

House Bill 552 was signed into law in 2014 requiring that physicians, certified nurse-midwives, and genetic counselors provide a fact sheet on Down syndrome from the Ohio Department of Health to patients with a test result indicating Down syndrome, or a prenatal or postnatal diagnosis of Down syndrome.

OVERVIEW OF DOWN SYNDROME

Down syndrome is a genetic condition that is usually caused by an extra copy of the twenty-first chromosome. Most people have two copies of each chromosome, for a total of 46. People with Down syndrome have 3 copies of the 21st chromosome, making a total of 47. Down syndrome is also called Trisomy 21. According to current data, about 250,000¹ people in the United States have Down syndrome. Studies show that about one in 830 babies are born with Down syndrome², and the chance of having a baby with the genetic condition increases with the age of the expectant mother. Down syndrome does not typically run in families and is not caused by anything either parent did or did not do. Some people can have “Translocation Down syndrome” or “Mosaic Down syndrome,” which are rare genetic variations of Down syndrome. If you have questions about the types of Down syndrome, ask your doctor or genetic counselor for more information.

Advances in medical care and research over the years have given people with Down syndrome better overall health. The traits, medical conditions, and abilities of people with Down syndrome vary widely and cannot be predicted before they are born. They generally have mild to moderate cognitive delays, low muscle tone, and higher chances for a variety of other health issues during their lifespan. Because of advances in health care, education, and public attitudes, the outlook for people with Down syndrome has improved significantly during the past few decades.

UNDERSTANDING SCREENING AND TESTING

The availability, types of prenatal screening and testing change rapidly. Currently, there are several screening and diagnostic testing options that are available. One option is to use either hormone levels in the blood and/or ultrasound markers to find out whether or not a pregnancy is at a higher chance of Down syndrome. This can be done in either the first- or second-trimester and these screenings adjust for the mother’s age-related risk of having a child with Down syndrome. For more information about prenatal screening and testing, please go to www.lettercase.org/prenataltesting.

One method of screening for Down syndrome and some other chromosome conditions is “non-invasive” prenatal screening, which uses cell-free fetal DNA. These are pieces of DNA from the pregnancy that are found in the mother’s bloodstream. By taking a sample of the mother’s blood, this test looks at the amount of DNA from the mother and the baby’s placenta for

chromosomes 13, 18 and 21 to see if there is the right amount. The results of this test can show if the baby is at a higher risk for trisomy 13, trisomy 18, or Down syndrome. Some labs also look at the amount of X and Y chromosomes and can detect extra or missing copies of these chromosomes.

Ultrasound can also provide information about whether a baby may have a higher chance of having Down syndrome. A detailed ultrasound at a high-risk hospital at 18 to 20 weeks of pregnancy looks closely at the baby to see if there any signs that can be seen more often in babies with Down syndrome. Half of babies who have Down syndrome will show one of these signs.³ **All of these tests are screening only. They do not pick up all cases of Down syndrome and cannot tell for sure whether a baby has Down syndrome.** If one of these screening tests shows a chance for Down syndrome, diagnostic testing should be offered to find out if the baby has Down syndrome if the parents want a more definitive answer.

Prenatal diagnosis is available through either a chorionic villus sampling (CVS) or amniocentesis. The CVS is done at approximately 10 to 12 weeks gestation and involves sampling a tiny piece of the placenta and performing chromosomes studies. The amniocentesis is done at approximately 16 to 18 weeks gestation and involves taking a sample of the amniotic fluid surrounding a baby. Chromosome studies are performed on the cells floating in the fluid. **These are the most accurate indicators of Down syndrome.** However, both of these tests have risks for complication that could potentially lead to miscarriage. The risk with the CVS is approximately 1/100 (1 percent) and the risk with the amniocentesis is less than 1/300 (less than 0.3 percent).

UNDERSTANDING DOWN SYNDROME⁴

- Children with Down syndrome are more similar to other children than they are different.
- Individuals with Down syndrome have a variable range of intellectual disability from mild to moderate (not typically severe).
- Babies with Down syndrome usually have developmental delays and benefit from early intervention, including physical, occupational, and speech therapy, to help them meet their milestones.
- 80 percent of babies with this condition have hypotonia or low muscle tone at birth. This usually improves with time, and physical therapy can help.
- 50 percent of babies with Down syndrome will have one or more health issues: 40 to 60 percent of babies with Down syndrome have a heart condition and 12 percent have a gastrointestinal condition that can be treated with surgery. Referrals to specialists are appropriate for identified complications.
- Babies with Down syndrome also have higher chances for feeding and digestive issues, hearing loss, vision impairment, and respiratory infections. Most of these conditions can be treated with good health care.

- Currently, the average life expectancy for people with Down syndrome is about 60 years.⁵
- Raising a child with Down syndrome may involve more time and commitment than raising one without.
- Breastfeeding is encouraged because breast milk helps protect the baby from some illness. Some babies with Down syndrome may have a weak and uncoordinated suck, so feeding can be harder and requires patience. Advice from a lactation expert may be helpful.
- Immunizations and regular health care are extremely important.
- Children with Down syndrome have higher chances of being obese; promoting weight management with an active and healthy lifestyle is important.

GETTING MORE SUPPORT

Patients can learn more from genetic counselors who can offer information about the genetics of Down syndrome, and local resources, such as local support organizations and connections to other families, recurrence chances for future pregnancies, testing options and results, the accuracy and limitations of different prenatal screens and tests, and reproductive options. This link will take you to the genetics centers in Ohio:

<http://www.odh.ohio.gov/odhprograms/cmh/genserv/genserv1.aspx>

Families can learn more about early intervention by contacting their county's *Help Me Grow* site at 1-800-755-GROW (4769) or visiting the *Help Me Grow* website at <http://www.helpmegrow.ohio.gov/>.

As a result of improved public attitudes and acceptance, many people with Down syndrome are thriving as active and valued members of the community. This includes children who are involved in social and school programs with their peers and many adults who are employed and live independently or with some support. Research shows that the majority of adults with Down syndrome report that they are happy with their lives.⁶

LOCAL/STATE/NATIONAL ORGANIZATIONS

- Down Syndrome Association of Central Ohio - www.dsaco.net/
- Down Syndrome Association of Greater Cincinnati - www.dsagc.com/
- Down Syndrome Association of Greater Toledo - www.dsagt.org/
- Miami Valley Down Syndrome Association (Dayton) - www.mvdsa.org
- Down Syndrome of the Valley (DSAV) - www.dsav.org
- National Down Syndrome Adoption Network - www.ndsan.org/
- The Up Side of Downs of Northeast Ohio - www.theupsideofdowns.org/

- Kids Health - <http://kidshealth.org/>
- National Center for Prenatal and Postnatal Down Syndrome Resources — <http://Downsyndromediagnosis.org> *A website with a resource directory for new and expectant parents learning about Down syndrome.*
- Center for Disease Control and Prevention (CDC), “Facts about Down Syndrome” — www.cdc.gov *A website with facts about Down syndrome.*
- American Academy of Pediatrics, “Health Care Information for Families of Children with Down Syndrome” — <http://healthychildren.org> *Guidelines focused on recommended healthcare and treatment.*
- National Down Syndrome Society — <http://www.ndss.org>
- National Down Syndrome Congress — <http://ndsccenter.org>
- International Mosaic Down Syndrome Association — <http://imdsa.org/>
- Down Syndrome Education USA — www.dseusa.org

PROFESSIONAL GENETICS ORGANIZATIONS

National Society of Genetic Counselors — <http://nsgc.org>

American College of Genetics and Genomics — <http://acmg.net>

NATIONAL DISABILITY ORGANIZATIONS

American Association Of People With Disabilities — <http://aapd.com>

ARC — <http://thearc.org>

Association of University Centers on Disability — <http://aucd.org>

March of Dimes — www.marchofdimes.org

Special Olympics International — <http://specialolympics.org>

*Some information used in this fact sheet was provided by the National Society of Genetic Counselors (NSGC)

1 Presson, A.P., Partyka, G., Jensen, K.M., Devine, O.J., Rasmussen, S.A., McCabe, L.L., McCabe, E.R.B. (2013). Current estimate of Down syndrome population prevalence in the United States. *J Pediatr*, 163(4):1163-1168.

2 Shin M., Besser L.M., Kucik J.E., Lu C., Siffle C., Correa A., et al. (2009) Prevalence of Down syndrome among children and adolescents in 10 regions of the United States. *Pediatrics* 124:1565-1571. (Indicative of the live birth numbers from data reported around 2003.)

3 <http://www.cdc.gov/ncbddd/birthdefects/DownSyndrome.html> and http://www.fetal.com/Genetic%20Sono/03_gen%20us%20color%20dopp.html

4 Sheets KB et al. 2011. Practice guidelines for communicating a prenatal or postnatal diagnosis of Down syndrome: recommendations of the national society of genetic counselors. *J Genet Couns*. 20(5):432-41.

5 Glasson, E.J., Sullivan, S.G., Petterson, B.A., Montgomery, P.D., Bittles, A.H. (2002). The changing survival profile of people with Down syndrome: implications for genetic counselling. *Clinical Genetics*, 62:390-393.

6 Skotko, B.G., Levine, S.P., Goldstein, R. (2011). Self-perceptions from People with Down Syndrome. *American Journal of Medical Genetics*, Part A: 155:2360-2369. See more research at brianskotko.com