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

**Handout #2: Healthwatch for the Person with Down Syndrome**

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

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.

### Genetic Counseling

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

Screen for acquired mitral or aortic valvular disease

### Cardiology

Echo

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

Annual ophthalmology appointment

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)

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.

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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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**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 2½ 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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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

Suggested Readings (Annotated):
Comprehensive and up-to-date summary by an outstanding clinician with experience caring for children with Down syndrome.


National Organizations

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