Down Syndrome

Overview of Health and Developmental Issues

Joseph D. Pinter, MD, MBA
Providence St Vincent Medical Center
Portland, Oregon

June 5, 2026

Providence

Disclosure Statement

- I am currently receiving no grant, commercial support or sponsorship.

Objectives

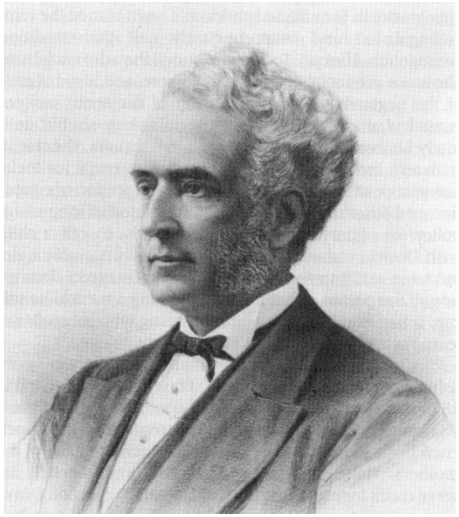
- Understand multiple systems involved and variable presentations, improving outlook for health
- Learn to use DS healthcare guidelines (for care providers and families) in the clinic to screen for common problems and optimize health
- Appreciate developmental variability
- Review some clinical research
- Even learn a few things you didn't know you needed to know

There are about 400,000 people in the U.S. with Down Syndrome (DS)! (1:700 births)

21 World Down Syndrome Day

World DS Day is on 3 21

Dr. John Langdon Down



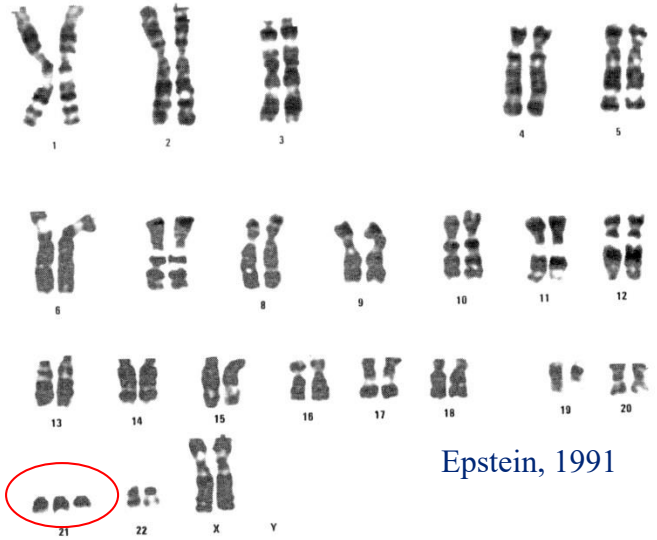
- Superintendent of Earlswood Asylum for Idiots, Surrey (1858-68)
- "Ethnic" classification of congenital idiocy: Mongolism (There terms are no longer acceptable, nor is mental retardation – preferred terms are Intellectual Disability or Cognitive Disability)
- Distinguished from cretinism (hypothyroidism) but no idea about genetics then
- "Down Syndrome" since 1961, after Lejeune discovered due to trisomy 21. (extra copy of chromosome 21)

1902

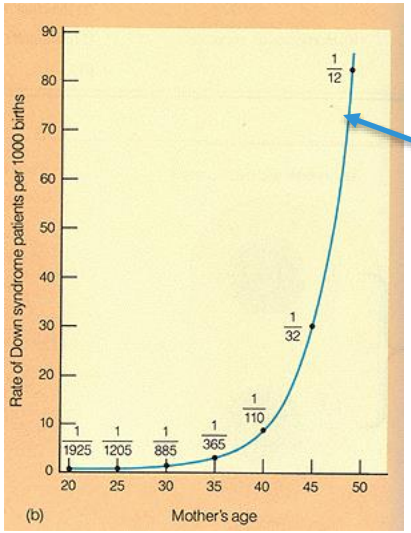
1930s

“Virgin and Child“ Andrea Mantegna (1430-1506)

Down Syndrome: Genetics



Epstein, 1991



Risk of having a child with DS increases with maternal age – 1:2000 at age 20, 1:100 at age 40, 1:8 at age 50.

http://www.micro.utexas.edu/courses/levin/bio304/genetics/motherage.gif

Key Points about Development in DS

- First words should always be: *Congratulations on your boy/girl/baby!*
- We expect children with DS to walk and talk, read, play sports, and have friends
- Children with DS don't stay like babies (and they are just as variable in personality and behavior as others)
- While some have more problems time will tell better than our guesses
- Every child with DS does not have problems in every area of possible concern

Images from
Thatdadblog.com
Noahsdad blog
Noahsdad.com

Development in DS

	Children with Down syndrome		"Normal" children	
	Average (months)	Range (months)	Average (months)	Range (months)
Smiling	2	1½–3	1	½–3
Rolling over	6	2–12	5	2–10
Sitting	9	6–18	7	5–9
Crawling	11	7–21	8	6–11
Creeping	13	8–25	10	7–13
Standing	10	10–32	11	8–16
Walking	20	12–45	13	8–18
Talking, words	14	9–30	10	6–14
Talking, sentences	24	18–46	21	14–32

	Children with Down syndrome		"Normal" children	
	Average (months)	Range (months)	Average (months)	Range (months)
Eating				
Finger feeding	12	8–28	8	6–16
Using spoon/fork	20	12–40	13	8–20
Toilet training				
Bladder	48	20–95	32	18–60
Bowel	42	28–90	29	16–48
Dressing				
Undressing	40	29–72	32	22–42
Putting clothes on	58	38–98	47	34–58

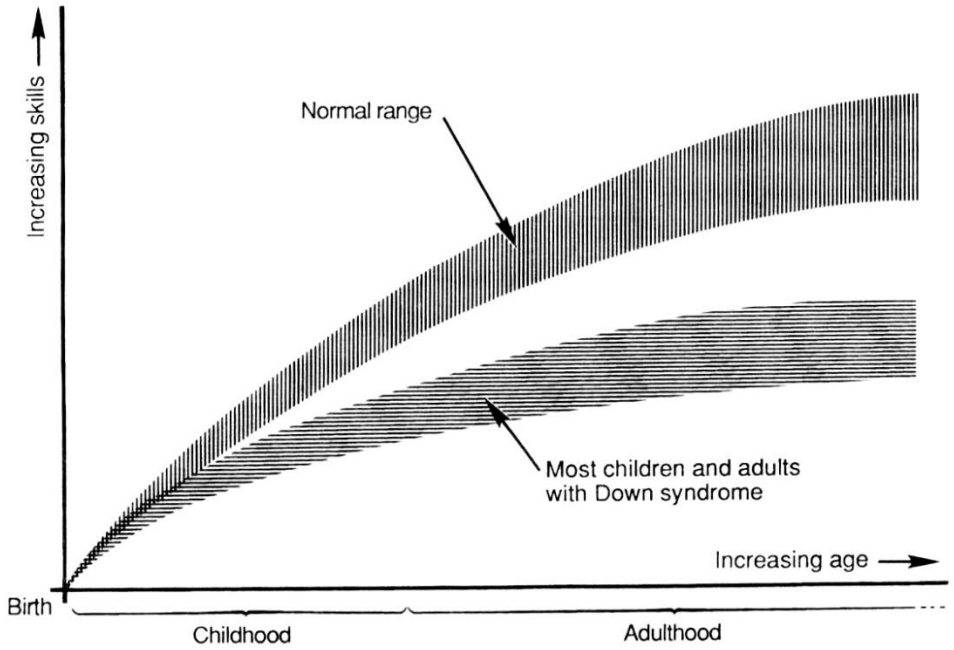


Figure 12 Rate of development.

Developmental delays are seen in most areas including language and motor skills – but most kids eventually get there and walk well, communicate well – most need help in many areas but many do learn to read, write, are physically active, have good friends and have their own strengths and weaknesses like all of us

Peabody Developmental Motor Scale- 2nd Edition (PDMS-2)

Over 500 children tested in our clinic ages 0 to 6 years.

FMQ (Fine motor quotient) is often reported, and score drops with age in Down syndrome, which is ***frustrating for many parents*** **since they know their skills are improving**

Our study shows that while FMQ decreases, the raw scores for each test increase and are parallel to the typical curve.
Kids with DS keep gaining skills! (Parents need to hear this!)

Journal of Child Neurology
OnlineFirst
© The Author(s) 2025, Article Reuse Guidelines
https://doi.org/10.1177/08830738251387929

Sage Journals

Original Article

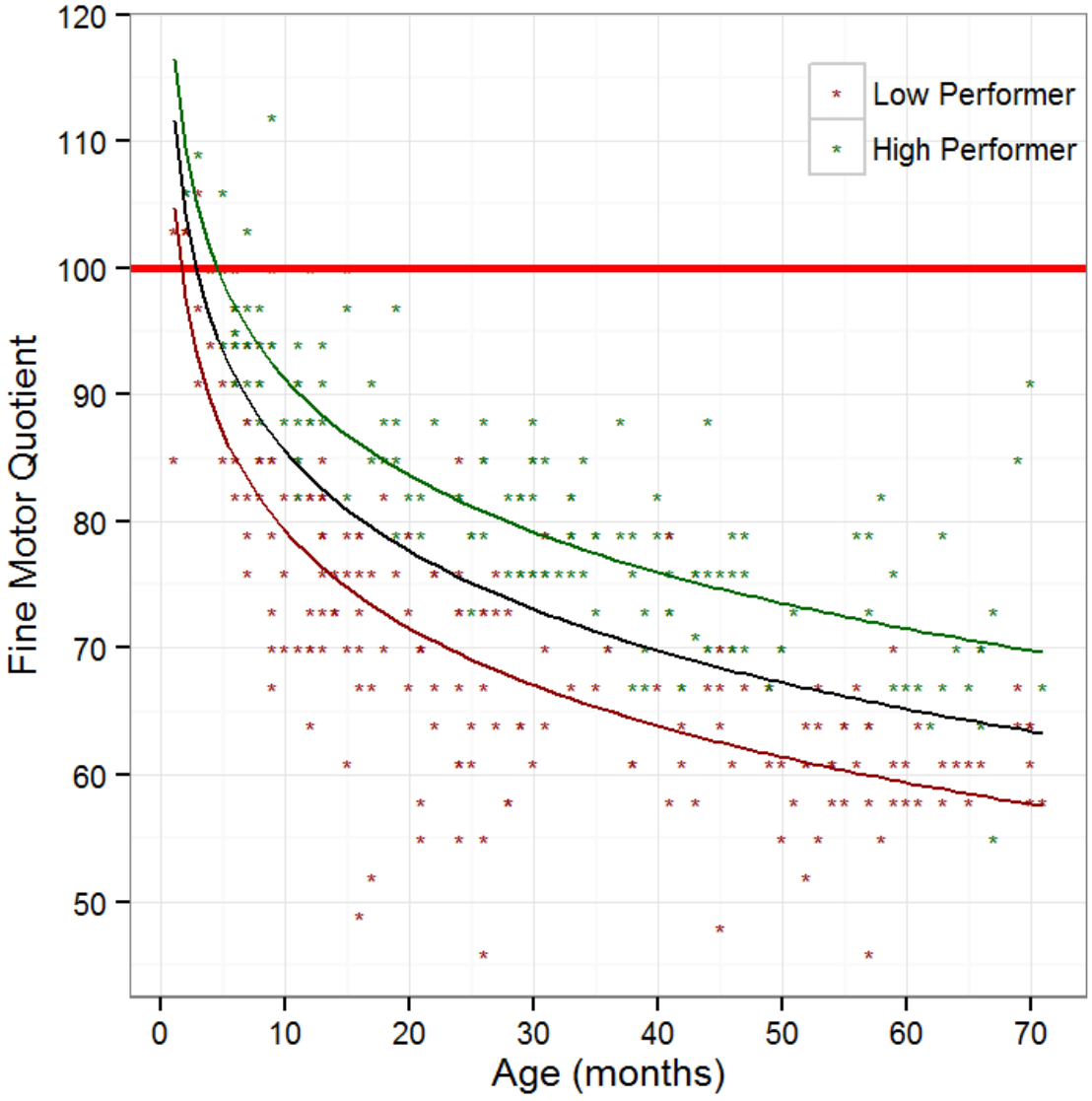
Fine Motor Development Growth Curves for Down Syndrome

Mandie Wiebers Jensen, MD, OT [1], Margaret Wolf, OTR/L [2], Alicia Feryn, MS [3], Thuan Nguyen, PhD [4], Isabel Katlaps, MD [5], and Joseph D. Pinter, MD, MBA [6]

Abstract

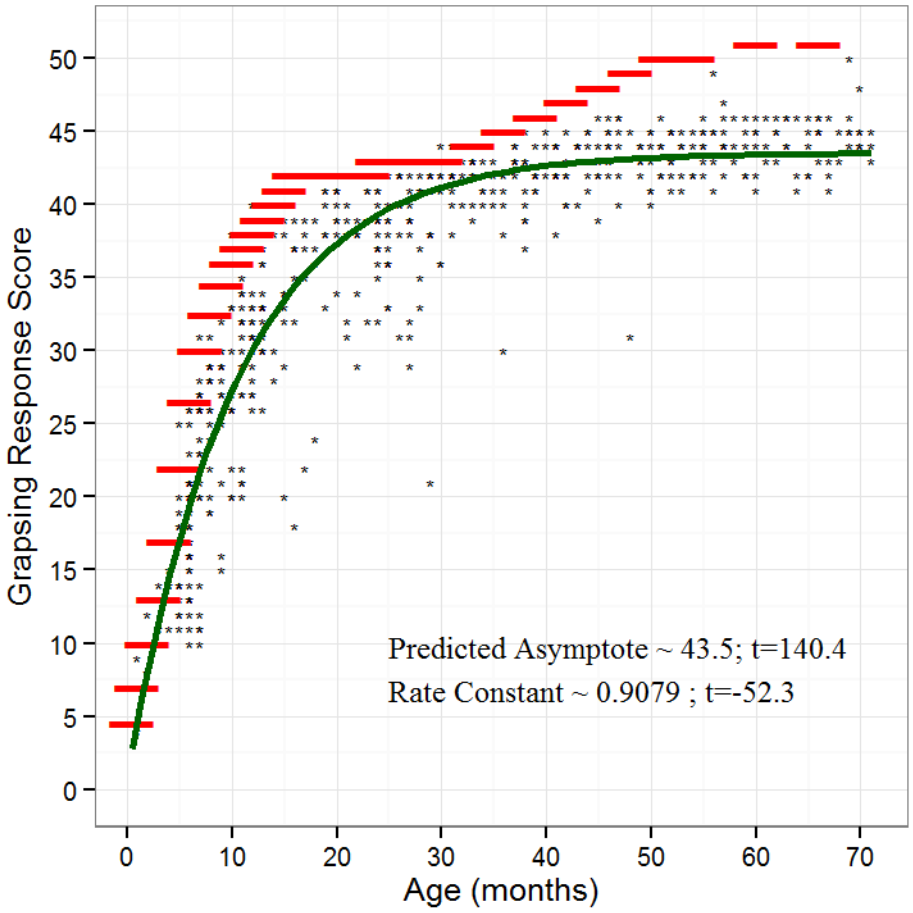
This study seeks to develop fine motor development growth curves for children with Down syndrome, specifically related to grasping and visual motor integration skills. A cross-sectional retrospective chart review was completed on a large cohort of children with Down syndrome from birth to 6 years of age who completed the Peabody Developmental Motor Scales–2nd Edition (PDMS-2). Although both fine motor and visual motor development were delayed in children with Down syndrome compared with typically developing peers, and a negative association between fine motor quotient and age was observed, grasping and visual motor integration raw scores increased with age. The Down syndrome–specific percentile curves developed here may enable providers to identify individuals requiring further investigation, and will be useful in future studies of factors that may influence fine motor development in Down syndrome. These data will also help parents better understand their children's development and interpret developmental test results.

Down Syndrome FMQ Scores with age (100 = typical average)

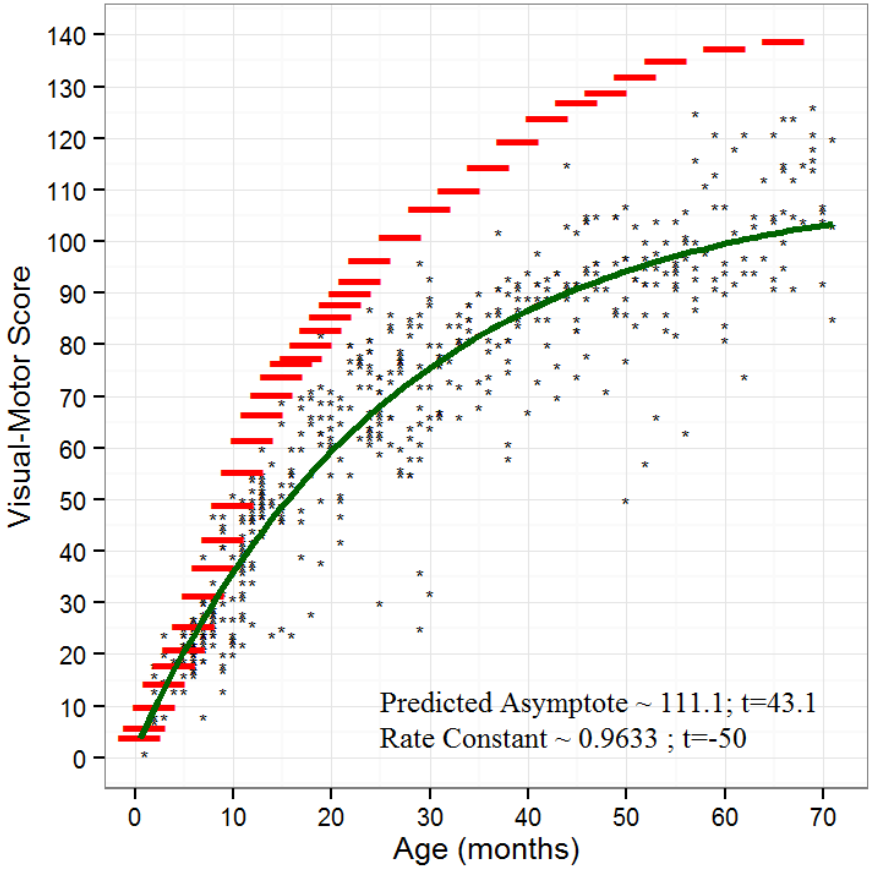


Down Syndrome Peabody Fine Motor Raw Scores

Grasping Raw Score



Visual Motor Raw Score



Systems affected in DS

- *Cardiac (50%)*
- *GI (10%) – duodenal atresia, Hirschsprung's, Celiac disease*
- *Ophthalmologic* strabismus, nystagmus, cataracts, vision problems – Ophthalmology exam by one year of age and yearly
- *Endocrine* hypothyroidism
- *Hematology/Oncology* leukemia, testicular cancer (but other types of cancer VERY RARE)
- *Infectious diseases: More frequent* otitis and upper respiratory infections
- *Audiologic – Middle ear fluid, hearing loss*
- *Pulmonary/Sleep* OSA* in at least half
- *Orthopedic* C1-2 instability (+ knees & hips & FEET) – in part due to ligamentous laxity

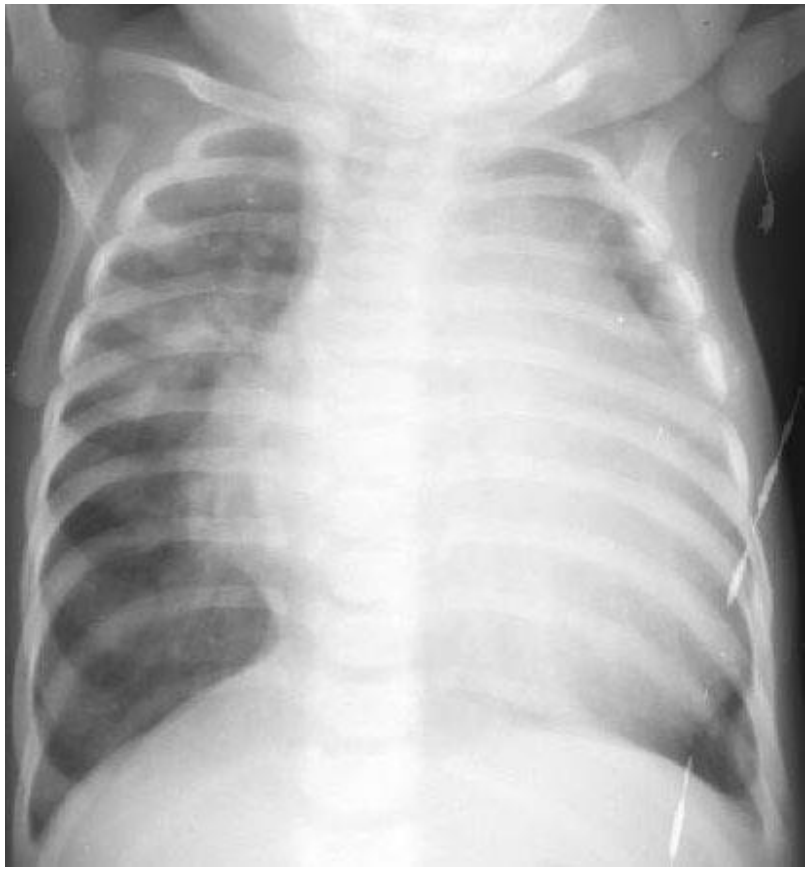
*OSA = obstructive sleep apnea

Medical Advances Have Improved Health in Many Areas

- Congenital heart defect: In ½; Early surgery needed in ¼; > 90% success rate
- Hematology: Leukemia in 1%, cure rate is 70-95%
- Hypothyroidism: Very common (1/2) and Usually easy to treat and important to make sure optimal development and growth

Cardiac Issues

- 50% with congenital heart defects (and half of these require surgery in first year)
- AV canal defects most common (60%)
- Isolated ASD, VSD, PDA (30%)
- Tetralogy of Fallot (7%)
- *Rapid resp, feeding diff, fatigue, congestive heart failure*



Hematology-Oncology

- Leukemia is much more common in DS, especially under age 5
- Solid tumors are much less common overall with exception of testicular cancer, which is seen at 5 to 10 x rate (at least) in DS
  - *Do/teach testicular exam for adolescents/young adults*

Leukemia

- Much more common in DS
- Overall very good survival/cure
  - 70-90+% depending on type but long-term toxicities can be problem
- *ALL: 60% of DS cases*
- *AML: 40%*
  - *Main type is AMKL (Acute megakaryoblastic leukemia)*
  - *500 x more common in DS than non-DS*

Hypothyroidism in DS

- Over 500 patients studied here:
  - 50% with thyroid abnormality by age 18
  - 1/5 of those are diagnosed by 6 months!
  - Important to get TSH/Free T4 by age 6 months at latest (we suggest even earlier will be better), then as per guidelines
  - TSH does tend to run a bit high – recheck in a few months if slightly high TSH and normal FT4 ....*or refer to Ped Endo*

Original Paper

HORMONE RESEARCH IN PÆDIATRICS

Horm Res Paediatr
DOI: 10.1159/000457952

Received: October 20, 2016
Accepted: January 24, 2017
Published online: March 3, 2017

Characterization of Thyroid Abnormalities in a Large Cohort of Children with Down Syndrome

Melinda J. Pierce[a] Stephen H. LaFranchi[a] Joseph D. Pinter[b]

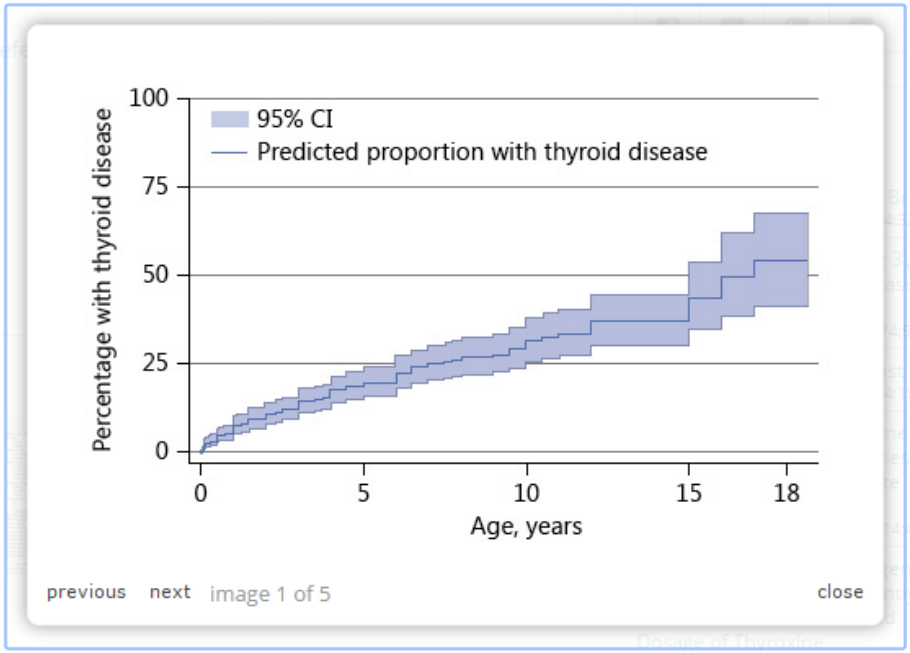
[a]Department of Pediatrics, Division of Endocrinology, Oregon Health and Science University, Portland, OR, USA;
[b]Institute on Development and Disability, Oregon Health and Science University, Portland, OR, USA

Keywords
Down syndrome · Thyroid diseases · Congenital heart malformation · Congenital hypothyroidism · Screening

Abstract
Background/Aims: Thyroid disease is a common comorbidity in individuals with Down syndrome (DS), but historical studies have multiple limitations. We assessed thyroid abnormalities in a large cohort of children with DS. ***Methods:*** Retrospective records review from a single institution. Calculated prevalence of common thyroid abnormalities and associations with common comorbidities. ***Results:*** Among 508 patients, 120 (24%) had a thyroid-related diagnosis, the majority having elevated thyrotropin treated with levothyroxine. A Kaplan-Meier estimate projects that 50% have thyroid disorder by adulthood, with 20% of hypothyroidism diagnosed before the age of 6 months. When tested, approximately 50% had positive antithyroid antibodies, though this rate was 100% in overt hypothyroidism. There was no association between congenital or acquired hypothyroidism and common comorbidities. ***Conclusion:*** Thyroid disease in DS is more common and occurs earlier than in the general population, and is often transient. Thyroid disease is unrelated to lated to autoimmunity; we recommend checking of antithyroid antibodies only in select cases. An additional screen for thyroid disease betwe[illegible] 6-month well-child visit roidism who passed the

Introduction

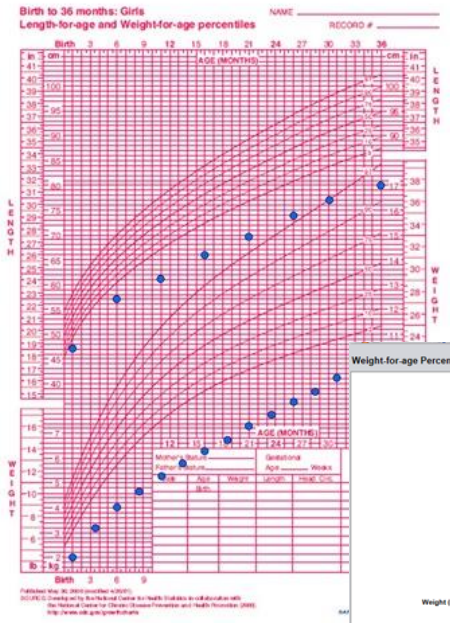
Down syndrome (D aneuploidy with a nati 10,000, or 1 in 691, live patients with DS are at of thyroid hormone ab alence between 8 and of abnormality [2–18]. disease: congenital hyp hypothyroidism; acqui perthyroidism. The w numbers in historical studies have greater nu Many studies also lack sociations of thyroid di

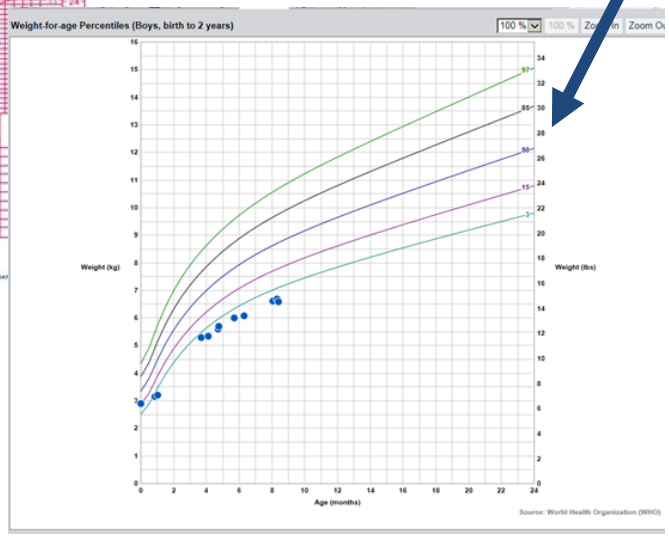


OHSU

Which curves to use?

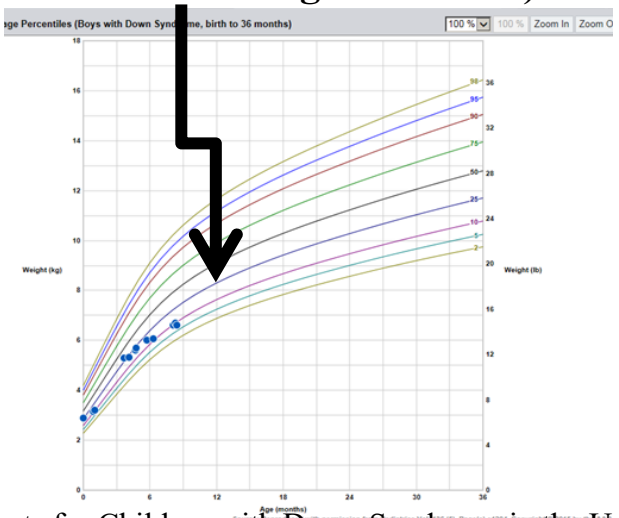
When young, standard growth curves result in most dropping below curves for Ht, Wt, HC





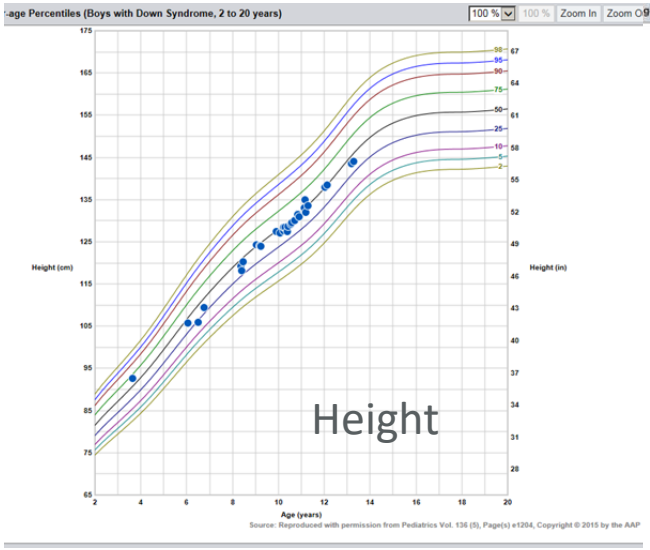
Final height is usually about 6 inches (15 cm) shorter than sib of same sex

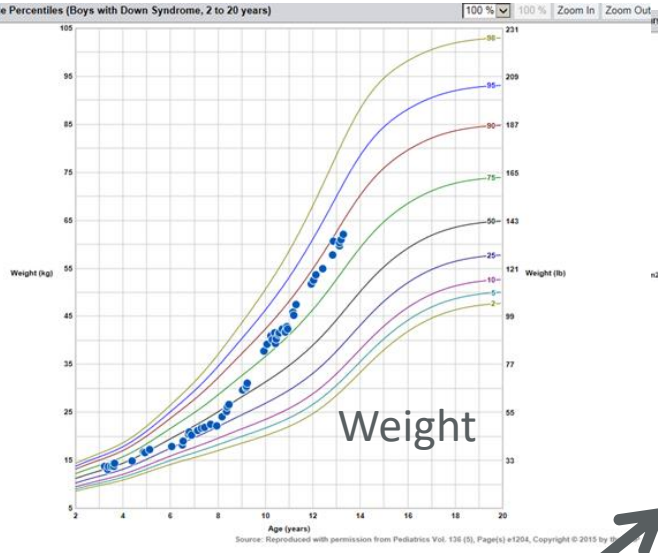
DS weight curve
Does not (usually) need a Nutrition consult
(but may need help with Feeding difficulties)

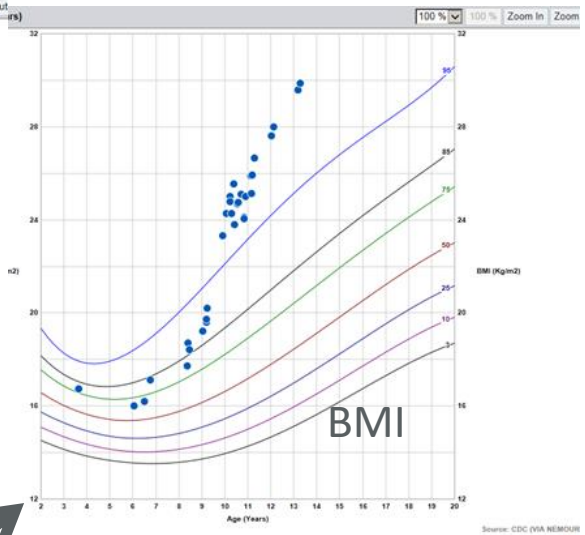


Zemel et al. Growth Charts for Children with Down Syndrome in the U.S. Pediatrics. 2015

Teenagers with DS often seem "normal" for weight on DS Curves







See: Hatch-Stein, et al, **Body Composition and BMI Growth Charts in Children With Down Syndrome**
Pediatrics 2016
Encourages use CDC BMI charts – a good start!

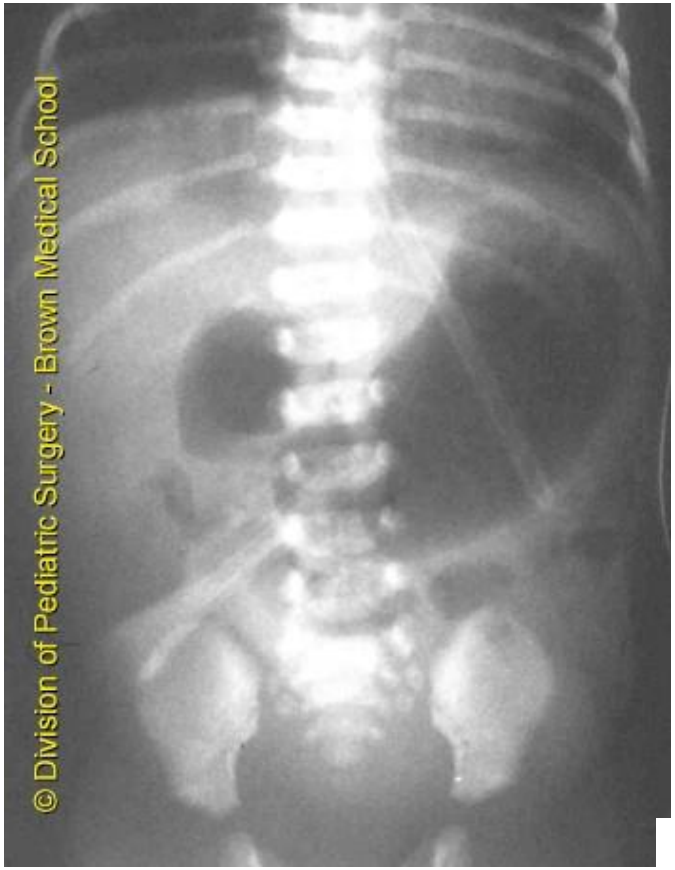
But the BMI ---and the physical exam – tell another story!

Pay attention to BMI (body mass index)

Obesity contributes to OSA and decreased fitness

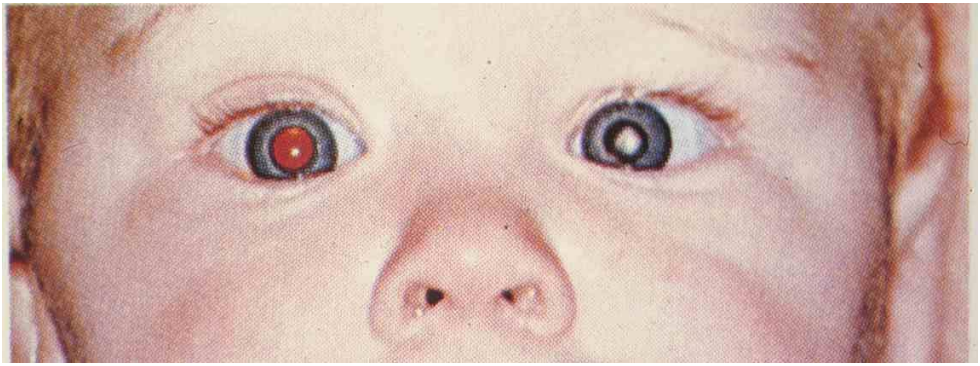
Gastrointestinal Problems

- GI malformations in 12%
  - Duodenal atresia/stenosis
  - Pyloric stenosis
  - Hirschprung's
- **Celiac Disease in up to 3%**
  - **Screen total serum IgA and anti-TTG IgA**



Ophthalmologic Problems

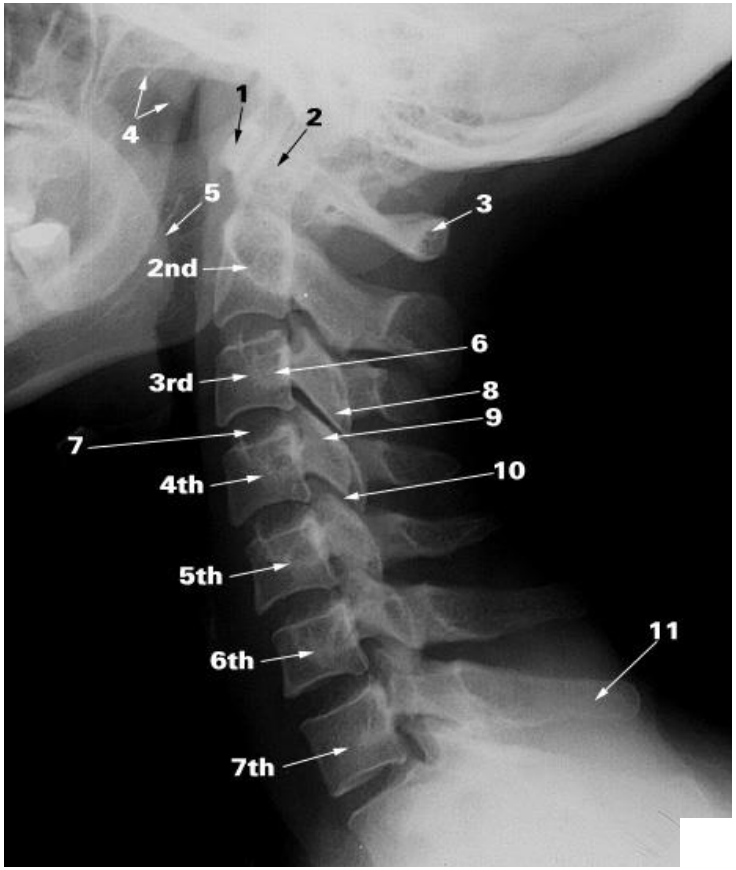
- **Cataracts in 1-2%**
- Nystagmus
- Strabismus
- *Note pseudo-strabismus very common due to epicanthal folds*
- *Poor visual acuity*



Orthopedic/Sports Participation

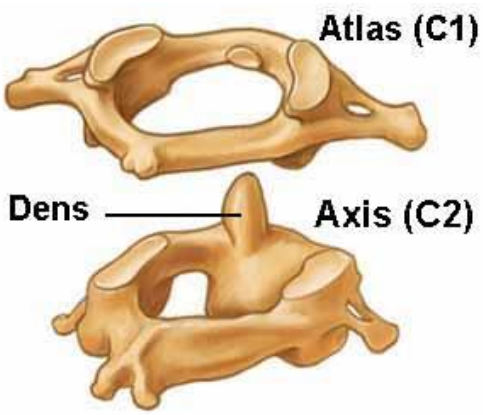
So why all the hubbub about neck xrays?

- **Atlanto-axial instability** common (20%) in DS, usually asymptomatic (Surgery needed in about 1%)
- Screening for Special Olympics, etc. (No longer required)
- Controversial if predicts symptomatic (BUT WE OFTEN SCREEN ANYWAY; ***ALWAYS CONSIDER IF NECK PAIN/SIGNS/SYMPTOMS OF CONCERN)***
- Ligamentous laxity (also hips, knees, ankles) is common

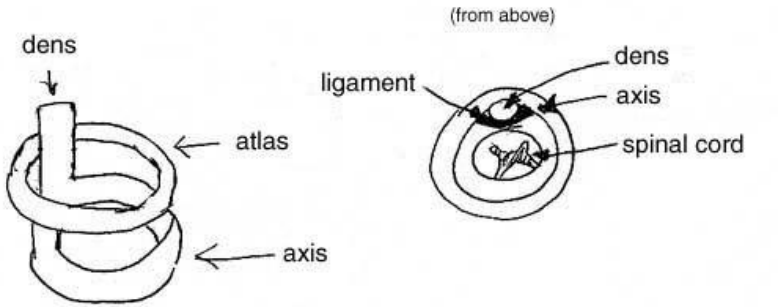


Orthopedic/Sports Participation

- Atlanto-dens interval: normal < 3-5 mm
- Neural canal width: normal > 14 mm
- 1-2% with symptomatic AAI or AAS (instability/subluxation)
- Sx: Neck pain, abnl gait, weakness, bowel/bladder probs
- Tx: Fusion of C1 and C2
- TAKE HOMES:
  - Neck xrays for high intensity actvities (gymastics, football) – If do, do them right
  - Even if not required, even if normal before, if there are new Sx then do them again and consider MRI and referral to Ped NSurg



https://www.spineuniverse.com/anatomy/vertebral-column



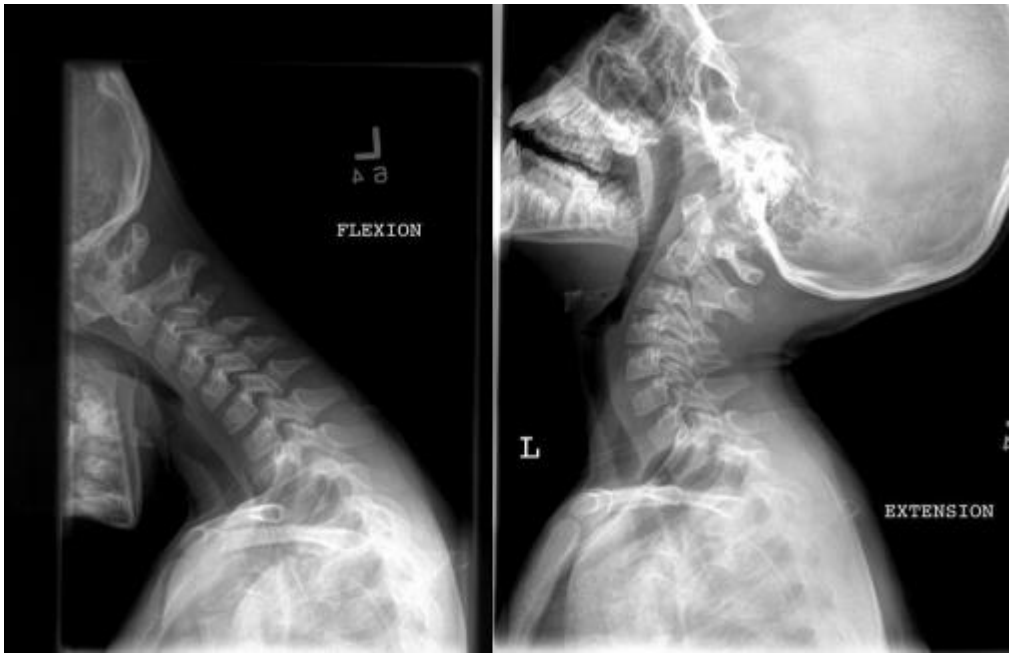
C spine X-rays:

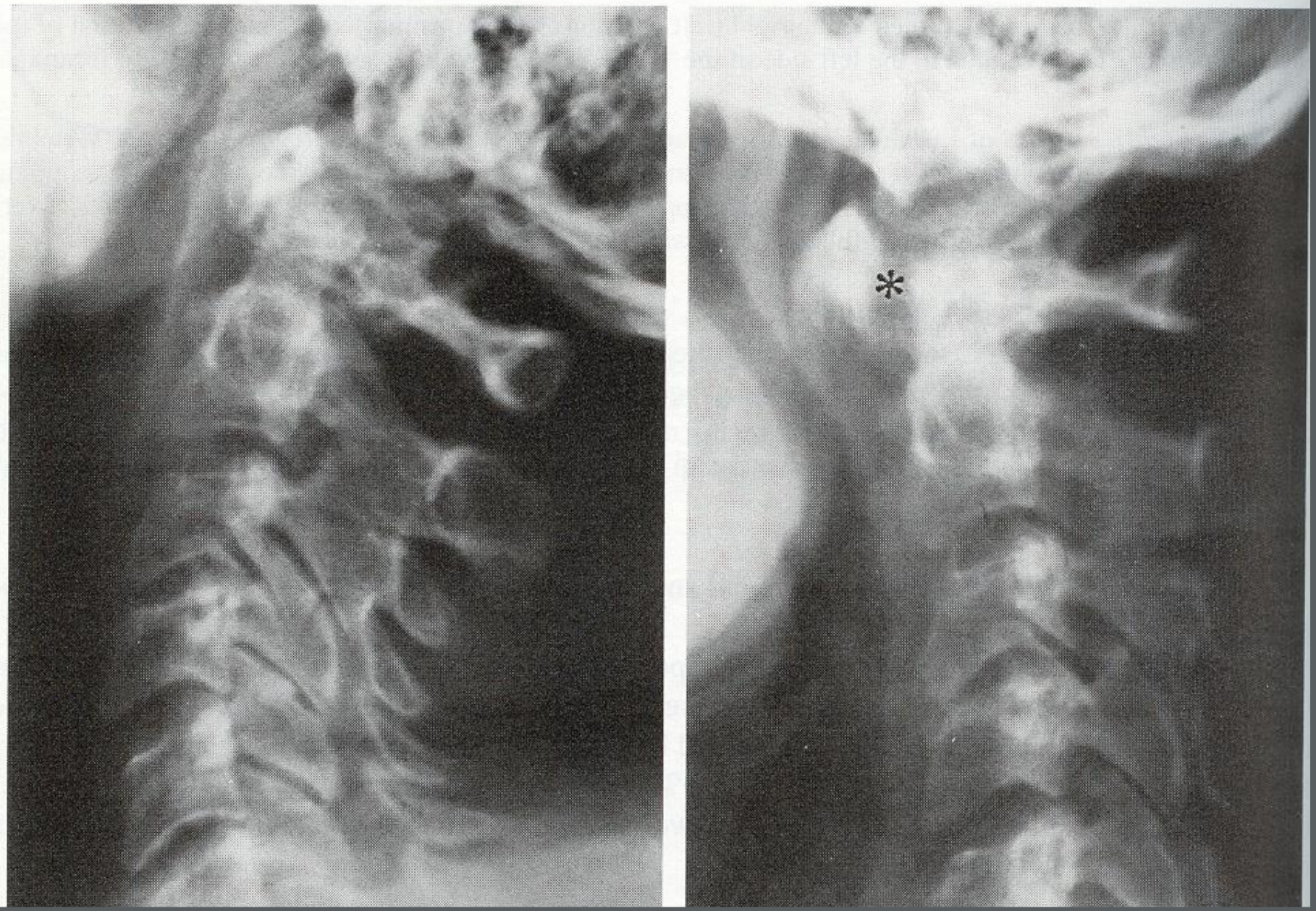
Specify in order:

"3 views, Lateral only, Neutral, flexion, extension Measure atlantodens interval and neural canal width in each view to rule out atlantoaxial instability"

--If you just order 3 view neck xrays, you usually get trauma series which does not evaluate dynamic instability

For young kids: in parent's lap with cell phone with video held by parent in front, above, below = >90% success rate

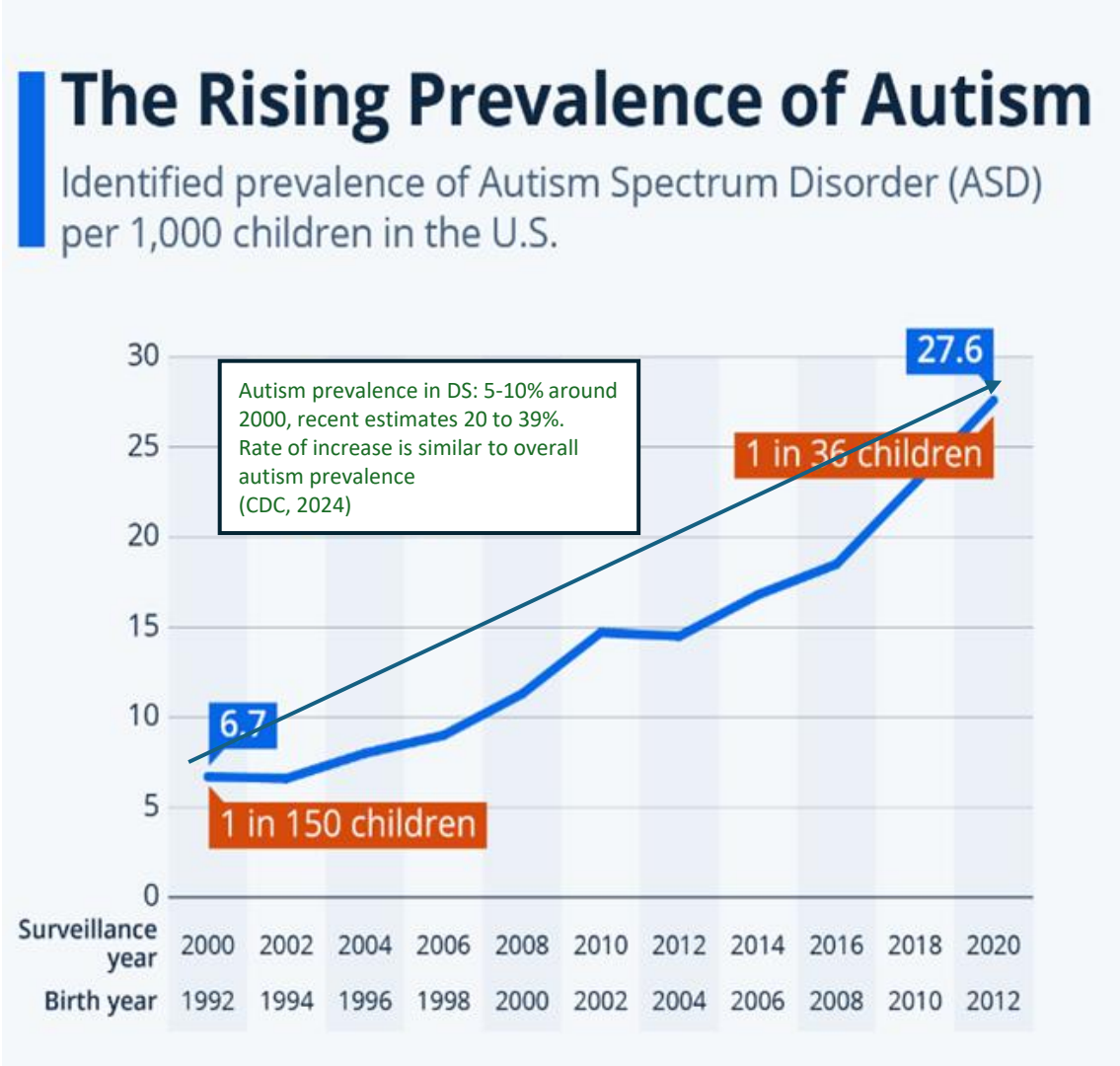




NEUROLOGIC (100%)

- **Hypotonia, motor delays**
- **Developmental delays, Cognitive/intellectual disability**
  - **Speech and Language especially impaired in most**
- **Seizures:**
  - Infantile spasms (IS) in 3 to 5% of kids with Down syndrome (usually around 6 months of age)
- Autism in 5-10% (or more)

•*Eisermann, et al. **Infantile spasms in Down syndrome--effects of delayed anticonvulsive treatment** Epilepsy Res. 2003


The Rising Prevalence of Autism
Identified prevalence of Autism Spectrum Disorder (ASD) per 1,000 children in the U.S.
Autism prevalence in DS: 5-10% around 2000, recent estimates 20 to 39%.
Rate of increase is similar to overall autism prevalence
(CDC, 2024)
27.6
1 in 36 children
6.7
1 in 150 children
30
25
20
15
10
5
0
Surveillance year
2000 2002 2004 2006 2008 2010 2012 2014 2016 2018 2020
Birth year
1992 1994 1996 1998 2000 2002 2004 2006 2008 2010 2012

Psychological/Behavioral

- Similar or increased incidence of most psychiatric conditions:
  - Depression, sleep disturbances (OSA – obstructive sleep apnea – often big tonsils in young children; often related to obesity in teens and adults)
  - Anxiety, ADHD, OCD
  - Autism
  - Dementia
- **Very much under-recognized**
  - **Not all kids with Down syndrome are always happy; just as variable as other kids**
- Consider underlying medical problems (Hypothyroidism; OSA – poor sleep quality can lead to behavioral and learning problems)
- **People with DS can have same conditions as anyone and should be offered same treatments and therapies**

Psychological/Behavioral

- Down Syndrome Regression Disorder (DSRD): A rare neuropsychiatric condition causing insomnia, catatonia, encephalopathy, and obsessive-compulsive behavior in otherwise healthy individuals with Down syndrome (DS).
- --Need to rule out other conditions – thyroid, depression, etc
- **Most cases with likely immunological basis, many respond to immune therapies**
- **See articles by Santoro (LA Children's)**
- **(and Dr. Christy of Prov St V)**

A Promising Future Together

A Guide for New and Expectant Parents

national down syndrome society
ndss

DS healthcare guidelines are helpful but LONG

- Guidelines available at www.ndss.org in publications and also published in *Pediatrics* (journal)
- Older, simpler checklists are easier to use in clinic and parents can be in charge of keeping track of what is needed at each age

APPENDIX 1 Health Supervision for Children With Down Syndrome

	Prenatal	Birth–1 mo	1 mo–1 y	1–5 y	5–13 y	13–21 y
Counseling regarding prenatal screening test & imaging results						
Plan for delivery						
Referral to geneticist						
Parent-to-parent contact, support groups, current books and pamphlets						
Physical exam for evidence of trisomy 21						
Chromosomal analysis to confirm dx						
Discuss risk of recurrence of Down syndrome						
Echocardiogram						
Radiographic swallowing assessment if marked hypotonia, slow feeding, choking with feeds, recurrent or persistent respiratory sx, FTT						
Eye exam for cataracts						
Newborn hearing screen and follow-up						
Hx and PE assessment for duodenal or anorectal atresia						
Reassure parents delayed and irregular dental eruption, hypodontia are common						
If constipation, evaluate for limited diet or fluids, hypotonia, hypothyroidism, GI malformation, Hirschsprung		Any visit				
CBC to R/O transient myeloproliferative disorder, polycythemia						
Hb annually; CBP & ferritin or CHr if possible risk iron deficiency or Hb <11 g				Annually		
Hemoglobin						Annually
TSH (may be part of newborn screening)			6 and 12 mo	Annually		
Discuss risk of respiratory infection						
If cardiac surgery or hypotonic: evaluate apnea, bradycardia, or oxygen desaturation in car seat before discharge						
Discuss complementary & alternative therapies		All health maint. visits				
Discuss cervical spine positioning, especially for anesthesia or surgical or radiologic procedures		All health maint. visits				
Review signs and symptoms of myelopathy		All health maint. visits				
If myopathic signs or symptoms: obtain neutral position spine films and, if normal, obtain flexion & extension films & refer to pediatric neurosurgeon or orthopedic surgeon with expertise in evaluating and treating atlanto-axial instability		Any visit				
Instruct to contact physician for change in gait, change in use of arms or hands, change in bowel or bladder function, neck pain, head tilt, torticollis, or new-onset weakness				Biennially		
Advise risk of some contact sports, trampolines				All health maint. visits		
Audiology evaluation at 6 mo						
If normal hearing established, behavioral audiogram and tympanometry until 3 behavioral ear specific testing possible. Refer child with abnormal hearing to ot				Every 6 mo		
If normal ear-specific hearing established, behavioral audiogram				Annually		
Assess for obstructive sleep apnea Sx				All health maint. visits		
Sleep study by age 4 years						
Opht thalmology referral to assess for strabismus, cataracts, and nystagmus						
Refer to pediatric ophthalmologist or ophthalmologist with expertise in with Down syndrome				Annually	Every 2 y	Every 3 y
If congenital heart disease, monitor for signs & Sx of Congestive heart failure		All visits				
Assess the emotional status of parents and intrafamilial relationships		All health maint. visits				
Check for Sx of celiac disease; if Sx present, obtain tissue transglutaminase IgA & quantitative IgA				All health maint. visits		
Early intervention: physical, occupational, and speech therapy				Health maint. visits		
At 30 months, discussion on transition to preschool and development of IEP						
Discuss behavioral and social progress				Health maint. visits		
Discuss self-help skills, ADHD, OCD, wandering off, transition to middle school					Health maint. visits	
If chronic cardiac or pulmonary disease, 23-valent pneumococcal vaccine at age >2 y						
Reassure regarding delayed and irregular dental eruption						
Establish optimal dietary and physical exercise patterns				Health maint. visits		
Discuss dermatologic issues with parents						
Discuss physical and psychosocial changes through puberty, need for gynecologic care in the pubescent female						
Facilitate transition: guardianship, financial planning, behavioral problems, school placement, vocational training, independence with hygiene and self-care, group homes, work settings						Health maint. visits
Discuss sexual development and behaviors, contraception, sexually transmitted diseases, recurrence risk for offspring						Health maint. visits
		Do once at this age				
		Do if not done previously				
		Repeat at indicated intervals				

Maint. indicates maintenance; dx, diagnosis; sx, symptoms; FTT, failure to thrive; Hx, history; PE, physician examination; GI, gastrointestinal; CBC, complete blood count; R/O, rule out; Hb, hemoglobin; ot, occupational therapy; CHr, reticulocyte hemoglobin; IgA, immunoglobulin A; IEP, Individualized Education Plan; ADHD, attention-deficit/hyperactivity disorder; OCD, obsessive compulsive disorder.

Downloaded from pediatrics.aappublications.org at Oregon Health & Science University on March 9, 2014

Bull, et al. American Academy of Pediatrics. Health Supervision for Children with Down Syndrome. *Pediatrics.* 2011;128(2):393–406

The older guidelines are more manageable in a clinic visit (1999)

Reprinted from *Down Syndrome Quarterly*, Volume 4, Number 3, September 1999

Down Syndrome Health Care Guidelines (1999 Revision) Record Sheet

Sheet #1: Birth to Age 12 Years

Name: _____ **Birthday:** _____

Medical Issues	Age, in years: At Birth or at Diagnosis	6-mo	1	1-1/2	2	2-1/2	3	4	5	6	7	8	9	10	11	12
Karotype & Genetic Counseling	____															
Usual Preventative Care	____	___	___	___	___	___	___	___	___	___	___	___	___	___	___	___
Cardiology	Echo															
Audiologic Evaluation	ABR or OAE	___	___	___	___	___	___	___	___	___	___	___	___	___	___	___
Ophthalmologic Evaluation	Red reflex	___	___	___	___	___	___	___	___	___	___	___	___	___	___	___
Thyroid (TSH & T_4)	State screening	___	___	___	___	___	___	___	___	___	___	___	___	___	___	___
Nutrition	____	___	___	___	___	___	___	___	___	___	___	___	___	___	___	___
Dental Exam[1]					___	___	___	___	___	___	___	___	___	___	___	___
Celiac Screening[2]					___											
Parent Support	____	___	___	___	___	___	___	___	___	___	___	___	___	___	___	___
Developmental & Educational Services	Early Intervention	___	___	___	___	___	___	___	___	___	___	___	___	___	___	___
Neck X-rays & Neurological Exam[3]							X-ray	___	___	___	___	___	___	___	___	___
Pneumococcal Conjugate Vaccine Series		_____														

Instructions: Perform indicated exam/screening and record date in blank spaces. The grey or shaded boxes mean no action is to be taken for those ages.

[1]Begin Dental Exams at 2 years of age, and continue every 6 months thereafter.
[2]IgA antiendomysium antibodies and total IgA.
[3]Cervical spine x-rays: flexion, neutral and extension, between 3-5 years of age. Repeat as needed for

Reprinted from *Down Syndrome Quarterly*, Volume 4, Number 3, September, 1999

Down Syndrome Health Care Guidelines (1999 Revision) Record Sheet

Sheet #2: 13 Years to Adulthood

Name: _____ **Birthday:** _____

Medical Issues	Age, in years: 13	14	15	16	17	18	19	20-29
Usual Preventative Care	___	___	___	___	___	___	___	___
Audiologic Evaluation	___	___	___	___	___	___	___	___
Ophthalmologic Evaluation	___	___	___	___	___	___	___	___
Thyroid (TSH & T_4)	___	___	___	___	___	___	___	___
Nutrition	___	___	___	___	___	___	___	___
Dental Exam[1]	___	___	___	___	___	___	___	___
Parent Support	___	___	___	___	___	___	___	___
Developmental & Educational Services	___	___	___	___	___	___	___	___
Neck X-rays & Neurological Exam[2]	___	___	___	___	___	___	___	___
Pelvic exam[3]				___	___	___	___	___
Assess Contraceptive Need[3]				___	___	___	___	___

Instructions: Perform indicated exam/screening and record date in blank spaces. The shaded boxes mean no action is to be taken for those ages.

[1]Begin Dental Exams at 2 years of age, and continue every 6 month thereafter.
[2]Cervical spine x-rays: flexion, neutral and extension, between 3-5 years of age. Repeat as needed for Special Olympics participation. Neurological examination at each visit.
[3]If sexually active.
© Down Syndrome Quarterly, 1999. This record sheet may be printed out for individual use but may not be reproduced on any website without prior permission.

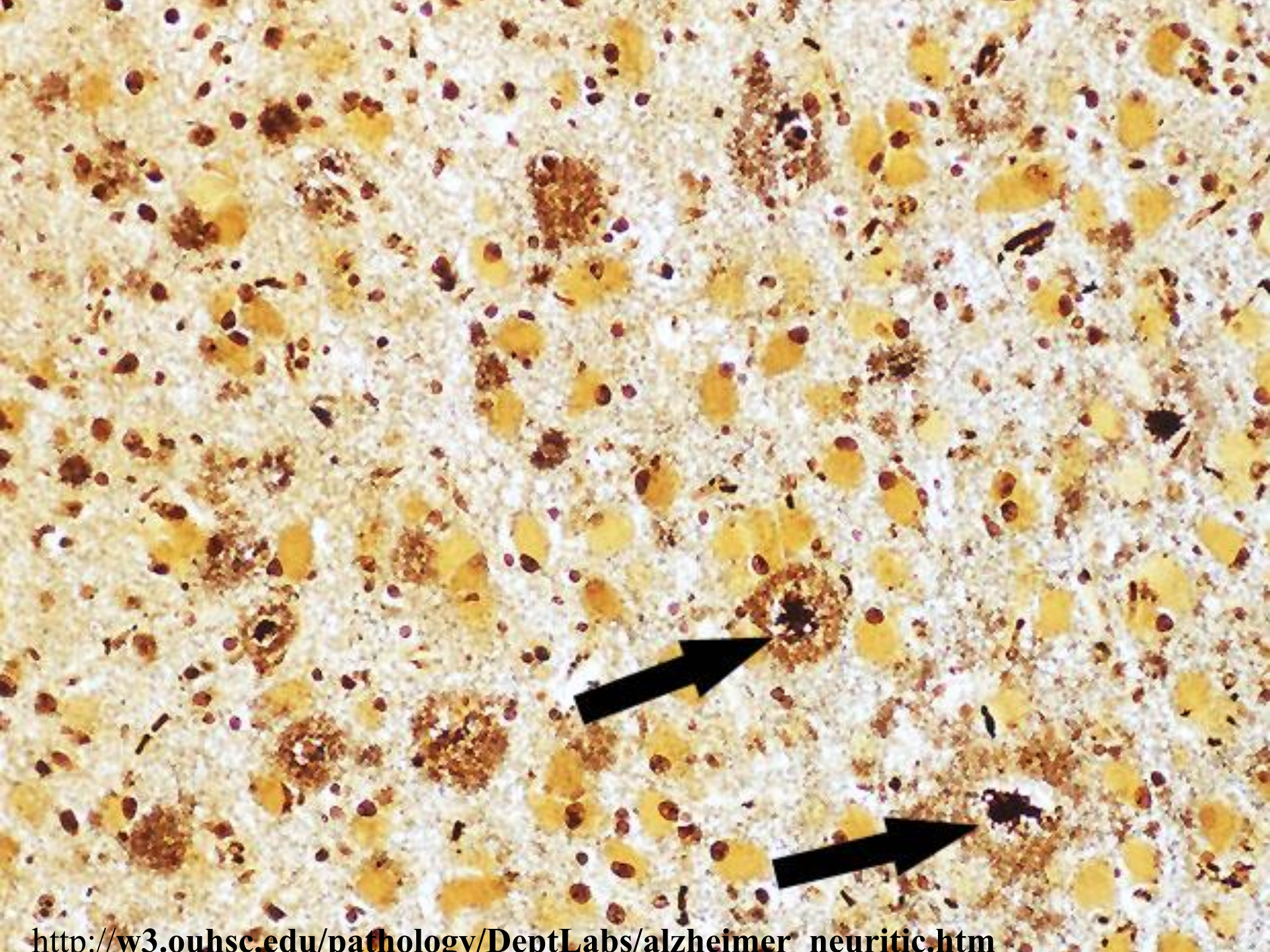
Go to Birth to Age 12 Years Record Sheet

Go to the Health Care Guidelines for People with DS full article

Back to Down Syndrome: Health Issues

Adults with Down Syndrome

- Be Aware of Medical Problems
  - Cardiac Problems (mitral valve prolapse/regurg; pulm HTN/heart failure (esp with obstructive sleep apnea)
  - OSA
  - Thyroid
  - Neck problems (AAI)
  - Psychosocial Issues
  - Psychiatric Issues: Depression
  - Dementia:
    - Early-onset Alzheimer's (50% by age 50)
    - Pseudodementia from other conditions



http://w3.ouhsc.edu/pathology/DeptLabs/alzheimer_neuritic.htm

Why interest in the DS-Alzheimer connection NOW?

- 5 million Americans NOW have AD
- 13.8 million by 2050
- Cost in 2015: $226 billion
- Cost by 2050: $1 trillion

http://www.alz.org/facts/overview.asp

The future of DS research and treatments

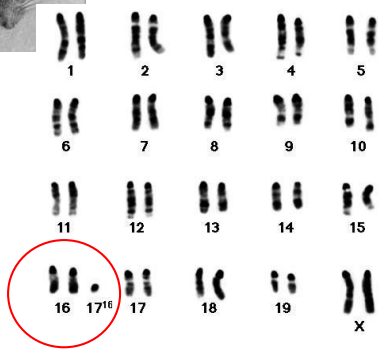
- Non-medical/educational/therapies

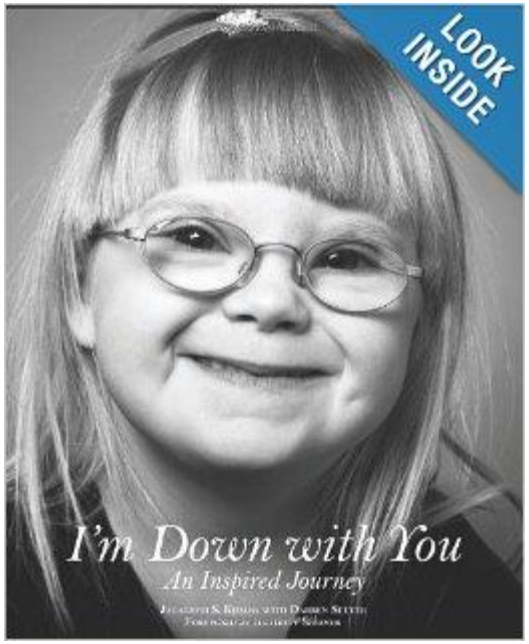
(Education/activity/experience do change your brain!)

- Non-specific/common sense (exercise/diet)
- Medical (thyroid, sleep, obesity)
- Targeted biological (much via mouse work)
  - Alzheimer disease-based treatments (cholinergic, beta amyloid modifiers)
  - Animal model-based therapies targeting neurotransmitters (SSRI's, GABA)?)

  (Caution: Lots of things work in mice but not in people)
- Gene therapy (NGF, APP, other targets (Xist))
- Prenatal therapy: with diagnosis in 1st trimester could affect course of later fetal brain development

  Prenatal maternal Folate helps prevent spina bifida – is that something like Folate to help prevent some of the problems in DS?

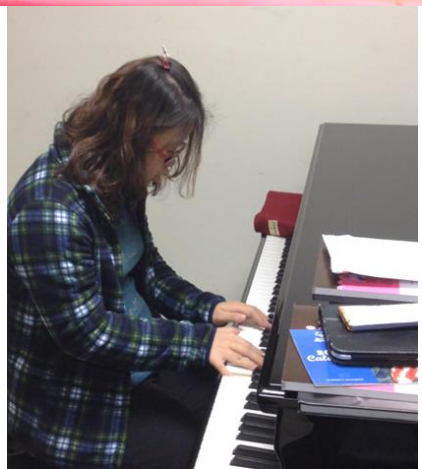



LOOK INSIDE
I'm Down with You
An Inspired Journey


still a battle to be won

What can a person with Down syndrome do?

Some become musicians



Some become artists

Fundacion John Langdon Down, Mexico City
http://www.fjldown.org.mx/

People with DS
100,000 in Mexico
30,000 in Mexico City

What can a person with Down syndrome do?

- Some grow up to be athletes

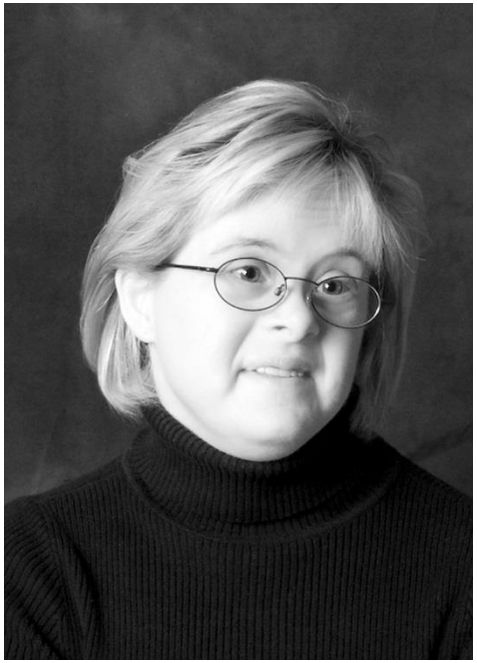
http://www.youtube.com/watch?v=flQZcosp2-k

What can a person with Down syndrome do?

- Some grow up to be athletes

http://www.youtube.com/watch?v=flQZcosp2-k

Karen Gaffney: Advocate, Motivational Speaker (TED Talk and international), Honorary Doctorate, Long-Distance Swimmer and…
OHSU Down Syndrome Clinic and CDRC team member

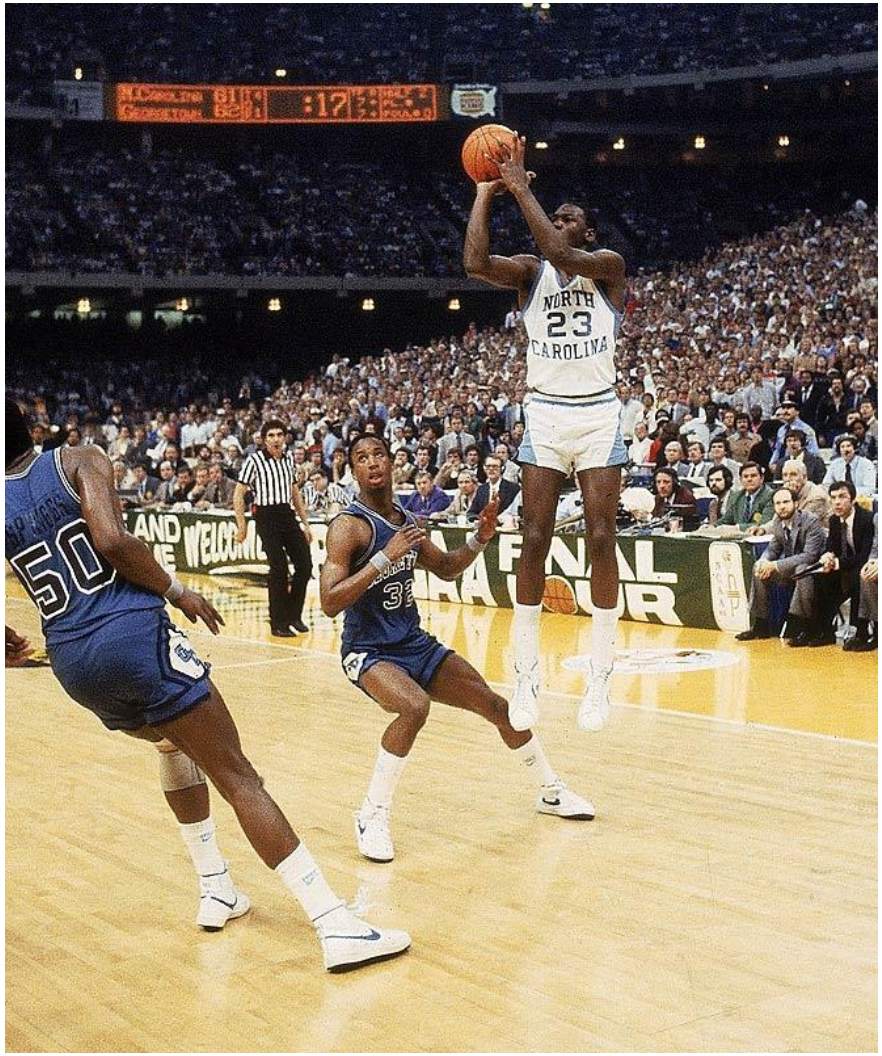


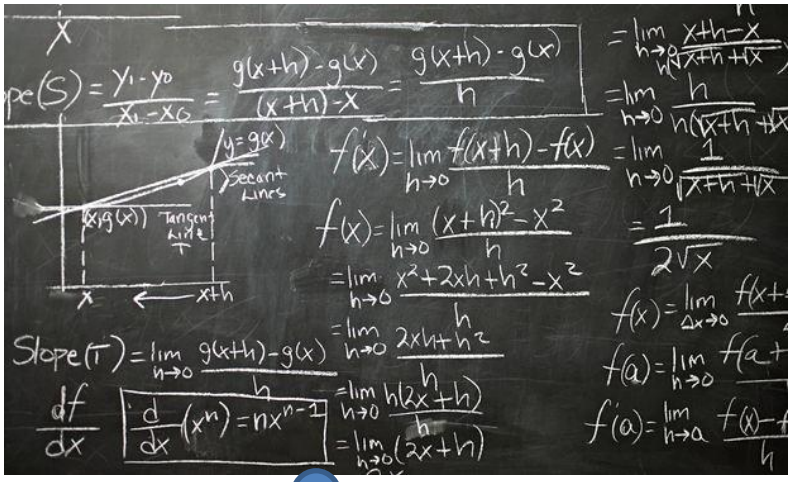
Teen with Down syndrome
becomes first ever to reach Mt. Everest basecamp (17,600 ft)
after hiking 70 miles over 19 days

What can a person with Down syndrome do?

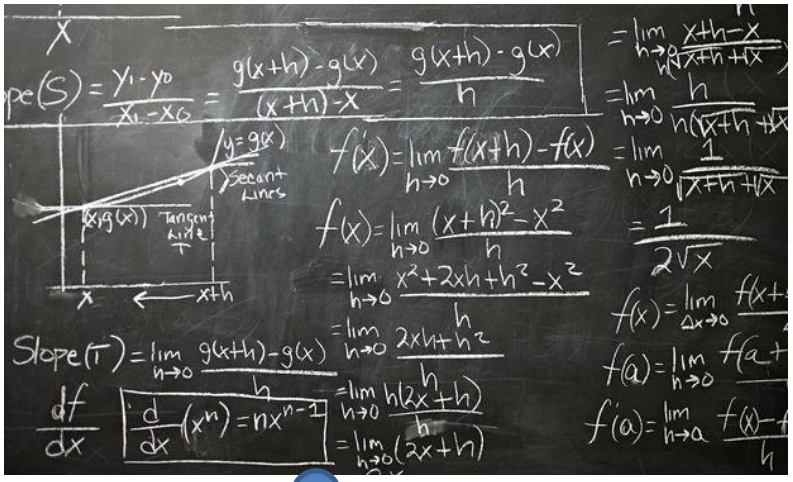
- Like all of us and all kids, not everyone can sing, dance or lift 200 kg
- (and NOBODY can if they are not given a chance)
- **ALL people with Down syndrome grow up with their own unique talents and challenges, and our goal should be (as with all our kids) to help them achieve their best!**

Physical activity has health and cognitive benefits!





Physical activity has health and cognitive benefits!

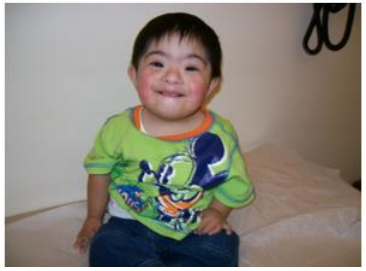


For EVERYONE!

Images from
Thatdadblog.com &
Noahsdad blog
Noahsdad.com

大綱

- 美國對於唐氏症患者治療指引
- 奧勒岡健康與科學大學唐氏症門診及其醫療照護與發展評估
- 唐氏症門診發展相關研究回顧
- 唐氏症腦部功能及治療研究回顧



سبب متلازمة داون هو تثلث الصبغي 21 *

- 95٪ من حالات متلازمة داون تحدث بسبب تثلث الصبغي 21 الكامل
- 3-2٪ انتقال موضعي أوتبادل للمادة الصبغية
- 2-1٪ حالات فسيفسائية
- الأكثر شيوعًا عدم انفصال الصبغيات الطبيعي لذ الأم في الانقسام الاختزالي
- الإصابة الإجمالية 1: 733
- يزيد الخطر بشكل كبير مع تقدم سن الأم

- * جيروم ليجون عام 1959

Fundacion John Langdon Down
Escuela de Arte Down
Mexico City http://www.fjldown.org.mx/

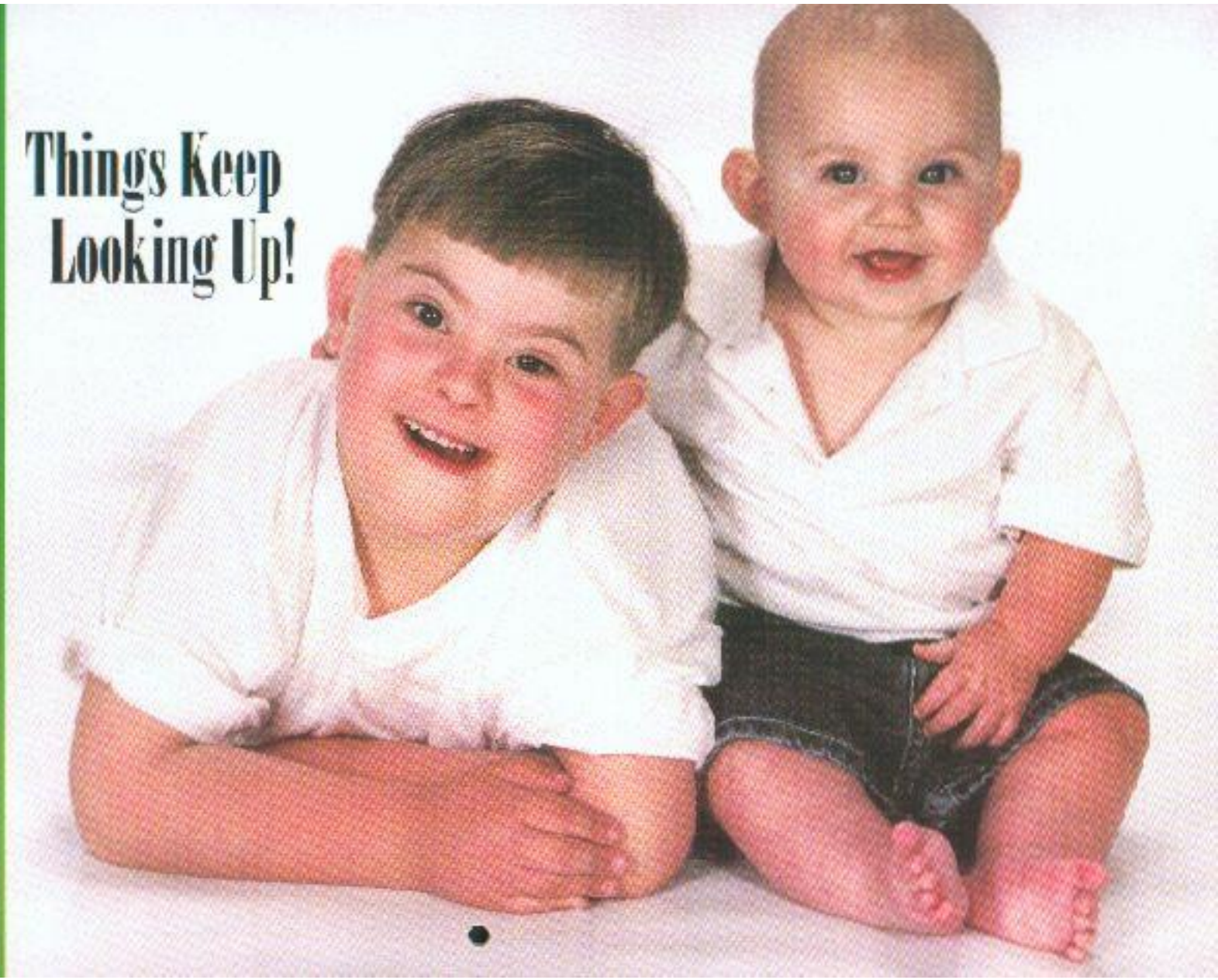
Improving Clinical Care – Simple Steps

- Use simple guidelines (www.ndss.org)
- Be positive!
- Look for and treat common conditions (Heart problems, Hypothyroidism, sleep apnea, obesity, hearing loss, hyperactivity, depression, autism)
- Get families information and involved with other families – encourage them to do everything with their kids that they would do if they did not have DS
- Get kids involved in play and activities, get them books/library cards (Dolly Parton's Imagination https://imaginationlibrary.com/
- Get kids into services/therapies (Early Intervention + school and private OT, PT, SLP) plus extras (GiGi's Playhouse | Down Syndrome Achievement Centers -- https://gigisplayhouse.org/)
- Refer to a Down syndrome clinic if one is available
- Email me for: Guidelines, Website list, PDF's of Down syndrome handouts (General Peds and Family Medicine can manage most and can become experts)
  - joseph.pinter@providence.org

2001 Calendar

Down Syndrome Association of Houston

Things Keep Looking Up!



Acknowledgements

OHSU

Melinda Pierce
Mandie Wiebers Jensen
Margaret Wolf
Alicia Feryn
Thuan Nguyen
Izzy Katlaps
Candace Ganz
Katherine Breithaupt
Erin Cochran
Amy Costanza Smith
Kerry Michaelis
Joan Medlen
Don Lollar
Christina Bethell
Randy Phelps
Liz Super
Jillian Sanford
David Oleson

Stanford

Allan Reiss
Stephan Eliez
Eric Schmitt
Vinod Menon

Johns Hopkins/ Kennedy Krieger

George Capone

UC Davis

Liz Reynolds-Losin
Susan Rivera
Frank Sharp

U of Washington

Elizabeth Aylward

****

Volunteers in my studies and their families in Seattle, Stanford, Sacramento and Portland

Dr. Pinter's research supported by:
NSADA, NIH NS 01692
Charles J. Epstein Down Syndrome Research Award (NDSS)
UCDMC M.I.N.D. Institute Intramural Research Award
OHSU/CDRC Internal Research Funding

Thank You

Providence