

Down Syndrome At a Glance

Down syndrome (DS) is a genetic condition resulting from an extra chromosome (number 21) in some or all of the cells. Instead of 46 chromosomes in each cell, there are 47 in each cell. Another name for Down syndrome is trisomy 21, referring to the extra copy of chromosome 21. Down syndrome can be diagnosed by physical features in most cases, and is confirmed by a karyotype usually done on a blood sample. Individuals who have Down syndrome have unique facial features and physical features, certain medical findings (such as heart defects, gastro-intestinal problems, etc.), and most have an intellectual disability. There is wide variability within individuals who have this condition.

About one in every 700 babies is born with Down syndrome.



Meet Courtney, Brendan, and Madeline starting on page 16.

Things to Think About

1. Medical / Dietary Needs

What you need to know

Individuals who have Down syndrome have variable medical needs:

Heart

- Approximately 40% have a heart defect
- Surgery is sometimes required
- After successful surgery, most children with DS have no limitations in activity, but this should be confirmed with family

Gastro-intestinal (GI)

Some individuals may have one or more of the following:

- Partial or total block in the first part of the intestine after stomach (Duodenal atresia)
- Esophagus not connected to stomach (Esophageal atresia)
- Reflux: (GERD or gastroesophageal reflux)
- Constipation
 - Due to the decreased muscle tone of intestinal tract
- Hirschsprung disease
 - This is caused by the lack of nerve cells in part of colon just above rectum and this lack impairs intestines ability to move stool to rectum.
- Celiac disease
 - A condition where the body can't process a protein in wheat and other grains known as gluten.
- Umbilical hernia
 - Gap between the muscle under the skin of the abdomen allowing belly button to protrude

Epilepsy

Seizures occur in 5-10% of individuals with Down syndrome.

Orthopedic problems

- Neck instability (atlantoaxial joint) found in 15% of individuals with Down syndrome.
 - Caused by underdevelopment of a bone called the odontoid process that usually anchors the head in position, resulting in too much movement between first and second vertebrae in neck
 - May be at risk of spinal cord damage
- Scoliosis (abnormal curvature of the spine)
- Hip instability
- Knee dislocation
- Foot problems
 - Flat feet
 - Ankles turning in and/or out

Ears, Nose and Throat

- Increased upper respiratory infections
 - This is secondary to having a smaller midface, smaller sinuses, smaller nasal passages, and a decreased immune response.
- Hearing loss
 - The risk of hearing loss is high (due mostly to middle ear problems) and regular audiologic testing is recommended.
 - Can cause speech delay

Sleep apnea

- May be due to the obstruction of upper airway by large adenoids and/or tonsils.

Blood problems

- Leukemia in 1% of individuals- cancer of white blood cells
 - Most leukemia occurs in first five years of life
 - AML (acute myelogenous leukemia)
 - ALL (acute lymphocytic leukemia)
 - Transient leukemia

Thyroid problems

- Hypothyroidism – low thyroid levels is more common
- Can have hyperthyroid

Eyes

- Cataracts
- Blocked tear ducts
- Strabismus (eyes turning in or out)
- Blepharitis (chronic infection of the eyelid)
- Nystagmus (repetitive involuntary movement of the eyeballs)
- Visual acuity
 - Nearsightedness
 - Farsightedness
 - Astigmatisms
 - About 50% of children with DS require corrective lenses.
 - If you have a new student with DS who is not in glasses, check with parents to ensure vision is regularly tested.

Dental

- Delayed eruption of teeth, missing teeth and small teeth with small roots are common.

Skin

- Mottled skin or cutis marmorata is very common in babies. In school age children, the skin is often dry and coarse, and atopic dermatitis (red, scaly and itchy skin) is common.
- The skin on palms and soles often becomes very thickened.

Obsessive/compulsive behaviors

- This is an ongoing area of study.
- Obsessive behaviors seem to be common in young children with Down syndrome and are thought to be developmentally important for their learning.
- However, some studies show that Obsessive Compulsive Disorder may be more common in older individuals with Down syndrome than in the general population but is still not a common feature.

Autism spectrum disorder

- Autism is seen in ~ 5-7% of children with Down syndrome
- The diagnosis is usually made at a later age (6-8 years of age).
- Interventions for autism in individuals with Down syndrome are same as the general population and are important to identify as early as possible.

Children who have Down syndrome do not usually require a special diet, other than a healthy, well balanced diet. If an individual has celiac disease they will require a gluten free diet. It is important to be sensitive to cultural differences in diet.

Physical characteristics and/or symptoms:

Not all people with Down syndrome have all of these characteristics.

Low muscle tone

This is called hypotonia.

Characteristic facial features

Nose

- Flat bridge of the nose

Eyes

- Appear to slant upward (upslanting palpebral fissures)
- Small folds of skin in the inner corners (epicanthal folds)
- Light colored spots (Brushfield spots) in the outer parts of the iris of the eye. These spots don't affect an individual's sight.

Mouth

- Tends to be small and the roof of mouth may be shallow
- Tongue often protrudes and appears large in relation to the mouth due to low muscle tone.

Teeth

- May come in late, be small, and appear in an unusual order

Ears

- Small, have small tops that fold over, absent ear lobes, and set lower on head
- Smaller ear passages that can lead to more frequent ear infections

Head shape

- Smaller than normal

Neck

- Appears shorter with excess folds of skin on the back of the neck

Stature:

- Rate of growth not as fast
- Average adult female height is 4 feet 9.
- Average adult male height is 5 feet 2 inches.

Hands and Feet:

- Small with short fingers
- Single crease extending across the palm of one or both hands
- 5th finger curves inward
- Toes have a gap between 1st and 2nd toes

Chest:

- Breastbone either sticks out or dents in.

Skin:

- Mottling is common, as is fair or sensitive skin.

Hair:

- Thin, soft, and/or sparse

What you can do

Encourage families to talk with their pediatricians or family physicians about the medical follow up. The *Health Supervision Guidelines* recommended by the American Academy of Pediatrics may be helpful; see “Resources”.

Otitis Media is common in children with Down Syndrome and signs of ear infection should be closely monitored to ensure that the child is not missing out on valuable language input.

2. Education Supports

What you need to know

It is important to have HIGH LEARNING EXPECTATIONS for learning for children who have Down syndrome. Encourage use of the core educational curriculum and modify it to meet the individual needs of the child.

Children who have Down syndrome have a unique cognitive profile with specific strengths and challenges. Children who have Down syndrome can be included in their neighborhood schools. They may do well when extra supports are provided. Being with their peers is good for developing social and communication skills. Their team can make decisions about the need for specific instruction and pace.

Development

- Intellectual Disability: The majority of Individuals have some degree of intellectual disability
 - Most have mild to moderate intellectual impairment
- When learning, may learn more slowly and have difficulties with complex reasoning and judgment
- Delayed developmental milestones (i.e. walking and talking)
- Reach goals on their own pace
- They are often much stronger visual learners with strong visual memory, visual discrimination, and visual reasoning compared to their overall intellectual ability and their auditory processing.
- Children with DS do better with simultaneous processing than with sequential processing tasks. They can get the "whole idea" but may have difficulty understanding a time sequence. In fact, time is a challenge for many.

- Children who have DS not only use self-talk to help themselves learn, but many seem almost incapable of silent thought.
 - Those who are readers may have difficulty reading silently and need to do so out loud.
 - Also, self-talk often involves the externalization of fantasy life. This can lead to embarrassing moments, and at worst, mistaken impressions that a child or youth is delusional or hallucinating as they audibly act out fantasy dialogues.

Attention

- They may be very distractible.

Motor development

- Visual problems
- Poor muscle tone
- Mild to moderate hearing loss
- Possible cardiovascular irregularities
- Neck instability (AAI or atlantoaxial instability)
 - This is a condition where there is increased mobility between the first and second cervical vertebra, allowing the vertebrae to slip out of alignment easily. This can cause damage to spinal cord.

Communication

- Language acquisition may be slow compared to age matched peers.
- Most children who have DS have an expressive language disability that is more significant than their receptive language or overall intellectual challenges.
- Low muscle tone can impact articulation and speech clarity. The child's message may be distorted due to distortion (altering) and omission (leaving out) of speech sounds.
- Children with Down syndrome often have difficulty with structure and organizing the order of words within a sentence. Constructing a sentence of increased length or complexity may be a challenge. Children may need guidance to recognize that sentences are grammatically correct.

What you can do

Each child should be looked at individually to find ideas that work for him or her. You may want to consider the following strategies and *decide* if any of these suggestions will work for the child. Strategies are adapted from the National Down Syndrome Society (<http://www.ndss.org/>).

Interventions to support academic differences:

Teaching strategies to help development

- Allow choice-making to build decision making skills
- Use routines to help learning
 - Give clear signals about the end of one activity and the beginning of a new activity.
 - Use picture symbols representing activities.
- Keep directions specific and brief
- Demonstrate skills to be learned
 - Use concrete objects/manipulatives along with verbal explanations
 - Use visual and auditory aids
 - Visual materials are important.
 - Reading might be approached initially using a sight reading method.
 - Breakdown in small simple steps
- For reading, may rely on a whole word/site word approach rather than decoding the individual sounds in a word
- Use peer partners
- Use positive behavioral support strategies
 - Provide positive reinforcement immediately
- Have high but realistic expectations
- Small group instruction may be more beneficial to the student than whole classroom
- Ask student to repeat or rephrase instructions
- Set aside time for review and practice of tasks
- Present only a few stimuli or objects at a time
- Be flexible with educational goals.

Teaching strategies to help with short attention span

- Direct instruction in short periods of time
- Teach smaller chunks of activities
- Give new material slowly
- Teach in a sequentially and step-by-step fashion
- Minimize distractibility
 - Keep away from windows
 - Keep a structured environment
 - Keep noise level down
 - Have clear expectations, routines, and rules

Considerations for neck instability

- Individuals need x-rays before physical education because there are no symptoms.

- Individuals who have AAI (atlanto-axial instability) need to avoid any exercise/activity that puts pressure on the muscles of the neck.
 - Gymnastics
 - Diving
 - Butterfly stroke
 - The high jump
 - Heading in soccer

Teaching strategies to help with communication

- If a child is difficult to understand, a speech-language pathologist (SLP) may recommend use of an augmentative and alternative communication device and/or the continued use of sign language.
 - These devices are programmed to the individual child to provide them with a voice and ensure that the child can relay messages to others.
 - iPads are useful for many.
- An SLP can assist with speech clarity and rhythm of speech. An SLP can also assist with grammatical aspects of language in both spoken and written forms.
 - Continued support through the school years will be important as literacy and pragmatic capabilities (the use of language for social communication) become increasingly important in the middle and high school years.
- Otitis Media is common in children with Down Syndrome and signs of ear infection should be closely monitored to ensure that the child is not missing out on valuable language input.

3. Behavior & Sensory Support

What you need to know

There is an increased risk for certain psychological conditions.

- Anxiety disorders
- Depression
- Obsessive compulsive disorders

Be aware of the potential for safety concerns.

- Wandering off
- Playground
- Field Trips

Behavioral Considerations:

Overly friendly affect

- Children with Down syndrome will often approach strangers in a friendly way.
- The overt friendliness with strangers may put them at an increased risk for exploitation.
- Reinforce the concepts of personal boundaries.

May engage in self talk when in uncomfortable or confusing situation

- Talking aloud to oneself is a way to process information and think things through.
- Children who have DS not only use self-talk to help themselves learn, but many seem almost incapable of silent thought.
- Those who are readers may have difficulty reading silently and need to do so out loud.
- Also, self-talk often involves the externalization of fantasy life. This can lead to embarrassing moments, and at worst, mistaken impressions that a child or youth is delusional or hallucinating as they audibly act out fantasy dialogues.

Stubborn/oppositional behavior

- Stubborn behavior usually prompted by not fully understanding what is expected or trying to gain control over their lives
- Pay attention to triggers
 - Frustration
 - Lack of understanding
 - Inability to communicate

Attention

Decreased attention span, impulsive behavior excessive fidgeting, and non-directed motor activity are common in all children at various ages. However, they are more commonly seen in individuals with Down syndrome.

- ADHD and impulsivity can be based on developmental age and not just strictly chronological age.
 - The frequency of ADHD in children with DS is not known with certainty.
 - However, ADHD like symptoms are more common in young children with Down syndrome compared to children from the general population.
 - Compounding symptoms such as repetitiveness, anxiety, or extreme irritability in the presence of ADHD-like symptoms may indicate another disorder such as autism, bipolar disorder or obsessive compulsive disorder.
 - Language processing problems and hearing loss may be involved.
- It is important to rule out other causes of ADHD.

Possible other causes of ADHD in children with Down syndrome

Medical problems can look like ADHD.

Visual and hearing problems

- Moderate hearing loss
- Near and far sightedness
- Cataracts

Gastrointestinal problems

- Typical symptoms of celiac disease include loose stools, diarrhea, and poor weight gain, however, the condition can presents with subtle effects on energy and behavior.
- People with Down syndrome are also more likely to have constipation, which when severe can cause abdominal pain, lack of appetite, and restlessness.

Thyroid Problems

- An underactive thyroid gland can, among other things, make a child very tired and lethargic.
- Too much thyroid activity can cause agitation and restlessness.
- Therefore, both conditions can look like poor attention and behavior.

Sleep problems

- Sleep disorders are extremely common in Down syndrome.
- When tired, children can be restless, whiny, and difficult to calm.
- When tired a child may have difficulty focusing and learning new information.

Communication problems can mimic ADHD.

- The receptive language skills of children with Down syndrome are often much stronger than their expressive language skills.
- This can make classroom participation more difficult.
- The child may express his frustration by acting out or by inattention.

Educational problems may resemble ADHD.

- If a child's learning style is not compatible with the teacher's educational method they may appear bored, fidgety, and hyperactive.
- If the material too difficult a child may "tune out" and appear inattentive.
- A child who is bored with overly easy material also may attend poorly and act out.

Emotional problems may resemble ADHD.

- If a child with Down syndrome has communication problems they may have difficulty talking about things that make them sad or angry.

What you can do

Interventions for psychological conditions

- Individuals with Down syndrome may need:
 - Behavioral supports
 - Counseling
 - Medication

Interventions for safety

- Safety considerations should be written into the classroom IEP.
- Consider using a visual supports such as a stop sign to act as a reminder to ask permission to leave.

Interventions for attention issues

- Direct instruction in short periods of time
- Teach smaller chunks of activities
- Give new material slowly
- Teach in a sequentially and step-by-step fashion
- Minimize distractibility
 - Keep away from windows
 - Keep a structured environment
 - Keep noise level down
 - Have clear expectations, routines, and rules

4. Physical Activity, Trips, Events

What you need to know

Any change in routine may produce anxiety, fears, and/or worry in some individuals. Crowds and loud noises may be overwhelming to some individuals. If the function is in a public place, the child may have more of an opportunity to interact with strangers, so monitoring may be important.

Physical education challenges may include:

- visual problems
- poor muscle tone
- mild to moderate hearing loss
- possible cardiovascular irregularities
- Neck Instability (AAI or atlantoaxial instability)
 - This is a condition where there is increased mobility between the first and second cervical vertebra, allowing the vertebrae to slip out of alignment easily. This can cause damage to spinal cord.

If you live in New England (USA) and qualify, Northeast Passage offers Therapeutic Recreation and Adaptive Sports programming (www.nepassage.org).

What you can do

Provide supports PRIOR to any new event or trip to help familiarize with the upcoming change. This could be a story about the event, photos, etc. Provide any supports that help them with sensory overload (iPod, ear phones, etc.). Because some children are overfriendly and overly familiar with strangers, a one-on-one aide on field trips may be required.

5. School Absences and Fatigue

What you need to know

Absences should not be a big factor for school aged children.

However, sleep problems are common in individuals with Down syndrome, and may lead to fatigue.

- When tired children can be restless, whiny, and difficult to calm.
- When tired a child may have difficulty focusing and learning new information.
- Children may have sleep apnea due to the obstruction of upper airway by large adenoids and/or tonsils.

What you can do

Contact parents if changes are noticed.

6. Emergency Planning

What you need to know

- Emergency plans should be made on an individual child, based on child's behaviors and needs.
- If any change is noticed in an individual who has Down syndrome, it is important to contact the parents.

7. Resources

Down Syndrome Education International - UK

<https://www.down-syndrome.org/en-us>

Explore this site for their excellent library of teaching resources.

PubMed Health- Down syndrome

<http://www.ncbi.nlm.nih.gov/pubmedhealth/PMH0001992/>

Learn more about the genetics of Down syndrome.

National Down Syndrome Society

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

The mission of the National Down Syndrome Society is to be the national advocate for value, acceptance, and inclusion of people with Down syndrome.

National Down Syndrome Congress

<http://ndsccenter.org/>

Parents, grandparents, self advocates and professionals are all invited to join this network, which builds on a shift from isolating people with disabilities to bringing them into communities. This site includes news, resources, government updates, and a special section for self advocates.

American Academy of Pediatrics (AAP) - Health Supervision Guidelines

<https://publications.aap.org/pediatrics/article/149/5/e2022057010/186778/Health-Supervision-for-Children-and-Adolescents>

The AAP endorses these Health Supervision Guidelines, and Health Care Information for Families of Children with Down Syndrome. Families may find these helpful when talking to their pediatricians or family physicians.

Down Syndrome Information Alliance

<http://downsyndromeinfo.org/about/>

Resources that teachers, resource specialists, school district staff, and aids might find useful when working with individuals with Down syndrome

Medline Plus

<https://medlineplus.gov/genetics/condition/down-syndrome/>

Consumer-friendly information about human genetics from the U.S. National Library of Medicine

8. Meet a Child with Down Syndrome



Catching up with Courtney and Brendan!

If you meet Courtney, you might be greeted with a shy smile followed by a big hug when she warms up. When she is not in school, Courtney might be found rock climbing in the local gym, cheerleading, or in a dance class. Her family is very important to her: she loves family gatherings, visiting her grandmother on Lake Winnepesaukee, and surprising her aunts with texting using very inventive spelling!

Her first grade experience has been wonderful, according to her mother. Courtney is a regular first grader with “beautiful friends” and she goes to lots of birthday parties. She uses words at times and uses her communication book and an iPad to communicate other ideas. Her classmates are important to her learning and social life. And when Courtney goes through the lunch line, she is learning to use her own Pin Number to pay, just like everyone else. Her friends lend a hand when she needs help and she also has a paraprofessional providing supports.

At home, Courtney is happy listening to classical music and Barney, and likes to dance to the music. She loves any sports that are associated with a ball!

Her mother has advice for teachers and parents: Choose full inclusion! “It is what is natural and why do something unnatural?” Courtney experiences everything her classmates experience. “It is heartwarming to see how beautiful it is,” she states when thinking of a recent visit to Courtney’s school. Her mother also feels that communication is a key factor in making inclusion successful. Monthly meetings between school and private therapists and specialists have been helpful for problem solving and keeping everyone on the same page.

Brother Brendan...

Brendan has just turned 21. “He is an amazing young man” says his mother, glowing about his life. Brendan has had a team of people helping him reach his goals. In high school, he became a 2nd LT in the JROTC Program and was presented with the Tuskegee Award. His colonel was a believer in Brendan and worked to figure out how Brendan could be supported, not if he should be supported. Brendan was prom king in high school and rode to the prom in a limo with his friends. As part of his work experience in high school, Brendan worked at Wal Mart, Sam’s Club, Walgreens, a pet store and a restaurant to see where his



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interests and successes were. Through the VocTech, he worked in the school store, cafeteria, restaurant, and the preschool program where he read books to young children. He even played Santa Claus!

But the latest achievement is an outstanding success for Brendan and his team. Brendan has recently found full time paid employment with NexDine, a food service company doing business at a high school in New Hampshire! He will have benefits and his supports will be the ‘all natural’ kind – his co-workers! We congratulate Brendan and his dedicated team and wish him great success in his new job.

Madeline for President!



Madeline is having a great year in school. She loves to sing and school has given her so much to sing about! She has shown great improvement in her language since starting at her local school’s preschool program. She is a fun-loving and independent 4 year old girl and her mother says she is like a “mini-teacher”, often directing her friends at school!

Madeline uses lots of words and uses some signs to supplement when needed. Although she uses an iPad at home, it is more for practice for possible use in the future. She has even developed skills in fixing the iPad! She knows the alphabet, recognizes many words and counts to 20. Her family has been reading to her even before she was born. Her vocabulary has grown by leaps and bounds since starting preschool.

Madeline recently missed some school due to her open-heart surgery but has recovered very quickly and hated missing school. She kept asking when she could go back. Her social interests have always been strong and she gets along very well with all kids. She is invited to birthday parties and her mother hopes to start more play dates now that she is recovering from her surgery.

She is great with her younger 8 month old brother often hugging, holding, and feeding him. And she loves to copy her mother when she does chores and will wash dishes, pull out her mini-broom and help sweep, or help clean-up her toys.

Madeline loves her family and her grandparents who play a great role in supporting her. In fact, her mother states that their immediate and extended family, their friends, and their neighbors have been an “extremely supporting and loving community from day one.”

From Madeline’s mother: Her biggest fear was what would happen when Madeline got to school. Her advice is to “Don’t let your fears hold you or your child back. Get on board with

inclusive education, even if you have to introduce the concept to your school.” On the advice of a wise pediatrician at her birth, they always treat Madeline like any other child. “Don’t expect anything less of your child. Give lots of opportunities. Don’t hold back- she is always surprising us on what she can do. I expect her to be President the way she is going.”

Her mother also attended the NH Leadership Series and trainings at the Parent Information Center. She feels more confident in her ability to be a good advocate, network with other families, and build a community of support due to her Leadership training. She says, “I feel lucky to have had Madeline. I have friends, connections and a purpose in life that I wouldn't otherwise have.”

Great wisdom and food for thought! We will be looking for Madeline in future elections!!!