Down Syndrome
A Clinical Overview

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OREGON HEALTH & SCIENCE UNIVERSITY

"About Down Syndrome" from Australia translated to Arabic

https://www.downsyndrome.org.au/wp-content/uploads/2021/02/About-Down-Syndrome-_Arabic_2021.pdf

World DS Day is on 3 21

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

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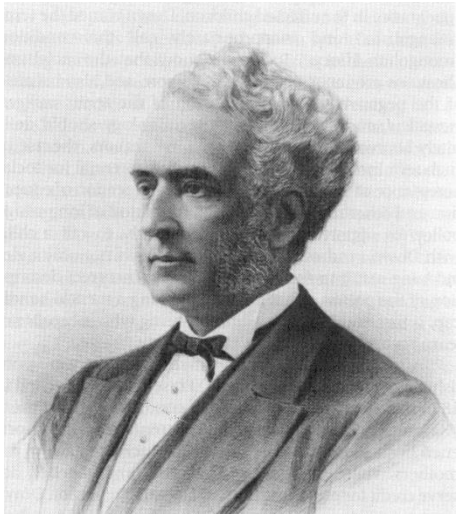
Dr. John Langdon Down



- 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 term is Intellectual 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

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

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DS is caused by trisomy 21*

- 95% of DS cases due to full trisomy 21
  - 2-3% translocation
  - 1-2% mosaicism
- Most commonly maternal non-disjunction in meiosis
- Overall incidence 1:733
- Risk increases dramatically with maternal age
- *Jerome Lejeune in 1959

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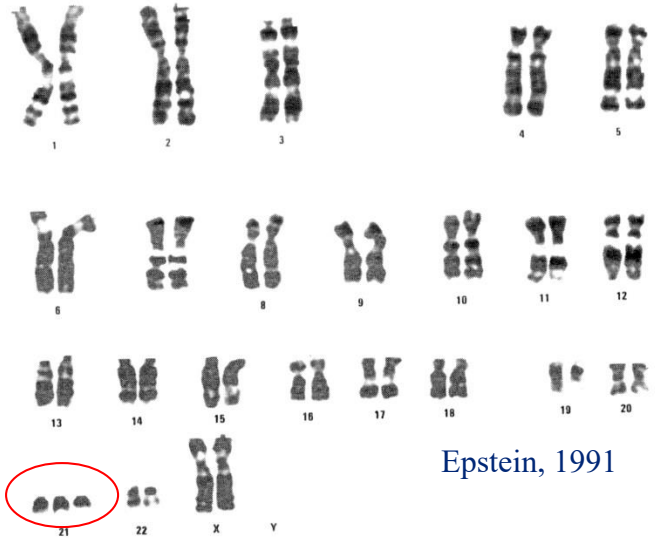
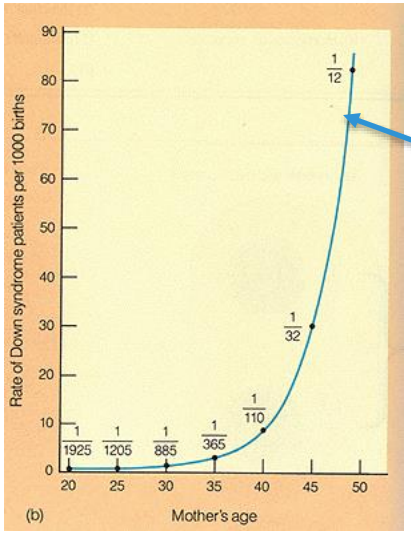
Down Syndrome: Genetics



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

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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

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%

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

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Psychiatric

- 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)

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Behavioral and Psychological Considerations for Children with DS

- Autism (1 in 59 kids (CDC, 2018) 3 x higher than 14 years ago -- 1:37 boys, 1:151 girls)
- Beyond Autism, we need to consider any and all conditions that occur among all children and determine if needs to be considered for a child with DS
  - ADHD
  - Anxiety, OCD
  - Depression
  - Pain

  People with DS can have same conditions as anyone and should be offered same treatments and therapies

Down Syndrome (DS) and Autism (ASD)

- 5 to 10% of children with DS have autism
- Autism:

  - A complex neurobehavioral condition that can include impairments in social interaction, developmental language, and communication skills combined with repetitive behaviors.

Why is Dual Diagnosis Important?

- Dual Diagnosis = DS + ASD
- Early intervention services and individualized educational plans (IEP) will be affected because children with ASD (autism) may need additional services and we know they help
- Helps us be alert about other common conditions like sleep problems, anxiety, mood disorders, behavior and to get the help needed
- Need to treat each child individually based upon his/her unique needs

Systems affected in DS

- *Cardiac (50%)*
- *GI (10%) – duodenal atresia, Hirschsprung's*
- *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

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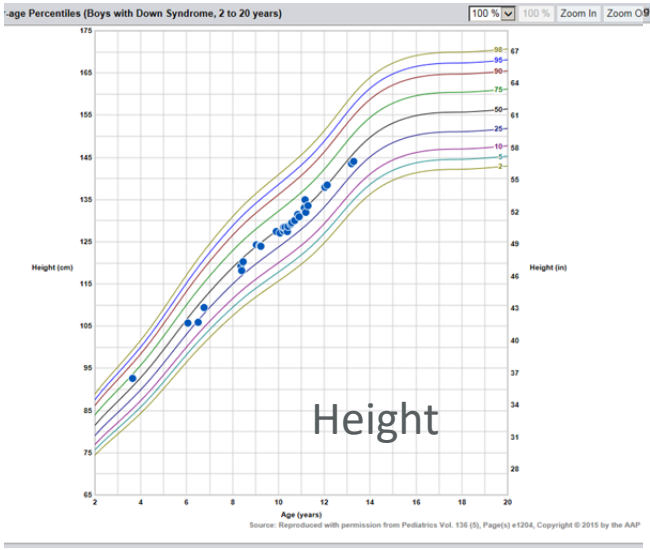
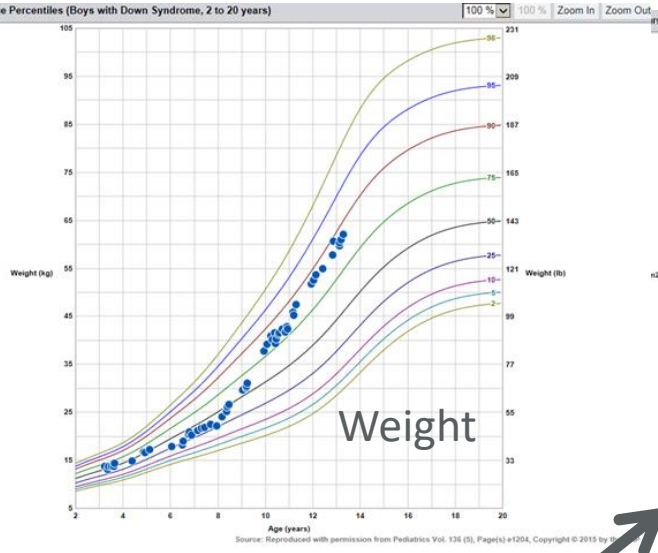
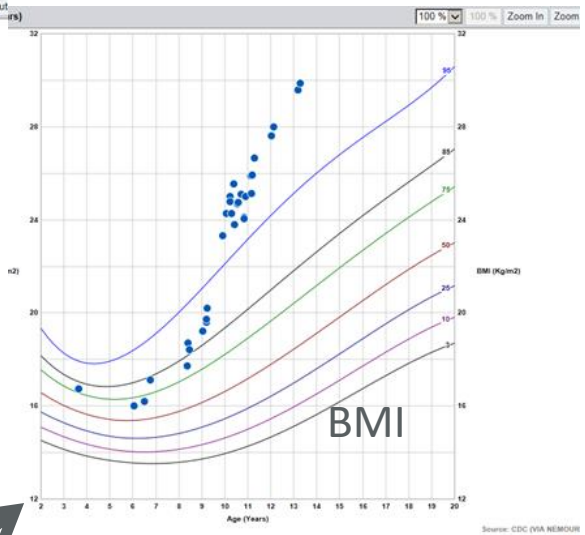
Teenagers with DS often seem "normal" for weight on DS Curves







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

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Healthcare for children with Down syndrome in the US

- How are we doing at providing healthcare for people with DS?
- Are we meeting families' needs?
- What strategies are used to assure good health maintenance?

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National Survey on Children's Health 2005-2006 (United States)

	SHCN (special healthcare needs)	**DS (Down Syndrome)**
4 or more health conditions	11%	42%
Family cut back/stopped work	23.5%	**55%**
Family provides > 11 hrs/week providing healthcare	9.5%	**30%**
Financial problems due to child's health needs	18%	**36%**
One or more unmet health service needs	16%	38%
Have family-centered care	66%	55%

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Health Care Needs of Children With Down Syndrome and Impact of Health System Performance on Children and Their Families

Randall A. Phelps, MD, PhD,* Joseph D. Pinter, MD,* Donald J. Lollar, EdD,*
Joan Guthrie Medlen, MEd, RD, Ld,† Christina D. Bethell, PhD, MPH, MBA‡

J Dev Behav Pediatr. 2012 Apr;33(3):214-20

- Families with children with Down syndrome in U.S. much more impacted financially than other health conditions – We believe that this will be true globally
- Very disruptive to families socially and financially
- We can do better to help families

Sponsored by the Maternal and Child Health Bureau (MCHB) and conducted by the National Center for Health Statistics for the CDC

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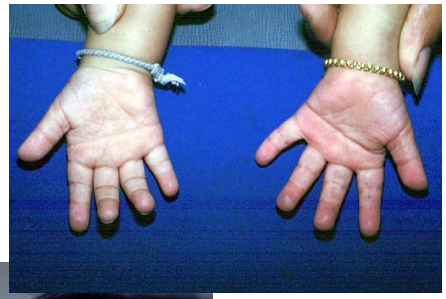
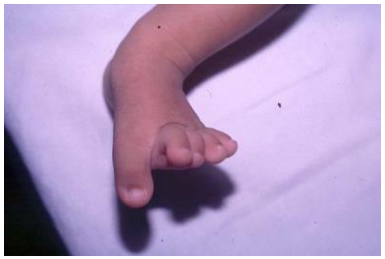
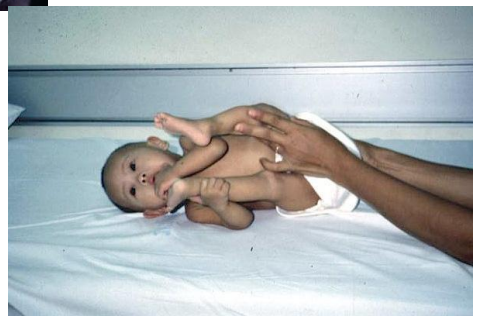
Diagnostic Physical Features in Newborns with DS

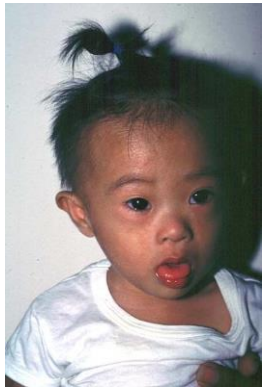
- Flat facial profile 90%
- Poor Moro reflex 85%
- Hypotonia 80%
- Hyperflexible joints 80%
- Excessive neck skin 80%
- Slanted palpebral fissures 80%
- Pelvic dysplasia 70%
- Anomalous auricles 60%
- Dysplastic midplalanx 5th digit 60%
- Single palmar crease 45%

- These findings HELP to diagnose but do not predict severity of health issues or degree of cognitive problems – appearance does NOT predict cognitive/intellectual outcomes









- Newborns with 4 or more features 100%
- Newborns with 6 or more features 90%

Photos from:
http://cai.md.chula.ac.th/lesson/down_syndrome/contents/q04a.htm

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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		■				
Hx 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 hypotonia: 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 bilateral 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						
Ophthalmology referral to assess for strabismus, cataracts, and nystagmus			■			
Refer to pediatric ophthalmologist or ophthalmologist with experience 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, discuss 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 though puberty, need for gynecologic care in the subsequent 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.

Do not translate this page – shown to demonstrate that the new guidelines are long and complicated

The reference below is a very good resource about care of children with Down Syndrome

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

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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:** _____

	Age, in years														
Medical Issues	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:** _____

	Age, in years							
Medical Issues	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.

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

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A Promising Future Together

A Guide for New and Expectant Parents

national down syndrome society
ndss

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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

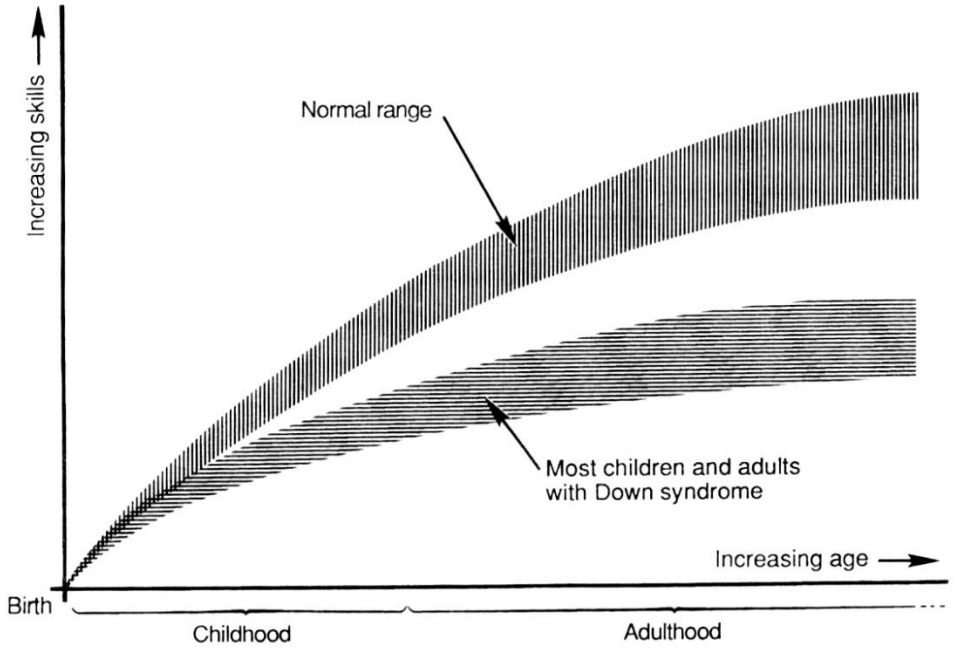
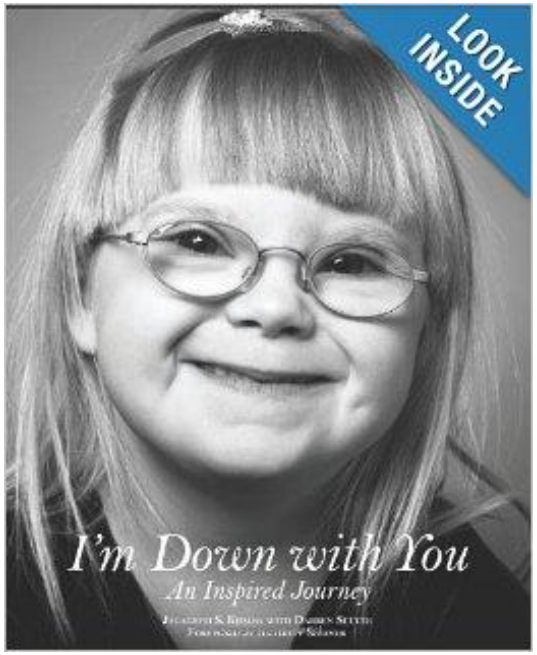


Figure 12 Rate of development.

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

What can a person with Down syndrome do?

- Some children with DS become entertainers



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Musicians with Down syndrome

Some become artists

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

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

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What can a person with Down syndrome do?

- Some grow up to be athletes (and have good jobs)

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

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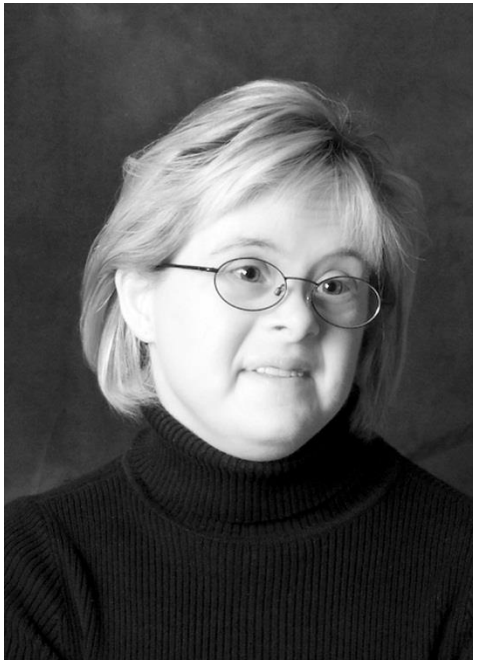
Karen Gaffney: Advocate, Motivational Speaker (TED Talk and international), Honorary Doctorate, Long-Distance Swimmer and…
OHSU Down Syndrome Clinic and CDRC team member



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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!

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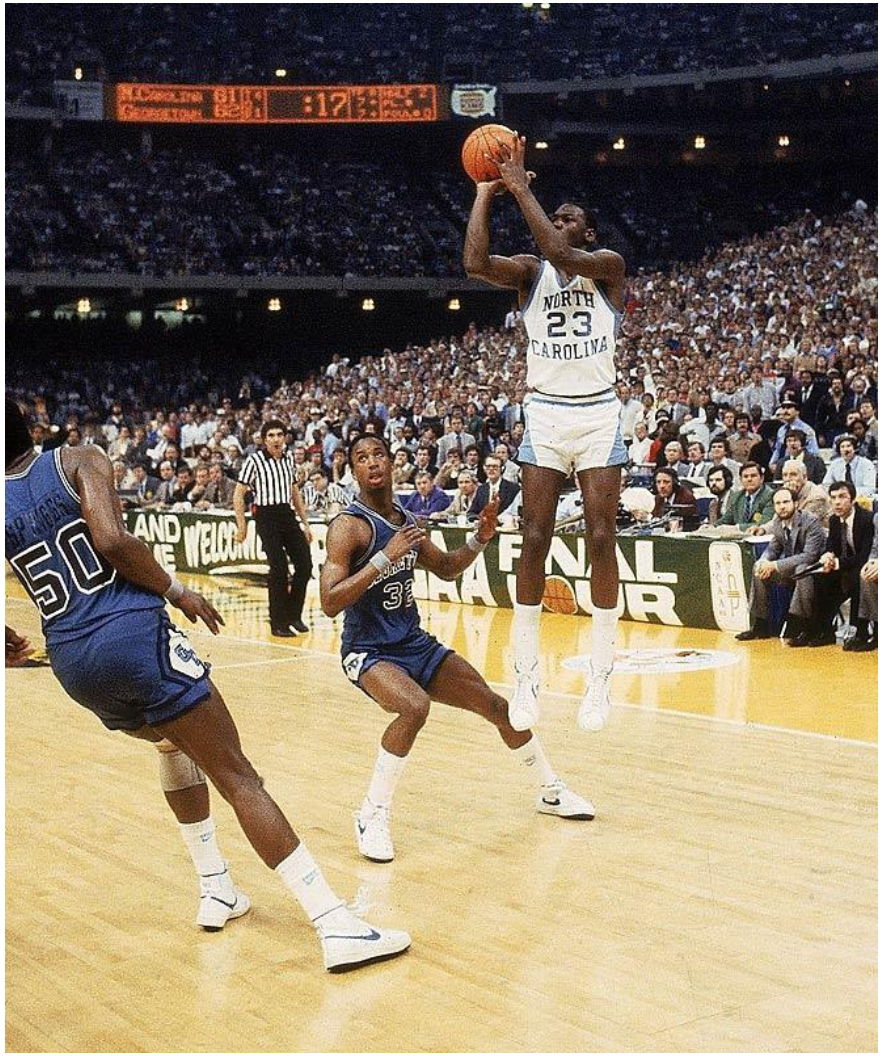
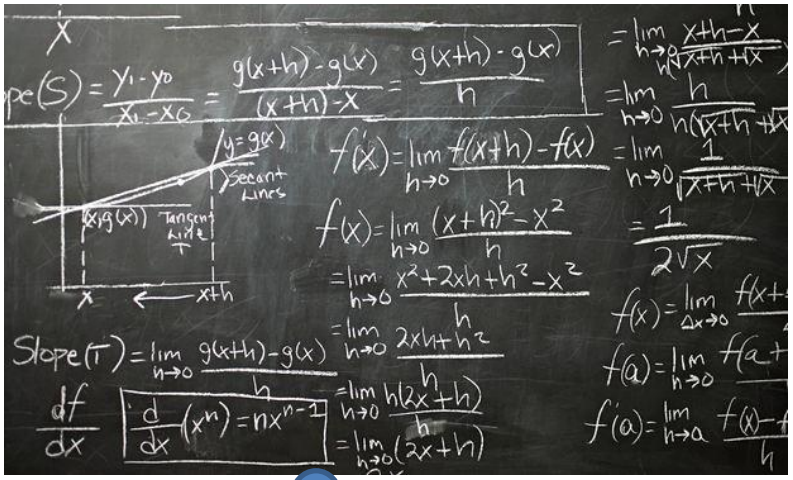
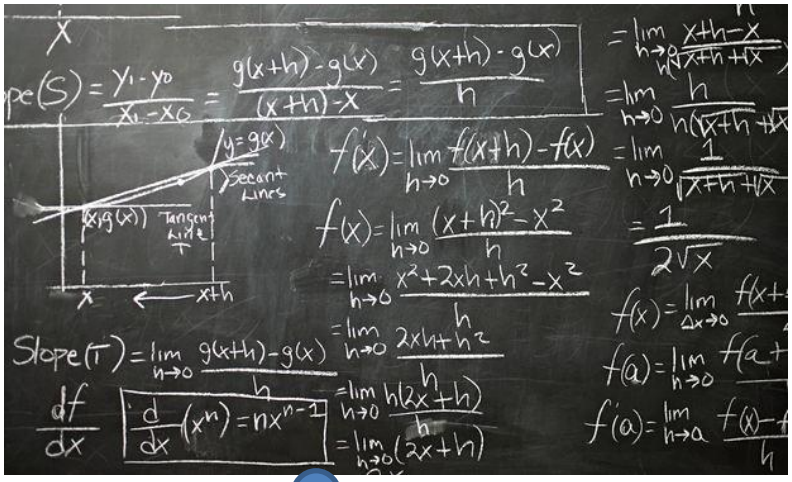
Physical Activity has Cognitive Benefit!





Physical Activity has Cognitive Benefit!

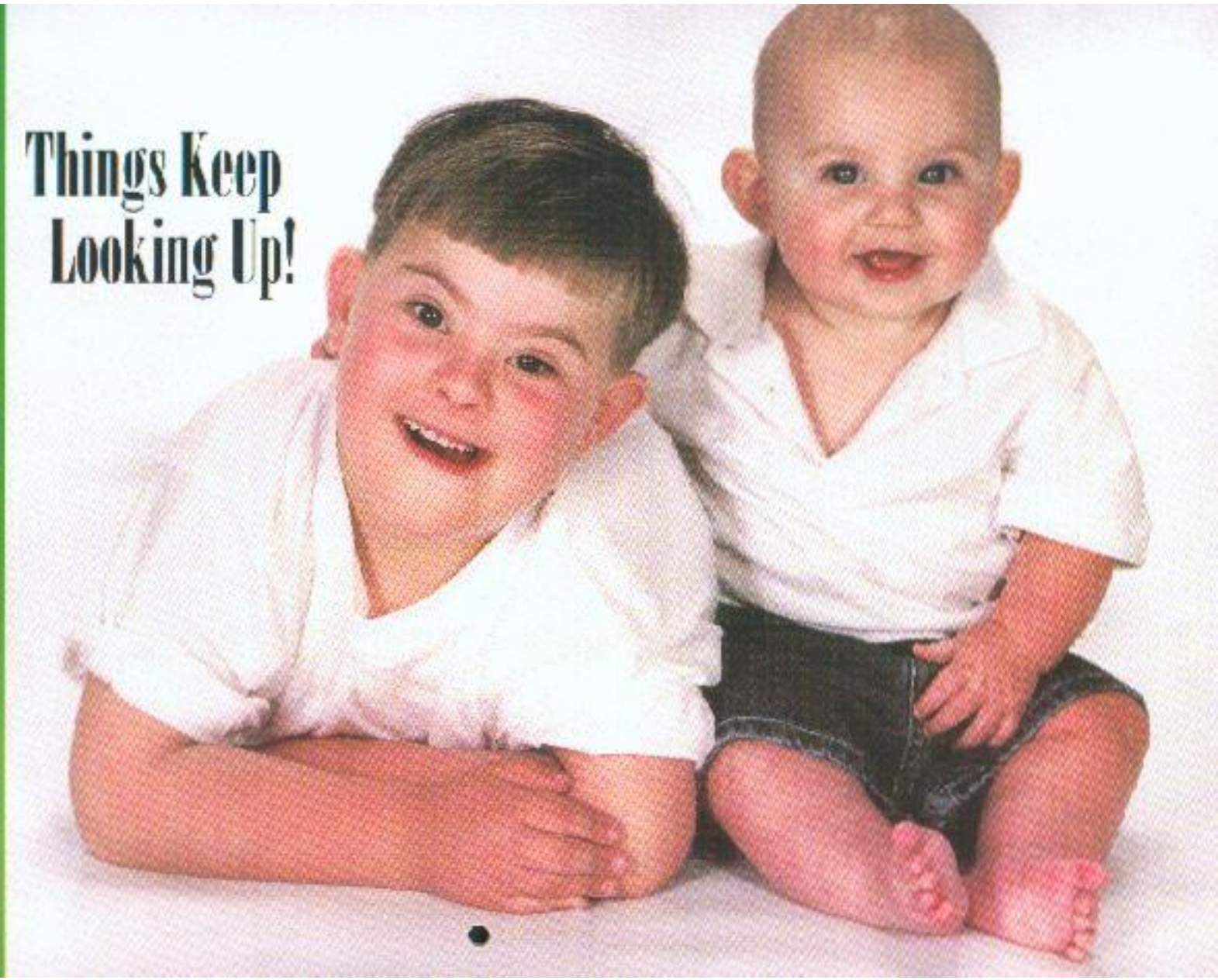
For EVERYONE!



2001 Calendar

Down Syndrome Association of Houston

Things Keep Looking Up!



Thank You