye and ear anomalies such as cataracts and hearing impairment.

- Eye and ear anomalies were more common among male than female infants during 1996-2002, but the increase in risk was not significant.

- Low birth weight infants were twice as likely to have an eye and ear anomaly as normal birth weight Alaskan infants during 1996-2002.

- Eye and ear anomalies were over twice as common in infants born of Alaska Native mothers than in infants born of other races during 1996-2002. Infants born of white, black and Asian or Pacific Islander mothers had equivalent rates of eye ear anomalies during the study period.

- While the differences in risk between maternal age groups were not statistically significant, there was a general trend toward less risk with increasing age.

- There was no association between the prevalence of eye and ear anomalies and trimester of prenatal care, prenatal maternal tobacco use or alcohol use for infants born during 1996-2002.

Prevalence of Eye and Ear Anomalies by Selected Birth Characteristics Alaska, 1996-2002

	n	Prevalence per 10,000 Live Births	Prevalence Ratio	95% CI
Child Sex				
Female	35	**10.3**	ref	-
Male	53	**14.8**	1.4	(0.9 - 2.2)
Birth Weight				
Low and Very Low	10	**25.1**	2.1	(1.1 - 4.1)
Normal	78	**11.8**	ref	-
Maternal Race				
White	43	**9.5**	ref	-
Alaska Native	38	**22.4**	2.4	(1.5 - 3.6)
Black	n < 5	**-**	-	-
Asian or Pacific Islander	n < 5	**-**	-	-
Maternal Ethnicity				
Hispanic	8	**17.8**	1.4	(0.7 - 2.9)
Non-Hispanic	77	**12.5**	ref	-
Maternal Age				
15-19 years	13	**16.9**	1.5	(0.8 - 2.9)
20-29 years	50	**13.2**	1.2	(0.7 - 1.9)
30-39 years	25	**11.3**	ref	-
40-45 years	n < 5	**-**	-	-
Prenatal Care				
First Trimester	66	**12.0**	ref	-
Second Trimester	12	**11.6**	1.0	(0.5 - 1.8)
Later or None	n < 5	**-**	-	-
Maternal Alcohol Use				
Reported	n < 5	**-**	-	-
Not Reported	84	**12.6**	ref	-
Maternal Tobacco Use				
Reported	15	**11.6**	0.9	(0.5 - 1.6)
Not Reported	71	**12.6**	ref	-

Specific Anomalies

Eye and ear anomalies are some of the least common major congenital anomalies. There are four specific eye or ear anomalies that are classified as major anomalies: congenital cataract, anotia/microtia, anopthalmia/micropthalmia and aniridia.

Congenital hearing loss is not necessarily related to defects in the physical structures of the ear, and Alaskan providers did not report newborn hearing loss to the ABDR during 1996-2002.

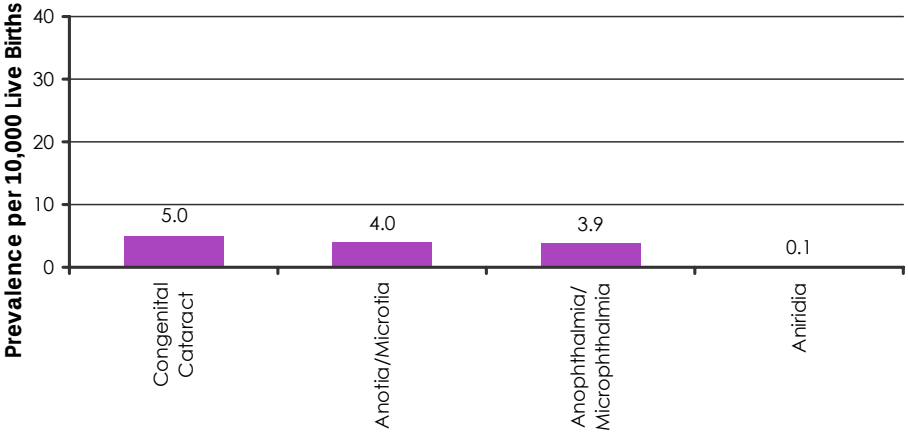
- Two percent of infants with major anomalies reported to the ABDR during 1996-2002 had eye or ear anomalies, an average of 13 Alaskan infants each year.

- The most common major anomaly of the eye or ear in Alaska was congenital cataract, which occurred in about 5 infants each year during 1996-2002.

- There was an average of 4 births affected annually by anotia/microtia or anopthalmia/micropthalmia during 1996-2002. Aniridia was rare.

- Thirty four percent of infants born with major eye and ear anomalies during 1996-2002 were reported with major anomalies in other anatomical groups.

Prevalence of Specific Eye and Ear Anomalies
Alaska, 1996-2002



Chapter References

Chapter 11: Eye and Ear Anomalies

1. eMedicine. Aniridia. Available at: http://www.emedicine.com/OPH/topic43.htm. Accessed November 23, 2005.

Detailed Tables

Detailed Trend Data

Major Anomalies	1996	1997	1998	1999	2000	2001	2002	1996-2002
Overall	542.3	583.2	592.4	595.2	563.1	512.7	502.8	556.0
Alaska Native	1030.8	1002.1	1215.8	1190.8	1034.6	1029.6	846.2	1050.0
Non Native	385.7	450.9	394.5	404.8	412.4	341.2	390.1	397.1
Cardivascular Anomalies	**1996**	**1997**	**1998**	**1999**	**2000**	**2001**	**2002**	**1996-2002**
Overall	212.0	199.7	178.3	226.8	200.4	213.9	251.4	211.8
Alaska Native	349.1	263.0	319.5	395.6	305.5	436.7	421.0	355.8
Non Native	166.6	180.4	133.7	175.2	167.7	140.8	196.4	165.8
Alcohol Related Anomalies	**1996**	**1997**	**1998**	**1999**	**2000**	**2001**	**2002**	**1996-2002**
Overall	136.6	140.5	188.4	161.6	147.3	105.9	80.4	137.2
Alaska Native	465.5	496.9	676.3	576.8	513.2	364.6	254.3	478.2
Non Native	32.8	27.9	32.1	25.8	28.4	20.3	25.7	27.6
Alimentary Tract Anomalies	**1996**	**1997**	**1998**	**1999**	**2000**	**2001**	**2002**	**1996-2002**
Overall	80.6	104.4	83.6	96.4	89.2	82.0	78.4	87.8
Alaska Native	116.4	112.7	116.2	152.5	146.6	120.2	125.1	127.1
Non Native	69.5	102.1	73.6	78.8	71.7	65.0	63.7	74.9
Genitourinary Anomalies	**1996**	**1997**	**1998**	**1999**	**2000**	**2001**	**2002**	**1996-2002**
Overall	57.7	87.3	66.5	67.2	63.1	69.0	72.4	69.0
Alaska Native	70.7	87.7	87.1	37.1	65.2	72.1	41.7	65.9
Non Native	53.8	86.2	60.2	77.4	62.2	69.1	81.3	70.0
Central Nervous System Anomalies			**1996-1998**	**1997-1999**	**1998-2000**	**1999-2001**	**2000-2002**	**1996-2002**
Overall			56.1	53.3	50.6	47.4	43.8	49.0
Alaska Native			88.8	87.1	82.3	90.8	84.4	84.7
Non Native			45.9	42.5	40.5	33.4	31.1	37.7

Musculoskeletal Anomalies			1996-1998	1997-1999	1998-2000	1999-2001	2000-2002	1996-2002
Overall			52.4	51.6	52.6	46.4	40.4	46.7
Alaska Native			49.9	56.7	52.1	58.3	46.3	51.1
Non Native			53.0	50.5	53.1	42.9	38.4	45.2
Chromosomal Anomalies			**1996-1998**	**1997-1999**	**1998-2000**	**1999-2001**	**2000-2002**	**1996-2002**
Overall			18.0	15.7	15.4	18.0	21.0	18.8
Alaska Native			12.5	9.7	16.5	23.0	31.3	19.9
Non Native			19.9	17.9	15.3	16.7	18.0	18.6
Eye & Ear Anomalies			**1996-1998**	**1997-1999**	**1998-2000**	**1999-2001**	**2000-2002**	**1996-2002**
Overall			13.4	13.4	12.0	12.7	12.0	12.6
Alaska Native			20.8	24.9	21.9	23.0	21.8	22.4
Non Native			11.0	9.8	9.0	9.5	9.0	9.6
Atrial septal defect	**1996**	**1997**	**1998**	**1999**	**2000**	**2001**	**2002**	**1996-2002**
Overall	67.7	71.3	70.5	117.4	100.2	82.0	133.7	91.8
Alaska Native	103.9	75.2	128.6	189.5	146.6	176.3	220.9	148.7
Non Native	56.4	70.3	52.2	95.1	86.5	51.5	102.9	73.6
Ventricular septal defect	**1996**	**1997**	**1998**	**1999**	**2000**	**2001**	**2002**	**1996-2002**
Overall	94.5	76.3	71.5	81.3	74.1	95.0	99.5	84.6
Alaska Native	178.7	133.6	153.5	148.3	154.8	208.3	195.9	167.6
Non Native	66.9	58.4	45.5	61.1	48.7	58.2	70.4	58.5
Patent ductus arteriosus			**1996-1998**	**1997-1999**	**1998-2000**	**1999-2001**	**2000-2002**	**1996-2002**
Overall			49.1	52.9	54.6	61.8	71.8	59.9
Alaska Native			45.8	70.5	79.5	100.3	102.0	78.0
Non Native			49.9	47.8	47.2	50.1	62.3	54.1
Hypospadias & Epispadias			**1996-1998**	**1997-1999**	**1998-2000**	**1999-2001**	**2000-2002**	**1996-2002**
Overall			35.1	41.9	38.8	36.7	35.8	33.6
Alaska Native			41.6	44.2	38.4	35.2	28.6	35.3
Non Native			32.7	41.1	39.1	37.5	37.9	36.7

Detailed Trend Data (continued)

Microcephaly	1996-1998	1997-1999	1998-2000	1999-2001	2000-2002	1996-2002
Overall	35.7	31.2	31.1	30.1	27.7	30.5
Alaska Native	49.9	52.5	54.9	59.6	54.4	51.1
Non Native	31.4	24.6	23.8	20.8	19.4	24.1
Obstructive genitourinary defect	**1996-1998**	**1997-1999**	**1998-2000**	**1999-2001**	**2000-2002**	**1996-2002**
Overall	32.4	29.5	24.4	27.0	30.7	30.3
Alaska Native	31.9	22.1	23.3	21.7	27.2	25.9
Non Native	32.7	31.7	24.7	28.9	31.6	31.6
Pyloric stenosis	**1996-1998**	**1997-1999**	**1998-2000**	**1999-2001**	**2000-2002**	**1996-2002**
Overall	30.7	32.5	29.5	30.1	27.7	30.1
Alaska Native	45.8	44.2	49.4	54.2	50.3	49.4
Non Native	26.1	29.0	23.4	22.1	19.9	23.8
Oral clefts	**1996-1998**	**1997-1999**	**1998-2000**	**1999-2001**	**2000-2002**	**1996-2002**
Overall	29.4	34.5	33.5	29.7	28.1	29.4
Alaska Native	43.0	58.1	63.1	51.5	43.5	47.1
Non Native	25.2	27.3	24.3	22.1	22.6	23.6
Pulmonary valve stenosis	**1996-1998**	**1997-1999**	**1998-2000**	**1999-2001**	**2000-2002**	**1996-2002**
Overall	35.4	31.5	24.8	23.7	20.7	27.9
Alaska Native	80.4	70.5	60.3	70.5	58.5	71.2
Non Native	21.2	19.2	13.0	8.1	8.1	13.9
Congenital hip dislocation	**1996-1998**	**1997-1999**	**1998-2000**	**1999-2001**	**2000-2002**	**1996-2002**
Overall	31.7	27.8	29.8	27.0	23.4	27.1
Alaska Native	19.4	23.5	23.3	31.2	21.8	22.9
Non Native	35.3	29.5	32.4	26.2	23.9	28.3

Down syndrome	1996-1998	1997-1999	1998-2000	1999-2001	2000-2002	1996-2002
Overall	14.0	12.4	12.7	15.4	17.7	15.3
Alaska Native	9.7	8.3	13.7	21.7	29.9	18.2
Non Native	15.5	13.9	12.6	13.5	14.0	14.6
Hydrocephaly	**1996-1998**	**1997-1999**	**1998-2000**	**1999-2001**	**2000-2002**	**1996-2002**
Overall	13.0	14.4	12.7	14.0	14.0	13.6
Alaska Native	22.2	20.7	20.6	32.5	31.3	26.4
Non Native	10.2	12.1	9.9	7.7	8.6	9.4
Hirschsprung's disease	**1996-1998**	**1997-1999**	**1998-2000**	**1999-2001**	**2000-2002**	**1996-2002**
Overall	15.4	12.7	11.7	13.4	12.0	13.3
Alaska Native	11.1	11.1	15.1	17.6	12.2	12.3
Non Native	16.8	13.0	10.3	11.3	11.7	13.4
Neural tube defects	**1996-1998**	**1997-1999**	**1998-2000**	**1999-2001**	**2000-2002**	**1996-2002**
Overall	9.7	9.7	9.0	6.7	5.7	7.7
Alaska Native	22.2	19.4	13.7	8.1	8.2	14.2
Non Native	5.7	6.7	7.6	6.3	5.0	5.7

Detailed Regional Data

	Anchorage / Mat-Su	Gulf Coast	Interior	Northern	Southeast	Southwest
Major Anomalies						
Prevalence per 10,000 live births	**504.4**	**509.3**	**485.4**	**1080.0**	**399.3**	**899.1**
95% CI	(480.7-528.1)	(457.1-561.4)	(445.7-525.0)	(974.8-1185.2)	(351.1-447.4)	(822.7-975.5)
Cardiovascular Anomalies						
Prevalence per 10,000 live births	**185.5**	**157.2**	**186.5**	**338.7**	**158.8**	**461.4**
95% CI	(171.2-199.9)	(128.2-186.2)	(162.0-211.1)	(279.8-397.6)	(128.4-189.2)	(406.7-516.1)
Alcohol Related Anomalies						
Prevalence per 10,000 live births	**111.6**	**141.9**	**88.6**	**528.0**	**43.9**	**234.9**
95% CI	(100.5-122.8)	(114.4-169.5)	(71.7-105.6)	(454.5-601.5)	(27.9-59.8)	(195.9-274.0)
Alimentary Tract Anomalies						
Prevalence per 10,000 live births	**90.2**	**77.9**	**80.2**	**141.3**	**59.0**	**99.7**
95% CI	(80.1-100.2)	(57.5-98.3)	(64.1-96.3)	(103.3-179.4)	(40.5-77.5)	(74.3-125.2)
Genitourinary Anomalies						
Prevalence per 10,000 live births	**80.6**	**61.2**	**56.6**	**56.0**	**57.5**	**59.2**
95% CI	(71.1-90.1)	(43.1-79.3)	(43.0-70.1)	(32.0-80.0)	(39.2-75.7)	(39.6-78.7)
Central Nervous System Anomalies						
Prevalence per 10,000 live births	**37.1**	**52.9**	**51.5**	**82.7**	**42.3**	**94.6**
95% CI	(30.7-43.5)	(36.1-69.7)	(38.6-64.4)	(53.6-111.8)	(26.7-58.0)	(69.9-119.4)
Musculoskeletal Anomalies						
Prevalence per 10,000 live births	**45.2**	**55.7**	**46.4**	**29.3**	**51.4**	**50.7**
95% CI	(38.1-52.3)	(38.4-72.9)	(34.2-58.7)	(12.0-46.7)	(34.1-68.7)	(32.6-68.8)

Chromosomal Anomalies						
Prevalence per 10,000 live births	**17.1**	**20.9**	**19.4**	**26.7**	**16.6**	**22.0**
95% CI	(12.7-21.5)	(10.3-31.4)	(11.5-27.3)	(10.1-43.2)	(6.8-26.5)	(10.0-33.9)
Eye & Ear Anomalies						
Prevalence per 10,000 live births	**12.8**	**8.3**	**10.1**	**8.0**	**9.1**	**28.7**
95% CI	(9.0-16.5)	(1.7-15.0)	(4.4-15.9)	(-1.1-17.1)	(1.8-16.3)	(15.1-42.4)
Atrial septal defect						
Prevalence per 10,000 live births	**84.4**	**82.1**	**81.0**	**173.3**	**46.9**	**167.3**
95% CI	(74.7-94.1)	(61.1-103.0)	(64.8-97.2)	(131.2-215.5)	(30.4-63.4)	(134.4-200.3)
Ventricular septal defect						
Prevalence per 10,000 live births	**67.8**	**47.3**	**71.7**	**176.0**	**75.6**	**206.2**
95% CI	(59.1-76.5)	(31.4-63.2)	(56.5-87.0)	(133.5-218.5)	(54.7-96.6)	(169.6-242.8)
Patent ductus arteriosus						
Prevalence per 10,000 live births	**56.2**	**48.7**	**59.9**	**85.3**	**51.4**	**87.9**
95% CI	(48.3-64.2)	(32.6-64.8)	(46.0-73.9)	(55.8-114.9)	(34.1-68.7)	(64.0-111.8)
Hypospadias & Epispadias						
Prevalence per 10,000 live births	**38.6**	**41.7**	**32.9**	**29.3**	**30.2**	**35.5**
95% CI	(24.8-42.3)	(35.1-48.4)	(27.2-38.7)	(20.3-38.4)	(23.0-37.5)	(21.8-49.1)
Microcephaly						
Prevalence per 10,000 live births	**23.5**	**25.0**	**32.9**	**61.3**	**22.7**	**62.5**
95% CI	(19.7-27.3)	(18.4-31.7)	(27.2-38.7)	(52.3-70.4)	(15.4-29.9)	(48.9-76.2)
Obstructive genitourinary defect						
Prevalence per 10,000 live births	**40.3**	**16.7**	**21.1**	**24.0**	**22.7**	**20.3**
95% CI	(36.5-44.1)	(10.0-23.4)	(15.4-26.8)	(14.9-33.1)	(15.4-29.9)	(6.6-33.9)
Pyloric stenosis						
Prevalence per 10,000 live births	**28.7**	**22.3**	**27.0**	**66.7**	**22.7**	**38.9**
95% CI	(24.9-32.5)	(15.6-28.9)	(21.3-32.7)	(57.6-75.7)	(15.4-29.9)	(25.2-52.5)

Detailed Regional Data (continued)

	Anchorage / Mat-Su	Gulf Coast	Interior	Northern	Southeast	Southwest
Oral clefts						
Prevalence per 10,000 live births	**29.6**	**26.4**	**26.9**	**53.3**	**18.1**	**33.8**
95% CI	(25.8-33.3)	(19.8-33.1)	(21.2-32.7)	(44.3-62.4)	(10.9-25.4)	(20.1-47.5)
Pulmonary valve stenosis						
Prevalence per 10,000 live births	**19.4**	**9.7**	**11.0**	**45.3**	**16.6**	**135.2**
95% CI	(14.8-24.1)	(2.5-17.0)	(5.0-16.9)	(23.8-66.9)	(6.8-26.5)	(105.6-164.8)
Congenital hip dislocation						
Prevalence per 10,000 live births	**28.1**	**29.2**	**31.2**	**16.0**	**27.2**	**16.9**
95% CI	(24.4-1.9)	(22.5-35.9)	(25.5-37.0)	(6.9-25.1)	(20.0-34.5)	(3.2-30.6)
Down syndrome						
Prevalence per 10,000 live births	**13.9**	**12.5**	**16.0**	**26.7**	**13.6**	**20.3**
95% CI	(10.1-17.7)	(5.8-19.2)	(10.3-21.8)	(17.6-35.7)	(6.4-20.9)	(6.6-33.9)
Hydrocephaly						
Prevalence per 10,000 live births	**9.0**	**20.9**	**12.7**	**16.0**	**13.6**	**32.1**
95% CI	(5.8-12.2)	(10.3-31.4)	(6.3-19.1)	(3.2-28.8)	(4.7-22.5)	(17.7-46.5)
Hirschsprung's disease						
Prevalence per 10,000 live births	**17.4**	**8.3**	**9.3**	**10.7**	**10.6**	**8.5**
95% CI	(13.0-21.8)	(1.7-15.0)	(3.8-14.8)	(0.2-21.1)	(2.7-18.4)	(1.0-15.9)
Neural tube defects						
Prevalence per 10,000 live births	**5.8**	**11.1**	**7.6**	**10.7**	**12.1**	**8.5**
95% CI	(3.3-8.3)	(3.4-18.8)	(2.6-12.6)	(0.2-21.1)	(3.7-20.5)	(1.0-15.9)

Detailed Prevalence Data

Detailed Prevalence Data for Major Anomalies with Prevalence Less Than 12 per 10,000 Live Births, Alaska, 1996 - 2002

Anomaly	Prevalence per 10,000 Live Births			Anomaly	Prevalence per 10,000 Live Births		
	Native	Non Native	Overall		Native	Non Native	Overall
Anencephalus	2.4	0.4	0.9	Endocardial Cusion Defect	7.7	5.8	6.2
Aniridia	0.0	0.2	0.1	Esophageal Atresia/ Tracheoesophageal Fistula	1.8	3.5	3.0
Anophthalmia/ Microphthalmia	8.8	2.3	3.9	Gastroschisis	9.4	5.8	6.6
Anotia/Microtia	8.8	2.5	4.0	Hypoplastic Left Heart Syndrome	2.4	3.8	3.4
Aortic Valve Stenosis	2.9	4.0	3.7	Omphalocele	0.6	1.3	1.1
Bilary Atresia	3.5	1.7	2.1	Rectal and Large Intestinal Atresia/ Stenosis	14.7	8.8	10.2
Bladder Extrophy	0.6	1.2	1.0	Reduction Deformity, Lower Limbs	7.1	5.2	5.7
Choanal Atresia	3.5	2.7	2.9	Reduction Deformity, Upper Limbs	5.3	2.7	3.3
Coarctation of Aorta	7.7	6.3	6.6	Renal Agenesis/ Hypoplasia	6.5	5.0	5.3
Common Truncus	3.5	1.0	1.6	Spina Bifida	7.1	3.5	4.3
Congenital Cataract	5.3	5.0	5.0	Tetralogy of Fallot	16.5	4.8	7.6
Diaphragmatic Hernia	7.7	5.0	5.6	Transposition of Great Vessels	4.7	6.3	6.2
Ebstein's Anomaly	2.9	0.2	0.9	Tricuspid Valve Atresia/Stenosis	4.1	2.5	2.9
Encephalocele	5.3	1.9	2.7	Trisomy 13	1.2	1.9	1.7
				Trisomy 18	1.8	2.5	2.3

Appendices

Reportable Birth Defects

Major Congenital Anomalies

ICD-9 Code	Reportable Condition
740.0 - 740.1	Anencephalus
741.0, 741.9 without 740.0 - 740.10	Spina bifida without anencephalus
742.0	Encephalocele
742.1	Microcephalus
742.3 without 741.0, 741.9	Hydrocephalus without Spina bifida
743.0, 743.1	Anophthalmia/Microphthalmia
743.30 – 743.34	Congenital cataract
743.45	Aniridia
744.01, 744.23	Anotia/Microtia
745.0	Common truncus
745.10 - 745.12, 745.19	Transposition of great arteries
745.2	Tetralogy of Fallot
745.4	Ventricular septal defect
745.5	Atrial septal defect
745.60, 745.61, 745.69	Endocardial cushion defect
746.01, 746.02	Pulmonary valve atresia and stenosis
746.1	Tricuspid valve atresia and stenosis
746.2	Ebstein's anomaly
746.3	Aortic valve stenosis
746.7	Hypoplastic left heart syndrome
747.0	Patent ductus arteriosus
747.1	Coarctation of aorta

Major Congenital Anomalies (Continued)

ICD-9 Code	Reportable Condition
748.0	Choanal atresia
749.0	Cleft palate without cleft lip
749.1, 749.2	Cleft lip with and without cleft palate
750.3	Esophageal atresia/tracheoesophageal fistula
750.5	Pyloric stenosis
751.2	Rectal and large intestinal atresia/stenosis
751.3	Hirschsprung's disease (congenital megacolon)
751.61	Biliary atresia
752.61, 752.62	Hypospadias and Epispadias
753.0	Renal agenesis/hypoplasia
753.5	Bladder exstrophy
753.2, 753.6	Obstructive genitourinary defect
754.30, 754.31, 754.35	Congenital hip dislocation
755.20 – 755.29	Reduction deformity, upper limbs
755.30 – 755.39	Reduction deformity, lower limbs
756.6	Diaphragmatic hernia
756.79	Gastroschisis
756.79	Omphalocele
758.0	Down syndrome
758.1	Trisomy 13
758.2	Trisomy 18
760.71	Fetus or newborn affected by maternal alcohol use
No code	Amniotic bands

Reportable Birth Defects

Reportable Congenital Conditions of Alaskan Interest

ICD-9 Code	Reportable Condition
237.7	Neurofibromatosis
243.0	Congenital hypothyroidism
255.2	Adrenogenital disorders
277.0 - 277.9	Other and unspecified disorders of the metabolism
	Cystic fibrosis
	Disorders of purine and pyrimidine metabolism
	Amyloidosis
	Disorders of bilirubin excretion
	Mucocpolysaccharidosis
	Other deficiencies of circulating enzymes
	Dysmetabolic syndrome X
	Other specified disorders of metabolism
	Unspecified disorder of metabolism
279.0 - 279.9	Disorders involving the immune mechanism
	Deficiency of humoral immunity
	Deficiency of cell-mediated immunity
	Combined immunity deficiency
	Unspecified immunity deficiency
	Autoimmune disease, not elsewhere classified
	Other specified disorders of the immune mechanism
	Unspecified disorder of the immune mechanism
282.0 - 282.9	Hereditary hemolytic anemias
	Pernicious anemia
	Other vitamin B_{12} deficiency anemia
	Folate deficiency anemia
	Other specified megaloblastic anemias
	Protein deficiency anemia
	Other specified nutritional deficiency anemia
	Unspecified deficiency anemia
284.0	Constitutional aplastic anemia
331.0 - 331.9	Alzheimer's disease
	Frontotemporal dementia
	Senile degeneration of brain
	Communicating hydrocephalus
	Obstructive hydrocephalus

Reportable Congenital Conditions of Alaskan Interest (Continued)

ICD-9 Code	Reportable Condition
331.0 - 331.9	Cerebral degeneration in diseases classified elsewhere
	Other cerebral degeneration
	Cerebral degeneration, unspecified
334.0 - 334.9	Spinocerebellar disease
	Friedreich's ataxia
	Hereditary spastic paraplegia
	Primary cerebellar degeneration
	Other cerebellar ataxia
	Cerebellar ataxia in diseases classified elsewhere
	Other spinocerebellar diseases
	Spinocerebellar disease, unspecified
335.0 - 335.9	Anterior horn cell disease
	Werdnig-Hoffman disease
	Spinal muscular atrophy
	Motor neuron disease
	Other anterior horn cell diseases
	Anterior horn cell disease, unspecified
343.0 - 343.9	Infantile cerebral palsy
	Diplegic, Monoplegic, Hemiplegic
	Infantile hemiplegia
	Other specified infantile cerebral palsy
	Infantile cerebral palsy, unspecified
359.0 - 359.9	Muscular dystrophies and other myopathies
	Congenital hereditary muscular dystrophy
	Hereditary progressive muscular dystrophy
	Myotonic disorders
	Familial periodic paralysis
	Toxic myopathy
	Myopathy in endocrine diseases classified elsewhere
	Other myopathies
	Myopathy, unspecified
362.74	Pigmentary retinal dystrophy
760.0 - 760.9, except 760.71	Fetus or newborn affected by maternal conditions
	which may be unrelated to present pregnancy
	Maternal hypertensive disorders
	Maternal renal and urinary tract diseases

Reportable Birth Defects

Other Reportable Congenital Conditions (Non-Major Anomalies)	
ICD-9 Code	Reportable Condition
742 except 742.0, 742.1, 742.3	Other congenital anomalies of nervous system
743 except 743.0, 743.1, 743.0 - 743.34, 743.45	Congenital anomalies of eye
744 except 744.01, 744.23	Congenital anomalies of ear, face and neck
745 except 745.0 - 745.2, 745.4 - 745.6	Bulbus cordis anomalies and anomalies of cardiac septal closure
746 except 746.01, 746.02, 746.1 - 746.3, 746.7	Other congenital anomalies of heart
747 except 747.0, 747.10	Other congenital anomalies of circulatory system
748 except 748.0	Congenital anomalies of respiratory system
750 except 750.3, 750.5	Other congenital anomalies of upper alimentary tract
751 except 751.2, 751.3, 751.61	Other congenital anomalies of digestive system
752 except 752.61, 752.62	Congenital anomalies of genital organs
753 except 753.0, 753.2, 753.5, 753.6	Congenital anomalies of urinary system
754 except 754.30, 754.31, 754.35	Certain musculoskeletal deformities
755 except 755.2, 755.3	Other congenital anomalies of limbs
756 except 756.6, 756.79	Other congenital musculoskeletal anomalies
757	Congenital anomalies of integument
758 except 758.0 - 758.2	Chromosomal anomalies
759	Other specified congenital anomalies

Technical Notes

All statistical analyses were performed at a significance level of alpha=.05. Any mention of a significant trend or significant difference between two groups implies that it is **statistically significant** at alpha=.05.

Trend Analyses

Trend analyses were performed using ordinary least squares regression of the natural log of the rate for years within a given time period. By convention, some trends (e.g., mortality rates) are graphed as three or five-year moving averages. However, all trend analyses are performed on the single year data, not the averaged data presented in the graph. Although the graphs of trends may show what appears to be a declining trend, it should be noted that these are moving averages and the decline may not be statistically significant since the regression is performed on single year data, not the averaged data.

Percent Change

Percent change between two time periods is calculated as follows:

$$PC = \frac{(P_n - P_o)}{P_o} \times 100$$

where P_n= later time period
P_o = earlier time period

Rate Ratios

Rate ratios, the ratio of two rates, are used to compare rates for two populations – calculated as follows:

where E_1 = number of events occurring in population 1

$$RR = \frac{(E_1 / P_1) \times 10^n}{(E_2 / P_2) \times 10^n} = \frac{Rate_1}{Rate_2}$$

E_2= number of events occurring in population 2
P_1 = number of people in population 1 at risk of an event
P_2= number of people in population 2 at risk of an event
n= base for multiplier
$Rate_1$= rate for population 1
$Rate_2$ = rate for population 2

so $n = 3 \Rightarrow 10^3$ would give a rate per 1,000

Note: The multiplier, 10^n, must be the same for both rates. A rate ratio of 1.0 indicates that there is no difference in the race-specific or age-specific rates for the two populations being compared. It is customary for the group of interest to be labeled as population 1 and the reference group as population 2, so, the group of interest is always in the numerator.

Moving Averages

Moving averages are overlapping sequences of time periods that are used to smooth out the year-to-year variability that is often observed when dealing with small numbers. A general formula for calculating the first and second time periods using the moving average method is as follows:

$$MA = \frac{\sum_{P_i-(w-1)}^{P_i} events}{\sum_{P_i-(w-1)}^{P_i} pop} \times 10^n, \quad \frac{\sum_{P_{i+1}-(w-1)}^{P_{i+1}} events}{\sum_{P_{i+1}-(w-1)}^{P_{i+1}} pop} \times 10^n$$

where P_i = time period of interest
w = width of interval
n = base for multiplier
pop = population

so $w = 3$ would be a three-year moving average
$n = 3 \Rightarrow 10^3$ would give a rate per 1,000

Example: The three-year moving average for the year 1991 is comprised of data from 1989-1991, 1992 is comprised of data from 1990-1992, and so forth. Using the formula, the rate per 1,000 for this example is:

$$\frac{(events_{1989} + events_{1990} + events_{1991})}{(pop_{1989} + pop_{1990} + pop_{1991})} \times 10^3, \quad \frac{(events_{1990} + events_{1991} + events_{1992})}{(pop_{1990} + pop_{1991} + pop_{1992})} \times 10^3$$

Prevalence Comparisons

Extreme caution should be exercised when comparing birth defects prevalence estimates. Ideal comparisons would be between programs with identical surveillance methods, case ascertainment protocols, population characteristics and analytic criteria. In the following tables, we present prevalence estimates published by other states, the US and Europe. These tables demonstrate that prevalence estimates for most birth defects are generally higher in Alaska than in states. The reader should be fully aware of the methodological differences in each of the surveillance programs featured (see insert, below) and the limitations of ABDR data (see Data Limitations section) when comparing the estimates presented in the tables.

Characteristics of Birth Defects Surveillance Programs

	Case ascertainment	Average size of annual birth cohort	Program reach	Reference Population per 10,000	Birth years included
Alaska	passive, with case confirmation for FAS	10,000	statewide	live births	1996-2002
California	active	60,000	sample	live births	1998-2002
Colorado	passive, active for FAS	68,798	statewide	live births	1998-2002
Georgia	active	51,676	metropolitan Atlanta	live births	1998-2002
Texas	active	377,000	statewide	live births	1998-2002
Europe	varied	varied	35 full member European registries	live births, fetal deaths and induced abortions	1998-2002
United States	varied	varied	varied	varied	varied

Appendix C: Comparison of ABDR Prevalence Estimates with Six Representative Birth Defects Surveillance Projects

	Alaska	California§	Colorado§	Georgia§	Texas§	Europe¥	United States
Cardiovascular							
Aortic valve stenosis	3.7	1.2	3.7	2.3	2.3	1.2	5.0[1]
Atrial septal defect	91.8	17.0	54.8	25.3	39.3	16.5	0.4[2]
Coarctation of aorta	6.6	3.4	9.2	5.9	4.3	3.3	1.0[3]
Common truncus	1.6	0.5	1.0	0.7	0.9	1.0	0.8[4]
Ebstein's anomaly	0.9		1.1	0.5	0.6	0.3	0.5[5]
Endocardial cushion defect	6.2	3.5	3.8	4.5	3.7		4.4[4]
Hypoplastic left heart syndrome	3.4	2.0	3.0	2.9	2.0	2.5	2.4[4]
Patent ductus arteriosus	59.9		39.0	30.8	42.2		5.0[6]
Pulmonary valve atresia/stenosis	27.9	0.9	10.9	7.1	6.5		
Tetralogy of Fallot	7.6	3.6	4.1	4.4	3.1	3.1	3.9[4]
Transposition of great arteries	6.2	3.9	4.4	5.6	4.6	3.1	4.7[4]
Tricuspid valve atresia/stenosis	2.9	0.8	1.4	2.1	2.5	1.0	
Ventricular septal defect	84.6	14.6	40.4	41.9	42.4	27.1	
Alcohol Related							
Fetus affected by maternal alcohol use	137.2	0.5		1.5	0.3		
Fetal alcohol syndrome*	16.5		0.03[7]				0.04[7]
Alimentary							
Biliary atresia	2.1	0.8	1.3	0.7	0.7		0.7[8]
Choanal atresia	2.9	0.2	1.3	1.4	1.2		1.4[9]
Cleft lip with & without cleft palate	16.5	8.8	11.8	8.8	10.6	9.1	10.5[4]
Cleft palate without cleft lip	12.9	3.6	8.0	7.2	5.7	5.8	6.4[4]

Appendix C (continued): Comparison of ABDR Prevalence Estimates with Six Representative Birth Defects Surveillance Projects

	Alaska	California§	Colorado§	Georgia§	Texas§	Europe¥	United States
Esophageal atresia	3.0	1.4	5.2	2.1	2.1	2.8	2.4[4]
Hirschsprung's disease	13.3		2.3	2.4	1.2		2.0[10]
Pyloric stenosis	30.1		17.3	12.7	18.6		20.0-30.0[11]
Rectal & large instesinal atresia/stenosis	10.2	3.0	5.7	3.7	4.9	2.7	4.8[4]
Genitourinary							
Bladder exstrophy	1.0		0.4	0.2	0.2	0.3	0.3[12]
Hypospadias & Epispadias	36.4	2.0	48.0	33.3	27.6		30.0[13]
Obstructive genitourinary defect	30.4		30.2	24.5	20.6		
Renal agenesis/hypoplasia	5.3	0.8	5.0	4.7	5.0	1.8	1.0-2.0[14]
Central Nervous System							
Anencephalus	0.9	3.1	1.3	3.2	2.8	3.9	4.0[15]
Encephalocele	2.7	0.6	1.0	1.2	1.0	1.2	1.0[16]
Hydrocephalus without spina bifida	13.6	2.3	9.0	8.1	7.0	5.4	20.0[17]
Microcephalus	30.5		5.0	7.7	6.5	2.0	16.0[18]
Spina bifida without anencephalus	4.3	3.8	3.2	3.5	3.6	5.0	4.2[19]
Musculoskeletal							
Congenital hip dislocation	27.1		17.4	6.4	4.6		10.0[20]
Diaphragmatic hernia	5.6	2.0	4.8	2.5	2.5	3.0	2.9[4]
Gastroschisis	6.6	3.7	5.2	2.6	3.9	2.0	3.7[4]
Omphalocele	1.1	0.7	2.8	2.3	2.2	2.9	2.1[4]
Reduction deformity, lower limbs	5.7	1.5	1.9	1.6	1.9	2.0	1.9[4]
Reduction deformity, upper limbs	3.3	3.5	3.9	4.2	3.9	4.0	3.8[4]

Chromosomal							
Down syndrome	15.3	13.1	19.2	13.4	12.0	19.5	13.7[4]
Trisomy 13	1.7	0.9	2.4	1.4	1.2	1.8	1.3[4]
Trisomy 18	2.3	1.6	4.9	2.6	2.1	4.2	2.4[4]
Eye and Ear							
Aniridia	0.1		0.2	0.1	0.1		0.1[21]
Anophthalmia/microphthalmia	3.9	0.7	1.2	3.0	2.8	1.1	2.1[4]
Anotia/microtia	4.0	2.9	2.0	1.5	2.7	0.8	0.5-2.0[22]
Congenital cataract	5.0	0.8	3.1	2.3	1.5		1.2-6.0[23]

§ Data source for California, Colorado, Georgia, Texas (except when otherwise noted): National Birth Defects Prevention Network 2005 Congenital Malformations Surveillance Report. Birth defects surveillance data from selected states, 1998-2002. Birth Defects Research (Part A): Clinical and Molecular Teratology. 2005;73(10):758-853.

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* ICD-9 code 760.71or Fetal alcohol spectrum disorder (FASD) surveillance case definition may vary.

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Glossary of Congenital Anomalies

Anencephaly
: Congenital absence of the skull and brain

Aniridia
: Congenital absence of the iris of the eye

Anophthalmia
: Congenital absence of the eye globe

Anotia
: Congenital absence of the ear

Aortic valve stenosis
: Congenital heart defect characterized by aortic valve narrowing reducing the flow of blood.

Atrial septal defect
: Congenital heart defect characterized by one or more openings in the atrial septum (wall between the right and left atria)

Biliary atresia
: Congenital absence of the ducts in the biliary tract

Bladder exstrophy
: Congenital exposure of the bladder mucosa caused by incomplete closure of the anterior bladder wall and the abdominal cavity

Choanal atresia
: Congenital absence of the passageway between the nose and pharynx due to a thick bone or thin "membranous" bone

Cleft lip
: Congenital defect of the upper lip in which there is incomplete closure

Cleft palate
: Congenital defect in the closure of the palate; the structure which separates the nasal cavities and the back of the mouth. May involve the soft palate, hard palate or alveolus (gum)

Coarctation of the aorta
: Congenital heart defect characterized by narrowing of the descending aorta

Common truncus
: Congenital heart defect characterized by a single great arterial trunk, instead of a separate aorta and pulmonary artery. Commonly known as truncus arteriosis

Congenital cataract
: Congenital clouding of the lens of the eye

Congenital hip dislocation
: Congenital dislocation of one or both hips

Diaphragmatic hernia
: Congenital defect of the muscular diaphragm resulting in herniation of the abdominal contents into the chest

Down syndrome (Trisomy 21)
: Distinctive and common chromosome abnormality syndrome caused by an extra copy of chromosome 21. Can be complete (Trisomy 21), attached to another chromosome (translocation), or mixed with cells containing normal chromosomes (mosaic)

Ebstein's anomaly
: Congenital heart defect characterized by downward displacement of the tricuspid valve into the right ventricle

Encephalocele
: Congenital defect of the skull resulting in herniation of the brain

Endocardial cushion defect
: Congenital heart defect characterized by a combined atrial and ventricular septal defect, and common atrioventricular valve (instead of distinct tricuspid and mitral valves)

Gastroschisis
: Congenital opening of the abdominal wall with protrusion of the abdominal contents. Can be distinguished from omphalocele by location usually to the right of the umbilicus

Epispadias
: Congenital defect of the genitals where the opening of the urethra is located on the upper side of the penis in boys and between the clitoris and labia in girls

Esophageal atresia/ tracheoesophageal fistula
: Congenital discontinuity of the lumen of the esophagus. Usually associated with a tracheoesophageal fistula, which is an abnormal connection between the esophagus and trachea

Hirschsprung's disease
: Congenital aganglionic megacolon (enlarged colon) due to absent nerves in the wall of the colon

Hydrocephalus
: Accumulation of fluid within the spaces of the brain. Can be congenital or acquired

Glossary of Congenital Anomalies

Hypoplastic left heart syndrome
: Congenital heart defect characterized by extreme smallness of left-sided structures. Classically, aortic valve/mitral valve atresia or marked hypoplasia, ascending aorta and left ventricle Hypoplasia

Hypospadias
: Congenital defect of the penis in which the urethral opening is on the underside of the penis

Microcephaly
: Small head, with corresponding smallness of the brain

Microphthalmia
: Congenital smallness of the eye globe

Microtia
: Congenital smallness or maldevelopment of the external ear, with or without absence or narrowing of the external auditory canal

Obstructive genitourinary defect
: Congenital narrowing or absence of the urinary tract structure at any level. Severity often depends upon the level of the obstruction

Omphalocele
: Congenital opening of the abdominal wall with protrusion of the abdominal contents. Can be distinguished from gastroschisis by location within umbilical ring

Patent ductus arteriosus
: Congenital heart defect characterized by persistence of the fetal blood vessel connecting the pulmonary artery and the aorta

Pulmonary valve atresia/ stenosis
: Congenital heart defect characterized by absence (or narrowing) of the pulmonary valve or pulmonary artery itself

Pyloric stenosis
: A congenital narrowing of the opening of the stomach into the small intestine

Rectal and large intestinal atresia/ stenosis
: Congenital absence, closure or constriction of the large intestine, rectum or anus

Reduction deformity, upper (arms) / lower (legs)
: Congenital absence of a portion or entire limb

Renal agenesis/hypoplasia
: Congenital absence of the kidney

Spina bifida
: Neural tube defect with protrusion of the spinal cord and/or Meninges

Tetralogy of Fallot
: Congenital heart defect composed of ventricular septal defect, pulmonary stenosis or atresia, displacement of the aorta to the right, and hypertrophy of right ventricle

Transposition of great vessels (arteries)
: Congenital heart defect in which the aorta arises from the right ventricle, and the pulmonary artery arises from the left ventricle (opposite of normal)

Tricuspid valve atresia/stenosis
: Congenital heart defect characterized by the absence (or narrowing of) of the tricuspid valve

Trisomy 13 (Patau syndrome)
: Chromosome abnormality caused by an extra chromosome 13

Trisomy 18 (Edwards syndrome)
: Chromosomal abnormality caused by an extra chromosome 18

Ventricular septal defect
: Congenital heart defect characterized by one or several openings in the ventricular septum

Glossary of Common Terms

Agenesis
: Congenital absence of a body part or organ, implying that the structure never formed. Result of an error in development as opposed to an external process.

Asymptomatic
: without symptoms.

Atresia
: Congenital absence or closure of a normal opening.

Autosomal
: Relating to an *autosome*, a chromosome that is not a sex chromosome.

Birth defect
: Congenital abnormalities of structure, function or metabolism present before birth.

Chromosomal
: Referring to defects caused by anomalies in the chromosome, either inherited or caused by mutation.

Congenital
: Abnormality or problem present at birth. Includes defects detected prenatally and those not recognized until after the newborn period.

Dysplasia
: Abnormal cell organization of an organ. Usually congenital, may be acquired.

Fistula
: Abnormal connection between an internal organ and the body surface, or between two internal organs or structures. Can be congenital or acquired.

Genetic
: Referring to familial inheritance via genes.

Hyperplasia
: Overgrowth due to an increase in the number of cells of tissue.

Hypertrophy
: Overgrowth due to enlargement of existing cells.

Hypoplasia
: Small size of organ or part due to arrested development

Meninges
: Membranes that cover the brain and spinal cord.

Parity
: The number of children borne by one woman

Prenatal
: Preceding birth. Used when referring to the mother or the child.

Stenosis
: Narrowing or constriction of the diameter of a bodily passage or orifice.

Teratogen
: A drug or other agent that causes abnormal prenatal development.

Trisomy
: Chromosome abnormality characterized by a third copy of a chromosome. Includes complete and partial formation of an extra chromosome.

Acronyms

MCH	Maternal and Child Health
PRAMS	Pregnancy Risk Assessment Monitoring System
ABDR	Alaska Birth Defects Registry
ICD-9	International Classification of Disease Version 9
NBDPN	National Birth Defects Prevention Network
ASD	Atrial septal defect
VSD	Ventricular septal defect
PDA	Patent ductus arteriosus
FASD	Fetal alcohol spectrum disorder
FAS	Fetal alcohol syndrome
ARND	Alcohol related neurodevelopmental disorders
ARBD	Alcohol related birth defects
CP	Cleft palate without cleft lip
CLP	Cleft lip with or without cleft palate

Appendix F

RP	Relative prevalence
OGD	Obstructive genitourinary defect
CNS	Central nervous system
NTD	Neural tube defect
CI	Confidence interval
CSF	Cerebrospinal fluid
CHD	Congenital hip dislocation

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