



# Understanding the Diagnosis

A Guide for Parents



[dsaneo.org](http://dsaneo.org)



**This booklet is designed  
to answer questions  
that you may have:**

**What is Down syndrome?**

**What health issues  
are associated with  
Down syndrome?**

**How will this affect  
our family?**

**What are the expectations  
for people with  
Down syndrome?**

**What resources  
are available?**



## Dear Parent,

You may have just been told that your baby most likely has Down syndrome. You probably have many questions, concerns and fears right now. **That is okay.** The most important thing to remember is that this diagnosis is not as “life changing” as the fact that you will have a new baby. In most ways, your baby will be just like other infants. Every baby needs to be fed, held and most of all, **loved.**

There will be challenges in raising your child, but there will also be many, many joys. It is normal to be nervous about what lies ahead but remember, Down syndrome is a condition your baby may have, it is not who your baby is.

**Down Syndrome Association of  
Northeast Ohio (DSANEO) wants you  
to know that you are not alone.**

**We support over 1,000 families in  
16 counties of Northeast Ohio who have  
a loved one with Down syndrome.**

**We are here to support you and provide  
you with information to help  
you on your journey.**

As a starting point, we have a comprehensive website ([www.dsaneo.org](http://www.dsaneo.org)) and a wonderful **New Parent Support** program that offers phone calls, home and/or hospital visits or e-mail contact, whichever you prefer. We also offer:

- Early Connections Parent Mentor Network
- Infant and toddler play groups
- Support groups for parents & grandparents
- Parent education programs
- Family grants

You can count on the Down Syndrome Association of Northeast Ohio to be supportive, informative, and available to your family, when you are ready.

## What is Down Syndrome?

In every cell in the human body there is a nucleus, where genetic material is stored in genes. Genes carry the codes responsible for all of our inherited traits and are grouped along rod-like structures called chromosomes. Normally, the nucleus of each cell contains 23 pairs of chromosomes, half of which are inherited from each parent. Down syndrome occurs when an individual has three, rather than two, partial or full copies of the 21st chromosome. This additional genetic material alters the course of development and causes the characteristics associated with Down syndrome.

**Down syndrome is the most commonly occurring chromosomal condition. According to current data, one in every 700 babies is born with Down syndrome and there are about 250,000 people living with Down syndrome in the United States.**

The incidence of births of children with Down syndrome increases with the age of the mother. But due to higher fertility rates in younger women, 80% of children with Down syndrome are born to women under 35 years of age.

A few of the common physical traits of Down syndrome are low muscle tone, small stature, an upward slant to the eyes, and a single deep crease across the center of the palm. Every person with Down syndrome is a unique individual and may possess these characteristics to different degrees, or not at all.

Many people with Down syndrome experience cognitive delays, but the effect is usually mild to moderate and is not indicative of the many strengths and talents that each individual possesses.

Down syndrome does not typically run in families and nothing that you or your partner did or did not do before or during your pregnancy caused your baby to have Down syndrome. Down syndrome occurs in people of all races and economic levels.



## What are the Types of Down Syndrome?

There are three main types of chromosome abnormalities that result in Down syndrome.

### **Trisomy 21 (Nondisjunction)**

Down syndrome is usually caused by an error in cell division called “nondisjunction.” Nondisjunction results in an embryo with three copies of chromosome 21 instead of the usual two. Prior to or at conception, a pair of 21st chromosomes in either the sperm or the egg fails to separate. As the embryo develops, the extra chromosome is replicated in every cell of the body. This type of Down syndrome, which accounts for 95% of cases, is called Trisomy 21.

### **Translocation**

In translocation, which accounts for about 4% of cases of Down syndrome, the total number of chromosomes in the cells remains 46; however, an additional full or partial copy of chromosome 21 attaches to another chromosome, usually chromosome 14. The presence of the extra full or partial chromosome 21 causes the characteristics of Down syndrome.

### **Mosaicism**

Mosaicism, or Mosaic Down syndrome, is noted in about one percent of individuals with Down syndrome. In this case, some cells have 47 chromosomes and others have 46 chromosomes. Mosaicism is the least common form of Down syndrome.

## What Health Issues are associated with Down syndrome?

Over the lifespan, there are various health issues that can arise in people with Down syndrome. In a newborn baby it is important to know that there can be problems with the heart or gastrointestinal tract. Some of these may require surgery.

As the baby grows it is common for development to be mildly or moderately delayed. Early intervention including physical, occupational and speech therapy is beneficial. These services are offered through state and county programs, such as Help Me Grow. Some families choose to find private therapists also. Individuals with Down syndrome can also experience a higher incidence of respiratory and breathing problems, childhood leukemia, thyroid conditions, and Alzheimer's disease. There are healthcare guidelines that families and physicians can follow to ensure that proper screening is being done.

**THANKS TO MEDICAL ADVANCES, LIFE EXPECTANCY FOR PEOPLE WITH DOWN SYNDROME HAS INCREASED DRAMATICALLY IN RECENT DECADES— FROM AGE 25 IN 1983 TO AGE 60 TODAY.**



## What are the Expectations For People with Down Syndrome?

No one, not even a medical professional, can predict your child's potential. However, it may be reassuring for you to know that there are many people with Down syndrome who have achieved great accomplishments in their lives, including graduating from high school, going on to college, living on their own, holding a meaningful job, and getting married. Know that your child will love you and be a valuable member of the family.

**With quality educational programs, a stimulating home environment, good health care, and positive support from family, friends and the community, people with Down syndrome can lead fulfilling lives, and contribute to society.**

At the Down Syndrome Association of Northeast Ohio, we offer many opportunities for children and adults with Down syndrome to learn & grow. Many of these programs are FREE of charge, or highly subsidized. Although our office is in Independence, we offer programs and activities in locations all across Northeast Ohio.



## **How will this Affect Our Family?**

Many parents are concerned about the potential impact that a child with Down syndrome may have on their marriage and/or on their other children. While raising any child offers its own unique set of challenges, it is realistic to acknowledge that there may be some unique challenges to raising a child with a disability.

Brothers and sisters of individuals with Down syndrome report that the positives typically outweigh the negatives in relationships with their siblings. According to some studies, siblings of children with Down syndrome tend to be more compassionate and well-adjusted than their peers. Additionally, a large number of siblings report close relationships with their brother or sister with Down syndrome into adulthood, and many of these siblings choose careers in the health care, support services and education fields.

## What Resources are Available?

### [www.dsaneo.org](http://www.dsaneo.org)

- DSANEO began in 1982 as a parent-led organization. Today we serve 1,000+ families in 16 counties in Northeast Ohio. Access our links and learn about the support that DSANEO provides.
- Visit our New & Expectant Parents page for healthcare and other information.

### [www.globaldownsyndrome.org](http://www.globaldownsyndrome.org)

### [www.ndss.org](http://www.ndss.org)

### [www.ndscenter.org](http://www.ndscenter.org)

International Down syndrome websites with resources for up-to-date statistics and facts about Down syndrome.

### [www.lettercase.org](http://www.lettercase.org)

Accurate information about Down syndrome and other genetic conditions. Information is available in Spanish and multiple other languages.

### [www.downsyndromepregnancy.org](http://www.downsyndromepregnancy.org)

This is a go-to site for honest, compassionate, medically based books and articles related to pregnancy with a Down syndrome diagnosis and an online Baby Center group.

## What are Suggested Next Steps?

### Contact the Down Syndrome Association of Northeast Ohio (DSANEO) at 216.447.8763.

- Request an Expectant Parent or New Parent packet.
- Speak to our New Parent and Family Support Coordinator who can help you through this sensitive time. They will be compassionate, listen to your feelings, and answer all of your questions honestly and without judgment. They can also put you in contact with other parents in our community who have agreed to share their journey.
- Request to be matched with a trained parent mentor from DSANEO who lives in your geographic area.
- Join the DSANEO Parent to Parent Network on Facebook. This private group is a safe space for families to connect and share.

### For Expectant Parents

- You may want to make an appointment for genetic counseling to gain more information about factors related to a Down syndrome diagnosis.

### For Parents of Newborns

- Review the American Academy of Pediatrics guidelines with your baby's doctor. There is a section on newborn care. <https://publications.aap.org/pediatrics/article/128/2/393/30609/Health-Supervision-for-Children-With-Down-Syndrome>
- Find out about early intervention services that can help encourage your baby's development. You can go to <https://ohioearlyintervention.org/about> to find your county contact for early intervention or ask your child's doctor for a referral to Help Me Grow or private therapies.

### For Adoption Information

- Contact the National Down Syndrome Adoption Network (NDSAN) at [www.ndsan.org](http://www.ndsan.org). NDSAN provides information and support to birth parents throughout the United States in order to ensure that every child born with Down syndrome grows up in a caring family.
- There are over 200 families that are home-study ready and eager to adopt a child with Down syndrome.



Scan the QR code for  
Adult Healthcare Guidelines.

We are committed to working  
alongside families to create  
a community in Northeast  
Ohio where people with Down  
syndrome have the opportunity  
to pursue their dreams and  
reach their full potential.



Scan the QR code for  
Pediatric Healthcare Guidelines.



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to visit our website.