Keeping Children and Adolescents with Down Syndrome Healthy:
All the Medical Updates that Parents Need to Know

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Disclosures

• My wife and I have no financial relationships with a commercial entity producing health-care related products and/or services.

• Volunteer in a non-paid capacity to National Down Syndrome Congress Massachusetts Down Syndrome Congress Band of Angels Foundation

• I have a sister with Down syndrome.
Goals

- How do you ensure that your son or daughter is getting the best medical care as it relates to Down syndrome?

- What are some common medical conditions that occur in people with Down syndrome during childhood and adolescence?

- What healthcare resources are available to you, as parents?
My child has some difficult to manage behaviors!
Down Syndrome Behavior Checklist

1. Exclude medical conditions.
   - Hearing difficulties
   - Vision difficulties
   - Thyroid problems
   - Celiac disease
   - Constipation
   - Obstructive sleep apnea

2. Maximize expressive language skills.

3. Think about behavior conditions.
Hearing Difficulties

FACTS: 75% of children with DS have hearing loss

DOCTOR: audiologist, otolaryngologist (ORL, ENT)

TESTS: audiology: birth, 6mo, 12mo, then yearly

1. **Conductive hearing loss**: consider ear wax removal, tympanostomy tubes for fluid build-up, preferential school seating

2. **Sensorineural hearing loss**: consider hearing aids, FM amplification system, preferential school seating

3. **Mixed hearing loss**: both of the above
Vision Difficulties

FACTS: ~60% of children with DS have eye conditions
DOCTOR: ophthalmologist
TESTS: birth, <1yo, annually 1-5yo; every two yrs 5-13yo; every three yrs 13-21yo

1. Myopia, hyperopia, astigmatism: eyeglasses
2. Nystagmus: working closely with eye doctor
3. Strabismus: patching or putting dilating drops in good eye to strengthen the weaker one; surgical correction
4. Cataracts: surgical correction
5. Lacrimal duct obstruction: warm compresses; surgery
Thyroid Problems

FACTS: ~15% of children with DS have thyroid problems
DOCTOR: endocrinologist
TESTS: birth, 6mo, 12mo, then annually

Symptoms: tired, sluggish, constipation, unusually cold, hyperactive, sweating, restless, diarrhea, behavior problems
Thyroid Problems

FACTS: ~15% of children with DS have thyroid problems
DOCTOR: endocrinologist
TESTS: birth, 6mo, 12mo, then annually

1. Hypothyroidism: levothyroxine
2. Hyperthyroidism: treat with medicines
3. Compensated hypothyroidism (hyperthyrotropinemia): needs to be followed more closely; consider treating with levothyroxine if persistently elevated
Clinical Report—Health Supervision for Children With Down Syndrome

Marilyn J. Bull, MD, and the COMMITTEE ON GENETICS
1. Go to NDSS’s web page: www.ndss.org
2. Click on “healthcare” on the left.
3. Then click on “health care guidelines.”
Down Syndrome: Healthcare Updates for the Primary Care Pediatrician

Course Description

Acknowledgement: This course is supported by educational grants from Celera and the Appieora Charitable Foundation.

Down syndrome is the most common chromosomal condition in humans, but do you know the latest evidence-based medicine on how to best manage these patients? Much has changed since medical school! Many healthcare guidelines and emerging literature are changing the way that individuals with Down syndrome are diagnosed, followed, and managed. Astute physicians can now detect associated conditions before they become symptomatic in an effort to maximize health care development. The goal of this course is to review the latest advances in healthcare management of children with Down syndrome so that primary care pediatricians and other interested health care professionals can provide exceptional care.

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Most Recent Update: 5/5/2010
Termination Date: 5/5/2013

NUMBER OF CREDIT HOURS: The Harvard Medical School designates this educational activity for a maximum of 1 AMA PRA Category 1 credit(s)**. Physicians should only claim credit commensurate with the extent of their participation in the activity.
Celiac Disease

FACTS: ~5% of children with DS have Celiac disease

DOCTOR: gastroenterologist (GI)

TESTS: beginning at 1 yo if symptomatic

Symptoms: diarrhea, constipation, bloating, behavioral problems, vomiting, abdominal pain

Diagnosis: start blood screen (IgA & TTG); intestinal biopsy

Treatment: 100% gluten-free diet
The Allen C. Crocker Speaker Series

The Allen C. Crocker Lecture Series is designed for parents and families members seeking quality information from experts on a range of topics related to Down syndrome. These lectures, honoring the legacy of Dr. Crocker, can be viewed, for free, from the links in the table below.

Information on Attending the Speaker Series:

* The Series will take place on the fourth Monday, unless otherwise noted.
* Families will arrive at 6:30pm
* Speaker will present from 7-8pm
* Question and Answer Session from 8-8:30pm
* All talks will be held in the Enders Auditorium at Children’s Hospital in Boston.
* Childcare is available as needed and will require RSVP’s in order to set this up!
* Validated Parking Fee is $9.
* Refreshments will be served!
Celiac Disease: Pathogenesis

Interaction of Gluten with

Environmental Factors

Immune Factors

Genetic Factors

Note 4
Our Talk on Celiac Disease will start at 7:00pm. Please email Angela, any questions you may have since we are not going to be accepting questions online! Thanks! :)

The NEW ENGLAND JOURNAL OF MEDICINE
Constipation
Constipation

Bulk-producing agents: dietary fiber; benefiber supplements

Softens stool: colace (docusate)

Lubricant: mineral oil

Motility/stimulant agent: senna, dulcolax, ex-lax

Waters-down stool: Miralax (polyethylene glycol), milk of magnesia, lactulose
Obstructive Sleep Apnea (OSA)

FACTS: up to 75% of children with DS have OSA
DOCTOR: otolaryngologist; sleep medicine doctor
TESTS: every child by the age of 4

Does your child snore at night?
Does your child gasp, choke, snort during sleep?
Does your child fall asleep on short drives? At school?
Does your child need to nap in an age-inappropriate way?
Does your child not seem refreshed during the day?

Sleep study (polysomnogram): only definitive diagnosis

Treatment: medicine; tonsil/adenoid surgery; surgery
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1. Exclude medical conditions.
   • Hearing difficulties
   • Vision difficulties
   • Thyroid problems
   • Celiac disease
   • Constipation
   • Obstructive sleep apnea

2. Maximize expressive language skills.

3. Think about behavior conditions.
Augmentative and Alternative Communication

Proloquo2Go: AAC in Your Pocket

Communication on the Go for iPhone, iPod touch and iPad

www.proloquo2go.com
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Specific Behavioral Conditions

**ADHD (Attention Deficit-Hyperactivity Disorder):** difficulty concentrating, paying attention, staying still in $\geq 2$ settings

**OCD (Obsessive Compulsive Disorder):** perseverations, repeated activities to the point of interference with daily activities

**Depression:** loss of interest, sleep changes, appetite changes, concentration problems, withdrawn

**Anxiety:** excessive nervousness, fearfulness to the point of interference with daily activities
Mental Wellness in Adults with Down Syndrome

A Guide to Emotional and Behavioral Strengths and Challenges

Dennis McGuire, Ph.D. & Brian Chicoine, M.D.

www.woodbinehouse.com
Autism Spectrum Disorder (ASD)

Pervasive Developmental Disability (PDD)

FACTS: up to 18% with ASD; about 6% with autism

DOCTOR: pediatric psychologist, pediatric psychiatrist

TESTS: neuropsychological evaluation; behavioral evaluation

1. Significant communication difficulties
2. Significant difficulties with social skills
3. Repetitive behaviors

TREATMENT: no less than 25 hours of ABA per week
I’m concerned about my child’s weight!
“The previously used Down syndrome-specific growth charts no longer reflect the current population styles and body proportion.” AAP, 2011
Overweight and Obesity

OVERWEIGHT: >85% on BMI or weight/height

OBESE: >95% on BMI or weight/height

TESTS: HgA1c for diabetes
        total cholesterol
        Liver function tests

NUTRITIONIST: once a month
The Down Syndrome Nutrition Handbook

A Guide to Promoting Healthy Lifestyles

Jeanne Babie-Edell, M.D., M.P.H.

www.downsyndromenutrition.com
My son’s/daughter’s skin is dry or has rashes!
Common Skin Findings

Xerosis
Eczema
Cheilitis
Onychomycosis
Tinea pedis
Alopecia areata
Vitiligo
Seborrheic dermatitis
Cutis marmorata
My child wants to participate in contact sports. Okay?
Atlantoaxial Instability (AAI) & Occipitoaxial instability (OAI)

FACTS: ~15%, consequences rare but dangerous
DOCTOR: neurosurgeon, orthopedic surgeon
TESTS: C-spine (3 views: lateral, flexion, extension)

- C-spines do not predict well which children are at risk
- Normal C-spines do not provide reassurance that a child will not develop spine problems later
- Routine C-spines not recommended in asymptomatic children
- However, Special Olympics might require them for participation
Atlantoaxial Instability (AAI) & Occipitoaxial instability (OAI)

SYMPTOMS

Change in gait
Change in use of arms or hands
Change in bowel or bladder function
    Neck pain
    Stiff neck
    Head tilt
    Torticollis
    New-onset weakness
    Hyperreflexia
Atlantoaxial Instability (AAI) & Occipitoaxial instability (OAI)

≥ 4.5mm
Atlantoaxial Instability (AAI) & Occipitoaxial instability (OAI)

ACTIVITY RESTRICTIONS

Contact sports (e.g., football, soccer, wrestling)
Gymnastics
Horse-back riding
Diving
Trampoline usage
Careful precautions during anesthesia
My child is limping and complaining of leg pain!
Hip Dysplasia/Dislocation

FACTS: ~1-4%: ages 2-10 years
DOCTOR: orthopedist
TESTS: Hip and knee X-rays, as warranted

Whenever a joint seems to hurt, always consider getting additional X-rays of the joints above and below.

TREATMENT: surgery, immobilization
Leukemia

FACTS: <1 % of children
DOCTOR: hematologist; oncologist
TESTS: CBC with differential; bone marrow biopsy

Acute lymphocytic leukemia (ALL):
• Between 3-6 years
• Less likely to have large spleen, chest mass, and central nervous system like typical population
• TREATMENT: Chemotherapy

Acute myelogenous leukemia (AML):
• Between 1-5 years
• History of TMD
• TREATMENT: Chemotherapy
My child is has some abnormalities in their blood cell types.
RBC
↑ polycythemia
↑ macrocytosis
↓ anemia

platelet
↑ thrombocytosis
↓ thrombocytopenia

WBC
↑ leukemia
↑ leukemoid reaction
↓ leukopenia
Transient Myeloproliferative Disorder (TMD)

- occurs 10-20% of babies with Down syndrome
- present at birth or within first week of life
- typically resolves in the first 3 months of life
- 20-30% will progress onto leukemia by age 4

If in this category: CBC ~3 months until age 4-6
Iron-deficiency anemia

FACTS: many with DS have low dietary intake of iron
DOCTOR: pediatrician
TESTS: Hemoglobin, annually at 1 yo

If hemoglobin < 11g, order (a) CRP and ferritin or (b) reticulocyte hemoglobin (CHr)

TREATMENT:
• improved dietary intake
• iron supplementation
Should my child be taking supplements?
• Has a **double-blind placebo-controlled** research been done?
• What are the potential **side effects**?
• What are the potential **long-term consequences**?
• What is the expense in **time and money**?
• Are there any **position statements** by physicians or national Down syndrome organizations?

1. Go to NDSS’ s web page:  [www.ndss.org](http://www.ndss.org)
2. Click on “**healthcare**” on the left.
3. Then click on “**alternative therapies**.”
Contact Information

Web page: www.brianskotko.com

Facebook: facebook.com/brian.skotko

Twitter: twitter.com/brianskotko

YouTube: http://www.youtube.com/brianskotko

Hospital: www.massgeneral.org/downsyndrome