



Prenatal Testing & Information

ABOUT DOWN SYNDROME

Helpful Resources

Every woman and every pregnancy is different. Pediatricians, genetic counselors, family members, friends, spiritual advisers and others can assist a pregnant woman who receives a prenatal diagnosis of Down syndrome.

EARLY INTERVENTION, EDUCATIONAL AND EMOTIONAL SUPPORTS

Woodbine House Books on Down Syndrome
www.woodbinehouse.com/product-category/down-syndrome/

MEDICAL CARE

American Academy of Pediatrics, “Health Supervision for Children with Down Syndrome”: www.publications.aap.org/pediatrics/article/128/2/393/30609/Health-Supervision-for-Children-With-Down-Syndrome

Anna & John J. Sie Center for Down Syndrome, “Pediatric Guideline Record Tool”: www.globaldownsyndrome.org/pediatrics-record-sheet/

NEW & EXPECTANT PARENTS

- www.globaldownsyndrome.org/prenatal-testing-pamphlet/
- www.ndsccenter.org/programs-resources/as/birthto2/
- www.ndss.org/lifespan/diagnosis
- www.downsyndromepregnancy.org
- Babies with Down Syndrome: A New Parents’ Guide (book is available in English and Spanish)
- The Parent’s Guide to Down Syndrome: Advice, Information, Inspiration, and Support for Raising Your Child from Diagnosis through Adulthood

IF YOU HAVE QUESTIONS ABOUT YOUR PREGNANCY OR DOWN SYNDROME VISIT US AT WWW.DOWNSYNDROMETEST.ORG.

Introduction

Pregnancy can be an exciting time...and one that can produce emotions and many questions. Will my baby be a boy or a girl? Who will he or she look like the most? Is my baby healthy? To help answer these questions, your doctor or healthcare provider may offer you a variety of testing options during your pregnancy.



Answering Your Questions

Should I have testing?

THE DECISION WHETHER TO HAVE a prenatal screening test or diagnostic test is a very personal one. Some women access these tests in order to gather more information about the chance for Down syndrome in their pregnancy, and/or so they can plan for the delivery of a child with Down syndrome. Others are comfortable proceeding without prenatal screening or diagnostic testing and decide that such tests are not necessary or not worth the risks. All of these tests are completely optional. If you have questions about any of these testing options, talk with your doctor, genetic counselor, maternal fetal specialist or other healthcare provider about the risks, benefits and limitations of these tests.



What is a “prenatal test” for Down syndrome?

GENERALLY THERE ARE TWO types of tests (screening tests and diagnostic tests) that you can have while you are pregnant to help determine if your baby has Down syndrome or another chromosome condition. Screening tests tell you the *chance* that the baby could have a chromosome condition, while diagnostic tests can tell you for sure.

SCREENING TESTS

Prenatal screening tests usually involve blood and/or ultrasound exams. There are two main types of screening tests that are based on measurements from 1-2 tubes of your blood - one measures proteins in the serum and the other measures small fragments of genetic material (DNA).

Only a diagnostic test will tell you for sure if your baby has Down syndrome

The serum test is also referred to as **maternal serum screening, biochemical screening, first trimester combined screening, the triple test and/or quad screening**. This blood test is often combined with results of a special ultrasound examination referred to as the “nuchal translucency” (or NT) measurement, which measures the amount of fluid at the back of the baby’s neck during early pregnancy (between 11 and 14 weeks). Maternal serum screening tests can be done in the late first trimester (between 10 and 14 weeks) or in the second trimester (between 15 and 20 weeks). The test gives you a risk number (such as “1 in 100”) for having a baby with Down syndrome or other chromosome conditions.

A newer test is based on counting fragments of DNA that float in your blood. This is called cell free DNA (cfDNA) screening, noninvasive prenatal testing (NIPT) or noninvasive prenatal screening (NIPS). cfDNA can be done any time in the pregnancy after 10 weeks of pregnancy up until delivery.

If your baby has Down syndrome, there will be a higher number of fragments from chromosome 21 in your blood sample. This test is very accurate in detecting the possibility that your baby has Down

syndrome, although all “positive”

tests should be confirmed by a

diagnostic test to be sure (see page

6). This test can also tell if the baby

is a boy or girl.

At the present time, the American

College of Obstetricians and

Gynecologists and other

professional societies recommend

that cfDNA screening tests can be

High risk factors include:

- Advanced maternal age (35 years of age or older)
- One or more relatives with Down syndrome
- Results from a different test that indicate a higher chance of having a baby with Down syndrome

offered to pregnant women who are at “high risk” for having a baby with Down syndrome. “High risk” includes women who are over the age of 35 at the time of their delivery, women whose babies have an abnormal ultrasound test, women who have an abnormal serum protein test, and women who have had a previous baby or a close family member with Down syndrome. Conventional screening tests are still the most appropriate first step for women not considered “high risk”.¹



There are no risks of miscarriage associated with the screening tests. Screening tests cannot tell you for sure if your baby has Down syndrome or another chromosome condition. However, if a screening test shows that the chances are high, your obstetrician or genetic counselor will offer you a “diagnostic” test. Regardless of your risk, you can request a diagnostic test if you want to know for sure if your baby has a chromosome condition.

DIAGNOSTIC TESTS

There are two diagnostic tests that can tell you with almost 100% accuracy if your baby will have Down syndrome or another chromosome condition. The amniocentesis test (“amnio” for short) is usually performed after the 15th week of pregnancy. The chorionic villus sampling test (“CVS” for short) is usually performed between the 10th and 13th weeks of pregnancy. Both tests have a 0.1 - 0.3% risk of causing miscarriage.²

What is Down syndrome?

DOWN SYNDROME, also known as Trisomy 21, is a condition where a person is born with three copies of chromosome 21 instead of two. In the United States, 1 in every 691 babies is born with the condition.³ There are hundreds of thousands of people with Down syndrome in the United States, and an estimated six million people with Down syndrome worldwide.

Down syndrome has nothing to do with race, nationality, socioeconomic status, religion, or anything the mother or father did before or during pregnancy

Why some babies have three copies of chromosome 21 instead of two is largely unknown. We do know that 98% of the time, Down syndrome is random and not hereditary. The chance of having a baby with Down syndrome increases with the mother's age; however, about half of babies born with Down syndrome are born to mother's under 35 years old simply because more young women are having babies.⁴

It is important to note that Down syndrome has nothing to do with race, nationality, socioeconomic status, religion, or anything the mother or father did before or during pregnancy.



Are my concerns and feelings normal?

MANY PARENTS ARE CONCERNED about how a child with Down syndrome will affect their family, including siblings. Every family is unique and may deal with the idea of, or the birth of, a baby with Down syndrome differently. It is natural to feel sad or worried if a prenatal test indicates your baby has or may have Down syndrome. Having a baby with Down syndrome is not something parents typically plan for, and any emotion may be expected.

At the time of diagnosis, it may be difficult to envision positive family impacts, but studies that focus on the long-term outcomes for families have been encouraging. Despite potential challenges, personal accounts and studies in the U.S. show that most families that have a child with Down syndrome are stable, successful, and happy.

In addition, many siblings respond positively and feel they have more fulfilled lives because of their sibling with Down syndrome. Studies focused on personal satisfaction for people with Down syndrome indicate that most older children, teens, and adults surveyed feel they lead happy and fulfilling lives.⁵

IF YOU HAVE QUESTIONS ABOUT YOUR PREGNANCY OR
ABOUT DOWN SYNDROME, PLEASE VISIT US AT
WWW.DOWNSYNDROMETEST.ORG

How will Down syndrome affect my baby?

THERE IS NO WAY TO KNOW what the future holds for any baby. In many ways, babies with Down syndrome are just like other babies. All babies need to be fed, have their diapers changed, and have playtime, but, most of all, they need to be loved. These needs are the same for a baby with Down syndrome.

However, it is important for families to know the healthcare and educational needs of a child with Down syndrome can be different or potentially more challenging than for other children.



Living with Down syndrome in the U.S.

BELOW ARE SOME FACTS ABOUT PEOPLE WITH DOWN SYNDROME THAT MAY BE HELPFUL TO KNOW:

- Since the human and civil rights movements began in the 1960s, people with Down syndrome are increasingly recognized as an important part of society.
- People with Down syndrome participate in school, religious groups, sports teams, performing and visual arts programs, volunteer organizations, and the workplace.
- Most children with Down syndrome function in the mild to moderate range of intellectual disability.⁶
- By law, people with Down syndrome must be provided a free, appropriate education through their public school system.⁷
- A small but growing number of people with Down syndrome are participating in post-secondary education and college programs.
- Most people with Down syndrome need extra support throughout their lives, but a growing number of people with Down syndrome live independently or semi-independently.
- A small but growing number of people with Down syndrome are choosing to get married.



MEDICAL INFORMATION RELATED TO PEOPLE WITH DOWN SYNDROME

- Currently, in the U.S., the average lifespan of a person with Down syndrome is 60 years, compared with 25 years in 1983^{8,9} (however, there are disparities by race and ethnicity).
- It is important to follow the American Academy of Pediatrics “Health Supervision for Children with Down Syndrome.” These recommendations can strengthen the physical and intellectual abilities of a person with Down syndrome.
- Up to 50% of babies with Down syndrome are born with a heart defect, and some will require surgery.¹⁰ The vast majority of these heart defects are correctable.
- People with Down syndrome have an increased risk for medical conditions such as respiratory issues, hearing and vision problems, ear infections, sleep apnea, thyroid conditions, and Alzheimer’s disease. All of these conditions occur in individuals who do not have Down syndrome, and some people with Down syndrome do not have any of these conditions. Most of these conditions are treatable, and the majority of people with Down syndrome lead healthy lives.¹¹
- People with Down syndrome are more likely to develop certain childhood leukemias,¹² but much less likely to develop solid tumor cancers as compared to the general population.¹³ People with Down syndrome are also less likely to suffer from heart attacks or stroke.^{14,15}

What should I expect the first year?

BABIES WITH DOWN SYNDROME require the same medical care needed for all babies (routine well-care visits and immunizations). In addition, the American Academy of Pediatrics (AAP) has created guidelines for infants and children with Down syndrome called “Health Supervision for Children with Down Syndrome.” In the first year, some of the AAP’s recommendations include an echocardiogram for heart defects, a blood count, hearing and vision screening, a blood test for thyroid hormone levels, and an examination for gastrointestinal issues.

Developmentally, children with Down syndrome typically experience delays in different areas. Gross motor delays can affect sitting, crawling and walking. Fine motor delays involve hand and finger coordination. Speech delays may impact the onset and quality of speech and language.

Developmental milestones vary widely in children with Down syndrome and are impossible to predict. Early intervention services, including physical, occupational, and speech therapies, should be started early on in life to optimize a child’s development.

In the U.S., a service known as “early intervention” supports the development of infants and toddlers with special needs, especially in the first three years of life. Services are implemented in each state under the guidance of a federal law called Individuals with Disabilities Education Act (IDEA, part C).

SOME TYPICAL MILESTONES

ACTIVITY	CHILDREN WITH DOWN SYNDROME	TYPICALLY DEVELOPING CHILDREN
Smiling	1.5 - 3 months	0.5 - 3 months
Rolling over	2 - 12 months	2 - 10 months
Sitting	6 - 18 months	5 - 9 months
Crawling	8 - 25 months	7 - 13 months
Standing	10 - 32 months	8 - 16 months
Walking	12 - 45 months	8 - 18 months
Talking, words	9 - 30 months	6 - 14 months
Talking, sentences	18 - 46 months	14 - 32 months

© 2001 Sigfried Pueschel





RESOURCES ARE AVAILABLE FOR MEDICAL CARE AND EARLY INTERVENTION SERVICES

If you have private health insurance, your baby with Down syndrome will be covered just like your other children. There should no longer be any health insurance denials based on “pre-existing conditions.” Every state will also have a program to assist in obtaining health insurance for children. This may be through the State Children’s Health Insurance Program (SCHIP) or through Medicaid’s Early and Periodic Screening, Diagnostic and Treatment program.

Federal law requires each state to offer early intervention services for infants with disabilities or at risk for developmental delays. An evaluation or assessment is offered at no charge, but the cost for early intervention services will depend on the policies of each state. In some states, there is no charge; in others, services are provided on a sliding scale, depending on your income.

For more information, contact your local Down syndrome organization for more details www.globaldownsyndrome.org/about-down-syndrome/resources/local-organizations/ or contact the NDSC at 770-604-9500 or the NDSS at 800-221-4602.

Other Resources

In addition to non-profit organizations such as the Global Down Syndrome Foundation, the National Down Syndrome Congress, and the National Down Syndrome Society, there are an estimated 250 local Down syndrome organizations in the United States, so chances are there is one in your area. Below is a short list of resources pregnant women and families may find helpful. For more detailed information please visit us at www.downsyndrometest.org.

GLOBAL DOWN SYNDROME FOUNDATION

www.globaldownsyndrome.org

NATIONAL DOWN SYNDROME CONGRESS

www.ndscenter.org

NATIONAL DOWN SYNDROME SOCIETY

www.ndss.org

DOWN SYNDROME WORLD MAGAZINE

www.downsyndromeworld.org

ADOPTION

Families expecting a child with Down syndrome may decide that making an adoption plan is the right choice for their family. The National Down Syndrome Adoption Network has a registry of families who are licensed to adopt and who want to adopt a child with Down syndrome. For more information visit www.ndsan.org or call 513-709-1751.



Footnotes

1. American College of Obstetricians and Gynecologists. (2015). Committee opinion number 640: Cell free-DNA screening for fetal aneuploidy. *Obstetrics and Gynecology*, 126, e31-37.
2. American College of Obstetricians and Gynecologists. (2016). Practice bulletin number 162: Prenatal diagnostic testing for genetic disorders. *Obstetrics and Gynecology*, 127(5), e108-122.
3. Parker, S. E., Mai, C. T., Canfield, M. A., Rickard, R., Wang, Y., Meyer, R. E.,...Correa, A. (2010). Updated national birth prevalence estimates for selected birth defects in the United States, 2004-2006. *Birth Defects Research Part A Clinical and Molecular Teratology*, 88, 1008-1016. <http://doi:10.1002/bdra.20735>
4. Resta, R. G. (2005). Changing demographic of advanced maternal age (AMA) and the impact on the predicted incidence of Down syndrome in the United States: Implications for prenatal screening and genetic counseling. *American Journal of Medical Genetics*, 133A(1), 31-36. <http://doi:10.1002/ajmg.a.30553>
5. Skotko, B. G., Levine, S. P., & Goldstein, R. (2011). Self-perceptions from people with Down syndrome. *American Journal of Medical Genetics. Part A*, 0(10), 2360-2369. <http://doi.org/10.1002/ajmg.a.34235>
6. Bull, M. J., & the Committee on Genetics. (2011). Clinical report- Health supervision for children with Down syndrome. *Pediatrics*, 123(2), 393-402. <http://doi:10.1542/peds.2011-1605>
7. Individuals with Disabilities Education Improvement Act, 20 U.S.C § 1400 (2004).
8. Glasson, E. J., Sullivan, S. G., Hussain, R., Petterson, B. A., Montgomery, P. D., & Bittles, A. H. (2002). The changing survival profile of people with Down's syndrome: Implications for genetic counselling. *Clinical Genetics*, 62(5), 390-393.
9. Weijerman, M. E., & de Winter, J. P. (2010). Clinical practice: The care of children with Down syndrome. *European Journal of Pediatrics*, 169(12), 1445-1452. <http://doi.org/10.1007/s00431-010-1253-0>
10. Bull, M. J., & the Committee on Genetics. (2011). Clinical report- Health supervision for children with Down syndrome. *Pediatrics*, 123(2), 393-402. <http://doi:10.1542/peds.2011-1605>
11. Hickey, F., Hickey, E., & Summar, K. L. (2012). Medical update for children with Down syndrome for the pediatrician and family practitioner. *Advances in Pediatrics*, 59(1), 137-157. <http://doi:10.1016/j.yapd.2012.04.006>
12. Hasle, H., Clemmensen, I. H., & Mikkelsen, M. (2000). Risks of leukaemia and solid tumours in individuals with Down's syndrome. *Lancet*, 355(9199), 165-169. [http://doi:10.1016/S0140-6736\(99\)05264-2](http://doi:10.1016/S0140-6736(99)05264-2)
13. Hasle, H., Friedman, J. M., Olsen, J. H., & Rasmussen, S. A. (2016). Low risk of solid tumors in persons with Down syndrome. *Genetics in Medicine*, 18(11), 1151-1157. <http://doi:10.1038/gim.2016.23>
14. Sobey, C. G., Judkins, C. P., Sundararajan, V., Phan, T.G., Drummond, G. R., & Srikanth, V.K. (2015). Risk of major cardiovascular events in people with Down syndrome. *PLoS One*, 10(9):e0137093. <http://doi:10.1371/journal.pone.0137093>
15. Alexander, M., Petri, H., Ding, Y., Wandel, C., Khwaja, O., & Foskett, N. (2015). Morbidity and medication in a large population of individuals with Down syndrome compared to the general population. *Developmental Medicine and Child Neurology*, 58(3), 246-254. <http://doi:10.1111/dmcn.12868>

Acknowledgments

The Global Down Syndrome Foundation, the National Down Syndrome Congress, and the National Down Syndrome Society would like to thank the following contributors for their valuable time and expert advice.

Campbell Brasington, MS

George Capone, MD

Dee Daniels, RN, MSN, CPNP

Penny Feldman, MD

Allan Fisher, MD

Barbara Gaffney

Karen Gaffney

Bryn Gelaro, LSW

Marjorie Greenfield, MD

Francis J. Hickey, MD

Dennis McGuire, PhD

Kishore Vellody, MD

Jeannie Visootsak, MD

Patricia C. Winders, PT



3239 E SECOND AVENUE
DENVER, CO 80206
GLOBALDOWNSYNDROME.ORG



30 MANSELL COURT
SUITE 108
ROSWELL, GA 30076
NDSCCENTER.ORG



8 E 41ST STREET
NEW YORK, NY 10017
NDSS.ORG

© 2017 Global Down Syndrome Foundation & National Down Syndrome Congress. All rights reserved.

Disclaimer. The information in this pamphlet is intended to serve only as an educational tool and is accurate to the best of the knowledge of the Global Down Syndrome Foundation and the National Down Syndrome Congress. The reader should not rely on this pamphlet as medical advice. The reader is urged to seek competent medical advice from a licensed physician and other medical professionals who are knowledgeable about Down syndrome. The Global Down Syndrome Foundation and the National Down Syndrome Congress disclaim any direct, indirect, consequential, special, exemplary, or other diagnosis arising from any person following the information provided in this brochure with respect to any diagnosis, treatment or action. The organizations do not endorse specific products or services, and any implications as such are unintentional.