Objectives

- Provide an overview of the 2011 AAP Health Supervision Guidelines for Children with Down Syndrome
- Identify some of the common medical vulnerabilities of children with Down syndrome
- Discuss available resources

Where do our guidelines for health care come from?

- American Academy of Pediatrics
- Guidelines are based on current available research and expert opinion
- Present the ideal standard of care – not always practical

Health supervision for children with Down syndrome.
Clinical report of the Committee on Genetics.
*Pediatrics.* 2011;128(2):393-406

I wish I'd known...

That my daughter would be so able: to clean her own room, to charm grown men, to plant the garden with me, to play dress-up with other little girls, to read and write, and to be a friend that cares and shares. 
—Jeni, mom to Joy Daisy (6 years)

I wish I'd known...
Not everything I read would happen to my child. I would love her more than anything else on Earth. Her hugs and kisses are real, from her heart. She is just like any other kid.
—Jennifer, mom to Brook (2 years)

I wish I'd known...
That it was OK to feel broken-hearted when we first heard the news. That having a child with Down syndrome is NOT a life sentence. That my son would have his own agenda for doing things and that it’s a privilege to be his mom.
—Sara, mom to Nathaniel (9 years)

**Suspecting the diagnosis**

- Flat nasal bridge: 83%
- Epicanthal folds: 60%
- Upslanting palpebral fissures: 98%
- Single palmar crease: 60%
- Gap between 1st & 2nd toes: 96%
- Low muscle tone: 100%
Medical Issues: Cardiac

Congenital heart disease
- 40-50% of babies
- Complete AV canal
- Most children with DS in the past died in early childhood

Cardiac disease

- Teens with history of heart defects need to see their cardiologist as scheduled
- Cardiac issues can appear in teens and adults – even without a prior history of heart problems
  - Symptoms
  - Changes in exam

Medical Issues: Blood Disorders

Increased risk of leukemia, especially in first 2 years of life
- Petechiae or multiple bruises
- Prolonged fever
- Significant weight loss
Iron Deficiency Anemia

- Iron deficiency common in children with Down syndrome
- Screen at every visit – New

My baby is not eating well - she throws up and is constipated

Feeding

- Oral motor feeding and swallowing issues
- Duodenal atresia
- Aspiration-Thickened feeds
- Modified barium swallow – New 2011

Medical Issues: Gastrointestinal

- Gastroesophageal reflux
- Constipation
- Celiac disease

GE Reflux

- Contents from the stomach come up into the esophagus (food pipe)
- The muscle that is at the junction of the esophagus and the stomach is the lower esophageal sphincter
- Reflux happens when the muscle/sphincter is not working well

Constipation

Passage of hard, painful stools. In severe cases, the child is unable to pass stools on his own.

Due to inadequate fiber in the diet (retains water in stool) or because the stool is kept too long in the rectum allowing more water to be reabsorbed leaving the stool dry and hard.
Celiac disease

- Estimated to occur in 7-15% of people with DS
- Exposure to gluten causes inflammation of the intestinal lining
- Symptoms
  - diarrhea, weight loss
  - constipation, bloating, flatulence, abdominal pain
  - weakness, fatigue
  - poor absorption of nutrients

How and when to test?

- Signs and symptoms of celiac disease should be assessed annually at routine physical exams
- Celiac disease should be considered anytime a person with DS has prolonged period of possible symptoms
- Initial screening is with a blood test; intestinal tissue may be needed to confirm the diagnosis
- In the CMH DS clinic we screen every patient at least once for celiac disease, then re-screen if indicated.

Medical Issues: Hypothyroidism

- May be congenital, acquired, or autoimmune
- Symptoms overlap with Down syndrome: constipation, dry skin, slow growth velocity, obesity, hair loss and developmental problems
- Screen annually
Why does my doctor test hearing at every visit?

- Children with DS may have neural, conductive 50-60% or mixed hearing loss
- Hearing loss affects language development
- Hearing loss is treatable

Why should my child be examined by the eye specialist?

- Strabismus (crossed-eyes)
- Myopia (near-sightedness)
- Congenital cataracts
- Nystagmus (rhythmic jerking movements)
- Vision issues often affect ability to perform therapies
- Examination by pediatric ophthalmologist by 6 months
- Yearly till 4 years of age and then every 2 years

When should the hearing be tested?

- Newborn hearing screen
- Rescreen within six months
- Screen every six months until normal hearing is established
- Then hearing test annually

Medical Issues: Neurologic

- Low muscle tone – improves in early years
- Seizures:
  - Occur in 10%
  - Myoclonic/hypsarrhythmia
  - Bimodal – early childhood and middle age

Developmental Expectations

Delays in all area of development
- Gross/fine motor
- Cognitive
- Language

Cognitive Disabilities

- Average IQ in the mild cognitive delay range
- 5% in severe/profound range
- Testing method can affect IQ score
**Language Disorder**
- 75% have an expressive language disorder
- Identifiable between ages 1-2 yrs
- Sign language
- Other communication device-iPads

**Skin Conditions**
- Skin is often very dry
  - Prone to secondary inflammation and eczema
- Cracking/fissuring of lips (esp. in winter)
- Folliculitis (esp. over buttocks)
- 10% develop alopecia areata (hair loss)
- Fungal infections of toe and finger nails are common

**Dental Issues**
- Later eruption of all teeth
- Irregular patterns of eruption
- One or more missing teeth
- No increased likelihood of dental disease (decay or gum disease) with good hygiene
- Poor dentition can affect feeding therapies
- Dental visits at 1 yr or with eruption of first tooth and every 6 months
Behavioral Impairments

- ADHD
- Autism

What is Autism?

- The autism spectrum disorders are a group of disorders of the brain that affect:
  - The ability to communicate
  - The ability to form relationships
  - Behavior

Autism and Down syndrome

- Sometimes it is difficult to distinguish learning disabilities or intellectual disability in children with Down syndrome from the characteristics of Autism
- The most distinguishing characteristics are:
  - Regression of achieved milestones like loss of language
  - Restrictive and repetitive behaviors
### Strengths of Children with DS

- Able to communicate with sign, gesture or pictures
- Enjoy peer interaction-play imitation & modeling
- Affectionate, joyful, eager to please
- Great visual learners
- Musicality

### Obesity

*Why are individuals with Down syndrome more prone to this?*

- Daily calorie needs are ~10-15% lower due to lower muscle tone and overall decreased activity levels
- Inactivity
- Decreased satiety
- Feeding issues, pickiness
- Use of food for reward and entertainment

### Height and weight
Obesity
Why should we be concerned with a few extra pounds?

• Overweight and obesity associated with many debilitating and sometimes life threatening medical conditions
  – Cardiovascular disease
  – Sleep apnea
  – Type 2 diabetes
  – Orthopedic problems
• Obesity often becomes a vicious cycle

Sleep apnea

• Pauses in breathing during sleep
• Results in decreased oxygen, increased carbon dioxide in the bloodstream
• Most sleep apnea in patients with DS is “obstructive,” increases with obesity
• Extremely common in people with DS

Sleep apnea
Signs and symptoms

- Snoring
- Odd sleep positions
- Excessive daytime sleepiness, decreased energy
- Behavioral issues, irritability
- Pauses in breathing are not always observed
Sleep apnea

*Evaluation*

- Guidelines suggest that every child with DS have a sleep study before age 4
  - Not always possible
- Every child with DS should be screened annually for signs and symptoms of sleep apnea, and referred to a sleep specialist as indicated.

Atlantoaxial instability (AAI)

- Related to general ligamentous laxity seen in people with DS
- AAI refers to the slipping of the vertebrae in the neck out of alignment
- Can damage nerves
- Occurs in relatively few patients

Atlantoaxial instability

*Signs and symptoms*

- Holding the head in unusual positions
- Pain – neck, area supplied by a particular nerve
- Weakness in arms or legs, change in gait
- Bowel and/or bladder incontinence
- Abnormal breathing pattern
- Symptoms can progress over time
Atlantoaxial instability
What is the best way to screen?

- Lots of controversy!
- Routine X-rays during adolescence no longer recommended by the health care guidelines
  *Evaluate when signs and symptoms appear*
- X-rays not yet removed from Special Olympics pre-participation forms

Immunizations

- All children with DS should be immunized according to ACIP recommendations
- Vaccine preventable diseases
  - are still with us
  - can be life-threatening
- Non-immunized individuals
  - are at risk of developing preventable diseases
  - can be carriers of life-threatening diseases to vulnerable people around them

Puberty
All children experience similar changes as they go through puberty, but... Adolescents with DS may face them without the same preparation or understanding

Emotional and behavioral changes of puberty

- All youth experience emotional and behavioral changes during puberty and adolescence.
- Emotional and behavioral changes may begin before or during physical changes and are highly variable and individual.

Emotional and behavioral changes of puberty

- Striving for greater independence
- Questioning family and school rules
- Moodiness
- “Attitude”
- “Drama”
- Increased aggression
- Non-compliance
- Need for more sleep
Menstruation

• Menstrual patterns in girls with DS are no different from those in typical girls
• Menstrual cycles are often irregular or unpredictable at first, but with time become more cyclic
• Girls with DS who have good self-care skills are able to take care of menstrual hygiene

Medical management of periods

• Various contraceptive medications can be used to try to shorten and lighten periods, or make them occur less often.
• No medication can completely prevent menstruation. Girls will still need to know how to care for their periods.

Children’s Mercy Hospital Down Syndrome Clinic

• Multidisciplinary clinic
  – Pediatrician and Adolescent Physician
  – Occupational Therapist
  – Speech Pathologist
  – Registered Dietician
  – Audiologist (ENT if needed)
  – Behavior Specialist
  – Social Work
  – Team Coordinator
  – Genetic Counselor
Resources

- American Academy of Pediatrics:
  Health Supervision for children with Down syndrome:
  http://pediatrics.aappublications.org/content/early/2011/07/21/peds.2011-1605
- National Down Syndrome Society:
  www.ndsccenter.org
- Down Syndrome-Autism Connection
  http://ds-asd-connection.org/