Band of Angels Foundation

Keeping Children and Adolescents with Down Syndrome Healthy

All the Medical Updates that Parents Need to Know

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Down Syndrome Program
Massachusetts General Hospital

Brian's Disclosures

- I receive research support for clinical drug trials from Hoffmann-La Roche, Inc.
- Volunteer in a non-paid capacity to
  - Massachusetts Down Syndrome Congress
  - Band of Angels Foundation
  - National Center for Prenatal and Postnatal Down Syndrome Diagnoses Resources
- I have a sister with Down syndrome.

"Mock My Pants, Not My Sister"

"Boston suffers from a kind of Style Down Syndrome, where a little extra ends up ruining everything."
—John Thompson, *GQ* Magazine, July 15, 2011

http://childrenshospitalblog.org/mock-my-pants-not-my-sister/

Goals

- How do can you ensure that your son or daughter is getting the best **medical care** as it relates to Down syndrome?
- What is the medical work-up for **behavioral problems**?
- What are some other **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 1-5 yo; every two years 5-13yo; every three years 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:

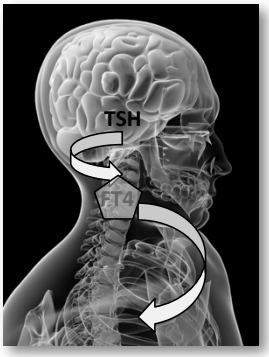
tired	hyperactive
sluggish	restless
constipation	diarrhea
unusually cold	sweating
	behavior problems

Thyroid Problems

Hypothyroidism
↓FT4, ↑TSH

Hyperthyroidism
↑FT4 ,↓TSH

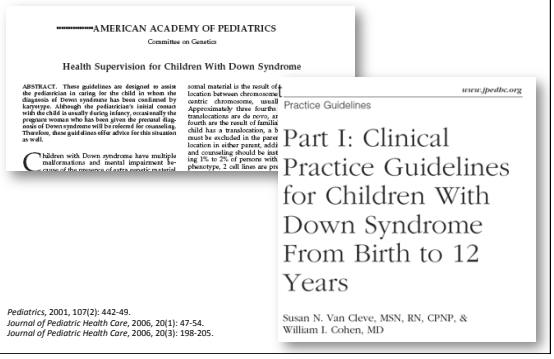
Compensated Hypothyroidism
Normal FT4, ↑TSH



Thyroid Problems

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

Old Healthcare Guidelines for Physicians


AMERICAN ACADEMY OF PEDIATRICS
Committee on Genetics

Health Supervision for Children With Down Syndrome

ABSTRACT. These guidelines are designed to assist the pediatrician in caring for the child in whom the diagnosis of Down syndrome has been confirmed by karyotype. Although the pediatrician's initial contact with the child is usually during infancy, occasionally the pregnant woman who has been given the prenatal diagnosis of Down syndrome will be referred for counseling. Therefore, these guidelines offer advice for this situation as well.

Children with Down syndrome have multiple malformations and mental impairment because of the presence of extra genetic material

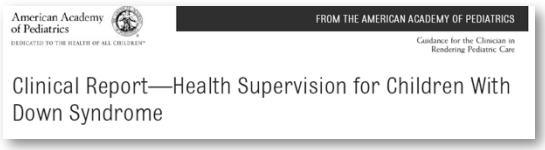
www.jpedhc.org
Practice Guidelines

Part I: Clinical Practice Guidelines for Children With Down Syndrome From Birth to 12 Years

Susan N. Van Cleve, MSN, RN, CPNP, & William I. Cohen, MD

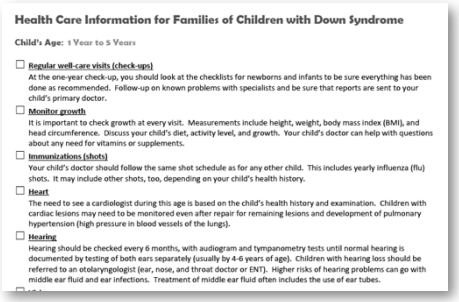
Pediatrics, 2001, 107(2): 442-49.
Journal of Pediatric Health Care, 2006, 20(1): 47-54.
Journal of Pediatric Health Care, 2006, 20(3): 198-205.

2011 Healthcare Guidelines for Physicians



Pediatrics Volume 128, Number 2, August 2011

Healthcare Guidelines (Parent-Friendly Version)



Health Care Information for Families of Children with Down Syndrome

Child's Age: 1 Year to 5 Years

- ☐ **Regular well-care visits (check-ups)**
  At the one-year check-up, you should look at the checklists for newborns and infants to be sure everything has been done as recommended. Follow-up on known problems with specialists and be sure that reports are sent to your child's primary doctor.
- ☐ **Monitor growth**
  It is important to check growth at every visit. Measurements include height, weight, body mass index (BMI), and head circumference. Discuss your child's diet, activity level, and growth. Your child's doctor can help with questions about any need for vitamins or supplements.
- ☐ **Immunizations (shots)**
  Your child's doctor should follow the same shot schedule as for any other child. This includes yearly influenza (flu) shots. It may include other shots, too, depending on your child's health history.
- ☐ **Heart**
  The need to see a cardiologist during this age is based on the child's health history and examination. Children with cardiac lesions may need to be monitored even after repair for remaining lesions and development of pulmonary hypertension (high pressure in blood vessels of the lungs).
- ☐ **Hearing**
  Hearing should be checked every 6 months, with audiogram and tympanometry tests until normal hearing is documented by testing of both ears separately (usually by 4-6 years of age). Children with hearing loss should be referred to an otolaryngologist (ear, nose, and throat doctor or ENT). Higher risks of hearing problems can go with middle ear fluid and ear infections. Treatment of middle ear fluid often includes the use of ear tubes.

http://www.healthychildren.org/

Online Course for Healthcare Professionals

cmeonline.med.harvard.edu

Celiac Disease

FACTS:	~5% of children with DS have Celiac disease
DOCTOR:	Gastroenterologist (GI)
TESTS:	Beginning at 1 yo if symptomatic

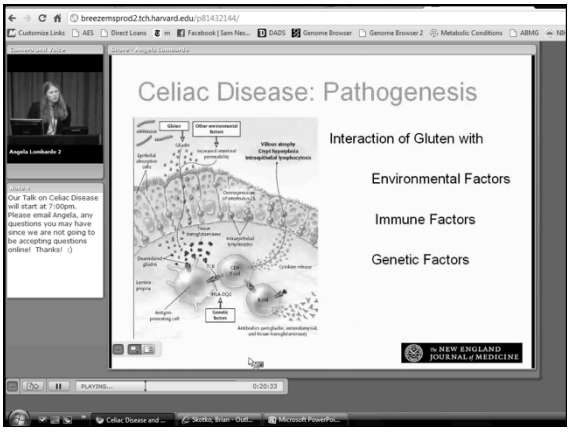
Symptoms: diarrhea, constipation, bloating, behavioral problems, vomiting, abdominal pain

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

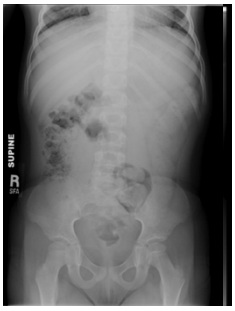
Treatment: 100% gluten-free diet

Healthcare Webinar Series

www.childrenshospital.org/downsyndrome



Constipation



Constipation

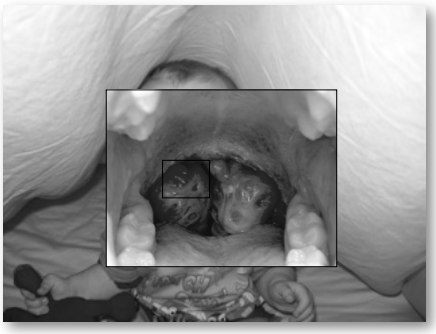
Bulk-producing agents: dietary fiber; benefiber supplements, metamucil

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 you 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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   - Vision difficulties
   - Thyroid problems
   - Celiac disease
   - Constipation
   - Obstructive sleep apnea
2. Maximize expressive language skills.
3. Think about behavior conditions.

www.signingtime.com

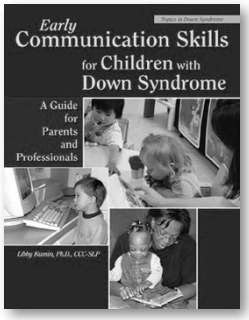
Augmentative and Alternative Communication

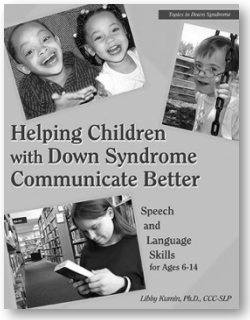
http://www.childrenshospital.org/dream/videos.html?bcpid=5558497001&bclid=5525144001&bctid=16391898001

iPads and iPhones

www.proloquo2go.com

www.woodbinehouse.com





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Down Syndrome Behavior Checklist

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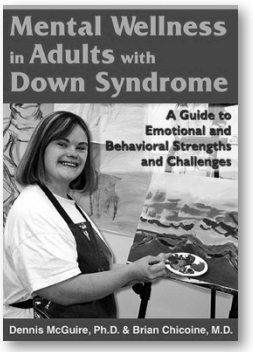
Specific Behavioral Conditions

ADHD (Attention Deficit-Hyperactivity Disorder): difficulty concentrating, paying attention, staying still in ≥ 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



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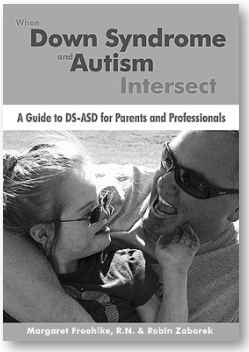
Autism Spectrum Disorder (ASD)

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



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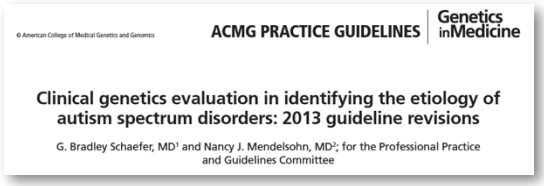


In about 30-40% of patients with autism spectrum disorders, a cause can now be identified.

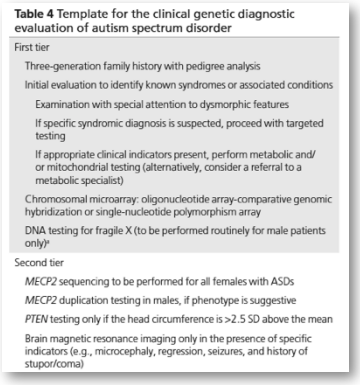


Table 4 Template for the clinical genetic diagnostic evaluation of autism spectrum disorder

First tier
Three-generation family history with pedigree analysis
Initial evaluation to identify known syndromes or associated conditions
Examination with special attention to dysmorphic features
If specific syndromic diagnosis is suspected, proceed with targeted testing
If appropriate clinical indicators present, perform metabolic and/or mitochondrial testing (alternatively, consider a referral to a metabolic specialist)
Chromosomal microarray: oligonucleotide array-comparative genomic hybridization or single-nucleotide polymorphism array
DNA testing for fragile X (to be performed routinely for male patients only)[a]
Second tier
MECP2 sequencing to be performed for all females with ASDs
MECP2 duplication testing in males, if phenotype is suggestive
PTEN testing only if the head circumference is >2.5 SD above the mean
Brain magnetic resonance imaging only in the presence of specific indicators (e.g., microcephaly, regression, seizures, and history of stupor/coma)

Disruptive Behavior Not Otherwise Specified

Functional Behavioral Assessment: http://cecp.air.org/fba/

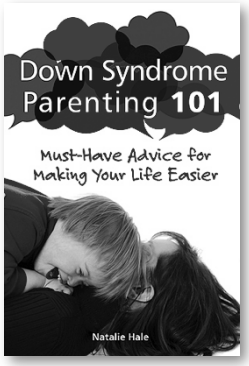
Pediatric psychologist, psychiatrist, Down syndrome specialist

TABLE VII. New Diagnoses for Patients Whose Parents Had an Initial Concern of Behavioral Issues (N = 56)*

Diagnosis	Number	%
Expressive language disorder	39	72.2
Disruptive behavior disorder NOS	37	66.1
Constipation	19	33.9
Hearing disorder	12	21.4
Ophthalmologic diagnosis	11	19.6
Obstructive sleep apnea	9	16.1
Thyroid disorder	5	8.9
Autism spectrum disorder	5	8.9
Celiac disease	4	7.1

*A patient could have more than one diagnosis; as such, percentages do not add up to 100%.

Skotko BG, Davidson EJ, Weintraub GS. 2012. Contributions of a specialty clinic for children and adolescents with Down syndrome. Am J Med Genet Part A 9999:1–8.



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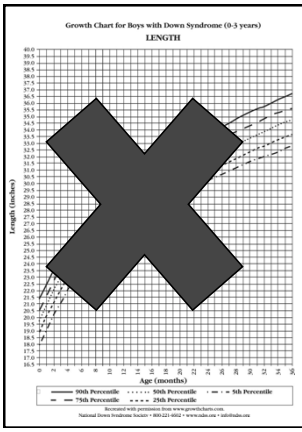
I'm concerned about my child's weight!



Down Syndrome Growth Curves

Weight
Height
Head circumference

"The previously used Down syndrome-specific growth charts no longer reflect the current population styles and body proportion." AAP, 2011

Weight/Height
BMI

Overweight and Obesity

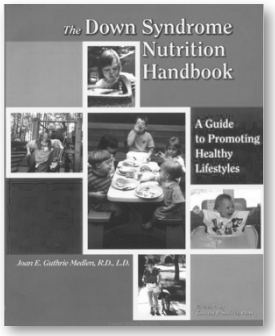
OVERWEIGHT: >85% on BMI or weight/height
OBESE: >95% on BMI or weight/height

TESTS: HgA1c for diabetes
total cholesterol
Liver function tests

NUTRITIONIST: once a month

Our Clinic: Majority Overweight or Obese

	Overweight	Obese	Total	Typical
Ages 2-19				
Female (N = 33)	36%	18%	54%	
Male (N = 40)	35%	30%	65%	
TOTAL (N = 73)	36%	25%	61%	32%
Ages 20-60				
Female (N = 88)	23%	58%	81%	
Male (N = 120)	38%	42%	80%	
TOTAL (N = 209)	32%	48%	80%	69%
All Ages 2-60				
Female (N = 121)	26%	47%	73%	
Male (N = 160)	38%	39%	77%	
TOTAL (N = 281)	33%	42%	75%	



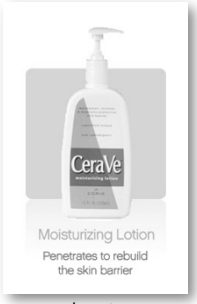
www.downsyndromenutrition.com

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

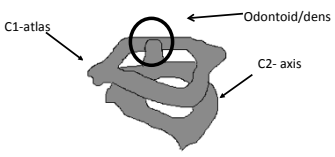


www.drugstore.com

My child wants to participate in contact sports. Okay?

Atlantoaxial Instability (AAI) & Occipitoaxial instability (OAI)

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



Atlantoaxial Instability (AAI) & Occipitoaxial instability (OAI)

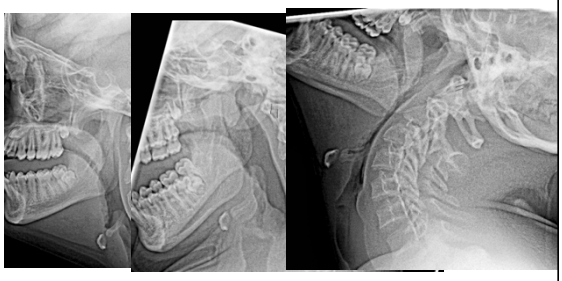
- C-spines do not predict well which children are at risk
- Normal C-spines do not provide reassurance that a child will not develop spine problems later
- Routine C-spines not recommended in asymptomatic children
- However, Special Olympics might require them for participation

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

I'm concerned that my child is having shaking movements!

Seizures

FACTS:	~8%: < 1 year old or > 30 years old
DOCTOR:	neurologist
TESTS:	EEG, as warranted

1. Infantile spasms
2. Partial seizures
3. Generalized tonic-clonic seizures

TREATMENT: same as general population

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.

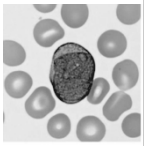
TREATMENT: surgery, immobilization

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 drinking a lot and going to bathroom excessively!

Diabetes

FACTS:	< 2% of children
DOCTOR:	endocrinologist
TESTS:	Glucose and HbA1c, as warranted

Type 1 diabetes:
- 75% of children with DS and diabetes
- TREATMENT: insulin injections

Type 2 diabetes:
- 25% of children with DS and diabetes
- TREATMENT: other medications

My child always has a runny nose and seems congested!

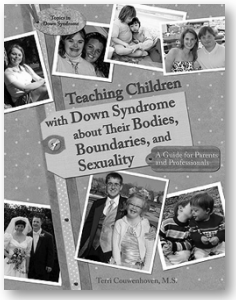
Seasonal Allergies

FACTS:	Many children with Down syndrome
DOCTOR:	allergist
TESTS:	RAST testing, as warranted

Systemic medication:
- Claritin
- Zyrtec

Local medication:
- Flonase
- Nasonex
- Saline spray drops

How should I prepare my child for puberty?



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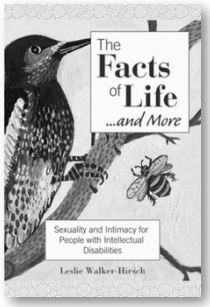
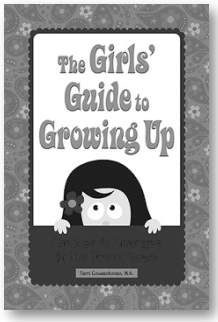
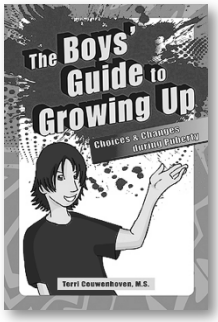


www.lesliewalker-hirsch.com





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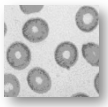
I can't get my child to go to the dentist.

Oral health conditions

FACTS:	Many children with Down syndrome
DOCTOR:	dentist
TESTS:	Dental exam every 6 months

Late eruption of teeth
Missing baby teeth
Cavities
Malocclusions
Periodontal disease

My child is has some abnormalities in their blood cell types.



RBC

↑ polycythemia

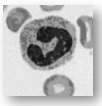
↑ macrocytosis

↓ anemia

platelet

↑ thrombocytosis

↓ thrombocytopenia

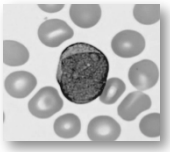


WBC

↑ leukemoid reaction

↓ leukopenia

Transient Myeloproliferative Disorder (TMD)

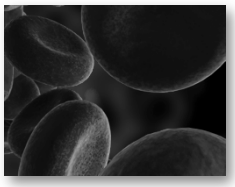


- occurs10-20% of babies with Down syndrome
- present at birth or within first week of life
- typically resolves in the first 3 months of life
- 20-30% will progress onto leukemia by age 4

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

Iron-deficiency anemia

FACTS:	Many children with low dietary intake of iron
DOCTOR:	pediatrician
TESTS:	Hemoglobin, annually beginning at 1 yo



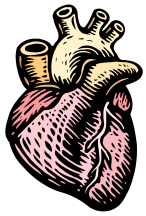
If hemoglobin < 11g, order
(a) CRP and ferritin or
(b) reticulocyte hemoglobin (CHr)

TREATMENT:
- improved dietary intake
- iron supplementation

What about my child's heart?

Cardiac concerns in childhood

FACTS:	Teens with DS can develop heart valve issues
DOCTOR:	cardiologist
TESTS:	Echocardiogram, as warranted



SYMPTOMS

Increasing fatigue
Shortness of breath
Exertional dyspnea
New murmur

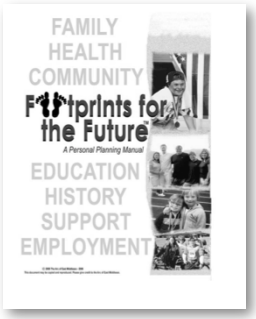
Should my child be taking supplements?

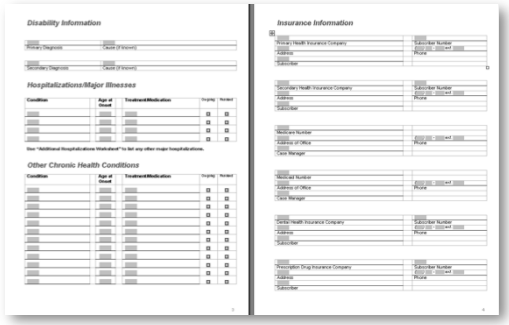
Questions to Ask About Supplements

- Has 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?

www.ndss.org/Resources/Therapies-Development/Alternative-Therapies/

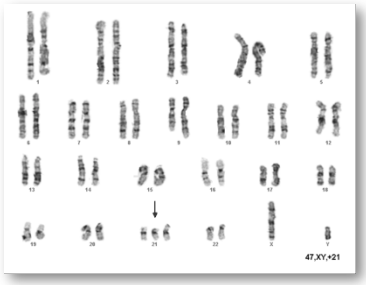
How do I keep my son's or daughter's health records organized?





www.theemarc.org

Keep a copy of the karyotype!



RESEARCH ARTICLE

AMERICAN JOURNAL OF medical genetics PART A

Contributions of a Specialty Clinic for Children and Adolescents with Down Syndrome

Brian G. Skotko,[1]* Emily Jean Davidson,[2] and Gil S. Weintraub[3]

Only 9.8% of patients up-to-date on Down syndrome healthcare guidelines prior to visiting our Down syndrome specialty clinic.

TABLE VI. New Diagnoses Made as a Result of Clinical Visit (N = 105)*

Diagnosis	Number	%
Xerosis	57	54.3
Expressive language disorder	56	53.3
Disruptive behavior NOS	39	37.1
Obesity (>95% BMI)	27	25.7
Overweight (85–95% BMI)	22	21.0
Constipation	20	19.0
Seasonal allergies	19	18.1
Obstructive sleep apnea	15	14.3
Thyroid conditions[a]	9	8.6
Celiac disease	6	5.7
Autism spectrum disorder	6	5.7
Sensorineural hearing loss	5	4.8
Hearing loss, unspecified	5	4.8
Atlantoaxial instability	5	4.8
Removed atlantoaxial instability diagnosis	5	4.8

*A patient could have more than one diagnosis; as such, percentages do not add up to 100%.
[a]Included one patient with hypothyroidism, four patients with Hashimoto's thyroiditis, and four patients with hyperthyrotropinemia (compensated hypothyroidism).

Where can I find a Down Syndrome Clinic?

www.ndss.org/Resources/Health-Care

MassGeneral Hospital Down Syndrome Program

Allie Schwartz, MD

Brian Skotko, MD, MPP

Jessica McCannon, MD

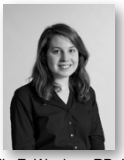
Jose Florez, MD, PhD

Tomi L. Toler, MS, CGC
Program Coordinator

Helen Weiner, MS, LICSW
Social Worker

Benjamin Majewski
Resource Specialist

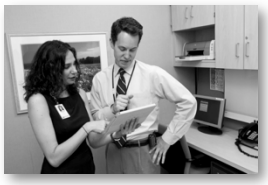
Sandy Massalski
Office Assistant



Caitlin E. Woglom, RD, LDN
Clinical Nutritionist

Prenatal Services

Dr. Allie Schwartz
Dr. Brian Skotko
Tomi Toler (genetic counselor)
Helen Weiner (social worker)
Ben Majewski (optional)



Infant & Toddler Clinic

Ages 0-5

Physicians
Nutritionist
Program coordinator
Social worker
Self-advocate resource specialist

Audiologist
Ophthalmologist
Physical therapist
Occupational therapist
Speech-language pathologist
Dentistry

Patricia Anne Chastain, PT, DPT, PCS

Sharon P. Serinsky, MS, OTR/L

Michele Psillos Donlon, MS, CCC-SLP

Child Clinic

Ages 5-13

Physicians
Nutritionist
Program coordinator
Social worker
Self-advocate resource specialist

Audiologist
Ophthalmologist
Dentistry

audiologists

Adolescent & Young Adult Clinic

Ages 13-21

Physicians
Nutritionist
Program coordinator
Social worker
Self-advocate resource specialist
Psychologist

Audiologist
Ophthalmologist

Julie Grieco, PhD.,
Kay Seligsohn, PhD &
Gretchen Timmel, MEd

Adult Clinic

Ages 21 and older

Physicians
Nutritionist
Program coordinator
Social worker
Self-advocate resource specialist

Audiologist
Ophthalmologist

Clinical Research Team

Mary Ellen McDonough, R.N.
Senior Clinical Research Coordinator

Christianne Sharr, B.A.
Research Assistant

Clinical and Research Web Pages

www.massgeneral.org/downsyndrome

Friend us!

www.facebook.com/MGHDownSyndrome

We tweet!

twitter.com/MGHDownSyndrome

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YouTube: www.youtube.com/brianskotko

Clinic: www.massgeneral.org/downsyndrome

Band of Angels Foundation