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Attention Deficit Hyperactivity Disorder (ADHD)

Attention Problems in Down Syndrome: Is This ADHD?

Attention deficit hyperactivity disorder, or ADHD, is a commonly diagnosed childhood problem. ADHD is characterized by consistent demonstration of the following traits: decreased attention span, impulsive behavior, and excessive fidgeting or other nondirected motor activity.

All children, including children with Down syndrome, display these traits from time to time. But the child with Down syndrome may exhibit these traits more often than other children his age.

Does that mean that your child has ADHD? It may, but more often it means that a medical problem needs to be addressed, or that your child's educational program or communication method needs some adjustment. In children with Down syndrome who have difficulty paying attention, ADHD is a diagnosis of exclusion. Other problems must be ruled out first. What follows is a discussion of those problems.

Medical Problems that Can Look Like ADHD

Hearing and vision problems

In order for a child to pay attention to classroom material, she has to be able to hear and to see it. Both hearing and visual problems are common in children with Down syndrome. Ear infections are overwhelmingly common and, even if treated, can cause hearing loss for weeks. People with Down syndrome have middle ear structural abnormalities that can cause lifetime mild to moderate hearing loss.

Both near- and far-sightedness are common in Down syndrome, as well as cataracts and "lazy eye."

How can we rule out significant hearing and/or visual loss as a cause of attentional problems? To monitor hearing, an auditory brainstem response test (ABR) or otoacoustic emission (OAE) should be performed early in the child's life--by 3 months of age at the latest--as a baseline. Hearing screens should be performed annually until three years of age, and every other year thereafter. Children with abnormal hearing evaluations should be seen by an ear-nose-throat physician (otolaryngologist) to manage treatable causes of hearing loss.

A child with Down syndrome should be evaluated by an eye doctor during the first year of her life, and yearly thereafter. Some children may need more frequent followup depending on their visual diagnosis.

Gastrointestinal problems

People with Down syndrome are at increased risk for an intestinal condition called celiac disease, which is a condition in which the body cannot process a protein found in wheat and certain other grains. While typical symptoms of celiac disease include loose stools, diarrhea, and poor weight gain,

the condition often presents only with subtle effects on energy and behavior. People with Down syndrome are also predisposed to significant constipation, which when severe can cause abdominal pain, lack of appetite, and restlessness.

Current recommendations for gastrointestinal monitoring include screening for celiac disease between 2 and 3 years of age. This screening should include measurement of IgA antiendomysium antibodies, as well as total IgA. Your child's primary care provider will want to review your child's bowel status with you at each visit as well.

Thyroid problems
About 30 percent of people with Down syndrome have thyroid disease at some point in life. Most have hypothyroidism, or underactive thyroid gland; a few have disease that results in overactive thyroid gland (Graves' disease). An underactive thyroid gland can, among other things, make a child very tired and apathetic.

Too much thyroid activity can cause agitation and restlessness. Therefore, both conditions can look like poor attention and behavior.

Because thyroid disease is so prevalent in this population, and because it is difficult for doctors to detect just by examining your child, an annual blood test for thyroid hormone is recommended by the Down Syndrome Preventive Checklist.

Sleep problems
Sleep disorders are extremely common in Down syndrome. These disorders are a group of conditions with many different causes but one thing in common: they all interfere with getting a good night's sleep. As a parent, you know that tired children can behave very differently from tired adults: they can become restless, whiny, and difficult to calm. And people of all ages have difficulty focusing and learning new information when they are sleep deprived.

Which sleep disorders are common in people with Down syndrome? Sleep apnea, or short periods of not breathing during sleep, is especially common. People with Down syndrome have small, often "floppy" airways, which can sometimes be completely or partially blocked during sleep by large tonsils and adenoids, or by the floppy walls of the airway collapsing as air is exhaled. Regardless of the cause of obstruction, the sleeper must awaken briefly to resume breathing. Some patients with sleep apnea awaken hundreds of times per night.

Symptoms associated with but not specific to sleep apnea include snoring, lots of "thrashing" while asleep, excessive daytime sleepiness, mouth breathing, and unusual sleep positions such as sleeping in a seated or hunched forward position.

Children suspected of having a sleep disorder should undergo a sleep study evaluation at an accredited sleep center.

Communication Problems that Can Look Like ADHD

People with Down syndrome may have many barriers to effective communication. The receptive language skills of children with Down syndrome--how well they understand what is being said--are often much stronger than their expressive language skills--how well they can say it. Parents often comment, "He knows what he wants to tell us, he just can't seem to put the words together or we can't make out what he is saying." Classroom participation is thus more difficult as well. The child may express his frustration by acting out or by inattention.

Educational problems
Children with Down syndrome have a wide range of learning styles. Your child's educational team may need to try more than one method of presenting material before finding the one that works best for your child. If material is presented in a way that is not compatible with a child's learning style--for example, oral lectures for a student who needs visual aids and prompts--that child may appear bored, fidgety, and hyperactive.

The level of the material may also be a problem. If a child is presented with concepts that are too difficult for his cognitive level, he might "tune out" and appear inattentive. A child who is bored with overly easy material also may attend poorly and act out.

Emotional problems

Because of the communication problems discussed above, people with Down syndrome may have difficulty talking about things that make them sad or angry. Major life changes such as loss or separation may prompt decreases in appropriate behavior at school or work.

ADHD and Down Syndrome

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 stereotypy (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.

Uncomplicated ADHD is common in the youngest children with Down syndrome. However many school age children with ADHD frequently have other behavioral conditions including oppositional defiant disorder, disruptive behavior disorder or obsessive compulsive traits.

Next Steps

If you are concerned about decreased attention span, impulsive behavior, and excessive fidgeting or other non-directed motor activity in your child, it is appropriate to consult your pediatrician, a developmental and behavioral pediatrician or child psychiatrist.

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The Health of Adults with Down Syndrome

There are some health conditions that occur more often in people with Down syndrome. There are also some health conditions that seem to occur less often. Adults with Down syndrome, their supporting loved ones and their health care providers should be aware of the main health issues related to trisomy 21. In addition, adults with Down syndrome should be offered all of the regular screening tests and health care maintenance interventions that are commonly provided to adults who do not have Down syndrome.

What health conditions occur more often in Down syndrome?

Adults with Down syndrome are at higher risk of developing diseases of autoimmunity, perhaps because of the number of immune system genes that reside in chromosome 21. These are conditions in which the person's own immune system attacks particular tissues that are mistakenly perceived as foreign. The most common autoimmune conditions that co-occur with Down syndrome are **hypothyroidism and celiac disease**. Graves' disease resulting in hyperthyroidism and **type 1 diabetes** also seem to have a higher incidence in Down syndrome. Other health conditions that can present in adulthood in Down syndrome include **atlantoaxial instability, obesity, obstructive sleep apnea and Alzheimer's disease.**

What is hypothyroidism? How is it diagnosed and treated?

Hypothyroidism is a dysfunction of the thyroid gland, by which it secretes lower amounts of thyroid hormone than are required to maintain normal body metabolism. Thyroid hormone helps regulate energy balance; when levels are low, the patient feels sluggish, tired and/or cold. He/she may develop constipation, gain weight and have lower intellectual functioning. It can be easily diagnosed with a blood test, and it is treated with daily thyroid hormone supplements. Because these supplements consist of the same hormone the body would have produced, there is no significant side effects when provided in adequate doses.

What is celiac disease? How is it diagnosed and treated?

Celiac disease results from an autoimmune reaction to gluten, a protein component of wheat products. The autoimmune reaction takes place in the gut, and as a result the small intestine becomes atrophied and loses absorbing capacity. Symptoms can be varied and include unintentional weight loss, bloating, abdominal pain, diarrhea or cramps. It can be screened for by measuring **tissue transglutaminase antibody**; supporting tests include **anti-gliadin and anti-endomysial** antibodies. A confirmatory diagnosis can only be obtained by a small bowel biopsy performed during upper endoscopy. Treatment involves the institution of a gluten-free diet, i.e. eliminating all wheat flour products from the diet.

What is atlantoaxial instability? How is diagnosed and treated?

Atlantoaxial instability results from laxity in the ligaments that hold together the first (atlas) and second (axis) cervical vertebrae in the neck. If the ligaments become loose, a bony process of the axis may impinge on the spinal cord and cause nerve damage, particularly during bending of the neck or an injury sustained in contact sports. The condition occurs in approximately 15 percent of people with Down syndrome: it is diagnosed by neck X-rays obtained in the flexion and extension positions. The only definitive treatment is surgical.

Why does obesity occur more often in Down syndrome?

Sometimes it is the result of untreated hypothyroidism. There is a suggestion that people with Down syndrome may have a lower level of metabolism, i.e. their bodies may consume less calories and therefore store more. More generally, it is due to the intake of **too many calories in relation to the level of physical activity**.

Treatment strategies involve lowering portion size, emphasizing healthy ingredients that increase bulk (i.e. fiber, fruits and vegetables), making smart choices on drinks and desserts, avoiding snacks, empowering young adults to monitor their own weight, involving supervisors at work and school on the treatment plan, preparing lunch at home, never using food as a reward, and introducing a daily exercise routine that is appropriate to the young adult's interest and skills.

What is obstructive sleep apnea? How is it diagnosed and treated?

In obstructive sleep apnea the airway closes transiently during sleep, resulting in frequent brief periods of poor oxygenation that lead to awakening and poor quality sleep. The patient is often unaware of these symptoms, but sleep fragmentation often results in daytime sleepiness, inability to concentrate, irritability, headaches, low energy and cognitive dysfunction. It occurs more often in Down syndrome because of the small size of the oral cavity in relation to the soft tissues it contains (tongue, palate, tonsils and adenoids), their low muscle tone, and their propensity to obesity. It is diagnosed by an overnight sleep study in a sleep lab. Non-invasive treatment options include a continuous positive airway pressure (CPAP) machine and weight loss. Invasive treatment options include resection of tonsils and adenoids, or more involved ENT procedures.

What about behavioral changes in adulthood?

These can be caused by a number of factors: difficulty with transitions into adolescence or young adulthood, with the loss of social networks, departure of older siblings, death of loved ones, move out of the home or transfer from a protective school environment into a work situation; sensory deprivation, either visual (e.g. cataracts) or auditory (hearing loss); emotional trauma; hypothyroidism; obstructive sleep apnea; depression; and **Alzheimer's disease**. While Alzheimer's disease occurs earlier and more often in adults with Down syndrome than in the general population, not every behavioral or cognitive change in an adult with Down syndrome should be ascribed to this form of dementia. The reversible causes enumerated above should be considered, sought after and treated.

What health conditions occur less often in Down syndrome?

It appears that adults with Down syndrome have a lower incidence of coronary atherosclerosis (which can cause heart attacks), stroke, high blood pressure and many solid tumors. This does not necessarily mean that they are exempt from them, especially if they do have a family history of any of the above. As a group, they do have a much lower incidence of substance abuse or death by violence. They also seem to have a higher threshold for pain.

What about health care maintenance?

Adults with Down syndrome should be offered the same screening procedures and preventive interventions that are recommended for the general population, including vaccinations, mammograms, diabetes and cholesterol screening, colon cancer screening, etc. These tests should be individualized to each patient's personal situation in consultation with their health care provider.

NDSS thanks special guest author Jose Florez, M.D., Ph.D. for preparing this piece.

The Health of Adults with Down Syndrome Resource List

Organizations and Websites

Adult Down Syndrome Center of Lutheran General Hospital
1999 Dempster Street
Park Ridge, IL 60068
Telephone: 847-318-2303
Website: http://www.advocatehealth.com/adultdown

Clearinghouse on Aging and Developmental Disabilities
Website: http://www.uic.edu/orgs/rrtcamr/clearinghouse.htm

Books

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Alantoaxial Instability in Down Syndrome

Atlantoaxial Instability in Down Syndrome: Controversy and Commentary

In 1984, the AAP issued its first position statement on Atlantoaxial Instability (AAI) in children with Down Syndrome (DS):

- All children with DS who wish to participate in sports should have cervical spine X-rays.
- When the distance on X-ray between the atlas (1st vertebra) and odontoid process (2nd vertebra) is more than 4.5 millimeters (mm), restriction on sports is advised.
- Repeated X-rays are not indicated for children with DS who have previously had normal neck X-rays.
- Persons with atlantoaxial subluxation or dislocation and neurologic signs should be restricted from "all strenuous activities."
- Persons with DS who have no evidence of AAI may participate in all sports.

The American Academy of Pediatrics' Committee on Sports Medicine released a revised statement in July 1995 regarding Atlantoaxial Instability (AAI) in children with DS. This was published in the journal Pediatrics, 96(1):151-154. Here's the text, edited for brevity's sake:

"In 1984, the Amer. Academy of Pediatrics (AAP) published a position statement on screening for AAI in youth with DS. In that statement, the AAP supported the requirement introduced by the Special Olympics (SO) in 1983 that lateral (side view) neck X-rays be obtained for individuals with DS before they participate in the SO's nationwide competitive program. Those participants with radiologic evidence of AAI are banned from certain activities that may be associated with increased risk of injury to the cervical spine....The Committee on Sports Medicine recently has reviewed the data on which this recommendation was based and has decided that uncertainty exists concerning the value of cervical spine X-rays in screening for possible catastrophic neck injury in athletes with DS. The 1984 statement therefore has been retired. This review discusses the available research data on this subject.

Background

AAI denotes increased mobility at the articulation of the first and second cervical vertebrae (atlantoaxial joint). The causes of AAI are not well understood but may include abnormalities of the ligaments that maintain the integrity of the articulation, bony abnormalities of the cervical vertebrae, or both.

In its mildest form, AAI is asymptomatic and is diagnosed using X-rays.....Symptomatic AAI results from subluxation (excessive slippage) that is severe enough to injure the spinal cord, or from dislocation at the atlantoaxial joint.

Approximately 15% of youth with DS have AAI. Almost all are asymptomatic. Some asymptomatic individuals who have normal X-rays initially will have abnormal X-rays later, and others with initially abnormal X-rays will have normal follow-up X-rays; the latter change is more common....

The neurologic manifestations of symptomatic AAI include easy fatiguability, difficulties in walking, abnormal gait, neck pain, limited neck mobility, torticollis (head tilt), incoordination and clumsiness, sensory deficits, spasticity, hyperreflexia...and {other spinal cord} signs and symptoms. Such signs and symptoms often remain relatively stable for months or years; occasionally they progress, rarely even to paraplegia, hemiplegia, quadriplegia, or death. Trauma rarely causes the initial appearance or the progression of these symptoms. Nearly all of the individuals who have experienced catastrophic injury to the spinal cord had weeks to years of preceding, less severe neurologic abnormalities....

Most importantly, symptomatic AAI is apparently rare in individuals with DS. In the pediatric age group, only 41 well-documented cases have been described in the published literature....

Asymptomatic AAI, which is common, has not been proven to be a significant risk factor for symptomatic AAI....

The efficacy of the intervention to prevent symptomatic AAI has never been tested. Sports trauma has not been an important cause of symptomatic AAI in the rare patients with this disorder; only 3 of the 41 reported pediatric cases had initial symptoms of AAI or worsening of symptoms after trauma during organized sports participation. Members of the SO Medical Advisory Committee think that more such sports-related injuries occur but that they are being overlooked because of a lack of information about the association of AAI and spinal cord injury among health care providers. This claim has not been substantiated with published research....

Tenative Conclusions

....it is reasonable to conclude that lateral neck X-rays are of potential but unproven value in detecting patients at risk for developing spinal cord injury during sports participation. It seems that identification of those patients who already have or who later have complaints or physical findings consistent with symptomatic spinal cord injury is a greater priority than obtaining X-rays. Recognition of these symptomatic patients is challenging and requires frequent interval histories and physical exams, including evaluations before participation in sports, preferably by physicians who have cared for these patients longitudinally. Their parents must learn the symptoms of AAI that indicate the need to seek immediate medical care.

The SO does not plan to remove its requirement that all athletes with DS receive neck X-rays. Pediatricians will therefore continue to be called on to order these tests. The information here can be used to interpret the results for family members...." (end excerpt)

Dr. Siegfried Pueschel wrote his opposition to the revised AAP statement in the Jan 1998 issue of the journal Archives of Pediatric and Adolescent Medicine.

Dr. Pueschel is the head of the DS Clinic in Providence, Rhode Island, and the author of several studies and textbooks on Down syndrome. In his article, Dr. Pueschel's main points are:

- While the X-ray may not be as good of a screen as we'd like, there is currently nothing better.
- AAI is not rare: it occurs in children with DS (10-30%) and symptomatic AAI may reach up to 1 to 2% of all children with DS.
- Symptomatic AAI is a serious disorder, which justifies the work and expense required to detect it.
- While it isn't known if asymptomatic AAI turns into symptomatic AAI, it hasn't been disproven yet, either.

- To date, there have been no reports of spinal cord injury from any activity associated with Special Olympics, Inc. This may mean that such an injury is a rare occurrence, or it may actually show that SO's precautionary measures are effective at preventing such injuries.
- If one waits for significant neurologic signs to appear, spinal cord damage may have already occurred. By waiting, an individual at risk with no symptoms will not be detected.
- Further, lateral neck X-rays may also detect the less common but more serious atlanto-occipital instability, or degenerative changes in the cervical spine.

Dr. Pueschel concludes, therefore, that lateral neck X-rays are still an important part of optimal care for individuals with DS.

Also in the Jan 1998 issue of the journal Archives of Pediatric and Adolescent Medicine is an editorial by Dr. Bill Cohen on the controversy.

Dr. Cohen is the head of the DS Clinic of the Children's Hospital of Pittsburgh and co-chair of the Down Syndrome Medical Interest Group, a collective of health professionals dedicated to the care of individuals with DS. Dr. Cohen summarizes both the 1995 AAP statement and Dr. Pueschel's statement, and then addresses the controversy thusly:

"If in fact asymptomatic AAI is not the precursor to symptomatic AAI, the current protocol [X-rays at 3, 12, and 18 years of age] should be abandoned. I would suggest that the devastating nature of cord compression and the technical difficulties in assessing children with developmental disabilities has led to the current quandary. Few organizations would be willing to take responsibility, however limited, for a recommendation that might lead or be perceived to lead to steps that would fail to protect individuals with DS from a spinal cord injury."

Dr. Cohen goes on to cite a recent article addressing the technical aspects of measuring for AAI, and adds that in his opinion, children with DS who have a narrowed neural canal or evidence of marked AAI should receive an MRI of the neck before restriction of activity or any surgical procedure requiring anesthesia. He ends his editorial by calling for a consensus meeting involving representatives of all medical fields that this topic encompasses.

Addendum, 1/14/00

This month, a valuable review on this topic was published by Dr. Douglas Brockmeyer, a neurosurgeon at Children's Hospital in Salt Lake City, Utah. In it, the author reviews the literature to date and gives solid recommendations for doctors for making decisions on the basis of X-rays. This paper, Down Syndrome and Craniovertebral Instability, is my Abstracted Paper for Jan 2000.

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Alzheimer's Disease & Down Syndrome

Alzheimer's Disease, a degenerative neurological disorder characterized by progressive memory loss, personality changes and loss of functional motor capabilities, is far more common in individuals with Down syndrome than the general population. However, not all individuals with Down syndrome will develop Alzheimer's disease, and even those showing Alzheimer's-type symptoms may not have Alzheimer's disease since other conditions can mimic the symptoms.

How common is Alzheimer's disease in individuals with Down syndrome?

Estimates vary, but a reasonable conclusion is that 25 percent or more of individuals with Down syndrome over age 35 show clinical signs and symptoms of Alzheimer's-type dementia. The percentage increases with age. In the general population, Alzheimer's disease does not usually develop before age 50, and the highest incidence (in people over age 65) is between five and 10 percent. The incidence of Alzheimer's disease in the Down syndrome population is estimated to be three to five times greater than in the general population, and oftentimes, symptoms begin much earlier.

What are the symptoms of Alzheimer's disease?

Early symptoms include loss of memory and logical thinking, personality change, decline in daily living skills, new onset of seizures, changes in coordination and gait, and loss of continence in bladder and bowel habits.

How is a final diagnosis made?

Alzheimer's disease is difficult to diagnose. It is important to be certain Alzheimer's-type symptoms do not arise from other conditions, namely thyroid disorders, clinical depression, brain tumor, recurrent brain strokes, metabolic imbalances and various neurological conditions.

The diagnosis of Alzheimer's disease is made on the basis of clinical history, showing a slow, steady decrease in cognitive function and a variety of laboratory tests which provide contributory evidence, including electroencephalogram, brain stem auditory evoked response, computerized transaxial tomography and magnetic resonance imaging, among other tests and measurements.

Is there a baseline test that can be repeated at intervals to determine specific decrease in cognitive function?

Psychologists often use questionnaires answered by family members, companions or caretakers that assist in the early detection of dementia. It is recommended that individuals with Down syndrome be tested at age 30 to provide a baseline reading, and periodically thereafter. If the tests show deterioration, further tests must be made to rule out conditions that present similar or overlapping symptoms.

What information has research yielded about a link between Alzheimer's disease and Down syndrome?

Current research investigating how certain genes on Chromosome 21 may predispose individuals with Down syndrome to Alzheimer's disease. A number of centers are testing therapies in Down syndrome that appear to benefit patients with Alzheimer's disease in the general population.

ndss Alzheimer's Disease & Down Syndrome

Alzheimer's and Down Syndrome Resource List

Organizations, Websites and Articles

Alzheimer's Association
225 North Michigan Avenue, Floor 17
Chicago, IL 60601
Telephone: 800-272-3900 (24-hour hotline)
Website: http://www.alz.org

Alzheimer's Disease International's fact sheet on dementia and intellectual disabilities:
http://www.alz.co.uk/adi/publications.html

Alzheimer's disease and people with mental retardation article:
http://www.thearc.org/netcommunity/document.doc

Developmental disabilities and Alzheimer's disease: What You Should Know
http://www.thearc.org → click "Publications"

Books on Alzheimer's Disease and Down Syndrome

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national down syndrome society

ndss Autism & Down Syndrome

Down Syndrome and Autistic Spectrum Disorder: A Look at What We Know
by George T. Capone, M.D.

From Disability Solutions
Volume 3, Issue 5 &6

During the past 10 years, I've evaluated hundreds of children with Down syndrome, each one with their own strengths and weaknesses, and certainly their own personality. I don't think I've met a parent who does not care deeply for their child at the clinic; their love and dedication is obvious. But some of the families stand out in my mind. Sometimes parents bring their child with Down syndrome to the clinic—not always for the first time—and they are deeply distraught about a change in their child's behavior or development. Sometimes they describe situations and isolated concerns that worry them such as their child has stopped learning new signs or using speech. He is happy playing by himself, seeming to need no one else to make the odd game (shaking a toy, lining things up) he is playing fun. When they call to him, he doesn't look at them. Maybe he isn't hearing well? He will only eat 3 or 4 foods. The suggestion of a new food, or even an old favorite, brings about a tantrum like no other. He is constantly staring at the lights and ceiling fans. Not just while they pass by, but obsessively. Getting him to stop staring at the lights is sometimes difficult and may result in a scene. He requires a certain order to things. Moving a chair to another spot in the room upsets him until it is returned to its usual spot.

Some families do their own research and mention they think their child may have autistic spectrum disorder (ASD) along with Down syndrome. Others have no idea what may be happening. They do know it isn't good and they want answers now. This article is for families in situations like this and other, similar ones. If your child has been dually-diagnosed with Down syndrome and autistic spectrum disorder (DS-ASD) or if you believe your child may have ASD, you will learn a little more about what that means, what we are learning through data collection, and insights to the evaluation process.

There is little written in the form of research or commentary about DS-ASD. In fact, until recently, it was commonly believed that the two conditions could not exist together. Parents were told their child had Down syndrome with a severe to profound cognitive impairment without further investigation or intervention into a diagnostic cause. Today, the medical profession recognizes that people with Down syndrome may also have a psychiatric-related diagnosis such as ASD or Obsessive Compulsive Disorder (OCD).
Because this philosophy is relatively new to medical and educational professionals, there is little known about children and adults with DS-ASD medically or educationally.

Over the past six years we have gathered data and studied DS-ASD at Kennedy Krieger Institute. We have collected and analyzed data from clinical medical evaluations, psychological and behavioral testing, and MRI scans of the brain. We now follow a cohort of approximately 30 children with DS-ASD through the Down syndrome Clinic, possibly the largest group of children with DS-ASD that has been gathered.

What Should I Look For?

Signs and Symptoms

As parents, it is common, if not expected, for you to worry at times about your child's development. It is also common to hear only part of the criteria for a particular label. This is especially true when it comes to DS-ASD because there is little information available on the topic. This can be especially troublesome if your child suddenly picks up a new habit you associate with ASD such as incessantly shaking toys. The children we have

seen at Kennedy Krieger Institute who have DS-ASD present symptoms in several different ways, which we have separated into two general groups:

Group One

Children in this first group appear to display “atypical” behaviors early. During infancy or toddler years you may see:

- Repetitive motor behaviors (fingers in mouth, hand flapping),
- Fascination with and staring at lights, ceiling fans, or fingers,
- Extreme food refusal,
- Receptive language problems (poor understanding and use of gestures) possibly giving the appearance that the child does not hear, and
- Spoken language may be highly repetitive or absent.
- Along with these behaviors, other medical conditions may also be present including seizures, dysfunctional swallow, nystagmus (a constant movement of the eyes), or severe hypotonia (low muscle tone) with a delay in motor skills.

If your child with Down syndrome is young, you may see only one or a few of the behaviors listed above. This does not mean your child will necessarily progress to have autistic spectrum disorder. It does mean that they should be monitored closely and may benefit from receiving different intervention services (such as sensory integration) and teaching strategies (such as visual communication strategies or discrete trial teaching) to promote learning.

Group Two

A second group of children are usually older. This group of children experience a dramatic loss (or plateauing) in their acquisition and use of language and social-attending skills. This developmental regression may be followed by excessive irritability, anxiety, and the onset of repetitive behaviors.

This situation is most often reported by parents to occur following an otherwise “typical” course of early development for a child with Down syndrome. According to parents, this regression most often occurs between ages three to seven years.

The medical concerns and strategies for these two groups may be different. There is not enough information available to know at this time. However, regardless of how or when ASD is first discovered, children with DS-ASD have similar educational and behavioral needs once they are identified.

ASD 101: A Crash Course

Signs and Symptoms Vary

Although we are documenting some similarities in the way DS-ASD presents, autism is what is considered a spectrum disorder. This means every child with DS-ASD will be different in one way or another. Some will have speech, some will not. Some will rely heavily on routine and order, and others will be more easy-going. Combined with the wide range of abilities seen in Down syndrome alone, it can feel mystifying. It is easier if you have an understanding of ASD disorders separate from Down syndrome.

Autism, autistic-like condition, autistic spectrum disorder (ASD), and pervasive developmental disorder (PDD) are terms that mean the same thing, more or less. They all refer to a neurobehavioral syndrome diagnosed by the appearance of specific symptoms and developmental delays early in life. These symptoms result from an

underlying disorder of the brain, which may have multiple causes, including Down syndrome. At this time, there is some disagreement in the medical community regarding the specific evaluations necessary to identify the syndrome or the degree to which certain “core-features” must be present to establish the diagnosis of ASD in a child with Down syndrome. Unfortunately, the lack of specific diagnostic tests creates considerable confusion for professionals, parents, and others trying to understand the child and develop an optimal medical care and effective educational program.

There is general agreement that:

- Autism is a spectrum disorder: it may be mild or severe.
- Many of the symptoms overlap with other conditions such as obsessive-compulsive disorder (OCD) or attention deficit hyperactivity disorder (ADHD).
- ASD is a developmental diagnosis. Expression of the syndrome varies with a child’s age and developmental level.
- Autism can co-exist with conditions such as mental retardation, seizure disorder, or Down syndrome.
- Autism is a life-long condition.

The most commonly described areas of concern for children with ASD include:

- Communication (using and understanding spoken words or signs),
- Social skills (relating to people and social circumstances),
- Repetitive body movements or behavior patterns.

Of course there is inconsistency in any of these areas in all children, especially during early childhood.

Children who have ASD may or may not exhibit all of these characteristics at any one time nor will they consistently demonstrate their abilities across similar circumstances. Some of the variable characteristics of ASD we have commonly observed in children with DS-ASD include:

- Unusual response to sensations (especially sounds,
- lights, touch or pain),
- Food refusal (preferred textures or tastes),
- Unusual play with toys and other objects,
- Difficulty with changes in routine or familiar surroundings,
- Little or no meaningful communication,
- Disruptive behaviors (aggression, throwing tantrums, or extreme non-compliance),
- Hyperactivity, short attention, and impulsivity,
- Self-injurious behavior (skin picking, head hitting or banging, eye-poking, or biting),
- Sleep disturbances, and
- History of developmental regression (esp. language and social skills).

Sometimes these characteristics are seen in other childhood disorders such as attention deficit hyperactivity disorder or obsessive compulsive disorder.

Sometimes ASD is overlooked or considered inappropriate for a child with Down syndrome due to cognitive impairment. For instance, if a child has a high degree of hyperactivity and impulsivity only the diagnosis of ADHD may be considered. Children with many repetitive behaviors may only be regarded as having stereotypy movement disorder (SMD), which is common in individuals with severe cognitive impairments.

Most parents agree that severe behavior problems are usually not easily fixed. Finding solutions for behavioral concerns is one reason families seek help from physicians and behavior specialists. Compared to other groups of children with cognitive impairment, those with Down syndrome, as a group, are less likely to have behavioral or psychiatric disorders. When they do, it is sometimes referred to as having a “dual-diagnosis.” It is important for professionals to consider the possibility of a dual-diagnosis (Down syndrome with a psychiatric condition such as ASD or OCD) because:

1. It may be responsive to medication or behavioral treatment, and

2. A formal diagnosis may entitle the child to more specialized and effective educational and intervention services.

If you think your child may have ASD disorder, share this before or during your evaluation. Don’t wait to see what might happen.

Incidence

Estimating the prevalence or occurrence of ASD disorder among children and adults with Down syndrome is difficult. This is partly due to disagreement about diagnostic criteria and incomplete documentation of cases over the years. Currently, estimates vary between 1 and 10%. I believe that 5-7% is a more accurate estimate. This is substantially higher than is seen in the general population (.04%) and less than other groups of children with mental retardation (20%). Apparently, the occurrence of trisomy 21 lowers the threshold for the emergence of ASD in some children. This may be due to other genetic or other biological influences on brain development.

A review of the literature on this subject since 1979 reveals 36 reports of DS-ASD (24 children and 12 adults). Of the 31 cases that include gender, an astonishing
28 individuals were males. The male-to-female ratio is much higher than the ratio seen for autism in the general population. Additionally, in reports that include cognitive level, most children tested were in the severe range of cognitive impairment.

Generally, the cause of ASD is poorly understood, whether or not it is associated with Down syndrome. There are some medical conditions in which ASD is more common such as Fragile-X syndrome, other chromosome anomalies, seizure disorder, and prenatal or perinatal viral infections. Down syndrome should be included in this list of conditions. The impact of a pre-existing medical condition such as Down syndrome on the developing brain is probably a critical factor in the emergence of ASD disorder in a child.

Brain Development and ASD

The development of the brain and how it functions is different in some way in children with DS-ASD than their peers with Down syndrome. Characterizing and recording these differences in brain development through detailed evaluation of both groups of children will provide a better understanding of the situation and possible treatments for children with DS-ASD.

A detailed analysis of the brain performed at autopsy or with magnetic resonance imaging (MRI) in children with autism shows involvement of several different regions of the brain:

- The limbic system, which is important for regulating emotional response, mood and memory,
- The temporal lobes, which are important for hearing and normal processing of sounds,
- The cerebellum, which coordinates motor movements and some cognitive operations, and
- The corpus callosum, which connects the two hemispheres of the cortex together.

At Kennedy Krieger Institute, we have conducted MRI studies of 25 children with DS-ASD. The preliminary results support the notion that the cerebellum and corpus callosum is different in appearance in these children compared to those with Down syndrome alone. We are presently evaluating other areas of the brain, including the limbic system and all major cortical subregions, to look for additional markers that will distinguish children with DS-ASD from their peers with Down syndrome alone.

Brain Chemistry and ASD

The neurochemistry (chemistry of the brain) of autism is far from clear and very likely involves several different chemical systems of the brain. This information provides the basis for medication trials to impact the way the brain works in order to elicit a change in behavior.

An analysis of neurochemistry in children with ASD alone has consistently identified involvement of at least two systems.

1. Dopamine: regulates movement, posture, attention, and reward behaviors; and

2. Serotonin: regulates mood, aggression, sleep, and feeding behaviors.

Additionally, opiates, which regulate mood, reward, responses to stress, and perception of pain, may also be involved in some children.

Detailed studies of brain chemistry in children with DS-ASD have not yet been done. However, our clinical experience in using medications that modulate dopamine, serotonin or both systems has been favorable in some children with DS-ASD.

How Do I Find Out?

Obtaining an Evaluation

If you suspect that your child with Down syndrome has some of the characteristics of ASD or any other condition qualifying as a dual-diagnosis, it is important for him to be seen by someone with sufficient experience evaluating children with cognitive impairment—ideally Down syndrome in particular. Some of the same symptoms which occur in DS-ASD are also seen in stereotypy movement disorder, major depression, post-traumatic stress disorder, acute adjustment reactions, obsessive-compulsive disorder, anxiety disorder, or when children are exposed to extremely stressful and chaotic events or environments.

Sometimes when children with Down syndrome are experiencing medical problems that are hidden—such as earache, headache, toothache, sinusitis, gastritis, ulcer, pelvic pain, glaucoma, and so on—the situation results in behaviors that may appear “autistic-like” such as self-injury, irritability, or aggressive behaviors. A comprehensive medical history and physical examination is mandatory to rule out other reasons for the behavior. When cooperation is elusive, sedation or anesthesia may be required. If so, use this “anesthesia time” effectively by scheduling as many specialty examinations as are feasible at one session.

In addition to the medical assessment, you will be asked to help complete a checklist to determine whether or not your child has ASD. I use the Autism Behavior Checklist (ABC), but there are others that are also used such as the Childhood Autism Rating Scale (CARS) and the Gilliam Autism Rating Scale (GARS). Each of

these is completed either in an interview with parents or done by parents before coming to the appointment. They are then scored and considered along with clinical observation to determine if your child has ASD.

Obstacles to Diagnosing DS-ASD

“If it looks like a duck, and it quacks like a duck... guess what?”

Parents sometimes face unnecessary obstacles in seeking help for their children. Parents have shared several reasons demonstrating this. Some of the more common include:

Failure to Recognize the Dual Diagnosis

Problem:

Failure to recognize the dual diagnosis except in the most severe cases.

Result:

This is frustrating for everyone who is actively seeking solutions for a child. If you are in this situation and feel that your concerns are not taken seriously, keep trying. The best advice is to trust your gut feeling regarding your child. Eventually you will find someone willing to look at all the possibilities with you.

Diagnostic Confusion

Problem:

Diagnostic confusion with other behavioral or psychiatric conditions such as ADHD, OCD, or depression.

Result:

Parents may feel forced into demanding a referral for another medical evaluation at a Down syndrome clinic or Child Development Center. This often is a considerable cost for families because of insurance concerns. Many HMOs and PPOs will not refer out or take on part of the cost for evaluations outside of their system. The same is true for educational evaluations. Many school systems may be hesitant to provide additional, intensive, and costly services for kids with DS-ASD. The combination of frustration and lack of acceptance by professionals (medical and educational) of the dual diagnosis may lead parents to abandon traditional services in favor of nontraditional solutions to their child’s medical and educational needs. This is not necessarily a bad thing. Individual, creative problem-solving is a great asset when support is elusive.

However, total withdrawal from “the system” may lead to feelings of abandonment and isolation, which makes it difficult for families to help their child and build the support systems needed to deal with stress. There will be plenty of frustrating and stressful moments in the future. Parents deserve support.

Lack of Acceptance by Professionals

Problem:

There is sometimes a lack of acceptance by professionals that ASD can coexist in a child with Down syndrome who has cognitive impairment. They may feel an additional label is not necessary or accurate. Parents may be told, “This is part of ‘low functioning’ Down syndrome.” We now know this is incorrect. Children with DS-ASD are clearly distinguishable from children with Down syndrome alone or those who have Down syndrome and severe cognitive impairment when standardized diagnostic assessment tools such as the ABC are used.

Result:

Parents become frustrated and may give up trying to obtain more specific medical treatment or behavioral intervention.

Confusion in Parents

Problem:

Lack of acceptance, understanding, awareness, or agreement on the part of parents or other family members, particularly of very young children, about what's happening. Initial reactions by families and parents vary considerably from, "This too shall pass" to "Why isn't he doing as much as other kids with DS?"

Result:

Parents in this situation may find themselves at odds with each other about the significance of their child's behavior and what to do about it. As a result, marriages are stressed, parenting relationships with other children are strained, and life is tough altogether. Unfortunately, I have found that parents in this situation almost universally withdraw from local Down syndrome support groups or other groups that may provide support. There are a variety of reasons for this including "the topics discussed don't apply to my child," It's just too hard to see all those children doing so much more than my child," and "I feel like people think I'm a bad parent because of my daughter's behavior."

Ideally someone in the parent group would recognize this when it is happening and offer additional support instead of watching them withdraw. What is worrisome is that the very parents who are most in need of support and assistance cannot or do not receive it within the context of their local parent group. In fact, there may not be another parent in the group with a child who is similar because DS-ASD is uncommon and not easily shared.

It is critical that parents have an opportunity to meet and learn from other parents whose children also have DS-ASD. Despite the underlying medical condition (trisomy 21), the neurobehavioral syndrome of ASD may mean that a support group for families of children with autism will be helpful as well. However, because of the lack of acceptance or knowledge about the dual diagnosis, these support groups can be equally daunting.

What Does it Mean?

Behavioral Findings

Obtaining a diagnosis of DS-ASD is rarely helpful in understanding how ASD effects your child. It is complicated by the lack of information available, making it difficult to discern appropriate medical and educational options. To determine what behaviors are most common in DS-ASD we are conducting case-control studies which randomly match (for gender and age) a child with DS-ASD with a child who has Down syndrome without ASD. These comparisons are based on the information obtained from the ABC together with a detailed developmental history and behavioral observation. Through this process we have been able to determine the following:

Children with DS-ASD were more likely to have:

- History of developmental regression including loss of language and social skills,
- Poor communication skills (many children had no meaningful speech or signing),
- Self-injurious and disruptive behaviors (such as skin picking, biting, and head hitting or banging),
- Repetitive motor behaviors (such as grinding teeth, hand flapping, and rocking),
- Unusual vocalizations (such as grunting, humming, and throaty noises),
- Unusual sensory responsiveness (such as spinning, staring at lights, or sensitivity to certain sounds),

- Feeding problems, (such as food refusal or strong preference for specific textures), and
- Increased anxiety, irritability, difficulty with transitions, hyperactivity, attention problems, and significant sleep disturbances.

Other observations include:

- Children with DS-ASD scored significantly higher than their peers with Down syndrome alone on all five subscales of the ABC: sensory function, social relating, body and object use, language use, and social skills.
- Children with DS-ASD show less impairment in social relatedness than those with ASD only.
- Children with DS-ASD show more preoccupation with body movement and object use than children with ASD alone.
- Children with DS-ASD scored higher on all five subscales of the ABC than children with severe cognitive impairment alone.
- Among children with Down syndrome only, even those with severe cognitive impairment do not always meet the criteria for ASD.

The conclusion I draw from this data is children DS-ASD are clearly distinguishable from both “typical” children with Down syndrome and those with severe cognitive impairment (including children with Down syndrome). Thus, it is probably incorrect to suggest autistic-like behaviors are entirely due to lower cognitive function. However, the fact that autistic features and lower cognition are associated indicates there is some shared determinant(s) that are common to both features (ASD and lower cognition) of the condition.

Associated Medical Conditions

There are questions about the possibility of similarities in the variety of medical conditions associated with Down syndrome in general in children with DS-ASD. To determine this we used the same matching scheme as described above. It is important to point out the number of matched pairs currently in our study is quite small and, as a result, some of these findings may not hold up as we examine more children.

DS/ASD children were more likely to have:

- Congenital heart disease and anatomical GI tract anomalies,
- Neurological findings,(i.e., seizures, dysfunctional swallow, severe hypotonia and motor delay),
- Opthamologic problems,
- Respiratory problems (i.e., pneumonia and sleep apnea), and
- Increased total number of medical conditions.

What Now?

After the Evaluation

If your child has DS-ASD, obtaining the diagnosis or label may be a relief of sorts. The addition of ASD brings new questions. From a medical perspective it is important to consider use of medication, particularly in older children, for specific behaviors. This is especially true if these behaviors interfere with learning or socialization. While there is no cure or remarkably effective treatment for Down syndrome and autistic spectrum, certain “target behaviors” may be responsive to medication. Some of these behaviors include:

- Hyperactivity and poor attention,
- Irritability and anxiety,
- Sleep disturbance,
- Explosive behaviors resulting in aggression/disruption (can sometimes be reduced),
- Rituals and repetitive behaviors (can sometimes be reduced), and
- Self-injury (can sometimes be reduced).

As you continue to take care of your child, make a point to take care of yourself and your family—in that order. You have a life and a family to consider. Recognize that there is only so much time, energy and resources that you can put into this “project.” Of course there will be cycles, of good times and bad, but if you can’t find some way to renew your emotional spirit, then “burn-out” is inevitable. There is a higher rate of anxiety, sleep problems, lack of energy, depression, and failed or struggling marriages under these circumstances. Learn to recognize your own difficulties and be honest with yourself and your spouse about the need for help. Counseling and medication may go a long way in helping you to be at your best, for everyone’s sake.

Conclusion

Clearly there is a great deal to be learned about children with Down syndrome who are dually diagnosed with autism spectrum disorder. In the meantime, it is essential for parents to educate themselves and others about this condition. Families must work on building a team of health-care professionals, therapists, and educators who are interested in working with their child to promote the best possible outcome. Research efforts must move beyond mere description to address causation, early identification, and natural history. Specific markers in the development of the brain which can distinguish DS-ASD from “typical” Down syndrome and “typical” autism need to be sought; and the possible benefits of various treatments need to be more carefully documented. Realizing these goals will take a very long time to accomplish and must be approached with a spirit of support, cooperation, and caring both for individual children and the larger community of children with DS-ASD.

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Resource Organizations

Autism Society of America
7910 Woodmont Avenue
Bethesda, MD 20814-3015
301/657-0881
www.autismsociety.org

National Association for Dual Diagnosis
132 Fair Street
Kingston, NY 12401-4802
www.thenadd.org

Kennedy Krieger Institute
Down Syndrome Clinic
707 N. Broadway
Baltimore, MD 21205
referrals: 888/554-2080

Cincinnati Center of Developmental Disorders
Down Syndrome Clinic
3333 Burnett
Cincinnati, OH 45208
513/636-6755

National Down Syndrome Society
666 Broadway
New York, NY 10012
800/221-4602
www.ndss.org

National Down Syndrome Congress
7000 Peachtree-Dunwoody Road, NE
Lake Ridge 400 Office Park
Bldg #5 - Suite 100
Atlanta, GA 30328

George T. Capone, M.D., is the Director of the Down Syndrome Clinic and Attending Physician on the Neurobehavioral Unit at Kennedy Krieger Institute in Baltimore, Maryland.

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3. Ghaziuddin, M., Tsai, L., and Gahziuddin, N. “Autism in Down’s Syndrome: Presentation and Diagnosis.” Journal of Intellectual Disability Research 36:449-456 (1992).

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6. Rogers, S. “Neuropsychology of Autism in Young Children and Its Implications for Early Intervention.” Mental Retardation and Developmental Disabilities Research Reviews 4:104-112 (1998).

7. Forness, S. and Kavale, K. “Autistic Children in School: The Role of the Pediatrician.” Pediatric Annals 13(4):319-328 (1984).

Dual Diagnosis of Autism and Down Syndrome Resource List

Organizations and Websites

Autism Society of America
7910 Woodmont Avenue, Suite 300
Bethesda, Maryland 20814-3067
Telephone: Phone: 301.657.0881 or 1.800.3AUTISM (1.800.328.8476)

Down Syndrome and Autism Internet Information Center for Creating Solutions
http://www.disabilitysolutions.org/dsaiic/downsyndrome.htm

National Association for the Dually Diagnosed
132 Fair Street
Kingston, NY 12401
Telephone: (845) 331-4336 or (800) 331-5362
Fax: (845) 331-4569
Website: http://www.thenadd.org
E-mail: info@thenadd.org

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Online Articles

Capone, G. **Down Syndrome and Autistic Spectrum Disorder: A Look at What We Know** (1999). Available online by the Kennedy Krieger Institute
http://www.kennedykrieger.org/kki_misc.jsp?pid=2141&bl=1

Gurthie Medlen, J. **More than Down Syndrome: A Parents View** (1999). Available online by the Kennedy Krieger Institute. http://www.kennedykrieger.org/kki_misc.jsp?pid=2140

Patterson, B. **Dual Diagnoses: the Importance of Diagnosis and Treatment** (1999).
Available online by the Kennedy Krieger Institute http://www.kennedykrieger.org/kki_misc.jsp?pid=2142&bl=1

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Blood Diseases & Down Syndrome

Hematologic Abnormalities in Down syndrome

Individuals with Down syndrome (DS) frequently show abnormalities in the blood cells which include the red cells (cells that carry oxygen throughout the body), white cells (infection-fighting cells) and platelets (cells that help to stop bleeding). Some of the changes found in the blood cells of individuals with DS can be associated to other medical complications seen among this patient population; many times however the same blood cell abnormalities are diagnosed without an apparent cause. In many instances the abnormalities resolve spontaneously after a period of time; this is especially common among newborn babies with DS; some patients will have persistence of the changes in the blood cells throughout their life. Sometimes the abnormalities seen in the blood cells in patients with DS can lead to serious medical problems; evaluation and treatment by a hematologist/oncologist (physician who specializes in disorders of the blood cells and the care of cancer patients) may be necessary.

What are the most common hematologic abnormalities identified in patients with DS?

The most common blood cell abnormalities diagnosed in patients with DS include: polycythemia (also known as erythrocytosis), macrocytosis, thrombocytopenia, thrombocytosis, leucopenia, leukemoid reactions and transient myeloproliferative disorder.

What is polycythemia?

Polycythemia or erythrocytosis means **elevated number of red blood cells**. This is frequently seen in newborn babies with DS. In some babies the elevation in the number of red blood cells can be associated with some types of congenital heart defects but the great majority of babies with DS who develop polycythemia don't have an associated heart defect. Typically the polycythemia will resolve within the first few months of life especially if it is not associated with an underlying medical condition. In general polycythemia, regardless of the cause, won't cause significant problems associated to the increased number of red cells. Occasionally some individuals may need to be treated if the number of red cells is extremely elevated and the blood gets too thick.

What is macrocytosis?

Macrocytosis means **enlargement of the red blood cells**. This is a very common finding in newborn babies with DS. This hematologic finding persists throughout life in about two-thirds of individuals with DS. Medical conditions that can be associated with macrocytosis include hypothyroidism (insufficient production of thyroid hormone), megaloblastic anemia (decreased number of red cells because of deficiency in vitamin B12 and Folic acid), increased number of reticulocytes (young red blood cells), some diseases of the liver and the bone marrow (the tissue found in the hollow interior of bones where all the blood cells are made). It is important to consider these medical conditions when an individual with DS is found with macrocytosis but in the great majority of cases an apparent cause won't be found. Macrocytosis shouldn't cause any adverse medical effect.

What is thrombocytopenia?

Thrombocytopenia means **decreased number of platelets**. This is a common diagnosis made in babies with DS. It can be seen associated with some types of congenital heart defects. Also it is a frequent hematologic finding among newborns with DS without any heart problems; in these cases typically the low platelet count usually resolves within few weeks. Having a platelet count that is too low may predispose to bleeding. Under some circumstances patients who have significant thrombocytopenia may require platelet transfusions.

What is thrombocytosis?

Thrombocytosis means **elevated number of platelets**. This is a rare hematologic finding in newborn babies with DS. When it occurs, it usually doesn't cause any medical problem and it goes away spontaneously within the first weeks of life.

What is leukopenia?

Leukopenia means **low number of white cells**. The number of white cells tends to be slightly lower than the normal range in about one-third of individuals with DS. There is some concern that this may increase the risk for infections among patients with DS but this has not been clearly proven through medical research yet.

What is a leukemoid reaction?

Leukemoid reaction means **very elevated number of white cells**. Babies born with DS can present with this blood cell abnormality within the first few months of life. This elevated white cell count typically goes away spontaneously. In some instances it may be associated with a more serious medical condition (see transient myeloproliferative disorder).

What is transient myeloproliferative disorder (TMD)?

This is a medical condition found almost exclusively in newborn babies with DS. Not every baby with DS will develop TMD, it is estimated that 10-20% (10 to 20 out of 100) babies born with DS are diagnosed with TMD. This condition results from rapid growth of abnormal white cells. The abnormal cells may go away without treatment, or they may need treatment. The choice of treatment or no treatment depends on whether certain harmful characteristics of the disease are observed in the baby. There are different ways to treat babies with TMD. The decision regarding what kind of therapy will be necessary depends on the type of harmful characteristics that are seen in the baby. Some of the treatments that have been used include 1) leukophoresis (a procedure that filters abnormal cells from their blood); and/or 2) chemotherapy (anti-cancer drugs). In the great majority of patients, the prognosis of TMD is good with complete resolution of the disease without any treatment; however, few patients with TMD don't show improvement and develop cancer in the blood or leukemia. In some patients leukemia develops years after TMD is resolved. In general babies who are diagnosed with TMD will need to be followed very closely to make sure that the disease goes away. Because TMD is potentially cancerous, children who are diagnosed with this disorder should be monitored closely for years after the signs and symptoms of the condition resolve. The evaluation and treatment of TMD should be discussed with a hematologist/oncologist.

Oncologic Disorders in Down syndrome

Individuals with DS have an increased risk for the development of precancerous conditions such as myelodysplastic syndrome, potentially cancerous conditions such as transient myeloproliferative disorder and cancerous conditions like leukemia.

What is myelodysplastic syndrome (MDS)?

Myelodysplastic syndrome (MDS) is a pre-cancerous condition that originates in the bone marrow in which there are cells that are abnormal. The bone marrow is the tissue found in the hollow interior of bones where all the blood cells are made.

The diagnosis of MDS is suspected when the blood cells start showing changes like a decrease in the platelet count (thrombocytopenia), an increase in the size of the RBC (macrocytosis), decrease in the number of red cells and level of hemoglobin (anemia) or a decrease or increase in the number of white cells.

If not treated MDS will progress to leukemia. The time frame for the development of leukemia ranges from months to years. The evaluation and treatment of MDS should be discussed with a hematologist/oncologist (physician who specializes in disorders of the blood cells and the care of cancer patients).

What is transient myeloproliferative disorder (TMD)?

This is a medical condition found almost exclusively in newborn babies with DS. Not every baby with DS will develop TMD, it is estimated that 10-20% (10 to 20 out of 100) babies born with DS are diagnosed with TMD. This condition results from rapid growth of abnormal white cells. The abnormal cells may go away without treatment, or they may need treatment. The choice of treatment or no treatment depends on whether certain harmful characteristics of the disease are observed in the baby. There are different ways to treat babies with TMD. The decision regarding what kind of therapy will be necessary depends on the type of harmful characteristics that are seen in the baby. Some of the treatments that have been used include 1) leukophoresis (a procedure that filters abnormal cells from their blood); and/or 2) chemotherapy (anti-cancer drugs). In the great majority of patients, the prognosis of TMD is good with complete resolution of the disease without any treatment; however, few patients with TMD don't show improvement and develop cancer in the blood or leukemia. In some patients leukemia develops years after TMD is resolved. In general babies who are diagnosed with TMD will need to be followed very closely to make sure that the disease goes away. Because TMD is potentially cancerous, children who are diagnosed with this disorder should be monitored closely for years after the signs and symptoms of the condition resolve. The evaluation and treatment of TMD should be discussed with a hematologist/oncologist.

What is leukemia?

Leukemia is a cancer of the blood cells. Leukemia develops when young and abnormal cells, called ‘blasts', crowd out normal bone marrow cells and spread into the blood stream. The ‘blasts' can also spread to the brain, spinal cord and other organs.

There are different subtypes of the leukemia; a patient who is diagnosed to have leukemia should be evaluated by a hematologists/oncologist who will determine the subtype of leukemia, and the type of therapy that will be administered to the patient.

How is the diagnosis of myelodysplastic syndrome and leukemia confirmed?

The diagnosis is confirmed by obtaining a bone marrow aspirate and biopsy. A bone marrow aspirate and biospy is a fairly simple procedure to remove a sample of the bone marrow for special testing. Typically it involves putting a needle into the marrow space of the patient's hip (pelvis) bone and removing bone marrow with a syringe. The marrow sample will be looked at under a microscope; also special tests will be performed in the sample in order to make a specific diagnosis.

How is cancer treated in patients with DS?

Patients who develop cancer should be evaluated and treated by a hematologist/oncologist. Chemotherapy agents (anti-cancer drugs) and other supportive therapies such as blood transfusions are used for the treatment of patients who have cancer. Individuals with DS are more sensitive to some chemotherapy agents and some of the side effects are more severe for patients with DS when compared to the general population. Therefore these patients should be watched more closely when receiving certain types of chemotherapy medicines.

Are there other types of cancers that patients with DS can develop?

Patients with DS probably have an increased risk for the development of germ cell tumors (a rare type of cancer found in the ovaries or the testes but may also be found in other areas of the body such as the brain, chest or abdomen). There is increasing concern that retinoblastoma (a rare type of cancer which develops in the eye, in the cells of the retina) may be diagnosed more frequently among people with DS. By far the most frequently type of cancer encountered in patients with DS is leukemia.

NDSS thanks Dr. Natalia Dixon for preparing this information.

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Organizations and Online Support

American Cancer Society
1599 Clifton Road NE
Atlanta, GA 30329
Telephone: 404-320-3333 or 800-227-2345 Website: www.cancer.org

Candlelighters Childhood Cancer Foundation *(support for parents of children with cancer)*
P.O. Box 498
Kensington MD 20895
Telephone: 800-366-2223
Email: staff@candlelighters.org Website: www.candlelighters.org

Down Syndrome and Leukemia Online Group
http://health.groups.yahoo.com/group/ds_leukemia/ *is an online support group for parents of children who have Down syndrome and Leukemia*

Children's Leukemia Research Association
585 Stewart Avenue
Garden City, NY 11530
Telephone: 516-222-1944 Website: www.childrensleukemia.org

Leukemia & Lymphoma Society
1311 Mamaroneck Avenue
White Plains, NY 10605
Telephone: 914-949-5213 or 800-955-4572
Email: infocenter@lls.org Website: www.lls.org

Rare Cancer Alliance
1649 North Pacana Way
Green Valley, AZ 85614
Telephone: 520-625-5495
Email: sharon.lane@rare-cancer.org Website: www.rare-cancer.org

National Cancer Institute
6116 Executive Blvd.
Bethesda MD 20892
Telephone: (301) 435-3848 or (800) 422-6237 Website: www.cancer.gov

Articles on Down Syndrome and Blood Diseases

Leshin, Len. “Diseases of the Blood in Down Syndrome”. Down Syndrome: Health Issues. www.ds-health.com/hemat.htm

Rajantie, J. “Review. Leukemia in Down's Syndrome” The Cancer Journal. May-June 1996. Volume 9 Number 3. Reprinted at: www.riverbendsds.org/rajantie.html

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Ear, Nose and Throat (ENT) Issues & Down Syndrome

Ear, Nose, and Throat (ENT) Issues and Down Syndrome

Ear, nose, and throat problems are common in children with Down syndrome. It is important for primary care physicians and caregivers to be aware of these problems, most of which are present throughout an individual's life. The ENT specialist (also called an otolaryngologist) plays an important role in the health of a child with Down syndrome, especially given that ENT problems are closely linked to physical, emotional, and educational development.

External Ear Canal Stenosis

Stenotic ear canals (narrow ear canals) can occur in up to 40-50 percent of infants with Down syndrome. Narrow ear canals can make the diagnosis of middle ear disease difficult. Cleaning of the ear canals by an ENT specialist is often necessary to ensure proper examination and diagnosis. Ear canals grow with age, and may no longer be of concern after age 3. If a child with Down syndrome has stenotic ear canals, he should see an ENT specialist every 3 months to avoid undiagnosed and untreated ear infections.

Chronic Ear Infections

Children with Down syndrome have an increased incidence of upper repertory tract infections, which predisposes chronic ear infections. The facial anatomy of Down syndrome also predisposes chronic ear disease.

The middle ear is aerated by the eustachian tube, a small tube that goes from the middle ear space to the area behind the nose in the nasopharynx. Upper airway infections or allergies can cause the eustachian tube to become swollen, trapping bacteria and causing ear infections. Low muscle tone (hypotonia) affects the opening and closing of the eustachian tube, as well, which can cause negative pressure to build up in the middle ear space, leading to fluid retention and infection.

Chronic eustachian tube dysfunction lasts longer in children with Down syndrome than in the general population; so the ears and potential infection should be monitored regularly. Some children may need repeated placement of pressure equalization (PE) tubes to eliminate chronic infections. Monitoring and treatment is critical, as there is a high rate of underdiagnosis and undertreatment of ear infections in children with Down syndrome.

Hearing Loss

Hearing loss can affect educational, language, and emotional development. Even mild hearing loss can affect a child's articulation. Monitoring and treatment of the ears and ear diseases can lessen the incidence of hearing loss. PE tubes can also improve hearing. The American Academy of Pediatrics and the Down Syndrome Medical Interest Group recommend audiologic testing at birth and then every 6 months up to age 3, or until the child can cooperate for an audiogram that includes ear-

specific testing (more frequently if hearing loss is present). After the age of 3, children with Down syndrome should have a hearing test performed annually. Hearing aids should be considered even with mild hearing loss to prevent delays in educational, emotional and language development.

Airway obstruction and Sleep Apnea

Airway obstruction is common in children with Down syndrome, with some studies suggesting that nearly all persons with Down syndrome have some form of sleep-related obstruction. Loss of sleep due to apnea and even poor quality sleep due to sleep disordered breathing can result in sleepiness, disturbances in fine motor skills, and also affects behavior and learning. Many with sleep disorders fall asleep with passive activities such as riding in the car or school bus.Long term complications of sleep apnea include systemic hypertension, pulmonary hypertension, heart failure, and even death.

Obstructive sleep apnea occurs when the airway is blocked during sleep. This can be caused by the small upper airway, large adenoids and tonsils, obesity, collapse of the airway due to hypotonia of the muscles of the throat, and increased secretions that can be characteristic of persons with Down syndrome. Obstruction can also occur from glossoptosis, a condition where a relatively large tongue falls back into a smaller airway during sleep.

Obstructive sleep apnea is often overlooked by caregivers and medical professionals, as sleep disturbances often occur unobserved or have been present for so long that parents assumed that was “normal” for their child.
A comprehensive clinical exam, X-ray, and thorough sleep study should be conducted if sleep apnea is suspected.

Airway obstruction can be treated both medically and surgically, and sometimes both treatments are necessary. Saline spray can keep the airway clear. Other medical options include the use of a Continuous Positive Airway Pressure (CPAP) machine during sleep, which provides some pressure with each breath, keeping the airway open while a person sleeps. Weight reduction may also help address sleep apnea. Surgically, removal of the tonsils and adenoids (T&A) is the first line of treatment of airway obstruction and sleep apnea in children with Down syndrome. Although removal of the tonsils and adenoids is usually curative of most sleep apnea in child, more recent studies suggest that this is not always the case with individuals with Down syndrome and further evaluation and treatment may be needed after T&A.

Chronic Rhinitis and Sinusitis

The facial anatomy of Down syndrome along with the developing immunological system predispose the child with Down syndrome to chronic rhinitis (inflammation of the mucus membranes of the nose and mucus discharge) and sinusitis (inflammation of the sinus membranes). Treatment includes the use of saline drops or spray to keep the smaller nasal passages clear as well as the use of antihistamine medications and steroid nasal sprays. These issues should improve with age and can usually be managed by the primary care physician, rather than the ENT specialist.

In children whose sinusitis fails to resolve with medical management, surgical removal of the adenoids and/or endoscopic sinus surgery may be necessary.

This piece was adapted from Down Syndrome: Common Otolaryngologic Manifestations by Dr. Sally Shott

Ear, Nose and Throat and Down Syndrome Resource List

American Speech-Language Hearing Association
10801 Rockville Pike
Rockville, MD 20852
Telephone: (301) 897.5700 or (800) 638-8255
Website: http://www.asha.org

Blind Children's Center
4120 Marathon Street
Los Angeles, CA 90029-0159
Telephone: (323) 664-2153 or (800) 222-3566
E-mail: info@blindchildrenscenter.org
Website: http://www.blindchildrenscenter.org

National Association for Parents of the Visually Impaired, Inc.
P.O. Box 317
Watertown, MA 02472-0317
Telephone: (617) 972-7441 or (800) 562-6265
E-mail: napvi@perkins.org
Website: http://www.napvi.org

National Institute on Deafness and Other Communication Disorders Clearinghouse
Telephone: (800) 241-1044 or (800) 241-1055
Website: http://www.nidcd.nih.gov

Books on Vision and Hearing

Holbrook, M.C. (Ed.). (1996). Children with Visual Impairments: A Parents' Guide. Bethesda, MD: Woodbine House. http://www.woodbinehouse.com

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Endocrine Conditions and Down Syndrome

Individuals with Down syndrome have a higher incidence of endocrine problems than the general population. The endocrine system refers to a set of glands that include the thyroid, adrenal and pituitary glands.

What is hypothyroidism?

Hypothyroidism results from a malfunctioning thyroid gland, which decreases the synthesis of the hormone thyroxin. Thyroxin is the hormone that promotes growth of the brain and other body tissue.

How common is hypothyroidism?

Hypothyroidism is the most common endocrine problem in children with Down syndrome. It is estimated that approximately 10 percent of children with Down syndrome have congenital or acquired thyroid disease. Studies of adults with Down syndrome vary widely, but the incidence of thyroid disease in adults with Down syndrome is believed to be between 13 and 50 percent. Hypothyroidism can occur at any time from infancy through adulthood.

How is hypothyroidism diagnosed?

All individuals with Down syndrome should be tested for hypothyroidism at birth and at periodic intervals (at least every two years) thereafter. The indicators of hypothyroidism -- enlarged tongue, constipation, poor circulation -- are also found in individuals who are not hypothyroid, so the blood test for thyroid function is an important diagnostic test. Because the thyroid hormone affects normal development of the brain, testing of infants is particularly crucial.

What treatment exists for hypothyroidism?

The thyroid hormone, thyroxin, is readily replaced through medication.

What is hyperthyroidism?

In this case, the thyroid gland is overactive. Symptoms are swelling in the neck, abnormal sweating and rapid pulse rate. No evidence exists as to whether hyperthyroidism is more prevalent in individuals with Down syndrome than in the general population.

What is the treatment for hyperthyroidism?

This thyroid malfunction can be treated pharmacologically, blocking the synthesis of thyroid hormone through medication. It can also be treated with radioactive compounds to destroy the gland, or by surgically removing a portion of the thyroid gland.

Are people with Down syndrome more prone to diabetes?

There is not sufficient data available at this point to know if there is increased risk for children with Down syndrome to develop type one diabetes as compared to the rate for their peers in the general population. However, research suggests that individuals who develop one type of endocrine autoimmune disorder, such as thyroiditis, are more likely to develop a second disorder, such as type one diabetes.

What is the status of research on use of the growth hormone for children with Down syndrome?

Use of growth hormone for children with Down syndrome is still in experimental stages. There are reports of increased rates of growth in children with Down syndrome who received the hormone for a brief period; however, these reports were not controlled studies, so there is no scientific evidence that long-term administration of the hormone would increase final height. To date, there is no convincing evidence that head circumference or mental functioning is improved by the use of growth hormone.

Can any growth hormone be administered to any Down syndrome child?

At present, doctors are licensed to prescribe growth hormone for individuals with Down syndrome only when there is a demonstrated deficiency of that hormone. It is strongly advised that such treatment be obtained only through a clinical trial until long-term benefits are demonstrated.

Endocrine Conditions and Down Syndrome Resource List

Organizations

American Thyroid Association
6066 Leesburg Pike, Suite 550
Falls Church, VA 22041
Telephone: 800-849-7643
Website: http://www.thyroid.org
Email: thyroid@thyroid.org

Chapters and Articles about Endocrine Conditions and Down Syndrome

Botero, D., Fleischman, A. "*Endocrinology*" in Medical Care for Children and Adults with Developmental Disabilities. Second Edition. Baltimore, MD: Brookes Publishing (2006). http://www.brookespublishing.com

Leshin, Len. "The Thyroid and Down Syndrome". Down Syndrome and Health Issues. http://www.ds-health.com/thyroid.htm

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Gastrointestinal Issues & Down Syndrome

Gastrointestinal Issues for Individuals with Down Syndrome

The gastrointestinal (GI) system includes all the parts of your body—from mouth to anus—that are involved in the digestion of food. Beginning in the newborn period, people with Down syndrome have an increased likelihood of developing medical conditions that interrupt or interfere with this digestion. Some of these medical issues can be managed by a person's primary care physician; others might require the added recommendations of a GI specialist.

What types of GI issues might my baby be born with? How do you correct them?

Approximately 3% of infants with Down syndrome are born with an **imperforate anus**, meaning that there is no open anus from which stool can be passed. This is easily identified when a physician examines a baby for the first time and can be corrected with a simple surgery.

Between 2-15% of infants with Down syndrome are born with **Hirschsprung Disease,** which results when the last part of their large intestine does not function properly due to a lack of certain nerve cells. As a result, children and cannot properly expel stool. Symptoms of Hirschsprung disease in early infancy include a swollen abdomen, vomiting, and an inability to expel stool. Children may also present later in life with severe constipation. If an infant has not stooled in the first few days, a physician might consider getting an X-ray. However, a definitive diagnosis is made by a rectal biopsy (removal of a small piece of rectal tissue for examination under the microscope). The treatment involves surgically removing the portion of colon that does not function properly.

If a newborn with Down syndrome has severe vomiting from birth, he or she might be among the approximately 5% of babies with Down syndrome who have a **duodenal obstruction,** which means that the first part of the small intestine—the duodenum—becomes blocked. Usually this occur because the duodenum developed differently during the fetal period. This can also occur when the pancreas, an organ that helps the body digest food, inappropriately wraps around the duodenum and occludes it. The end result is that digested food cannot pass through the duodenum; surgery is curative.

If a newborn with Down syndrome is bubbling up milk during feeds and has frequent choking episodes, he or she might be among the 1% of babies with Down syndrome who have a **tracheoesophageal fistula—**that is, an inappropriate connection between the trachea (the body's wind pipe) and the esophagus (the body's swallowing tube). With this condition, food inappropriately enters the wind pipe and the lungs. The diagnosis can be made by passing a tube down a baby's nose and/or through a chest X-ray. Surgical correction is curative.

Will by baby with Down syndrome be able to breastfeed?

Babies with Down syndrome oftentimes have low muscle tone (hypotonia) and sometimes have trouble forming a latch to breastfeed. However, with the help of lactation specialists, many mothers are able to provide all of the known benefits of breastfeeding to their baby with Down syndrome. More information is available from La Leche League International at: http://www.llli.org/FAQ/down.html.

What is reflux? What are the available treatments?

Reflux—medically known as gastroesophageal reflux disorder (GERD)—is a condition that results when acidic stomach contents travel backwards up the esophagus, a person's swallowing tube. All babies with and without Down syndrome have GERD; for some babies, however, the severity of symptoms warrant special attention.

Based on the current studies available, between 1-5% of people with Down syndrome have GERD and experience symptoms such as heartburn or intolerance with certain foods. In babies, reflux is typically expressed as intense back arching and crying during feeds. GERD typically results when the muscular ring at the end of the esophagus becomes relaxed, allowing the stomach contents to track backwards. Medication therapies result in significant improvements for most individuals. In certain cases, a consultation with a GI specialist might be helpful as additional testing can be done.

If my child is constipated, what might be the causes? And what are the treatment options?

Children with Down syndrome can be constipated for all of the same reasons that kids without Down syndrome become constipated—poor diet and lack of exercise, among many reasons. However, children with Down syndrome are also prone to three conditions that can result in constipation: (1) hypothyroidism, (2) Hirschsprung disease, and (3) Celiac Disease. **Hypothyroidism** results when the body's thyroid gland does not produce enough thyroid hormone, which regulates many activities in the body, including stooling. This can be diagnosed with a simple blood test and treated with synthetic thyroid hormone. **Hirschsprung disease** is described above and most often diagnosed within the first year of life. **Celiac disease** is a condition where the body cannot properly digest certain foods and is described next.

If none of the above three conditions explain your child's constipation, you shoudl work closely with your child's doctor to explore some laxative medications that are safe for children. In certain occasions, where the cause of constipation might be due to behavioral concerns, working with a developmental-behavioral specialist could also be helpful.

What is Celiac disease? Should my child be tested?

Celiac disease is a condition where the body is unable to properly digest barley, rye, and wheat products. As the condition can range from mild to severe, the symptoms can also vary including difficulty gaining weight, diarrhea, vomiting, or constipation. An initial diagnosis can be made through a simple blood test, but a definitive test requires a special procedure from a GI specialist.

Up to 16% of individuals with Down syndrome are believed to have Celiac disease. Because of this high percentage, all infants with Down syndrome between the ages of 2 and 3 should be screened for Celiac disease with the simple blood test. Left untreated, Celiac disease could result in malnutrition, decreased growth, and, in rare cases, intestinal cancer (lymphoma). The treatment is dietary and involves eliminating all barley, rye, and wheat from a person's diet. Working with a dietician is often helpful to families.

Is obesity a problem for people with Down syndrome? What are the options?

There have not been any large-scale studies quantifying the percentage of people with Down syndrome who are either overweight or clinically obese. However, most families and clinicians would agree that weight problems are common. Sometimes there are medical reasons to explain the obesity, such as hypothyroidism or a lower rate of metabolism. Additionally or alternatively, people with Down syndrome frequently consume too many calories and have little to no exercise.

Building healthy eating habits while a person is young is key to preventing obesity in adolescence and adulthood. Obesity has been linked to secondary health problems such as high blood pressure, obstructive sleep apnea, and diabetes. As such, paying close attention to the weight of a person with Down syndrome is of prime importance to his or her lifelong health. Oftentimes, working closely with a nutritionist is beneficial to families.

Special thanks to Brian Skotko, M.D., M.P.P., for preparing this piece.

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Gastrointestinal Issues & Down Syndrome

Organizations and Websites

Celiac Disease Foundation
13251 Ventura Blvd., #1
Studio City, CA 91604
Telephone: 818-990-2354
Fax: 818-990-2379
Email: cdf@celiac.org Website: http://www.celiac.org

Celiac dot Com
Online contact available at website: http://www.celiac.com

International Foundation for Functional Gastrointestinal Disorders
P.O. Box 170864
Milwaukee, WI 53217
Telephone: 414-964-1799 or 888-964-2001
Email: iffgd@iffgd.org Website: http://www.iffgd.org

National Organization for Rare Disorders (NORD)
55 Kenosia Avenue
P.O. Box 1968
Danbury, CT 06813-1968
Telephone: 203-744-0100 or 800-999-6673 (*voicemail only*)
Email: orphan@rarediseases.org Website: http://www.rarediseases.org

NIH's National Digestive Diseases Information Clearinghouse
Office of Communications and Public Liaison, NIODK, NIH
Building 31, Room 9A06
31 Center Drive, MSC 2560
Bethesda, MD 20892-2560
Telephone: 301-654-3810 or 800-891-5389
Email: nddic@info.niddk.nih.gov Website: http://www.niddk.nih.gov

Books and Articles on Gastrointestinal Issues and Down Syndrome

Guthrie Medlen, J.E. The Down Syndrome Nutrition Handbook. Lake Oswego, OR: Phronesis Publishing (2006). http://www.downsyndromenutrition.com/phronesis

Korn, D. Kids with Celiac Disease: A Family Guide to Raising Happy, Healthy, Gluten-Free Children. Bethesda, MD: Woodbine House Publishing (2001). http://www.woodbinehouse.com

Leshin, Len. "*Celiac Disease and Down Syndrome*". Down Syndrome and Health Issues. http://www.ds-health.com/celiac.htm

Sanderson, Sheri L. Incredible Edible Gluten-Free Food for Kids. Bethesda, MD: Woodbine House Publishing (2002). http://www.woodbinehouse.com

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The Heart & Down Syndrome

Abnormalities of the cardiovascular system are common in Down syndrome. Approximately half of all infants born with Down syndrome have a heart defect. Many of these defects have serious implications and it is important to understand them and how they may affect the child so that appropriate medical may be provided.

What are the most common heart defects in children with Down syndrome?

The most common defects are Atrioventricular Septal Defect (formally called Endocardial Cushion Defect), Ventricular Septal Defect, Persistent Ductus Arteriosus and Tetralogy of Fallot.

What is an Atrioventricular Septal Defect?

An Atrioventricular Septal Defect is caused by a failure of tissue to come together in the heart during embryonic life. This results in a large opening in the center of the heart, with usually a hole between the two pumping chambers (a Ventricular Septal Defect) and between the two collecting chambers (an Atrial Septal Defect) as well as abnormalities of the two atrioventricular valves, the mitral and tricuspid valves. Of those children with Down syndrome who are born with congenital heart disease, an antrioventricular septal defect is the most common. In less severe cases, Ventricular Septal Defects and Atrial Septal Defects can also occur separately.

What is Persistent Ductus Arteriosus?

The ductus arteriosus is a channel between the pulmonary artery and the aorta. During fetal life it diverts blood away from the lungs because prenatal blood is already oxygenated from the mother. After birth this channel usually closes on the first day of life. If it does not close, it is termed "persistent" and results in an increased flow of blood into the lungs.

What is Tetralogy of Fallot?

This is a term given to a heart condition composed of four abnormalities: 1) ventricular septal defect 2) a narrowing of the passage from the right venrtical to the lungs 3) an overenlarged right ventrical because of the backup of blood and 4) an overenlarged aorta, which carries blood from the left ventrical to the body.

What is the relationship of heart defects to the respiratory system?

The lungs of children with Down syndrome do not develop as fully as in the general population. Consequently, the growth of blood vessels throughout the lungs is limited. The narrowed arteries of the lungs hold potential for lasting consequences due to the increased pressure and flow of blood through the lungs.

How are the defects diagnosed?

Some children with Down syndrome and major heart defects will present with heart failure, difficulty breathing and failure to thrive in the newborn period; however, because in some children the defect may not be at first apparent, it is important that all children born with Down syndrome, even those who have no symptoms of heart disease, should have an echocardiogram in the first two or three months of life. Some heart conditions are also identified during prenatal ultrasounds.

What is the recommended treatment?

Heart surgery to correct the defects is recommended and it must be done before age five or six months in order to prevent lung damage. Although the complexity of the defects raises the risk of surgery slightly above that of surgery on children without Down syndrome, successful surgery will allow many children with heart conditions to thrive as well as any child with Down syndrome who is born with a normal heart. There may be residual defects (such as imperfect valves, in cases of Atrioventricular Septal Defect), but their effect on health is often minimal.

What are the guidelines for choosing a hospital?

Look for a medical center in a major metropolitan area which has experience in open heart surgery on infants, and experience in operating on infants with Down syndrome in particular. A hospital which conducts at least 10 such operations a year, with a good survival rate, would be considered an experienced hospital.

What should be considered in selecting a surgical team?

Again, experience, together with the parents' rapport with the physician, should be the determining factor in the choice. It is important that an experienced anesthesiologist be selected, as children with Down syndrome often have airway problems.

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Resources & Organizations:

American Heart Association
7272 Greenville Avenue
Dallas, TX 75231
Telephone: 800-242-8721
Website: http://www.americanheart.org

Down's Heart Group
Email: info@dhg.org.uk
Website: http://www.dhg.org.uk
http://groups.yahoo.com/group/downs-heart *is an online support group for parents of children who have Down syndrome and heart defects*

Mended Little Hearts *(support for parents of children with heart defects)*
7272 Greenville Avenue
Dallas, Texas 75231
Telephone: 888-432-7899
Email: mlh@mendedhearts.org
Website: www.mendedhearts.org

Books and Chapters About the Heart and Down Syndrome

Cohen, W., Nadel, L., Madnick, M. (Eds.). Down Syndrome: Visions for the 21st Century. Hoboken, NJ: John Wiley and Sons, Inc. (2002).

"History of Management of Congenital Heart Disease". Dooley, K. In: Medical Care for Children and Adults with Developmental Disabilities. Second Edition. Rubin, I. and Crocker, Allen (Eds.) Baltimore, MD: Paul H. Brookes Publishing. (2006). www.brookespublishing.com

Kramer, G. and Maurer, S. The Parent's Guide to Children's Congenital Heart Defects: What They Are, How to Treat Them, How to Cope with Them. New York, NY: Three Rivers Press. (2001).

"Medical Concerns and Health Issues" Pueschel, S. In: A Parent's Guide to Down Syndrome: Toward a Brighter Future. Revised Edition. Pueschel, S. (Ed.) Baltimore, MD: Paul H. Brookes Publishing. (2000). www.brooksepublishing.com

"Spectrum of Heart Disease". Mahle, W. In: Medical Care for Children and Adults with Developmental Disabilities. Second Edition. Rubin, I. and Crocker, Allen (Eds.) (2006). Baltimore, MD: Paul H. Brookes Publishing. www.brookespublishing.com

ndss Mental Health & Down Syndrome

Mental Health Issues and Down Syndrome

What are the major mental health related concerns in persons with Down syndrome?

At least half of all children/adults with Down syndrome face a major mental health concern during their life span. Children/adults with multiple medical problems experience an even higher rate of mental health problems. The most common mental health concerns include: general anxiety, repetitive and obsessive-compulsive behaviors; oppositional, impulsive, and inattentive behaviors; sleep related difficulties; depression; autism spectrum conditions; and neuropsychological problems characterized by progressive loss of cognitive skills.

The pattern of mental health problems in Down syndrome vary depending on the age and developmental characteristics of the child/adult with Down syndrome as follows.
Young and early school age children with limitations in language and communication skills, cognition, and non-verbal problem solving abilities present with increased vulnerabilities in terms of:

- Disruptive, impulsive, inattentive, hyperactive and oppositional behaviors (raising concerns of coexisting oppositional disorder and ADHD)
- Anxious, stuck, ruminative, inflexible behaviors (raising concerns of co-existing generalized anxiety and obsessive-compulsive disorders)
- Deficits in social relatedness, self-immersed, repetitive stereotypical behaviors (raising concerns of co-existing autism or pervasive developmental disorder)
- Chronic sleep difficulties, daytime sleepiness, fatigue, and mood related problems (raising concerns of co-existing sleep disorders and sleep apnea)

Older school age children and adolescents, as well as young adults with Down syndrome with better language and communication and cognitive skills presenting with increased vulnerability to:

- Depression, social withdrawal, diminished interests and coping skills
- Generalized anxiety
- Obsessive compulsive behaviors
- Regression with decline in loss of cognitive and social skills
- Chronic sleep difficulties, daytime sleepiness, fatigue, and mood related problems (raising concerns of co-existing sleep disorders and sleep apnea)

Older adults present with increased vulnerability to:

- Generalized anxiety
- Depression, social withdrawal, loss of interest, and diminished self-care
- Regression with decline in cognitive and social skills
- Dementia

All these changes in behavior often seem to occur as a reaction to (or triggered by) a psychosocial or environmental stressor, e.g., illness in, separation from, or loss, of a key attachment figure.

Who should you turn to for help for assessment and treatment of mental health concerns?

Many families live in areas without a mental health professional skilled in working with children/adults with Down syndrome. We therefore recommend the following approach for families:

- Consider making a preliminary search in your area for potential providers with experience in working with children/adults with developmental disorders. This may include asking your primary care provider,

inquiring at work with your employee benefits officer responsible for your medical coverage to give you a list of providers who indicated an interest in evaluating children/adults with developmental disorders. It always helps greatly if you already have a primary care physician who can make referral recommendations or who already has someone in mind who similarly can make an appropriate referral for you. If you have access to the Internet you visit the website for your medical coverage provider and search for professionals in your geographic region who indicated an expertise in developmental disorders. Finally, you may consider calling the local department or case coordinator in your district for additional services that may be available in your area.

- It is always worth making an initial consultation visit to familiarize the child/adult with the professional and to see if this is a good match for your needs. Such an introductory visit is helpful as it enables the child/adult with Down syndrome also to feel comfortable with the place, provider, and it also enable you to get a timely appointment in a crisis situation in the future when a critical situation arises. It is often much more difficult to get an initial appointment and to be able to do so in an acute situation has become increasingly difficult, especially in well known centers.
- Please remember that the ideal mental health provider skilled in Down syndrome is someone who has knowledge of developmental disorders and who also has had experience in working with children. It may be advisable to first seek a mental health provider who works in a pediatric medical center or who works in close proximity to a pediatric practice.
- In geographic locations with limited proximity to such services it is always worth remembering that each state in the U.S. has what is known as a University Center of Excellence in Developmental Disabilities (UCEDD) which is part of the Association of University Centers in Developmental Disabilities (www.aucd.org). Many of these programs have been in existence for over 30 years and are located in tertiary care centers with interdisciplinary services that include mental health professionals (child psychiatrists, psychologists, social workers), as well as developmental-behavioral pediatricians. The UCEDD programs can also provide advice regarding referral to adult service in the community and help locate mental health providers that have expertise in working with individuals with Down syndrome.
- If it is very important to select a psychiatric provider with expertise in medication management with individuals with developmental disorders, it is critical that you find someone who has worked in close proximity to a medical practice, or agency serving the needs of individuals with developmental disorders.

If my child has a new "behavior problem," are there some medical causes that we should rule out first?

This is a common question that many medical as well as mental health providers are asked by concerned parents. There are a number of baseline tests that need to be completed to rule our medical conditions that are often associated with children/adults with Down syndrome presenting with a "behavioral problem". Among these we recommend considering the following conditions:

- Thyroid function tests can be completed by the primary care provider or by a developmental and behavioral pediatrician or even a psychiatrist as part of an initial assessment.
- Sleep related difficulties need to be evaluated by the primary care provider, developmental-behavioral pediatrician, or a psychiatrist as part of an initial assessment with referral to a sleep disorders clinic or laboratory as needed to rule obstructive sleep apnea - see further discussion below).
- Underlying contribution of constipation or bowel successful related difficulties need also to be ruled out by the primary care or developmental-behavioral pediatrician with interventions as may be necessary and referral to a nutritionist for counsel. There is a great opportunity to use healthy diet as a tool to reinforce positive behaviors.
- As part of the comprehensive checklist of potential medical conditions it is important also to make sure that the child/adult with Down syndrome has been evaluated for hearing (audiology), vision (ophthalmology), anemia (hematology) and GERD (GI).

Finally, caveats or steps to consider in addressing any of the above potential medical concerns in the context of treatment of "behavioral problems" include the following:

- Step 1: Emotional/behavioral problems in children/adults with Down syndrome occur commonly and are not always due to an underlying medical condition. Nevertheless, these medical conditions associated

in children/adults with Down syndrome need to be ruled out as part of a comprehensive assessment approach.

- Step 2: The medical conditions, even if they may in themselves not cause the emotional/behavioral issues, may nevertheless *exacerbate* them, or make the child/adult with Down syndrome *resistant* to treatment of the emotional/behavioral problem.
- Step 3: Correction of a medical condition, e.g., hypothyroidism, may not remove the underlying emotional/behavioral issues, and the emotional/behavioral issues still need to be treated concurrently. Therefore, a child/adult with hypothyroidism plus depression is unlikely to respond to treatment of depression with antidepressant medication alone, unless the hypothyroidism is corrected, i.e., there is an "interaction" between the medical and emotional/behavioral conditions.

What are the symptoms of generalized anxiety, obsessive-compulsive disorder, and depression in Down syndrome? How are they diagnosed and treated?

Generalized Anxiety: These are the most prominent presentations among children/adults with Down syndrome. This manifests as an increased level of both *baseline* as well as *situational* anxiety with clear cut stressors for each. Situational anxiety is often manifest during transitions and anticipation of new situations, e.g., transitions from home to school, transit, meal or bed times, as well as during novel and unfamiliar situations with uncertain expectations in the environment.

Obsessive Compulsive Symptoms: Increased level of restlessness and worry may lead the child/adult to behave in a very rigid manner, even resulting in a state of being "*stuck," as* is often reported by caregivers where the child or adult needs to follow familiar routines in these situations. They also engage in repetitive, compulsive, as well as ritualistic behaviors that raise the question of *obsessive-compulsive disorder*. The child/adult under these circumstances tends often to be unhappy, fearful, and the two states - generalized anxiety and obsessive-compulsive behaviors - may often co-exist. The disruptive, oppositional and inattentive child with Down syndrome often does not tend to be unhappy, but rather quite silly, happy, and excited. The problems are quite challenging for parents/caregivers to navigate, as the child/adult with Down syndrome with generalized anxiety or obsessive-compulsive profile has a tendency to be stuck, frozen, and require great degree of negative attention that, in turn, is reinforced, and continues in a vicious cycle.

Differentiating Anxiety and ADHD Symptoms in Young Children: Unlike in children with Down syndrome with impulsive, oppositional, and attention deficit profile, the restlessness, fidgeting, and compulsiveness associated with generalized anxiety state has an identifiable onset with a more intermittent course. There is a need to take a detailed history in all these situations in order to identify the source or environmental triggers contributing to the anxiety in relation to change in immediate home/school/work environment. In such circumstances assessment of antecedents, behaviors, and consequences (ABCs) and development of a behavioral modification and management plan is essential. The use of antidepressants or anti-anxiety medications may help and should be reserved for more persistent and serious level of symptoms.

Depressive Symptoms: The children/adults with symptoms of *depression* often present with extreme social withdrawal, sad (but not labile) affect, and inability to enjoy many activities they used to love. The parents/caregivers often report that the child/adult's demeanor had not previously been like that. Disrupted sleep commonly co-occurs in both depression and anxiety states and do not necessarily help us to distinguish between them. A most remarkable aspect of depression in children/adults with Down syndrome is its association with environmental noxious triggers. These may include previously unrecognized medical illness or pain or psychosocial stressors, e.g., older sibling moving to college, sudden or chronic illness in a family member, death of a long household pet, absence of a teacher (leave, illness). All these ordinary events seem extraordinary for children/adults with Down syndrome with a disproportionate psychological impact, as compared to a typical person under similar circumstances. In summary, children/adults with Down syndrome remain exquisitely sensitive to changes in their environment which they often perceive unfavorably. We recommend that if any negative changes are to be anticipated that supportive counseling services and supports be put in place in anticipation of their impact. Attempt to treat persistent depression in the context of ongoing stress with pharmacological intervention is often futile without individual supports. Combination treatment involving both psychosocial and pharmacological components is needed. The argument for pharmacological intervention is strengthened if the child/adult with Down syndrome is deemed

to already have a biological vulnerability (e.g., positive family history, previous episode of depression, concurrent medical illness).

What are the inattentive, impulsive, hyperactive, and disruptive symptoms in persons with Down syndrome? How are they diagnosed and treated?

Children/adults with Down syndrome often have significant processing difficulties and present with a very remarkable difficulty in sustaining attention on tasks. In children with greater cognitive and receptive-expressive language deficits, especially for younger age groups, the difficulties in attention are often accompanied with impulsive and hyperactive behaviors. This pattern of inattention, impulsivity and motor hyperactivity is consistent with a diagnosis of Attention Deficit Hyperactivity Disorder. For this reason, many children with such characteristic behaviors are treated with stimulant medications.
The response of children/adults with Down syndrome has not yielded encouraging results. In a sub-population of children/adults with Down syndrome there is a definite adverse behavioral activation in response to these medications. The most commonly observed adverse effects include: irritability, agitation, aggressive behaviors, transitional anxiety, and sleep related problems. Parents/caregivers need to be forewarned about these adverse effects since it can occur very soon after the initiation of treatment and can be very troubling for them to witness. A small group of children with ADHD symptoms may nevertheless benefit from stimulant medications, but even for them they may increase in anxiety, as well as obsessive compulsive symptoms. For this reason, the main emphasis in treatment of ADHD-like symptoms needs to focus on behavioral and therapeutic strategies to enhance adaptive functioning and performance in the home and classroom settings.
For children with high-degree impulsivity and disruptive behaviors the use of low dose of clonidine has been helpful, but this medication may also be limited in its efficacy as it may lead to daytime drowsiness in some children. It should be emphasized that the use if clonidine, per se, can be effective only in addressing impulsive, hyperactive and disruptive behavior, and does not necessarily improve primary attention. Clonidine administered at bed time may also help the child to settle down to sleep.

What are the behavioral concerns related commonly associated with chronic sleep difficulties? How is this assessed and treated?

Children/adults with Down syndrome commonly experience a range of sleep-related difficulties either as primary sleep disorder or associated with mental health problems (e.g., generalized anxiety and mood disturbances). Irrespective of the etiology, sleep difficulties impair the ability of a child/adult with Down syndrome to maintain alertness and attention during the day, as well as maintain better control, e.g., frustration tolerance. Chronic sleep difficulties in children/adults with Down syndrome need to be evaluated thoroughly by interdisciplinary team in order to rule out any contributory medical conditions.
Children/adults with Down syndrome, in particular, are at increased risk for development of obstructive sleep apnea with mild to moderate cessation of breathing during sleep that leads to reduction of oxygen saturation in the blood. Although the diagnosis of sleep apnea is suspected on the basis of history that often includes evidence for periods of daytime sleepiness, fatigue, it is necessary to conduct further tests to confirm this diagnosis by means of referral for a sleep study at a sleep disorder laboratory often available in major medical centers.

What are major environmental triggers of behavioral and emotional difficulties?

Children/adults with Down syndrome are often exquisitely sensitive to psychosocial and environmental stressors. Illness or loss of close loved ones and family is particularly devastating and almost invariably leads to a complicated grief reaction during which time the child/adult with Down syndrome may experience regressive change in their ability to think, reason, remember, process information, and learn. The psychosocial and environmental triggers also lead to a state of generalized anxiety, obsessive compulsive symptoms, and depression and sleep difficulties. They may be associated with weight loss, poor self care, and inability to be motivated to attend school or go to work placements. If the situation persists and there is no concerted attempt to intervene with psychosocial counseling, treatment with appropriate medications, and behavioral interventions, the mental state may persist and be associated with longer term decline in psychosocial and cognitive functioning.

Is oppositional defiant disorder common in children/adults with Down syndrome?

Many children/adults with Down syndrome have a wonderful disposition: they are fun loving and their interactions generally involve teasing, making jokes, giggly outbursts that often may also lead to intrusive, uninhibited social behaviors. Many respond to structure and behavioral interventions with clear-cut reinforcements and rewards. Sometimes these behaviors become out of control and take over the overall interactions. They become increasingly oppositional, unable to listen, and quite single-minded and self-immersed, e.g., sitting down or lying down and refusing to get up, or continuing in a self-directed activity with disregard of consequences at meal, bath and bed times, or during transitions. The oppositional behaviors occur in individuals with all levels of cognitive and language skills, but is more difficult to manage in those with greater receptive-expressive communication skills. In the classroom setting, behavioral management and 1:1 aide may help to keep the situation in better control and to enable learning. Oppositional behavioral problems in children with receptive-expressive and cognitive limitations also tend to be associated with increased level of impulsive and hyperactive behaviors and often co-occur with ADHD symptoms.

Are mood and bipolar disorders common in children/adults with Down syndrome?

A comprehensive approach is needed in assessment of a child/adult with Down syndrome presenting with mood instability. It is essential to rule out any underlying medical and neurological conditions, and especially to consider the possibility of adverse effect of medications that may lead to secondary mood instability.
A young child with Down syndrome that presents with persistent oppositional, impulsive, disruptive, irritable, and aggressive behaviors should be considered under a possible mood disorder. In our clinical experience the coexistence of true bipolar disorder and Down syndrome is relatively unknown. The use of anticonvulsant medications (as mood stabilizers) need only be considered under careful supervision. Likewise, the use of atypical neuroleptic medications ought to be considered only as a last resort, again, with careful monitoring of their potential side effects. These latter medications tend to be limited in efficacy and should be used sparingly and in low doses. Since children/adults with Down syndrome are already at increased risk of weight gain over their lifespan, the increased appetite leading to weight gain associated with atypical neuroleptic medications, can be destabilizing. Concurrent behavioral and nutritional interventions are therefore always essential.

Are we entering a new age with improved assessment of mental health concerns in children/adults with Down syndrome?

The assessment of mental health concerns in children/adults has improved considerably in recent years. There is now wider range of available screening and diagnostic tools for assessment of mental conditions across different developmental age groups in terms of measurement of domains such as, non-verbal problem-solving abilities, language and communication and adaptive and behavioral functioning. Much of our current knowledge is based on clinical experience and although emphasis varies depending on the orientation of each provider, e.g., behavioral modification, pharmacological intervention, and social skills training, parents would be wise in seeking a holistic philosophy for integrated care (medical-mental health, behavior-pharmacology-social skills).
Despite the fact that many individuals with Down syndrome experience significant cognitive delays and other associated physical conditions, they have a very wide range of abilities, and each individual develops at his or her own pace. Even though they may be delayed in their progression, many achieve meaningful developmental milestones and lead enjoyable and highly enriching lives. There is a need to develop a better evidence base with expanded research in mental health aspects of Down syndrome. The increased awareness of the mental health issues bodes very well for the future.

NDSS thanks special guest author Kerim Munir, M.D., MPH, D.Sc., for preparing this piece.

ndss **Mental** Health & Down Syndrome

Mental Health and Down Syndrome Resource List

Organizations and Websites

The Association for Positive Behavior Support
P.O. Box 328
Bloomsburg, PA 17815
Telephone: (570) 389-4081
Fax: (570) 389-3980
E-mail: tknoster@bloomu.edu Website: http://www.apbs.org/

National Information Center for Children and Youth with Disabilities (NICHCY)
P.O. Box 1492
Washington, DC 20013
Telephone: (800) 695-0285
Fax: (202)884-8441
E-mail: nichcy@aed.org Website: http://www.nichcy.org

Positive Behavioral Interventions and Support Technical Assistance Center
Website: http://www.pbis.org

Research and Training Center on Family Support and Children's Mental Health
Portland State University
P.O. Box 751
Portland, OR 97207-0751
Telephone: 503-725-4040
Fax: 503-725-4180
E-mail: janetw@pdx.edu Website: http://www.rtc.pdx.edu

Books about Mental Health

Bambara, L. and Kern, L. Individualized Supports for Students with Problem Behaviors. New York, New York: The Guilford Press. (2005).

McGuire,D,& Chicoine, B. Mental Wellness in Adults with Down Syndrome. Bethesda, MD: Woodbine House (2005). http:// www.woodbinehouse.com.

ndss

The Neurology of Down Syndrome

Neurology, the study of the human nervous system and its disorders, is important in the clinical care of persons with Down syndrome. The nervous system is always affected in Down syndrome. Among the most common involvements are development disabilities (retardation in mental development and motor capabilities), hypotonia, atlantoaxial dislocation and seizures.

What does the term "developmental disabilities" mean in relation to Down syndrome?

This term refers to incomplete development of the brain, which leads to both intellectual disability and to slowed and/or incomplete mastery of physical coordination.

What is known about brain pathology in regard to developmental difficulties?

Pathology in the Down syndrome brain includes a slightly smaller brain size for age, a shorter diameter for the anterior-posterior brain measurement, an unusually steep slope to the posterior portions of the brain and an insufficiently developed superior temporal gyrus. It is not known in what way these features contribute to the developmental disabilities of Down syndrome.

Through research has any progress been made in the pathology of the brain of a person with Down syndrome?

Research in this area is being actively pursued, especially since it may yield clues to Alzheimer's disease and to the neuroscientific effects on learning and memory.

Is IQ (Intelligence Quotient) a meaningful measure in Down syndrome?

No. IQ is not an adequate measure of the functional status of people with Down syndrome. For example, individuals with Down syndrome may have difficulty with grammar, but understand individual words, or have a sense of when to speak, at a par with their mental age. IQ tests to not measure many of the abilities that people with Down syndrome possess. However, obtaining an IQ test is sometimes necessary to receive appropriate services.

What can be done to help people with Down syndrome to achieve their full potential in the face of such disabilities?

A variety of intervention programs, designed to begin in infancy and continue throughout learning years, help children with Down syndrome maximize their capabilities.

What is hypotonia?

Hypotonia refers to the reduced muscle tone that occurs in virtually all infants with Down syndrome. It is commonly seen in the flexor group, muscles which act to flex a joint. The muscle weakness ranges from mild to moderate to severe.

How is hypotonia diagnosed?

Common hypotonic symptoms are a lagging head when the child is pulled into a sitting position and arching of the back when carried upright or lying on the stomach. In addition, hypotonic children will tire more easily and adopt movement patterns requiring the least expenditure of energy.

Are there any effective interventions for hypotonia?

Hypotonic conditions tend to improve with age. Gross motor programs offered by occupational and physical therapists have been shown to diminish hypotonic symptoms. These programs improve large body movements, such as walking, turning, sitting, standing and climbing stairs. Enrollment of children in an early intervention program which includes gross motor programs is recommended at the earliest feasible age, generally between four and six weeks of age.

What is atlantoaxial dislocation?

Atlantoaxial dislocation refers to a problem caused by hypotonia. The ligaments at the first two cervical vertebra are more relaxed than they should be, putting the individual at risk of spinal cord compression and injury.

How common is atlantoaxial dislocation?

The cervical spine instability occurs in 10 to 20 percent of children with Down syndrome; however, actual spinal compression is rare.

What are the symptoms of atlantoaxial dislocation?

If symptoms are present, they include neck pain, change in gait, onset of weakness in the extremities, spasticity, limited neck movement and bowel/bladder incontinence (particularly after toilet training has been accomplished). However, most children with x-ray evidence of atlantoaxial dislocation have no apparent symptoms.

What preventive measures should be taken?

Since most children with atlantoaxial dislocation do not exhibit symptoms, lateral cervical x-rays are recommended for all children with Down syndrome between the ages of 3 and 5. If the instability is present, but there are no symptoms, the appropriate precaution is to limit "high risk" activities which might over-stress the neck. These activities include high-jumping, diving, gymnastics, trampoline, and butterfly strokes in swimming. To devise a safe exercise program for a symptom-free child with atlantoaxial dislocation, consult your doctor.

Is there any treatment for atlantoaxial dislocation?

If symptoms are present, the atlantoaxial joint can be surgically fused.

What is the incidence of seizures in Down syndrome?

In the young child with Down syndrome, seizures are no more common than in the general population. Beginning at age 20-30, the incidence of seizure disorder rises substantially in the Down syndrome population. Research is ongoing as to whether the frequencies of seizures in individuals with Down syndrome is related to the aging of the brain.

What is the typical seizure for a person with Down syndrome?

Seizures for individuals with Down syndrome commonly look like epileptic seizures: jerking of arms and legs and loss of consciousness. Seizures can also take a mixed form, with staring spells and momentary lapses of attention.

What is the treatment for seizures?

Seizures can be controlled with standard anti-convulsant medication.

ndss The Neurology of Down Syndrome

The Neurology of Down Syndrome Resource List

Books on Neurology

Lubec, G. Protein Expression in Down Syndrome Brain (Journal of Neural Transmission Supplementum). New York, NY: Springer Publishing (2002). http://www.springerpub.com

Rondal. J. and Perera, J. Down Syndrome: Neurobehavioural Specificity. San Francisco, CA: Wiley Publishing (2006). http://www.wiley.com/wileycda

Rubin, L. and Crocker, A. Medical Care for Children and Adults with Developmental Disabilities. Second Edition. Baltimore, MD: Brookes Publishing (2006). http://www.brookespublishing.com

Wellness and Healthy Living Resource List

Organizations and Websites

Academy of Dentistry for Persons with Disabilities
Special Care Dentistry
401 North Michigan Avenue, Suite 2200
Chicago, Il 60611
Telephone: (312) 527-6764
Fax: (312) 673-6663
E-mail: SCDA@SCDonline.org Website: http://www.scdonline.org

American Speech-Language Hearing Association
10801 Rockville Pike
Rockville, MD 20852
Telephone: (301) 897.5700 or (800) 638-8255
Website: http://www.asha.org

LaLeche League International
1400 North Meacham Road
Schaumberg, IL 60173
Telephone: (800) LALECHE or (847) 519-7730
Website: http://www.lalecheleague.org

National Center on Physical Activity and Disability
1640 W. Roosevelt Road
Chicago, IL 60608-6904
Telephone: (800) 900-8086
E-mail: ncpad@uic.edu Website: http://www.ncpad.org

Books and DVDs

Durand, V. M.. Sleep Better: A Guide to Improving Sleep for Children with Special Needs, Baltimore, MD; Brookes Publishing (1998). http://www.brookespublishing.com

Guthrie Medlen, J.E. The Down Syndrome Nutrition Handbook. Lake Oswego, OR
Phronesis Publishing (2006). http://www.downsyndromenutrition.com/phronesis/

McGuire,D,& Chicoine, B. Mental Wellness in Adults with Down Syndrome. Bethesda, MD: Woodbine House (2005). http://www.woodbinehouse.com.

Sanderson, Sheri L. Incredible Edible Gluten-Free Food for Kids. Bethesda, MD: Woodbine House Publishing (2002). http://www.woodbinehouse.com

Schermerhorn, Will (Producer). Kids with Down Syndrome: Staying Healthy and Making Friends. Bethesda, MD: Woodbine House (2005). http://www.woodbinehouse.com

Feeding Resource List

Organizations and Websites

LaLeche League International
1400 North Meacham Road
Schaumberg, IL 60173
Telephone: (800) LALECHE or (847) 519-7730
Website: http://www.lalecheleague.org

New Visions
1124 Roberts Mountain Road
Faber, VA 22938
Telephone: (800) 606-7112
E-mail: mealtime@new-vis.com
Website: http://www.new-vis.com

Books and Articles

Fiocca, Sharon. Oral Motor & Feeding Skills. (1999; Updated 2007) Reprinted at http://www.riverbendds.org/fiocca.html

Lowman, D. K, Murphy, S.M. The Educator's Guide to Feeding Children with Disabilities. Baltimore, MD: Paul H. Brookes Publishing Co (1998). http://www.brookespublishing.com/

Sleep Issues Resource List

Organizations and Websites

American Sleep Apnea Association.
1424 K Street NW, Suite 302
Washington, DC 20005
Telephone: (202) 293-3650
Fax: (202) 293-3656
Email: asaa@sleepapnea.org Website: http://www.sleepapnea.org/

American Sleep Disorders Association
1610 14th Street NW
Rochester MN 55901
Telephone: 507-287-6006
Email: asda@millcomm.com

Center for Research in Sleep Disorders
1275 East Kemper Rd.
Cincinnati OH 45246
Telephone: 513-671-3101
Email: ggaz@tristatesleep.com

National Sleep Foundation
1522 K Street, NW, Suite 500
Washington, DC 20005
Telephone: (202) 347-3471
Fax: (202) 347-3472
Website: http://www.sleepfoundation.org/site/c.huIXKjM0IxF/b.2417141/k.C60C/Welcome.htm

Books and Articles

Durand, V. M.. Sleep Better: A Guide to Improving Sleep for Children with Special Needs, Baltimore, MD; Brookes Publishing (1998). www.brookespublishing.com

Leshin, Len. “Obstructive Sleep Apnea and Down Sydrome”. Down Syndrome: Health Issues. http://www.ds-health.com/apnea.htm

“Obstructive Sleep Apnea in Children with Down Syndrome”. Capitol Sleep Medicine Newsletter. September 2007. Volume 2 Number 9. www.capitolsleepmedicine.com

Obstructive Sleep Apnea & Down Syndrome

Airway obstruction and Sleep Apnea

Airway obstruction is common in children with Down syndrome, with some studies suggesting that nearly all persons with Down syndrome have some form of sleep-related obstruction. Loss of sleep due to apnea and even poor quality sleep due to sleep disordered breathing can result in sleepiness, disturbances in fine motor skills, and also affects behavior and learning. Many with sleep disorders fall asleep with passive activities such as riding in the car or school bus.Long term complications of sleep apnea include systemic hypertension, pulmonary hypertension, heart failure, and even death.

Obstructive sleep apnea occurs when the airway is blocked during sleep. This can be caused by the small upper airway, large adenoids and tonsils, obesity, collapse of the airway due to hypotonia of the muscles of the throat, and increased secretions that can be characteristic of persons with Down syndrome. Obstruction can also occur from glossoptosis, a condition where a relatively large tongue falls back into a smaller airway during sleep.

Obstructive sleep apnea is often overlooked by caregivers and medical professionals, as sleep disturbances often occur unobserved or have been present for so long that parents assumed that was “normal” for their child.
A comprehensive clinical exam, X-ray, and thorough sleep study should be conducted if sleep apnea is suspected.

Airway obstruction can be treated both medically and surgically, and sometimes both treatments are necessary. Saline spray can keep the airway clear. Other medical options include the use of a Continuous Positive Airway Pressure (CPAP) machine during sleep, which provides some pressure with each breath, keeping the airway open while a person sleeps. Weight reduction may also help address sleep apnea. Surgically, removal of the tonsils and adenoids (T&A) is the first line of treatment of airway obstruction and sleep apnea in children with Down syndrome. Although removal of the tonsils and adenoids is usually curative of most sleep apnea in child, more recent studies suggest that this is not always the case with individuals with Down syndrome and further evaluation and treatment may be needed after T&A.

Sleep Apnea Confirmed Common in Children with Down Syndrome

Parents of children with Down syndrome weak to identify signs of sleep apnea

Monday, May 08, 2006

Children with Down syndrome are known to have a greater risk for obstructive sleep apnea. A new study by researchers at Cincinnati Children's Hospital Medical Center shows that few parents of children with Down syndrome are in tune with whether their child has obstructive sleep apnea.

The study appears in the April 17 issue of the Journal of the American Medical Association.

Obstructive sleep apnea syndrome is defined by sleep abnormalities that consist of complete and partial upper airway obstruction during sleep, a reduction in the amount of air entering the lungs with an abnormally high level of carbon dioxide in the blood, and oxygen loss.

Although obstructive sleep apnea is seen in only 0.7 to 2 percent of all children, a previous study based on 53 children and adults with Down syndrome between the ages of four weeks and 51 years old (an average of seven years old), found an incidence of sleep abnormalities as high as 100 percent in some cases. With this very high incidence in children, Sally R. Shott, MD, a pediatric otolaryngologist at Cincinnati Children's and principal investigator of the JAMA study, questioned whether all children with Down syndrome should be objectively evaluated for sleep abnormalities, obstructive sleep apnea, and at what age this evaluation should be done.

"Our results point to the need for objective testing for the presence of obstructive sleep apnea in children as young as three to four years old. Because there is a high incidence of sleep disorders in children with Down syndrome, clinical sleep studies are recommended even if the parents report no sleep problems in their child," Dr. Shott said.

Dr. Shott and her colleagues investigated the incidence of obstructive sleep apnea in 65 children with Down syndrome who were followed in a longitudinal, five-year study from the ages of two to five years old. The children in the study underwent a polysomnogram, or sleep study, between three and four years old. Researchers also examined the ability of parents to identify sleep abnormalities in their child.

They found that in general, parents of children with Down syndrome significantly underestimated the severity of their child's sleep disturbances. Overall, 69 percent of parents reported no sleep problems. Yet, 57 percent of children expressed abnormal sleep patterns. In the children with abnormal sleep study results, only 23 percent correctly predicted a problem. In a previous study, only 32 percent of parents suspected their child had obstructive sleep apnea, despite a 100 percent incidence of abnormal studies.

There is concern that an excessive number of arousals may lead to fragmented sleep and sleep deprivation. Excessive sleep arousals and sleep fragmentation have been linked to symptoms usually associated with sleep deprivation such as daytime sleepiness, lack of energy and lack of initiative.

Increased arousals have been associated with decreased neurocognitive abilities and lower results on IQ testing. It has been suggested that the increased arousal rate seen in children with Down syndrome may affect their daytime function and could exacerbate learning or behavior disorders. In children with Down syndrome, behavior and learning disabilities associated with sleep loss tend to be overlooked and assumed to be associated with the limited intellectual abilities associated with Down syndrome.

Because children with Down syndrome have an increased risk of sleep abnormalities, in 2001 the American Academy of Pediatrics recommend that primary care physicians question parents about possible sleep disorders when the children are five years old and older. However, no recommendations are made for specific testing. This study recommends objective testing by polysomnography, or sleep study, in all children with Down syndrome.

Cincinnati Children's is a 475-bed institution devoted to bringing the world the joy of healthier kids. Cincinnati Children's is dedicated to transforming the way health care is delivered by providing care that is timely, efficient, effective, family-centered, equitable and safe. It ranks third nationally among all pediatric centers in research grants from the National Institutes of Health. The Cincinnati Children's vision is to be the leader in improving child health.

Contact Information

Amy Caruso, 513-636-5637, amy.caruso@cchmc.org

http://www.cincinnatichildrens.org/about/news/release/2006/5-down-syndrome-sleep-apnea.htm

Sleep Apnea Tests Advised for Down's Children

By Judith Groch, MedPage Today Senior Writer
Published: April 18, 2006
Reviewed by Robert Jasmer, MD; Assistant Professor of Medicine, University of California, San Francisco .

CINCINNATI, April 18 - Because of high rates of obstructive sleep apnea in young children with Down's syndrome, researchers here have recommend baseline testing between ages three and four.

Overnight polysomnograms performed on 56 children, ages 3.5 to four, found that 57% of the children had abnormal results and evidence of obstructive sleep apnea syndrome, according to a study in the April issue of the *Archives of Otolaryngology-Head and Neck Surgery*.

When the researchers included an elevated arousal index, which is associated with increased difficulty breathing, the abnormal percentage rose to 80%, said Sally Shott, M.D., of the University of Cincinnati here, and colleagues.

Because of a lack of expertise in evaluating sleep disturbances, the parents are often oblivious to the problem. Sixty-nine percent of parents who filled out a questionnaire about their child's sleep patterns reported no problems, whereas 54% of the children had abnormal polysomnograms, Dr. Shott said. Parents and children came from a tertiary-care pediatric referral center.

The polysomnograms were classified as abnormal if the obstructive apnea index was greater than 1, if the carbon dioxide level was greater than 45 mm Hg for more than two-thirds of the study or greater than 50 mm Hg for more than 10% of the study. Also included was unexpected hypoxemia (oxygen saturation less than 92% during sleep or repeated intermittent desaturations less than 90%), the researchers said.

For purpose of analysis, the results were categorized in three groups, the researchers said. Group 1 (n=21) consisted of abnormal results because of an elevated obstructive sleep apnea index. These children also had hypercarbia, hypoxemia, or any combination, with or without hypoventilation and an elevated arousal index, according to the researchers.

In this category, they said, hypercarbia and hypoxemia, in addition to an abnormal obstructive apnea index, led to a statistically high obstructive apnea index compared with the index for children who did not have these add-on's (17.15, ±4.63 vs. 2.9±1.86, respectively; *P*=.02).

In group 2 (n=11), results were reported as abnormal because of hypoventilation with hypercarbia and/or hypoxemia, with or without an elevated arousal index. The apnea obstructive index was in the normal range. However, results from other studies show an increased risk of hypertension and abnormal cardiac rates as well as sleep fragmentation with prolonged hypercarbia, the researchers commented.

The third group (n= 24) included children with normal polysomnograms, but further inspection found that 13 of these children had an arousal index greater than 10 (mean index 15.6).

Commenting on the significance of the arousal response, Dr. Shott said that ordinarily an arousal is a protective reflex that helps curtail the upper airway obstruction and reestablish a patent airway.

However, there is concernthat an excessive number of arousals may lead to fragmented sleep and sleep deprivation. The increased arousal rate in Down's children may affect daytime function, ability to learn, and resultant behavior, often misattributed to a child's limited intellectual abilities, she said.

The parental questionnaire cast doubt on the parents' ability to assess their child's sleep problems. In general, these parents underestimate the severity of their child's sleep disturbances, Dr. Shott said. Thirty-five parents completed a questionnaire at the study's outset asking whether their child snored, stopped breathing while sleeping, and if there were snorts and gasps for air during sleep.

Overall, 11 (31%) parents reported that their child had sleep problems, but these parents were correct about a sleep abnormality in only four cases. The other seven children, believed by parents to have abnormalities, had normal polysomnograms. Of the 24 parents who reported no sleep problems, 13 children (54%) had abnormal tests, the researchers reported.

In a further analysis, for children in Groups 1 and 2 with major sleep disorders, 13 parents (77%) said their child had no sleep problems, and in group 3, in which the children were normal, seven (39%) said their child had sleep problems.

> "Our results point to the need for objective testing for obstructive sleep disorders in children as young as three or four years," Dr. Shott said. Because there is a high incidence of sleep disorders in Down's syndrome children, "baseline studies, using full overnight polysomnograms, are recommended even if parents report no sleep problems in their child," she said.
> **http://www.medpagetoday.com/PrimaryCare/SleepDisorders/3111**

Obstructive sleep apnea : Should all children with Down syndrome be tested?

Abstract

A study was done to determine the incidence of obstructive sleep apnea syndrome in children aged 2 to 4 years with Down syndrome and to determine parents' ability to predict sleep abnormalities in this patient population. Sixty-five children participating in a 5-year longitudinal study in which the otolaryngologic problems seen in Down syndrome were evaluated. Of those studied fifty-six completed overnight polysomnography (PSG) between 4 and 63 months of age (mean age, 42 months). Overnight PSG was performed. Parents also completed a questionnaire regarding their impressions of their child's sleep patterns before PSG. The PSGs revealed that 57% of the children had abnormal results and evidence of obstructive sleep apnea syndrome. If we also include an elevated arousal index, 80% of the PSGs had abnormal results. Sixty-nine percent of parents reported no sleep problems in their children, but in this group, 54% of PSGs had abnormal results. Of the parents who reported sleep problems in their children, only 36% had abnormal sleep study results. Conclusion: Because of the high incidence of obstructive sleep apnea syndrome in young children with Down syndrome, and the poor correlation between parental impressions of sleep problems and PSG results, baseline PSG is recommended in all children with Down syndrome at age 3 to 4 years.

Author(s)

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http://www.riverbendds.org/index.htm?page=sleepab.html

American Academy of Family Physicians has noted concerns regarding sleep apnea in children:

According to the American Academy of Family Physicians, one to twelve percent of typical children are affected by obstructive sleep apnea while experts studying sleep apnea in children with Down syndrome report that as many as 57% to 80% of the young children with Down syndrome studied experienced sleep apnea. Sleep apnea in children can have serious consequences. The American Academy of Family Physicians has advised, “Sleep-disordered breathing in children is a timely public health concern…” On their website, they further noted, “Consequences of untreated obstructive sleep apnea include failure to thrive, enuresis, attention-deficit disorder, behavior problems, poor academic performance, and cardiopulmonary disease.” Parents and physicians of children with Down syndrome need to address this serious health concern.

. http://www.aafp.org/afp/2004/0301/p1147.html

ndss Vision & Down Syndrome

Down Syndrome and the Eye

Trisomy 21 has effects on the developing eye, which could impact the proper development of vision. Eye disease is reported in over half of patients with Down Syndrome, from less severe problems such as tear duct abnormalities to vision threatening diagnoses, such as early age cataracts. Particular attention should be given to vision in people with Down syndrome. ----NDSS interviews Danielle Ledoux, MD Assistant in Ophthalmology at Children's Hospital, Boston and Instructor in Ophthalmology at Harvard Medical School.

What is different about the eyes in Down Syndrome?
Dr. Ledoux: As any family member of a person with Down syndrome knows, there are characteristic features about the eyes. This includes upward slanting of the eyelids, prominent folds of skin between the eye and the nose, and small white spots present on the iris (the colored part of the eye) called Brushfield's spots. These spots are harmless, and can be seen in people without Down syndrome as well.

Do most children with Down Syndrome need glasses?
Dr. Ledoux: Refractive error (the need for glasses) is much more common in children with Down syndrome than in the general population. This refractive error can be hyperopia (farsightedness), astigmatism, or myopia (nearsightedness). Another problem is weak accommodation (difficulty changing the focusing power of the eye from distance to near). We can test this easily in the office, and if detected, we will prescribe glasses that have bifocals. Some of my patients have difficulty adjusting to glasses, but once they get accustomed to having the glasses on their face, their vision is significantly better and often their eye alignment improves as well.

What are common, but less serious, eye abnormalities affecting Down Syndrome patients?
Dr. Ledoux: In addition to the need for eyeglasses, many children with Down syndrome have tear duct abnormalities. Family members will notice this as frequent discharge and tearing from the eyes, worsened by colds. We generally recommend firm massage over the space between the eye and the nose (tear sac region) 2-3 times a day to attempt to open the tear duct. If this continues beyond a year of age, the tear ducts may need to be opened by a surgical procedure. Strabismus (eye misalignment) is also more common. Family members may notice that the eyes do not line up well with each other, but often the strabismus can be subtle, even to the pediatrician. The folds of skin I mentioned between the eyes and the nose can also cover up the underlying strabismus, or make the eyes appear as if they are crossing even if they are not. It is important to diagnose strabismus as a child, as crossed eyes can result in amblyopia (loss of vision also known as lazy eye) and loss of stereopsis (the use of the two eyes together, or depth perception).

How can the strabismus be treated?
Dr. Ledoux: Sometimes, simply glasses alone are enough to straighten eyes with strabismus. If glasses are needed, we always start there. If the eyes continue to have strabismus despite the correct pair of eyeglasses, then we recommend strabismus surgery (eye muscle surgery). This is a one to two hour procedure, which can often be done as an outpatient unless there are other reasons the person would need to be admitted, such as a serious heart condition. Unfortunately, our patients with Down syndrome are more likely to require more than one surgery to align their eyes as they don't always respond as predictably to strabismus surgery as the general population with strabismus would.

What are the more severe eye problems that might develop?
Dr. Ledoux: My greatest concern is congenital cataracts (lack of clearness to the lens of the eye). If visually significant cataracts are present early in a child's eye, then a clear image is not delivered to the brain and therefore the brain can never "learn" to see. This is a severe form of amblyopia known as deprivational amblyopia. While we can take our time removing a cataract in an adult patient, significant cataracts present

very early in a child's life that is not removed can result in lifelong poor vision. In that situation, even if the cataract is removed when the child is older, the vision never improves significantly. This is what makes early detection of cataracts in infants and children so important. A child with Down syndrome will be evaluated by the pediatrician at birth, and referred to an ophthalmologist if something abnormal is detected. There is also a unique form of cataract in Down syndrome patients that we have found in our research. However, depending on how developmentally delayed the person is, they may not be able to communicate that they can't see. For this reason, I recommend any patient with Down syndrome, no matter what age, have a complete eye examination if they are starting to show reduced cognitive function, or changes in their normal activities.

Are there other eye conditions in Down Syndrome that can cause loss of eyesight?
Dr. Ledoux: I mentioned amblyopia (commonly called "lazy eye" which is decreased vision) which can be caused by multiple different eye problems such as strabismus, severe ptosis (eyelid droop), cataracts, or even uncorrected refractive error, especially if one eye needs a much stronger eyeglass prescription than the other. Ptosis is usually easier to appreciate but strabismus and significant refractive error can be very difficult for the pediatrician to diagnose. There are other more rare problems which can occur with the optic nerve or retina of the eye which can sometimes cause vision loss and unfortunately are generally not treatable. Nystagmus (a rhythmic shaking of the eyes) can also occur.

What is the recommended eye care for children with Down Syndrome?
Dr. Ledoux: The American Academy of Pediatrics (AAP) and the United States Down Syndrome Medical Interest Group (DSMIG) recommend evaluation of the red reflex of the eyes at birth to look for cataracts, as well as to assess the eyes for strabismus or nystagmus. The red reflex is essentially the "red eye" seen in photography, which is the normal reflex of the retina when struck by direct light. If the eyes don't look normal, then the infant will be referred to a pediatric ophthalmologist – a physician who has completed specialty training in medical and surgical management of the child's eye. We, along with the AAP and the DSMIG, recommend a child with Down syndrome has their first eye exam by an ophthalmologist experienced in patients with special disabilities (for example, a pediatric ophthalmologist) by six months of age. After that, children with Down syndrome, even if they are without symptoms, should see an ophthalmologist every one to two years. If any eye problems are detected, they will be followed more frequently.

What sort of symptoms might we see if a child has an eye problem?
Dr. Ledoux: Unfortunately, children with Down Syndrome often do not complain about their eye problems, either because they don't notice the problem or because they can't communicate the problem well enough. Signs to look for include squinting or closing one eye shut, an unusual head tilt, crossing or wandering of one or both eyes, or light sensitivity. In some severe cases, the sign of vision problems may be a regression in overall function or loss of developmental milestones. Ptosis will be seen as a lid droop, and a blocked tear duct will result in daily tearing and discharge.

Any thoughts for parents of a child with Down Syndrome who are concerned about the eye or vision?

Dr. Ledoux: Getting regular eye exams is very important in children with Down Syndrome because eye disorders are so common and are difficult for the pediatrician to diagnose. Because the examination can be difficult for both the child and the doctor, it is best to have the examination done by an ophthalmologist skilled in dealing with children with developmental delays. Don't be surprised to find out your child needs glasses; if needed, the glasses will help the vision, and possibly the eye alignment, as well as to help in the development of normal vision pathways in the brain. This will help with your child's learning and functioning. Our research is looking at just how common eye problems are in Down syndrome, as well as the development of cataracts in these patients.

Dr. Danielle Ledoux can be reached at Danielle.ledoux@childrens.harvard.edu , or through the Children's Hospital, Boston website at www.childrenshospital.org/eyes.

ndss Vision & Down Syndrome

Vision and Down Syndrome Resource List

Organizations and Websites

American Speech-Language Hearing Association
10801 Rockville Pike
Rockville, MD 20852
Telephone: (301) 897.5700 or (800) 638-8255
Website: http://www.asha.org

Blind Children's Center
4120 Marathon Street
Los Angeles, CA 90029-0159
Telephone: (323) 664-2153 or (800) 222-3566
E-mail: info@blindchildrenscenter.org Website: http://www.blindchildrenscenter.org

National Association for Parents of the Visually Impaired, Inc.
P.O. Box 317
Watertown, MA 02472-0317
Telephone: (617) 972-7441 or (800) 562-6265
E-mail: napvi@perkins.org Website: http://www.napvi.org

National Institute on Deafness and Other Communication Disorders Clearinghouse
Telephone: (800) 241-1044 or (800) 241-1055
Website: http://www.nidcd.nih.gov

Books on Vision and Hearing

Holbrook, M.C. (Ed.). (1996). Children with Visual Impairments: A Parents' Guide. Bethesda, MD: Woodbine House. http://www.woodbinehouse.com

Wellness and Healthy Living Resource List

Organizations and Websites

Academy of Dentistry for Persons with Disabilities
Special Care Dentistry
401 North Michigan Avenue, Suite 2200
Chicago, Il 60611
Telephone: (312) 527-6764
Fax: (312) 673-6663
E-mail: SCDA@SCDonline.org Website: http://www.scdonline.org

American Speech-Language Hearing Association
10801 Rockville Pike
Rockville, MD 20852
Telephone: (301) 897.5700 or (800) 638-8255
Website: http://www.asha.org

LaLeche League International
1400 North Meacham Road
Schaumberg, IL 60173
Telephone: (800) LALECHE or (847) 519-7730
Website: http://www.lalecheleague.org

National Center on Physical Activity and Disability
1640 W. Roosevelt Road
Chicago, IL 60608-6904
Telephone: (800) 900-8086
E-mail: ncpad@uic.edu Website: http://www.ncpad.org

Books and DVDs

Durand, V. M.. Sleep Better: A Guide to Improving Sleep for Children with Special Needs, Baltimore, MD; Brookes Publishing (1998). http://www.brookespublishing.com

Guthrie Medlen, J.E. The Down Syndrome Nutrition Handbook. Lake Oswego, OR
Phronesis Publishing (2006). http://www.downsyndromenutrition.com/phronesis/

McGuire,D,& Chicoine, B. Mental Wellness in Adults with Down Syndrome. Bethesda, MD: Woodbine House (2005). http://www.woodbinehouse.com.

Sanderson, Sheri L. Incredible Edible Gluten-Free Food for Kids. Bethesda, MD: Woodbine House Publishing (2002). http://www.woodbinehouse.com

Schermerhorn, Will (Producer). Kids with Down Syndrome: Staying Healthy and Making Friends. Bethesda, MD: Woodbine House (2005). http://www.woodbinehouse.com

Feeding Resource List

Organizations and Websites

LaLeche League International
1400 North Meacham Road
Schaumberg, IL 60173
Telephone: (800) LALECHE or (847) 519-7730
Website: http://www.lalecheleague.org

New Visions
1124 Roberts Mountain Road
Faber, VA 22938
Telephone: (800) 606-7112
E-mail: mealtime@new-vis.com
Website: http://www.new-vis.com

Books and Articles

Fiocca, Sharon. Oral Motor & Feeding Skills. (1999; Updated 2007) Reprinted at http://www.riverbendds.org/fiocca.html

Lowman, D. K, Murphy, S.M. The Educator's Guide to Feeding Children with Disabilities. Baltimore, MD: Paul H. Brookes Publishing Co (1998). http://www.brookespublishing.com/

Sleep Issues Resource List

Organizations and Websites

American Sleep Apnea Association.
1424 K Street NW, Suite 302
Washington, DC 20005
Telephone: (202) 293-3650
Fax: (202) 293-3656
Email: asaa@sleepapnea.org Website: http://www.sleepapnea.org/

American Sleep Disorders Association
1610 14th Street NW
Rochester MN 55901
Telephone: 507-287-6006
Email: asda@millcomm.com

Center for Research in Sleep Disorders
1275 East Kemper Rd.
Cincinnati OH 45246
Telephone: 513-671-3101
Email: ggaz@tristatesleep.com

National Sleep Foundation
1522 K Street, NW, Suite 500
Washington, DC 20005
Telephone: (202) 347-3471
Fax: (202) 347-3472
Website: http://www.sleepfoundation.org/site/c.huIXKjM0IxF/b.2417141/k.C60C/Welcome.htm

Books and Articles

Durand, V. M.. Sleep Better: A Guide to Improving Sleep for Children with Special Needs, Baltimore, MD; Brookes Publishing (1998). www.brookespublishing.com

Leshin, Len. “Obstructive Sleep Apnea and Down Sydrome”. Down Syndrome: Health Issues. http://www.ds-health.com/apnea.htm

“Obstructive Sleep Apnea in Children with Down Syndrome”. Capitol Sleep Medicine Newsletter. September 2007. Volume 2 Number 9. www.capitolsleepmedicine.com