Facilitator Preparation: Facilitators should thoroughly review this module. They should also prepare or photocopy handouts to distribute during the course of the case presentation and the "Materials for Learners" packet.

Open the Discussion: Introduce the case title and the objectives of the session. Explain that this will be an interactive case discussion prompted by a series of questions and not a lecture. Distribute Part I of the case and ask one or more of the participants to read it aloud.

Finding New Friends

Down Syndrome

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Dedicated to, and in Memory of:
Allen C. Crocker, MD

Objectives:

- List the diagnostic features of Down syndrome and its associated complications.
- Describe the role of the clinician in management of Down syndrome, including: monitoring of growth/nutrition, recognition and prevention of medical complications, and participation in educational planning.
- Describe the psychosocial stresses faced by children with Down syndrome and their parents.

Part I

Introduction:

Jamie Peters is an 8-year-old girl who comes to your clinic with a complaint of headaches and neck pain. She was born at full term to a 28-year-old Gravida 2/Para 1 mother. Pregnancy was unremarkable, and birth was by normal spontaneous vaginal delivery. Apgar scores were 8 and 9. A pediatrician was called to the delivery room because the nurses noticed that Jamie had distinctive palpebral fissures, flat occiput, and transverse palmar crease on her left hand. Subsequent chromosomal analysis indicated that Jamie had trisomy 21 (Down syndrome).

Her clinic record reveals that she had a small muscular ventricular septal defect that closed by the age of 18 months. She underwent surgical correction of duodenal atresia at age 3 days, and had bilateral tympanostomy tube placement for recurrent otitis media at age 3 years. Aside from intermittent bouts of constipation, she has been well since that time.

Developmentally, she spoke her first words at 17 months, walked at age 22 months, and completed toilet training at 4 1/2 years of age. She is currently in the third grade, in a mainstream classroom, and her mother reports she is able to "read at grade level." A full-time teacher's aide is assigned to Jamie and one other child. Jamie also receives occupational therapy once weekly and speech therapy twice weekly at school. Last year, she was quite happy and well behaved in the classroom. This year, however, she has demonstrated occasional school avoidance and noncompliant behavior. She becomes somewhat oppositional when asked to transition from a favorite activity to one that is less enjoyable. For example, when recess is over she will sit on the ground and refuse to go back to class.

She began to complain of headaches and neck pain three weeks ago. Her mother tells you, "*I think Jamie is complaining about these things so she won't have to go to school*."

Following this reading, ask all participants "*So what do you think about this case? What would you like to focus on during our discussion today*?" List agenda items on a blackboard or flipchart. Then use the questions below to guide the discussion. Remember that the key to successfully leading a small group is facilitation of the discussion rather than lecture. Draw as many participants as possible into the discussion. Allow silences while group members think about questions. Present material from the discussion guide only when needed to complement or redirect the group discussion.

Potential Questions for Discussion:

What are the diagnostic features of Down syndrome?
Down syndrome is the most common chromosomal abnormality affecting children. The syndrome involves multiple anomalies resulting from either complete or partial trisomy of chromosome 21, and is reported to have an incidence of 1/691 live births. Prenatal screening is offered to all expectant women. The majority of children with Down syndrome are born to women *under* age 35

Clinical findings associated with Down syndrome include: hypotonia, almond shaped eyes with epicanthal folds, upslanted palpebral fissures, Brushfield spots (speckling on the iris), small nose with low nasal bridge, flat facial profile, brachycephaly with flat occiput, short neck, excess skin on the posterior neck, small ears, hyper-flexibility of the joints, wide space between first and second toe, single transverse palmar crease, dysplasia of middle phalanx which may result in incurving of the fifth finger (clinodactyly), small stature, and others.

The diagnosis is confirmed by chromosome analysis - with trisomy of all or part of chromosome 21. To expedite the diagnosis, fluorescent in-situ hybridization (FISH) study is often performed. The most common chromosomal finding is full trisomy 21 (95%), with translocation of 21 (3 to 4%) and trisomy 21/normal mosaicism (1 to 2%) accounting for the rest. Because FISH testing cannot distinguish among these possibilities

a full karyotype must also be performed. Parents of children who have a translocation should have genetic counseling and a chromosome analysis themselves.

What are the associated developmental and medical problems seen in children with Down syndrome?
Developmentally, children with Down syndrome usually have mild to moderate intellectual disability, with a small percentage of children with severe intellectual disability. While cognitive impairment is nearly universal in children with Down syndrome, all areas of development are not affected to the same degree. Social development and visual processing skills tend to be less affected, while they experience considerable difficulty in the area of expressive language.

These children are also prone to a number of medical problems. Associated cardiac problems (40-50%) include atrial-ventricular canal defects, ventricular septal defects, atrial septal defects, Tetralogy of Fallot, and patent ductus arteriosis. Associated gastrointestinal malformations (12%) include Hirschsprung Disease, imperforate anus, duodenal atresia, and others. Constipation occurs in over 50% of children with Down syndrome. Celiac disease occurs in 5% of individuals. During the newborn period, children with Down syndrome are more likely to be jaundiced. Associated orthopedic problems include atlantoaxial instability (potentially life-threatening) and late hip dislocations. Ophthalmologic problems are quite common, and include refractive errors, myopia, congenital cataracts, nystagmus, and strabismus. Children with Down syndrome have a relatively high incidence of recurrent otitis media, chronic sinusitis, and 70-80% may experience hearing loss - mostly conductive in nature. Obstructive sleep apnea occurs in 50-75%. The increased risk of primary hypothyroidism occurs in 10-20 %. Leukemias occur at a rate of approximately 1 % and transient myeloproliferative disorder occurs in 4 to 10% of newborns. More recently autism has been found to occur in approximately 5% of children with Down syndrome, and must be considered in a child with impairments in social skills, communication, and the presence of repetitive behaviors or restricted interests.

What further questions would you like to ask Jamie, her parents and teachers?

- Questions for Jamie: How is school going? What subjects do you like/not like? What things are hard for you? Do you have friends at school? Tell me what your teachers are like. Tell me a little more about this pain you have in your neck.
- Questions for her parents: How do you think Jamie is doing in school? Does her schoolwork seem too hard for her? Does she complain (of neck pain) before going to school? Does she ever complain of neck pain after school or on weekends? Has Jamie had any falls or injuries? Has Jamie ever suddenly fallen down without apparent cause (i.e. drop attack)? Does she do strenuous exercise? Does Jamie see friends outside of school? How many parties has she been invited to over the past year?
- Questions for her teachers: How is Jamie doing academically, socially, and behaviorally? Does she need more classroom support than she is receiving? Does she complain of pains or ailments while at school? What happens when she complains?

Distribute Part II of the case narrative and have the participant(s) read it aloud. During the reading, distribute Handout #1: Girls with Down Syndrome: Physical Growth 2 to 18 Years, and allow a minute for participants to review it.

Part II:

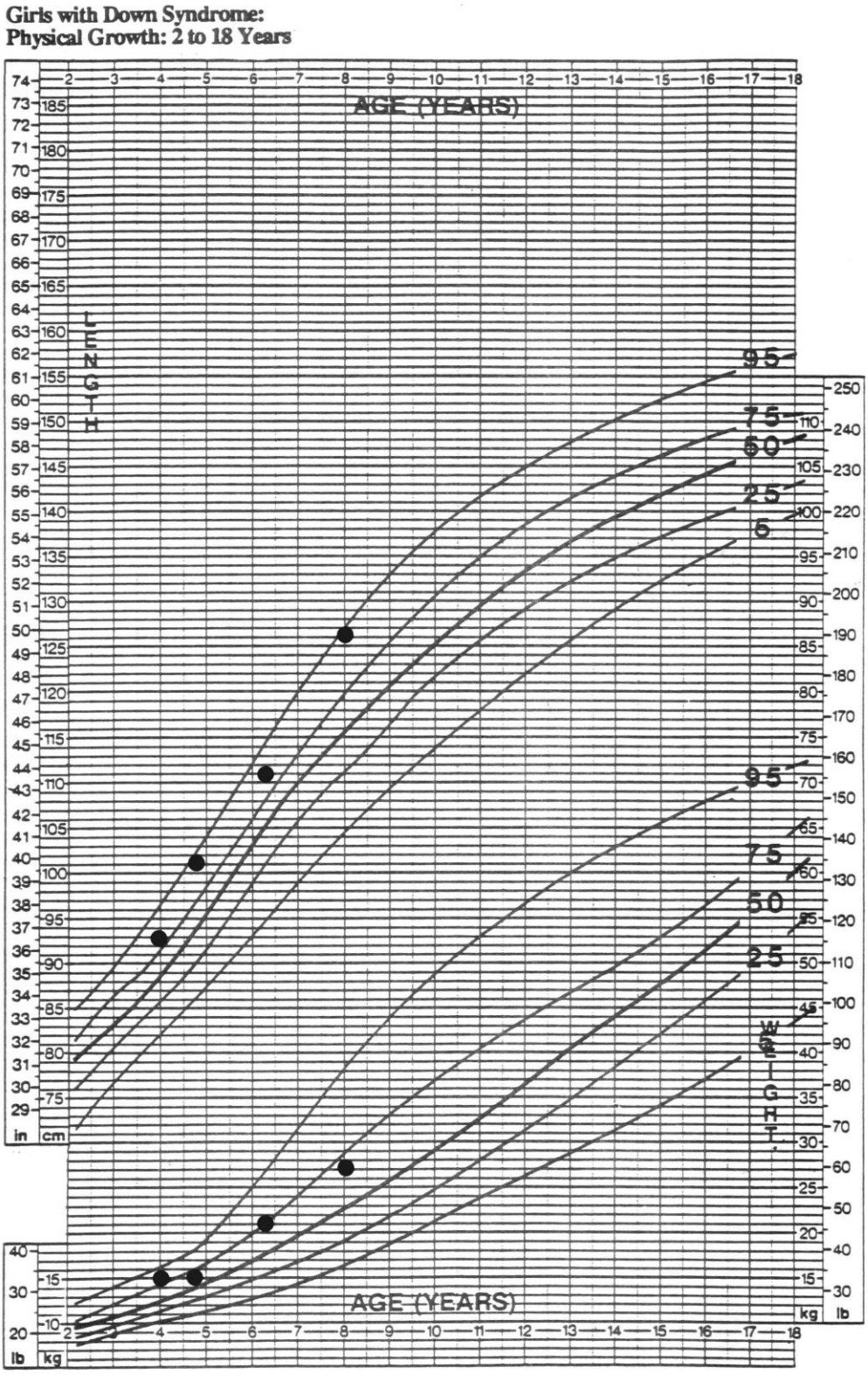
Next Steps:

Jamie points to her head and over the right side of her neck when asked about the location of the pain. You ask, "*Is the pain there all the time*?"
"*No*."
"*Does the pain go anywhere else on your body? Do your legs, arms or fingertips ever feel funny or tingly*?"
"*No*."
"*What time of the day is the pain the worst*?"
"*After lunch*" she replies.
"*Is there anything that makes the pain worse*?"
"*I don't know.*"
When asked about school, she responds that she likes her teachers and the classroom activities.
"*Do you have friends in school*?"
Jamie says, "*Yes*."
"*Can you tell me the name of one of your friends*?"
She is silent.
You turn to Jamie's mother and ask, "*Does she have any play dates outside of school*?"
Mom replies, "*She occasionally invites a younger girl from the neighborhood to come and play with her.*

Jamie's mother later tells you in private that while the younger neighbor occasionally does come over to play, she never invites Jamie to come to her house. She also lets you know that her schoolmates have never invited Jamie to any birthday parties or out-of-school activities.

On physical exam Jamie is a warm and friendly 8-year-old with the typical facial features of Down syndrome. Her height is 126 cm. and her weight is 27 kg. Her body mass index is 17.0 (71 % on the NCHS growth chart). Her skin is dry, and she has mild atopic dermatitis. She wears glasses and has epicanthal folds and Brushfield spots on the outer third of the iris. She has an open bite with a tongue thrust. Her tympanic membranes are dull but not injected, and the motility is mildly reduced. Her thyroid gland is non-palpable. Heart and lung sounds are normal. Abdominal exam reveals a midline surgical scar, and a moderate amount of palpable stool in the LLQ. Her breast and pubic hair are Tanner Stage I. You note a bilateral hallux valgus deformity. Her overall muscle tone is diminished with hyperextensibility of joints, and she has decreased truncal stability and rounding of the shoulders. There are no lateralizing neurological signs. Her gait is slightly immature although symmetric. .

Girls with Down Syndrome: Physical Growth 2 to 18 Years



Pause and begin next set of discussion questions.

Potential discussion questions:

What other evaluations are indicated?

- Thyroid function tests, which should be obtained annually, can be obtained to further assess the possible causes of her constipation, although her normal linear growth is quite reassuring.
- To evaluate the neck pain, an examination and radiograph of the lateral cervical spine, in the neutral position; if this is normal, lateral flexion and extension films can be obtained as well. If the neutral film is abnormal, urgent referral to orthopedics or neurosurgery is indicated. Otherwise, she also should be referred to neurosurgery or orthopedics.
- Jamie should have audiometry and tympanometry, also recommended yearly, to exclude further hearing loss as a cause of diminished classroom function. In addition to conductive hearing loss children with Down syndrome are at increased risk for developing sensorineural hearing loss

Distribute results and have a participant read aloud.

Results:

Lateral neck x-ray shows no evidence of occipito-atlanto or atlanto-axial instability. Her thyroid function tests are all normal. Audiometry reveals that her left ear is normal, and her right ear has mild low frequency conductive hearing loss. Her tympanogram is normal for the left, and shows negative middle ear pressure on the right.

*Describe a management plan for Jamie including medical, educational, and psychological interventions and support**?***

- Medical: Jamie's x-rays indicate no need for restriction of most physical activity. Orthopedic surgeons, however, recommend children with Down syndrome avoid somersaults, vigorous gymnastics, use of trampolines, and other sports that may cause neck strain or injury. Since thyroid function tests are normal, constipation can be managed with dietary interventions such as fiber, fluids, fruits and vegetables. Her middle ear effusion should be re-evaluated by a consulting otorhinolaryngologist.
- Educational: Jamie should continue receiving help from the teacher's aide within the classroom. A daily program including a high degree of structure and assistance with transitions between activities is needed. Although her parents report she is "*reading at grade level*," the recent problems with behavior may indicate the need for an educational re-evaluation and subsequent review of her Individualized Educational Program (IEP). Jamie may be able to *decode* words at an age appropriate level; however, she may not fully *comprehend* the meaning of what she has read.
- Psychological: Jamie may benefit from a behavioral medicine consultation to assist with management of oppositional behaviors at school and at home. In addition, Jamie would benefit from social skills training, group activities outside of school (Brownies, 4-H Club, church group, Special Olympics, etc.), and contact with another family in the area that has a child with Down syndrome who

is close to Jamie's age. The aim is to promote peer interaction in an adult-supervised setting, to foster a peer friendship, and thus enhance feelings of self-esteem.

What are the relative advantages and disadvantages of inclusion model of educational programs as opposed to separate programs?
Inclusion programs are considered to provide positive experiences for children with disabilities and their classmates. Many children with Down syndrome benefit from peer modeling. Typical peers may have better understanding of and acceptance of diversity. However, these programs need to be able to adequately meet the needs of all students, as well as have the support of the special educators, general teachers, and school administrators.

Mainstream educational programs may be too difficult or too confusing for certain children with complex developmental and behavioral needs. These children may be too distracted by the large class size, and may also be disruptive to other children. Some teachers may be unfamiliar or uncomfortable with teaching children who have significant disabilities. However, this tends to be the exception. For the vast majority of children with developmental disabilities, mainstream education offers clear advantages and few disadvantages.

Distribute Part III and ask a participant to read Epilogue aloud.

Part III
Epilogue

Following her benign medical work-up, you reassured Jamie and her mother that there was no serious medical problem underlying her neck pain. Following a consultation with a Behavioral Medicine specialist, Jamie's parents instituted a behavior management plan that included positive reinforcement (e.g., a sticker chart with subsequent reward) for staying in class and following teacher directions. Jamie continued to complain of pain in her head and neck. Her parents were initially reluctant to involve her in activities with other children who have developmental disabilities, as they feared that Jamie could learn additional negative behaviors. They ultimately decided to try *Special Olympics*, which has on-going activities during the entire year. Jamie found acceptance there and had a wonderful time. She subsequently joined a dance class with typically developing peers at the local community center. Her complaints of head and neck pain have progressively diminished. She is not always compliant in school and the behavioral management plan needs to be reactivated from time to time.

Refer back to group's learning agenda and summarize the key teaching points that were made. This will give the group a sense of accomplishment, and emphasize the important messages. Suggest further sources of reading or other information if there are agenda items that were not covered in the discussion.

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

Knowledge Questions:

Ask learners to complete the knowledge questions in their packet. If time allows, questions and answers can be discussed as a group, or learners can complete and review answers on their own.

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

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.

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.
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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

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

Finding New Friends

Down Syndrome

Part I

Introduction:

Jamie Peters is an 8-year-old girl who comes to your clinic with a complaint of headaches and neck pain. She was born at full term to a 28-year-old Gravida 2/Para 1 mother. Pregnancy was unremarkable, and birth was by normal spontaneous vaginal delivery. Apgar scores were 8 and 9. A pediatrician was called to the delivery room because the nurses noticed that Jamie had distinctive palpebral fissures, flat occiput, and transverse palmar crease on her left hand. Subsequent chromosomal analysis indicated that Jamie had trisomy 21 (Down syndrome).

Her clinic record reveals that she had a small muscular ventricular septal defect that closed by the age of 18 months. She underwent surgical correction of duodenal atresia at age 3 days, and had bilateral tympanostomy tube placement for recurrent otitis media at age 3 years. Aside from intermittent bouts of constipation, she has been well since that time.

Developmentally, she spoke her first words at 17 months, walked at age 22 months, and completed toilet training at 4 1/2 years of age. She is currently in the third grade, in a mainstream classroom, and her mother reports she is able to "read at grade level." A full-time teacher's aide is assigned to Jamie and one other child. Jamie also receives occupational therapy once weekly and speech therapy twice weekly at school. Last year, she was quite happy and well behaved in the classroom. This year, however, she has demonstrated occasional school avoidance and noncompliant behavior. She becomes somewhat oppositional when asked to transition from a favorite activity to one that is less enjoyable. For example, when recess is over she will sit on the ground and refuse to go back to class.

She began to complain of headaches and neck pain three weeks ago. Her mother tells you, "*I think Jamie is complaining about these things so she won't have to go to school*."

Finding New Friends

Down Syndrome

Part II:

Next Steps:

Jamie points to her head and over the right side of her neck when asked about the location of the pain. You ask, "*Is the pain there all the time*?"
"*No*."
"*Does the pain go anywhere else on your body? Do your legs, arms or fingertips ever feel funny or tingly*?"
"*No*."
"*What time of the day is the pain the worst*?"
"*After lunch*" she replies.
"*Is there anything that makes the pain worse*?"
"*I don't know*."
When asked about school, she responds that she likes her teachers and the classroom activities.
"*Do you have friends in school*?"
Jamie says, "*Yes*."
"*Can you tell me the name of one of your friends*?"
She is silent.
You turn to Jamie's mother and ask, "*Does she have any play dates outside of school*?"
Mom replies, "*She occasionally invites a younger girl from the neighborhood to come and play with her.*

Jamie's mother later tells you in private that while the younger neighbor occasionally does come over to play, she never invites Jamie to come to her house. She also lets you know that her schoolmates have never invited Jamie to any birthday parties or out-of-school activities.

On physical exam Jamie is a warm and friendly 8-year-old with the typical facial features of Down syndrome. Her height is 126 cm. and her weight is 27 kg. Her body mass index is 17.0 (71 % on the NCHS growth chart). Her skin is dry, and she has mild atopic dermatitis. She wears glasses and has epicanthal folds and Brushfield spots on the outer third of the iris. She has an open bite with a tongue thrust. Her tympanic membranes are dull but not injected, and the motility is mildly reduced. Her thyroid gland is non-palpable. Heart and lung sounds are normal. Abdominal exam reveals a midline surgical scar, and a moderate amount of palpable stool in the LLQ. Her breast and pubic hair are Tanner Stage I. You note a bilateral hallux valgus deformity. Her overall muscle tone is diminished with hyperextensibility of joints, and she has decreased truncal stability and rounding of the shoulders. There are no lateralizing neurological signs. Her gait is slightly immature although symmetric.

Finding New Friends

Down Syndrome

Results:

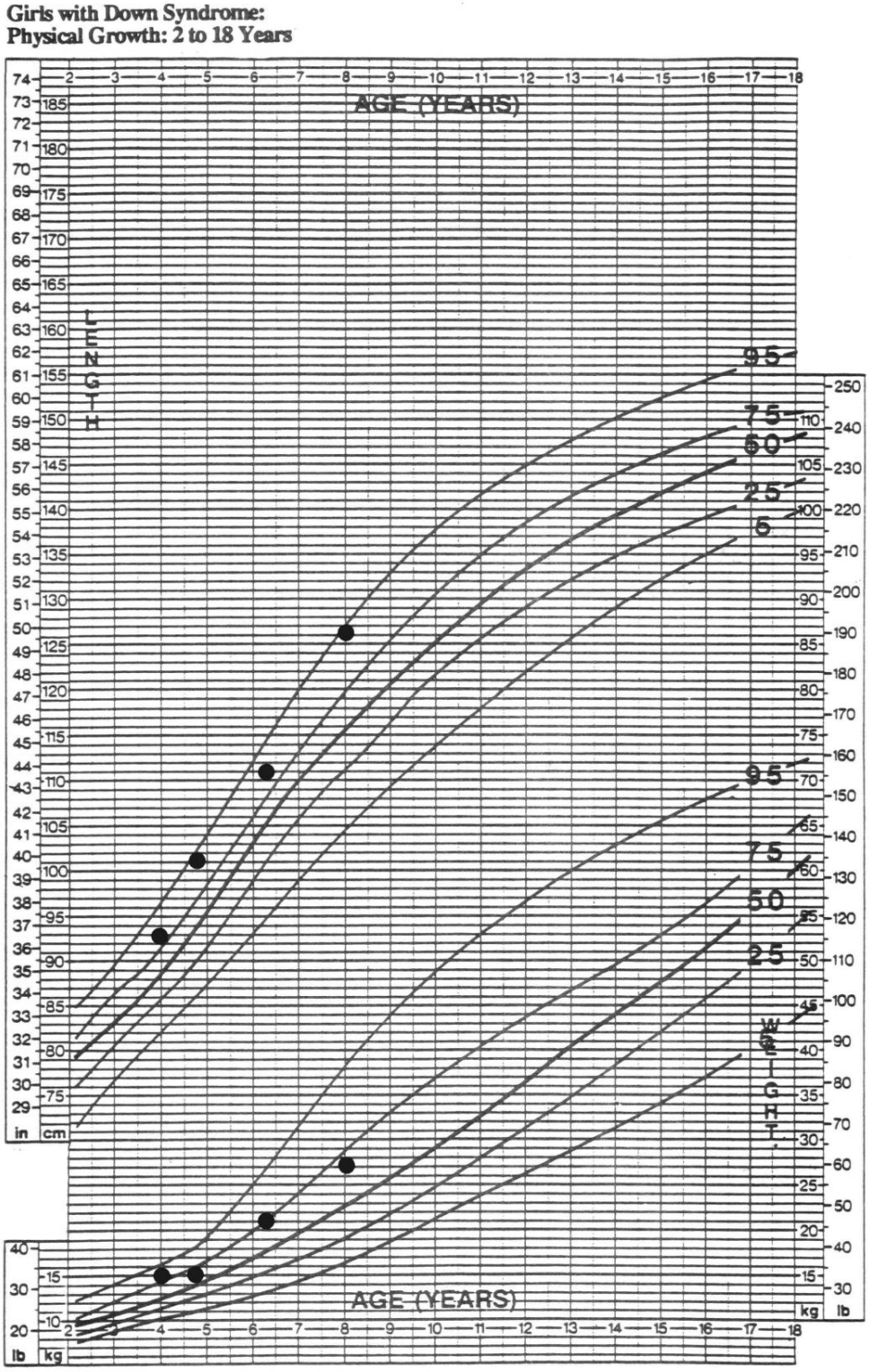
Lateral neck x-ray shows no evidence of occipito-atlanto or atlanto-axial instability. Her thyroid function tests are all normal. Audiometry reveals that her left ear is normal, and her right ear has mild low frequency conductive hearing loss. Her tympanogram is normal for the left, and shows negative middle ear pressure on the right.

Finding New Friends
Down Syndrome

Part III
Epilogue

Following her benign medical work-up, you reassured Jamie and her mother that there was no serious medical problem underlying her neck pain. Following a consultation with a Behavioral Medicine specialist, Jamie's parents instituted a behavior management plan that included positive reinforcement (e.g., a sticker chart with subsequent reward) for staying in class and following teacher directions. Jamie continued to complain of pain in her head and neck. Her parents were initially reluctant to involve her in activities with other children who have developmental disabilities, as they feared that Jamie could learn additional negative behaviors. They ultimately decided to try *Special Olympics*, which has on-going activities during the entire year. Jamie found acceptance there and had a wonderful time. She subsequently joined a dance class with typically developing peers at the local community center. Her complaints of head and neck pain have progressively diminished. She is not always compliant in school and the behavioral management plan needs to be reactivated from time to time.

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



Finding New Friends
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