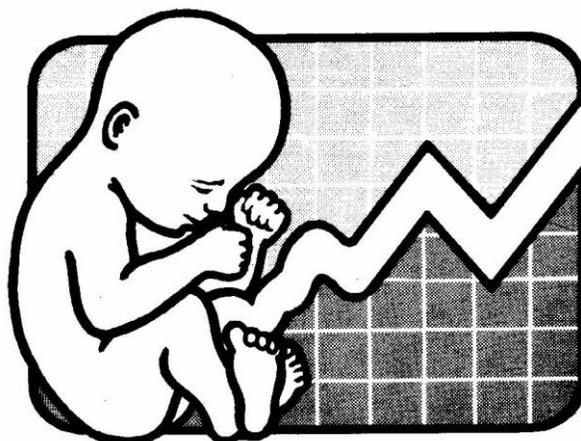




**2008 - 2009
Arizona Birth Defects
Monitoring Program Report**





Health and Wellness for all Arizonans

Janice K. Brewer, Governor
State of Arizona

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Arizona Department of Health Services

MISSION

Health and Wellness for all Arizonans

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2008 - 2009
ARIZONA BIRTH DEFECTS MONITORING
PROGRAM REPORT

Arizona Birth Defects Monitoring Program
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Bureau of Public Health Statistics
Arizona Department of Health Services

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The Arizona Department of Health Services (ADHS) does not discriminate on the basis of disability in the administration of its programs and services as prescribed by Title II of the Americans with Disabilities Act of 1990 and Section 504 of the Rehabilitation Act of 1973.

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EXECUTIVE SUMMARY

The Arizona Birth Defects Monitoring Program (ABDMP) is a population-based registry which provides information on the occurrence of birth defects (also referred to as congenital anomalies) throughout the state of Arizona. This report includes information about the occurrence of 31 categories of structural birth defects diagnosed in children born to Arizona residents from 2008-2009 (see Appendix A for the definitions of birth defects reported by the ABDMP). The specific birth defects included in this report are significant because they require medical or surgical intervention, considerably affect the child's appearance, and/or seriously affect the health and development of the child.¹

General Conclusions

- The total number of birth defects *cases* reported from 2008-2009 is 1,399. This includes infants, live-born and stillborn, with one or more reportable and confirmed birth defects, and accounts for individual babies identified with multiple defects.
- The total number of specific *defects* reported 2008-2009 is 1,673.
- There were 191,831 live births in Arizona from 2008-2009 (99,215 in 2008; 92,616 in 2009).²
 - From 2008-2009; 1,341 live-born infants born were confirmed to have one or more reportable birth defects.
- In Arizona, from 2008-2009, there were 1,055 stillborns of 20 weeks or greater gestation (spontaneous fetal losses, at 20+ weeks and 350g+).³
 - Of those who were stillborn at 20 weeks or more gestation, 58 had a reportable birth defect.
- The most common congenital anomalies were Trisomy 21 (Down syndrome); cleft lip with and without cleft palate; cleft palate without cleft lip; gastroschisis; and several specific heart defects, including pulmonary valve atresia and stenosis, tetralogy of Fallot, transposition of great arteries, and coarctation of aorta.

Birth defects affect thousands of families in the state of Arizona. The Arizona Birth Defects Monitoring Program plays a vital role in accurately identifying congenital anomalies that

occur throughout the state, and provides this data to state and community leaders and health care professionals to plan, implement, and evaluate programs for prevention, treatment, and referral to services in Arizona.

THE IMPORTANCE OF ARIZONA'S BIRTH DEFECTS REGISTRY

Introduction

Birth defect surveillance programs in the United States were first established in response to concerns over environmental pollutants and the finding that severe birth defects were associated with pregnant women who, without knowing the harmful effects on the fetus, used medications such as thalidomide. At present, birth defect surveillance programs are being used to monitor trends in birth defect rates over time, evaluate birth defect prevention programs, and facilitate research efforts to identify the etiology of birth defects.^{4,5}

The Arizona Birth Defects Monitoring Program (ABDMP) is a population-based registry which provides information on the occurrence of 31 categories of birth defects. These specific birth defects are monitored at the recommendation of the National Birth Defects Prevention Network and Centers for Disease Control and Prevention (CDC). The defects were selected because of a combination of factors: they require medical or surgical intervention, they considerably affect the child's appearance, they seriously affect the health and development of the child, they have a significant public health impact, their frequency of occurrence, and/or the level of existing knowledge on their etiology and risk factors.⁶ The ABDMP provides ongoing surveillance to monitor trends in the occurrence of these birth defects and detect the onset of possible problems.^{7,8,9} The information is used to plan and evaluate birth defect prevention efforts and to direct allocation of resources for health services. Such a registry is necessary because other systems for reporting birth defects, such as birth certificates and hospital discharge data, tend to be inaccurate or incomplete due to under-reporting of cases, lack of specificity of the type of birth defect, and/or incomplete demographic data.¹⁰

In 2003, the ABDMP entered into a five-year Cooperative Agreement with CDC to continue and improve Arizona's population-based birth defects surveillance system and to provide accurate and current information for health planning and prevention activities. The Cooperative Agreement was extended twice, through 2008 and 2009. ABDMP received a new 5-year grant from the CDC in 2010 to continue surveillance, prevention, and referral (to treatment and social services) activities.

Economic Cost

Birth defects are expensive, both in their costs in human life and productivity, as well as monetary resources. They remain the leading cause of infant mortality in both Arizona and the United States. Arizona data shows that from 2008 through 2009, 23% of infant deaths (through 1 year of age) were due to a congenital anomaly.¹¹ Birth defects are the fifth leading cause of years of potential life lost.¹² Although many infants with birth defects survive beyond their first birthday, many require special medical services, education and rehabilitation services, vocational training, and/or custodial care. Lifetime costs for these services is estimated to be \$75,000 to more than \$500,000 per child.¹³ These costs contribute to a lifetime of potential hardship for the affected children, their families, and our communities.

Known Causes of Birth Defects

Despite extensive research efforts, the causes of most birth defects remain unknown. It is known that both genetic and environmental factors play a role in birth defect etiology. There are three major categories of known causes of birth defects. The first category is “chromosomal errors,” which may cause defects such as Down syndrome. The second is “environmental factors,” such as maternal alcohol consumption, which may cause fetal alcohol syndrome. The third category is “maternal illness, infections, or medical conditions,” such as German measles, which may cause congenital rubella. While there are some known teratogens (specific factors that cause birth defects), research suggests that most birth defects are caused by complex combinations of genetic and environmental factors that are very difficult to identify – particularly in studies of relatively rare conditions. This is likely the reason researchers have been largely unsuccessful in identifying the etiology of birth defects.^{14,15,16}

Interventions

Although the causes of birth defects are largely unknown, there are many important steps women can take to minimize the risk of such defects. For example, maternal intake of the B-vitamin folic acid has been shown to be necessary for proper fetal development. Several prospective case-controlled studies have shown that the consumption of 400 mcg of folic acid daily starting prior to conception through the first trimester reduced the incidence of neural tube defects (NTDs) by at least 50%.¹⁷ Some research suggests that folic acid may reduce the incidence of

certain types of heart defects, urinary tract defects, and oral clefts.^{18,19,20} It is also known that maternal alcohol consumption may result in negative fetal outcomes, to include Fetal Alcohol Syndrome, facial defects, and certain heart defects.²¹ Education about folic acid intake and alcohol abstinence before and during pregnancy are two examples of public health interventions that could have a significant impact on improving the health of the population.

A considerable amount of research still needs to be undertaken to augment our knowledge of congenital anomalies, their etiology, and their impact on different subpopulations. Through surveillance and documentation of the occurrence of birth defects in the state, the ABDMP plays a vital role in this process. Making this information available to researchers and the public positively contributes to the well-being of Arizona's children.

METHODS

The ABDMP is a statewide, population-based, active ascertainment program, pursuant to Arizona Revised Statute §36-133, which mandates the surveillance of chronic diseases, including birth defects. The funding for the ABDMP comes from appropriations of the Arizona State Legislature, federal funds from the Maternal Child Health Block Grant, and a Cooperative Agreement with the CDC. Trained ABDMP staff members collect data from Arizona hospitals and medical facilities. Ascertainment procedures used by the ABDMP are similar to those used by the CDC's Metropolitan Atlanta Congenital Defects Program.²²

Hospital case-finding sources include the Hospital Discharge Database, disease indices from hospitals, and birth and fetal death certificates. All records for children up to one year of age with recorded birth defects become part of a listing of possible cases. ABDMP staff review the medical records of possible cases to determine which records meet the case definition. For each confirmed, reportable case ABDMP staff create an abstract (case report) of the medical record. If the nature of a defect diagnosed in the first year of life is more precisely diagnosed later in the child's life and this information is contained in the chart at the time of our review, then the more precise diagnosis is used. ABDMP staff assigns a six-digit classification code to each reportable defect. The classification system is CDC's modification of the British Pediatric Association (BPA) Classification of Disease. BPA coding is an extension of the World Health Organization's International Classification of Disease, 9th Revision, Clinical Modification (ICD-9-CM) (See Appendix B for a list of BPA codes used by the ABDMP). The abstracts of unique cases identified from multiple sources are added to the registry. Duplicate cases are compared, merged, and new information is added to the registry database.

CASE DEFINITION

The following are the criteria for inclusion in the Arizona Birth Defects Monitoring Program case file:

- A. The mother's place of residence at the time of birth must be in Arizona.
- B. The child must have a birth defect that is reported by the ABDMP (see Appendix B for the list of reported birth defects).
- C. The health care provider must have had a high level of confidence in the diagnosis (defined as a precision of diagnosis code of eight or greater, as explained in Appendix C.)
- D. The defect must be diagnosed, or signs and symptoms of a potential defect recognized, within the first year of life.
- E. The child must have an Arizona-issued birth or fetal death certificate.
- F. The date of birth (or delivery for stillbirths 20 weeks or greater gestational age) must be on or after January 1, 1986.
- F. For a list of exclusions, see Appendix D.

For 2008-2009, the ABDMP collected data on 31 categories of birth defects. The list of 31 categories includes many of the major congenital anomalies recommended by The International Clearinghouse for Birth Defects Monitoring Systems and the Centers for Disease Control and Prevention (CDC). The 31 categories of birth defects permit the ABDMP to compare its rates with other state registries for the major birth defect categories.

The data for the ABDMP Report (for 2008-2009 cases) has been analyzed using the 31 birth defect categories as defined by the CDC (see Appendix B). Of note, the data for previous Arizona reports contained analysis that used 32 to 44 birth defect categories defined by the ABDMP.

INTERPRETING THE DATA

The tables presented in this report represent data collected on birth defects in Arizona for the period 2008 to 2009. This report also includes some comparison data from prior years to help illustrate trends, compare incidence rates from recent years, and to improve understanding of the counts. Each table presents the reported counts and incidence rates on selected congenital anomalies. Below is an explanation of how cases, counts, and incidence rates, were calculated.

Cases

Cases account for the number of children who, in the first year of life, were diagnosed with at least one reportable birth defect within the defect category.

Counts

The counts represent the number of specific defects diagnosed in children in the first year of life. Children born with multiple reportable defects may be counted in more than one defect category, but will not be counted as more than one case.

Incidence Rates

Incidence rates of birth defects were calculated by dividing the number of children (cases) with a particular reportable defect by the total number of live births (and in some cases live births plus fetal deaths) for the specific year of interest, and then multiplying by 10,000. Most tables and figures show rates that are calculated by including live births and fetal deaths in the numerator and only live births for the denominator (the inclusion of fetal deaths in the denominator does not change the rates significantly). For example:

$$\frac{115 \text{ live borns or stillborns with Down syndrome born in the year 2009}}{92,616 \text{ live births in the year 2009}} * 10,000 = 12.41 \text{ cases of Down syndrome per 10,000 live births}$$

Small Numbers and a Note of Caution

While the intent of this data is to provide useful information on birth defects in Arizona, it is equally important not to mislead data users. Rates, confidence intervals, and any other analyses based on fewer than 10 reported cases cannot be considered statistically stable and are not shown for local areas.

DISCUSSION OF STATE DEFECTS

The Arizona Birth Defects Monitoring Program has been in operation since 1986. This is the fourteenth report of data compiled by the ABDMP in its mission to collect, analyze, and disseminate information on children with birth defects. Currently, the ABDMP has data from 1986 through 2009 for all the major reportable birth defects conditions.

Overview of Select Birth Defects

Neural Tube Defects

- There were 124 infants born with neural tube defects (NTDs) in this two year period. For the purposes of this report, NTDs include anencephaly, encephalocele, and spina bifida.
- Neural tube defects are serious conditions that may cause paralysis, severe mental retardation, or death.
- Intrauterine diagnosis and selective termination of affected pregnancies was hypothesized as one of the important contributors to the declining incidence of NTDs.²³
- The incidence rate of NTDs in the 3 most populated Arizona counties (Maricopa, Pinal, and Pima) remained relatively constant from 2007-2009. For all other counties across the state, there was some rate variability during this same time period; the variability is most likely due to a small number of NTDs and births per county.

Gastroschisis

- There were 113 infants born with gastroschisis in this two year period.
- For 2008-2009, the rates of gastroschisis among Native Americans (6 per 10,000 live births) and Hispanics (7 per 10,000 live births) were higher than the rate for White non-Hispanics (about 4 per 10,000 live births).
- The rate of gastroschisis for births to mothers less than 20 years of age (17 per 10,000 live births) was significantly higher than the state rate (6 per 10,000 live

births). This supports the finding that the younger the maternal age, the higher the risk of gastroschisis.^{24,25}

Heart Defects

- Heart defects represent the most common *category* of birth defects.
- From 2008-2009, there were 467 infants born with a reportable heart defect in Arizona.
- For this two year period, Arizona included eight types of heart defects in its list of reportable defects. These eight include aortic valve stenosis, coarctation of aorta, common trunkus, Ebstein anomaly, hypoplastic left heart syndrome, pulmonary valve atresia and stenosis, Tetralogy of Fallot, and transposition of great arteries.
- The heart defect rates varied across the state and appear unstable during this period because of the low case counts from counties with small numbers of births.

Fetus/newborn Affected by Maternal Alcohol Use

- For 2008-2009, ABDMP reports the rates for fetus/newborn affected by maternal alcohol use, rather than Fetal Alcohol Syndrome (FAS) as a defect category.
- FAS is a significant public health concern, with prevalence rates comparable with or above other common developmental disabilities, such as Down syndrome or spina bifida, have been reported.²⁶
- This category was collected and reported as a defect from 1986 through 2009.
- 2009 is the last year of surveillance and reporting of this category, due to several factors, including difficulty in confirming, late diagnoses, and high number of reports of exposure not related to defects:
 - Specific developmental delays and functioning are part of the diagnostic criteria for FAS, many of which are not normally identifiable until late preschool or early school age.
 - FAS is difficult to identify in the neonatal and infancy stages, and may not be correctly diagnosed or confirmed until age 4 to 7.²⁷
 - While many newborns may have been exposed to alcohol prenatally, not all are significantly affected.

- Research has directly linked prenatal alcohol exposure to other serious birth defects.^{28,29} Alcohol is a known teratogen, and promoting abstinence from alcohol during pregnancy remains a crucial intervention to reduce the number of babies affected by birth defects each year.

TABLES AND FIGURES

The following pages include various tables and figures representing different data collected and reported by ABDMP. Below is a summary of the information presented.

Table 1-A (pages 13-14) presents data on the 31 categories of birth defects collected by the ABDMP among live born and stillborn infants born statewide, analyzed by race/ethnicity, for 2008. **Table 1-B** (pages 15-16) presents similar data for 2009.

Table 2 (page 17) displays the number of live born and stillborn infants with all reportable birth defects and the average number of defects among live born and stillborn infants analyzed by county of maternal residence for 2008 and 2009.

Table 3 (page 18) displays the number of stillborn and live born infants, by the number of defects they were born with for 2008 and 2009.

Table 4 (pages 19-21) presents the number of cases and the incidence rates of the 31 categories of anomalies by year for 2000 through 2009.

The series of graphs in **Figure 1** (pages 22-26) display the trends for 2000 through 2009 for most of the defects listed in Table 4. Those without graphs are not reportable in 2008-2009.

Figure 2 (page 28) shows the counts and incidence rates from 2000-2009 for Neural Tube Defects (NTDs), combining anencephaly, encephalocele, and spina bifida.

Figure 3 (page 29) represent anencephaly, encephalocele, and spina bifida by race and ethnicity for births 2007 to 2009.

Figure 4 (page 30) graphs the incidence rates for critical congenital heart defects by race and ethnicity for births 2007 to 2009.

Figure 5 (page 31) compares rates of Neural Tube Defects by maternal age group for births 2008-2009.

Figure 6 (page 32) shows the incidence rates for Trisomy 21 by maternal age group for births 2008-2009.

Figure 7 (page 32) shows the incidence rates for Trisomy 13, 18, and 21 (combined) by maternal age group for births 2008-2009.

Figure 8 (page 33) compares incidence rates of gastroschisis by maternal age compared to all other defects for births 2008-2009.

Table 1 – A
Arizona Birth Defects Monitoring Program
Live-born and Stillborn Cases of Congenital Anomalies^a by Race/Ethnicity - Arizona 2008
Incidence Rate ^{b,c} Per 10,000 Live Births

<u>CONDITION</u>	<u>TOTAL^d</u>	<u>RATE</u>	<u>WHITE NON- HISP.</u>	<u>RATE</u>	<u>HISP.</u>	<u>RATE</u>	<u>BLACK</u>	<u>RATE</u>	<u>NATIVE AMER.</u>	<u>RATE</u>	<u>OTHER^e</u>	<u>RATE</u>
Amniotic bands	9	0.91	2	0.48	2	0.47	1	2.32	1	1.57	3	7.52
Anencephalus	15	1.51	4	1.00	8	1.88	1	2.32	1	1.57	1	2.51
Aniridia	1	0.10	1	0.24	0	0.00	0	0.00	0	0.00	0	0.00
Anophthalmia/microphthalmia	12	1.21	4	0.95	6	1.41	0	0.00	0	0.00	1	2.51
Anotia/microtia	17	1.71	4	0.95	9	2.11	0	0.00	2	3.14	2	5.02
Aortic valve stenosis	13	1.31	3	0.72	8	1.87	0	0.00	2	3.14	0	0.00
Biliary atresia	4	0.40	0	0.00	3	0.70	1	2.32	0	0.00	0	0.00
Bladder exstrophy	4	0.40	1	0.24	3	0.70	0	0.00	0	0.00	0	0.00
Choanal atresia	12	1.21	6	1.43	4	0.94	1	2.32	0	0.00	1	2.51
Cleft lip with and without cleft palate	118	11.89	46	10.97	42	9.85	6	13.95	16	19.44	8	20.08
Cleft palate without cleft lip	63	6.35	17	4.05	27	6.33	4	9.30	4	6.29	11	27.61
Coarctation of aorta	56	5.64	29	6.92	17	3.99	2	4.65	5	7.86	3	7.52
Common truncus	3	0.30	1	0.24	1	0.23	0	0.00	0	0.00	1	2.51
Congenital cataract	7	0.71	3	0.72	3	0.70	0	0.00	0	0.00	1	2.51
Diaphragmatic hernia	28	2.81	11	2.62	10	2.34	2	4.65	4	6.29	1	2.51
Down syndrome (Trisomy 21)	160	15.12	74	17.65	52	12.20	6	13.95	20	15.72	8	20.08
Edwards syndrome (Trisomy 18)	22	2.22	10	2.39	9	2.11	0	0.00	1	1.57	2	5.02
Ebstein anomaly	6	0.60	2	0.48	3	0.70	0	0.00	0	0.00	1	2.51
Encephalocele	11	1.11	3	0.72	3	0.70	2	4.65	2	3.14	1	2.51
Esophageal atresia/tracheoesophageal fistula	22	2.21	13	3.10	8	1.88	0	0.00	1	1.57	0	0.00

^a ABDMP collects and reports on some defects not shown in this table.

^b Incidence rates include live-born and stillborn cases in numerator and only live-born in denominator.

^c Incidence rates based on counts of less than 10 events are not statistically reliable.

^d Total includes cases of unknown race/ethnicity, therefore the total may be greater than the sum of the cases listed in the individual race/ethnicity categories.

^e 'Other' race/ethnicity category includes Asian, Pacific Islander, missing, and unknown classifications.

Table 1 – A (continued)
Arizona Birth Defects Monitoring Program
Live-born and Stillborn Cases of Congenital Anomalies^a by Race/Ethnicity - Arizona 2008
Incidence Rate^{b,c} Per 10,000 Live Births

<u>CONDITION</u>	<u>TOTAL^d</u>	<u>RATE</u>	<u>WHITE NON- HISP.</u>	<u>RATE</u>	<u>HISP.</u>	<u>RATE</u>	<u>BLACK</u>	<u>RATE</u>	<u>NATIVE AMER.</u>	<u>RATE</u>	<u>OTHER^e</u>	<u>RATE</u>
Fetus or newborn affected by maternal alcohol use	3	0.30	0	0.00	0	0.00	0	0.00	3	4.71	0	0.00
Gastroschisis	49	4.94	15	3.57	22	5.16	1	2.32	5	7.86	6	15.06
Hirschsprung disease (congenital megacolon)	15	1.51	7	1.67	4	1.41	0	0.00	0	0.00	2	5.02
Hypoplastic left heart syndrome	21	2.12	14	3.34	5	1.17	0	0.00	2	3.14	0	0.00
Omphalocele	23	2.32	9	2.15	10	2.35	2	4.65	0	0.00	2	5.02
Patau syndrome (Trisomy 13)	13	1.31	3	0.72	6	1.41	1	2.32	0	0.00	3	7.52
Pulmonary valve atresia and stenosis	39	3.93	12	2.86	22	5.16	0	0.00	2	3.14	3	7.52
Reduction deformity, lower limbs	19	1.92	5	1.19	6	1.41	4	9.30	1	1.57	3	7.52
Reduction deformity, upper limbs	22	2.22	5	1.19	9	2.11	2	4.65	2	3.14	4	10.04
Spina bifida without anencephalus	33	3.33	12	2.86	14	3.28	0	0.00	5	7.86	2	5.02
Tetralogy of Fallot	44	4.43	20	4.77	15	3.52	2	4.65	4	6.29	3	7.52
Transposition of great arteries	64	6.45	27	6.44	29	6.80	4	9.30	3	4.72	1	2.51

^a ABDMP collects and reports on some defects not shown in this table.

^b Incidence rates include live-born and stillborn cases in numerator and only live-born in denominator.

^c Incidence rates based on counts of less than 10 events are not statistically reliable.

^d Total includes cases of unknown race/ethnicity, therefore the total may be greater than the sum of the cases listed in the individual race/ethnicity categories.

^e 'Other' race/ethnicity category includes Asian and Pacific Islanders, missing, and unknown classifications.

Table 1 – B
Arizona Birth Defects Monitoring Program
Live-born and Stillborn Cases of Congenital Anomalies^a by Race/Ethnicity - Arizona 2009
Incidence Rate^{b,c} Per 10,000 Live Births

<u>CONDITION</u>	<u>TOTAL^d</u>	<u>RATE</u>	<u>WHITE NON- HISP.</u>	<u>RATE</u>	<u>HISP.</u>	<u>RATE</u>	<u>BLACK</u>	<u>RATE</u>	<u>NATIVE AMER.</u>	<u>RATE</u>	<u>OTHER^e</u>	<u>RATE</u>
Amniotic bands	2	0.22	0	0.00	0	0.00	1	2.28	1	1.62	0	0.00
Anencephalus	18	1.94	4	1.01	12	3.12	1	2.28	1	1.62	0	0.00
Aniridia	1	1.01	0	0.00	1	0.26	0	0.00	0	0.00	0	0.00
Anophthalmia/microphthalmia	10	1.08	2	0.50	7	1.82	0	0.00	0	0.00	1	2.55
Anotia/microtia	9	0.97	2	0.50	5	1.30	0	0.00	0	0.00	2	5.09
Aortic valve stenosis	17	1.84	4	1.01	9	2.35	2	4.57	2	3.24	0	0.00
Biliary atresia	1	0.11	1	0.25	0	0.00	0	0.00	0	0.00	0	0.00
Bladder exstrophy	1	0.00	0	0.00	1	0.26	0	0.00	0	0.00	0	0.00
Choanal atresia	9	0.97	5	1.25	3	0.78	0	0.00	0	0.00	1	2.55
Cleft lip with and without cleft palate	97	10.47	40	10.05	36	9.38	2	4.57	12	19.44	7	17.85
Cleft palate without cleft lip	53	5.72	21	5.27	20	5.21	0	0.00	7	11.34	5	12.75
Coarctation of aorta	40	4.32	20	5.03	14	3.65	2	4.57	1	1.62	3	7.64
Common truncus	5	0.54	2	0.50	3	0.78	0	0.00	0	0.00	0	0.00
Congenital cataract	4	0.43	2	0.50	2	0.52	0	0.00	0	0.00	0	0.00
Diaphragmatic hernia	9	0.97	3	0.75	5	1.30	0	0.00	1	1.62	0	0.00
Down syndrome (Trisomy 21)	115	12.41	47	11.81	54	14.07	3	6.85	3	4.86	8	20.38
Ebstein anomaly	8	0.86	2	0.50	4	1.04	0	0.00	1	1.62	1	2.55
Edwards syndrome (Trisomy 18)	15	1.62	8	2.01	2	0.52	2	4.57	3	4.86	0	0.00
Encephalocele	5	0.54	3	0.75	1	0.26	0	0.00	0	0.00	1	2.55
Esophageal atresia/tracheoesophageal fistula	24	2.59	14	3.52	8	2.08	1	2.28	1	1.62	0	0.00

^a ABDMP collects and reports on some defects not shown in this table.

^b Incidence rates include live-born and stillborn cases in numerator and only live-born in denominator.

^c Incidence rates based on counts of less than 10 events are not statistically reliable.

^d Total includes cases of unknown race/ethnicity, therefore the total may be greater than the sum of the cases listed in the individual race/ethnicity categories.

^e 'Other' race/ethnicity category includes Asian and Pacific Islanders, missing, and unknown classifications.

Table 1 – B (continued)
Arizona Birth Defects Monitoring Program
Live-born and Stillborn Cases of Congenital Anomalies^a by Race/Ethnicity - Arizona 2009
Incidence Rate^{b,c} Per 10,000 Live Births

<u>CONDITION</u>	<u>TOTAL^d</u>	<u>RATE</u>	<u>WHITE NON- HISP.</u>	<u>RATE</u>	<u>HISP.</u>	<u>RATE</u>	<u>BLACK</u>	<u>RATE</u>	<u>NATIVE AMER.</u>	<u>RATE</u>	<u>OTHER^e</u>	<u>RATE</u>
Fetus or newborn affected by maternal alcohol use	4	0.43	0	0.00	0	0.00	0	0.00	3	4.86	1	2.55
Gastroschisis	64	6.91	21	5.27	34	8.86	4	9.13	3	4.86	2	5.09
Hirschsprung disease (congenital megacolon)	12	1.29	7	1.76	4	1.04	1	2.28	0	0.00	0	0.00
Hypoplastic left heart syndrome	29	3.13	14	3.51	11	2.87	2	4.57	2	3.24	0	0.00
Omphalocele	17	1.84	8	2.01	7	1.82	0	0.00	0	0.00	2	5.09
Patau syndrome (Trisomy 13)	14	1.51	4	1.01	5	1.30	2	4.57	0	0.00	3	7.64
Pulmonary valve atresia and stenosis	38	4.10	17	4.27	14	3.65	3	6.85	1	1.62	3	7.64
Reduction deformity, lower limbs	6	0.65	1	0.25	4	1.04	1	2.28	0	0.00	0	0.00
Reduction deformity, upper limbs	21	2.27	7	1.76	9	2.35	2	4.57	1	1.62	2	5.09
Spina bifida without anencephalus	42	4.53	17	4.27	17	4.43	1	2.28	3	4.86	4	10.20
Tetralogy of Fallot	47	5.07	19	4.77	19	4.95	2	4.57	5	8.10	2	5.09
Transposition of great arteries	35	3.78	17	4.27	13	3.39	1	2.28	3	4.86	1	2.55

^a ABDMP collects and reports on some defects not shown in this table.

^b Incidence rates include live-born and stillborn cases in numerator and only live-born in denominator.

^c Incidence rates based on counts of less than 10 events are not statistically reliable.

^d Total includes cases of unknown race/ethnicity, therefore the total may be greater than the sum of the cases listed in the individual race/ethnicity categories.

^e ‘Other’ race/ethnicity category includes Asian and Pacific Islanders, missing, and unknown classifications.

Table 2
Arizona Birth Defects Monitoring Program
Birth Defects by County of Residence, 2008-2009

YEAR	2008				2009			
	LIVE-BORNS (LB) WITH DEFECTS		STILLBORNS (SB) WITH DEFECTS		LIVE-BORNS (LB) WITH DEFECTS		STILLBORNS (SB) WITH DEFECTS	
	Number with defect	% LB with defect	Number with defect	% SB with defect	Number with defect	% LB with defect	Number with defect	% SB with defect
COUNTY								
ARIZONA TOTAL	712	0.72	26	4.78	629	0.71	32	6.26
APACHE	8	0.01	0	0.00	5	0.01	0	0.00
COCHISE	10	0.01	0	0.00	13	0.01	0	0.00
COCONINO	20	0.02	1	0.18	11	0.01	3	0.59
GILA	6	0.01	0	0.00	3	0.00	1	0.20
GRAHAM	4	0.00	0	0.00	5	0.01	1	0.20
GREENLEE	3	0.00	0	0.00	0	0.00	0	0.00
LA PAZ	3	0.00	0	0.00	0	0.00	0	0.00
MARICOPA	438	0.44	12	2.21	376	0.40	16	3.13
MOHAVE	13	0.01	0	0.00	9	0.01	2	0.39
NAVAJO	19	0.02	1	0.18	16	0.02	0	0.00
PIMA	103	0.10	4	0.74	99	0.10	6	1.17
PINAL	43	0.04	6	1.10	48	0.05	1	0.20
SANTA CRUZ	6	0.01	1	0.18	7	0.01	0	0.00
YAVAPAI	17	0.02	0	0.00	9	0.01	0	0.00
YUMA	19	0.02	1	0.18	28	0.03	2	0.39
TOTAL ARIZONA BIRTHS	99215		544		92616		511	

Stillborns classified as 'reportable spontaneous fetal deaths.'

Table 3
Arizona Birth Defects Monitoring Program
Birth Defects by Number of Defects, 2008-2009

YEAR	2008				2009			
	LIVE-BORNS (LB)		STILLBORNS (SB)		LIVE-BORNS (LB)		STILLBORNS (SB)	
	Number with defect	% LB with defect	Number with defect	% SB with defect	Number with defect	% LB with defect	Number with defect	% SB with defect
ARIZONA TOTAL	712	0.72	26	4.78	629	0.71	32	6.26
1 DEFECT	604	0.60	16	2.75	552	0.60	25	4.89
2 DEFECTS	86	0.10	6	1.10	61	0.07	5	0.98
3 OR MORE DEFECTS	22	0.03	4	0.74	16	0.02	2	0.39
TOTAL ARIZONA BIRTHS	99215		544		92616		511	

Stillborns classified as 'reportable spontaneous fetal deaths.'

Table 4
Congenital Anomalies^a by Year, Live-Borns and Stillborns, 2000- 2009
Incidence Rates Per 10,000 Live Births, Arizona

CONDITION^b		2000	2003	2004	2005	2006	2007	2008	2009
Amniotic bands	Cases ^c	14	10	7	*	*	*	*	*
	Rate	1.65	1.10	0.75	*	*	*	*	*
Anencephaly	Cases	15	20	15	11	13	16	15	18
	Rate	1.77	2.20	1.61	1.15	1.23	1.56	1.51	1.94
Aniridia	Cases	1	0	0	0	0	1	1	1
	Rate	0.12	0.00	0.00	0.00	0.00	0.10	0.10	0.11
Anophthalmia/microphthalmia	Cases	22	24	14	10	7	5	12	10
	Rate	2.59	2.64	1.50	1.04	0.69	0.48	1.21	1.01
Anotia/microtia	Cases	18	16	23	16	13	10	17	9
	Rate	2.12	1.76	2.46	1.67	1.27	0.97	1.71	0.92
Aortic valve stenosis	Cases	33	34	30	17	17	26	13	17
	Rate	3.89	3.74	3.21	1.77	1.67	2.53	1.31	1.83
Biliary atresia	Cases	6	1	4	1	3	3	4	1
	Rate	0.71	0.11	0.43	0.10	0.29	0.29	0.40	0.10
Bladder exstrophy	Cases	2	3	2	2	1	1	4	1
	Rate	0.24	0.33	0.21	0.21	0.10	0.10	0.40	0.10
Choanal atresia	Cases	20	13	12	7	8	6	12	9
	Rate	2.36	1.43	1.28	0.73	0.78	0.58	1.21	0.97
Cleft lip with or without cleft palate	Cases	96	106	115	126	111	97	118	97
	Rate	11.31	11.68	12.31	13.15	10.88	9.45	11.89	10.47
Cleft palate without cleft lip	Cases	53	61	67	66	73	57	63	53
	Rate	6.24	6.71	7.17	6.89	7.15	5.55	6.35	5.72
Coarctation of aorta	Cases ^b	37	40	59	47	34	49	56	40
	Rate	1.77	4.41	6.32	4.91	3.33	4.77	5.64	4.31
Common truncus	Cases	6	11	8	6	4	8	3	5
	Rate	0.71	1.21	0.86	0.63	0.39	0.77	0.30	0.54
Congenital cataract	Cases	13	10	17	10	11	2	7	3
	Rate	1.53	1.10	1.82	1.04	1.08	0.19	0.71	0.43
Diaphragmatic hernia	Cases	29	22	19	23	18	24	28	9
	Rate	3.42	2.42	2.03	2.40	1.76	2.34	2.82	0.97
Down syndrome (Trisomy 21)	Cases	111	117	120	134	103	107	150	115
	Rate	13.06	12.89	12.85	13.99	10.09	10.42	15.12	12.42

^a ABDMP collects and reports on some defects not shown in this table. ^b See Appendix A and Appendix B for definitions of the conditions. ^c Cases" is the number of live-born and stillborn infants \geq 20 weeks gestation.

* Denotes a year in which the defect was non-reportable.

Table 4 (continued)
Congenital Anomalies^a by Year, Live-Borns and Stillborns, 2000- 2009
Incidence Rates Per 10,000 Live Births, Arizona

CONDITION^b		2000	2003	2004	2005	2006	2007	2008	2009
Ebstein anomaly	Cases ^c	9	4	10	10	4	8	6	8
	Rate	1.06	0.44	1.07	1.04	0.39	0.78	0.60	0.86
Edwards syndrome (Trisomy 18)	Cases	26	24	25	20	17	23	22	15
	Rate	3.06	2.64	2.68	2.09	1.67	2.24	2.12	1.62
Encephalocele	Cases	10	13	9	7	4	7	11	5
	Rate	1.78	1.43	0.96	0.73	0.39	0.68	1.11	0.54
Esophageal atresia/tracheoesophageal fistula	Cases	18	27	21	13	17	16	22	24
	Rate	2.12	2.97	2.25	1.36	1.67	1.56	2.22	2.59
Fetus or newborn affected by maternal alcohol use	Cases	3	7	5	2	2	0	*	*
	Rate	0.35	0.77	0.54	0.21	0.20	0.00	*	*
Gastroschisis	Cases	47	41	57	47	55	50	49	64
	Rate	5.53	4.51	6.10	4.91	5.39	4.87	4.94	6.91
Hirschsprung disease	Cases	12	13	12	17	6	14	15	12
	Rate	1.41	1.43	1.28	1.77	0.59	1.36	1.51	1.30
Hypoplastic left heart syndrome	Cases	15	20	16	29	20	37	21	29
	Rate	1.77	2.20	1.71	3.03	1.96	3.60	2.12	3.13
Omphalocele	Cases	19	17	13	5	15	24	23	17
	Rate	2.24	1.87	1.39	0.52	1.47	2.34	3.32	1.84
Patau Syndrome (Trisomy 13)	Cases	18	17	13	7	8	11	13	14
	Rate	2.12	1.87	1.39	0.73	0.78	1.07	1.31	1.51
Pulmonary valve atresia and stenosis	Cases	78	77	95	56	66	33	39	38
	Rate	9.19	8.48	10.17	5.85	6.47	3.21	3.93	4.10
Pyloric stenosis	Cases	159	178	164	*	*	*	*	*
	Rate	18.74	19.61	17.56	*	*	*	*	*
Rectal and large intestinal atresia/stenosis	Cases	33	36	33	*	*	*	*	*
	Rate	3.89	3.97	3.53	*	*	*	*	*

^a ABDMP collects and reports on some defects not shown in this table. ^b See Appendix A and Appendix B for definitions of the conditions. ^c Cases" is the number of live-born and stillborn infants \geq 20 weeks gestation.
* Denotes a year in which the defect was non-reportable.

Table 4 (continued)
Congenital Anomalies^a by Year, Live-Borns and Stillborns, 2000- 2009
Incidence Rates Per 10,000 Live Births, Arizona

CONDITION^b		2000	2003	2004	2005	2006	2007	2008	2009
Reduction deformity, lower limbs	Cases ^c	22	15	6	10	6	6	19	6
	Rate	2.59	1.65	0.64	1.04	0.59	0.58	1.92	0.65
Reduction deformity, upper limbs	Cases	37	31	23	29	24	15	22	21
	Rate	4.36	3.41	2.46	3.03	2.35	1.46	2.22	2.26
Renal agenesis/hypoplasia	Cases	36	36	42	*	*	*	*	*
	Rate	4.24	3.97	4.50	*	*	*	*	*
Spina bifida without anencephalus	Cases	29	40	31	33	28	38	33	42
	Rate	3.42	4.41	3.32	3.44	2.74	3.70	3.33	4.53
Tetralogy of Fallot	Cases	33	41	42	42	37	40	46	47
	Rate	3.88	4.52	4.50	4.50	3.63	3.89	4.67	4.74
Transposition of great arteries	Cases	42	38	48	36	35	44	64	35
	Rate	4.95	4.19	5.14	3.76	3.43	4.28	6.45	3.78

^a ABDMP collects and reports on some defects not shown in this table. ^b See Appendix A and Appendix B for definitions of the conditions. ^c Cases" is the number of live-born and stillborn infants \geq 20 weeks gestation.

* Denotes a year in which the defect was non-reportable.

The rates are calculated as the number of live-born and stillborn cases of each defect in each year divided by the total number of live births in each year. Live births for each year are as follows:

Year	2000	2003	2004	2005	2006	2007	2008	2009
# of live births in Arizona to Arizona residents	84,985	90,783	93,396	95,798	102,042	102,687	99,215	92,616

Figure 1:
Trends of Selected Congenital Anomalies, Incidence Rates, 2000-2009
(Live-Born and Stillborn Cases per 10,000 Live Births)

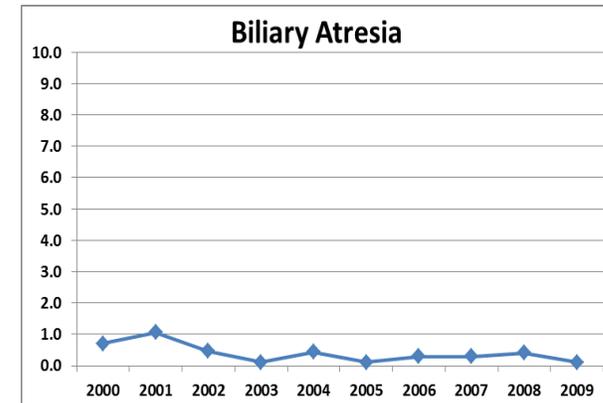
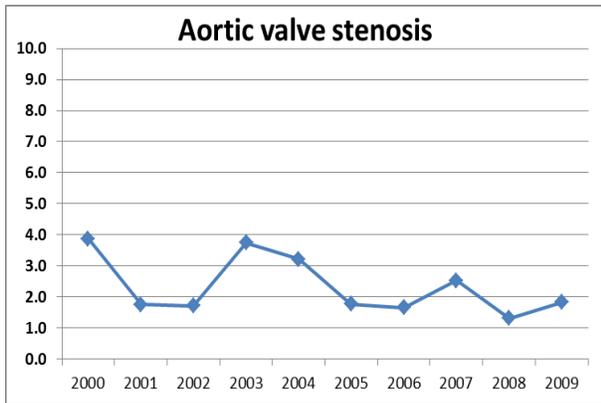
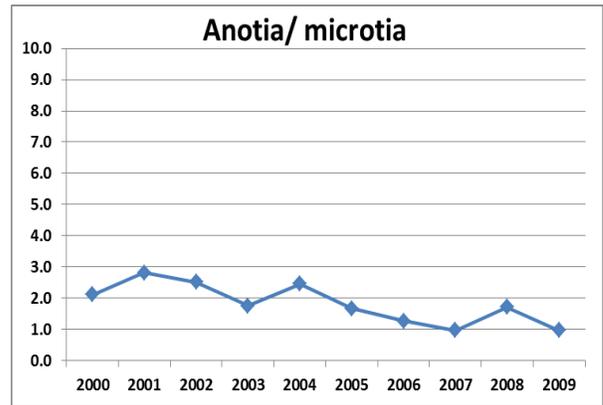
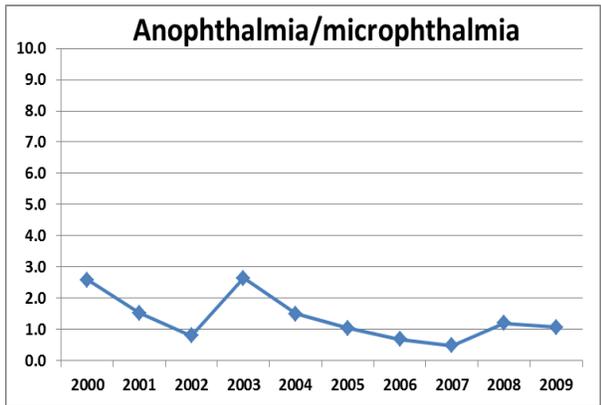
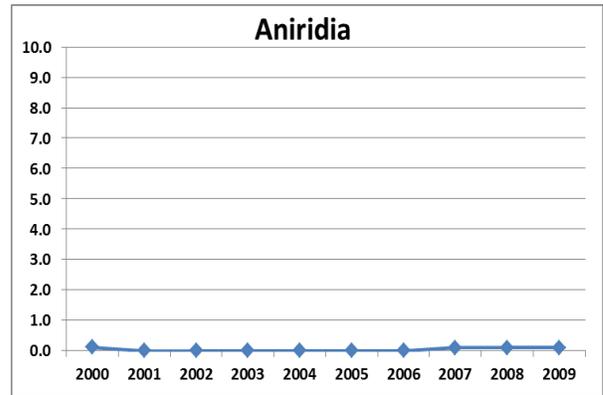
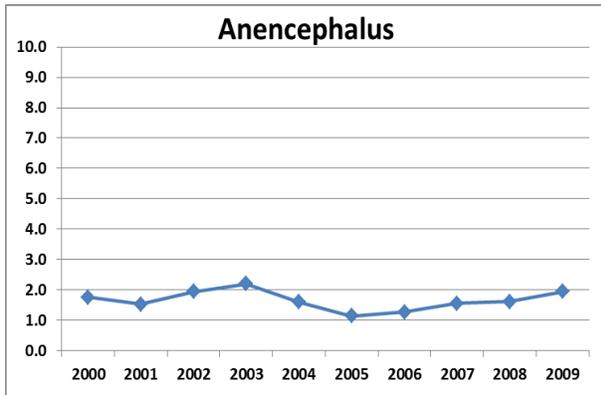
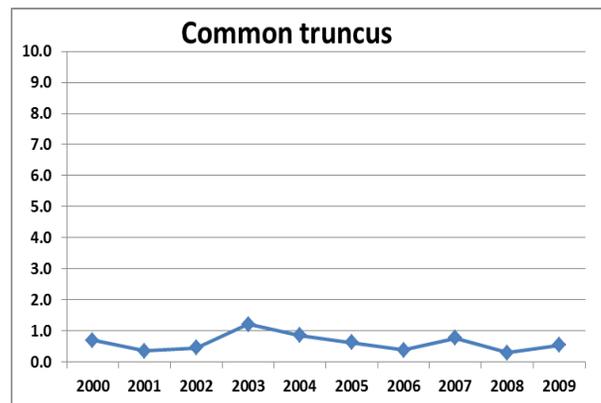
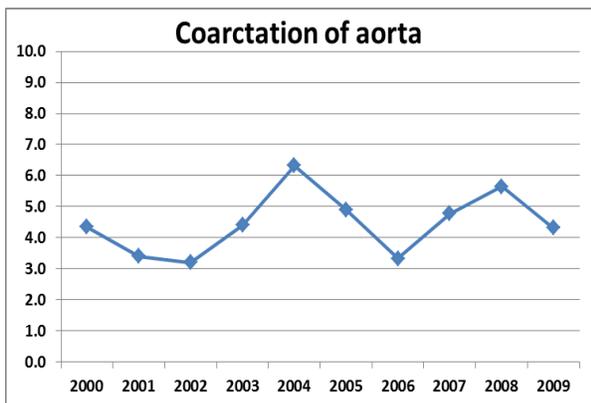
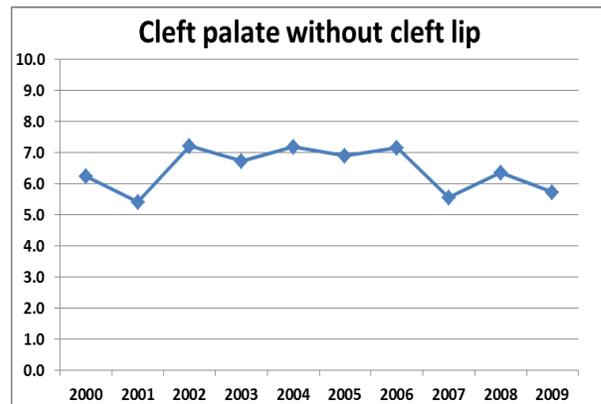
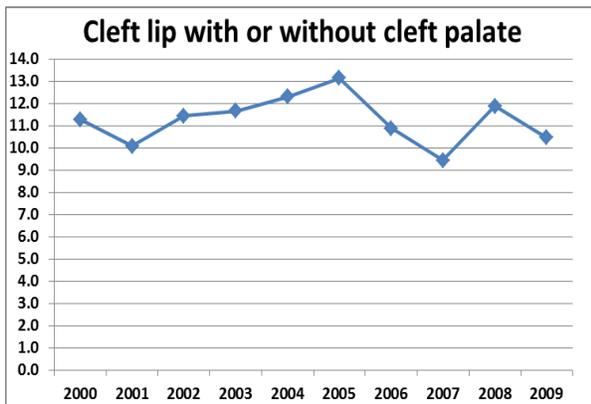
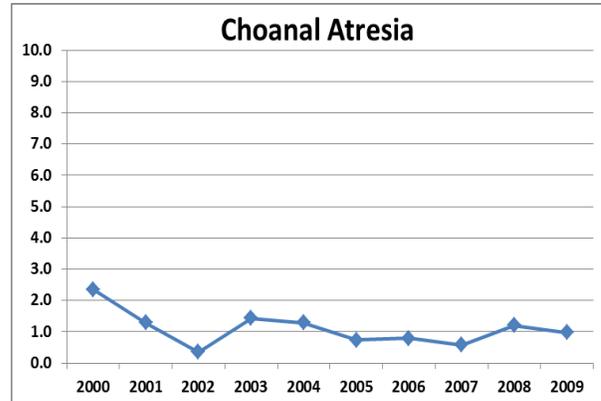
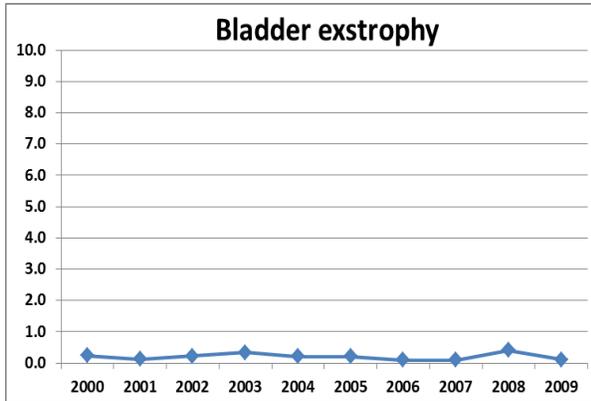
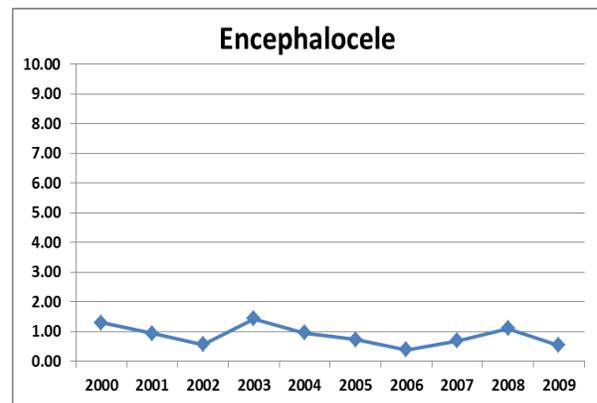
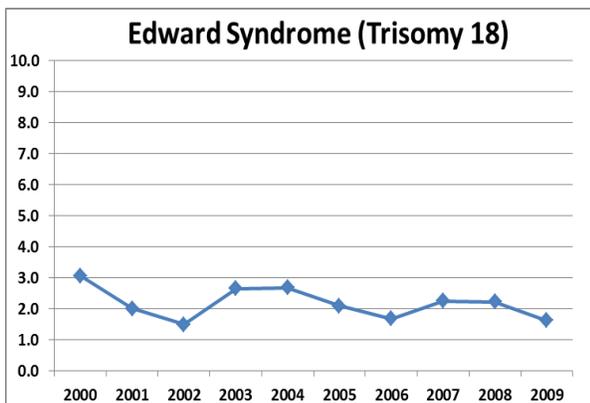
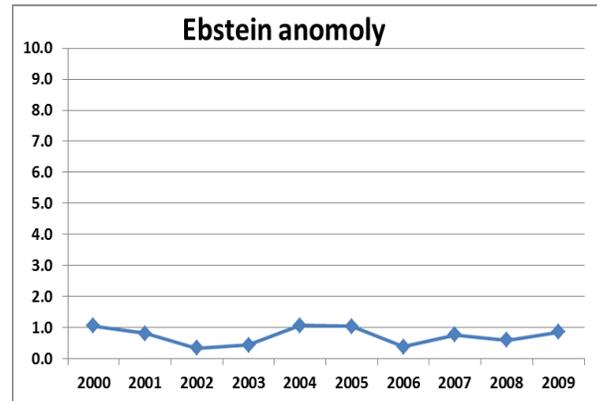
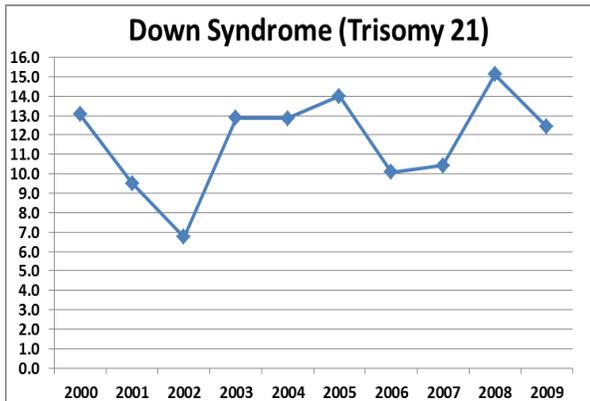
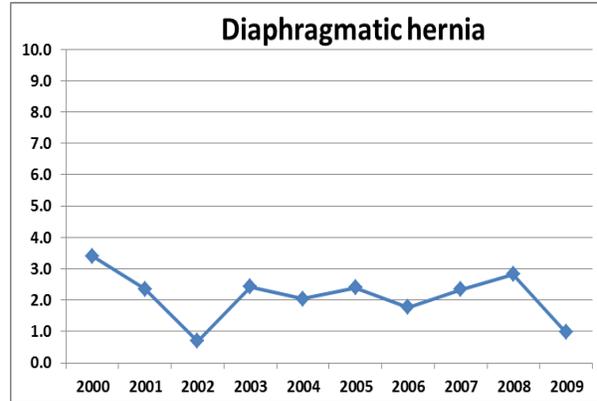
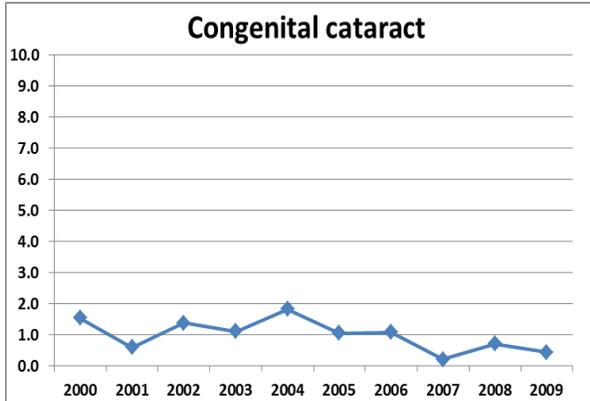


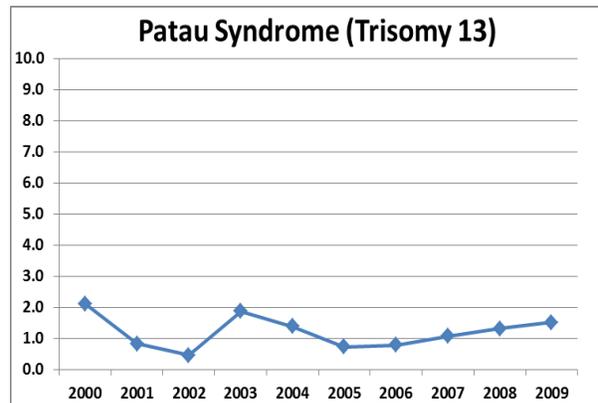
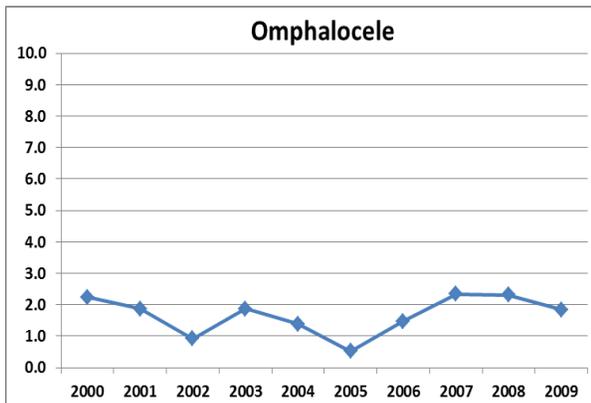
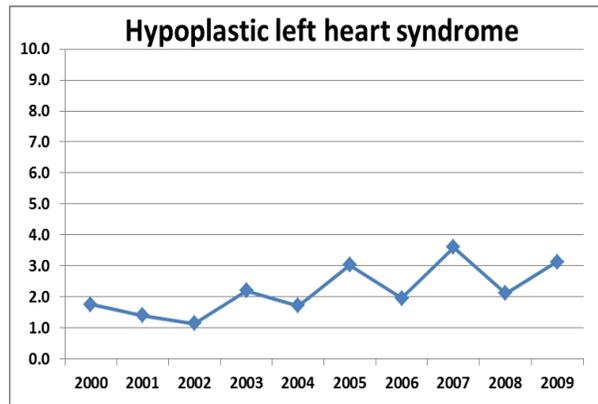
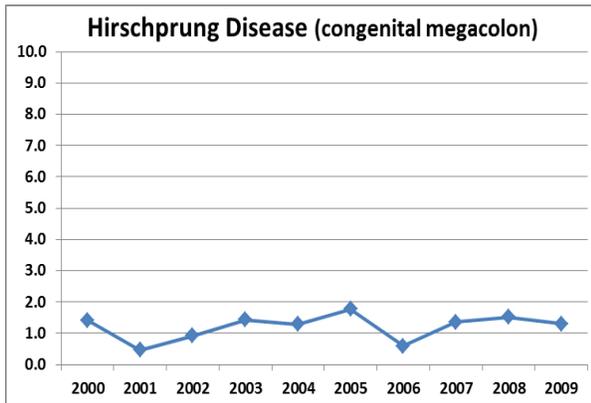
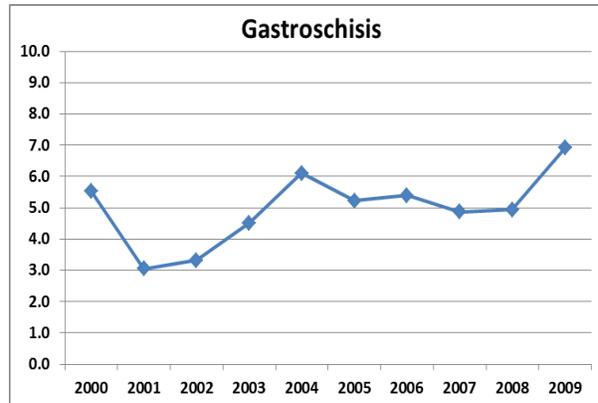
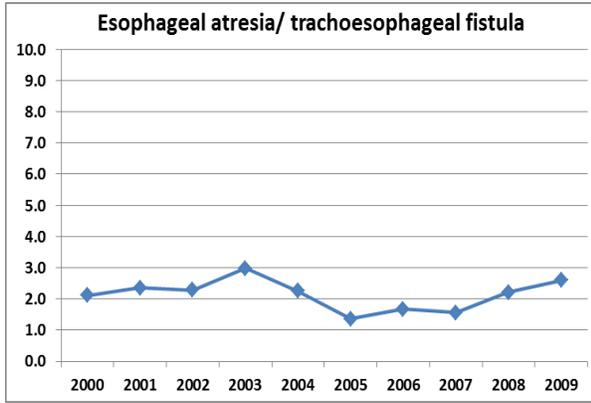
Figure 1 (continued):
Trends of Selected Congenital Anomalies, Incidence Rates, 2000-2009
(Live-Born and Stillborn Cases per 10,000 Live Births)



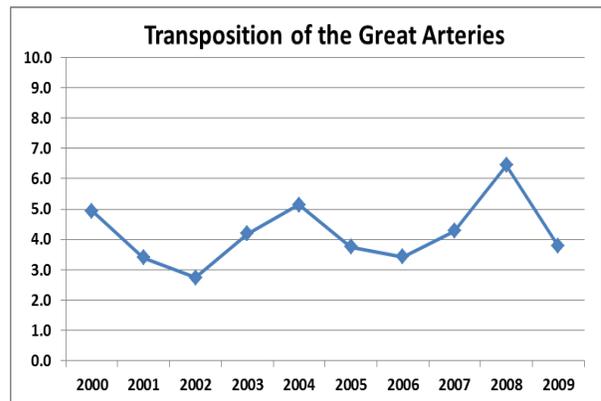
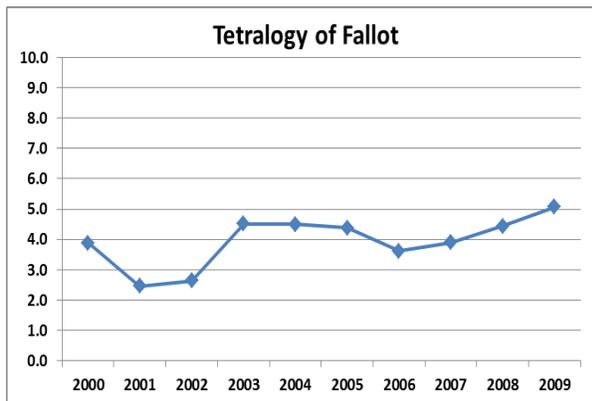
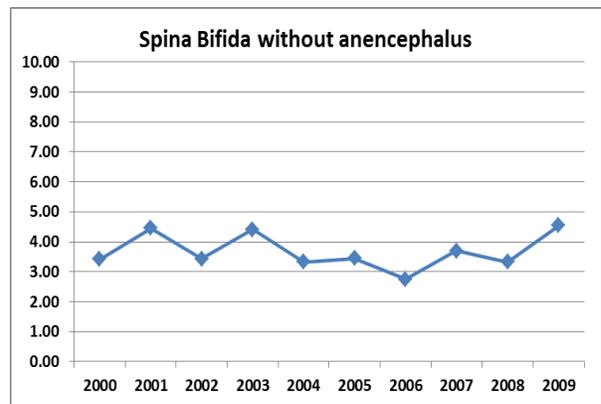
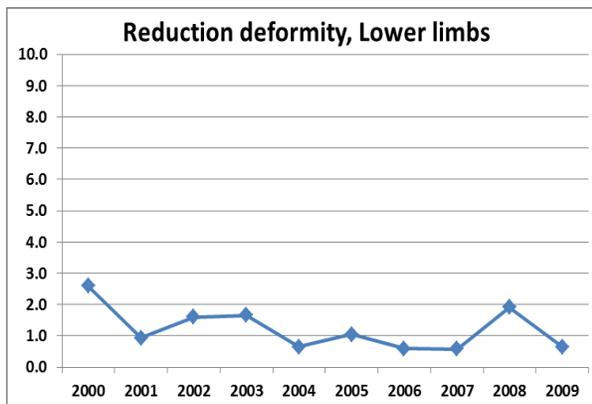
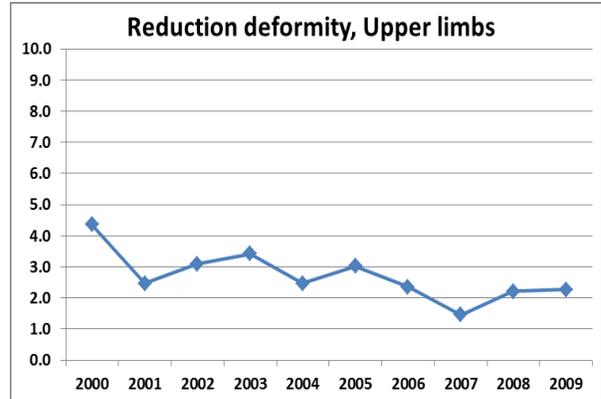
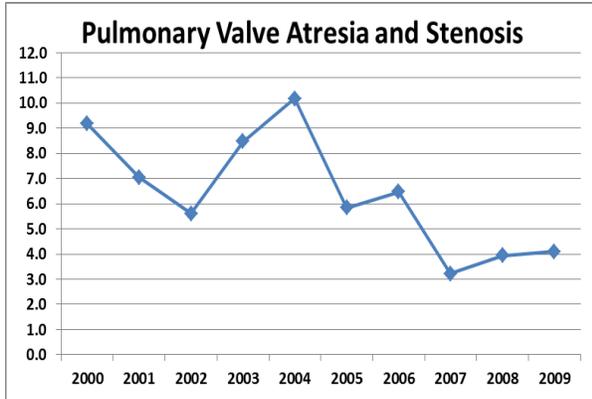
**Figure 1 (continued):
Trends of Selected Congenital Anomalies, Incidence Rates, 2000-2009
(Live-born and Stillborn Cases per 10,000 live births)**



**Figure 1 (continued):
Trends of Selected Congenital Anomalies, Incidence Rates, 2000-2009
(Live-born and Stillborn Cases per 10,000 live births)**



**Figure 1 (continued):
Trends of Selected Congenital Anomalies, Incidence Rates, 2000-2009
(Live-born and Stillborn Cases per 10,000 live births)**



Neural Tube Defects

Neural tube defects (NTDs) result from the failure of the neural tube to close properly during fetal development, occurring up to approximately four weeks gestation.³⁰ Under the NTD classification, there are three major defect categories and several smaller defects. For the purposes of this report, the focus is on the three major NTDs, which include anencephaly, encephalocele, and spina bifida (without anencephalus). Anencephaly is an absence of part or all of the brain. Encephalocele is the herniation of brain tissue through a gap in the skull. Spina bifida is a defective closure of the bones of the spine, through which the spinal cord and meninges may or may not protrude.

Research indicates that maternal obesity, socioeconomic status and neighborhood social conditions, prior spontaneous and elective terminations, and short periods of time between pregnancies (interpregnancy intervals) are associated with an increased risk for an NTD-affected pregnancy.^{31,32,33} However, compelling data from randomized clinical trials shows that daily intake of 400 mcg of folic acid starting before conception and continuing through the first trimester can reduce the risk of an NTD-affected pregnancy by at least 50 percent.^{34,35} Because of the overwhelming evidence that folic acid helps to prevent these birth defects, the Food and Drug Administration (FDA) mandated fortification of cereal grain products with 140 mcg/100 g of folic acid starting in January of 1998.³⁶

On average, there was approximately a 47% decrease in NTD-affected pregnancies in the United States from 1991-2006.³⁷ There was a 46% decline in NTD rates during the same time period in Arizona. The decreases are associated with the FDA's fortification mandate.³⁸ The pre-fortification NTD rate in Arizona (1990 to 1999 data combined) was 8.84 cases per 10,000 live births. The post-fortification NTD rate in Arizona (2000 to 2009 data combined) was 6.09 cases per 10,000 live births. Although there was an increase from 2006 to 2007, overall, the data shows a 31% decline in NTD rates in Arizona post-fortification through 2009.

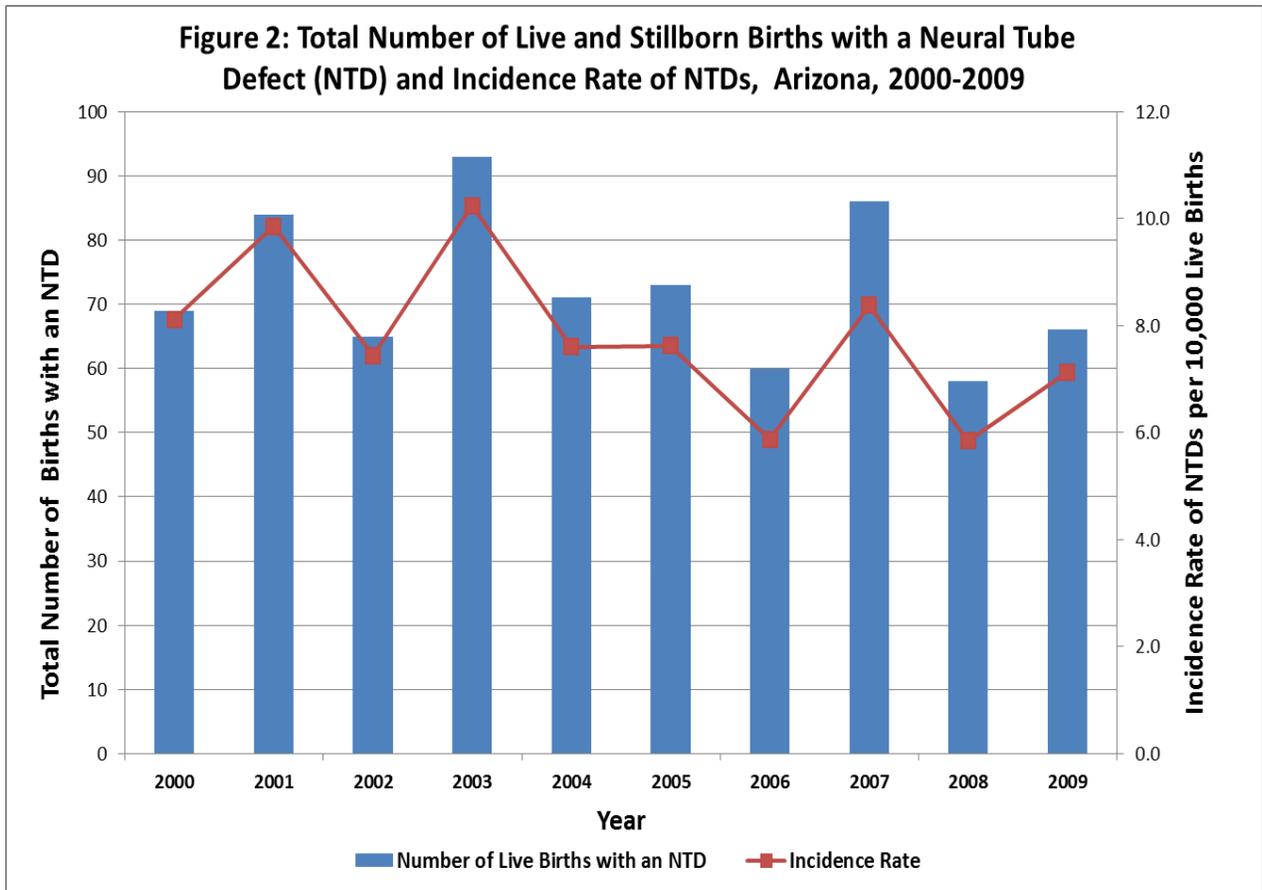
While Arizona data shows an overall decrease in NTDs post fortification, the incidence rate of NTDs in the 3 most populated Arizona counties (Maricopa, Pinal, and Pima) remained relatively constant from 2007-2009. For all other counties across the state, there was some rate variability

during this same time period; however, the variability is most likely due to a small number of NTDs and births per county.

NTDs occur very early in pregnancy, before most women know they are pregnant. While fortification has made an impact on the rates of NTDs, the current public health recommendation includes folic acid supplementation for all women between 15 and 45 years of age.³⁹ With supplementation, evidence suggests that 50% to 70% of these birth defects can be prevented if all women of childbearing age take 400 mcg of folic acid daily, starting before they become pregnant.^{40,41,42}

Figure 2 shows the counts and incidence rates from 2000-2009 for NTDs, combining anencephaly, encephalocele, and spina bifida.

Figure 2 shows neural tube defects (NTDs) rates for 2000-2009.

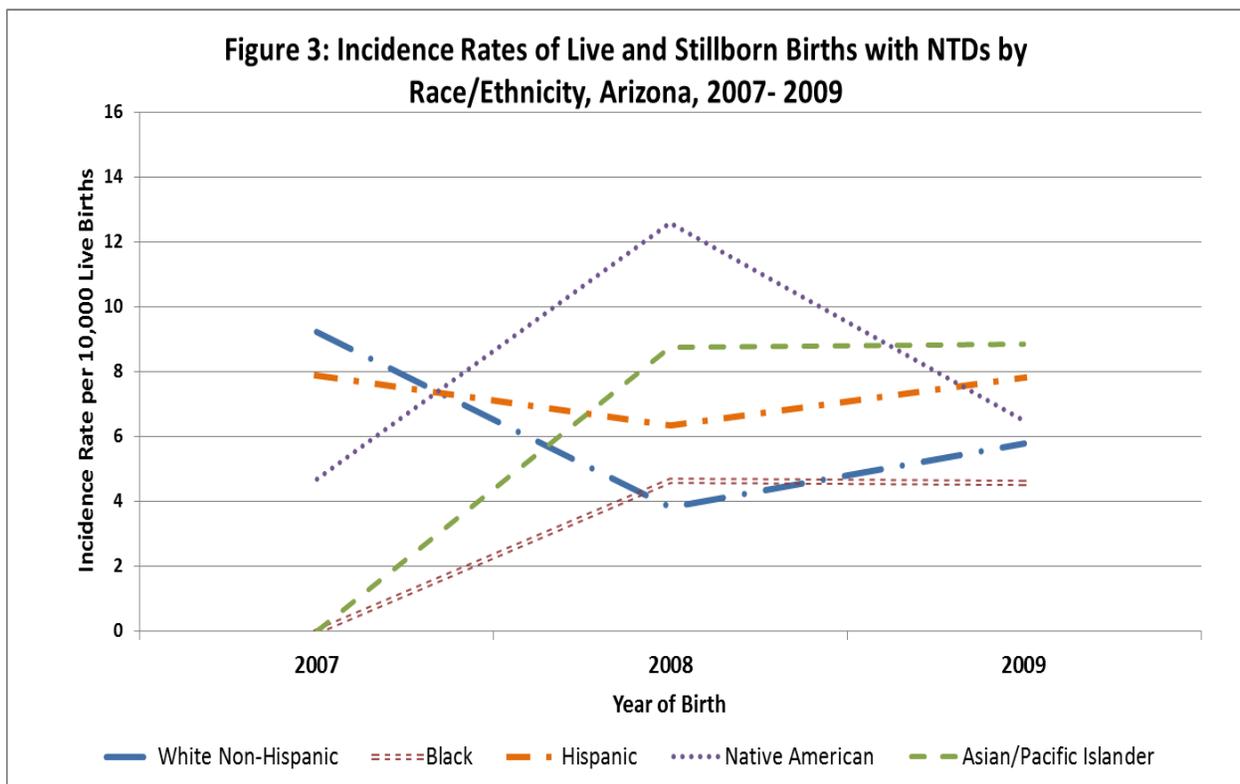


Race/Ethnicity

Birth defects occur in all racial and ethnic groups, but the frequency and types of these defects vary by race and ethnicity.^{43,44} The race and ethnicity information collected in the Arizona birth and fetal death certificates allow for the analysis of birth defects by race and ethnicity. See Appendix E for an explanation of how race/ethnicity is determined for each case.

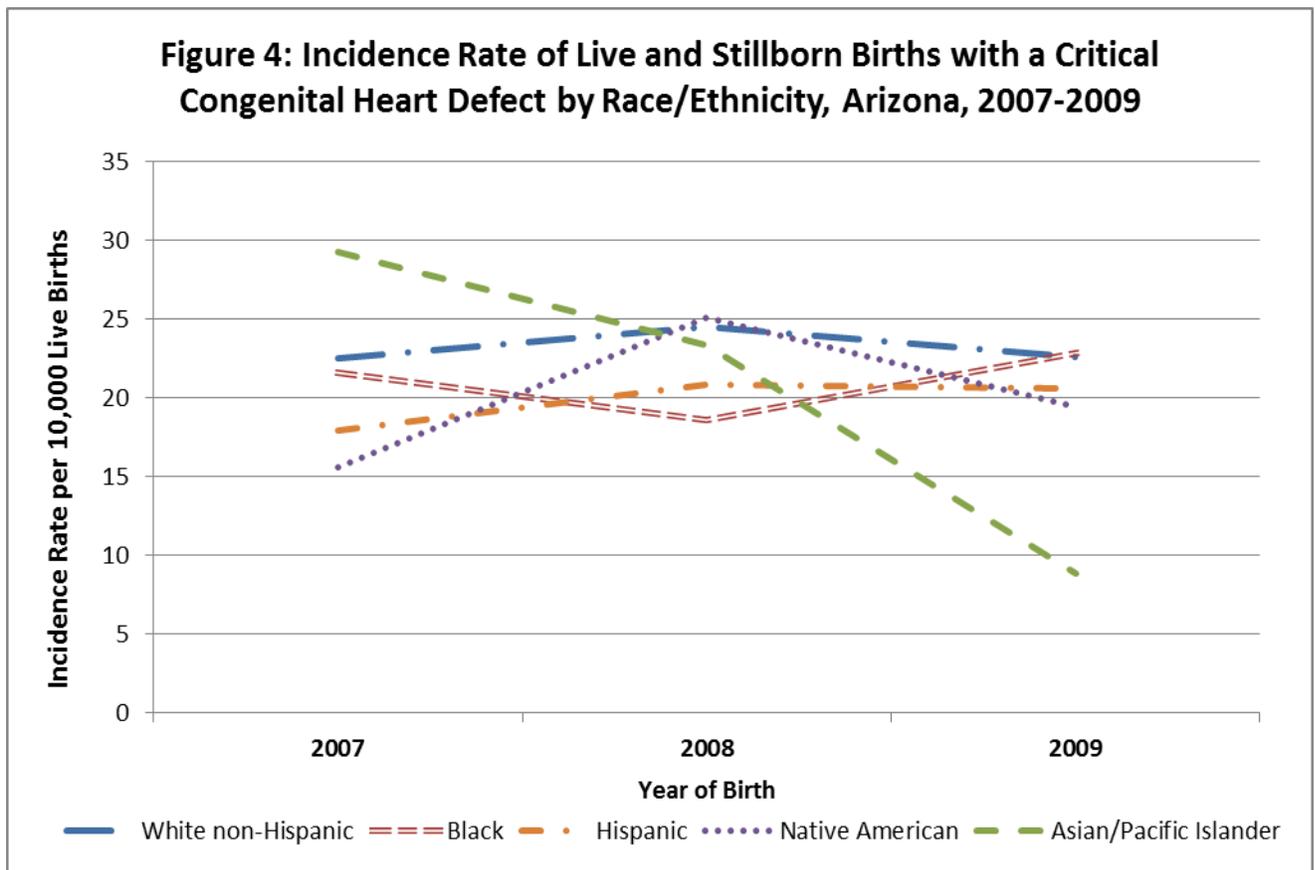
The rates in **Figure 3** represent anencephaly, encephalocele, and spina bifida by race and ethnicity for births 2007 to 2009. (See Tables 1-A and 1-B for data). The rate of NTDs among Native Americans was higher in 2008 than the rates for White non-Hispanics (12.5 for Native Americans vs. 3.8 for White non-Hispanics per 10,000 live births). Despite this sharp increase in 2008 from the previous year, the rate in 2009 shows an equally important decrease in the NTD incidence rates for Native Americans - making the overall increase in NTDs quite small (from 4.5 in 2007 to 6.5 in 2009 per 10,000 live births). In contrast, the rate of NTDs for Asians/Pacific Islanders shows an increase over this time period (0.0 in 2007 to 9.0 in 2009 per 10,000 live births). The yearly counts are relatively small, also making the rates appear unstable.

Figure 3 shows the time trends of NTDs by race/ethnicity for 2007 to 2009.



In **Figure 4** incidence rates for critical congenital heart defects have been stratified by race and ethnicity. The rate for Asians/Pacific Islanders shows a significant decrease from 2008 to 2009 (from 24 per 10,000 live births to 9 per 10,000 live births). The cause of this decrease is unknown. All other races in the state have had no significant rate change during this time period. Similar to the trend seen for NTDs, there was a slight increase in the rates of congenital heart defect in 2008 for all races, except Black and Asian/Pacific Islander, with a decrease in 2009 for all races, except Black. For the purposes of this graph, the critical congenital heart defects included are: coarctation of aorta, total anomalous pulmonary venous return, common truncus, hypoplastic left heart syndrome, pulmonary valve atresia and stenosis, tetralogy of Fallot, transposition of the great arteries and tricuspid valve atresia and stenosis.

Figure 4 shows the rate of certain critical congenital heart defects (CCHDs) by race/ethnicity for 2007 to 2009.

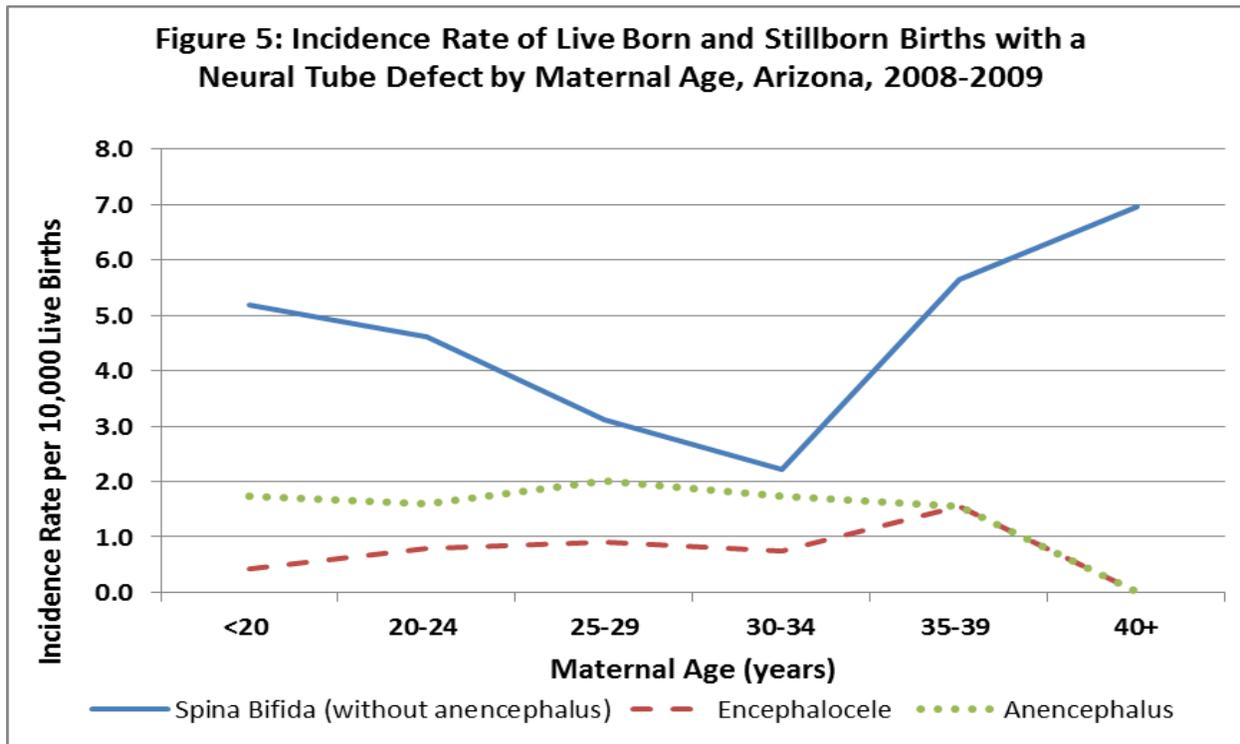


Maternal Age

Maternal age is commonly cited as a risk factor for congenital anomalies. The age of the mother has an impact on the occurrence of chromosomal and other types of defects. Relating to specific anomalies, risk may increase or decrease with advancing maternal age.⁴⁵

Figure 5 demonstrates that for anencephaly and encephalocele, the rate of occurrence remains relatively consistent until the maternal age of 40 years and older; after which the rate decreases. Spina bifida (without anencephalus) has a significantly higher rate as maternal age increases—increasing from an incidence rate of just over 2 per 10,000 live births in the age group 30-34, to 7 per 10,000 live births in mothers age 40 years and over.

Figure 5 compares rates of Neural Tube Defects occurring in 2008-2009 among different maternal age groups.



The risks for chromosomal defects (e.g., Trisomies 13, 18, and 21) are commonly noted to increase with maternal age. This correlation is most often discussed in relation to Down syndrome (Trisomy 21). **Figure 6** illustrates the incidence rates for Trisomy 21 by maternal age, while **Figure 7** combines data to show the incidence rates for Trisomy 13 (Patau syndrome), Trisomy 18 (Edward syndrome), and Trisomy 21 (Down syndrome) collectively, by maternal age. It is interesting to note that almost identical patterns are seen when combining the three anomalies (Trisomies 13, 18, and 21).

Figure 6 shows the incidence rates for Trisomy 21 by maternal age group.

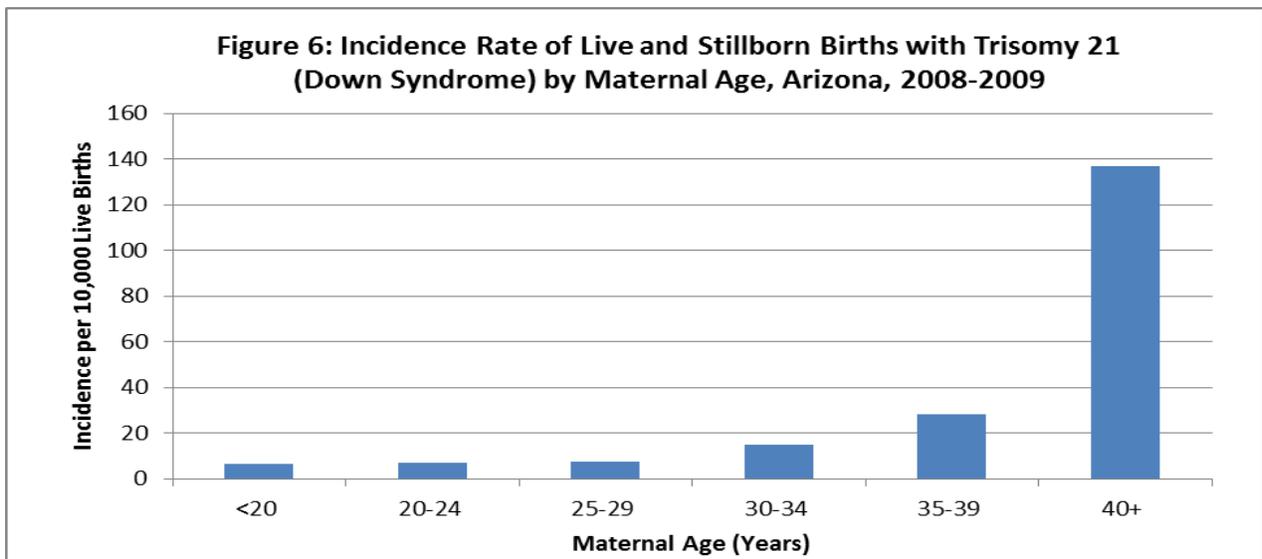
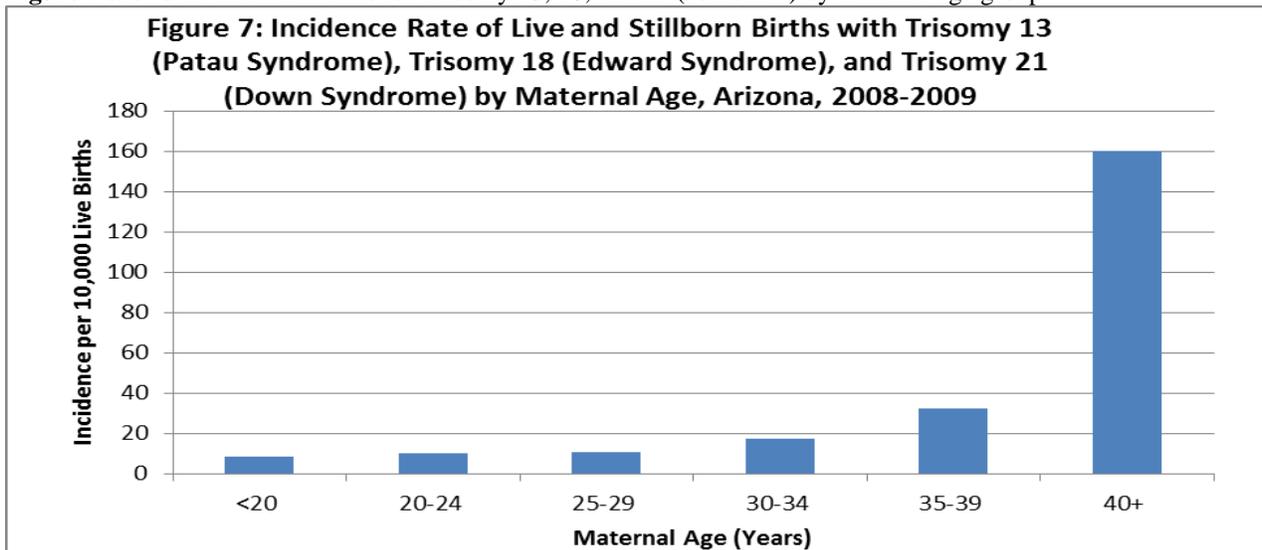
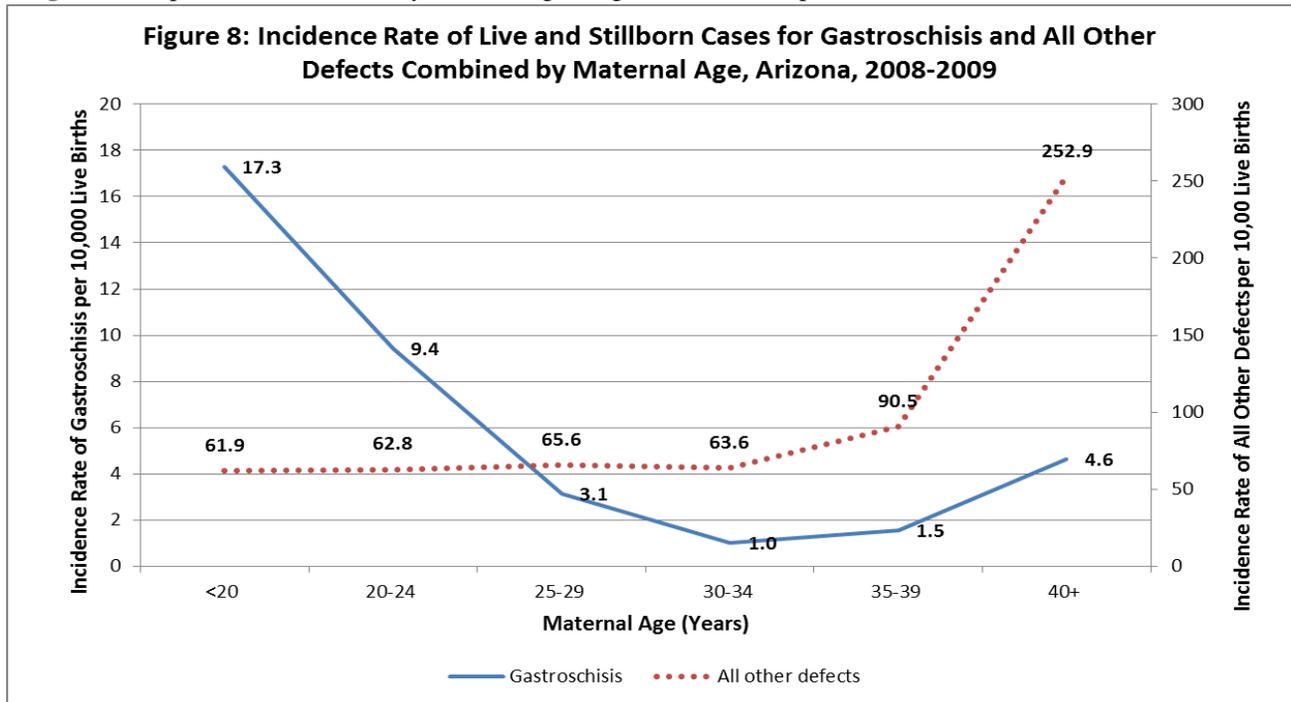


Figure 7 shows the incidence rates for Trisomy 13, 18, and 21 (combined) by maternal age group.



The opposite trends are noted for gastroschisis. As seen in **Figure 8**, the incidence rates for gastroschisis are significantly higher for moms age 20 and below, and decrease consistently through maternal ages of 30-34, with only slight increases in rates with increasing maternal age.

Figure 8 compares incidence rates by maternal age for gastroschisis compared to all other defects.



APPENDIX A

Definitions of Reported Birth Defects

(As defined in the National Birth Defects Prevention Network's *Guidelines for Conducting Birth Defect Surveillance*, Appendix 3.2)

Amniotic Bands

Strands of tissue that float in the amniotic fluid as a consequence of tears or ruptures in the amniotic membrane which surrounds the fetus during development.

Anencephaly (anencephalus)

Partial or complete absence of the brain and skull.

Aniridia

Hypoplasia of the iris of both eyes.

Anophthalmia/Microphthalmia

Anophthalmia – Total absence of eye tissue or apparent absence of the globe in an otherwise normal orbit.

Microphthalmia – Reduced volume of the eye. The corneal diameter is usually less than 10 millimeters, or the anteroposterior globe diameter is less than 20 millimeters.

Anotia/Microtia

Anotia – Total absence of the external ear and canal.

Microtia – Malformation or hypoplasia of the external ear (auricle, pinna).

Aortic Valve Stenosis

Obstruction or narrowing of the aortic valve, which may impair blood flow from the left ventricle to the aorta.

Biliary Atresia

Congenital absence of the lumen of the extrahepatic bile ducts.

Bladder Exstrophy

A defect in the lower abdominal wall and anterior wall of the bladder through which the lining of the bladder is exposed to the outside.

Choanal Atresia

Congenital obstruction of the opening of the nasal cavity into the nasopharynx on either side. This prevents communication of the nasal cavity with the pharynx.

Cleft Lip with and without Cleft Palate

A defect in the upper lip resulting from incomplete fusion of the parts of the lip.

Cleft Palate without Cleft Lip

An opening in the roof of the mouth resulting from incomplete fusion of the shelves of the palate. The opening may involve the hard palate only, the soft palate only, or both.

Coarctation of the Aorta

Narrowing of the descending aorta, which may obstruct blood flow from the heart to the rest of the body. The most common site of coarctation occurs distal to the origin of the left subclavian artery in the region of the ductus arteriosus.

Common Truncus (Truncus Arteriosus or TA)

Failure of separation of the aorta and the pulmonary artery, resulting in a single common arterial trunk carrying blood from the heart to both the body and lungs.

Congenital Cataract

An opacity of the lens of the eye that has its origin prenatally.

Diaphragmatic Hernia

Incomplete formation of the diaphragm through which a portion of the abdominal contents herniate into the thoracic cavity.

Down Syndrome (Trisomy 21)

The presence of three copies of all or a large part of chromosome 21.

Ebstein Anomaly

Downward displacement of the tricuspid valve into the right ventricle. The tricuspid valve is usually hypoplastic and regurgitant.

Edwards Syndrome (Trisomy 18)

The presence of three copies of all or a large part of chromosome 18.

Encephalocele

Herniation of brain tissue and/or meninges through a defect in the skull. The hernia sac is usually covered by skin.

Esophageal Atresia/ Tracheoesophageal Fistula

Esophageal atresia – A condition in which the esophagus ends in a blind pouch and fails to connect with the stomach.

Tracheoesophageal fistula – An abnormal communication between the esophagus and the trachea. This is almost always associated with some form of esophageal atresia.

Fetus/Newborn Affected by Maternal Alcohol Use (Fetal Alcohol Syndrome/FAS)

A spectrum of abnormalities resulting from exposure to alcohol *in utero*. While the specific abnormalities vary among individuals, the hallmarks include growth deficiency, microcephaly, facial dysmorphisms, and neurodevelopmental abnormalities.

Gastroschisis

A congenital opening or fissure in the anterior abdominal wall lateral to the umbilicus through which the small intestine, part of the large intestine, and occasionally the liver and spleen, may herniate. The opening is separated from the umbilicus by a small bridge of skin, and the herniating organs are not covered by a protective membrane. Gastroschisis usually occurs on the right side of the umbilicus, although it may occur on the left.

Hirschsprung Disease (Congenital Megacolon)

Hirschsprung disease – Absence of the parasympathetic ganglion nerve cells (aganglionosis) of the wall of the colon or rectum, which may result in congenital megacolon.

Hypoplastic Left Heart Syndrome (HLHS)

A condition in which the structures on the left side of the heart and the aorta are extremely small. Classically, this condition includes hypoplasia of the left ventricle, atresia or severe hypoplasia of the mitral and aortic valves, and hypoplasia and coarctation of the aorta.

Omphalocele

A defect in the anterior abdominal wall in which the umbilical ring is widened, allowing herniation of abdominal organs, including the small intestine, part of the large intestine, and occasionally the liver and spleen, into the umbilical cord. The herniating organs are covered by a nearly transparent membranous sac.

Patau Syndrome (Trisomy 13)

The presence of three copies of all or a large part of chromosome 13.

Pulmonary Valve Atresia and Stenosis

Pulmonary valve atresia – Lack of patency, or failure of formation altogether, of the pulmonary valve, resulting in obstruction of blood flow from the right ventricle to the pulmonary artery.

Pulmonary valve stenosis – Obstruction or narrowing of the pulmonary valve, which may impair blood flow from the right ventricle to the pulmonary artery.

Reduction Deformity, Lower Limbs

Complete or partial absence of the upper leg (femur), lower leg (tibia and/or fibula), ankle (tarsals), foot (metatarsals), or toes (phalanges).

Reduction Deformity, Upper Limbs

Complete or partial absence of the upper arm (humerus), lower arm (radius and/or ulna), wrist (carpals), hand (metacarpals), or fingers (phalanges).

Spina Bifida without Anencephalus

Incomplete closure of the vertebral spine (usually posteriorly) through which spinal cord tissue and/or the membranes covering the spine (meninges) herniate.

Tetralogy of Fallot

The simultaneous presence of a ventricular septal defect (VSD), pulmonic stenosis, a malpositioned aorta that overrides the ventricular septum, and right ventricular hypertrophy.

Transposition of the Great Arteries (TGA)

Transposition of the aorta and the pulmonary artery such that the aorta arises from the right ventricle (instead of the left) and the pulmonary artery arises from the left ventricle (instead of the right).

APPENDIX B
ICD-9 and CDC/BPA Codes Defining Conditions in the ABDMP Annual Report

The birth defect categories analyzed in this report are listed below, along with the code ranges that define each category. The center column shows the World Health Organization's International Classification of Disease, 9th Revision, Clinical Modification (ICD-9-CM) (1979) code ranges for each category. The right column specifies the British Pediatric Association Classification of Diseases (BPA) (1979) code ranges for the same conditions.

Condition	ICD-9 codes	BPA codes
Anencephaly	740.0 – 740.1	740.00 – 740.10
Amniotic bands	No code	658.8
Aniridia	743.45	743.42
Anophthalmia/microphthalmia	743.0, 743.1	743.00 – 743.10
Anotia/microtia	744.01, 744.23	744.01, 744.21
Aortic valve stenosis	746.3	746.3
Biliary atresia	751.61	751.65
Bladder exstrophy	753.5	753.5
Choanal atresia	748	748
Cleft lip with and without cleft palate	749.1, 749.2	749.10 – 749.29
Cleft palate without cleft lip	749	749.00 – 749.09
Coarctation of aorta	747.1	747.10 – 747.19
Common truncus	745	745.00 – 745.01
Congenital cataract	743.30 – 743.34	743.32 – 743.326
Diaphragmatic hernia	756.6	756.610 – 756.617
Down syndrome (Trisomy 21)	758	758.00 – 758.09
Ebstein anomaly	746.2	746.2
Edwards syndrome (Trisomy 18)	758.2	758.20 – 758.290
Encephalocele	742	742.00 – 742.09
Esophageal atresia/tracheoesophageal fistula	750.3	750.30 – 750.35
Fetus/newborn affected by maternal alcohol use	760.71	760.71
Gastroschisis	756.79	756.71
Hirschsprung disease	751.3	751.30 – 751.34
Hypoplastic left heart syndrome	746.7	746.7
Omphalocele	756.79	756.7
Patau syndrome (Trisomy 13)	758.1	758.10 – 758.19
Pulmonary valve atresia and stenosis	746.01, 746.02	746.00 – 746.01
Reduction deformity, lower limbs	755.30 – 755.39	755.30 – 755.39
Reduction deformity, upper limbs	755.20 – 755.29	755.20 – 755.29
Spina bifida without anencephalus	741.0, 741.9 (except 740.0 - 740.10)	741.00 – 741.99 (except 740.0 – 740.10)
Tetralogy of Fallot	745.2	745.20 – 745.21, 746.84
Transposition of great arteries	745.10, 745.11, 745.12, 745.19	745.10 – 745.19

APPENDIX C
Precision of Diagnosis Codes

Often health care professionals qualify a diagnosis, using words to express their level of confidence that the particular diagnosis explains what has been observed when examining, testing, or performing a procedure on a patient. If a professional makes a diagnosis using a qualifying term, the ABDMP assigns that diagnosis a “precision code,” based on the table below. Higher code numbers indicate higher “levels of precision.” Generally, if a diagnosis is made several times with different levels of precision, the diagnosis is assigned the precision code consistent with the most certain diagnosis.

Precision Code Qualifying Terms

1	not stated
2	probably not
3	vs, or
4	rule out, equivocal, questionable, r/o, uncertain, concern for, doubtful
5	suggestive of
6	suspected, suspicious
7	possible, may have, could be, felt to be, perhaps, consider, may be, question of, question
8	consistent with, most likely
9	compatible with, like, appears, evidence of, seems, apparent, believe
10	probable, presume
11	(code not currently used)
12	precise diagnosis, characteristic of (even if qualified with "mild," "somewhat," "relatively," or "borderline")

APPENDIX D
Exclusion List
Non-reportable Birth Defect Cases

The following potential cases are not included in the ABDMP Report:

- “Possibles” abstracted for review and consideration and subsequently determined to have conditions or defects that were not reportable according to the ABDMP lists of “excludable conditions.”
- Babies born to mothers whose residences are out-of-state or out-of-country (i.e., nonresident cases).
- “Negatives,” that is, conditions that were ruled-out during case-finding and medical record review.
- “No match” cases. A birth certificate was not on file and the state of birth could not be confirmed as Arizona.
- Cases among aborted fetuses less than 20 weeks gestation and weighing less than 500 grams. These cases were excluded because there was no reliable denominator that could be used to generate a birth defect rate.
- Prenatally diagnosed cases that did not result in a known live birth or stillbirth are not included. The ABDMP is not currently visiting prenatal diagnostic centers to identify cases.
- Defects with a “precision of diagnosis” code 1-7 are excluded. Only those defects diagnosed at the higher levels of precision (8 or above) are included. Refer to Appendix B for list of precision of diagnosis codes.
- Cases among aborted fetuses less than 20 weeks gestation and weighing less than 500 grams. These cases were excluded because there was no reliable denominator that could be used to generate a birth defect rate.

APPENDIX E
Race and Ethnicity

The Arizona Birth Defects Monitoring Program does not collect race and ethnicity data directly from hospital or clinic records. However, all case files are matched with a birth or fetal death certificate prior to being included in the dataset. For statistical purposes, the ABDMP defines the race and ethnicity of the child as equivalent to the mother’s race and ethnicity as recorded on the child’s birth or fetal death certificate. In this report race and ethnicity combinations are classified the same as in other Arizona Department of Health Services publications, as follows:

**Classification of Race/Ethnicity Combinations in Arizona
Statistical Reports**

	Hispanic	Non-Hispanic
White	Hispanic	White
Black	Black	Black
Native American	Native American	Native American
Asian	Asian	Asian
Other	Other	Other

- “White” refers to White non-Hispanics.
- “Hispanic” refers to White Hispanics.
- “Black” refers to African Americans, whether or not they are also Hispanic.
- “Native American” includes people of all Native American tribes, Aleuts, and Eskimos, whether or not they are living on reservation land and whether or not they are Hispanic
- “Asian” refers to people who classify themselves as Asian, Chinese, Japanese, Hawaiian, Filipino, or other Asian or Pacific Islander descent.
- “Other” includes persons who are unclassified or did not provide a response to race question on the certificate.

NOTE: Arizona Birth Defects Monitoring Program data provided to the Centers for Disease Control and Prevention (CDC) and which may be published in other documents are analyzed using different classifications for race and ethnicity combinations, as follows.

**Classification of Race/Ethnicity Combinations in National
Statistical Reports Related to Birth Defects**

	Hispanic	Non-Hispanic
White	Hispanic	White
Black	Hispanic	Black
Native American	Hispanic	Native American
Other	Other	Other

APPENDIX F

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