

Definition and Types

Down syndrome is a genetic disease resulting from chromosomal abnormality in which an individual has all or part of an extra copy of chromosome 21¹. There are three types of Down syndrome caused by three types of abnormal cell division mechanisms involving chromosome 21: Trisomy 21, Mosaicism and Robersonian Translocation.

- Trisomy 21- accounts for 90% cases of Down syndrome and occurs when an extra full copy of chromosome 21 presents in every cells.
- Mosaicism - accounts for less 2% of Down syndrome cases and the extra copy of chromosome 21 presents in some, but not all, cells of the individual.
- Robersonian Translocation - accounts for about 3-4% cases of Down syndrome and is due to translocation of chromosome 21 to another chromosome (usually the chromosome 14) before or at conception. Thus, individuals with Robersonian translocation have the usual two copies of chromosome 21, they also have additional material from chromosome 21. stuck to the translocated chromosome².

Some common physical features of Down syndrome include a flat face with an upward slant to the eye, a short neck, small ears, and a large tongue; tiny white spots on the iris of the eye; small hands and feet; a single crease across the palm of the hand; small pinky fingers that sometimes curve toward the thumb; poor muscle tone or loose ligaments³.

Babies and adults with Down syndrome can have IQ range from mild to moderate intellectual disability. They might also have physical problems such as congenital heart diseases, stomach problems, celiac disease, dementia, hearing problems, skeleton problems, eye problems, and thyroid problems¹.

United States Estimates

CDC estimates that about 6,000 babies in the United States are born with Down syndrome each year. The frequency in the United States is about 1 case in 691 live births⁵. The risk of having a child with Down syndrome increases with maternal age, especially among mothers older than 35 years of age. Also the death rate for the infants with Down syndrome among the African-American population seems to be higher than death rates among the White (non-Hispanic) population.

ABDMP Data Collection

The ABDMP staff review hospital discharge records, birth, and death certificates in order to identify potential cases with Down syndrome. An abstraction list serves to identify each Down syndrome case. After potential cases are identified, the medical records are reviewed to confirm that the child is one year old or younger and has Down syndrome. Once the case report is completed, it will be entered into the Arizona Birth Defects Registry⁴.

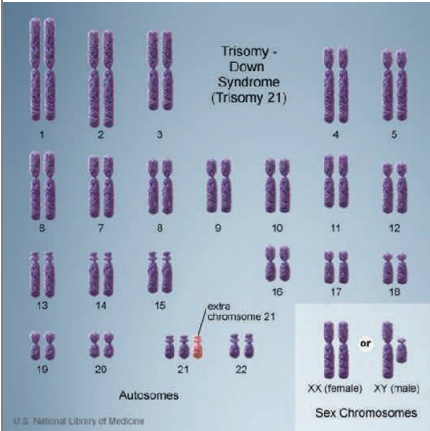
Down Syndrome in Arizona

The average incident rate of Down syndrome in Arizona is between 1995 and 2007* was 12.87 (95% CI: 8.96-16.77) per 10,000 live births for all race/ethnicity. The rates remained relatively constant over time with a slight decline from 1998 to 2000 (Fig. 1). The exact cause of decline of the incident rate of Down syndrome is not known. It is likely that folic acid fortification might have had an impact.

The incident rate of Down syndrome from 1995 to 2007* fluctuated immensely among the Native American population (Fig.2) as well as the African-American population (Fig.3). The average incident rate of Down syndrome for Native American is 14.32 (95% CI: -1.83-30.47) and 13.33 (95% CI: -2.13-28.79) for African-American. The incident rate of Down syndrome for the White (non-Hispanic) population and the Hispanic population are relatively constant with average rates of 11.9 (95% CI: 8.41-15.38) (Fig. 4) and 13.67 (95% CI: 8.24-19.1) (Fig.5), respectively. Statistically, there is no significant difference between the incident rates among all race/ethnicity groups (Fig.6).

Down Syndrome in Arizona

Figure 1 illustrates The incident rate of Down syndrome for all races/ethnicity in Arizona between 1995 and 2007*.



An illustration of the genetic basis of Down syndrome (trisomy 21)

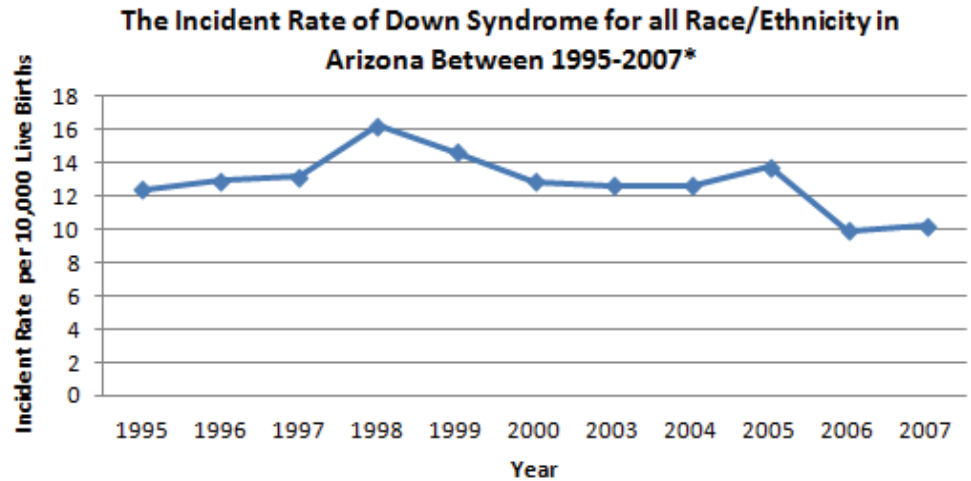
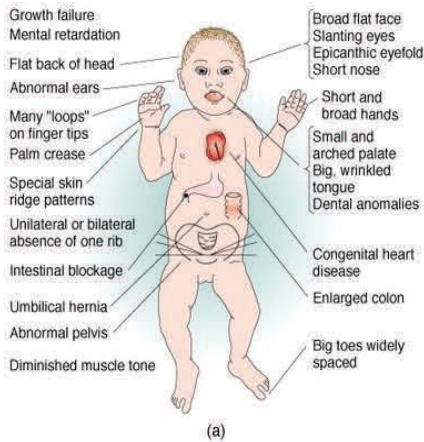
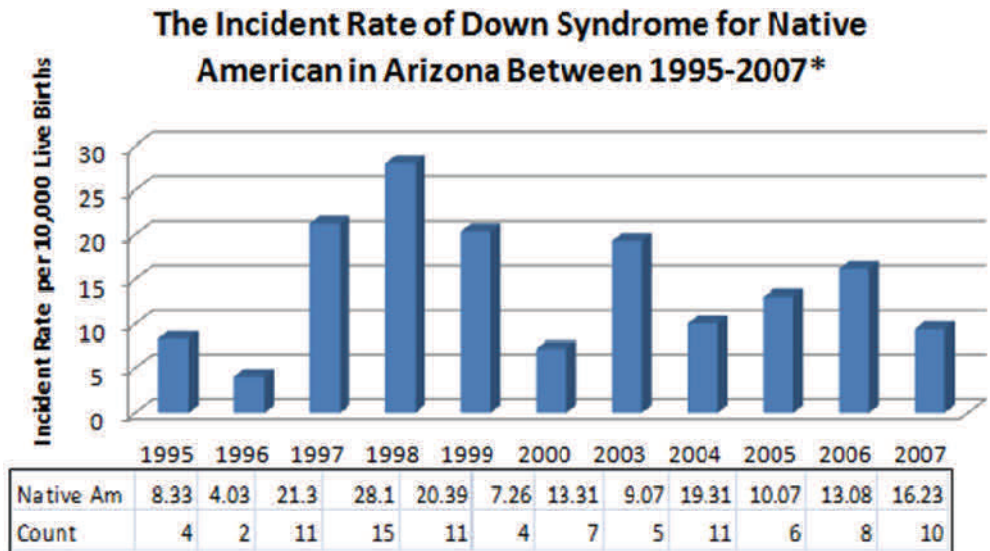


Figure 2: The average incident rate of Down syndrome in the Native American population between 1995 and 2007* is 14.32 cases per 10,000 live births.



Down syndrome associates with malformations of organs and systems in 45% of patients¹³.



* 2000 and 2001 data were omitted due to incompleteness



Down Syndrome in Arizona

Figure 3: The average incident rate of Down syndrome in the African-American population between 1995 and 2007* is 13.33 cases per 10,000 live births.



Infants exhibiting the symptoms of Down syndrome such as flattened nose and face, upward slanting eyes, open mouth with tendency of tongue protrusion, small ear with overfolded helix, single palmer crease, short fifth finger that curves inward, and widely separated first and second toes and increased skin crease³.

The Incident Rate of Down Syndrome for African-American in Arizona Between 1995-2007*

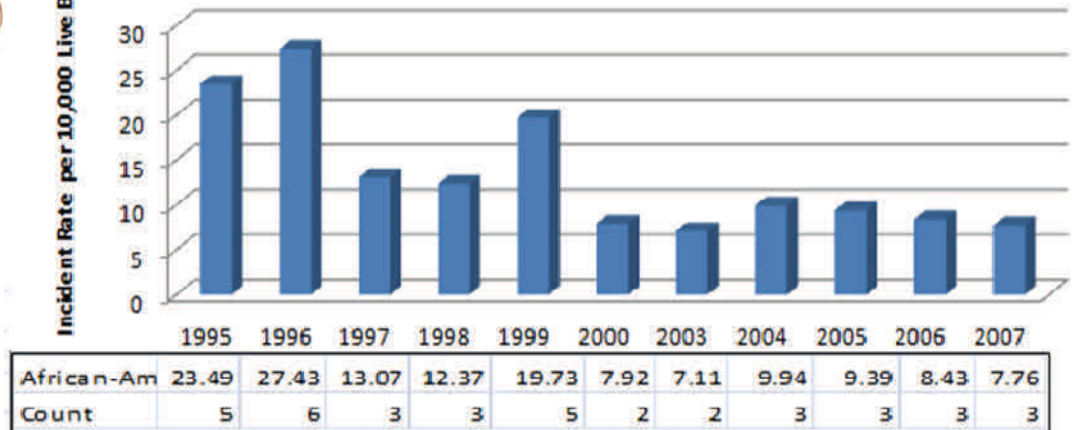
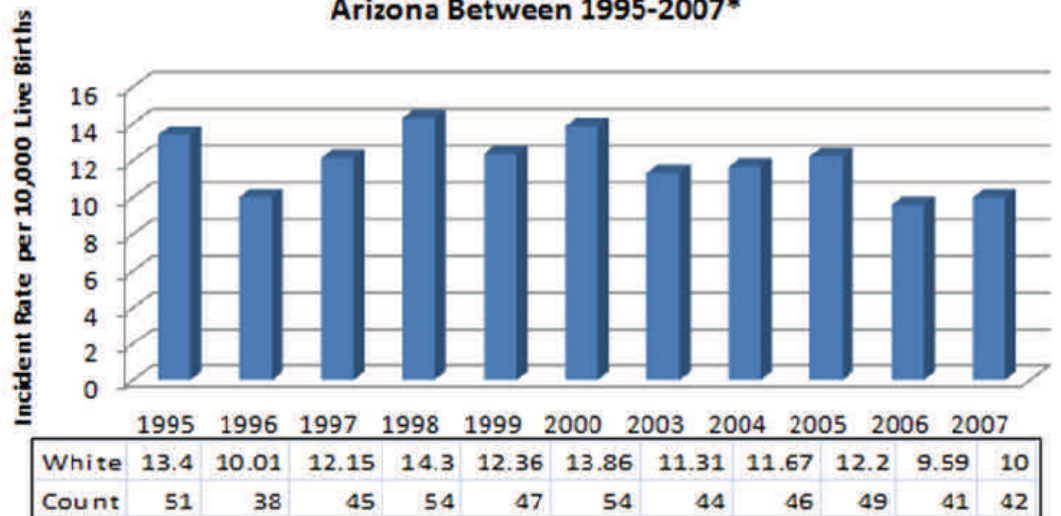


Figure 4: The average incident rate of Down syndrome in the White (non-Hispanic) population between 1995 and 2007* is 11.9 cases per 10,000 live births.

The Incident Rate of Down Syndrome for White (non-Hispanic) in Arizona Between 1995-2007*



* 2000 and 2001 data were omitted due to incompleteness



Down Syndrome in Arizona

Figure 5: The average incident rate of Down syndrome in the Hispanic population between 1995 and 2007* is 13.67 cases per 10,000 live births.



Kids with Down syndrome.

The Incident Rate of Down Syndrome for Hispanic in Arizona Between 1995-2007*

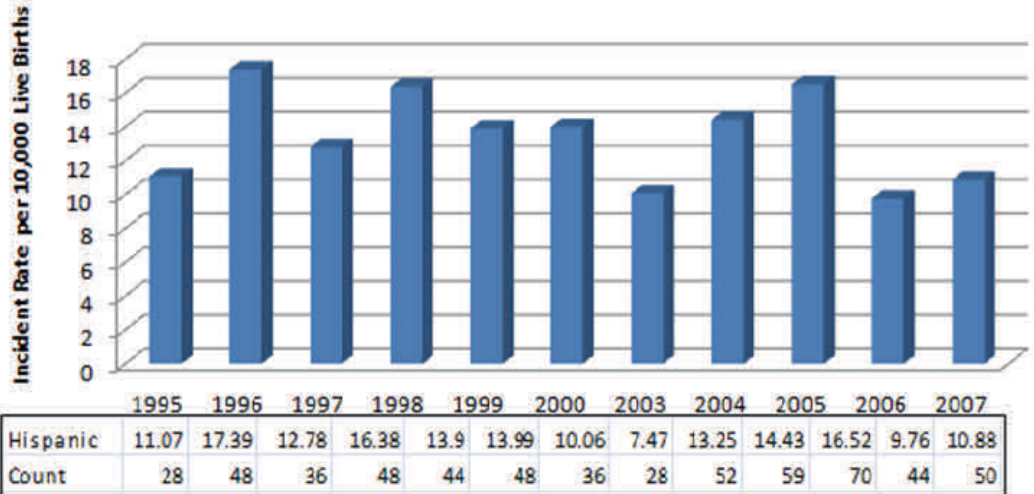
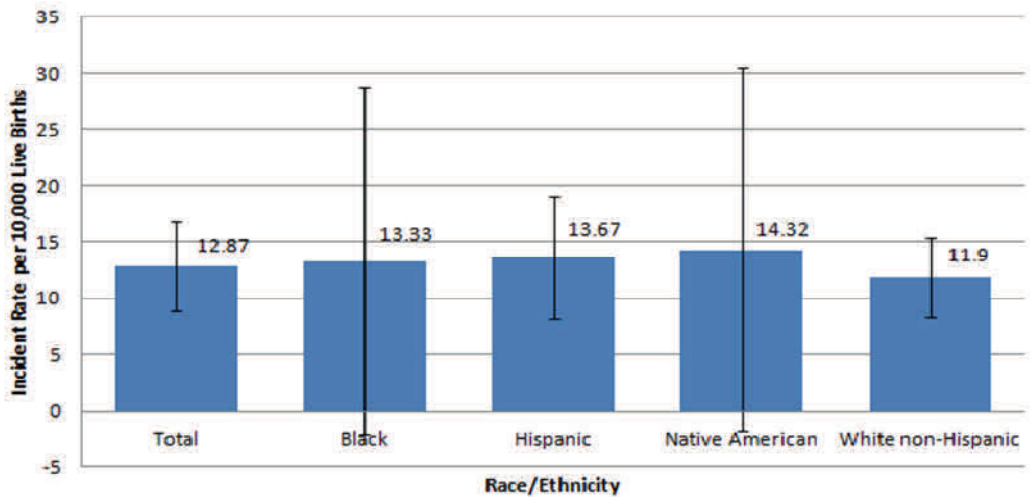


Figure 6: The incident rate of Down syndrome for selected race/ethnicity between 1995 and 2007*.

The Incident Rate of Down Syndrome for Selected Race/Ethnicity in Arizona Between 1995-2007*



* 2000 and 2001 data were omitted due to incompleteness

Prevention

The occurrence of Down syndrome is due to a random event that occurs during formation of the reproductive cells, the egg, the sperm or the embryo. The age of the mother is the only factor that has been shown to increase the risk of having a baby with Down syndrome. This risk increases with every year, especially after the mother is 35 years old. By the age of 35, the risk for a woman having a baby with Down syndrome is about 1 in 400 whereas the risk is 1 in 2000 for a mother of 20 years of age. The risk increases to 1 in 35 when the mother is 45 years old. When the mother already has a baby with Down syndrome, there is a 1% chance of having another baby with Down syndrome. Also, when one of the parents is a balance carrier of translocation, the risk of having a baby with Down syndrome is greatly increased. A balance carrier has some rearranged genetic material but no extra genetic material, hence has no signs or symptoms of Down syndrome ^{7,8}.

There is no known way to prevent Down syndrome. However, a study in 1999 indicated that impairments in folate/homocysteine metabolism could increase the risk of having an infant with Down syndrome, suggesting a potential link between folic acid and Down syndrome ⁹. Folic acid is essentially a water-soluble vitamin B that contributes to cell division and growth; therefore it is particularly important for women during their pregnancy. The Centers for Disease Control and Prevention (CDC) and other organizations recommend that all women of child bearing age take 400 micrograms of folic acid 1-3 months prior to pregnancy and keep taking it throughout the pregnancy ¹⁰.

*Folic acid might
have some
impact on
preventing Down
syndrome*



Dietary folate and folic acid can ensure the health of mother and child ¹⁰.

Screening

Screening for Down syndrome is offered as a routine prenatal care. In fact, the American Congress of Obstetricians and Gynecologists recommends offering various screening tests for Down syndrome to all pregnant women, regardless of age. Some of those tests are carried out in the first trimester and some in the second trimester.

First trimester screening includes a blood test and a special ultrasound scan called nuchal translucency (NT) test. The blood test measures the levels of two proteins in the maternal serum: free Beta-hCG and PAPP-A. This test is done between 9 weeks and 13 weeks. The NT test measures the translucent (clear) space in the tissue at the back of the baby. The NT test can only be done between 11 and 13 weeks. The accuracy of the first trimester screening range from 79% to 90% ¹¹. The first trimester screening provides information about the risk for the chromosomal abnormalities of the baby in the early stage of pregnancy.

Second trimester screening involves a blood test measures the levels of four substances in the maternal serum: AFP, hCG, uE3 and inhibin A. This test could detect about 80% of babies with Down syndrome ¹¹.

The combination of the first and second trimester screening is called integrated screening or sequential screening. The integrated screening can detect 94-96% of babies with Down syndrome; therefore it is the most accurate noninvasive screening test currently available ¹².

Diagnostic Tests

Although screening tests offer some certainty about the risk of having a baby with Down syndrome, they are not confirmative tests and have a high rate of false positive and false negative.

The diagnostic tests include amniocentesis, chorionic villus sampling (CVS), and percutaneous umbilical blood sampling (PUBS), which could identify babies with more than 99% accuracy¹². Unlike the screening tests, the diagnostic tests are invasive and pose some risk of miscarriage.

Amniocentesis is a test that a sample of amniotic fluid surrounding the baby is withdrawn through a needle inserted into the uterus. This sample is used to analyze the chromosome of the baby. This test is usually done in the fifteenth week of pregnancy and carries a 1 in 200 risk of miscarriage.

Chorionic villus sampling (CVS) is a test that takes the cell sample from the mother's placenta and uses the sample to analyze the baby's chromosome. This test is typically performed between ninth and fourteenth week of pregnancy and carries a 1 in 100 risk of miscarriage.

Percutaneous umbilical blood sampling (PUBS) is a blood test in which the blood is taken from a vein in the umbilical cord and tested for chromosomal abnormalities. This test is usually done at the eighteenth week of pregnancy and carries higher risk of miscarriage than amniocentesis and CVS. It is only performed when the result from other tests are not clear.

Coping and Support

There are no cures for Down syndrome. Hence the treatment for the condition focus on controlling symptoms and any medical conditions that result from Down syndrome. Moreover, counseling and support that also recognized as important interventions in treating individuals affected with Down syndrome. Although treatments and therapies are for the physical, medical and cognitive problems associated with Down syndrome, the following resources are starting places for parents and families of children affected with Down syndrome:



- **Sharing Down syndrome Arizona (<http://sharingds.org/>)-** a 501(C) (3) organization that tries to integrate all individuals who are affected by Down syndrome by advocating positive acceptance and inclusion.
- **National Association for Down syndrome (<http://www.nads.org/>)-** gives educational resources and counseling and support for parents of infants newly-diagnosed with Down syndrome.
- **Down Syndrome Network Arizona (<http://dsnetworkaz.org/>)-** an organization that tries to educate, support and advocate for those in our community impacted by Down syndrome.
- **National Down Syndrome Society (<http://www.ndss.org/>)-** offers research news and searchable resource database and message boards to advance research on Down syndrome.
- **Parent to Parent link (<http://www.parenttoparent.org/>)-** a network of parents of children with special needs can be matched for support and sharing experiences.
- **The Down Syndrome Research Foundation (<http://dsrf.org/index.cfm?fuseaction=publications.dsq>)-** a foundation dedicated to the advancement of research on ways of increasing the cognitive and social development skills of children affected by Down syndrome.

- **Medical Home Portal:** <http://www.medicalhomeportal.org/diagnoses-and-conditions/down-syndrome/description> - offers more information about Down syndrome.



With appropriate intervention, some patients with Down syndrome can live normally: go to mainstream school, read and write, and have jobs.

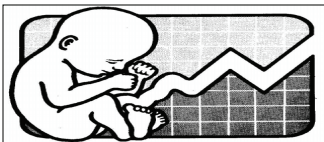
ABDMP Goals

The Arizona Birth Defects Monitoring Program (ABDMP) is a statewide, population-based, active surveillance program that collects and analyzes information on children with reportable birth defects diagnosed within the first year of life.

The goals of the ABDMP include :

- To reduce the incidence of birth defects in Arizona from preventable causes.
- To produce accurate statistics regarding the occurrence of birth defects in Arizona.
- To identify, report, and investigate various birth defects trends, high-risk populations, and high risk locations.
- To provide a resource for information about the incidence and epidemiology of birth defects for researchers, health professionals, hospitals, local health agencies, and others with a valid scientific or public health interest.³

Arizona Birth Defects Monitoring Program



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We are on the web!

[http://www.azdhs.gov/phs/
phstats/bdr/index.htm](http://www.azdhs.gov/phs/phstats/bdr/index.htm)



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