

Fact Sheet about Down Syndrome for New and Expectant Parents

Reviewed by the National Society of Genetic Counselors Down Syndrome Information Act Working Group, with assistance from the National Center for Prenatal and Postnatal Down Syndrome Resources

OVERVIEW OF DOWN SYNDROME

Down syndrome is a genetic condition that is usually caused by an extra copy of the twenty-first chromosome. According to current data, about 250,000¹ people in the United States have Down syndrome. Studies show that about 1 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.

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 over their lifespan. Because of advances in health care, education, and public attitudes, the outlook for people with Down syndrome has improved significantly over the past few decades.

UNDERSTANDING DOWN SYNDROME³

- 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% of babies with this condition have hypotonia or low muscle tone at birth. This usually improves with time, and physical therapy can help.
- 50% of babies with Down syndrome will have one or more health issues: 40–60% of babies with Down syndrome have a heart condition and 12% have a gastrointestinal condition, which may require 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 impairments, 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.⁴

- Raising a child with Down syndrome may involve more time commitment than typical children.
- Individuals with developmental disabilities can participate in community sports, activities, and leagues.
- Individualized education programs can help children with Down syndrome reach their potential. Special education services at school can range from inclusion in the typical classroom with extra help to small group instruction. There are also over 250 college programs for people with intellectual disabilities.
- Individuals can be employed competitively or with supports; live independently or in a group home; and have friends and intimate relationships.

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.⁵

GETTING MORE SUPPORT

Patients can learn more from genetic counselors who can offer information about testing options and results, the accuracy and limitations of different prenatal screens and tests, reproductive options (continuing a pregnancy, termination, and adoption), recurrence chances for future pregnancies, the genetics of Down syndrome, and local resources, such as local support organizations and connections to other families. A directory of genetic counselors can be found at the National Society of Genetic Counselors (NSGC) website: nsgc.org.

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 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.

⁴ 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.

⁵ 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.

RECOMMENDED RESOURCES FOR NEW AND EXPECTANT PARENTS

All of the following materials have been reviewed by medical experts and are included in the “NSGC Practice guidelines for communicating a prenatal or postnatal diagnosis of Down syndrome.”

National Center for Prenatal and Postnatal Down Syndrome Resources — downsyndromediagnosis.org

A website with a resource directory for new and expectant parents learning about Down syndrome.

“Understanding a Down Syndrome Diagnosis” — lettercase.org

A booklet reviewed by representatives of national medical and Down syndrome organizations for patients first learning about a prenatal diagnosis of Down syndrome.

“Diagnosis to Delivery: A Pregnant Mother’s Guide to Down Syndrome” — downsyndromepregnancy.org

A book and website with detailed information for new and expectant parents anticipating the birth of a child with Down syndrome.

“A Time to Decide, A Time to Heal” — pineapplepress.org and “A Heartbreaking Choice” — aheartbreakingchoice.com:

Books and websites for patients who do not plan on continuing a pregnancy following a prenatal diagnosis.

“Planning for a Loving Future” and “A Loving Choice: Making an Adoption Plan” — ndsan.org
Pamphlets and website for parents considering placing their child for adoption.

Brighter Tomorrows — brightertomorrow.org

A website for both new and expectant parents.

“Facts about Down Syndrome” Center for Disease Control and Prevention (CDC) — cdc.gov
A website with facts about Down syndrome.

“Babies with Down Syndrome,” Edited by Susan J. Skallerup — woodbinehouse.com

An extensive book for new parents.

“Gifts: Mothers Reflect on How Children with Down Syndrome Enrich Their Lives,” and “What Parents Wish They’d Known: Reflections on Parenting a Child with Down Syndrome,” Ed. by Kathryn Soper — woodbinehouse.com

Book with personal essays and quotes by new and expectant parents.

American Academy of Pediatrics, “Health Care Information for Families of Children with Down Syndrome” — healthychildren.org

Guidelines focused on recommended healthcare and treatment.

RECOMMENDED RESOURCES ABOUT PRENATAL SCREENING AND TESTING

“Understanding Prenatal Screening and Testing for Chromosome Conditions” — lettercase.org

“A Patient’s Guide to Understanding Non-Invasive Prenatal Testing” — nsgc.org

NATIONAL DOWN SYNDROME ORGANIZATIONS

National Down Syndrome Society — ndss.org

National Down Syndrome Congress — ndscenter.org

International Mosaic Down Syndrome Association — imdsa.org

Focused on Adoption

National Down Syndrome Adoption Network — ndsan.org

Focused on Education

Down Syndrome Education USA — dseusa.org

Focused on Research

Jerome Lejeune Foundation — lejeuneusa.org, Global Down Syndrome Foundation — globaldownsyndrome.org, and Research Down Syndrome — researchds.org

DS-Connect — dsconnect.nih.gov

Research on cognition and health can also greatly improve outcomes for people with Down syndrome in the future. DS-Connect, sponsored by The National Institutes of Health (NIH), is a voluntary registry for families who want to participate in research opportunities, receive updates on research initiatives, and be included in the collection of data about Down syndrome.

PROFESSIONAL GENETICS ORGANIZATIONS

National Society of Genetic Counselors — nsgc.org

American College of Genetics and Genomics — acmg.net

NATIONAL DISABILITY ORGANIZATIONS

American Association Of People With Disabilities — aapd.com

ARC — thearc.org

Association of University Centers on Disability — aucd.org

March of Dimes — marchofdimes.org

Special Olympics International — specialolympics.org

LOCAL RESOURCES

Enter Children's Hospital Down Syndrome Program

<http://www.childrenshospital.org/centers-and-services/down-syndrome-program>

The Massachusetts General Hospital Down Syndrome Program

<http://www.massgeneral.org/children/services/treatmentprograms.aspx?id=1701>

Massachusetts Down Syndrome Congress

<http://www.mdsc.org/>