PRC

Down Syndrome

What is it?

Down syndrome (DS) is a condition that happens when an infant is born with an extra chromosome. "Typically, a baby is born with 46 chromosomes. Babies with Down syndrome have an extra copy of one of these chromosomes, chromosome 21. A medical term for having an extra copy of a chromosome is 'trisomy: Down syndrome is also referred to as Trisomy 21 :' (https://www.cdc.gov)

There are three types of Down syndrome. From the Centers for Disease Control we learn:

- **Trisomy 21**: About 95% of people with Down syndrome have Trisomy 21. With this type of Down syndrome, each cell in the body has three separate copies of chromosome 21 instead of the usual two copies.
- **Translocation Down syndrome**:This type accounts for a small percentage of people with Down syndrome (about 3%). This occurs when an extra part or a whole extra chromosome 21 is present, but it is attached or "trans-located" to a different chromosome rather than being a separate chromosome 21.
- **Mosaic Down syndrome**: This type affects about 2% of the people with Down syndrome. Mosaic means mixture or combination. For children with mosaic Down syndrome, some of their cells have three copies of chromosome 21, but other cells have the typical two copies of chromosome 21. Children with Mosaic Down syndrome may have the same features as other children with Down syndrome. However, they may have fewer features of the condition due to the presence of some (or many) cells with a typical number of chromosomes.

The physical features for each type of Down syndrome are similar and therefore it is only with further testing that a distinction can be made.

What are the symptoms?

The Department of National Health and Human Services offers this listing of common physical symptoms children with Down syndrome may exhibit: (https://www.nichd.nih.gov/health/topics/down/conditioninfo/symptoms)

- Decreased or poor muscle tone
- Short neck, with excess skin at the back of the neck
- Flattened facial profile and nose

- Small head, ears, and mouth
- Upward slanting eyes, often with a skin fold that comes out from the upper eyelid and covers the inner corner of the eye
- White spots on the colored part of the eye (called Brushfield spots)
- Wide, short hands with short fingers
- A single, deep, crease across the palm of the hand
- A deep groove between the first and second toe

When is Down Syndrome typically diagnosed?

An infant with Down syndrome can be diagnosed during pregnancy or soon after birth. There are several diagnostic tests that can confirm the additional chromosome as well the perceived impact of Down syndrome.

> "There are two types of antenatal (before birth) tests for Down syndrome: screening tests, which allow your doctor to analyze the risk that your child will be born with Down syndrome; and diagnostic tests, which determine definitively whether your baby has the condition. If results from a screening test suggest a high risk of Down syndrome, diagnostic testing will most likely be recommended:' (https://www.sequenom.com/down-syndrome-testing)

What is the prognosis?

Down syndrome is a lifelong condition, and most people with OS are able to live vibrant and fulfilling lives. Some of the cognitive challenges someone with OS may experience are:

- Short attention span
- Poor judgment
- Impulsive behavior
- Slow learning
- Delayed language and speech development

> "Today, because of improved medical technology and early intervention, 80 percent of people with Down syndrome live to age 55 and many live even longer:' (http://www.healthcommunities.com)

How does it impact communication skills?

People with OS are able to communicate in many visual and social ways; however, many do struggle with verbal communication. The Integrated Treatment site offers this explanation:

> One of the areas of communication difficulty that most people are familiar with in people with OS is speech intelligibility. This is often one of the most difficult areas for people with Down syndrome at all ages. Speech is highly complex. It involves coordinating breathing (respiration), voice (phonation), and the production of speech sounds (articulation).

Factors that can contribute to speech intelligibility problems include: a small oral cavity, articulation problems with specific sounds, low oral- facial muscle tone, difficulty with sensory processing and oral tactile feedback and difficulties in motor planning for speech.

In addition, people with OS are significantly more likely than the general population to have hearing and/or vision disorders. For them, receptive language is typically higher than expressive.

However, there are a number of therapies and training that caregivers and parents can begin at an early age to help their child with communication. For those whose verbal communication abilities are severely challenged, PRC- Saltillo offers a number of devices to help in the process of communicating.

Language Preferences

As with other diseases and conditions, there are appropriate ways to refer to and communicate with those that have OS. Following is a listing provided by the National Down Syndrome Society (NOSS) of the dos and don'ts for communication with someone who has OS, and/or their caregiver:

- People with Down syndrome should always be referred to as people first.
- Instead of"a Down syndrome child;' it should be"a child with Down syndrome:' Also avoid "Down's child" and describing the condition as "Down's;' as in, "He has Down's:'
- Down syndrome is a condition or a syndrome, not a disease.
- People"have" Down syndrome, they do not"suffer from" it and are not"afflicted by" it.
- "Typically developing" or"typical" is preferred over"normal:'
- "Intellectual disability" or"cognitive disability" has replaced "mental retardation" as the appropriate term.
- NOSS strongly condemns the use of the word "retarded" in any derogatory context. Using this word is hurtful and suggests that people with disabilities are not competent.

Resources

Primary source of information, resources, research and support: https://www.ndss.org
National Down Syndrome Society
Center for Disease Control: https://www.cdc.gov/ncbddd/birthdefects/downsyndrome.html
US Department of Health: https://www.nichd.nih.gov/health/topics/down/conditioninfo/symptoms
Integrated Treatment services:
http://integratedtreatmentservices.co.uk/blog/downs-syndrome-communication-perspective/
42 Resources for Parents of Children with DS:
https://www.friendshipcircle.org/blog/2013/03/20/42-top-down-syndrome-resources-you-should-know-about/
Health Communities:
http://www.healthcommunities.com/down-syndrome/children/overview-of-down-syndrome.shtml
Local Support NDSS – 375 Local Chapters worldwide: https://www.ndss.org/resources/local-support/

Support

Family Support through Better Health:
https://www.betterhealth.vic.gov.au/health/servicesandsupport/down-syndrome-and-family-support

Facebook Parents of children with Down syndrome: https://www.facebook.com/groups/132540303533527/

Facebook Kids with Down https://www.facebook.com/kidswithdownsyndrome/