Finding New Friends

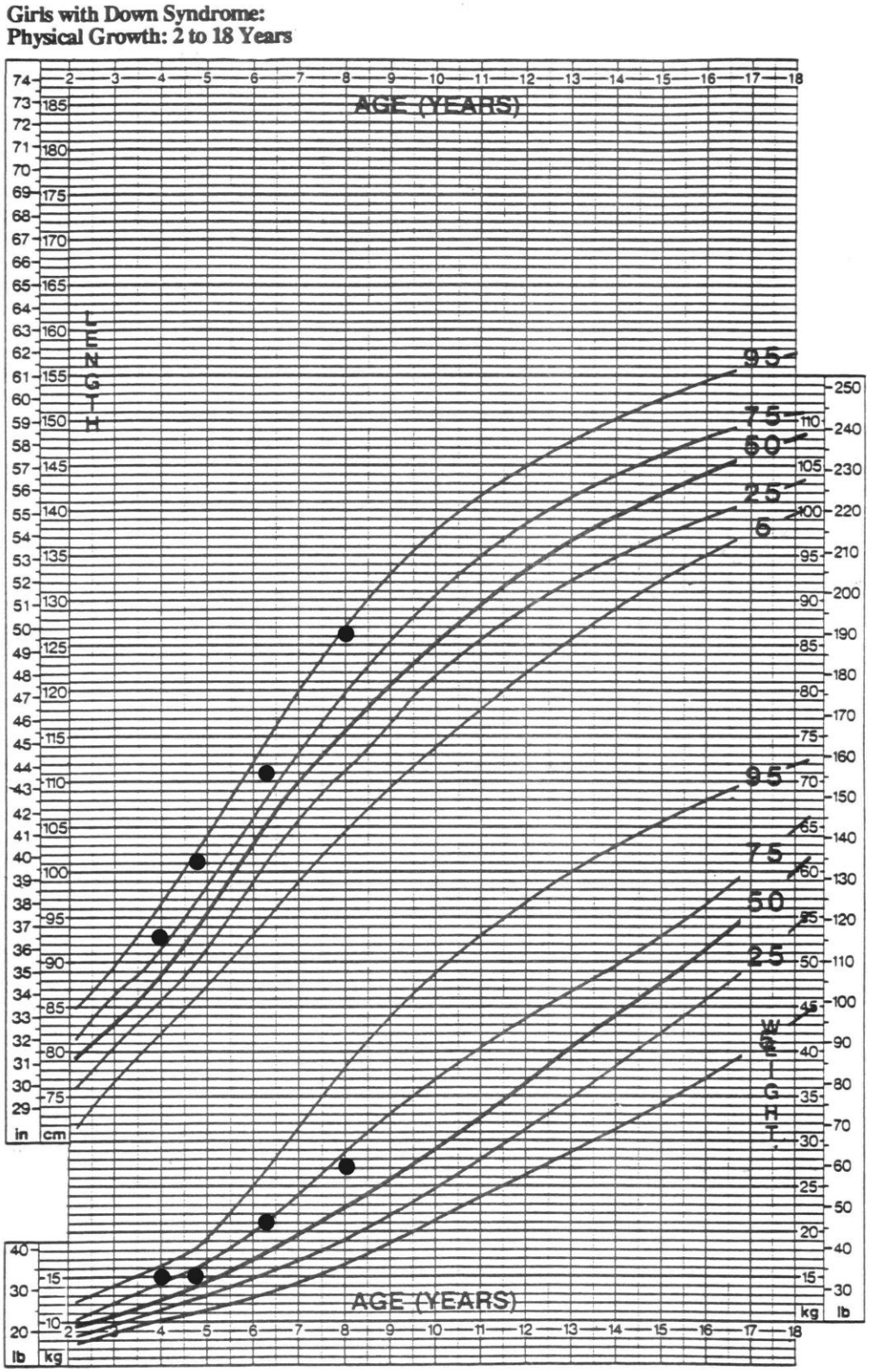
Down Syndrome

Materials for Learners

Packet should include the following:

- Handout #1: Girls with Down Syndrome: Physical Growth 2 to 18 years
- Handout #2: Healthwatch for the Person with Down Syndrome
- Handout #3: Screening Timeline in Down Syndrome
- Clinical Pearls
- Knowledge questions and answers
- References

Down Syndrome
Handout # 1: Girls with Down Syndrome: Physical Growth 2 to 18 Years



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Handout #2: Healthwatch for the Person with Down Syndrome

Concern	Clinical Expression	When Seen	Prevalence	Management
Congenital heart disease	AV canal defects, auricular or ventricular septal defects, Tetralogy of Fallot	Newborn or first year	40-50%	Cardiac consultation, echocardiography, surgical repair, SBE prophylaxis
Hypotonia	Reduced muscle tone, increased range of joints, motor function problems	Throughout life; improvement with maturity	All	Guidance by physical therapy; early intervention; adapted physical education
Delayed growth	Typically at or near third percentile for general population	Throughout	All	Use DS growth charts, early nutritional support; check thyroid, heart
Developmental delays	Some global delay, variable degrees; specific language problems	1st year; continues	All	Early intervention, educational planning, speech/language therapy
Hearing concerns	Serous otitis media, small ear canals, mostly conductive impairment	Check at birth or by 3 months; assess annually	Up to 50% at some times, ? 10% sensorineural	Audiology, tympanometry. ENT consultation
Ocular problems	Refractive errors Strabismus Cataracts	Eye exam at birth or by 6 months, then annual follow-ups	50% 35% 5%	Look for cataract; ophthalmologic consultation
Cervical spine abnormality	Atlantoaxial instability Potential neck of long-tract signs	X-ray at 3-5 years; repeat if symptomatic	10%± 1-2%+	Orthopedic referral; possible restriction, fusion
Thyroid disease	Hypothyroidism (rare hyper-), decreased growth and development	Some congenital; most 2nd+ decade; check at 0, 6 months, 12 months, then annually	15%	Endocrine consult, replacement therapy as needed
Overweight	Excessive weight gain	Preschool and adolescent years	Common	Lifestyle adjustment, including diet and activity
Seizure disorders	Primary generalized (also hypsarrhythmia)	Any time	5-10%	EEG, neurologic consultation
Emotional problems	Inappropriate behavior, depression, other emotional disturbances	Mid to late childhood, adult life	Common	Family guidance, aid in transitions, mental health assistance
Premature senescence	Behavioral changes, functional losses	5th, 6th decades	Unknown (increased rate)	Special support

Additional variable occurrences include congenital intestinal obstruction, Hirschsprung disease, leukemia, alopecia areata, keratoconus, hip dysplasia, diabetes mellitus, missing teeth, obstructive sleep apnea, celiac disease, and mitral valve prolapse.

Adapted from: Crocker, AC. *The Spectrum of Medical Care for Developmental Disabilities.* In: Rubin, IL, and Crocker, AC, editors. *Developmental Disabilities: Delivery of Medical Care for Children and Adults.* Philadelphia: Lea and Febinger; 1989. p.15-17, and AAP Committee on Genetics. Health supervision of children with Down Syndrome. *Pediatrics* 2001;107:442-449.

Finding New Friends Handout #3: Screening Timeline in Down Syndrome*

	Birth	6 mo	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18	19	20
Genetic Counseling[1], Karyotype																						
Parent Group Info and Support		Parent-to-parent contact, support groups, current books and pamphlets																				
CBC to R/O transient myeloproliferative disorder, polycythemia																						
Swallowing assessment if feeding problems or aspiration																						
Hemoglobin			Hemoglobin annually beginning at 1 year old. If Hg<11, do (a) CRP and ferritin, or (b) Reticulocyte Hemoglobin Content (CHr). If possible risk for iron deficiency, do (a) or (b) regardless.																			
23-valent pneumococcal vaccine[2]																						
Cardiology	Echo[4]														Screen for acquired mitral or aortic valvular disease							
Audiological Evaluation	ABR or OAE	Every 6 months till 3 years of age. Annually thereafter.																				
Ophthalmologic Evaluation	Red Reflex	Optho appt	Annual ophthalmology appt					Q2 Ophthalmology appointment								Q3 Ophthalmology appointment						
Celiac Disease Screening					(Only test if signs and symptoms present)																	
Thyroid – TSH, T4	State Screen	Test	Test	test TSH and T4 annually																		
Neck X-ray (AAI)[3]					✓[3]																	
Dental Exam			Annual Dental Exams. Reassure parents that delayed or irregular eruption, hypodontia are common.																			
Sleep Study by age 4 years	Done prior to 4 years of age																					
Early Intervention																						
Childhood					Discuss self-help, ADHD, OCD, wandering off, transition to middle school																	
Puberty															Discuss physical and psychosocial changes through puberty, need for gynecologic care (pelvic exams) in pubescent female							
Facilitate transition															Guardianship, financial planning, behavioral problems, school placement, vocational training, independence with hygiene and self-care, group home, work settings							
Sexual development and behaviors															Discuss Contraception, STDs, recurrence risk for offspring							
Preventive care	Annually monitor for signs and symptoms of constipation, OSA, and aspiration.																					

1. Discuss Recurrence Rate of future pregnancies with parents
2. 23-valent pneumococcal vaccine if chronic or pulmonary disease
3. AAI: See AAP Guidelines page 399 - X-rays only if myopathic signs or symptoms
4. Follow-up to be determined by cardiologist

* Peds 2011;128 :393-406 Chart by Sie Center for Down Syndrome

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Clinical Pearls:

- Although the risk of having a child with Down syndrome increases significantly in women 35 years of age and older, most children with Down syndrome are born to younger mothers.
- Most children with Down syndrome can be integrated within a regular classroom setting, with appropriate educational and therapeutic supports.
- Although social skills are relatively strong in children with Down syndrome, a subset of children with Down syndrome meet behavioral criteria for the diagnosis of an autism spectrum disorder (ASD). It is important to identify those children, as the diagnosis of an ASD will affect the types of educational curricula and therapies provided.
- Medical providers should follow Down syndrome-specific health maintenance guidelines and screenings, which vary with age of the child.
- As with all individuals exercise is of clear benefit to the individual with Down syndrome, both in terms of cardiovascular and neuromuscular responses. Also, participation in the community activities is vital to their progress.

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Down Syndrome

Knowledge Questions:

1. *Select the best statement pertaining to a newborn with Down syndrome.*

a. Infants with Down syndrome often are able to breastfeed.
b. Medical complications, such as cardiac co-morbidity, may affect an infant's early ability to feed.
c. A radiographic swallow study is recommended for infants with feeding difficulty, aspiration pneumonia, or marked hypotonia.
d. All infants should have an echocardiogram.
e. All of the above

2. *Why are routine cervical X-rays in asymptomatic children with Down syndrome no longer recommended?*

a. Radiation exposure.
b. Plain radiographs predict which children are at increased risk of developing spine problems.
c. Risk of injury from the previous 3 view X-rays caused their discontinuation.
d. Current evidence does not support routine X-rays for atlanto-axial instability in asymptomatic children
e. CT is the preferred imaging method.

3. *Positive celiac screening tests occur in 5-7 % of individuals with Down syndrome. After the age of 2 ½ years, which of the following symptoms* ***is not*** *suggestive of celiac disease?*

a. Protracted constipation
b. Slow growth or failure to thrive
c. Reactive airway disease
d. Abdominal bloating or pain
e. Anemia

4. *Which one of the following is NOT recommended as part of annual routine screening for children between 1 to 5 years with Down syndrome?*

a. TSH
b. Audiology evaluation
c. Ophthalmology exam
d. EKG
e. Hemoglobin

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Down Syndrome

Answers to knowledge questions:

1. *Select the best statement pertaining to a newborn with Down syndrome*
   Preferred response: e "All of the above"
   Many infants with Down syndrome are able to breast feed and parents should be supported in their choice to breast feed. However, congenital cardiac lesions, low muscle tone and swallowing dysfunction can impact feeding. All children with Down syndrome should be screened for congenital cardiac conditions and should have an echocardiogram.

   Babies with Down syndrome are often sleepy and placid, so you may need to interest your baby through frequent breast feedings throughout the day, wake him or her fully before breastfeeding, or provide extra touch and stimulation to keep him alert. Signs of feeding dysfunction such as choking, coughing or cyanosis with feeds require further medical assessment such as a swallow study to identify any structural problems and provide guidance to parents on safe feeding practices. This information is provided in the AAP clinical report, *Health Supervision for Children with Down Syndrome*, as noted in the reference list.

2. *Why are routine cervical X-rays in asymptomatic children with Down syndrome no longer recommended?*
   Preferred response: d "Current evidence does not support routine X-rays"
   There is no evidence to support problems with radiation exposure from cervical spine radiographs, or benefits of X-rays in asymptomatic children. For symptomatic children, a cervical X-ray in the neutral position is done first. If there are positive findings, the child is quickly referred to a pediatric neurosurgeon or pediatric orthopedic surgeon. If the X-ray is negative, then X –rays in flexion and extension can be obtained before a prompt referral for further evaluation. CT is not required or preferred method of imaging.

3. *Positive celiac screening tests occur in 5-7 % of individuals with Down syndrome. After the age of 2 ½ years, which of the following symptoms* ***is not*** *suggestive of celiac disease?*
   Preferred response: c "Reactive airway disease"
   Celiac disease is commonly associated with gastrointestinal symptoms. Untreated celiac disease affects the lining of the small intestine, with decreased absorption of iron. There are no associated pulmonary symptoms with celiac disease.

4. *Which one of the following is NOT recommended as part of annual routine screening for children between 1 to 5 years with Down syndrome?*
   Preferred response: d "EKG"
   It is important that infants undergo a cardiac evaluation. However, there is no need to obtain routine EKG tests on people with Down syndrome in the absence of symptoms or concerns for a cardiac problem. Given the relatively high incidence of thyroid, hearing, and eye problems, there should be annual screening for problems in these areas. Children with Down syndrome have been found to have lower dietary intake of iron therefore routine screening is recommended.

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References

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2. Bull, MJ and the Committee on Genetics. Health supervision for children with Down syndrome. Pediatrics 2011;128(2):393-406.
3. Carroll KN, Arbogast PG, Dudley JA, et al. Increase in incidence of medically treated thyroid disease in children with Down syndrome after rerelease of American Academy of Pediatrics Health Supervision guidelines. Pediatrics 2008; 122(2):e493-498.
4. Castillo h, Patterson B, Hickey F, Kinsman A, Howard JM, Mitchell T, Molloy CA. Difference in age at regression in children with autism with and without Down syndrome. J Dev Behav Pediatr 2008;29(2):89-93.
5. Cohen, WI. Down syndrome: Care of the child and family, Developmental and Behavioral Pediatricss, 4th edition, (eds).Carey, WB, Crocker, AC, Elias, ER, Feldman, HM, Coleman, WL, Philadelphia: Saunders Eselvier, 2009.
6. Fitzgerald, DA, Paul, A, Richmond, C. Severity of obstructive sleep apnea in children with Down syndrome who snore. Archives of Diseases in Childhood 2007;92:423 – 436.
7. Hankinson T, Anderson R. Craniovertebral junction abnormalities in Down syndrome. Neurosurgery 2010;66(3):A32 – 38.
8. McCabe, LL and McCabe ERB. Call for change in prenatal counseling for Down syndrome. American Journal of Medical Genetics 2012;158A:482 – 484.
9. Murphy, NA, Carbone, PS, and Council on Committee on Children with Disabilities. Pediatrics 2008;121(5);1057-1061.
10. Reilly, C. Autism spectrum disorders in Down syndrome. Research in Autism Spectrum Disorders 2009;3:829- 839.
11. Skotko, BG, Capone, GT, Kishani, PS, Down Syndrome Diagnosis Study Group. Postnatal diagnosis of Down syndrome: Synthesis of the evidence on how best to deliver the news. Pediatrics 2009;124(4);e751. Available at: www.pediatrics.org/cgi/full/124/4/e751.

Suggested Readings (Annotated):

1. Bull, MJ and the Committee on Genetics. Health supervision for children with Down syndrome. Pediatrics 2011; 128(2):393-406.
   Essential reading of new guidelines in the care of children with Down syndrome.
2. Hankinson T, Anderson R. Craniovertebral junction abnormalities in Down syndrome. Neurosurgery 2010; 66(3):A32-8.
   Good discussion of atlanto-axial instability.
3. Cohen, WI. Down syndrome: Care of the child and family, Developmental and Behavioral Pediatrics, 4th edition, eds. Carey, WB, Crocker, AC, Elias, ER, Feldman, HM, Coleman, WL, Philadelphia: Saunders Eselvier, 2009.

Comprehensive and up-to-date summary by an outstanding clinician with experience caring for children with Down syndrome.

4. Roizen, NJ. Down syndrome, in Children with Disabilities, sixth edition, eds. Batshaw, M, Pelligrino, L, Roizen, NL, Baltimore: Paul Brookes, 2007. Useful diagrams of phenotypical features seen in children with Down syndrome, and recommendations for preventative care.

National Organizations

- National Down Syndrome Congress http://www.ndsccenter.org/
- National Down Syndrome Society http://www.ndss.org/

Education Support

- Council for Exceptional Children
- Wrightslaw Special Education Law and Advocacy
- Down Syndrome Education International

Web-based Parent Support

- Brighter Tomorrows
- Parent 2 Parent
- Down Syndrome Pregnancy
- Babycenter Down Syndrome Community